ABP Content Specs for Developmental-Behavioral Pediatrics Fellow Collaboration Project

Developmental-Behavioral Pediatrics Fellow Collaboration Project
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A Word of Thanks and Acknowledgement

It is such an honor to be able to write a small note of thanks and acknowledgement to all the DBP fellows and staff who contributed to this wonderful project. I think we can all agree that 2020 has been, in a word, challenging. We all have been overwhelmed with juggling so much over this year with managing patients virtually, finding much needed resources and supports for the families we work with, maintaining momentum on research and PI/QI projects that were threatened to come to a halt, and all while trying to keep our lives at home afloat during such tumultuous and uncertain times. One area I was worried about for myself at the start of the COVID-19 situation was keeping up with my own independent learning and study. I knew that with four young children at home I would need to figure out a way to stay organized and systematic if I was to remain focused at all with life's inevitable and constant interruptions. When I talked about my plan to start writing out the ABP DBP Content Specifications with Dr. Flake though, he was the major force behind taking an idea from small scale to BIG scale by introducing me to the DBP fellowship programs and departments that volunteered their own time to generate this amazing collaboration!

One of the greatest unexpected joys of getting to organize this project was being able to connect with so many of my DBP colleagues across the country. Having an opportunity to work with all of you and to stay in communication about your progress on your sections truly bolstered my spirits. It is ironic that at a time in my life when I felt so isolated from so much of my usual "normal," I still had a chance remotely to feel connected with so many people who will be a huge part of my professional life. It really affirmed to me that I had picked the right "tribe" to be a part of for the rest of my medical career. You all inspire me!

All I hope is that this collaboration will continue on in the future! The purpose of this project from the very beginning was simply to freely share an end product with all DBP fellows and staff across the country for the benefit of shared/joint learning. There was never any other intent. I would love this to become a living, breathing document that DBP programs can continue to work on, update, revise, review, and share with each other for years to come! The next page lists all of the individuals from the 18 (yes, 18!) different DBP fellowship programs and departments who contributed to this finished product. Seeing all of our names listed out really reminds me how awesome it is that we were able to get this done working together as such a large and impressive group! A very special "thank you" has to be sent out as well to our DBP fellowship administrative coordinator, Norbilyn Bernardo, for compiling all of our work into one compressed document that could be sent out via email!

Thank you all again so much for your tireless efforts and for sticking through this undertaking until the end. I sincerely hope we all meet together face to face very soon in a not-too-distant happy and healthy future!

Dr. Rebecca (Becky) Christi, MD, FAAP

Lt Col, USAF, MC

Developmental Behavioral Pediatric Fellow (FEL-3)

Madigan Army Medical Center

DBP Program/Academic Center/Hospital	Faculty Contributor/Reviewers	Contributing Fellows/Residents
1-Madigan Army Medical Center	1. Dr. Eric Flake 2. Dr. Bonnie Jordan	Fellows: 1. Dr. Matthew Scott 2. Dr. Jonathan Chooey 3. Dr. Meagan Butsch 4. Dr. Paul Patterson 5. Dr. Kira Belzer 6. Dr. Josh Strait 7. Dr. Rebecca Christi
2-Baylor Developmental Behavioral Pediatrics	Dr. Noel Mensah-Bonsu	Fellows: 1. Dr. Ann Kennelly 2. Dr. Maja Katusic 3. Dr. Veronica Villarreal
3-Boston Children's	1. Dr. Demetra Pappas 2. Dr. Leonard Rappaport	Fellows: 1. Dr. Liesl Windsor 2. Dr. Cassie Conrad 3. Dr. Audrey Christiansen 4. Dr. Helene Pinches 5. Dr. Lianna Lipton
4-UCLA	Dr. Irene Koolwijk	Fellows: 1. Dr. Melissa Harada 2. Dr. Preeya Desai
5-Nebraska	1. Dr. Howard Needelman 2. Dr. Grace Winningham	Fellow: Dr. Beatrice Egboh
6-UW/Seattle Children's Hospital	1. Dr. Samuel Zinner 2. Dr. Emily Myers	Fellows: 1. Dr. Erika Phelps- Nishiguchi 2. Dr. Anisha Srinivasan
8-Developmental and Behavioral Pediatrics: Atrium Health-Levine Children's Hospital	Dr. Yasmin Senturias	Pediatric Residents: 1. Dr. Shailly Gaur 2. Dr. Alexa Ernst 3. Dr. Anna Rees
9-OHSU	Dr. Randall Phelps	
10-UC Davis	Dr. Kathleen Angkustsiri	Fellows: 1. Dr. Petrina Kaluzhny 2. Dr. Rosa Denisse Rodriguez 3. Dr. Ruchi Punatar
11-Minnesota	Dr. Andrew Barnes	Fellow: Dr. Adam Langenfeld

12-UH Rainbow Babies and Children's-Cleveland	Dr. Shanna Kralovic	Fellows: 1. Dr. Carrie Cuffman 2. Dr. Tanaporn 'Jasmine' Wilaisakditipakorn
13-Brown/Hasbro Children's	1. Dr. Pam High 2. Dr. Carrie Kelly	Fellows: 1. Dr. Devina Savant 2. Dr. Dalal Elsori 3. Dr. Starrina Gianelloni **4.** Dr. Stephanie Klees **5.** Dr. Rene Bartos
14- UChicago	Dr. Michael Msall	
15- Boston Medical Center	Dr. Naomi Steiner	Fellows: 1. Dr. Ana Treadaway 2. Dr. Mediatrix Mbamalu 3. Dr. Christine McGivney
16- Children's Hospital Colorado	1. Dr. Abigail Angulo 2. Dr. Sandra Friedman	Fellows: 1. Dr. Sara Williams 2. Dr. Puji Jonnalagadda
17- UCSD	1. Dr. Theodora Nelson 2. Dr. Lauren Gist	Fellows: 1. Dr. Caroline Sawyer 2. Dr. Cecilia Rhodus 3. Dr. Anthony Kuleto 4. Dr. Petrina Kaluzhny
18- Michigan	Dr. Barbara Felt	Fellows: 1. Dr. Kimberley Levitt 2. Dr. Nicole Hamp 3. Dr. Chioma Torres

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ABP Content Specs for Developmental-Behavioral Pediatrics Fellow Collaboration Project

Prepared by Rebecca Christi, MD, Madigan Army Medical Center DBP Fellow

Reviewed by Abigail Angulo, MD, MPH, Colorado Children's DBP Fellowship Director

Foundations of Developmental-Behavioral Pediatrics

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      b. Know that the progression of gross motor development proceeds in a cephalo- caudal fashion
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I. Foundations of Developmental-Behavioral Pediatrics

A. Domains of development

1. Motor

a. Know the developmental milestones of normal gross motor development in the first year after birth.

1) The ultimate goal of gross motor development is to gain independent and volitional movement

a) During gestation, primitive reflexes develop and persist for several months after birth to prepare the infant for the acquisition of specific skills

b) These brainstem and spinal reflexes are stereotypic movements generated in response to specific sensory stimuli- include Moro reflex, asymmetric tonic neck (ATNR) (Figure 1), and positive support reflexes

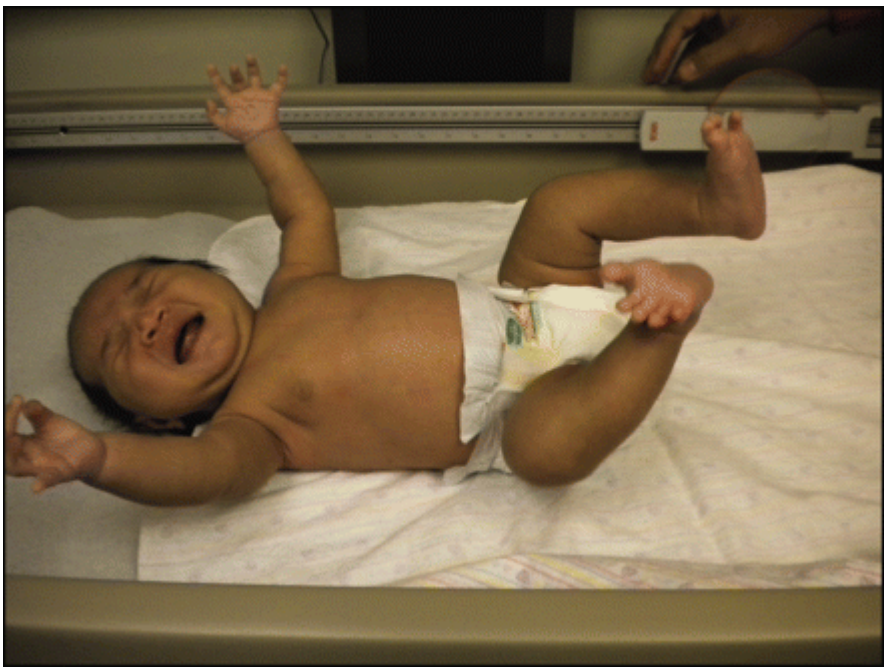


Figure 1. Image of Moro reflex. This reflex occurs spontaneously to loud noises or by simply holding the supine infant's hand and releasing the hand suddenly. Classically, the reflex is elicited while holding the infant supine, with the head dropped slightly backward. This produces sudden extension and abduction of the upper extremities with hands open, followed by flexion of the upper extremities to midline (the "startle reflex"). (Image from Gerber and Erdie-Lalena, 2010)

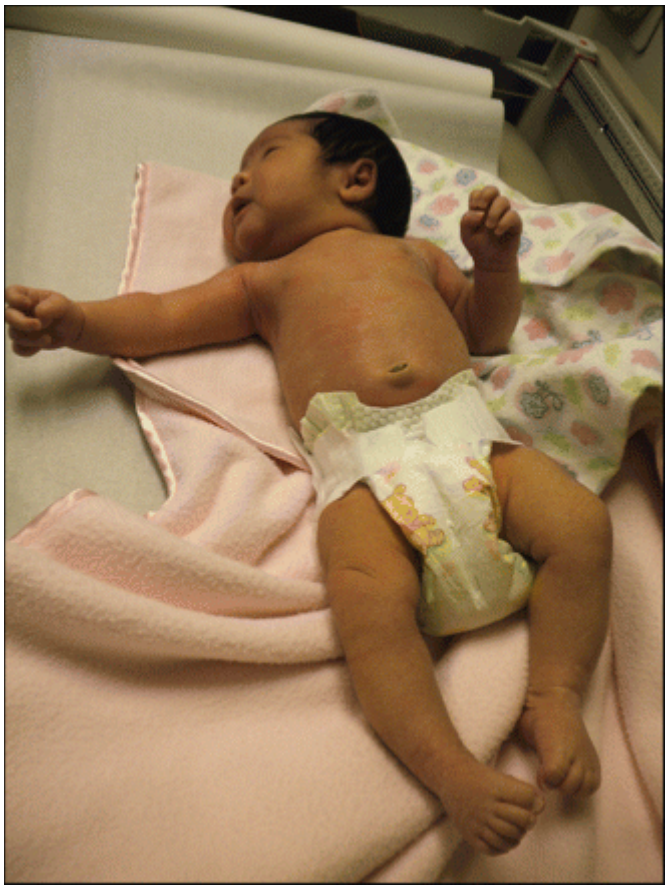


Figure 2. Image of Asymmetric tonic neck reflex (ATNR). The sensory limb of the ATNR involves proprioceptors in the cervical vertebrae. With active or passive head rotation, the baby extends the arm and leg on the face side and flexes the extremities on the contralateral side (the “fencer posture”). There also is some subtle trunk curvature on the contralateral side produced by mild paraspinous muscle contraction. (Image from Gerber and Erdie-Lalena, 2010)

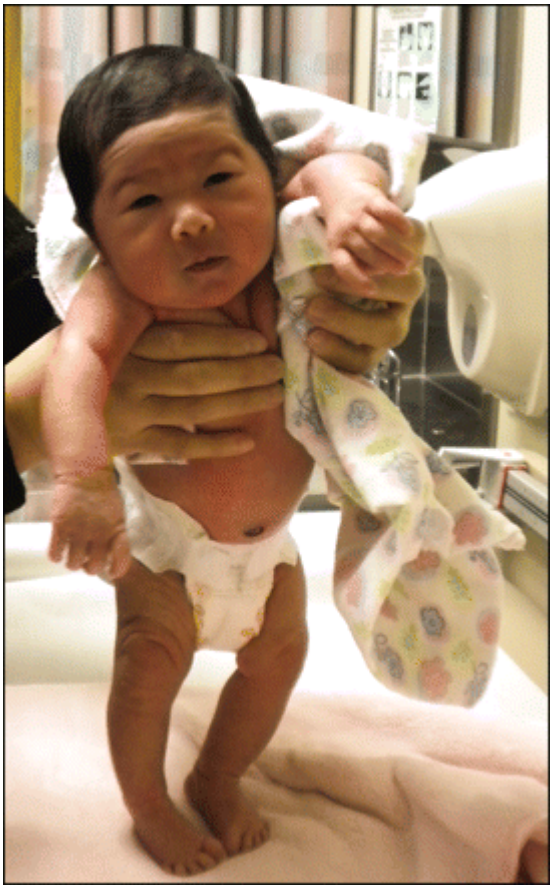


Figure 3. Positive support reflex. With support around the trunk, the infant is suspended, then lowered to touch the feet gently on a flat surface. This produces reflex extension at the hips, knees, and ankles so the infant stands up, completely or partially bearing weight. Mature weight-bearing lacks the rigid quality of this primitive reflex. (Image from Gerber and Erdie-Lalena, 2010)

2) As the central nervous system matures, the reflexes are inhibited to allow the infant to make purposeful movements

a) For example, during the time when the ATNR persists, an infant is unable to roll from back to front, bring the hands to midline, or reach for objects→this reflex disappears between 4 and 6 months of age, however, at the same time that these skills begin to emerge

b) Along the same lines, the Moro reflex interferes with head control and sitting equilibrium→as this reflex lessens and disappears by 6 months of age, the infant gains progressive stability in a seated position

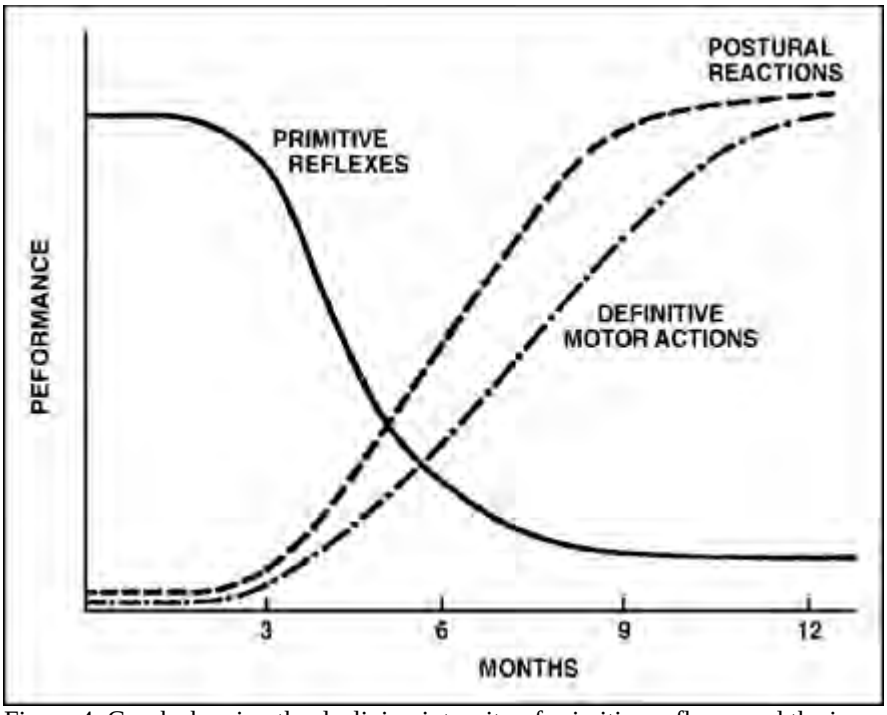


Figure 4. Graph showing the declining intensity of primitive reflexes and the increasing role of postural reactions represent at least permissive, and possibly necessary, conditions for the development of definitive motor reactions. (Johnson CP, Blasco PA. Infant growth and development. *Pediatr Rev.* 1997;18: 225–242.)

3) In addition to primitive reflexes, postural reactions, such as righting and protection responses, also begin to develop after birth
    a) These reactions, mediated at the midbrain level, interact with each other and work toward the establishment of normal head and body relationship in space.
    b) Protective extension, for example, allows the infant to catch him- or herself when falling forward, sideways, or backwards
    c) These reactions develop between 6 and 9 months, the same time that an infant learns to move into a seated position and then to hands and knees.

Figure 5. Image depicting Lateral protection. In the seated position, the child is pushed gently but rapidly to one side. The reaction is present if the child puts out his or her hand to prevent a fall. (Image from Gerber and Erdie-Lalena, 2010)

4) Soon afterward, higher cortical centers mediate the development of equilibrium responses and permit the infant to pull to stand by 9 months of age and begin walking by 12 months.

a) Additional equilibrium responses develop during the second year after birth to allow for more complex bipedal movements, such as moving backward, running, and jumping.

b) During the first postnatal year, an infant thus moves from lying prone, to rolling over, to getting to hands and knees, and ultimately to coming to a seated position or pulling to stand (Fig. 6).

c) It is important to note that crawling is not a prerequisite to walking; pulling to stand is the skill infants must develop before they take their first steps.

d) The ultimate goal of this timeframe is to develop skills that allow for independent movement and freedom to use the hands to explore, manipulate, and learn from the environment.

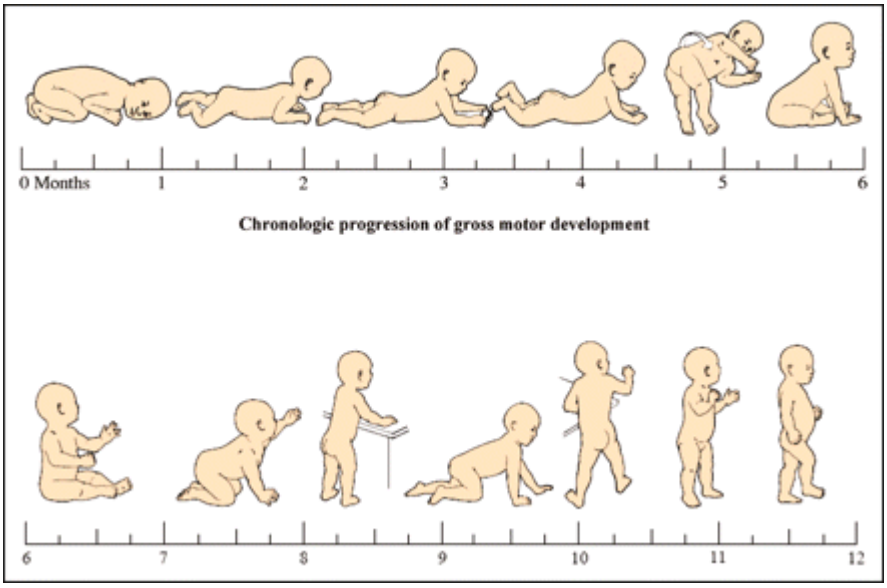


Figure 6. Image showing the Chronologic progression of gross motor development during the first 12 postnatal months. (Johnson CP, Blasco PA. Infant growth and development. *Pediatr Rev.* 1997;18: 224–242.)

5) Gross motor development in subsequent years consists of refinements in balance, coordination, speed, and strength.
a) The wide-based, slightly crouched, staccato gait of a 12-month-old evolves into a smooth, upright, and narrow-based style. The arms change from being held abducted and slightly elevated for balance to swinging in a reciprocal fashion as the gait reaches an adult pattern by age 3 years.
b) Similarly, running develops soon after walking, starting as a stiff-legged approximation and changing into a well-coordinated movement that includes rapid change of direction and speed by 18 months of age.
c) Simultaneous use of both arms and legs occurs after successful use of each limb independently.
d) At age 2 years, a child can kick a ball, jump with two feet off the floor, and throw a big ball overhand. Milestones for succeeding ages reflect progress in the length of time, number of repetitions, or the distance each task can be performed successfully.
e) By the time a child starts school, he or she is able to perform multiple complex gross motor tasks simultaneously (such as pedaling, maintaining balance, and steering while on a bicycle).

Gross Motor Milestone	Mean Age
Lifts head only	1 mo
Lifts head prone to wrists	4 mo
Rolls over prone to supine	4 mo
Rolls over supine to prone	5 mo
Sits supported	5 mo
Sits alone	6 mo
Comes to sit	8 mo
Crawls	8 mo
Pulls to stand	9 mo
Cruises	9 mo
Stands well. Walks alone	12 mo

Table 1. Gross motor milestones

b. Know that the progression of gross motor development proceeds in a cephalo-caudal fashion.

1) The pattern of motor development and gain of skills progresses in what is described as cephalocaudal- or the cephalocaudal principle → describes the direction of motor growth and development
2) The cephalocaudal theory states that muscular control develops from the head downward: first the neck, then the upper body and the arms, then the lower trunk and the legs.
3) Motor development from birth to six months of age includes initial head and neck control (within first 2 months), then hand movements and eye-hand coordination, followed by preliminary upper body control.

4) Coordination of the arms always precedes coordination of the legs- By 6 to 12 months of age, infants start to gain leg control and may be able to crawl, stand, or walk

c. Know the developmental milestones of normal fine motor development in the first year after birth.

1) Fine motor skills relate to the use of the upper extremities to engage and manipulate the environment.
2) They are necessary for a person to perform self-help tasks, to play, and to accomplish work.
3) Like all developmental streams, fine motor milestones do not proceed in isolation but depend on other areas of development, including gross motor, cognitive, and visual perceptual skills.
4) At first, the upper extremities play an important role in balance and mobility.
    a) Hands are used for support, first in the prone position for lifting the head and then in sitting. Arms help with rolling over, then crawling, then pulling to stand.
    b) Infants begin to use their hands to explore, even when in the supine position.
    c) When gross motor skills have developed, such that the infant is more stable in upright positions and can move into these positions easily, the hands are free for more purposeful exploration.
5) At birth, infants do not have any apparent voluntary use of their hands
    a) They open and close them in response to touch and other stimuli, but movement otherwise is dominated by a primitive grasp reflex.
    b) Because of this, infants spend the first 3 months after birth "contacting" objects with their eyes rather than their hands, fixating on faces and objects and then visually tracking objects.
6) Gradually, they start to reach clumsily and bring their hands together
    a) As the primitive reflexes decrease, infants begin to prehend objects voluntarily, first using the entire palm toward the ulnar side (5 months – "raking") and then predominantly using the radial aspect of the palm (7 months – "palmar-radial grasp")
7) At the same time, infants learn to release objects voluntarily.
    a) In the presence of a strong grasp reflex, objects must be removed forcibly from an infant's grasp or drop involuntarily from the hand.
    b) Voluntary release is seen as the infant learns to transfer objects from one hand to the other, first using the mouth as an intermediate stage (5 months) and then directly hand-to-hand (6 months).
8) Between 6 months and 12 months of age, the grasp evolves to allow for prehension of objects of different shapes and sizes
    a) The thumb becomes more involved to grasp objects, using all four fingers against the thumb (a "scissors" grasp) at 8 months, and eventually to just two fingers and thumb (radial digital grasp) at 9 months.
    b) A pincer grasp emerges as the ulnar fingers are inhibited while slightly extending and supinating the wrist.
    c) Voluntary release is awkward at first, with all fingers extended. By 10 months of age, infants can release a cube into a container or drop things onto the floor.

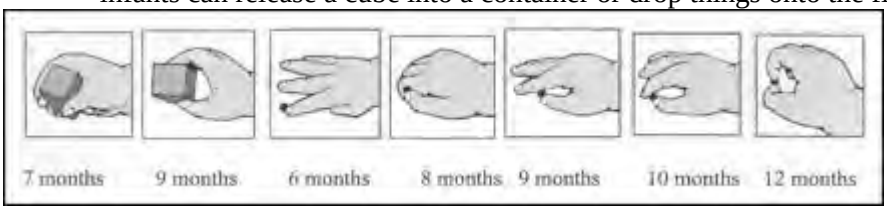


Figure 7. Development of pincer grasp. Illustrations from the Erhardt developmental prehension. In Erhardt RP. *Developmental Hand Dysfunction: Theory Assessment, Treatment.* 2nd ed. San Antonio, Tex: Therapy Skill Builders; 1994.

9) Object permanence reinforces the desire to practice this skill over and over

a) Intrinsic muscle control develops to allow the isolation of the index finger, and infants will poke their fingers into small holes for exploration

10) By 12 months of age, most infants enjoy putting things into containers and dumping them out repeatedly.

a) They also can pick up small pieces of food with a mature pincer grasp and bring them to their mouths.

11) As infants move into their second year, their mastery of the reach, grasp, and release allows them to start using objects as tools. Fine motor development becomes more closely associated with cognitive and adaptive development, with the infant knowing both what he or she wants to do and how he or she can accomplish it.

a. Intrinsic muscle refinement allows for holding flat objects, such as crackers or cookies.

b. By 15 months of age, voluntary release has developed further to enable stacking of three to four blocks and releasing small objects into containers.

c. The child starts to adjust objects after grasping to use them properly, such as picking up a crayon and adjusting it to scribble spontaneously (18 months of age) and adjusting a spoon to use it consistently for eating (20 months of age).

12) In subsequent years, fine motor skills are refined further to draw, explore, problem-solve, create, and perform self-help tasks.

a. By age 2 years, children can create a six-block tower, feed themselves with a spoon and fork, remove clothing, and grasp and turn a door knob.

b. They have sufficient control of a crayon to imitate both vertical and horizontal lines. In-hand manipulation skills permit them to rotate objects, such as unscrewing a small bottle cap or reorienting a puzzle piece before putting it in place.

c. They are able to wash and dry their hands.

d. By 36 months of age, they can draw a circle, put on shoes, and stack 10 blocks.

e. They make snips with scissors by alternating between full-finger extension and flexion. Their grasp and in-hand manipulation skills allow them to string small beads and unbutton clothes.

f. At age 4 years, a palmar tripod grasp allows for finer control of pencil movements, and the child can copy a cross, a square, and some letters and numerals and can draw a figure of a person (the head and a few other body parts). Scissor skills have progressed to permit the cutting of a circle.

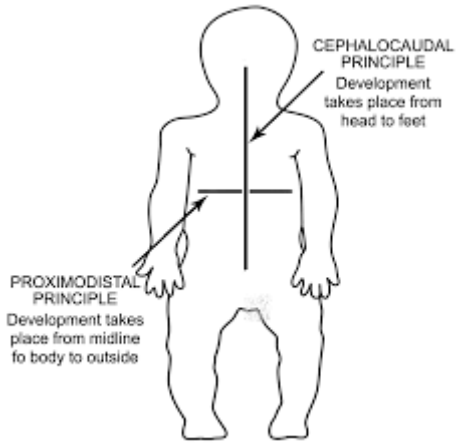
13) When a child reaches the age of 5 years, he or she can dress and undress independently, brush the teeth well, and spread with a knife. More precise in-hand manipulation skills enable the child to cut a square with mature scissor movements (independent finger use) and to print his or her own name and copy a triangle using a mature tripod pencil grasp (using the fingers to move the pencil rather than the forearm and wrist).

Fine Motor Milestones	Mean Age Achieved
Hands fisted near face	1 mo
Hands unfisted 50%	2 mo
Retains rattle if placed in hand	2 mo
Holds hands together	2 mo
Hands unfisted 75%	3 mo
Inspects fingers	3 mo
Bats at objects	3 mo
Clutches at clothes	4 mo
Hands to mouth	4 mo
Reaches persistently	4 mo
Plays with rattle	4 mo
Palmar grasp/cube	5 mo
Transfers object: hand-mouth-hand	5 mo
Holds hands together	5 mo

Attains dangling ring	5 mo
Transfers hand to hand	6 mo
Rakes pellet	6 mo
Takes 2nd cube, holds onto 1st	6 mo
Radial/palmar grasp	7 mo
Bangs spoon w/demo	8 mo
Scissor grasp of pellet	8 mo
Takes cube out of cup	8 mo
Pulls large peg out	8 mo
Scissor pincer grasp of pellet	9 mo
Radial-digital grasp of cube	9 mo
Bangs 2 cubes together	9 mo
Clumsy release of cube	10 mo
Immature pincer grasp of pellet	10 mo
Isolates index finger and pokes	10 mo
Throws objects	11 mo
Stirs with spoon	11 mo
Marks after demo	12 mo
Fine pincer grasp of pellet	12 mo
Holds crayon	12 mo
Attempts tower of 2	12 mo

d. Know that the progression of fine motor development proceeds in a midline to lateral fashion.

1) The principle of **proximodistal (midline to lateral) development, similar to the principle of cephalocaudal development for gross motor skill acquisition,** describes the direction of fine motor skill development and acquisition.
2) This principle reflects the direction of myelination in the brain and spinal cord that occurs in the first two years of life
3) The myelin sheath is set down in the nervous system in the body in two directions: from the head downward (in a *cephalocaudal* direction, from head to tail) and from the torso out to the extremities of the fingers and toes (in a **proximodistal** direction, from the center of the body out toward the extremities).
    a. Proximodistral direction of myelination, from the central axis of the body out to the extremities, results in following steps in development: torso→arms→hands→fingers
    b. The child's arms, therefore, demonstrate coordinated motor function before the hands and the hands and feet develop before the fingers and toes.
    c. Finger and toe muscles (used in fine motor dexterity) are the last to develop in physical development.



e. Know the developmental milestones of normal gross motor development in the toddler age group (12-36 months of age) - see chart below.

1) Gross motor development in subsequent years consists of refinements in balance, coordination, speed, and strength.
2) The wide-based, slightly crouched, staccato gait of a 12-month-old evolves into a smooth, upright, and narrow-based style.
3) The arms change from being held abducted and slightly elevated for balance to swinging in a reciprocal fashion as the gait reaches an adult pattern by age 3 years.
4) Similarly, running develops soon after walking, starting as a stiff-legged approximation and changing into a well-coordinated movement that includes rapid change of direction and speed by 18 months of age.
5) Simultaneous use of either arms or legs occurs after successful use of each limb independently. At age 2 years, a child can kick a ball, jump with two feet off the floor, and throw a big ball overhand.

Gross Motor Milestones	
Walks w/high guard	13 mo
Stands w/o pulling up	14 mo
Walks well	14 mo
Stoops to pick up toy	15 mo
Creeps up stairs	15 mo
Runs stiff-legged	15 mo
Walks carrying toy	15 mo
Climbs on furniture	15 mo
Stands on 1 foot w/slight support	16 mo
Walks backwards	16 mo
Walks up stairs, 1 hand held	16 mo
Creeps down stairs	18 mo
Runs well	18 mo
Seats self in small chair	18 mo
Throws ball standing	18 mo
Squats in play	20 mo
Carries large object	20 mo
Kicks ball w/demo	22 mo
Walks down stairs w/rail	24 mo
Jumps in place, both feet off floor	24 mo
Kicks ball w/o demo	24 mo
Throws overhand	24 mo
Stands one foot, tries	24 mo
3 hops in place	36 mo
Walks backward, 10’	28 mo
Jumps from bottom step, 1 foot landing	28 mo
Walks on toes after demo	28 mo

f. Know the developmental milestones of normal gross motor development in the preschool age group (36-60 months of age) – see chart below

1) Milestones for succeeding ages reflect progress in the length of time, number of repetitions, or the distance each task can be performed successfully.
2) By the time a child starts school, he or she is able to perform multiple complex gross motor tasks simultaneously (such as pedaling, maintaining balance, and steering while on a bicycle).

Gross Motor Milestones	
Balances 1 foot x 3 sec	3 y
Upstairs, alternating feet, no rail	3 y
Pedals trike, 10’	3 y
Standing broad jump 1-2 ft	4 y
Gallops	4 y
Throws ball 10’, 1 or 2 arms	4 y
Catches bounced ball	4 ½ y
Down stairs, alternating feet, w/rail	5 y
Skips	5 y
Stand 1 foot x 8 sec	5 y
Hops 10 times, 15 feet forward	5 y
Running broad jump 2-3 ft	5 y

Walks backward heel-toe	5 y
Jumps backward	5 y
Gross Motor Milestones	
Balances 1 foot x 4-8 sec	4 y
Walks down stairs, alt. feet	4 y
Hops 1 foot x 2-3	4 y
Standing broad jump 1-2 ft	4 y
Gallops	4 y
Throws ball 10', 1 or 2 arms	4 y
Catches bounced ball	4 ½ y
Down stairs, alternating feet, w/rail	5 y
Skips	5 y
Stand 1 foot x 8 sec	5 y
Hops 10 times, 15 feet forward	5 y
Running broad jump 2-3 ft	5 y
Walks backward heel-toe	5 y
Jumps backward	5 y

g. Know the developmental milestones of normal fine motor development in the toddler age group (12-36 months of age)

1) As infants move into their second year, their mastery of the reach, grasp, and release allows them to start using objects as tools.

2) Fine motor development becomes more closely associated with cognitive and adaptive development, with the infant knowing both what he or she wants to do and how he or she can accomplish it.

3) Intrinsic muscle refinement allows for holding flat objects, such as crackers or cookies.

4) By 15 months of age, voluntary release has developed further to enable stacking of three to four blocks and releasing small objects into containers.

5) The child starts to adjust objects after grasping to use them properly, such as picking up a crayon and adjusting it to scribble spontaneously (18 months of age) and adjusting a spoon to use it consistently for eating (20 months of age).

Fine Motor Milestone	Mean Age
Attempts to release pellet in bottle	13 mo
Imitates back & forth scribble	14 mo
Attains 3rd cube by combining 2	14 mo
2 cube tower	14 mo
1 round peg in and out	14 mo
3-4 cube tower	15 mo
Places 10 cubes in cup	15 mo
Releases pellet into bottle	15 mo
All round pegs in with urging	16 mo
Scribbles spontaneously	16 mo
4 cube tower	18 mo
Crudely imitates vertical stroke	18 mo
Completes round peg board w/o urging	20 mo
5-6 cube tower	20 mo
Completes square peg board	20 mo
Closes box with lid	22 mo
Imitates vertical line	22 mo
Imitates circular stroke	22 mo
Train of cubes w/o stack	24 mo
Imitates single circle	24 mo
Imitates horizontal line	24 mo
Strings beads awkwardly	28 mo
Unscrews jar lid	28 mo
8 cube tower	30 mo
Train of cubes w/stack	30 mo
9-10 cube tower	33 mo
6 square pegs in pegboard	33 mo
Imitates cross	33 mo

h. Know the developmental milestones of normal fine motor development in the preschool age group (36-60 months of age). – See chart below

1) In subsequent years, fine motor skills are refined further to draw, explore, problem-solve, create, and perform self-help tasks
2) By age 2 years, children can create a six-block tower, feed themselves with a spoon and fork, remove clothing, and grasp and turn a door knob
    a. They have sufficient control of a crayon to imitate both vertical and horizontal lines. In-hand manipulation skills permit them to rotate objects, such as unscrewing a small bottle cap or reorienting a puzzle piece before putting it in place.
    b. They are able to wash and dry their hands.
3) By 36 months of age, they can draw a circle, put on shoes, and stack 10 blocks.
    a. They make snips with scissors by alternating between full-finger extension and flexion.
    b. Their grasp and in-hand manipulation skills allow them to string small beads and unbutton clothes.
4) At age 4 years, a palmar tripod grasp allows for finer control of pencil movements, and the child can copy a cross, a square, and some letters and numerals and can draw a figure of a person (the head and a few other body parts).
    a. Scissor skills have progressed to permit the cutting of a circle.
5) When a child reaches the age of 5 years, he or she can dress and undress independently, brush the teeth well, and spread with a knife.
    a. More precise in-hand manipulation skills enable the child to cut a square with mature scissor movements (independent finger use)
    b. Also able to print his or her own name and copy a triangle using a mature tripod pencil grasp (using the fingers to move the pencil rather than the forearm and wrist).

Copies circle	3 y
Cuts w/scissors: side to side, awkwardly	3 y
Imitates bridge	3 y
Strings smaller beads well	3 y
Copies triangle	5 y
Builds stairs from model (4/3/2/1)	5 y
Puts paper clip on paper	5 y
Builds stairs from memory	6 y
Draws diamond	6 y
Copies flag	6 y

i. Know the age ranges for the developmental milestones of normal gross motor development in children above 60 months of age.

5 to 6 years	6 to 7 years	7 to 8 years
▪ Walks down stairs, alternating feet, without using rail ▪ Balances on 1 foot: More than 8 seconds ▪ Hops on 1 foot: 15 feet ▪ Skips ▪ Running broad jump: 2 to 3 feet ▪ Walks backward heel-toe ▪ Jumps backward	▪ Tandem walks ▪ Skips	▪ Rides bicycle independently ▪ Bats ball placed on cone ▪ Does somersaults

j. Know the developmental milestones of normal fine motor development in children above 60 months of age

- Copies triangle 5-6 y
- Builds stairs with cubes from model 5-6 y
- Puts paper clip on paper 5-6 y
- Can use clothespins to transfer small objects 5-6 y
- Cuts with scissors 5-6 y
- Writes first name 5-6 y
- Builds stairs with cubes from memory- 6 to 7 years
- Draws diamond- 6 to 7 years
- Copies flag – 6 to 7 years
- Writes first and last name – 6 to 7 years
- Creates and writes short stories – 6 to 7 years
- Forms letters with down-going and counter-clockwise strokes – 6 to 7 years
- Writing rate increases – 7 to 8 years
- Stays on line when writing – 7 to 8 years
- Spaces between words – 7 to 8 years
- Size of letters becomes uniform – 7 to 8 years
- Letter reversals disappear- 7 to 8 years

2. Speech and language

a. Know the developmental milestones of normal language/speech development in the first year after birth

Language/Social (E = Expressive; R = Receptive; S = Social) Milestone	Mean Age
Alerts to voice/sound (R)	1 mo
Throaty noises (E)	1 mo
Coos, vowel-like noises (E)	2 mo
Social smile	2 mo
Regards speaker (R)	3 mo
Chuckles (R)	3 mo
Vocalizes when talked to (E)	3 mo
Orients to voice (R)	4 mo
Stops crying to soothing voice (R)	4 mo
Laughs out loud (E)	4 mo
Vocalizes when alone (E)	4 mo
“Ah-goo” (E)	5 mo
Orients bell I (R)	5 mo
Begins to respond to name (R)	5 mo
Razz, squeal (E)	5 mo
Responds to own name (R)	6-9 mo
Looks, stops, or withdraws to “no” (R)	6 mo
Listens, then vocalizes, when adult stops (E)	6 mo
Smiles/vocalizes to mirror (E)	6 mo
Babbles (E)	6 mo
Orients to bell II (R)	7 mo
Responds to “come here” (R)	8 mo
Looks for family members (R)	8 mo
Responds to “Where’s mama?” (R)	8 mo
Uses some gestures (“up”) (R)	6-9 mo
“Dada” indiscrim. (E)	7 mo
Shakes head for “no” (E)	8 mo
Enjoys gesture games (R)	9 mo
Orients to name well (R)	9 mo
Orients bell III (R)	9 mo
“Mama” indiscrim. (E)	9 mo
Imitates sounds (E)	9 mo

Gesture games (pat-a-cake, peekaboo, so big, I get you) (S)	10 mo
Waves bye back (R)	10 mo
Mama/dada discrim. (E)	10 mo
Follows command w/gesture (R)	11-12 mo
1st word (E)	11-12 mo

b. Know the developmental milestones of normal language/speech development in the toddler age group (12-36 months of age)

Immature jargon- inflection w/o real words (E)	13 mo
3rd word (E)	13 mo
Responds appropriately to "Where's ball?" (R)	13 mo
1-step command w/o gesture (R)	14 mo
Names 1 object	14 mo
Points to 1 body part (R)	15 mo
Points to 1 object of 3 (R)	15 mo
Gets object from another room on demand (R)	15 mo
3-5 words (E)	15 mo
Mature jargoning w/real words (E)	15 mo
Understands simple commands, e.g., "Bring to mommy" (R)	16 mo
Points to 1 picture (R)	16 mo
5-10 words (E)	16 mo
Points to 2 of 3 objects (R)	18 mo
Points to 3 body parts (R)	18 mo
Points to self (R)	18 mo
Understands "mine" (R)	18 mo
10-25 words (E)	18 mo
Giant words; e.g., "all gone," "stop that" (E)	18 mo
Imitates environmental sounds (E)	18 mo
Names 1 picture on demand (E)	18 mo
Points to 3 pictures (R)	20 mo
Begins to understand him, her, me (R)	20 mo
Holophrases, e.g., "mommy?" & points to keys- "these are mom's keys" (E)	20 mo
2 word combinations (E)	20 mo
Answers requests with "no" (E)	20 mo
Points to 4-5 pictures (R)	22 mo
Points to 4-5 body parts (R)	22 mo
Points to 4-5 pieces of clothing (R)	22 mo
25- 50 words (E)	22 mo
Asks for more (E)	22 mo
Adds 1-2 words/week (E)	22 mo
Follows 2-step command (R)	24 mo
Understands me, you (R)	24 mo
2 word sentence (noun/verb) (E)	24 mo
Points to 5-10 pictures (R)	24 mo
Telegraphic speech (E)	24 mo
50+ words (E)	24 mo
Intelligibility to stranger 50% (E)	24 mo
Refers to self by name (E)	24 mo
Names 3 pictures (E)	24 mo
Understands "just one" (R)	28 mo
Repeats 2 digits (E)	28 mo
Begins to use pronouns I, me, you (E)	28 mo
Names 10-15 pictures (E)	28 mo
Follows 2 prepositions ("put block in..on box") (R)	30 mo
Understands action words ("playing…washing…blowing") (R)	30 mo
Echolalia & jargoning gone (E)	30 mo
Knows gender (R)	30 mo
Names objects by use (E)	30 mo

Refers to self with correct pronoun (E)	30 mo
Understands 3 prepositions (R)	33 mo
Understands dry, wet (R)	33 mo
Points to objects by use (ride in…put on feet…write with) (R)	33 mo
Gives 1st, last name, age, sex (E)	33 mo
Counts to 3 (E)	33 mo
Begins to use past tense (E)	33 mo

c. Know the developmental milestones of normal language/speech development in the preschool age group (36-60 months of age)

Sentence length: 3 – 4 words/2.5 – 5 (E)	3 y
200+ words (E)	3 y
Uses plurals (E)	3 y
Uses pronouns correctly (E)	3 y
Intelligibility to stranger: 75% (E)	3 y
Names body parts by use (E)	3 y
Points to parts of pictures (nose of cow, door of car) (R)	3 y
Understands long/short (R)	3 y
Names body parts w/function (R)	3 y
Uses prep. commands (under, behind, in front) (R)	3 y
Identifies 3 colors (R)	3 y
Names 2 colors (E)	3 y
Follows 3 step commands (R)	4 y
Understands action words (swims in water; cut with; read; sit at; tells time) (R)	4 y
Understands adjectives (busy, long, thin, pointed) (R)	4 y
Digits: 3 forward (E)	4 y
300-1000 words (E)	4 y
Tells stories (E)	4 y
Using “feeling” words (E)	4 y
Counts to 4 (E)	4 y
Names 4 colors (E)	4 y
100% intelligible (E)	4 y
Uses past tense (E)	4 y
Digits: 4 forward (E)	5 y
Counts to 10 (E)	5 y
Colors: Names 4-6 (E)	5 y
Defines simple words (E)	5 y
2,000 words (E)	5 y
Knows phone number, home address (E)	5 y
Responds to why questions (E)	5 y
Follows 3-step commands (R)	4-5 y
Points to things that are the same versus different (R)	4-5 y
Names things when actions are described (e.g. it swims in water, you cut with it, it is something you read, it tells time) (R)	4-5 y

d. Know the developmental milestones of normal language/speech development in children above 60 months of age (R= receptive; E= expressive)

- Knows right and left on self- -5-6 y (R)
- Points to different one in a series 5-6 y R
- Understands "er" endings (e.g., batter, skater) 5-6 y R
- Understands adjectives (e.g., bushy, long, thin, pointed) 5-6 y R

- Enjoys rhyming words and alliterations 5-6 y - R
- Produces words that rhyme 5-6 y- R
- Points correctly to "side," "middle," and "corner" 5-6 y – R
- Repeats 6- to 8-syllable sentences – 5-6 y E
- Defines simple words – 5-6 y E
- 2000 word vocabulary – 5-6 y E
- Knows telephone number – 5-6 y E
- Responds to "why" questions – 5-6 y E
- Retells stories with clear beginning, middle, and end – 5-6 y E
- Asks what unfamiliar words mean – 6-7 y R
- Can tell which words do not belong in a group – 6-7 y R
- Repeats 8- to 10-word sentences – 6-7 y E
- Describes events in an orderly way- 6-7 y E
- Knows days of the week – 6-7 y E
- 10,000 word vocabulary – 6-7 y E
- Understands opposites and word analogies – 7-8 y R
- Knows right and left on others – 7-8 y R
- Understands days and months – 7-8 y R
- Masters "r" sound in speech – 7-8 y E
- Tells time – 7-8 y E
- Uses complex and compound sentences -7-8 y E
- Talks about a range of topics – 7-8 y E

e. Understand the typical progression of normal speech and language development

1) Language is a broad concept that involves the representation of thoughts and ideas using culturally agreed-upon arbitrary signals for the exchange of ideas.
    a. Language encompasses both expressive and receptive processes.
    b. Medical literature reflects that early language skills are related to later reading skills.
        i. In school-age children who have specific language impairments→ up to 50% also have a learning disorder involving reading.
2) Clinicians often assess language development incompletely by asking how many words a child knows, but speech, or vocal communication, is only one form of expressive language.
    a. Other forms include the use of gestures, manual signs, facial expressions, body postures, pictures, diagrams, and written symbols.
    b. Each of these has a strong cultural overlay, although some, such as facial expressions, can be understood more universally.
    c. The production of speech is a specific motor skill and requires complex control of air flow, mouth shape, and tongue position.
    d. Impairments in motor planning and execution or anatomic abnormalities can impair speech yet spare other language abilities.
    e. A hearing-impaired child using manual sign language can demonstrate normal expressive language without the use of speech. Rather than asking parents how many words their child uses, an alternative question is, “How does your child communicate with you?” The question can be clarified to ask how the child shows displeasure or happiness or requests a want or a need.
3) Receptive language is the ability to understand communication. It is evaluated most often by a response to a request or question.

   a. However, a request to perform a gross motor task by the child who has gross motor impairment, such as cerebral palsy, may give a false impression of a child's receptive language skills.
   b. A more useful method of assessing receptive language is to ask how the child responds to parental communication.
   c. Language and social development are largely intertwined, and conversations regarding communication help the clinician survey both developmental domains.
4) Language development is molded by the type of interactions the infant has with his environment.
   a. Although the ability to learn language is innate, environmental exposure to language is essential. For example, an infant who has a significant hearing impairment can startle to sound, laugh, and even babble.
   b. Hearing impairment would be easily missed by using observational measures alone, highlighting the importance of systematic hearing screening programs.
   c. With intervention by 6 months of age (e.g., with hearing aids), language outcomes in children who have hearing impairments are similar to children who have no hearing deficits, emphasizing that exposure to language is the key to language development.
   d. The importance of language exposure is demonstrated most strongly and simply by children beginning to speak the language they hear, despite being born with the ability to learn any language.
   e. Ideally, the language to which they are exposed should be as rich and diverse as possible.
   f. Multiple studies have demonstrated that a robust linguistic environment is critical to language development.
5) Interventional approaches, such as reading to infants and children, try to counter this risk by promoting increased frequency and complexity of linguistic exposure.
   a. Other, less successful strategies have involved “educational” videos. These videos fail to understand the interdependence of language and social development and have been shown to impair language development if not paired with social interactions.
   b. Conversational turn-taking and appropriate communicative interaction is not provided through video or television media; these crucial aspects can be provided only by interactions with people who are present and responsive.
6) Infants communicate long before they speak their first words or phrases.
   a. At birth, crying is the primary form of communication. It is nonspecific but very effective in initiating a response from a caregiver.
   b. Expressive communication then progresses through cooing and babbling.
7) In a trial-and-error process, the infant begins making vowel and consonant sounds that she can put together into “mama” and “dada” by 9 months of age.
   a. Although she is not using the words discriminately, if her caregivers respond to the sounds she makes, she will continue to use them.
   b. As her attempts to communicate become more precise and the outcome more predictable, by her first birthday she can say her first word and can point to communicate a request.
   c. The first words attained often are labels for things with which the infant commonly interacts.
   d. There is great variety among children of the same age as to what words they use, emphasizing the importance that environmental stimulus plays in language development.
8) By 15 months, the toddler is able to give a clear “no” with a headshake.
   a. His ability to imitate sounds increases, and he can repeat an entire word and even mimic environmental sounds.
9) By 18 to 24 months of age, he is starting to use pronouns such as “me,” and his vocabulary has expanded to 50 words.
   a. New words are learned quickly, and he begins to combine them into flexible, two-word phrases (noun + verb).
   b. He now is able to communicate basic wants (“more drink”) and social interest (“bye, mama”).
10) Between 2 and 3 years of age, his vocabulary continues to increase, and the phrases he uses increase to 3 to 4 words in length.

a. He begins asking "what" questions frequently. His ability to pronounce words also has improved, and by 2 years of age, at least 50% of his speech is understandable to a stranger.
b. At 3 years of age, 75% is understandable, and the beginning of the "why" questioning occurs. He is able to tell others what he did while they were apart
c. His sentence structure continues to gain complexity, and by 4 to 5 years of age, his speech is completely understandable to strangers.

11) Although the infant's expressive communication is more observable, receptive language skills also are present at birth.
    a. A neonate shows preference for voices and interest in faces. She will begin to turn toward sound.
    b. Early evidence of receptive language ability is her response to "no" and to her name.
    c. Once she can isolate her index finger she can work on pointing.
    d. By 12 to 15 months, she can point to body parts and familiar objects when named.
    e. Her understanding of grammar increases, and she is able to understand pronouns by 18 to 24 months.
    f. By 2 to 3 years of age, she can answer questions, understands the concept of "one," and follows two-step commands.
    g. Her natural ability to categorize has matured, and by 3 to 4 years, she can point to an object in a requested category rather than just by name. She understands much of what is said, including negatives. Her ability to follow complex instructions continues to improve as she begins to prepare for early school experiences.

3. Social-emotional

a. Know the developmental milestones of normal social-emotional development in the first year after birth

Joint attention skills:

- 8-10 months: gaze monitoring
- 10-12 months: following a point
- 12-14 months: proto-imperative pointing
- 14-16 months: proto-declarative pointing
- 14-16 months: showing of objects

Social & play milestones:

- 4 months: shows interest in watching people's faces, smiles back, initiates smile
- 6 months: relates to parents with real joy, smiles often while playing with parent
- 9 months: back and forth smiles/sounds/gestures…giving and taking activities
- ~4-12 months: sensorimotor play(mouth, manipulate, bang, toss)
- 12 months:
    - orients to name
    - uses gestures to get needs met
    - plays peek-a-boo/patty cake
    - repeats actions clapped for
- 12+ months: functional or relational play
    - use of simple objects according to intended function, e.g., brush for the hair
    - seeing relations and combining toys that are functionally related, e.g., bowl and spoon, truck and driver
    - making objects do what they were made to do, e.g., turn handle on jack-in-box

- 12-15 months: early pretend play- child pretends to engage in real-life activities centered around child's own body and actions (e.g., pretend eating, drinking)
- 12-18 months: responds to facial expressions of emotion in other familiar people
- 15 months:
    - checks parents' facial expression after unexpected stimulus and then reacts accordingly
    - uses sounds, pointing, or other "showing" gestures to draw attention to objects of interest
    - begins to show empathy (becomes concerned when others cry)
- ~16 months: simple pretend play- child is able to focus pretend play on other people- child first directs actions towards persons, e.g., feeding mother
- 18 months:
    - simple pretend play directed towards inanimate objects (feeding baby doll with play bottle)
    - attracts parents attention by looking up at them or gesturing during play
- 24 months: more complex pretend and social play
    - 18- 24 months: use object to represent something else (wooden peg = bottle to feed doll)
    - engaging in 2-step pretend play (feed doll and then put doll to sleep) without prompt from adult
    - enjoys being next to other children
    - shows interest in playing, offers child a toy
- 36 months:
    - talking for doll or action figure
    - extensive use of imaginary objects
    - plays with children
    - shows/tells another child about a favorite toy
    - talks about feelings (hungry, sleepy, sad) past and future
- 3- 5 years:
    - child imagines self as different imaginary characters in play

Table 1 General developmental tasks that should be assessed in each dimension of social/emotional development for each developmental period

	Developmental milestones in social-emotional domain dimensions				
Developmental period	**Social competence**	**Attachment**	**Emotional competence**	**Self-perceived competence**	**Temperament/personality**
Infancy (birth to 18 or 24 months)	Interest in people; shows desire for personal attention Capable of coordinated interaction Initiates contact with age mates	Formation of attachment bond with adults Inception of "internal working model" of attachment (ie, security or insecurity of attachment emerges)	Expression of basic emotions Differential reaction to adult emotions Emotion regulation; some self-soothing, much assistance by adults	Responds to own name; recognises self Expresses ownership or possession (Note: these milestones are really more closely allied with self-concept than perceived competence)	Shows distinct dimensions of self-regulation and reactivity
Toddler period (18–24 months through 3 years)	Plays alongside age mates Participates in group play	"Goal-corrected partnership" in attachment (ie, the beginning of autonomy as well as connectedness)	Expression of more social emotions (eg, guilt, shame, empathy) Begins to comprehend "good" and "bad" feelings More independent emotion regulation	Speaks positively of self Desires autonomy Begins to have some idea of distinct domains of self-competence	Moderate continuity seen in dimensions of temperament, but some change seen Regulatory dimensions become more important due to anterior cortical brain development
Preschool period through kindergarten (3 to 5–6 years)	Beginning peer interaction while managing emotional arousal Beginning of specific friendships and peer status Prosocial behaviours and interactions emerge	Enjoys familiar adults Separates easily from parents	Expression of "blended" emotions Understands expressions and situations of basic emotions More independent emotion regulation	Shows awareness of differentiated physical, social and cognitive abilities Speaks positively of self Asserts self in socially acceptable ways	Temperament beginning to be differentiated into personality
Grade school	Formation of dyadic friendships Solidification of peer status General diminution of physical aggression	Begins to balance connection to parents and peers	Use of display rules Understands complex emotions (eg, ambivalence, unique perspectives) Begins independently to use cognitive strategies to regulate emotions	Greater differentiation of self-perceptions of physical, social and cognitive abilities Social comparison becomes even more important	Personality traits becoming more differentiated
Early adolescence (12–14 years)	Achieving new and more mature relations with others, both boys and girls, in their age group	Continues balancing connections with parents and peers (in some ways peers now "come out on top", but parents are still important)	More subtle experience and expression emotion Ever more sophisticated understanding of unique emotional perspectives Broader array of emotion regulatory strategies	Begins a period of heightened self-awareness Also begins a period of heightened self-consciousness	Personality traits becoming more differentiated Continuity from earlier years Temperament dimensions of reactivity and regulation remain important
Middle adolescence (15–17 years)	Achieving new and more mature relations with others, both boys and girls, in their age group Achieving emotional independence from parents and other adults	Moves into even more intimate relationships with friends of the same and opposite sex	Same as above	Achieving a masculine or feminine social role Accepting one's physique	Continuity from earlier years Temperament dimensions of reactivity and regulation remain important
Late adolescence/early adulthood	Achieving emotional independence from parents and other adults Desiring and achieving socially responsible behaviour Preparing for marriage, family life, and career	Same as above	Same as above	Acquiring a set of values and an ethical system as a guide to behaviour – developing an ideology and other forms of identify	Continuity from earlier years Temperament dimensions of reactivity and regulation remain important

b. Know the developmental milestones of normal social-emotional development in the toddler age group (12-36 months of age)

See above chart

c. Know the developmental milestones of normal social-emotional development in the preschool age group (36-60 months of age)

See above chart

d. Know the developmental milestones of normal social-emotional development in the school age group (5-12 years of age)- See above chart

1) At 5 and 6 years of age, the child can follow simple rules and directions.

a. He learns adult social skills like giving praise and apologizing for unintentional mistakes.
b. He likes to spend more time in peer groups and relates to a group of friends.
c. Imaginative play gets more complex and cooperative, and he likes to play dress up and act out his fantasies.
d. Begins to understand imagination is not real

2) At 7 and 8 years of age, the child fully understands rules and regulations.
   a. He shows a deeper understanding of relationships and responsibilities and can take charge of simple chores.
   b. Moral development furthers, and he learns more complex coping skills. At this age, a child explores new ideas and activities and peers may test his beliefs. Children identify more with other children of similar gender and finding a best friend is common.
3) At 9 and 10 years of age, peer and friend groups take precedence over family.
   a. Children at this age will show increasing independent decision-making and a growing need for independence from family.
   b. Parents can use responsibilities and chores to earn time with friends. A positive nurturing relationship with a caregiver with praise and affection and setting up a reasonable balance between independence and house rules builds self-confidence and self-assurance.
   c. Promoting supportive adult relationships and increasing opportunities to take part in positive community activities increases resilience.

e. Know the developmental milestones of normal social-emotional development in adolescents (13-21 years of age)- See above chart

1) Greater independence and commitment to peer groups drives the transition to adolescence.
   a. This will include indulging in risky behavior to explore uncertain emotions and impress peer groups.
2) Social interactions include complex relationships, disagreements, breakups, new friendships, and long-lasting relations.
3) Normally the adolescent will learn to cope with these stresses with healthy adult relationship and guidance to make independent decisions.
4) As young adulthood approaches, school success and work-related activities become important.
5) For a healthy transition to adulthood positive and supportive adult guidance and opportunities to take part constructively in the community play a pivotal role.

f. Understand the typical progression of normal social-emotional development

1) Social-emotional development covers 2 important concepts of development including: development of self or temperament and relationship to others or attachment.
   a. Temperament is an innate attribute that defines the child's approach to the world and his interaction with environment across 9 dimensions which are activity level, distractibility, the intensity of emotions, regularity, sensory threshold, the tendency to approach versus withdraw, adaptability, persistence, and mood quality
      i. Can be defined as the child's "style" or "personality," and it is intrinsic to a child.
      ii. As early as birth, all children demonstrate individual characteristics and patterns of behavior that constitute that individual child's temperament.
      iii. Temperament influences how an infant responds to routine activities, such as feeding, dressing, playing, and going to sleep.

iv. There seems to be a biologic basis to these characteristics, although how a child learns to regulate her emotional state also depends on the interactions between child and caregiver.

v. It influences a child behavior and interaction with others. Based on the above attributes that define temperament, researchers have categorized young children's temperament into 3 broad temperamental categories:

1. *Easy or flexible:* This category includes children who are friendly and easygoing, comply with routines such as sleep and meal times, adapt to changes, and have a calm disposition.
2. *Active or feisty:* Children who are fussy, do not follow routines and have irregular feeding and sleeping schedules, are apprehensive of a new environment and new people, have intense reactions, and get easily upset.
3. *Slow to warm up or cautious:* Children who may be less engaged or active, have a shy disposition to a new situation and new people, may withdraw or have a negative reaction. They become more comfortable and warm up with repeated exposure to a new environment or person.

vi. This classification is for the ease of discussion, and all temperaments will not fit into one or other categories exactly.

vii. Discussion about temperament with parents and caregivers can better identify the child's strengths and needs. Based on this, caregivers can adapt their management and caregiving styles to match the child's temperament.

viii. This can mold a child's behavior and facilitate the child's successful interaction with the environment, defined as "goodness of fit."

b. Emotional development involves three specific elements: neural processes to relay information about the environment to the brain, mental processes that generate feelings, and motor actions that include facial expressions, speech, and purposeful movements.

i. The limbic system is responsible primarily for receiving, processing, and interpreting environmental stimuli that produce emotional responses.

ii. During development, the repertoire of specific emotions remains constant, but the stimuli that produce them become more abstract

iii. All infants demonstrate universal facial expressions that reveal these emotions, although they do not use these expressions discriminately before the age of 3 months.

c. Eventually, however, cognitive skills play a role as emotional expressions become connected to specific occurrences.

i. For example, an 8-month-old infant can let his parents know that he is upset about being left alone in his crib or happy about playing with a toy.

ii. Because he now has object permanence, he demonstrates fear in new situations due to the ability to shift attention and recognize "familiar" from "unfamiliar."

d. Emotional development continues as the toddler learns to identify different emotions in other people

i. At 15 months, a child demonstrates empathy by looking sad when she sees someone else cry.

ii. She also develops self-conscious emotions (embarrassment, shame, pride) as she evaluates her own behavior in the context of the social environment.

iii. Having once performed cute tricks on demand, she suddenly seems embarrassed and refuses to perform when she realizes that others are watching.

iv. She may hide behind a chair to have a bowel movement and become upset if someone catches her in the act.

e. As language skills develop, the child can label different emotional states in others and even associate language with emotions and memory.

i. For example, if he had a tantrum when he didn't get a toy from the store, he may have an identical emotional outburst when he hears a verbal reminder of the situation.
ii. By age 2 years, he starts to mask emotions for social etiquette

f. During the preschool years, children learn more and more behavioral strategies to manage their emotions, depending on a given situation.
    i. They begin to understand that their expressed emotion—whether a facial, vocal, or behavioral expression—does not necessarily need to match their subjective emotional experience.
    ii. They demonstrate an increased understanding and use of "display rules." These are "culturally defined rules that guide a person's decision to alter emotional behavior consistent with the demands of the social context"

g. Children learn to substitute their expressions (smile and say "thank you" even though they are disappointed in the birthday present), amplify expressions (exaggerate a painful response to get sympathy), neutralize expression (put on a "poker face" to hide true feelings), or minimize emotion (look mildly upset when feeling extremely angry). By the time they enter kindergarten, children have started to master many of the emotional nuances of social interactions.

2) In terms of attachment, most children are born with an inherent drive to connect with others and share feelings, thoughts, and actions.

a. The earliest social milestone is the bonding of a caregiver with the infant, characterized by the caregiver's feelings for the child.

b. Even before acquiring language, babies learn to communicate through emotions.
    i. There is a rapid growth in social and emotional areas of the brain during the first 18 months of life. The nonverbal parts of the right brain, including the amygdala and the limbic system, receives, processes and interprets stimulus from the environment that produce an emotional response and build emotional and stress regulatory systems of the body.
    ii. The lower limbic system, outside the cortex, dictates most of our spontaneous, instinctive emotional responses, like fear resulting in a racing heart or weak knees.
    iii. The upper limbic system part of the cerebral cortex, known as the limbic cortex, controls conscious awareness of emotions and refines the responses according to the environmental culture of the individual.
    iv. The amygdala which lies at the junction of the cortex and subcortical areas of the brain and plays a pivotal role in sensing emotions and connects them both to higher and lower limbic structures.
    v. During the second half of infancy, emotional information from the lower limbic system moves up and becomes part of the babies conscious.
    vi. Frontal lobe activity increases and myelination of the limbic pathways also begins during this time.
    vii. With this gain in the limbic system, a caregivers soothing and consistent response to the child's emotions develops into the child's attachment to the caregiver, usually the mother.
    viii. Attachment is regarded as a pivotal event in a person's emotional development. It lays the foundation of a child's security, harbors self-esteem, and builds emotional regulation and self-control skills.

c. The consistent availability of the caregiver and the bonding that comes with this results in the development of "basic trust" and confidence in the infant for the caregiver during the

first year of life. Basic trust is the first psychosocial stage described by Erickson. This allows the infant to seek for parents or the caregiver during times of stress, known as the attachment

i. The infant learns to discriminate his mother's voice during the first month after birth.

ii. He cries to express distress from hunger, fatigue, or a wet diaper.

iii. Attachment theory suggests that as the caregiver responds to these cries and other behaviors, the infant gains confidence in the caregiver's accessibility and responsiveness.

iv. This behavior system promotes the parent–child relationship that some researchers believe facilitates parental protection, and thus infant survival.

v. From this relationship comes the first measureable social milestone: the smile.

d. The infant smiles at first in response to high pitched vocalizations ("baby talk") and a smile from his caregiver; but over time, less and less stimulation is required. Ultimately, just seeing the caregiver elicits a smile.

e. The infant learns that he can manipulate the environment to satisfy personal needs by flashing a toothless grin or, alternatively, by crying.

f. His interactions then begin to involve to-and-fro vocalizations by 4 months.

g. Visual skills develop as well, and he can recognize his caregivers by sight at 5 months.

h. Stranger anxiety, or the ability to distinguish between familiar and unfamiliar people, emerges by 6 months. Whereas the 4-month-old infant smiles at any adult, the slightly older infant cries and looks nervously between his caregiver and other adults.

i. Joint attention is the quintessential social milestone that develops towards the end of the first year after birth.

i. Joint attention is the process whereby an infant and caregiver share an experience and recognize that the experience is being shared.

ii. The earliest demonstration of joint attention occurs around 8 months of age, when an infant follows a caregiver's gaze and looks in the same direction.

iii. In a few months, the infant looks back at the caregiver as an indication of a shared interaction.

iv. The infant consistently turns her head to the speaker when her name is called by 10 months, further demonstrating a connectedness with her environment.

j. Play skills also follow a specific developmental course

i. Initially, an infant holds blocks and bangs them against each other or on the table, drops them, and eventually throws them.

ii. Object permanence allows her to realize that the blocks are still present, even if she cannot see them. She learns that dropping the blocks from her highchair will cause her caregiver to pick them up and return them to her; ~~so~~so, she repeats this "game" over and over.

iii. As fine motor and cognitive skills develop, she starts to use objects for more specific purposes, such as using those blocks to build a tower.

iv. By 18 months, she engages in simple pretend play, such as using miniature representative items in a correct fashion→For example, she pretends to talk on a toy phone or "feeds" a doll by using a toy spoon or bottle.

k. After his second birthday, the child begins to play with others his own age.

i. A rule of thumb is that a child can play effectively only in groups of children in the same number as his age in years.

ii. Thus, a 2-year-old can play well only with one other child.

l. Two-year-old play often is described as “parallel” because a child of this age often plays next *to* another child but not with him.
m. However, the 2-year-old frequently looks at his playmate and imitates his actions. He has not yet mastered the skill of cooperation; so, aggression often is the tool of choice to obtain a desired object.
n. By 30 months, the child uses complex pretend play, such as using generic items to represent other objects.
    i. A block may be used as a telephone in one scenario or used as a bottle to feed a doll in another.
    ii. The scenarios themselves also increase in complexity, from merely feeding the doll to dressing the doll and putting her to “sleep.”
o. By age 3 years, a child has mastered her aggression to some extent, and she is able to initiate a cooperative play experience with one or two peers. Most of the time, they are able to have joint goals and take turns. She also moves into simple fantasy or imaginative play. She may pretend to be a dog or an airplane. However, she cannot yet distinguish between what is real and what is make-believe; so fear of imaginary things is common at this time.
p. Four-year-olds usually have mastered the difference between real and imaginary.
    i. They become interested in tricking others and concerned about being tricked themselves.
    ii. They are able to play effectively with up to three other children, although some may have a preferred friend.
    iii. Imaginary scenarios increase in complexity: a cardboard box may become a sailboat, and toilet paper rolls may become binoculars.
q. By age 5, children have learned many adult social skills, such as giving a positive comment in response to another's good fortune, apologizing for unintentional mistakes, and relating to a group of friends.
    i. Their imaginative play is increasingly more complex, and they love to dress up and act out their fantasies.

3) Close attention must be paid to the red flags in the domain of social-emotional development as in any other realm of development; regression always a concern as well as a lack of age-expected/appropriate joint attention

Table.

Social-Emotional Red Flags

Age	Red Flag
6 mo	Lack of smiles or other joyful expressions
9 mo	Lack of reciprocal (back-and-forth sharing of) vocalizations, smiles, or other facial expressions
12 mo	Failure to respond to name when called
	Absence of babbling
	Lack of reciprocal gestures (showing, reaching, waving)
15 mo	Lack of proto-declarative pointing or other showing gestures
	Lack of single words
18 mo	Lack of simple pretend play
	Lack of spoken language/gesture combinations
24 mo	Lack of two-word meaningful phrases (without imitating or repeating)
Any age	Loss of previously acquired babbling, speech, or social skills

4. Cognitive

a. Know the developmental milestones of normal cognitive development in the first year after birth.- see chart below

b. Know the developmental milestones of normal cognitive development in the toddler age group (12-36 months of age)- see chart below

c. Know the developmental milestones of normal cognitive development in the preschool age group (36-60 months of age)- see chart below

d. Know the developmental milestones of normal cognitive development in the school age group (5-12 years of age)- see chart below in addition

1) Probably best summarized by consideration of the concrete operational stage – the third stage of Piaget's theory of cognitive development.

2) Occurs between the ages of 7-11 and is characterized by the appropriate use of logic.

3) During this stage, a child's thought processes become more mature and "adult like"–start solving problems in a more logical fashion.

4) Abstract, hypothetical thinking is not yet developed in the child, and children can only solve problems that apply to concrete events or objects.

5) At this stage, the children undergo a transition where the child learns rules such as conservation- e.g. water level task, see figure # below

6) Also are able to incorporate inductive reasoning→inductive reasoning involves drawing inferences from observations in order to make a generalization

7) In contrast, children struggle with deductive reasoning at this age which involves using a generalized principle in order to try to predict the outcome

of an event—common to experience difficulties with figuring out logic in their heads.

8) For example, a child will understand that "A is more than B" and "B is more than C"→However, when asked "is A more than C?" …the child might not be able to logically figure the question out mentally.

9) Two other important processes in the concrete operational stage are logic and the elimination of egocentrism.

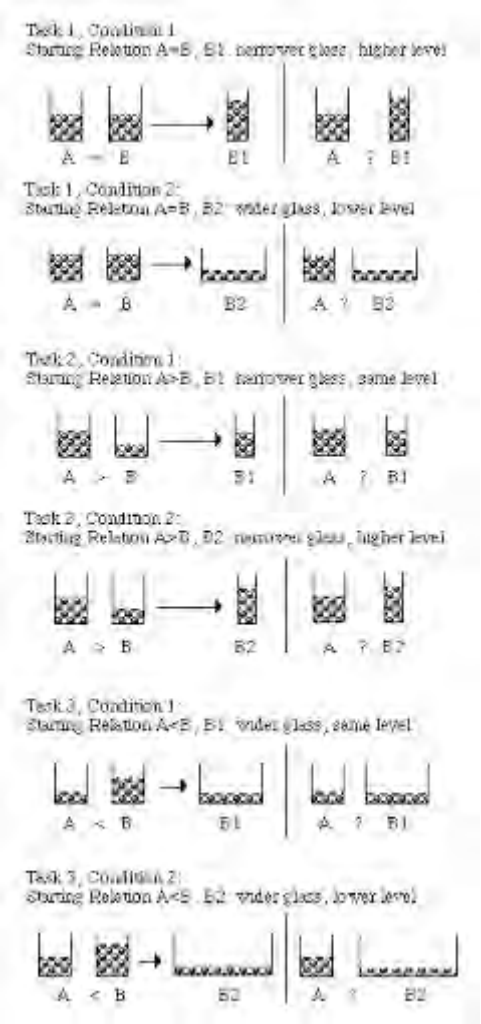


Figure showing Piaget's water level task illustrating principle of conservation.

e. Know the developmental milestones of normal cognitive development in the adolescent age group (13-21 years of age)

1) Best illustrated with consideration of the formal operational stage- the final stage of Piaget's theory of cognitive development (from roughly age 12 to adulthood)

2) Piaget stated that hypothetico-deductive reasoning becomes important during this stage →concept of counterfactual thinking (hypothetical "what if" situations not always rooted in reality)

3) Intelligence is demonstrated through the logical use of symbols related to abstract concepts and using logic to draw specific conclusions from abstract concepts

4) During this time, people develop the ability to think about abstract concepts and begin to consider possible outcomes and consequences of their actions

5) It can be possible for an individual to use formal operations in some aspects, but not all, of their lives

1. Example- ADHD and other medical conditions associated with executive functioning deficits

f. Understand the typical progression of normal cognitive development.

i. Cognitive development is the foundation of intelligence.

a) Intelligence is defined as the ability to learn or understand or to deal with new situations→but it's a much broader concept that involves the interaction of multiple different factors

b) The best efforts to quantify this concept come through use of standardized intelligence tests that attempt to measure multiple areas, such as problem-solving, language, attention, memory, and information processing.

c) Scores in these domains are used to determine specific subset strengths and weaknesses in addition to a composite score.

d) The subset scores and composite score do not represent the individual in isolation accurately and require careful analysis.

e) Accurate interpretation of intelligence testing requires more than a casual understanding of normal ranges.

ii. Standardized intelligence testing is not available for infants.

a. Therefore, the assessment of infant and child intelligence depends on progression through two developmental domains: problem-solving and language
b. Children advance through these domains by learning and exposures to novel experiences that dictate this learning
c. Learning requires the ability to direct and to sustain attention as well as the ability to manipulate information.

iii. The foundational aspects of cognitive development include memory, representational competence, attention, and processing speed.

a. Successful cognitive development requires progress in all these domains.
b. Memory involves the proper encoding, storing, and retrieving of information.
c. Representational competence is the ability to create and manipulate a mental image of an object or idea that is not seen.
d. Appropriate advancement in the control of attention–learning how to focus and shift focus–also is important.
e. Processing speed may be the central limiting factor of intelligence because it links the other functions together.

iv. Many theorists have tried to create a framework to understand better the cognitive development of infants and children.

a. No categorization of developmental stages has been able to describe accurately what parents have been observing for centuries.

b. The current overarching construct is that infants are not merely passive learners; they are very active in the observation and modification of their environment.
c. Learning is a product of this observation and occurs when there is disequilibrium of assimilation (taking in information) and accommodation (revising existing mental structures).
d. The cognitive development of an infant is more subtle than other areas of development and ~~b~~predictable than the motor development described in the first article of this series. The progression, however, is more subtle, and child behavior often is mistaken as abnormal rather than evidence of advancement to a new developmental stage.

v. Object permanence is one of the subtle developments and is not an all-or-nothing phenomenon.
   a. It may be most noticeable around 9 to 10 months of age, when the infant understands that his mother still exists even when she is not visible.
   b. The object permanence is not yet mature enough, however, to allow the infant to make a judgment about where the mother might be when she is not visible, so he cries and demonstrates separation anxiety.
   c. By 15 to 18 months, the toddler's understanding has matured, and he will be able to make predictions regarding the mother's location.
   d. Rather than crying when separated, the child will use advancing motor skills to seek her out.
   e. Object permanence allows him to form concepts and ideas about things, even if the items themselves cannot be seen, and then to create schemes for relating to objects encountered in the future.
   f. For example, successfully interacting with a cup involves use of different skills than throwing a ball.
   g. The toddler's early organizational ability also allows him to separate objects into language-specific categories.

vi. Causality refers to the infant's gradual understanding of her role in changing or acting on her environment.
   a. The infant discovers that she can use objects to her benefit; she even can use her voice to make objects (e.g., her parents) respond in ways that directly meet her needs or wants (e.g., being picked up).
   b. Her actions become increasingly more intentional.
   c. Between 4 and 8 months of age, she purposefully begins to repeat effects that were discovered accidently.
   d. For example, a random kick causes toys hanging over her activity mat to move, so she kicks again.
   e. Eventually, she will try different interactions to see what effects she can create.

vii. Symbolic thinking allows for expansion of play by using one object to represent another object.
   a. The toddler may use blocks as cars and often reenacts past events.
   b. The toddler may “cook” his favorite food in a toy kitchen and use paper to represent his “food.”
   c. Imitation of the actions of others is an important part of play, and his food preparation reflects what he sees his caregivers do for him.

viii. Object permanence, causality, and symbolic thinking are fundamental concepts in the current understanding of cognitive development.
    a. Such foundational skills combine with a toddler's gross and fine motor advancement to permit the ability to problem-solve and develop language.

ix. Problem-solving involves the manipulation of objects to achieve a specific goal.
    a. An infant's first exploration of her environment is performed visually
    b. At first, the infant is able to follow a face and then objects.
    c. Tracking starts with horizontal and vertical movements but soon advances to tracking circular motion.
    d. Three-dimensional awareness and response is seen at 3 months, when an infant reacts to a visual threat.
    e. Initially, the infant only regards and stares intently, but as she gains improved control of her arms and hands, she begins batting at and reaching for objects.
    f. She begins to inspect items placed in her hand visually and by mouthing as a means of environmental exploration.
    g. As hand control improves, she is able to hold an object in one hand while manipulating it with the other.
    h. At around 5 months of age, her vision has matured to allow her to focus on smaller objects.
    i. Her attempts to pick them up help develop a pincer grasp and eventually lead to isolation of her index finger, which allows her to explore objects by poking.

x. With maturation of object permanence and causality, the toddler begins to play more purposefully
    a. Will be positively reinforced by reactions from caregivers when she plays with items and shows pleasure in the play
    b. Looks for dropped objects
    c. Her attention span continues to increase, and she is able to work on obtaining objects that initially are partially hidden and then hidden completely.
    d. She works on removing lids and formulating different strategies for obtaining objects out of her reach.
    e. She soon begins to learn through her manipulation instead of merely learning how to manipulate.

Milestone	Mean Age
Fixes on ring	1 mo
Follows face	1 mo
Follows ring	2 mo
Recognizes mother	2 mo
Reaches for face	3 mo
Follows ring in circle (supine)	3 mo
Regards cube	3 mo
Mouths objects	4 mo
Aware of strange situation	4 mo
Shakes rattle	4 mo
Reaches for ring/rattle	4 mo
Attains dangling ring	5 mo
Turns head-looks for dropped spoon	5 mo
Regards pellet	5 mo
Touches reflection & vocalizes	6 mo
Removes cloth on face	6 mo
Bangs & shakes keys	6 mo
Inspects ring	7 mo
Observes cube in each hand	7 mo
Finds partially hidden object	7 mo
Seeks object after it falls silently to floor	8 mo
Inspects bell	9 mo
Rings bell	9 mo
Pulls string to obtain ring	9 mo
Uncovers toy under cloth	10 mo
Pokes at pellet in bottle	10 mo
Tries to put cube in cup, but may or may not let go	10 mo
Finds toy under cup	11 mo
Looks at pictures in book	11 mo
Rattles spoon in cup	12 mo
Lifts box lid to find toy	12 mo
Dangles ring by string	13 mo
Solves glass frustration test	13 mo
Unwraps toy in cloth	13 mo
Dumps pellet out of bottle after demo	14 mo
Turns pages in book	15 mo
Places circle in single shape puzzle	15 mo
Dumps pellet out w/o demo	16 mo
Places circle in form board	16 mo
Finds toy under layered covers	16 mo
Matches pairs of objects	18 mo
Circle reversed after searching	18 mo
Deduces location of hidden object	20 mo
Places circle, square in formboard	20 mo
Adapts to formboard reversal w/4 trials	22 mo
Completes formboard	22 mo
Sorts objects	24 mo
Matches objects to pictures	24 mo
Shows use of familiar objects	24 mo
Matches shapes	28 mo
Matches colors	28 mo
Reverses formboard spontaneously	30 mo
Points to small details in pictures	30 mo
Points to self in photos	33 mo
Points to body parts and to function (“what do you hear with/”)	33 mo
Adds 2 parts to DAP	3 y
Understands long/short, big/small, more/less	3 y
Knows own age, gender	3 y
DAP = 4-5 parts	4 y
Number concepts to 2	4 y
Simple analogies: dad/boy, mother/?; ice/cold, fire/?; ceiling up/floor/?	4 y
Points to 4 colors	4 y
DAP = 8-10 parts	5 y
Number concepts to 3	5 y
Identifies coins	5 y
Standard IQ test needed	5 y
DAP = 12 – 14 parts	6 y
Number concepts to 10	6 y
Simple addition	6 y
Understands seasons	6 y

Content Category 1B- Foundations of Developmental Behavioral Pediatrics- Theoretical Frameworks

ABP Content Specs for Developmental-Behavioral Pediatrics Fellow Collaboration Project

Prepared by Rebecca Christi, MD, Madigan Army Medical Center DBP Fellow

Reviewed by Abigail Angulo, MD, Colorado Children's DBP Fellowship Director

B. Theoretical frameworks

1. Criteria for evaluating theories

a. Recognize the characteristics of a good developmental theory

2. Maturational theory (Gesell)

a. Know that maturational theory (Gesell) proposes that development proceeds in a systematic direction (principle of developmental direction)

b. Understand the major components of Gesell's maturational theory

c. Know the limitations of Gesell's maturational theory in explaining child development

d. Understand the relationship between the maturational theory and developmental testing

e. Describe developmental tests based on Gesell's theory

3. Cognitive (Piaget)

a. Differentiate between assimilation and accommodation as cognitive processes described by Piaget to facilitate the learning of new information

b. Understand how disciplinary strategies are best adapted to the conceptual development of the child (pre-operational, concrete operational, formal operational)

c. Know the implications of a belief in immanent justice and magical thought for young children's understanding of illness and loss

d. Understand the characteristics of the four stages of conceptual development as described by Piaget

e. Formulate a developmentally appropriate explanation of illness or loss for a child in any one of the conceptual stages described by Piaget

f. Know how to design treatment plans for common medical conditions in ways that would facilitate the adjustment of children at various conceptual stages described by Piaget

g. Understand the relevance of object permanence to the development of stranger anxiety and sleep problems in infancy

h. Understand the limitations of Piaget's theory of cognitive development

I. Describe developmental tests based on Piaget's theory

4.Psychodynamic (Freud)

a. Understand the three primary motivating forces of psychosexual development

b. Distinguish between the concepts of conscious/unconscious/ subconscious awareness

c. Recognize the role of denial in response to a death, chronic illness, disability, or other significant stress

d. Differentiate between signs of regression and of projection

e. Understand the impact of transference and counter-transference on the relationship of physician and patient

f. Know the characteristics of the stages of psychosexual development as described by Freud

g. Understand the limitations of Freudian theory of psychosexual development

h. Understand that parental anger toward healthcare providers may be a result of projection

5. Social-emotional (Erikson)

a. Know the parallels between Erickson's and Freud's stages of development from infancy to adolescence

b. Know the components of the stages of Erickson's paradigm

c. Appreciate the impact of unsuccessful resolution of one of Erickson's stages on children's ability to meet the challenges of subsequent stages

d. Know the implications of succeeding at the various stages described by Erickson for success in relationships and school

6. Attachment (Mahler, Bowlby, Ainsworth)

a. Appreciate the reciprocal contributions of infants and caregivers on the development of attachment

b. Understand how attachment theory and cognitive developmental theory together explain the phenomena of separation protest and stranger anxiety

c. Differentiate among securely attached, insecurely attached, and ambivalently attached infants using Ainsworth's "strange situation" paradigm

d. Recognize the behavioral hallmarks of securely attached infants in the first year of life

e. Know the components and parenting styles of child-rearing that promote the development of secure attachment
f. Recognize signs of insecure attachment seen in infants
g. Know how to advise parents based on Mahler's construct of separation-individuation to help promote psychological separation-individuation
h. Understand the limitations of attachment theory

7. Behavioral theory
a. Distinguish between classical and operant conditioning paradigms
b. Understand how modeling and shaping can be used to facilitate the expression of a novel and complex behavior
c. Understand how various schedules of reinforcement facilitate learning of a new behavior and maintenance of an established behavior
d. be able to counsel a family on the likelihood of post-extinction burst
e. Understand the concept of positive reinforcement in behavior theory
f. Understand that reinforcement during a post-extinction burst will render the behavior more resistant to extinction
g. Understand the problems associated with punishment-based behavior management plans
h. Know the limitations of behavioral theory
I. Recognize the role of modifying antecedents in changing behavior
j. Understand the importance of obtaining a functional behavioral assessment to determine an effective behavior management plan
k. Understand the importance of a child's developmental level on implementing behavioral strategies

8. Social learning theory
a. Know that social learning theories posit that the main determinants of individual behavior are observations and interactions with others
b. Understand that according to social learning theories, social hierarchies, family bonds, sex, age, motivation, and perception all influence the rate of an individual's behavior change
c. Differentiate methods of behavior change based on operant conditioning versus social learning theory
d. Understand how social learning theory is used during parent training programs
e. Understand the concepts taught in parent training programs
f. Understand the differences between cognitive behavioral and social influence theories
g. Understand the distinction between behavioral norms and behavioral values
h. Know how social influence theory predicts behavioral norms
I. Know the limitations of social learning theory for behavior change
j. Distinguish between methods of toilet training based on social learning versus behavioral therapy
k. Know how behavioral values are related to the adoption and maintenance of high-risk behaviors

9. Moral reasoning (Kohlberg)
a. Know that moral reasoning in Kohlberg's theory progresses from explanations based on what feels good to an individual to explanations based on abstract principles of fairness and justice
b. Understand the poor association between the level of moral reasoning and their individual's moral behavior
c. Understand the stages of moral reasoning as defined by Kohlberg
d. Know how to interpret a child's explanations for moral decisions based on Kohlberg's theory

10. Temperament/Individual variation (Chess, Thomas, Birch, Carey)
a. Understand the concept of "goodness of fit" and implications for interpreting parents' perceptions of their children's temperament
b. Identify the major categories used for describing temperament
c. Know the temperament characteristics of the "easy child," the "slow to warm child," and the "difficult child" syndrome
d. Plan appropriate management for a family concerned by one of their child's temperament characteristics
e. Describe long-term behavioral patterns associated with the three categories of temperamental clusters (easy, slow to warm, difficult child)

f. Know the limitations of temperament as an explanatory concept

11. Theories of behavior change
a. Know the key health beliefs that, according to the Health Belief Model, produce a readiness to act
b. Understand how perceived barriers or costs and perceived benefits of behavior change can influence the efficacy of risk reduction efforts
c. Understand the stages of change described by Prochaska in the Trans theoretical Model
d. Know how assessment of an individual's stage of change helps in the selection of an appropriate intervention to facilitate behavior change (e.g., weight reduction, safe sex practices, stopping cigarette smoking)
e. Know the limitations of the Trans theoretical Model of behavior change
f. Understand the concept of motivational interviewing and its relationship to theories of behavioral change

1. Criteria for evaluating theories

a. Recognize the characteristics of a good developmental theory

- Theory=group of logically related statements (e.g. rules/ideas), used to explain past events and predict future occurrences (**explanatory and predictive)**
- Value rests upon ability to be
    - stimulating: amount of research generated
    - inclusive: number and type of phenomena encompassed
    - parsimonious: simplicity, explanation with fewest assumptions is best
    - operational: testable; reliable methods of measuring the concepts of the theory
    - precise: does it help to understand observations of behavior or in generating predictions
    - empirically valid: *crucial;* ability to explain and predict available findings

2. Maturational theory (Gesell)

a. Know that maturational theory (Gesell) proposes that development proceeds in a systematic direction (principle of developmental direction) -->self-explanatory

b. Understand the major components of Gesell's maturational theory

- *Five* basic principles largely influenced by Darwin's theory of evolution
    - Principle of Developmental Direction: proceeds in systematic direction as a function of preprogrammed genetic mechanisms; 2 patterns
        - Cephalocaudal (head down)=gross motor development
        - Proximodistal (near to far)=fine motor development
    - Principle of Reciprocal Interweaving: modeled after physiologic principle of reciprocal innervations; inhibition and excitation of different muscles operate in complementary fashion-->efficient movement (e.g. walking)
    - Principle of Functional Asymmetry: behaviors often go through period of asymmetric development in process of achieving maturity (e.g. ATNR is precursor of symmetrical reaching)
    - Principle of Individualizing Maturation: development is a process of sequential patterning; prerequisite structures must be present for other development/learning to occur
    - Principle of Self-Regulatory Fluctuation: development is alternating periods of stability and instability; distinct sequence of stages allowing function and growth

c. Know the limitations of Gesell's maturational theory in explaining child development

- Proposed that most important determinant in development is biologic maturation
- Rate at which one progresses is determined by heredity and altered only somewhat by experience
- Thereby minimizes effects of environmental/social factors-->in fact believed that environmental influences do not change the basic pattern of development, and ignored the effect of learning
- Inadequately explains cognitive, language, and socio-emotional development or the development of personality/personal identity

d. Understand the relationship between the maturational theory and developmental testing

- Based on systematic progression, "milestones" could be determined
- Some developmental testing is based on the proportion of children who attain these milestones at a particular age, allowing for calculation of a Developmental Quotient (e.g. *Bayley Scales of Infant Development, Gesell Developmental Schedules)*

e. Describe developmental tests based on Gesell's theory (below is NOT all-inclusive)

- *Bayley Scales of Infant Development*
    - Currently Third Edition (BSID-III): provides a comprehensive assessment with 5 different scales (Cognitive, Language, Motor, Social-Emotional and Adaptive Behavior) for children ages 1-42 months
- *Gesell Developmental Schedules*
    - Historically: Gesell Developmental Schedules (GDS), the Gesell Maturity Scale, Yale Tests of Child Development, Gesell Preschool Test, Gesell Kindergarten Screener, and the Gesell Developmental Observation-->meant to measure the development of infants and young children ages 4 weeks to 36 months
    - Included measurement of GM and FM skills, and assessed adaptive, language, and personal-social abilities
    - Current assessment from the Gesell Institute of Child Development is known as the Gesell Developmental Observation-Revised for ages 2 ½ to 9 years

3. Cognitive (Piaget)

a. Differentiate between assimilation and accommodation as cognitive processes described by Piaget to facilitate the learning of new information

- Children take an active role in adapting to their environment-->adaptation involves two complementary processes
    - Assimilation: process through which individual incorporates new knowledge into existing cognitive frameworks/schemas; schema into which new event/experience is assimilated expands but does not qualitatively change
    - Accommodation: adjustment or modification of existing schemas to incorporate new knowledge or information; process through which changes in intellectual development correspond to changes in reality
    - Equilibrium: reached when a child understands what he or she perceives in the world and can deal with most new information through assimilation; drive to equilibrium is the force that drives the learning process

b. Understand how disciplinary strategies are best adapted to the conceptual development of the child (pre-operational, concrete operational, formal operational)

- To Piaget, morality develops because of life experiences and increasing ability to appreciate others' perspective
    - Premoral Stage (birth-4 years, sensorimotor and part of preoperational): little understanding of rules; behavior regulated from outside the child; little ability to understand right from wrong-->discipline focuses on identifying and addressing the cause of the misbehavior, redirection, modeling appropriate behaviors, and building positive/warm relationships

    - Heteronomous/Moral Reasoning Stage (5-9 years, part of preoperational and concrete operational): rule are rigid and given by adults/teachers/higher being; rules tell you right from wrong; severity of behavior determined by consequences and not intention-->discipline focuses on encouraging children to problem solve with guidance when misbehavior occurs rather than just telling children what to do or immediately punishing; rule-breaking events are opportunities for children to contribute to rule-making and active discussion to see other perspectives and learn to collaboratively problem solve
    - Autonomous Morality/Moral Relativism Stage (10+ years, formal operational): emphasis placed on cooperation and rules are changeable under certain circumstances with mutual consent; intent and consequence considered when judging if act is moral or not-->can clearly separate intent from consequences, identify own morals/values, and collaborate with parents/teachers in problem solving
- Goal of discipline is development of moral autonomy (ability to make decisions for oneself regarding right/wrong behaviors); decisions made regardless of rewards and punishment, take into account all relevant factors while taking responsibility for one's behavior; rules/values are instilled into conscience as child grows and experiences life-->goal of discipline is NOT obedience but to learn to make wise choices
- Role of parent/teacher is to facilitate or guide child in learning while maintaining safety and setting appropriate limits, as well as being good role model; role is NOT to reward/punish, but to teach

c. Know the implications of a belief in immanent justice and magical thought for young children's understanding of illness and loss

- Immanent Justice=immediate punishment will happen when rules are broken; belief that illness or other "bad" events occur as punishment for misbehavior
- Magical thinking/Animism=nonliving objects taking on living characteristics
- Immanent justice and magical thinking are characteristic of children in preoperational stage; additionally they believe that events that occur at the same time have a cause and effect relationship
- Children in this stage may believe that an illness or loss was caused by their recent naughty behavior or that their parents are separating because the children refused to eat vegetables

d. Understand the characteristics of the four stages of conceptual development as described by Piaget

- Piaget's theory acknowledges influences of biology and impact of the environment; maturation controlled by innate mechanisms (biologic factors), but potential outcome of development can be affected by other environmental factors (including direct learning and social transmission)
- Assumes development occurs in series of qualitatively distinct stages that are hierarchically organized and follow the same invariant sequence for all individuals-->forms basis of behavior
- *Four* distinct stages (see Table 1-3 in Wolraich, Third Edition)
    - Sensorimotor (Birth-2 years): infants understand and organize the world through sensory information and motor activity-->*six* substages (Early Reflex Reactions, Primary Circular Reactions, Secondary Circular Reactions, Coordination of Secondary Schemas, Tertiary Circular Reactions, Emergence of Representational Thought); object permanence develops
    - Preoperational (2-7 years): children use symbolic representation for events, places, and people; language and pretend play develop-->main characteristics

        - rigidity of thought-centration (focus on one salient feature and ignore other features even when this leads to illogical conclusions), confusion of appearance and reality (not realize that a change in appearance does not change basic nature or identity)
        - egocentric worldview
        - semi-logical reasoning-transductive thinking (from the specific/particular to the specific/particular; events occurring at the same time are thought to be causally related (e.g. fight with brother caused his illness); animism (attributing human characteristics and actions to inanimate objects
        - limited social cognition-judges wrongness of an act according to *external* variable (amount of damage), less likely to consider *internal* variable (person's intentions)
    - Concrete Operational (7-11 years): children can solve logical problems about concrete physical subjects; <u>conservation and hierarchical thinking</u> develop
        - flexibility of thought-decentration and reversibility (can focus on more than one aspect at a time, can reverse operations mentally)
        - declining egocentrism-social perspective taking, understand that a person can feel one way and act another
        - better understanding of temporal and spatial relations-better able to reason about causality of events
        - able to regulate their interaction with each other through rules-rule-based games; able to take intentions into account when making judgments of good and bad behavior
    - Formal Operational (12 years-adulthood): adolescents can reason logically about abstract topics, hypothetical problems and possible outcomes of a situation
        - ability to consider many different solutions to a problem before acting on any one
        - begin to adopt physiologically-based conception of illness-->increased understanding of varying degrees of illness as well as personal control over onset/severity of illness
        - renewed egocentrism-lack of differentiation between own thoughts and others; self-consciousness, self-criticism, self-administration-->over time, take into account how others judge them, how they judge the judgment process of others, and how all this corresponds to social categories in culture (politic and law as abstract principles; beneficial side of laws rather than just punitive)

e. Formulate a developmentally appropriate explanation of illness or loss for a child in any one of the conceptual stages described by Piaget

- *The following is taken from a table referenced in DBPeds PREP, April 2016*
- Infant (Sensorimotor): No cognitive understanding; reaction to separation, change of routine; sensitive to anxiety of caregiver
    - Keep routines and structure; provide physical affection, reassurance; caretaker should model healthy coping and also ensure they meet their own needs related to grief
    - Allow caretaker to hold/provide physical affection
- Preschool (Preoperational): Death is reversible, temporary, and/or punishment; “Magical thinking,”- may think thoughts, words or actions may have caused death; may ask questions repeatedly; may offer a contagion explanation (attributing cause to objects/people in close proximity to but not touching child; "got sick because I stood close to a sick tree") or

phenomenistic logic (attributing cause to external concrete event/object occurring simultaneously; "got sick because the tree fell down")
    - Explain in simple and clear language with concrete terms; avoid euphemisms (e.g., went to sleep); gently address magical thinking; assure child was not responsible for death; be tolerant of repeated questions; encourage pretend/imaginary play to help cope with traumatic events
    - Provide child with limited choices and control in daily life
- School age (Concrete Operational): May personify death (e.g., ghost, angel); may start to understand death is permanent and universal, but may not realize personal self death; may be interested in process of death and associated rituals ; use concepts of contamination (direct contact with person/action/object that is potentially harmful; "got a cold because went outside without a hat and the cold got on my head") or internalization (taking in or swallowing something harmful that affects internal organs; "got a cold because breathed in too much air with germs in it that went to my lungs")
    - Honest, simple explanations; accurate yet understandable explanations; allow opportunities for them to express themselves
    - Inform child of plans for treatment and take care to prepare the child for any medical procedures
- Adolescent (Formal Operational): Understands death is irreversible and universal; may not acknowledge own risk of death; physiologic (includes outside trigger such as virus that causes internal organs to malfunction and cause illness) and psychophysiological (recognizes role of emotional states in affecting functioning of body, as well as physiological causes ["nervousness made my BP high"]) reasoning
    - Provide support by being available; ensure there are also peers and other adults that can provide them support; discuss feelings, avoid clichés of placing adult responsibilities on the teenager
    - Provide clear physiological and psychophysiological explanation of illness-helps with understanding and treatment compliance; discuss impact of illness on school, social, emotional, and physical functioning

f. Know how to design treatment plans for common medical conditions in ways that would facilitate the adjustment of children at various conceptual stages described by Piaget

- *See section 3e above*

g. Understand the relevance of object permanence to the development of stranger anxiety and sleep problems in infancy

- Related to Sensorimotor Substage 4 (Coordination of Secondary Schemas), a hallmark of which is infant's realization that objects are clearly separate from him/her, and if removed from the visual field, it does not cease to exist
- Stranger Anxiety: child can detect discrepancy between known (care taker) and unknown (stranger)
- Sleep Problems: similar concept as child can keep parent/care taker in mind even when disappeared from view

h. Understand the limitations of Piaget's theory of cognitive development

- Evidence that children can improve performance on cognitive tasks through training
- Theory doesn't explain why some children perform differently on logically equivalent tasks

- Does not account for influences of different cultural, emotional, and social experiences or differences in education or motivation
- Does cognitive development stop in adolescence? Are there distinct stages versus continuous/gradual process?
- Underestimated abilities of younger children-confusion between motor/memory abilities and cognition

i. Describe developmental tests based on Piaget's theory

- Piagetian tools describe strengths and weaknesses; do not provide a numerical score (unlike standard intelligence tests)
- Some tasks (object permanence, sorting/classification tasks) are part of tests like *Bayley Scales of Infant and Toddler Development-3* or the *Mullen Scales of Early Learning*
- Alternative measures of cognitive evaluation in young children include *The Program Evaluation Measure* and *Southern California Ordinal Scales of Development*-->advantage over traditional intelligence testing as can be given at frequent intervals to provide formative feedback and helpful in guiding educational interventions

4. Psychodynamic (Freud)

a. Understand the three primary motivating forces of psychosexual development

- Id: present from birth, driven by the pleasure principle
    - Consists of instincts and impulses; all needs/wants must be immediately met or tension develops; primarily in unconscious mind and is source of energy for later development of ego and superego
- Ego: develops later (and floats between 3 states of mind described below), decision-maker and operates from reality principle
    - Consists of perceptual, cognitive, executive, memory and defensive functions of the mind; weighs costs and benefits of an action before deciding what to do; does not take into account whether action is right/wrong
- Superego: last to develop; upholds the moral standard, communicates with ego through feelings (pride, shame, guilt)
    - Consists of conscience and ego ideal; conscience is compilation of punishments and warnings that child has experienced (role is to punish); ego ideal is compilation of rewards and positive reinforcement child has received (role is to reward); tends to reside in subconscious

b. Distinguish between the concepts of conscious/unconscious/ subconscious awareness

- Freud imagined the mind as an iceberg
    - Conscious: one's awareness at a particular moment; "tip of the iceberg"
    - Subconscious (Preconscious): thoughts available to the conscious mind through focused attention; "part of iceberg just below the waterline"
    - Unconscious: contains wishes, fear, impulses and repressed memories; cannot directly communicate with conscious mind, but contents could dramatically impact life; "largest part of iceberg, well under waterline"

c. Recognize the role of denial in response to a death, chronic illness, disability, or other significant stress

- Internal conflict of 3 forces produces anxiety-Ego attempts to reduce anxiety using defense mechanisms (indirect ways of dealing or coping)
- Denial is one of many defense mechanisms-entails ignoring or refusing to believe an unpleasant reality, to protect one's psychological well-being in a traumatic situation
- Serves to protect person from emotional shock and intense grief that often accompanies news of death/chronic illness/disability-->may lead to refusal to make any lifestyle changes

d. Differentiate between signs of regression and of projection

- Regression: reversion back to childlike emotional state in which your unconscious fears, anxieties and "angst" reappear, particularly under conditions of stress; behaviors become more childish or primitive (e.g. sucking thumb, wetting the bed, road rage)
- Projection: attributing one's own unacceptable emotions to others; defense mechanism by which individual directly projects own undesirable thoughts, motivations or desires onto another (e.g. parent of child with significant illness may not be able to express anger and instead say physician is angry with them)

Other defense mechanisms include:

- Displacement: transfer of emotions from an unacceptable person or objects to a different person or objects; satisfying an impulse with a substitute object
- Sublimation: acting out unacceptable impulses by converting them to acceptable ones
- Repression: unconscious mechanism employed by ego to keep disturbing or threatening thoughts from becoming conscious; although hidden, will create anxiety

e. Understand the impact of transference and counter-transference on the relationship of physician and patient

- Transference: unconscious redirection of feelings from the past to a person or situation in the present; usually some aspect of feelings and attitudes of the previous relation is being transferred, not everything about the relationship (e.g. father angry at pediatrician because he reminds him of his highly controlling father)
    - Important to recognize and manage transference when interferes with or jeopardizes care of the patient
    - May coexist with emotions appropriate to situation, so emotion directed toward HCP may be appropriate response
    - Occurs more frequently when someone ill or afraid and settings where patients seen and emotionally attended to with greater frequency
    - Managed by recognizing importance of relationship of clinician to individual, maintaining professional boundaries/clear limits, and helping individual recognize the transference when they have the capacity to understand it
- Countertransference: feelings or attitudes evoked in a therapist/healthcare provider as a result of transference from the patient
    - Normal, but needs to be dealt with when affects care of child and family, as well as HCP's mental health
    - Clinician should address through reflection on occurrence, including thinking and questioning own feeling and motivation; can also discuss with other members of

healthcare team; consider transfer of care to another clinician when not resolved and affects care of patient or mental health of clinician

f. Know the characteristics of the stages of psychosexual development as described by Freud

- Freud believed parent's behavior toward child in early years largely determined child's personality, which was largely developed by 5-6 years of age; postulated that biological drives (especially pleasure-seeking psychosexual drive/libido) propelled child development
- Believed if psychosexual stages completed successfully-->healthy personality; if not resolved, will continue to have fixations related to that stage
    - Oral Stage (birth-1 year): mouth is main source of pleasure and interaction; sucking and eating are dependent on mother; conflict over weaning-->success results in distinction between other and self; otherwise oral fixation or overly dependent personality
    - Anal Stage (1-3 years): anus is main source of gratification; primary focus is toilet training; overly punitive parents cause child to develop rigid personality traits, while overly lenient parents cause child to become disorganized and defiant
    - Phallic Stage (3-6 years): genitals are main source of gratification; unconscious attraction to opposite gender parent and view same gender parent as rival; gender role and moral development as interactions between id/ego/superego form basic personality; boys=castration fears, girls=penis envy; stage resolves by child identifying with same gender parent
    - Latent Stage (6-12 years): psychosexual energy and instincts suspended, allowing focus on academic/social skills; superego continues to develop through social interaction
    - Genital Stage (12 years-adult): primary task is initiation of mature sexual activity and development of dyadic partner relationship; sexual instincts of phallic stage reappear

g. Understand the limitations of Freudian theory of psychosexual development

- Freud's psychosexual theory has been subject of much criticism as being unscientific, unproven, limited, sexist, and outdated
- Notion that development primarily motivated by sexual and aggressive drives very limited; does not take into account other possible reasons such as innate drive toward mastery or social interaction
- No accounting for individual differences or genetic contribution to development of personality
- Idea of erogenous zones being important at different times in development criticized as reflective of morals of Victorian era culture-->not generalizable to other times/cultures
- Too much importance on Oedipal/Electra complex and toilet training issues in the development of personality
- No consideration for adult development, especially women
- Logic of theory is retrospective (based on adult memories of early childhood experiences)-->unreliable and may be explained by other experiences; also difficult to test with empiric scientific methods

h. Understand that parental anger toward healthcare providers may be a result of projection

- *See sections 4d and 4e above.*

5. Social-emotional (Erikson)

a. Know the parallels between Erickson's and Freud's stages of development from infancy to adolescence

- Erikson's psychosocial theory of development had its basis in Freud's psychosexual theory
    - Shared features such as critical influence of emotions and relationships
    - Agreed with maturational order of stages, that developmental conflicts need to be resolved before moving on to next stage, development occurs gradually, and childhood traumatic experiences can have adverse effect on development
    - Also accepted Freud's theory that unconscious motive can drive behavior (id, ego, superego)
    - Differences: believed social and cultural experiences greatly influenced development, and development occurs over entire lifespan; did not support idea of fixations (rather each stage has own crisis/conflict, and regardless of outcome, individual moves on to other stages)
    - Emphasized role of ego as well as natural drive for competence
- First 5 stages of Erikson's psychosocial theory parallel Freud's 5 psychosexual stages (birth through adolescence, approximately same age ranges) but are broader-->overall experience with the caregiver determines the outcome of the stage
- Last 3 stages of Erikson's theory concern adulthood and have no counterpart in Freud's theory

b. Know the components of the stages of Erickson's paradigm

Table 1–8 Erikson's Psychosocial Stages and Developmental Issues

Age	*Psychosocial Stage*	*Description (Virtue)*
Birth–1 year	Trust vs mistrust	Responsive care giving gives infants a sense of trust in others and self and that the world is a good place. (Hope)
1–3 years	Autonomy vs shame and doubt	Children become more self-sufficient and want independence; reasonable freedom of choice leads to autonomy. (Will)
3–6 years	Initiative vs guilt	Pretend play and acceptance of responsibilities help to foster a sense of direction; children must balance this with the demands of parents. (Purpose)
6–12 years	Industry vs inferiority	Children learn to cooperate with peers and master academic tasks; competency and productivity are important. (Skill)
12–18 years	Identity vs role confusion	Adolescents strive to develop a coherent and lasting personal identity. (Fidelity)
Young adulthood	Intimacy vs isolation	Young adults work to achieve intimate relationships and commitments to other people. Those who have not formed a strong sense of self may have difficulty. (Love)
Adulthood	Generativity vs stagnation	The focus is on child rearing and work productivity to contribute to the next generation. (Care)
Late adulthood	Ego integrity vs despair	Older adults attempt to reflect on their lives and feel satisfied with their successes and failures. (Wisdom)

- *Eight* stage theory characterized by an ego conflict that needs to be resolved successfully to become competent in a specific psychological quality
- Healthy ego development stems from satisfactory resolution of crisis and requires balancing of positive characteristic (predominant) and related negative characteristic (healthy to some degree)-->if successful, particular strength/virtue emerges
    - Early Infancy (birth-1 year): Trust vs. Mistrust-->responsive care giving gives infants sense of trust in self and others and world is a good place; safety and security (HOPE)

- Late Infancy/Toddlerhood (1-3 years): Autonomy vs. Shame/Doubt-->become more self-sufficient and want independence, primary task is control over bodily function (toileting, eating, walking, talking); reasonable freedom of choice leads to autonomy (WILL)
- Early Childhood (3-6 years): Initiative vs. Guilt-->preschoolers start to assert power/control over world through directed play and other social interactions; pretend play and acceptance of responsibilities help foster sense of direction; must balance with SKILL)
- Adolescence (12-18 years): Identity vs. Role Confusion-->strive to develop coherent and lasting personal identity; "Who am I and where am I going?" (FIDELITY)
- Young Adulthood: Intimacy vs. Isolation-->work to achieve intimate relationships and commitments to others (LOVE)
- Adulthood: Generativity vs. Stagnation-->focus on child rearing and work productivity to contribute to next generation (CARE)
- Late Adulthood: Ego Integrity vs. Despair-->attempt to reflect on one's life and feel satisfied with successes and failures (WISDOM)

c. Appreciate the impact of unsuccessful resolution of one of Erickson's stages on children's ability to meet the challenges of subsequent stages

- *See section 5b above.*

d. Know the implications of succeeding at the various stages described by Erickson for success in relationships and school

- *See section 5b above.*

6. Attachment (Mahler, Bowlby, Ainsworth)

a. Appreciate the reciprocal contributions of infants and caregivers on the development of attachment

- Object Relations Theories: focus is the relationships a child has with important people (objects) in his/her life and the idea that characteristics of these people become integrated into own personality and mental processes
  - Mahler and McDevitt examined ways in which attachment and experiences with care givers (objects) affect mental representations child has of those objects and how these images impact development of personality through adulthood
  - Proposed *four* stages in development of psychological sense of self, all in first 3 years of life
    - Autistic Phase (birth-2 months): self-absorbed, little awareness of outside world
    - Symbiotic Phase (2-6 months): psychological blending with primary care giver (sights and sounds of world are fused with self), complete dependence, developing mental image of care giver-->response care giving promotes separation in next phase; harsh/unresponsive care giving have difficulty separating
    - Separation-Individuation Phase (6-24 months): develops separate sense of self and begins to function individually; onset of crawling/moving away from care giver (view care giver from distance); walking/exploration leads to consciousness of self as distinct from care giver
      - Hatching Subphase (6-10 months): begins to respond differently to primary care giver than others

        - Practicing Subphase (10-16 months): experiences disengagement and safety in separation
        - Rapprochement Subphase (16-24 months): more complete experimentation of separating from and returning to primary care giver
    - Object Constancy (24-36+ months): develops stable sense of self founded on availability of reliable mental representations of primary care giver; greater ability to use language and mental representation of care giver and their relationship as a comfort in times of separation
- Attachment Theory: initiated by John Bowlby as evolution to ethologic theory (all mammals exhibit species-specific behaviors that are innate and have evolved over time-->predisposed to behave in certain ways, and to learn certain skills at critical/sensitive periods of development); Bowlby applied this to the maternal bond
    - Mother and child prewired to respond in fixed ways to each other (babies inherently seek contact with mother and many infant reflexes serve to retain proximity by signaling needs; mothers respond to crying/clinging/vocalizations)
    - Infants will attach to any adult, but by 6-9 months, principal attachment will be to primary care giver
    - Bowlby's research focused on what happens with bond interrupted by separation or neglect-->extended by Ainsworth and others
- Attachment is bidirectional between parent and child, and occurs over first several years after birth; Bonding is unidirectional relationship parent develops with infant early days after birth; Temperament describes personality of both parent and child

b. Understand how attachment theory and cognitive developmental theory together explain the phenomena of separation protest and stranger anxiety.

1. Stranger Anxiety - Attachment in the making by 6-9 months attachment to primary care giver (Discriminate social responsiveness), cognitively at 6 months is able to relates to parents with real joy, smiles often while playing with parent at 9 months: back and forth smiles/sounds/gestures…giving and taking activities

2. Separation Anxiety Clear Cut Attachment - 8 mos – 2 yrs Active initiation of proximity seeking and contact – cognitive skills of object permanence has evolved.

c. Differentiate among securely attached, insecurely attached, and ambivalently attached infants using Ainsworth's "strange situation" paradigm

- Ainsworth defined secure attachment as affectional tie between parent and child characterized by seeking contact with object of attachment by proximity or communication; mother is secure base from which child able to explore surroundings without distress, checking back for reassurance to continue
- Attachment behaviors include proximity seeking, clinging, smiling, crying; may increase when infant feels stress or fear; disrupted relationship leads to insecure attachments
- "Strange Situation" used series of interactions with stranger including separations and reunions with mother; looked at Exploratory Behavior before Separation, Behavior during Separation, Reunion Behavior, and Behavior with Stranger (see Table 1-12 in Wolraich, Third Edition)
    - Secure Attachment: child comfortable using mom as base from which to explore, looks back for reassurance but then could explore; when separated gets upset but can be calmed; mom and child both truly pleased at reunion-->**parent is secure base so child better able to explore environment**

- Insecure/Avoidant Attachment: rarely cry upon separation, appear more mature and emotionally controlled than age; upon reunion will avoid mother, failing to reach out and appearing angry/indignant; mother's behaviors angry as well; physical distancing, tenseness, irritability; mother lacks confidence in and derives little pleasure from parenting role-->**manage separations easily, don't seek care or consolation, parents ignore child's negative emotions and fail to respond to child/inconsistent response**
- Ambivalent-Resistant Attachment: most upset upon separation and difficult to console; ambivalent reaction upon reunion (wants to be picked up, but then pushes away); little exploration; parents don't avoid contact, but contact ill-timed and less contingent on cues-->uncoordinated acceptance and rejection, well-meaning but less capable parents-->**significant separation anxiety, inconsistent responses to care giver, and difficulty settling into routines**
- **Disorganized/Disoriented Attachment: could have distress upon exit of caregiver, but response inconsistent; look happy upon reunion but almost dazed/confused, not make direct eye contact; mothers felt to be giving dual or mixed messages leading to noncontingent and disorganized quality of relationship; seen in dyads with psychiatric difficulties (e.g. depression, drug abuse)-->**contradictory behaviors such as manifesting signs of stress even in the presence of the parent, seeking proximity of a stranger rather than the parent, or ignoring a parent after showing distress about the absence of that parent**

d. Recognize the behavioral hallmarks of securely attached infants in the first year of life

- Mother's behavior characterized as "contingent": attuned to infant's needs and respected them; prompt to pick up on distress signals and the tempo of the interaction-smiling and holding close when child indicated, less contact/more space when child indicated
- Good deal of physical closeness

e. Know the components and parenting styles of child-rearing that promote the development of secure attachment

- Parental behaviors that promote secure attachment include sensitive and responsive care; clear, consistent, developmentally appropriate expectations and supervision; warm, positive, and responsive verbal interaction; and viewing the child as an individual with unique responses and needs
- Tips to parents include (*taken from a table referenced in DBPeds PREP, July 2015)*:
  - Enjoy and take pleasure in your relationship with your baby.
  - Observe and pay attention to your baby's subtle cues for attention, exploration, and comfort.
  - Don't worry about "spoiling" your baby.
  - Use nonverbal communication to bond with your baby
  - Make eye contact, touch, smile, and move rhythmically such as by rocking or swaying.
  - Hold and cuddle your baby often.
  - Take care of yourself.
  - Find a childcare provider who works to promote secure attachments with the children in her care and supports your infant's social and emotional development.
  - Provide your toddler with freedom and space to safely explore new environments.
  - Remember that you do not need to be a perfect parent.

f. Recognize signs of insecure attachment seen in infants

- *See section 6c above.*

g. Know how to advise parents based on Mahler's construct of separation-individuation to help promote psychological separation-individuation

- Around 6-7 months, infants start to develop understanding of independence from caregivers; around 8 months, separation and stranger anxiety may emerge and may last through 24 months
- Advise parent to start with brief, predictable separation as first step-will foster successful psychological separation between parent and child (e.g. walker to different room, but continue to talk to child and reappear after short time)
- Incorporate period of introduction time with parent present and providing reassurance, prior to leaving in care of babysitter

h. Understand the limitations of attachment theory

- Other factors that may modify observed attachment behavior include temperament, cultural issues, degree of familiarity, and current physical symptoms
- Many have questioned methodologic integrity of observational studies (e.g. Strange Situation has been criticized); need to develop other empirically valid ways to measure attachment beyond early childhood as well as ways in which attachment security can be measured in non-research settings
- Exact mechanism behind sensitive period for attachment to care giver and how can be empirically validated
- Does not explain how attachment is adaptive for infants and why our species uses these specific behaviors

7. Behavioral theory

a. Distinguish between classical and operant conditioning paradigms

- **Classic conditioning** occurs when two different events occur in such a way that one of the events begins to signal or elicit the other event. For example, a young child may begin to connect frightening experience, such as nightmare, with darkness. Over time this connection might result ina fear of the dark.
- **Operant behaviors** are those that are controlled by what follows them, not by what precedes them. attention is directed toward what happens to the child after entering the darkened room (eg, having a positive experience, such as being entertained in a theater or having a negative experience, such as being frightened by a nightmare).

b. Understand how modeling and shaping can be used to facilitate the expression of a novel and complex behavior

- **Shaping** is formally defined as *reinforcement of successive approximations to the terminal goal*. **Shaping** - assists in **discrimination**, the ability to tell the difference between stimuli that are and are not reinforced and **generalization** the application of a response learned in one situation to a different but similar situation
- **Modeling** demonstrated correct behavior and then rewards accordingly until desired behavior is obtained.

c. Understand how various schedules of reinforcement facilitate learning of a new behavior and maintenance of an established behavior

- **Positive reinforcement** is the addition of an event that increases the behavior (eg, parental attention

when a child has a tantrum may increase tantruming). A **negative reinforcer** (not to be confused with punishment) is the withdrawal of some negative eventthat serves to increase behavior.

d. Be able to counsel a family on the likelihood of post-extinction burst:

- **Extinction** refers to the withholding of reinforcement for a previously reinforced behavior in order to eliminate that behavior. For example, the child who climbs under his desk to gain attention is ignored until he returns to his seat.
- An Extinction Burst occurs when the child repeats the behavior over and over again, in a burst of activity, then stops permanently because of lack of reinforcment.

e. Understand the concept of positive reinforcement in behavior theory

- **Positive reinforcement** is the addition of an event that increases the behavior (eg, parental attention

when a child has a tantrum may increase tantruming).

f. Understand that reinforcement during a post-extinction burst will render the behavior more resistant to extinction – see above

g. Understand the problems associated with punishment-based behavior management plans
Negative Punishment - student lose recess or suspension
Positive Punishment - Chores or corporal punishments
Arguments against non-violent modification of behavior include the issue of ethics, and whether your will should be forced on your children. Positive Parenting and Taking Children Seriously is a way to address the concerns mentioned in this paragraph

h. Know the limitations of behavioral theory
Limitations:

1. Lack of concern with the child's inner experiences
2. No stages are implied: Children, adults and animals all respond the same.
3. If a misbehavior is a result of an underlying emotional problem then exclusive use of behavior therapy may leave the cause untreated.

Strengths:

1. Simplicity
2. Wide application
3. Verifiable

i. Recognize the role of modifying antecedents in changing behavior:
The antecedent (trigger) if modified may decrease the resulting behavior from occurring

8. Social learning theory

a. Know that social learning theories post that the main determinants of individual behavior are observations and interactions with others

- This learning method without direct or obvious reinforcement is called vicarious learning. A process by which imitation or modeling is used. This can be as powerful as direct reinforcement.

b. Understand that according to social learning theories, social hierarchies, family bonds, sex, age, motivation, and perception all influence the rate of an individual's behavior change:

- Social learning/cognitive theory favors a model of causation involving triadic reciprocal determinism. In this model of reciprocal causation, behavior, cognition and other personal factors, and environmental influences (such as social hierarchies, family bonds, and other demographics; inherent personality and temperament) all operate as interacting determinants that influence each other bidirectionally
- Reciprocal causation does not mean that the different sources of influence are of equal strength. Some may be stronger than others. Nor do the reciprocal influences all occur simultaneously. It takes time for a causal factor to exert its influence and activate reciprocal influences.
- The relationship between personal factors and behavior reflects the interaction between thought, affect and action. Expectations, beliefs, self- perceptions, goals and intentions give shape and direction to behavior. What people think, believe, and feel, affects how they behave
    - The natural and extrinsic effects of their actions, in turn, partly determine

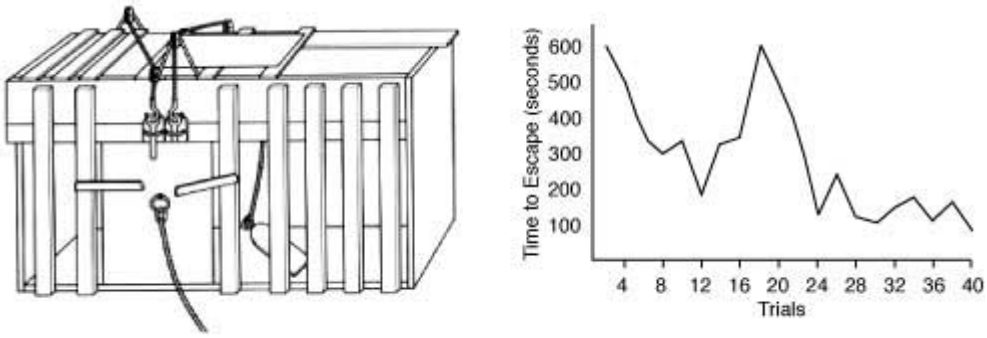
their thought patterns and emotional reactions.

The personal factor also encompasses the biological properties of the organism.
Physical structure and sensory and neural systems affect behavior and impose constraints on capabilities. Sensory systems and brain structures are, in turn, modifiable by behavioral experiences

- The environmental and personal segment of reciprocal causation is concerned with the interactive relationship between personal characteristics and environmental influences.
    - Human expectations, beliefs, emotional bents and cognitive competencies are developed and modified by social influences that convey information and activate emotional reactions through modeling, instruction and social persuasion also evoke different reactions from their social environment by their physical characteristics, such as their age, size, race, sex, and physical attractiveness, quite apart from what they say and do (Lerner, 1982).
    - People similarly activate different social reactions depending on their socially conferred roles and status. For example, children who have a reputation as tough aggressors will elicit different reactions from their peers than those reputed to be unassertive.
    - Thus, by their social status and observable characteristics people can affect their social environment before they say or do anything. The social reactions so elicited affect the recipients' conceptions of themselves and others in ways that either strengthen or alter the environmental bias

c. Differentiate methods of behavior change based on operant conditioning versus social learning theory –

- Operant Conditioning: Operant Conditioning is essentially learning from the consequences of your actions or behavior.
    - "Operant conditioning deals with the modification of 'voluntary behavior' or operant behavior."
    - Operant conditioning uses reinforcement (positive and negative), punishment, and extinction
    - The first person to describe operant conditioning was an American psychologist named Edward Thorndike. He was originally questioning the intelligence of animals→puzzle box with cats → "law of effect."
    - The law of effect states "that the consequences of a response determine whether the response will be performed in the future."



Adapted from Domjan, 1993 (modified from Thorndike, 1898 [left] and Imada & Imada, 1983 [right])

- The Social Learning Theory originated from Albert Bandura. "Bandura's Social Learning Theory posits that people learn from one another, via observation, imitation, and modeling." (Social Learning Theory (Bandura) Learning-Theories.com, 2009).
  - His theory is considered the 'bridge' cogitative and behaviorist learning theories.
  - The Social Learning Theory uses three things. These are observing, imitating, and reinforcement
  - SLT place importance on the mediational process when an individual will change their behavior based on foresight of the consequence of their behavior. Bandura called this "reciprocal determinism"
  - more to learning than just direct reinforcements in learning→people were able to learn by watching other people. This was called observational learning or modeling.
  - Modeling can be used to explain a number of different behaviors. There are four conditions that are necessary for modeling. These conditions are attention, retention, reproduction and motivation.
  - "Bobo" doll experiment

d. Plan a parent group aimed at improving child care practices using social learning theory

A parent group would discuss experience of others that have demonstrated successful or not childcare practices with an emphasis on imitating the good practices.

e. Understand the differences between cognitive behavioral and social influence theories

- Social influence is the change in behavior that one person causes in another, intentionally or unintentionally, as a result of the way the changed person perceives themselves in relationship to the influencer, other people and society in general.
- Cognitive behavioral theory focuses on how people's perceptions of, or spontaneous thoughts about, situations influence their emotional, behavioral (and often physiological) reactions

f. Understand the distinction between behavioral norms and behavioral values.

- Behavioral Norms: see g. below
- Behavioral Values: internally driven/personal as compared to norms which are group driven

g. Know how social influence theory predicts behavioral norms

- Behavioral Norms: The rules that a group uses for appropriate and inappropriate values, beliefs, attitudes and behaviors. These rules may be explicit or implicit.
  - Failure to stick to the rules can result in severe punishments, the most feared of which is exclusion from the group. A common rule is that the some norms must frequently be displayed; neutrality is seldom an option.
  - Other norms include:

- *Injunctive Norms* are behaviors which are perceived as being approved of by other people.
- *Descriptive Norms* are perceptions of how other people are actually behaving, whether or not these are approved of.
- *Explicit Norms* are written or spoken openly.
- *Implicit Norms* are not openly stated (but you find out when you transgress them).

Example

A common group norm amongst academics is that dress is casual (with the underlying implication that what goes on in the mind is more important than what goes on the body).

h. Know the limitations of social learning theory for behavior change

- This theory states that the likelihood that a person will respond to social influence will increase with:

- *Strength*: how important the influencing group of people are to you.
- *Immediacy*: how close the group are to you (in space and time) at the time of the influence attempt.
- *Number*: How many people there are in the group.

- Increasing the numbers has a decreasing incremental effect (going from 2 to 3 has more effect than going from 66 to 67). In fact beyond four or five, the effect tails off rapidly. This is the *Social Influence Model*.
- The effect is most powerful when everyone in the group (apart from the person being persuaded) clearly agree.
- Example - In meetings in the workplace, few will speak out if their opinion differs from the majority. →So what? Using it Convince one person about something. Then collaborate with them on persuading a friend (find out first who will most easily be convinced). →Then work through the group, one at a time. →Also work out through interconnected groups.

i. Distinguish between methods of toilet training based on social learning versus behavioral therapy.
   - Behavioral Therapy – pottie stickers, rewards, parents give increase attention
   - Social Learning – social pressure for hygiene reason to learn to go in a toilet not anywhere one pleases

j. Know how behavioral values are related to the adoption and maintenance of high-risk behaviors

Values are the rules by which we make decisions about right and wrong, should and shouldn't, good and bad. They also tell us which are more or less important, which is useful when we have to trade off meeting one value over another.

Dictionary.com defines values as: n : beliefs of a person or social group in which they have an emotional investment (either for or against something); "he has very conservatives values"

9. Moral reasoning (Kohlberg)

a. Know that moral reasoning in Kohlberg's theory progresses from explanations based on what feels good to an individual to explanations based on abstract principles of fairness and justice.

b. Understand the poor association between the level of moral reasoning and the individual's moral behavior

This is one of the deficiencies in Kohlberg's theory. People argue that we should be more focused on what occurs in real life. The other one is that there are gender differences in moral reasoning and Kohlberg's theory focuses on middle aged white male (Kohlbergs test subjects) moral reasoning.
c. Understand the stages of moral reasoning as defined by Kohlberg
3 Basic Levels-Each with 2 substages:

- **Preconventional Level of Morality – depend on standards of alder authority figures -**

Stage 1: Obedience and Punishment Orientation: it is moral to obey authority and avoid punishment

Stage 2: Individualism and Exchange: morality is relative; one should pursue one's own interests

- **Conventional Level – adolescents appreciate moral codes of society -**

Stage 3: Good Interpersonal Relationships: being moral includes being helpful to others

Stage 4: Maintaining the Social Order: morality is in preserving the laws of society

- **Post-Conventional Level –more abstract principles are consider above social conventions -**

Stage 5: Social Contract and Individual Rights: democratic process and basic human rights are moral

Stage 6: Universal Principles: universal principles of justice and individual rights are moral

d. Know how to interpret a child's explanations for moral decisions based on Kohlberg's theory
Stage 1 Childhood (pre K – early elementary school) – avoid getting in trouble at all cost, others interests are not considered or is the room for differing opinions. Right and wrong is base on how the parent reacts to a situation.
Stage 2 (elementary school) rules are based on rewards and ensuring fairness.

10. Individual variation

a. **Understand the concept of "goodness of fit" and implications for interpreting parents' perceptions of their children's temperament.**

The concept of matching temperament with learning styles to create a "goodness of fit" as proposed by Thomas and Chess (1977) is summarized by the authors as follows:

> *"Stated briefly, there is a goodness of fit when the person's temperament and other characteristics such as motivation and levels of intellectual and other abilities, are adequate to master the successive demands, expectations, and opportunities of the environment.*
>
> *This formulation stems from the conviction that normal or pathologic psychologic development does not depend on temperament alone. Rather, it is the nature of the interaction between temperament and the individual's other characteristics with specific features of the environment which provides the basic dynamic influence for the process of development.*
>
> *If there is a goodness of fit between child and environment, the foundation for a healthy self-concept and stabel self-esteem is laid down. If there is a poorness of fit, a negative, denigrated self-evaluation begins to crystalize. If, in latter childhood or even in adult life, a poorness of fit can be altered, such as by the emergence of new positive capacities or a favorable change in the environment, then a negative self-image may be transformed into a positive one* (p.15-16).

Another Study concluded (Child Psychiatry Hum Dev. 1981 Spring;11(3):167-78**)** The degree of adult control or demands is an important component in understanding the "goodness of fit" between parent behavior and child characteristics and that a child's temperament and its relationship to adult behavior cannot be considered in isolation from other child characteristics, particularly the child's sex. Children's sex and temperament were found to interact in relationship with adult controlling behavior

while no child behaviors varied as a function of sex or temperament alone. Adults were found to adjust their controlling behavior as a function of the child's sex and temperament.

b. Identify the major categories used for describing temperament

Activity Level: This is the child's "idle speed or how active the child is generally. Does the infant always wiggle, more squirm? Is the infant difficult to diaper because of this? Is the infant content to sit and quietly watch? Does the child have difficulty sitting still? Is the child always on the go? Or, does the child prefer sedentary quiet activities? Highly active children may channel such extra energy into success in sports; may perform well in high-energy careers and may be able to keep up with many different responsibilities.

Distractibility: The degree of concentration and paying attention displayed when a child is not particularly interested in an activity. This trait refers to the ease with which external stimuli interfere with ongoing behavior. Is the infant easily distracted by sounds or sights while drinking a bottle? Is the infant easily soothed when upset by being offered alternate activity? Does the child become sidetracked easily when attempting to follow routine or working on some activity? High distractibility is seen as positive when it is easy to divert a child from an undesirable behavior but seen as negative when it prevents the child from finishing school work.

Intensity: The energy level of a response whether positive or negative. Does the infant react strongly and loudly to everything, even relatively minor events? Does the child show pleasure or upset strongly and dramatically? Or does the child just get quiet when upset? Intense children are more likely to have their needs met and may have depth and delight of emotion rarely experienced by others. These children may be gifted in dramatic arts. Intense children tend to be exhausting to live with.

Regularity: The trait refers to the predictability of biological functions like appetite and sleep. Does the child get hungry or tired at predictable times? Or, is the child unpredictable in terms of hunger and tiredness? As grown-ups irregular individuals may do better than others with traveling as well as be likely to adapt to careers with unusual working hours.

Sensory Threshold: Related to how sensitive this child is to physical stimuli. It is the amount of stimulation (sounds, tastes, touch, temperature changes) needed to produce a response in the child. Does the child react positively or negatively to particular sounds? Does the child startle easily to sounds? Is the child a picky eater or will he eat almost anything? Does the child respond positively or negatively to the feel of clothing? Highly sensitive individuals are more likely to be artistic and creative.

Approach/Withdrawal: Refers to the child's characteristic response to a new situation or strangers. Does the child eagerly approach new situations or people? Or does the child seem hesitant and resistant when faced with new situations, people or things? Slow-to-warm up children tend to think before they act. They are less likely to act impulsively during adolescence.

Adaptability: Related to how easily the child adapts to transitions and changes, like switching to a new activity. Does the child have difficulty with changes in routines, or with transitions from one activity to another? Does the child take a long time to become comfortable to new situations? A slow-to-adapt child is less likely to rush into dangerous situations, and may be less influenced by peer pressure.

Persistence: This is the length of time a child continues in activities in the face of obstacles. Does the child continue to work on a puzzle when he has difficulty with it or does he just move on to another activity? Is the child able to wait to have his needs met? Does the child react strongly when interrupted in an activity? When a child persists in an activity he is asked to stop, he is labeled as stubborn. When a child stays with a tough puzzle he is seen a being patient. The highly persistent child is more likely to succeed in reaching goals. A child with low persistence may develop strong social skills because he realizes other people can help.

Mood: This is the tendency to react to the world primarily in a positive or negative way. Does the child see the glass as half full? Does he focus on the positive aspects of life? Is the child generally in a happy mood? Or, does the child see the gall as half empty and tend to focus on the negative aspects of life? Is the child generally serious? Serious children tend to be analytical and evaluate situations carefully.

c. Know the temperament characteristics of the "difficult child" syndrome

Difficult children tend to display power and revenge behaviors. They are typically confrontational, often hostile, and definitely disruptive. They are the ones we are least likely to want to assist, manipulative, they spot and exploit insecurities.

d. Plan appropriate management for a family upset by one of their child's temperament characteristics

Parenting Strategies For Very Intense Children: Provide activities that are soothing such as warm bath, massage, water play, stories. Recognize cues that signal that intensity is rising. Help child learn to recognize cues that signal that intensity is rising. Use humor to diffuse intensity. Teach child to use time-out as a time to calm self-down. Avoid escalating intensity of child be reacting intensely to his/her behavior. Give calm, clear, brief feedback.

Parenting Strategies for Slow-to-Adapt Children: Establish clear routines. Prepare child by discussing plans for the day when routine changes. Prepare child for transitions. Give warnings a few minutes before transition from one activity to next occurs. Allow time for closure of one activity before going on to next. Stay aware of number of transitions required, and keep transitions to minimum if possible.

e. Understand the prevalence of the "difficult child" cluster of temperament characteristics

- Thomas, Chess, Birch, Hertzig and Korn found that many babies could be categorized into one of three groups: *easy*, *difficult*, and *slow-to-warm-up*. (Thomas & Chess 1977). Not all children can be placed in one of these groups. Approximately 65% of children fit one of the patterns. Of the 65%, 40% fit the easy pattern, 10% fell into the difficult pattern, and 15% were slow to warm up. Each category has its own strength and weakness and one is not superior to another
- Thomas, Chess, Birch, Hertzig and Korn showed that *Easy* babies readily adapt to new experiences, generally display positive moods and emotions and also have normal eating and sleeping patterns. *Difficult* babies tend to be very emotional, irritable and fussy, and cry a lot. They also tend to have irregular eating and sleeping patterns. *Slow-to-warm-up* babies have a low activity level, and tend to withdraw from new situations and people. They are slow to adapt to new experiences, but then slowly accepts them after repeated exposure
- Thomas , Chess, Birch, Hertzig and Korn found that these broad patterns of temperamental qualities are remarkably stable through childhood. These traits are also found in children across all cultures

f. **Understand the natural history of infants with the "difficult child" cluster of temperament characteristics**

- Thomas and Chess also studied temperament and environment. One sample consisted of white middle class families with high educational status and the other was of Puerto Rican working class families. They found several differences. Among those were:
- Parents of middle class children were more likely to report behavior problems before the age of nine and the children had sleep problems. This may be because children start preschool between the ages of three and four. Puerto Rican children under the age of five showed rare signs of sleep problems, however, sleep problems became more common at the age of six.

- Middle class parents also placed great stress on the child's early development, believing that problems in early ages were indicative of later problems in psychological development, whereas Puerto Rican parents felt their children would outgrow any problems.
- At the age of nine, the report of new problems dropped for middle class children but they rose in Puerto Rican children, possibly due to the demands of school

g. Know the limitations of temperament as an explanatory concept:

- Most experts agree that temperament does have a genetic and biological basis; but researchers also agree that environmental experiences can modify a child's temperament.
- Differences of temperament or behavior styles among each individual are important in family life. They affect the interactions among family members.
- While some children can adapt quickly and easily to family routines and get along with siblings, others who are more active or intense may have a difficult time adjusting.
- It is the interactions between these children and their parents and/or siblings that can lead to stress and friction within the family life.

11. Theories of behavior change

a. Know the key health beliefs that, according to the Health Belief Model, produce a readiness to act.

- The HBM was originally developed as a systematic method to explain and predict preventive health behavior. It focused on the relationship of health behaviors, practices and utilization of health services. In later years, the HBM has been revised to include general health motivation for the purpose of distinguishing illness and sick-role behavior from health behavior. Originated around 1952- measures including x-rays for screening for TB. It is generally regarded as the beginning of systematic, theory-based research in health behavior.
- The HBM states that the perception of a personal health behavior threat is itself influenced by at least three factors: general health values, which include interest and concern about health; specific health beliefs about vulnerability to a particular health threat; and beliefs about the consequences of the health problem. Once an individual perceives a threat to his/her health and is simultaneously cued to action, and his/her perceived benefits outweighs his/her perceived benefits, then that individual is most likely to undertake the recommended preventive health action

b. Understand how perceived barriers or costs and perceived benefits of behavior change can influence the efficacy of risk reduction efforts ----

- The Health Belief Model proposes that people are most likely to take preventative action if they perceive the threat of a health risk to be serious, if they feel they are personally susceptible and if there are fewer costs than benefits to engaging in it
- Therefore, a central aspect of the Health Belief Model is that behavior change interventions are more effective if they address an individual's specific perceptions about susceptibility, benefits, barriers, and self-efficacy
- Interventions focusing on this model may involve risk calculation and prediction, as well as personalized advice and education.
- The model was based on an assumption that people fear diseases, and that health actions are motivated in relation to the degree of fear (perceived threat) and expected fear-reduction potential of actions, as long as that potential outweighs practical and psychological obstacles to taking action (net benefits)
- The four key constructs of the health belief model are identified as perceived susceptibility and perceived severity (two dimensions of "threat"), and perceived benefits and perceived barriers (the components of "net benefits")→it would lead to the understanding then, that if net benefits did not outweigh perceived threats due to barriers (costs of risk reduction efforts) & if you were not able to influence behavioral change regarding a perceived threat (i.e. convincing people who have

never gotten a flu vaccine or had any personal adverse outcomes related to influenza that it is still a good preventative measure to take every year)

c. Understand the stages of change described by Prochaska in the Transtheoretical Model.

d. Know how assessment of an individual's stage of change helps in the selection of an appropriate intervention to facilitate behavior change (eg, weight reduction, safe sex practices, stopping cigarette smoking).—answer to c. and d. below

According to Dr. Prochaska's original book on *Systems of Psychotherapy: A Transtheoretical Analysis* published in 1979, there were so many theories in the field of psychotherapy that this encouraged him to pursue his own research. In this book, he did a comparative analysis of 18 major theories of psychotherapy and behavioral change such as consciousness raising from the Freudian school of thought, contingency management from the Skinnerian tradition, and helping relationships from the Rogerians. Thus, the term **transtheoretical.**

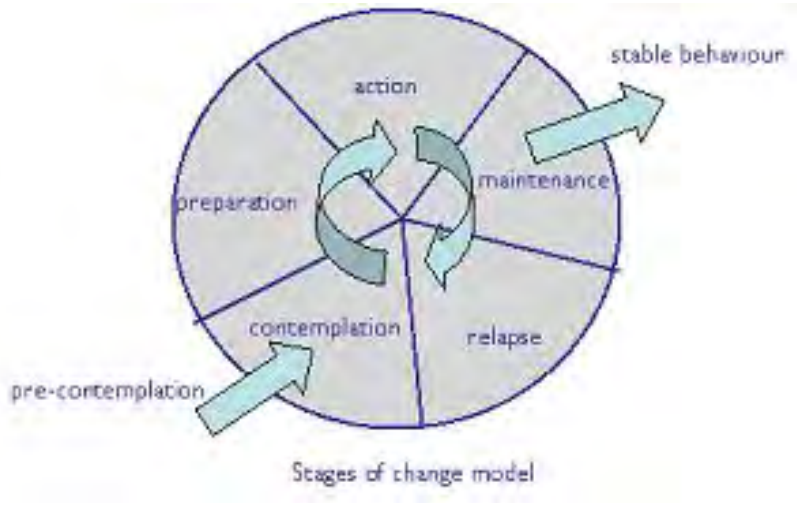
The Transtheoretical Model (also called the Stages of Change Model), developed by Prochaska and DiClemente in the late 1970s, evolved through studies examining the experiences of smokers who quit on their own with those requiring further treatment to understand why some people were capable of quitting on their own. It was determined that people quit smoking if they were ready to do so. Thus, the Transtheoretical Model (TTM) focuses on the decision-making of the individual and is a model of intentional change. The TTM operates on the assumption that people do not change behaviors quickly and decisively. Rather, change in behavior, especially habitual behavior, occurs continuously through a cyclical process. The TTM is not a theory but a model; different behavioral theories and constructs can be applied to various stages of the model where they may be most effective.

The TTM posits that individuals move through six stages of change: precontemplation, contemplation, preparation, action, maintenance, and termination. Termination was not part of the original model and is less often used in application of stages of change for health-related behaviors. For each stage of change, different intervention strategies are most effective at moving the person to the next stage of change and subsequently through the model to maintenance, the ideal stage of behavior.

Stages of Change under the TTM:

1. Precontemplation - In this stage, people do not intend to take action in the foreseeable future (defined as within the next 6 months). People are often unaware that their behavior is problematic or produces negative consequences. People in this stage often underestimate the pros of changing behavior and place too much emphasis on the cons of changing behavior.
2. Contemplation - In this stage, people are intending to start the healthy behavior in the foreseeable future (defined as within the next 6 months). People recognize that their behavior may be problematic, and a more thoughtful and practical consideration of the pros and cons of changing the behavior takes place, with equal emphasis placed on both. Even with this recognition, people may still feel ambivalent toward changing their behavior.
3. Preparation (Determination) - In this stage, people are ready to take action within the next 30 days. People start to take small steps toward the behavior change, and they believe changing their behavior can lead to a healthier life.
4. Action - In this stage, people have recently changed their behavior (defined as within the last 6 months) and intend to keep moving forward with that behavior change. People may exhibit this by modifying their problem behavior or acquiring new healthy behaviors.
5. Maintenance - In this stage, people have sustained their behavior change for a while (defined as more than 6 months) and intend to maintain the behavior change going forward. People in this stage work to prevent relapse to earlier stages.

6. Termination - In this stage, people have no desire to return to their unhealthy behaviors and are sure they will not relapse. Since this is rarely reached, and people tend to stay in the maintenance stage, this stage is often not considered in health promotion programs.



Stages of change model

e. Know the limitations of the Trans-theoretical Model of behavior change –

- A model based on multiple major theories
- **Limitations** of the **model** include the following:
  1) The theory ignores the social context in which change occurs, such as SES and income.
  2) The lines between the stages can be arbitrary with no set criteria of how to determine a person's stage of change. The questionnaires that have been developed to assign a person to a stage of change are not always standardized or validated.
  3) There is no clear sense for how much time is needed for each stage, or how long a person can remain in a stage.
  4) The model assumes that individuals make coherent and logical plans in their decision-making process when this is not always true.

f. Understand the concept of motivational interviewing and its relationship to theories of behavioral change

- Motivational Interviewing (MI) was developed by William R. Miller and Stephen Rollnick, and is defined as a collaborative, person-centered directive counseling method for addressing the common problem of ambivalence about behavior change.
- It is designed to strengthen personal motivation for and commitment to a specific goal by eliciting and exploring the person's own reasons for change, within an atmosphere of acceptance and compassion.
- The concept of Motivational Interviewing evolved from experiences with problem drinkers and now is applied to a wide range of concerns such as healthcare improvement, high-risk sexual behaviors, diabetes management, smoking cessation, and mental health problems.
- MI recognizes and accepts the fact that clients who need to make changes in their lives approach counseling at different levels of readiness to change their behavior-→tied in heavily, therefore, with the stages of changes under the Trans-Theoretical Model and where a person is in those stages to structure MI model for individual patient
- MI involves collaboration rather than confrontation, evocation rather than education, autonomy rather than authority, and exploration rather than explanation.
- Effective processes for positive change focus on goals that are small, important to the client, specific, realistic, and oriented in the present and/or future.

- An emergent theory of MI is proposed (Miller, 2009), emphasizing two specific active components: a *relational* component focused on empathy and the interpersonal spirit of MI, and a *technical* component involving the differential evocation and reinforcement of client change talk

Key Components of MI

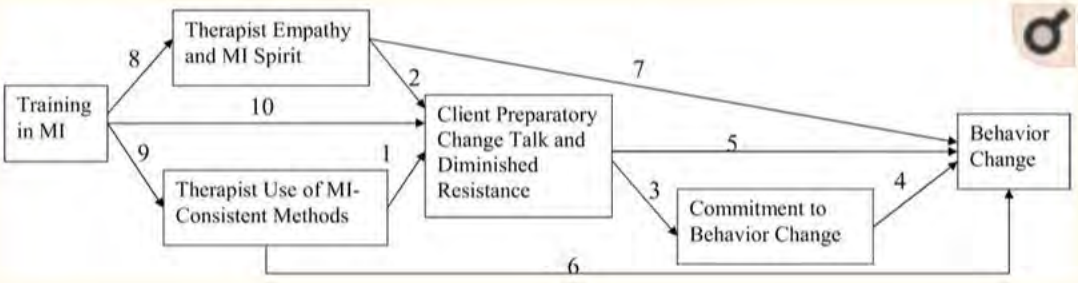
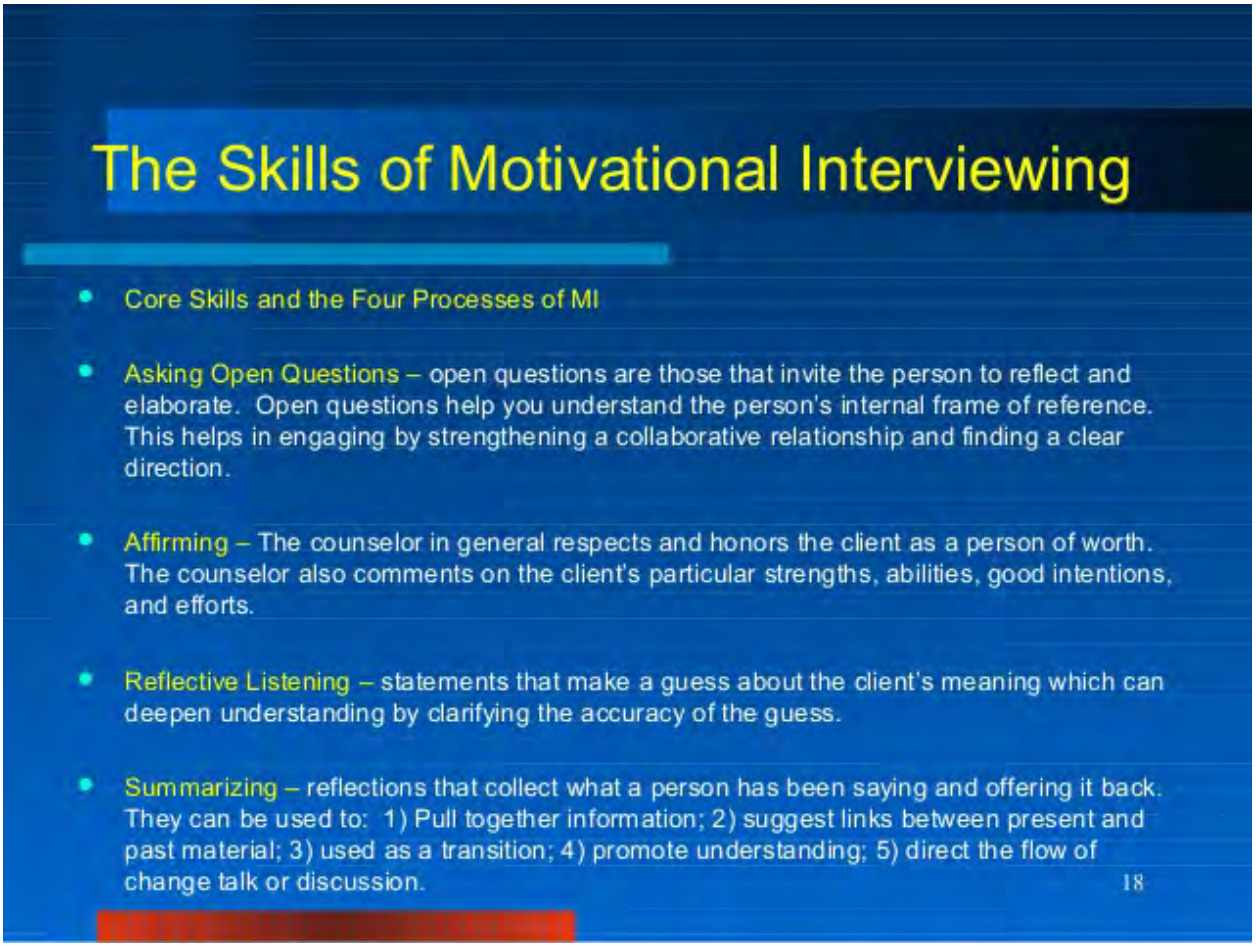


Figure 1

Hypothesized Relationships Among Process and Outcome Variables in Motivational Interviewing


The Skills of Motivational Interviewing
Core Skills and the Four Processes of MI
Asking Open Questions – open questions are those that invite the person to reflect and elaborate. Open questions help you understand the person's internal frame of reference. This helps in engaging by strengthening a collaborative relationship and finding a clear direction.
Affirming – The counselor in general respects and honors the client as a person of worth. The counselor also comments on the client's particular strengths, abilities, good intentions, and efforts.
Reflective Listening – statements that make a guess about the client's meaning which can deepen understanding by clarifying the accuracy of the guess.
Summarizing – reflections that collect what a person has been saying and offering it back. They can be used to: 1) Pull together information; 2) suggest links between present and past material; 3) used as a transition; 4) promote understanding; 5) direct the flow of change talk or discussion.
18

Content Category 2- Biological Mechanisms in Developmental & Behaviors-Part A-C

ABP Content Specs for Developmental-Behavioral Pediatrics Fellow Collaboration Project

Prepared by Rebecca Christi, MD, Madigan Army Medical Center DBP Fellow

Reviewed by, Eric Flake, MD, Staff Developmental Behavioral Pediatrics Madigan Army Medical Center

2. Biological Mechanisms in Development and Behavior

A. Early development of the central nervous system
   1. Understand the neuroanatomic anomalies that arise from perturbations in neural tube development
   2. Understand current theories on the genetic control of central nervous system development
   3. Recognize the importance of folate in preventing defects in neural tube closure
   4. Understand the neuroanatomic abnormalities that arise from perturbations in neuronal migration
   5. Understand that developing neurons form complex dendritic arbors with synaptic connections to many other neurons
   6. Understand the timing and the process of myelination in the central nervous system
   7. Understand the influence of steroid hormones on the organization of the developing brain
   8. Understand the effects of stress on the developing brain

B. Later development of the central nervous system
   1. Understand that the post-natal maturation of the central nervous system includes a subtractive process (pruning)
   2. Understand that the period of time that synaptic pruning is most active may vary according to region of the brain
   3. Realize that synaptic pruning often results in loss of cortical plasticity
   4. Know how molecular processes and synaptic activity reflect learning at a cellular level
   5. Know the definition of "plasticity" and the neurobiological mechanisms that are believed to underlie plasticity
   6. Know the effect of environmental enrichment on neurobiological development

C. Functional organization of the central nervous system
   1. Understand that the cortex is highly interconnected, facilitating interactions between parts of the brain that are engaged in different types of information processing
   2. Realize that the brain systems responsible for a given function may be different in children than in adults
   3. Know the range of functions that are served by the frontal lobes
   4. Know the range of functions that are served by the parietal lobes
   5. Know the range of functions that are served by the temporal lobes
   6. Know the range of functions that are served by the occipital lobes
   7. Know the range of functions that are served by the cerebellum
   8. Know the range of functions that are served by the basal ganglia
   9. Explain the theory of dual cortical streams for visual-spatial processing
   10. Understand the layered and columnar organization of the normal neocortex

2. Biological Mechanisms in Development and Behavior

A. Early development of the central nervous system

1. Understand the neuroanatomic anomalies that arise from perturbations in neural tube development

a. Neural tube defects (NTDs) affect 0.5-2 per 1000 established pregnancies, worldwide and are the second commonest group of birth defects, after congenital heart defects, and are the most common congenital malformation of the CNS. Neural tube defects are major birth defects of the brain and spine that occur early in pregnancy due to improper closure of the embryonic neural tube, which may lead to a range of disabilities or death.

i) The most common neural tube defects **are anencephaly (an underdeveloped brain and an incomplete skull) and spina bifida (incomplete closing of the spinal cord)**

ii) Based on 2009-2011 data, the estimated average annual prevalence of anencephaly and spina bifida combined was 6.5 cases per 10 000 live births.

iii) Result of failure of neural tube closure in the 3rd and 4th week of intrauterine development

iv) **Spina bifida** is a group of common NTDs in which the spinal cord is exposed or protrudes to the surface with the meninges into a sac-like through a defect in the vertebral wall. The 4 types of spina bifida are: **myelomeningocele, meningocele, myelocele, and spina bifida occulta.**
   (1) When this closure defect involves herniation of only cerebrospinal fluid, it is called **myelocele**
   (2) A myelocele that contains meninges is a **meningomyelocele**
   (3) With both meninges and spinal cord it is called **myelomeningocele**
   (4) Commonly associated conditions with spina bifida are hydrocephalus and Arnold-Chiari malformation type II (a combination of myelomeningocele and cerebellar tonsil herniation).

v) **Anencephaly** is one of the common types of NTDs with a congenital absence of the brain or parts of the brain and cranium→it occurs as a result of the failure of the cranial portion of neural tube closure

vi) **Encephalocele** is a rare NTD in which the brain protrudes through an abnormal opening of the cranium with or without the meninges, leaving a projection of a bag-like structure handing on the head
   (1) It is also frequently associated with other abnormalities of the CNS like hydrocephalus, especially with the posterior encephaloceles
   (2) This condition may be the result of aqueductal stenosis or torsion and may also be a post-surgical complication of encephalocele

vii) Understanding of NTDs requires an understanding the embryological development of the central nervous system (brain and spinal cord)- specifically the formation of the neural tube

b. CNS develops from the embryonic ectoderm alongside other structures like the skin.

e-TABLE 24.1 Schematic Chronology of the Major Events During Human Neocortical Development

Event	Time Event Occurs
Neuroectoderm induction	Third GW
Neurulation	Third to end of fourth GW
Proencephalic and hemispheric formation	Fifth to tenth GW
Neuronal proliferation	Tenth to twentieth GW
Neuronal migration	Twelfth to twenty-fourth GW
Programmed neuronal cell death	Twenty-eighth to forty-first GW
Neurogenesis	Fifteenth to twentieth GW to well into postnatal months or years
Synaptogenesis	Twentieth GW to puberty
Gliogenesis	Twenty to twenty-fourth GW to well into postnatal years
Myelination	Twenty-sixth to twenty-eighth GW to 2 to 3 postnatal years
Angiogenesis	Fifth to tenth GW to well into postnatal years

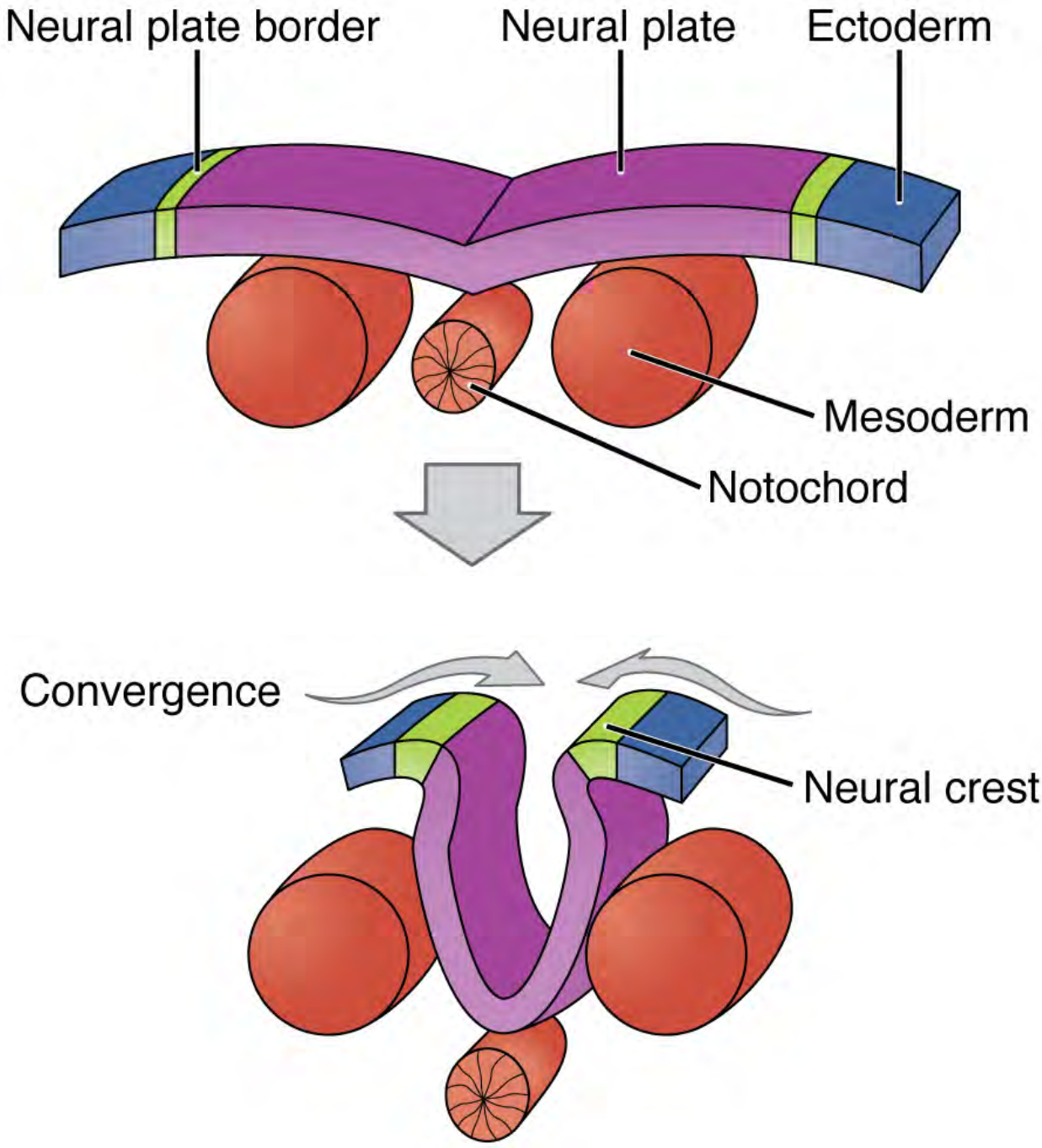
GW, Gestational week.

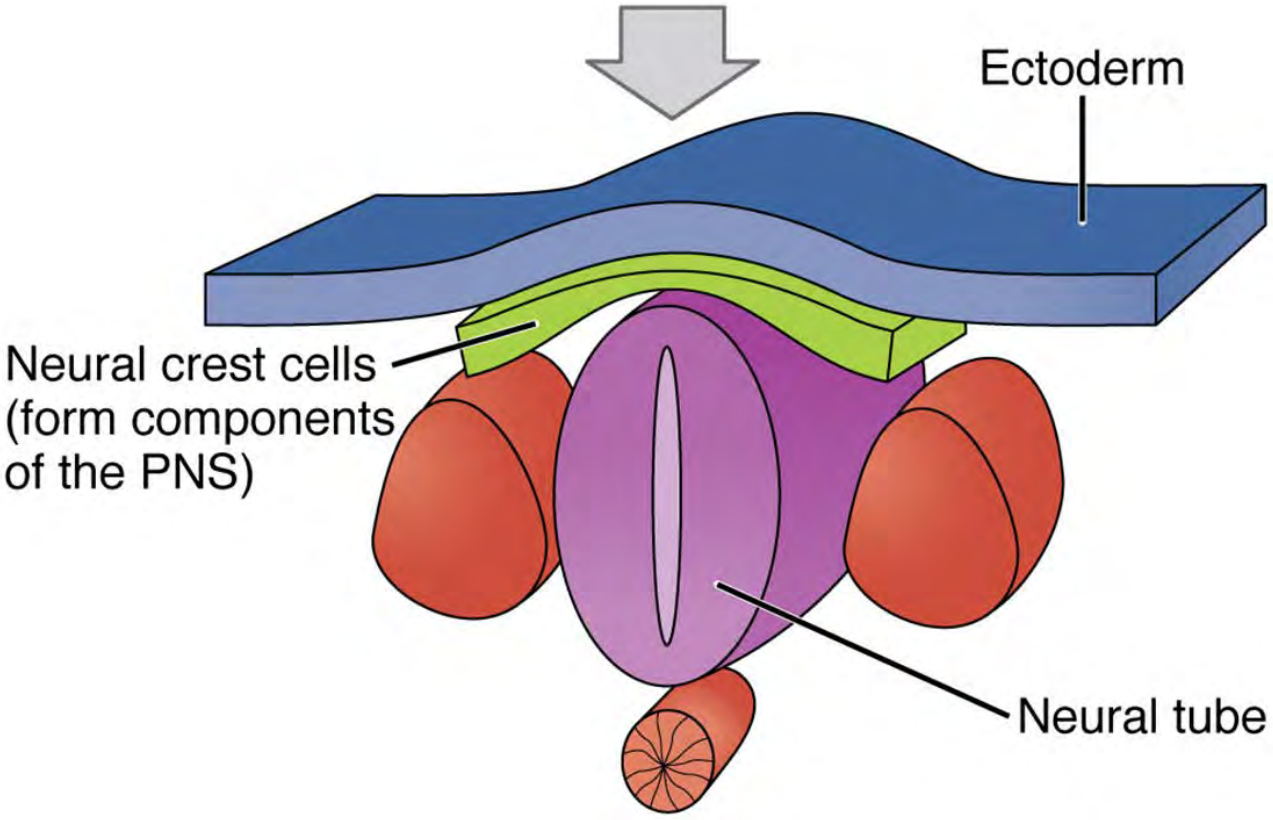
c. Development of the CNS begins in 3rd-4th weeks of embryonic life

i) development of the CNS initially begins with process of neuralation (development of the neural tube)-

- About 2 weeks after conception, the developing embryo has organized itself into a three-layered, spherical structure

- In one area of this sphere, the cells thicken to form what is called the *neural plate*. This plate then folds over onto itself, forming a tube that gradually closes first at the bottom and then at the top, much like a zipper.


Neural plate border
Neural plate
Ectoderm
Mesoderm
Notochord
Convergence
Neural crest



- The neural groove proliferates, and ultimately forms the neural tube. The inner cells of the neural tube will lead to the formation of the central nervous system (brain and spinal cord) while the outer cells will give rise to the autonomic nervous system (nerves outside the brain and spinal cord).

- The neural tube closes ROSTRALLY and CAUDALLY.

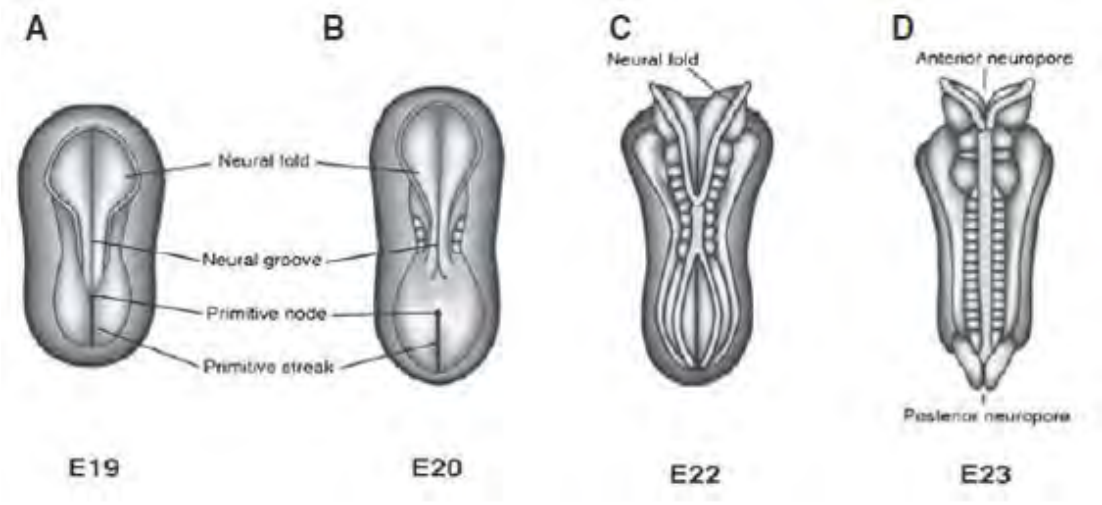
- The defects of neural tube fusion consist of encephalocele, meningocele, myelomeningocele, and spina bifida occulta

- Specifically, alterations in the **closure of the rostral neural tube (anterior neuropore) result in conditions like anencephaly or encephalocele**.

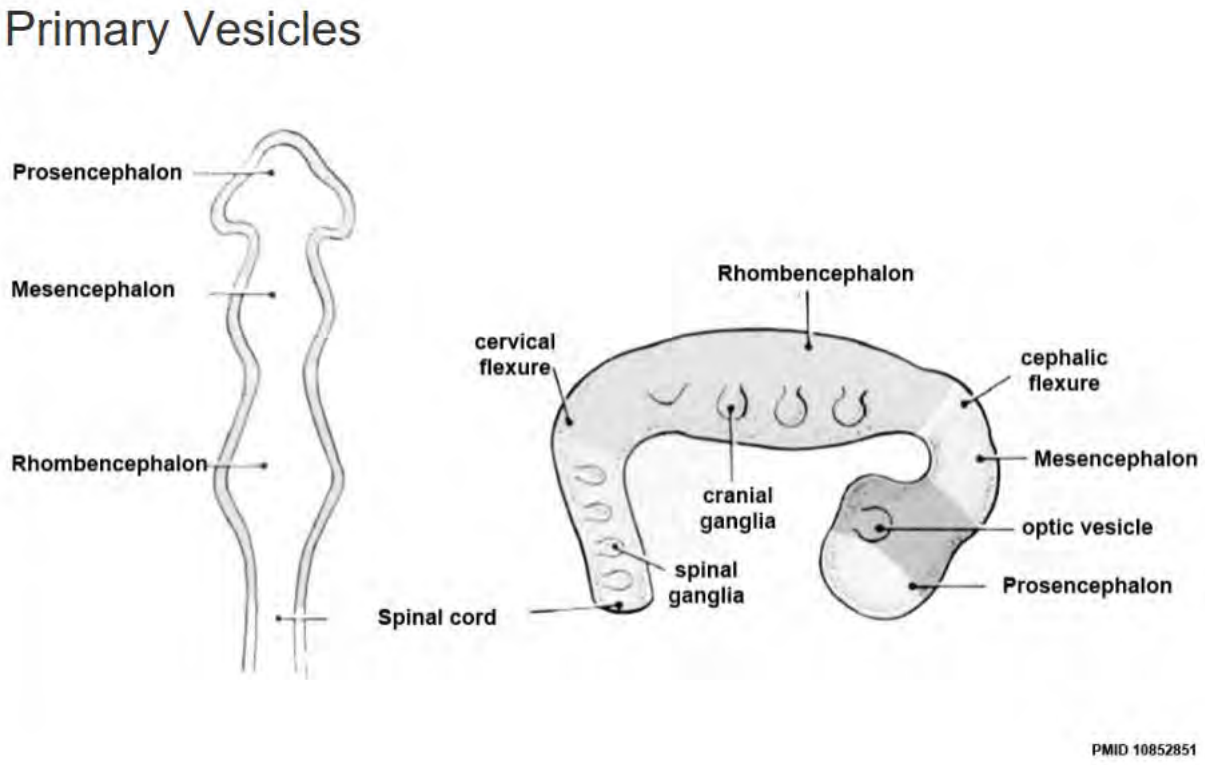
- Myelomeningocele occurs from the **incomplete caudal fusion of the neural tube (posterior neuropore) .**

- See figure below for graphic depicting these stages.

-Anencephaly typically occurs before the 24th day of life and encephalocele and myelomeningocele occur about the 26th day of life.

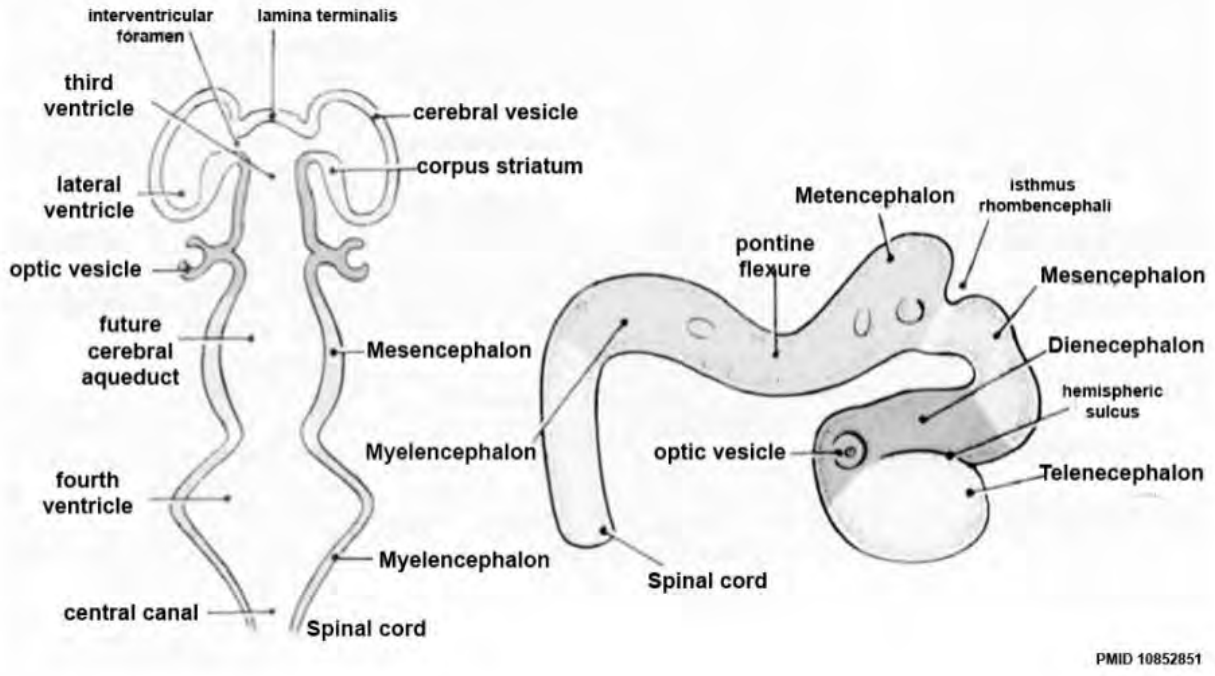


d. Once the neural tube is closed, it becomes a three-vesicle structure (the "primitive brain" which is comprised of the prosencephalon, mesencephalon, and rhombencephalon)→this happens around 5th-6th weeks of gestation (see figure below)



- The PROSENCEPHALON divides further into the telencephalon and diencephalon→ happens through a series of developmental stages, namely: formation, cleavage, and development of the midline; 3 primary vesicles→5 secondary vesicles (see figure below)

Secondary Vesicles



- Once five vesicle structure has formed, the different regions of tissue around the ventricles will become distinct brain structures: 1) The anterior portion of the neural tube will become the *forebrain*, which includes the cerebral hemispheres; 2) the *diencephalon* (the thalamus and the hypothalamus); and 3) the basal ganglia. The cells around the middle vesicle will become the 4) *midbrain*, a structure that connects the diencephalon to the hindbrain. The rear-most portion of the tube will give rise to the 5) *hindbrain*, which will consist of the medulla oblongata, the pons, and the cerebellum. Finally, the cells that remain will give rise to the spinal cord.

- any form of developmental alteration in these steps leads to the malformation of the developing brain

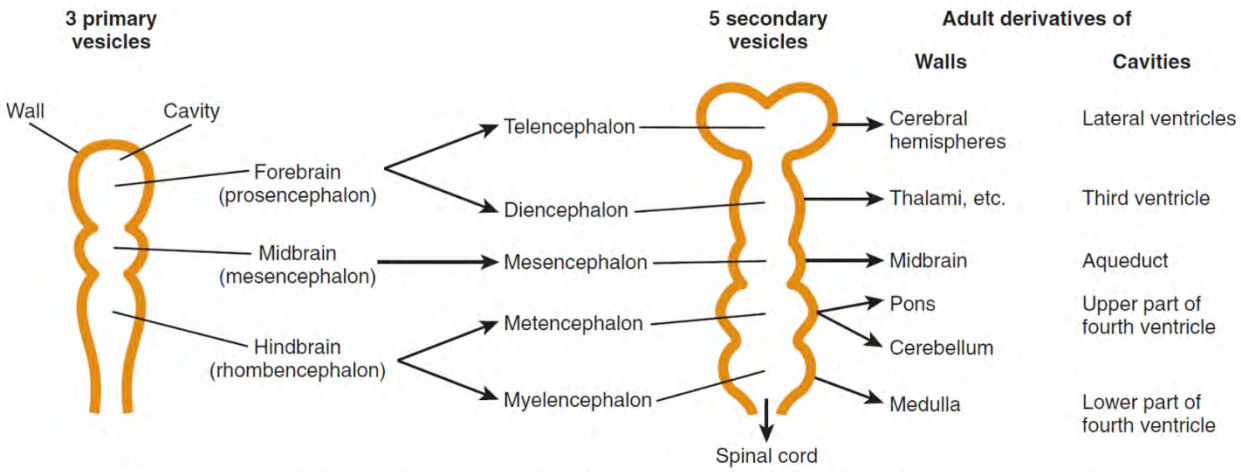


Figure 24.5. Ventral induction. *(From Moore KL, Persaud TVN.* Before We Are Born. *5th ed. Philadelphia: WB Saunders; 1988.)*

f. Stages of the development of the cerebral cortex- happens in 3 major steps and defects in one or a combinations of these steps form the basis of classification of abnormality of cortical development (1) proliferation and differentiation of the neuronal stem cells into neuroblasts and glia cells, (2) migration of neuronal precursors toward the cortical plate by either radial or tangential movements, and (3) cortical organization into six layers associated with synaptogenesis and apoptosis

i) Proliferation of neural cells: an abnormally high proliferation of the neural cells can lead to megalencephaly and decreased proliferation leads to microcephaly

ii) Neuronal migration: (also see A4. Below)

- After the cells are born, they travel to their final destinations. The cerebral cortex is composed of multilayered tissue several millimeters thick.
- It is formed by the movement of cells in an inside-out direction, beginning in the ventricular zone and migrating through the intermediate zone, with the cells eventually reaching their final destination on the outside of the developing brain.
- The earliest migrating cells occupy the deepest cortical layer, whereas the subsequent migrations pass through previously formed layers to form the outer layers.
- About 25 weeks after conception, all six layers of the cortex will have formed→ neocortex 6 layers; allocortex 3-5 layers
- The inside-out pattern of migration described here is that of *radial migration*, which applies to about 70%–80% of migrating neurons, most of which are pyramidal neurons and glia.
- *Pyramidal neurons* are the large neurons in the cortex that are responsible for sending signals to different layers of the cortex and other parts of the brain.
- *Glia* are nonneuronal brain cells that are involved in the support of neuronal processes (such as producing myelin or removing debris, such as dead brain cells).
- In contrast, *interneurons*—relatively smaller neurons that are involved in communication between pyramidal cells within a particular layer of the cortex—follow a pattern of tangential migration.

iii) Postmigrational cortical organization and connectivity: irregular events in the post-migrational cortical organization causes focal cortical dysplasias and polymicrogyria.

2. Understand current theories on the genetic control of central nervous system development.

a. the human cerebral cortex is made up of ~20 billion neurons, each of which makes an average of 7,000 synaptic contacts.

b. to add to this complexity, each neuron contains 46 chromosomes, made up of ~3 billion nucleotides, and up to 25,000 individual genes are contained in one set of 23 chromosomes→illustrates the potential for complex genetic regulation of the human brain

c. Several clinical examples that illustrate this point: (see figure and table below illustrating genetic regulation of cortical development and function)

i) primary microcephaly—a disorder of neurogenesis

ii) classical lissencephaly—a disorder of neuron migration

iii) horizontal gaze palsy with progressive scoliosis—a disorder of axon outgrowth

iv) primary epilepsy—a disorder of circuit formation and function

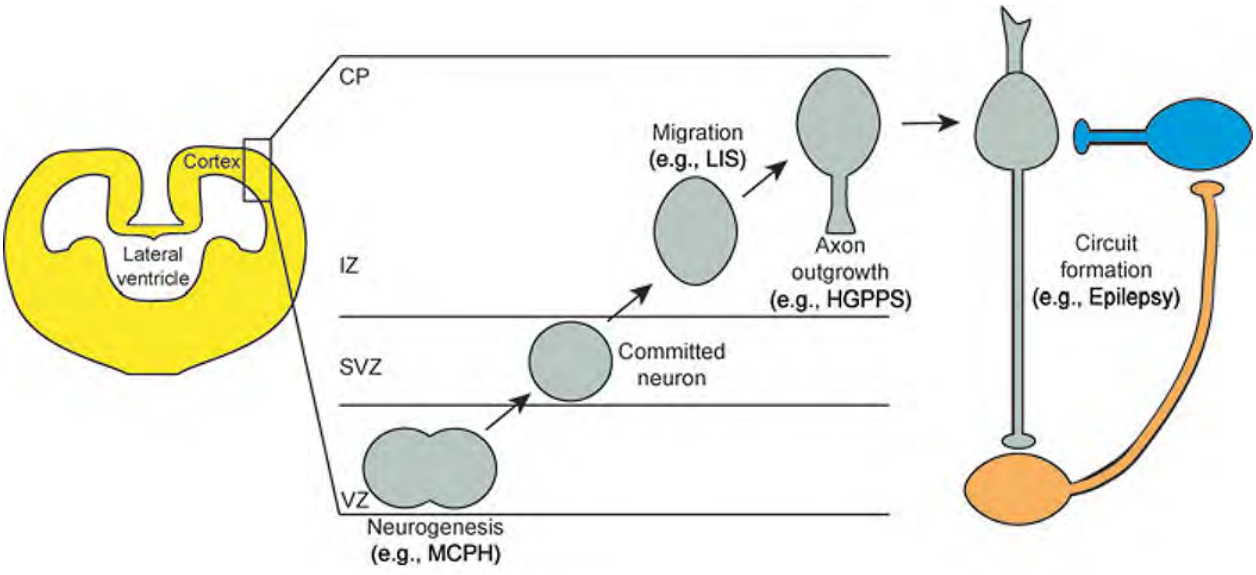


Figure of Overview of early cortical neuron development. Cortical neurons are born and undergo fate determination in the ventricular zone (VZ) and subventricular zone of the developing cortex. These neurons then migrate through the intermediate zone (IZ) and into the cortical plate (CP), where they come to reside in their specific cortical layer. Maturing neurons then send out axons, which grow outward until they reach their designated target. This process is repeated with each newly born neuron. Ultimately, neurons form many synaptic connections to create functional neural circuits. Mendelian disorders can occur at any of these developmental stages.

Table outlining genes involved in Mendelian disorders of cortical development and function (table from Dixon-Salazar TJ, Gleeson JG. Genetic regulation of human brain development: lessons from Mendelian diseases. *Ann N Y Acad Sci*. 2010;1214:156-167. doi:10.1111/j.1749-6632.2010.05819.x)

Disorder	**Gene name**	**Localization**	**Cellular function**
Neurogenesis primary microcephaly (MCPH)	*MCPH1*	Centrosome	Coordinates entry into mitosis
	ASPM	Pericentrosome	Regulates mitotic spindles
	CDK5RAP2	Centrosome	Coordinates centrosome during mitosis
	CENPJ	Centrosome	Coordinates centrosome during mitosis
	STIL	Pericentrosome	Regulates mitotic spindles
Neuron migration isolated lissencephaly sequence (ILS)	*DCX*	Microtubules	Bundles microtubules during migration
	LIS1	Microtubules	Component of dynein motor complex
	TUBA1A	Microtubules	Major constituent of microtubules
Axon outgrowth horizontal gaze palsy with progressive scoliosis (HSPPS)	*ROBO3*	Growth cone receptor	Guidance of projection axons
Congenital mirror movements (CMM)	*DCC*	Growth cone receptor	Guidance of projection axons
Circuit formation primary epilepsy	*SCN1A*	Axonal receptor	Action potential initiation
	SCN1B	Axonal receptor	Action potential initiation
	SCN2A	Axonal receptor	Action potential initiation
	GABRA1	Postsynaptic receptor	Synaptic inhibition
	GABRAG2	Postsynaptic receptor	Synaptic inhibition
	CHRNA2	Presynaptic receptor	Synaptic transmission
	CHRNB2	Presynaptic receptor	Synaptic transmission
	CHRNA4	Presynaptic receptor	Synaptic transmission
	CACNA1A	Presynaptic receptor	Synaptic transmission

Disorder	Gene name	Localization	Cellular function
	KCNQ2	Axonal receptor	Axon potential propagation
	KCNQ3	Axonal receptor	Axon potential propagation
	KCNA1	Presynaptic receptor	Synaptic transmission
	KCNMA1	Presynaptic receptor	Synaptic function

3. Recognize the importance of folate in preventing defects in neural tube closure.

a. Both genetic predisposition and environmental influences are thought to contribute to neural tube defects.
b. Clues to the genetic basis of NTDs come from chromosomal anomalies that can pinpoint critical genes within aberrant chromosomal regions, family studies that have identified single gene-related NTDs, and mouse models where more than 200 different genes are known to be essential for neural tube closure
c. the interplay, however, between these genetic and environmental influences and in how they influence the development of NTD's is still an ongoing investigation
d. an important known environmental influence in the development of NTDs, however, is the consumption of folate- folic acid is the synthetic form of folate, a water-soluble B vitamin (B9).
i) the exact mechanism of action of folate in the prevention of neural tube defects is unknown.
ii) folate acts as a coenzyme in the synthesis of nucleic acids and the metabolism of amino acids
iii) an important function of folate is its role in single-carbon transfers, which are important in methylation reactions and in purine and pyrimidine synthesis
v) folate is necessary for the regulation of DNA synthesis and function; reduced concentrations of folate may limit the number of methyl groups available for DNA replication and methylation
vi) further evidence suggests that mutation in the *MTHFR* gene, which encodes the enzyme methylenetetrahydrofolate reductase, is a risk factor for neural tube defects→this enzyme regulates folate and homocysteine levels
vii) persons who have this gene mutation have decreased folate levels, which reduces the conversion of homocysteine to methionine and may increase the risk of neural tube defects→therefore, folic acid consumption may help diminish the effects of the gene mutation
d. that being said, the majority of neural tube defects are still sporadic→genetic factors remain strongly implicated in the pathogenesis of NTD, but the usual form of inheritance is multifactorial or polygenic
e. bottom line is that maternal folate deficiency may contribute to NTD development in genetically susceptible individuals, but maternal folate level is actually a direct risk factor in an inconsequential number of cases
f. the potential benefits of reducing risk of NTD's by maternal supplementation of folic acid (at recommended dosing of 400 IU/day; and in women who have had a pregnancy affected by an NTD 4000 IU/day in month before becoming pregnant and during first 3 months of pregnancy) far outweigh any risks of said supplementation→thus why we recommend it ☺

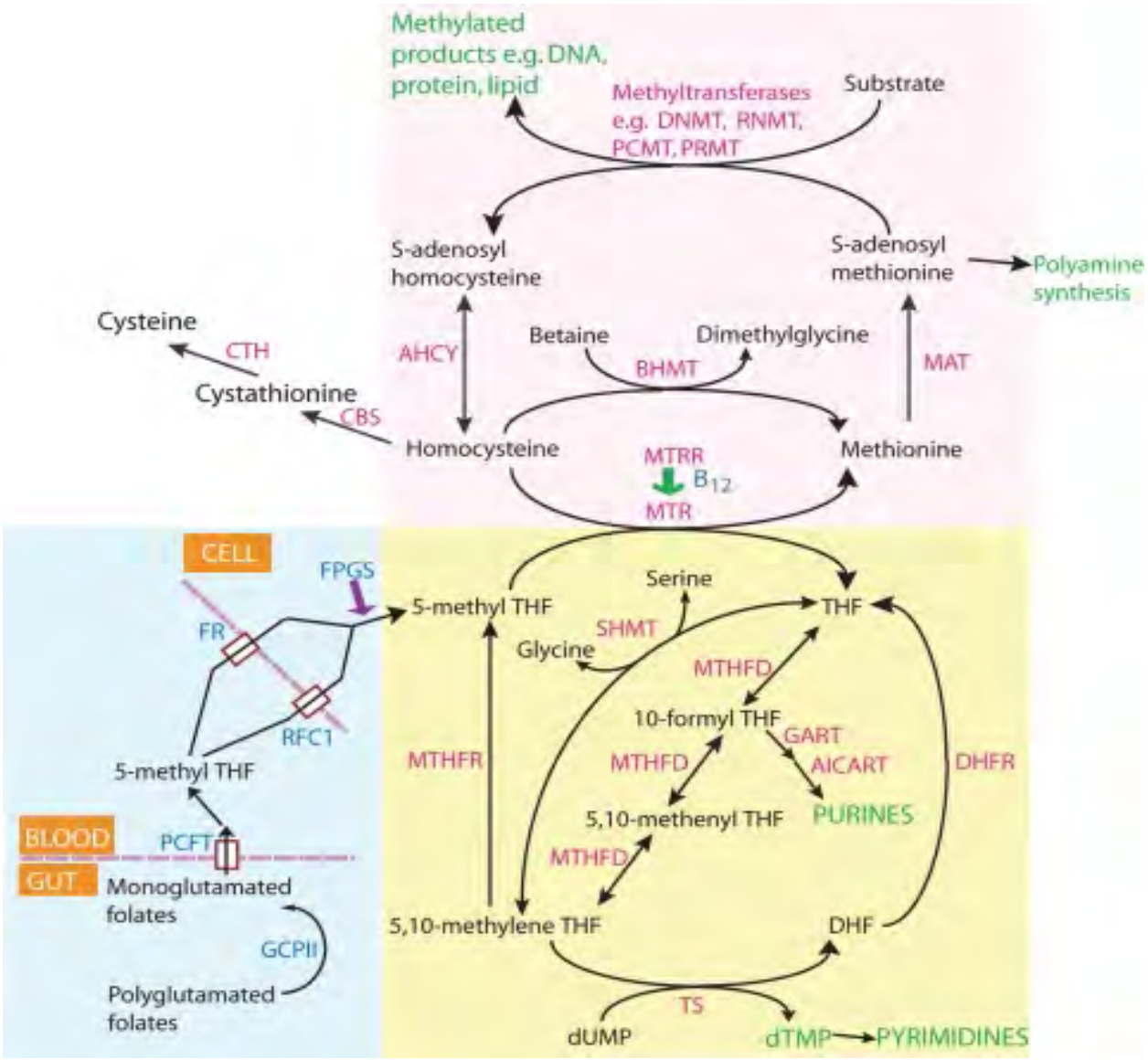


Figure above: Summary of folate one-carbon metabolism showing the main pathways and reactions that have been subject to analysis in the context of NTDs. Blue shading: proteins involved in processing of folates in the digestive tract, transport and cellular retention (by addition of glutamates). Yellow shading: the major part of the cycle involving transfer of 1C groups between folate molecules, as required for purine and pyrimidine biosynthesis. Pink shading: reactions of the methylation cycle. For clarity, mitochondrial reactions that include generation of formate and cleavage of glycine have been omitted.

4. Understand the neuroanatomic abnormalities that arise from perturbations in neuronal migration.

Table 1. Classification scheme for malformations of cortical development (MCDs)

Affected step of development	MCDs resulting from the disturbance	Short definition of the MCD
Progenitor cell proliferation and apoptosis	Microcephaly	Abnormally small head and brain
	Macrocephaly	Abnormally big head and brain
	Hemimegalencephaly	Overgrowth of (part of) a cerebral hemisphere
	Focal cortical dysplasia	Disturbed lamination and dysmorphic neurons
Neuronal migration	Lissencephaly type I	Absence of normal convolutions/folds
	Periventricular heterotopia (PH)	Neurons accumulating at the ventricles underneath a normal cortex
	Subcortical band heterotopia/double cortex	Band of grey matter located between the lateral ventricular wall and the cortex
	Cobblestone lissencephaly/lissencephaly type II	Overmigration of neurons to localize on the surface of a brain with reduced gyri
Neuronal organisation	Polymicrogyria	Too many (usually small) folds/convolutions
	Schizencephaly	Fluid-filled cleft from ventricle(s) to pia lined by heterotopic grey matter

a. Neuronal Migration Disorders can be classified into these major groups:

i) True Migration Disorders: Neurons fail to reach their intended destination

a) Lissencephaly, which literally means “smooth brain” from Ancient Greek derivation (type I lissencephaly)- group of related disorders all characterized by being the outcome of PARTIAL or DEFECTIVE NEURONAL MIGRATION or

(1) agyria (complete lissencephaly) =without gyria

(2) pachygyri= simplied gyri (associated strongly with Zellweger Syndrome)

b) Heterotopias, characterized by abnormally positioned neurons, including:

(1) subcortical band heterotopia or SBH and

(2) periventricular heterotopia-associated most commonly with afebrile seizures, but also associated with EDS, Cri du Chat, and Williams Syndrome)

c) Cobblestone malformation/lissencephaly (type II lissencephaly)- characterized by being the outcome of neuronal OVERMIGRATION →also typically associated with hypoplasia and/or malformation of the cerebellum; commonly seen in Walker-Warburg Syndrome, muscle eye brain Disease, and Fukuyama-type congenital muscular dystrophy

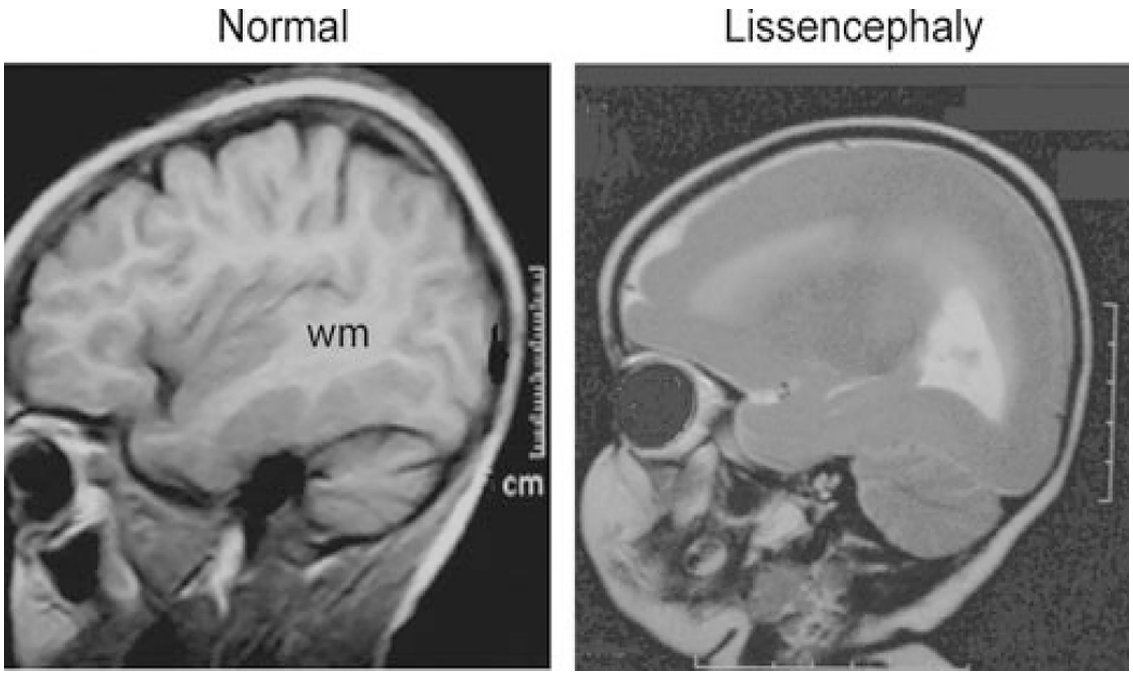


Figure above- MRI imaging comparing typically developing brain with complete lissencephaly

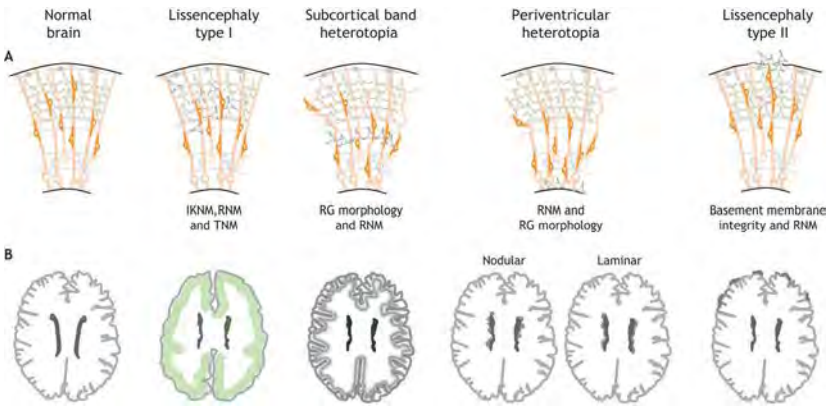


Figure above outlining the neuronal migration defect that occurs in each type of true neuronal migration disorder

ii) Malformation of cortical organization –microscopic abnormality in cortical arrangement

(1) Polymicrogyria- abnormally formed cerebral cortex that has multiple small gyri; mutations in genes controlling major molecular pathways like the phosphatidylinositol 3-kinase (PI3K/AKT) have been implicated; the severity of the symptoms is strongly related to the extent of brain involvement with the unilateral focal variant as the mildest form of this disease which has little or no symptoms and mostly controlled with antiepileptic medications→the most severe is the bilateral frontoparietal polymicrogyria with significant neurological manifestations- this

severe form gets inherited in an autosomal recessive pattern, and the defect is on chromosome 16q12-21 (polymicrogyria strongly associated with Aicardi, Delleman, DiGeorge 22q11.2 (deletion), Sturge–Weber syndromes

(2) Schizencephaly- generally classifies as a subtype of Polymicrogyria; rare brain malformation described as a split-brain or cleft that transverse the brain pia mater to the ventricles

iii) Defective cellular proliferation and differentiation of progenitor cells:

(1) Focal cortical dysplasias (FCDs)- umbrella term consisting of several subgroups of abnormal lamination of the cerebral cortex→Most common cause of medically refractory epilepsy in the pediatric population →overlap with TSC
(2) Megalencephaly – technically increased head size above two standard deviations, but clinically, it is more applicable to define it as brain size greater than three standard deviations above the mean to exclude familial megalencephaly; megalencephaly requires differentiation from **macrocephaly,** which is an unusual increase in occipitofrontal circumference (OFC) at least two standard deviations caused by structural abnormalities of the cranium, brain or cerebrospinal fluid (CSF) and related structures
(3) Hemimegaloencephaly- a one-sided cerebral hemisphere enlargement involving part of or the whole cerebral hemisphere
    a. Commonly seen in association with neurocutaneous syndromes like linear sebaceous syndrome, tuberous sclerosis, and neurofibromatosis.
    b. common presentation of this disease includes psychomotor retardation, intractable seizures, cranial nerve palsies, and hemiparesis
    c. presence of seizures in the first year of life is indicative of poor prognosis
(4) Microcephaly- referring to primary microcephaly or autosomal recessive primary microcephaly (often shortened to MCPH, which stands for "microcephaly primary hereditary")- associated with mild to moderate ID, mild dysmorphic features, shorter stature, and possibility of seizures

5. Understand that developing neurons form complex dendritic arbors with synaptic connections to many other neurons

a. Axons and dendrites are the means by neurons communicate.

b. Dendrites receive signals and are usually relatively localized but the structure and arborization of dendrites is what has a profound impact on the processing ultimately of neuronal information→the characteristic dendritic pattern of a neuron is coincident with its functional role

c. Axons can project long distances with the longest in humans extending from pyramidal neurons in layer 5 to anterior horn cells in the lumbar spinal cord

d. Axons are able to reach their correct target through axon elongation and extension that occurs at portion of the axon called the growth cone (at the tip)

e. Growth cone samples the environment for signaling molecules, some of which are “attractive” and some of which are “repulsive”→once the growth cone arrives at the target, it forms a synapse… synapses are the major sites of information input into neurons

f. this process occurs both prenatally and postnatally

g.. The pattern of dendritic arborization is critical because it determines the synaptic input field of the dendrite→distinct dendritic regions receive synaptic input from different sources

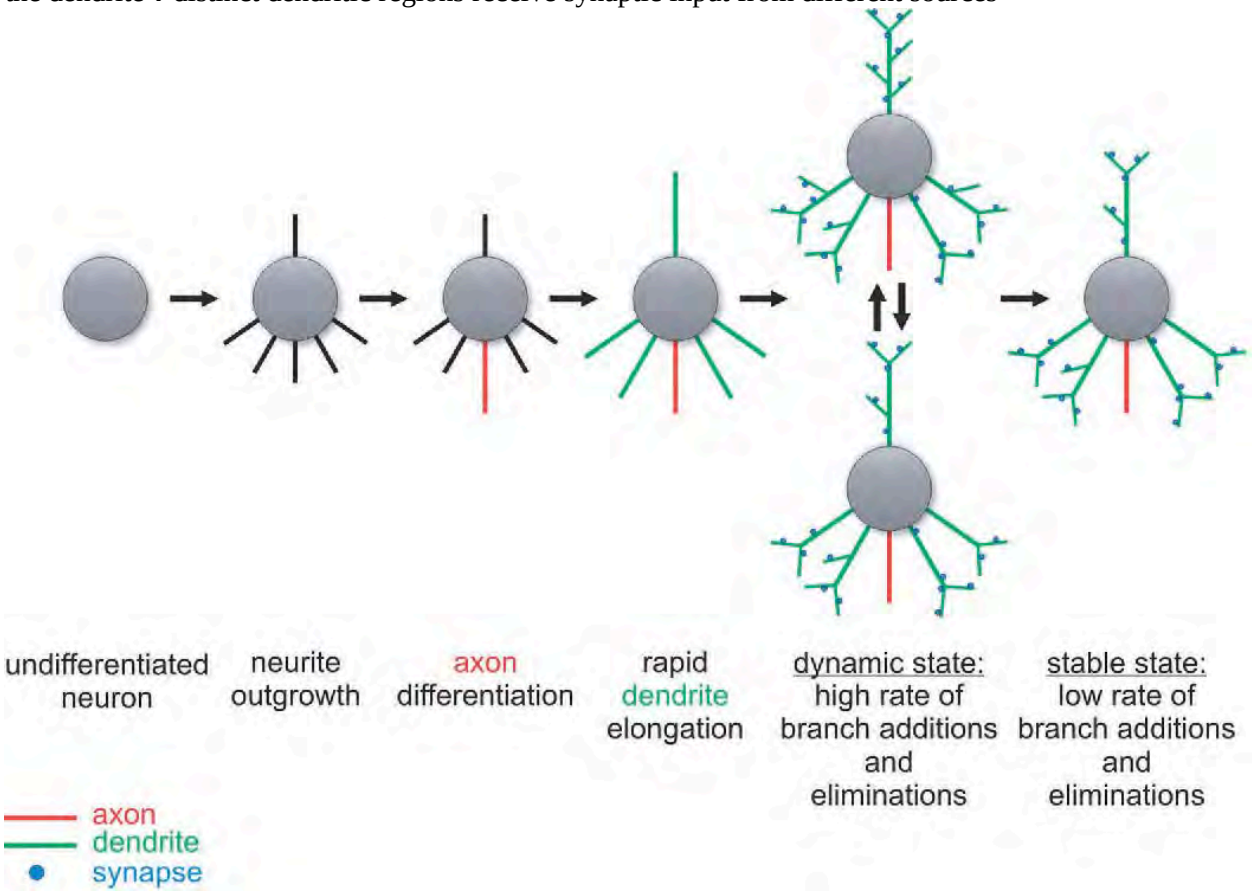


Figure above- Development of dendritic arbor consists of several overlapping stages

6. Understand the timing and the process of myelination in the central nervous system.

a. the final process involved in the development of the brain is called *myelination*

b. myelination follows a very orderly and predicable process→ generally proceeds from caudal to rostral and from posterior to anterior

i) in this process the axons of neurons are wrapped in fatty cells, which ultimately facilitates neuronal activity and communication because this insulation allows myelinated axons to transmit electrical signals faster than unmyelinated axons

ii) The timing of myelination is dependent on the region of the brain in which it occurs

iii) Regions of the brain in certain sensory and motor areas are myelinated earlier in a process that is complete around the preschool period; in contrast, regions involved in higher cognitive abilities, such as the prefrontal cortex, are not fully myelinated until adolescence or early adulthood

c. Reaches adult appearance, however, on MRI imaging in T1 by about 12 months and in T2 by about 24 months

TABLE 24.2 Sequence of Myelination Based on Histologic Analysis and Magnetic Resonance Imaging

Anatomic Region	Median Age for Detection of Myelin Histology	Age for Detection of Myelin: Magnetic Resonance Imaging	
		T1-Weighted Images	T2-Weighted Images
Ventrolateral thalamus	28–30 wk	32–34 wk	
Posterior limb of internal capsule	38–44 wk*	38–40 wk	Posterior portion 40–48 wk Anterior portion 56–70 wk
Anterior limb of the internal capsule	50–87 wk	48–53 wk	70–90 wk
Central corona radiata	37–52 wk	28–56 wk	52–65 wk
Genu corpus callosum	50–53 wk	56–64 wk	64–72 wk
Splenium corpus callosum	54–65 wk	52–56 wk	56–64 wk
Occipital white matter			
Central	47–87 wk	52–60 wk	76–96 wk
Peripheral	56–122 wk	56–70 wk	90–102 wk
Frontal white matter			
Central	50–119 wk	52–64 wk	90–106 wk
Peripheral	72–119 wk	70–90 wk	96–114 wk

*The first number corresponds to earliest identification of some myelin tubules by microscopic examination of hematoxylin and eosin stained sections. The second number corresponds to mature myelin stained with blue dye by eye observation.

From Martin RJ, Fanaroff AA, Walsh MC, eds. Fanaroff and Martin's Neonatal Perinatal Medicine. *8th ed. Philadelphia: Mosby Elsevier; 2006.*

7. Understand the influence of steroid hormones on the organization of the developing brain.

a. The brain is a target organ for the actions of hormones secreted by the gonads, adrenals, and thyroid gland, and this sensitivity to hormones begins in embryonic life with the appearance of hormone receptor sites in discrete populations of neurons

b. Because the secretion of hormones is also under control by its neural and pituitary targets, the brain-endocrine axis during development is in a delicately balanced state that can be upset in various ways

c. Thus, any agent that disrupts normal hormone secretion can upset normal brain development. Likewise, exogenous substances that mimic the actions of natural hormones can also play havoc with CNS development and differentiation

d. The brain responds to all six classes of steroid hormones (androgens, estrogens, progestins, glucocorticoids, mineralocorticoids, and vitamin D) and contains receptors for them, as well as for thyroid hormone

e. All of these hormone receptors are proteins that contain a hormone-recognizing domain and a domain that binds to specific DNA sequences →thus, these receptors exert their effects by binding to specific enhancer-like elements of the genome and modulating (increasing or decreasing) gene expression

f. because most of these receptors begin to be expressed in neurons during embryonic life, their presence allows hormones or other molecules that mimic hormone actions (pseudohormones) to affect brain development→generally speaking, such effects involve induced growth or inhibition of growth of selected groups of neurons, as well as promotion of differentiation of neurotransmitter phenotype or regulatory phenotype

g. some of the most relevant clinical examples of the possible harms of steroid exposure on the developing brain are in looking at the potential risks of administering antenatal corticosteroids to late preterm and term gestations (especially after 34 weeks)

h. A surge in endogenous cortisol occurs near term when the fetus is in a critical period of brain development in preparation for parturition and transition to life ex utero

(i) High levels of 11β-hydroxysteroid dehydrogenase-2 (11β-HSD-2) in the fetal brain help to protect it from the effects of the physiological rise in endogenous cortisol, but do not protect it from maternally administered betamethasone or dexamethasone due to the resistance of these drugs to metabolism by 11β-HSD-2

(ii) Thus, these steroids may cause overactivation of glucocorticoid receptors in the fetal brain near term

(iii) It has been proposed that exogenous steroids may have different effects at different gestational ages due to the multiple factors that change with advancing gestational age and the complex regulation of glucocorticoid receptor-mediated responses
(iv) Because the human brain grows by 35 percent, cortical volume increases by 50 percent, and 25 percent of cerebellar development occurs between 34 weeks of gestation and term exposure to exogenous betamethasone or dexamethasone during this time period is likely to have greater adverse consequences on brain development than at any other period of development
(v) In particular, disruption of the normal fetal environment at this critical time may lead to changes in development of the neuroendocrine system, life-long effects on endocrine, behavioral, emotional, and cognitive function, and increased risks for development of a wide range or metabolic, cardiovascular, and brain disorders in later life

8. Understand the effects of stress on the developing brain.

a. the brain undergoes dynamic changes throughout the course of development, with important implications for how stress influences the brain and the efficacy of treatments targeting stress-related mental illness at different developmental time points

b. Sensitive periods refer to times in development when heightened neuroplasticity renders the brain especially amenable to environmental influences

c. timing of sensitive periods differs by neural circuit and behavioral system, but it may be that sensitive periods occur when brain development is most dynamic, such as infancy and adolescence

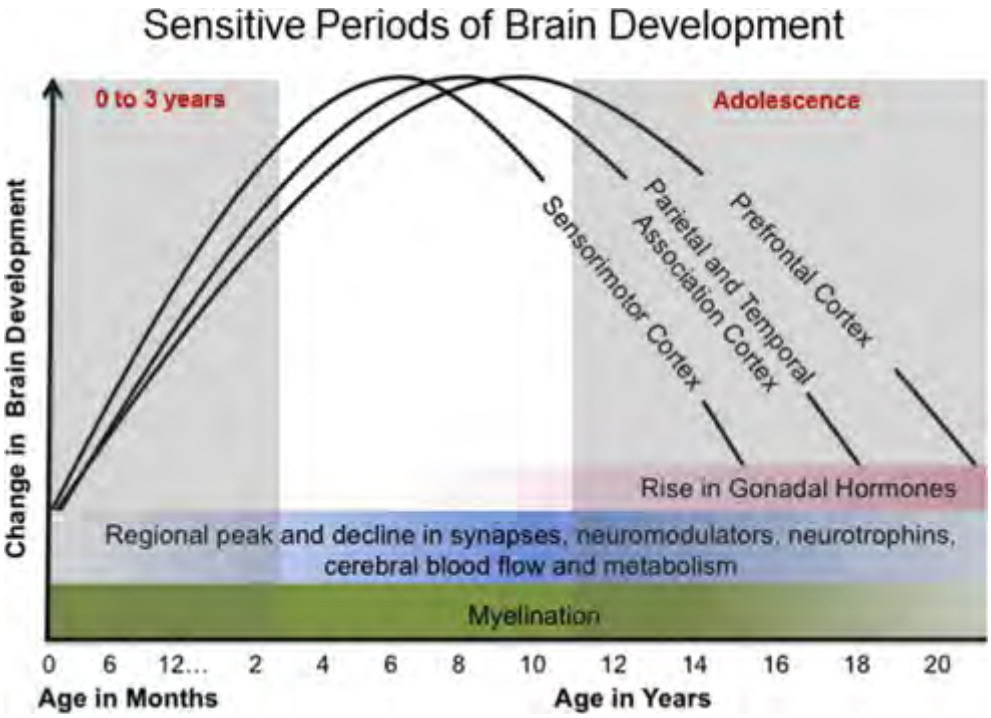
d. During these periods, environmental input can lead to a series of developmental cascades (Masten and Cicchetti, 2010) that ultimately have significant influences on behavior, of a positive *or* negative nature

e. A sensitive period may render the brain more capable of responding to stress in adaptive ways

f. It could also magnify consequences of stressful life events in maladaptive ways.

g. By contrast, stress that occurs during windows of reduced plasticity (e.g., after the closing of a sensitive period) may yield a brain that is less capable of remodeling itself.

h. Thus, sensitive periods in neurodevelopment may render the developing brain more vulnerable to the effects of later stress, but they could also serve as windows of opportunity, during which there is increased potential for positive adaptation or effective intervention (see figure below)



B. Later development of the central nervous system

1. Understand that the post-natal maturation of the central nervous system includes a subtractive process (pruning).

a. Synaptic pruning refers to the process by which extra neurons and synaptic connections are eliminated in order to increase the efficiency of neuronal transmissions

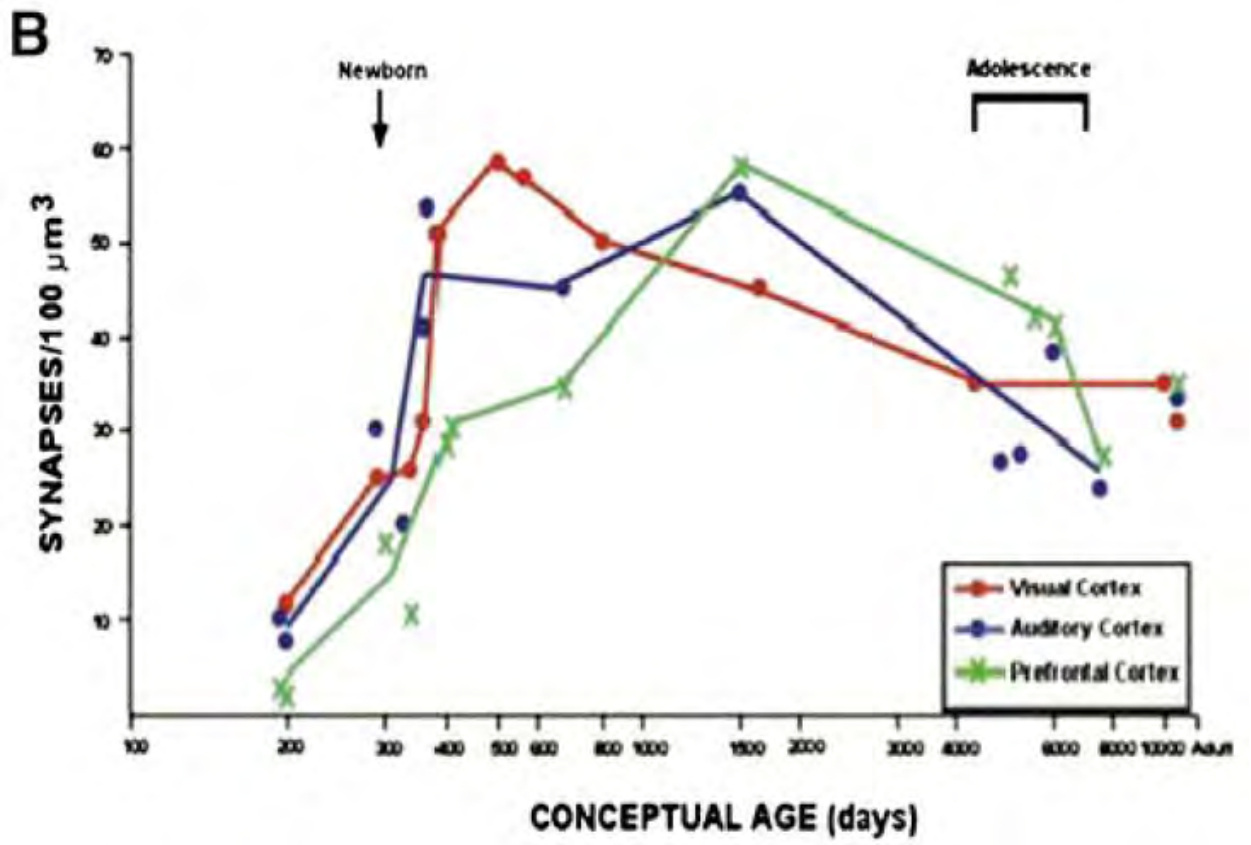
b. the overproduction of synapses in the first few years of life is followed by a pruning back of the unused and overabundance of synapses.

c. Many connections are formed- many of which are not to correct or functional locations

d. About 50% of synapses are ultimately pruned

e. Neurotrophic hypothesis—neurons that form stable synapses receive neurotrophic factors and are more likely to survive

f. Synaptic pruning is a postnatal process that extends through adolescence



Graph above illustrating how period of time of synaptic pruning varies according to region of the brain

2. Understand that the period of time that synaptic pruning is most active may vary according to region of the brain.

a. just as the time course of synaptogenesis differs across cortical regions, so does the time of synaptic pruning → sensory and motor cortices undergoing dramatic fine-tuning after birth, followed by association cortices and the corpus callosum, and later by regions that serve higher cognitive functions

b. Until the stage of synaptogenesis, the stages of brain development are largely gene driven.

c. However, once the brain reaches the point where synapses are eliminated, the balance shifts; the process of pruning is largely experience driven.

d. The elimination of axons, dendrites, and synapses, and the death of neurons through apoptosis, are important counterpart processes to the elaboration of supernumerary axons, dendrites, and synapses.

(i) Pruning processes begin in late gestation and become increasingly active postnatally

(ii) Makes sense in terms of emotional development of adolescents- prefrontal cortex is in charge of judgment,

planning, and regulation of impulse control- adolescents are more likely to react based on their emotions as supposed to reasoning through the situation

(iii) Likewise, the connections between centers for reasoning and centers for emotion (i.e. amygdala) are also still actively developing in adolescence- reflected in adolescent's emotional responses being less tempered by reasoning

e. In the primary visual cortex→after a burst of synapse formation between age 3 and 4 months, synaptic density reaches its peak at 140–150% of adult levels between the ages of 4 and 12 months, after which the mean number of synapses per neuron decline and pruning is complete b/t 4^{th}-6^{th} year of life

f. In comparison, synaptogenesis in the prefrontal cortex begins about the same time as in visual cortex, but it does not reach its peak period until age 8 months, continuing thereafter through the second year of life→pruning in PFC continues through adolescence

g. The processes of overproduction of synapses and subsequent synaptic reduction are essential for the flexibility required for the adaptive capabilities of the developing mind

(i) It allows the individual to respond to the unique environment in which he or she is born. Those pathways that are activated by the environment are strengthened while the ones that go unused are eliminated.

(ii) In this way, the networks of neurons involved in the development of behavior are fine-tuned and modified as needed

3. Realize that synaptic pruning often results in loss of cortical plasticity.
a. synaptic pruning and its impact on the loss of cortical plasticity is seen most significantly in instances of cortical injury during critical periods of time when synaptic pruning is occurring and how this can emphasize the impact of a cortical injury

b. this would be in comparison with a cortical injury that occurred during a time of neuronal proliferation where the brain can take recruit from this proliferation to aid in functional recovery of the injured area

4. Know how molecular processes and synaptic activity reflect learning at a cellular level

a. learning and memory require the formation of new neural networks in the brain and a key mechanism underlying this process is synaptic plasticity at excitatory synapses, which connect neurons into networks

b. excitatory synaptic transmission happens when glutamate, the excitatory neurotransmitter, activates receptors on the postsynaptic neuron

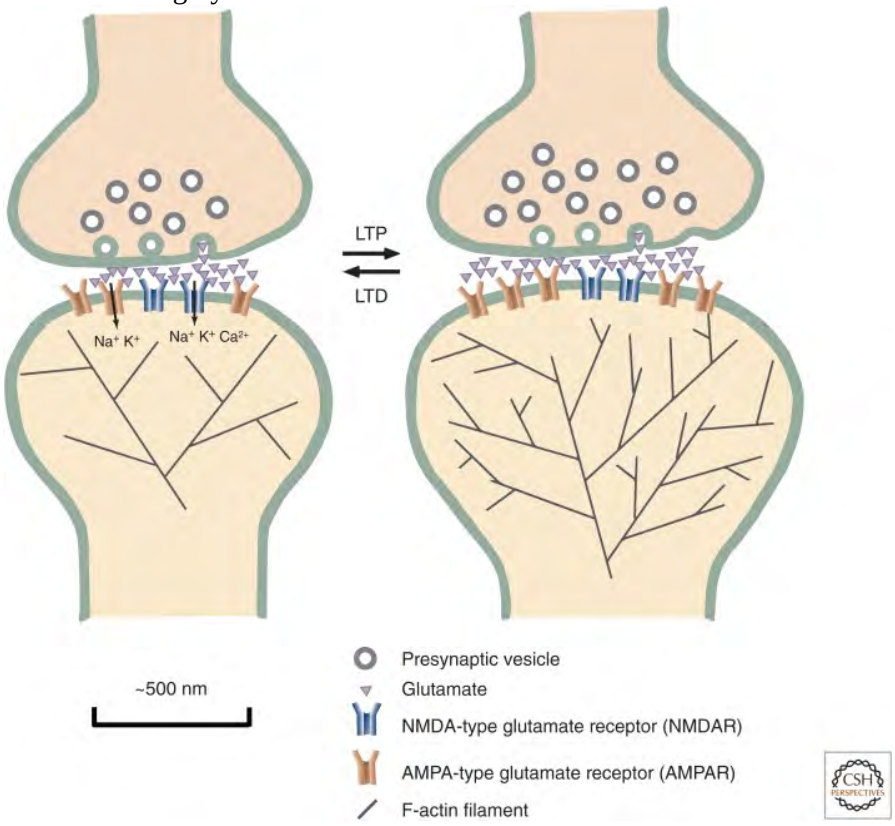
c. synaptic plasticity is a higher-level process in which the strength of excitatory synapses is altered in response to the pattern of activity at the synapse

(i) at the cellular level, one of the most essential elements of memory formation is the adjustment in synaptic strength of excitatory synapses between neurons

(ii) it is initiated in the postsynaptic compartment, where the precise pattern of influx of calcium through activated glutamate receptors leads either to the addition of new receptors and enlargement of the synapse (long-term potentiation=LTP) or the removal of receptors and shrinkage of the synapse (long-term depression=LTD)

(iii) AMPA-type glutamate receptors (yellow in the figure below) allow passage of sodium and potassium through their channel. Their principal function is to depolarize the membrane, producing an excitatory postsynaptic potential (EPSP). NMDA-type glutamate receptors (blue in the figure below) also depolarize the membrane, but in addition to sodium and potassium, calcium flows through their channel and can initiate synaptic plasticity

(iv) Calcium/calmodulin-regulated enzymes and small GTPases collaborate to control this highly tuned mechanism.



5. Know the definition of "plasticity" and the neurobiological mechanisms that are believed to underlie plasticity

a. Neuroplasticity is the biological capacity of the nervous system to modify its structure and functioning to adapt to both physiological and pathological variations in the environment

(i) Its main physiological consequences are learning and memory, and its pathological outcome is neurological rehabilitation→The continuous change and initial fragility of the

developing brain make the embryonic and fetal periods especially plastic (what is known as developmental neuroplasticity).

(ii) The progressive reduction in plasticity, however, is never complete and the capacity to modify the brain circuits in response to new learning (adaptive neuroplasticity) or brain injuries (reactive neuroplasticity) remains throughout the individual's entire lifespan

(1) Highlights important feature of plasticity that is found in the cells lining the subventricular zone of the lateral ventricles and cells in the hilus of the dentate gyrus
(2) Both regions contain stem cells that remain active throughout life. The cells in the subventricular zone produce both glial and neural progenitor cells that can migrate into cerebral grey or white matter, even in adulthood. In humans, the subventricular zone cells appear mostly quiescent but can become activated, largely in response to cerebral perturbations. Stem cells in the dentate gyrus generate new neurons at a slow but steady pace throughout life in humans, although there is a decline with aging. The functional role of these cells is not totally understood, but they do integrate with the existing neurons and likely play a role in the formation of new memories

(iii) **f**rom a neurobiological perspective, neurogenesis, synaptogenesis and synaptic pruning represent the building blocks for CNS plasticity

(iv) these biological processes are subjected to genetically programmed, time-limited periods, called the critical or sensitive periods during which the brain is most amenable to change

(v) Neurogenesis is most prominent during early fetal development→this is followed by robust synaptogenesis that starts as early as 27 weeks post gestational age and intensifies over the first 2 years of life, depending on area of the cortex

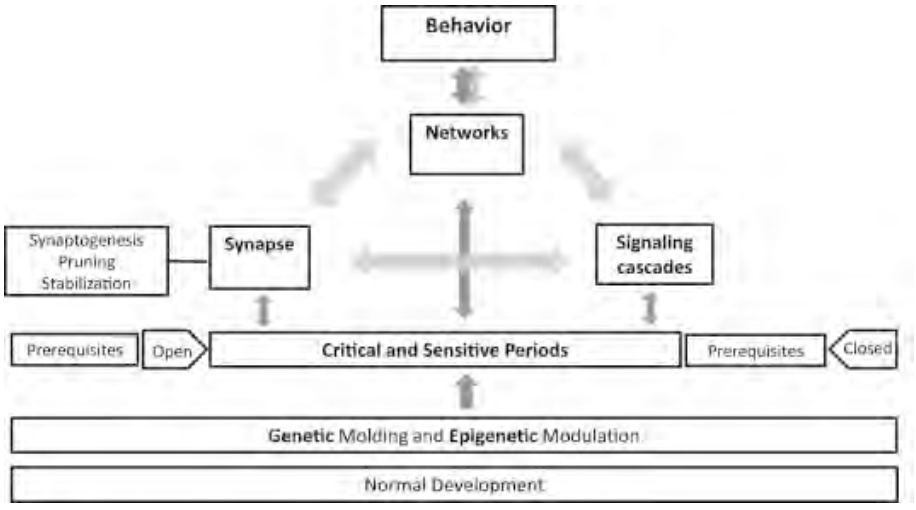


Figure illustrating the time-sensitivity and multi-level integration of neuroplasticity in the developing brain

b. Pathological states that disturb normal developmental homeostasis and/or induce aberrant developmental neuroplasticity and lead to an abnormal neurophysiological and behavioral phenotype illustrate the complex relationship between different levels of the developing nervous system that are subjected to plasticity

(i) Distinct abnormal “plasticity patterns” have been implicated in many neurological disorders of childhood

(ii) for example, abnormal dendritic spine structural plasticity has been implicated in Rett syndrome, intellectual disabilities and epilepsy

(iii) Disorders of over- or under-pruning of the synapse have been described as basic etiologies for certain neurobehavioral disorders such adolescence-onset schizophrenia b. Pathological states that disturb normal developmental homeostasis and/or induce aberrant developmental neuroplasticity and lead to an abnormal neurophysiological and behavioral phenotype illustrate the complex relationship between different levels of the developing nervous system that are subjected to plasticity

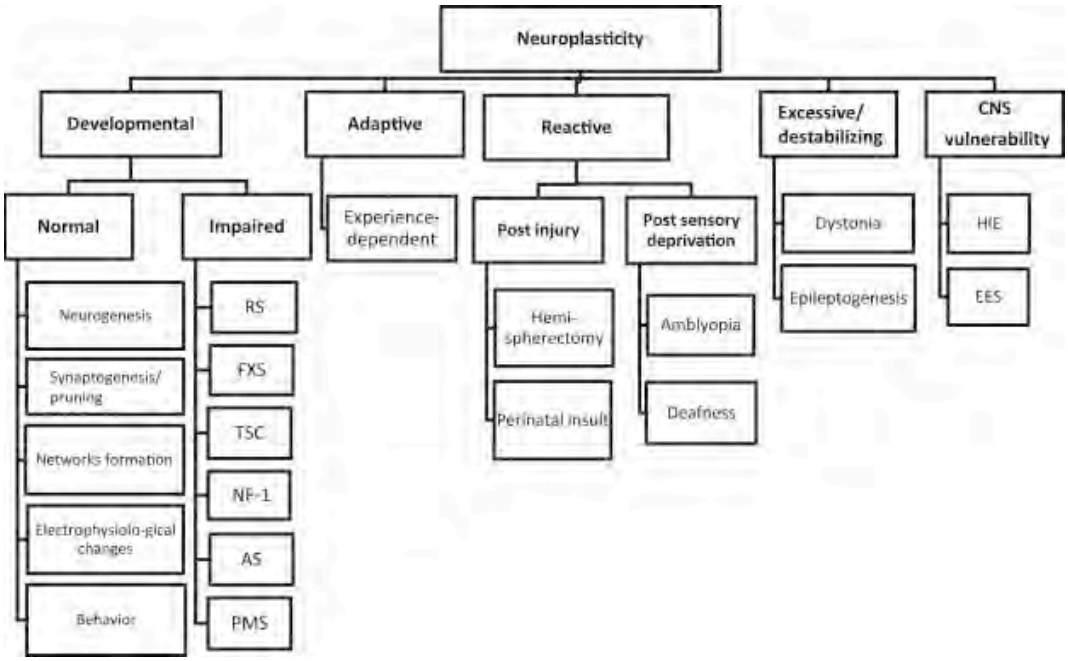


Figure showing Patterns of neuroplasticity in the developing brain- both normal and impaired. RS: Rett Syndrome; FXS: Fragile X Syndrome, TSC: Tuberous Sclerosis Syndrome; NF-1: Neurofibromatosis Syndrome; AS: Angelman Syndrome; PMS: Phelan-McDermid Syndrome. HIE: Hypoxic Ischemic Encephalopathy; EES: Epileptic Encephalopathy Syndromes.

Disorder/gene defect	Impaired plasticity mechanisms	Excitatory/Inhibitory imbalance	Clinical cortical electrophysiology and plasticity by TMS
RS/MeCP2	Abnormal homeostatic synaptic scaling	↑↑ Glutamate and NMDAR density, [26] impaired LTP, absent LTD [25]	↓ CMCT [29]
FXS/FMR1	Delayed maturation and stabilization of dendritic plasticity and increased spine density. [31]	Impaired Late-phase LTP and dysregulation of mGluR- dependent LTD, [33] downregulation of GAB-A receptors [35]	Impaired LTP/LTD-like plasticity after TBS. [36] Reduced/absent GABA-A mediated parameters in asymptomatic carriers [37]
TSC/TSC 1 and 2	Defective dendritic spine pruning and increase spine density [43]	Abnormally increased LTP and impaired mGluR-mediated LTD [43]	No available data *
AS/UBE3A	Restricted dendritic spines growth and maturation and reduced synaptic density	Impaired AMPA-mediated plasticity, [50] upregulation of mGLuRS-mediated LTD [51]	No available data *
PMS/SHANK3	Decrease membrane expression of AMPA and NMDA receptors [40]	Impaired activity-dependent LTP [40]	No available data *
NF-1	Reduced spine density, [63] Abnormal morphology of synapses [64]	Abnormal increase in presynaptic GABA release of inhibitory neurons leading to impaired LTP [65]	Abnormal GABA mediated inhibition during motor learning [67]
DYT1/TOR1A	Defective cerebellar synaptogenesis [111]	Abnormally increased LTP and failure to induce LTD [114]	Cortical hyperexcitability, Reduced ICI, SP and RI. [120] Exaggerated LTP [122] and LTD responses [121]

Abnormal developmental plasticity in selected genetic disorders: from genes to cortical electrophysiology. RS: Rett Syndrome; FXS: Fragile X Syndrome, TSC: Tuberous Sclerosis Syndrome; AS: Angelman Syndrome; PMS: Phelan-McDermid Syndrome; NF-1: Neurofibromatosis Syndrome; DYT1: Early onset generalized torsion dystonia. LTP: long term potentiation; LTD: long term depression; CMCT: Central motor conduction time; TBS: Theta Burst Stimulation; ICI: Intracortical Inhibition; SP: Silent Period; RI: Reciprocal inhibition.

6. Know the effect of environmental enrichment on neurobiological development

a. In general, three types of plasticity can be distinguished in the developing brain: experience-independent, experience-expectant, and experience-dependent

(i) experience-independent plasticity results from the fact that the genome generates a rough approximation of connectivity that is modified by both internal and external events→neurons that are active together increase their connections, whereas those that are not coincidentally active weaken their connections

(ii) Experience-expectant plasticity occurs mostly during early postnatal development (example of an infant and language→ Early in life, infants can discriminate the speech sounds of all languages, but over the first year the auditory system begins to change such that the infant becomes expert in discriminating sounds in its language environment but loses the ability to discriminate sounds that are not experienced)

(iii) experience-dependent plasticity, a process whereby the connections of ensembles of neurons are modified by experience, begins in early postnatal life and continues for a lifetime→it is here that we see the effects of so-called "environmentally enriched" experiences

c. foundation of studies on enriched environments seen in the work of Donald Hebb (1949), who is credited with discovering the connection between enriched environments and improvements in cognition and behavior in animal models

(i) Hebb noticed that the animals he occasionally brought home for his children to play with performed the best in later behavioral tasks→replicated in other research with enrichment of the housing environments of lab animals

d. Rosenzweig et al. (1978) defined environmental enrichment as "a combination of complex inanimate and social stimulation"

(i) Rosenzweig determined that social grouping of rats was not sufficient to produce enhancements in cognition→availability of inanimate objects along with a social environment as well as a living space to allow for more physical activity was what promoted "better" cognitive functioning overall

e. majority of studies looking at environmental enrichment (EE) and its impact on neurobiological development have been done in animal models for obvious ethical reasons, but parallels b/t these models and humans grows in strength and further extension has been applied to human research in part due to growing interest in its potential therapeutic benefits for children with neurodevelopmental disorders

(i) animal studies looking at sensory deprivation (namely in the visual cortex) and how it can inhibit the development of sensory systems and prolong typical temporal windows of experience-expectant plasticity→provision of EE can compensate for/counteract some of these effects (i.e. studies looking at rats raised in dark environments when given standard housing vs enriched surroundings)

(ii) other animal studies looking at maternal separation and anxiety and "rescuing" neural plasticity and amelioration of its effects on the growth and eventual size of the basolateral surface of the amygdala

g. Human studies indicating similar findings (albeit with shortcomings involving the inability to show changes at anatomic or molecular levels):
    - (i) Bucharest Early Intervention Project (BEIP) (2007)- longitudinal study looking at how institutionalization at a young age leads to severe consequences in the development of both brain and behavior
    - (ii) Study followed three groups of children:
        - (1) an institutionalized group, children who have lived virtually all their lives in an institutional setting in Bucharest, Romania;
        - (2) a foster care group, which includes children who were institutionalized at birth and then placed in foster care (at a mean age of placement of 22 months); and a
        - (3) never institutionalized group, which includes children living with their biological families in the Bucharest region
    - (iii) Study indicated that the institutionalized children showed stunted and delayed patterns of cognitive and physical growth, as well as different patterns of brain activity when compared to children who had never been institutionalized
    - (iv) Timing also played an effect as children placed in foster care <2 yo showed patterns of brain activity that were more in line with children

who had never been institutionalized; same was found for cognitive and language scores

(v) While the Bucharest study indicates that deprivation can lead to negative outcomes in terms of development, it still does not give a standardized definition of what a standardized enriched environment should look like compared to the given "norm" in developed societies

C. Functional organization of the central nervous system

1. Understand that the cortex is highly interconnected, facilitating interactions between parts of the brain that are engaged in different types of information processing.

a. One of the most prominent features of the human brain is the size of the cerebral cortex and its intricate folding

b. Cortical folding (process called corticalization) takes place during embryonic development and is important to optimize the functional organization and wiring of the brain, as well as to allow fitting a large cortex in a limited cranial volume

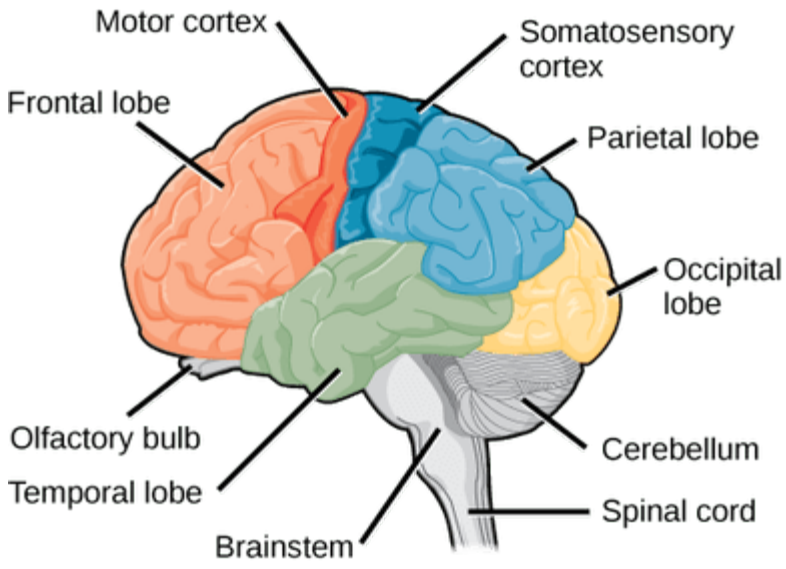
c. the complexity and interconnected nature of the cortex is illustrated in reviewing the parts of the cortex and how they are engage in different types of information processing (see figures below)

i) the **primary sensory areas** receive somesthetic, auditory, visual, and gustatory stimuli from the thalamus, which receives stimuli from specialized sensory organs and peripheral receptors; Olfactory pathways bypass the thalamus and go directly to specialized areas of the cortex; Sensory stimuli are further processed in association areas that relate to one or more senses.

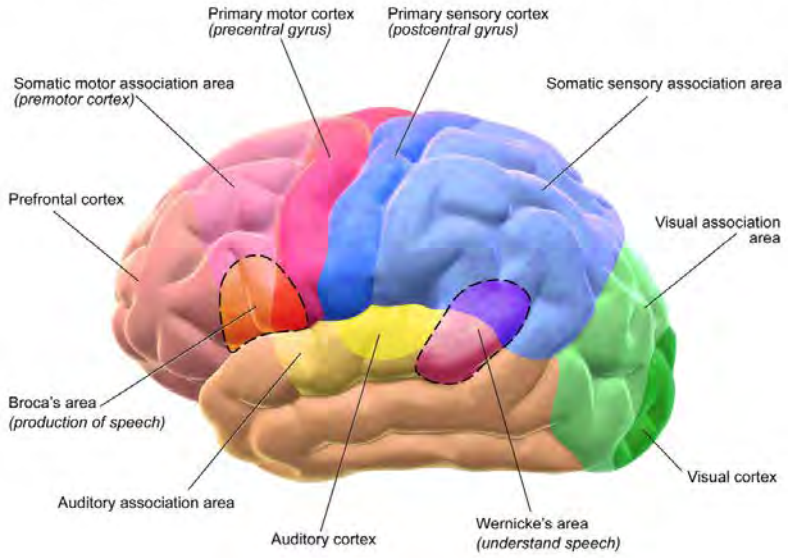
ii) The **primary motor cortex** generates volitional body movements; motor association areas help plan and execute complex motor activity

iii) Each **unimodal association area** is adjacent to its corresponding primary sensory area and processes information from that area at a higher level than the primary sensory area

iv) **Heteromodal association areas** are not restricted to any single motor or sensory function but receive convergent information from multiple sensory and motor areas of the brain; heteromodal association areas in the frontal, temporal, and parietal lobes integrate sensory data, motor feedback, and other information with instinctual and acquired memories. This integration facilitates learning and creates thought, expression, and behavior.



Motor and Sensory Regions of the Cerebral Cortex



d. the motor cortex is responsible for planning, controlling and executing voluntary movements. Moreover, the associative cortex integrates generated visual, auditory, gustatory and other general sensory signals

e. sensory cortex is defined as all cortical areas linked with sensory functions or in another definition, the sensory cortex is a section of the cerebral cortex which is responsible for receiving and interpreting sensory information from different parts of the body

(i) Stimuli received from different receptors such as nociceptors and thermoreceptors are transduced to an action potential which is conveyed along one or more afferent neuron to a specific section of the brain→the sensory cortex is comprised of the visual cortex, auditory cortex, the primary olfactory cortex, the gustatory cortex and the primary somatosensory cortex

f. The visual cortex – part of sensory cortex found in the occipital lobe; Furthermore, the occipital lobe is one of the four primary lobes of the human brain and it acts as the visual processing center

(i) For the visual cortex to respond, visual information from the eyes passes through the lateral geniculate nucleus found in the thalamus. The section of the visual cortex **that** receives sensory input from the thalamus is called the primary visual cortex, also referred to as visual area 1 (V1) or striate cortex

(ii) Both the right and left hemispheres of the human brain contain the visual cortex

(iii) the visual cortex found in the left hemisphere receives radiations from the right visual field whereas the visual cortex in the right hemisphere receives radiations from the left field of vision

(iv) therefore, the visual cortex is responsible for processing visual information. Visual nerves run from eye to primary visual cortex, then to the visual association cortex

g, the auditory cortex is positioned on the upper side of the temporal lobe

(i) Its main role is to process auditory information (1).

(ii) In humans, the temporal lobe processes sensory input to clear meanings which facilitates language comprehension, visual memory and emotion association.

(iii) The auditory cortex is one part of the auditory system that does common and higher roles in hearing like language switching.

(iv) the auditory cortex is comprised of sections belongs to the transverse temporal gyri and the superior temporal gyrus

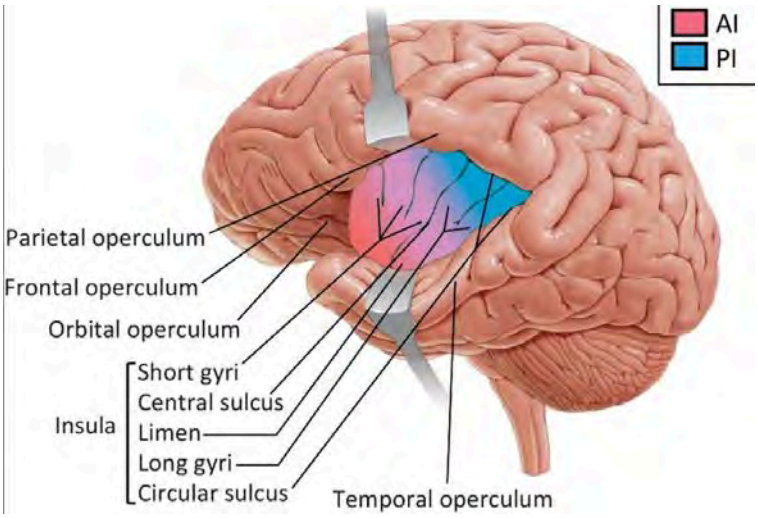
(v)Transverse temporal gyri- Also known as Heschl's gyri, these are gyri located in the primary auditory cortex. It is found in varying numbers in the right and left hemispheres →transverse temporal gyri are the first cortical structures to process incoming auditory information

(vi) superior temporal gyrus' main function is to process sounds; furthermore, some sections of the superior temporal gyrus have been designed to process the combination of frequencies and others are designed to process changes in amplitude or frequency→The superior temporal gyrus is also part of the Wernicke's area, found in the left hemisphere; It is involved in language comprehension and social cognition.

h. gustatory Cortex- part of sensory cortex for taste perception; Neurons in the gustatory cortex respond to sourness, sweetness, saltiness, and bitterness and also code the intenseness of the taste stimulus

(i) The gustatory cortex is made up of 2 substructures namely 1) The anterior insula and 2) The frontal operculum

1. Frontal operculum (FO)- it is found in the frontal lobe which is located on the front part of the brain just directly behind the forehead.
2. Anterior insula (AI)- it is found on the insular lobe which is located deep within the cerebral cortex, beneath the frontal-parietal and temporal lobes



h. Olfactory cortex- portions of the cerebral cortex that receive direct projections from the olfactory bulb

(i) Gustatory and olfactory systems work hand-in-hand and are a time called the chemosensory system since they provide the brain with information on the chemical composition of items

(ii) The olfactory cortex is housed in the uncus and relies on communication of sensory input between the piriform cortex, amygdala, olfactory tubercle, and parahippocampal gyrus

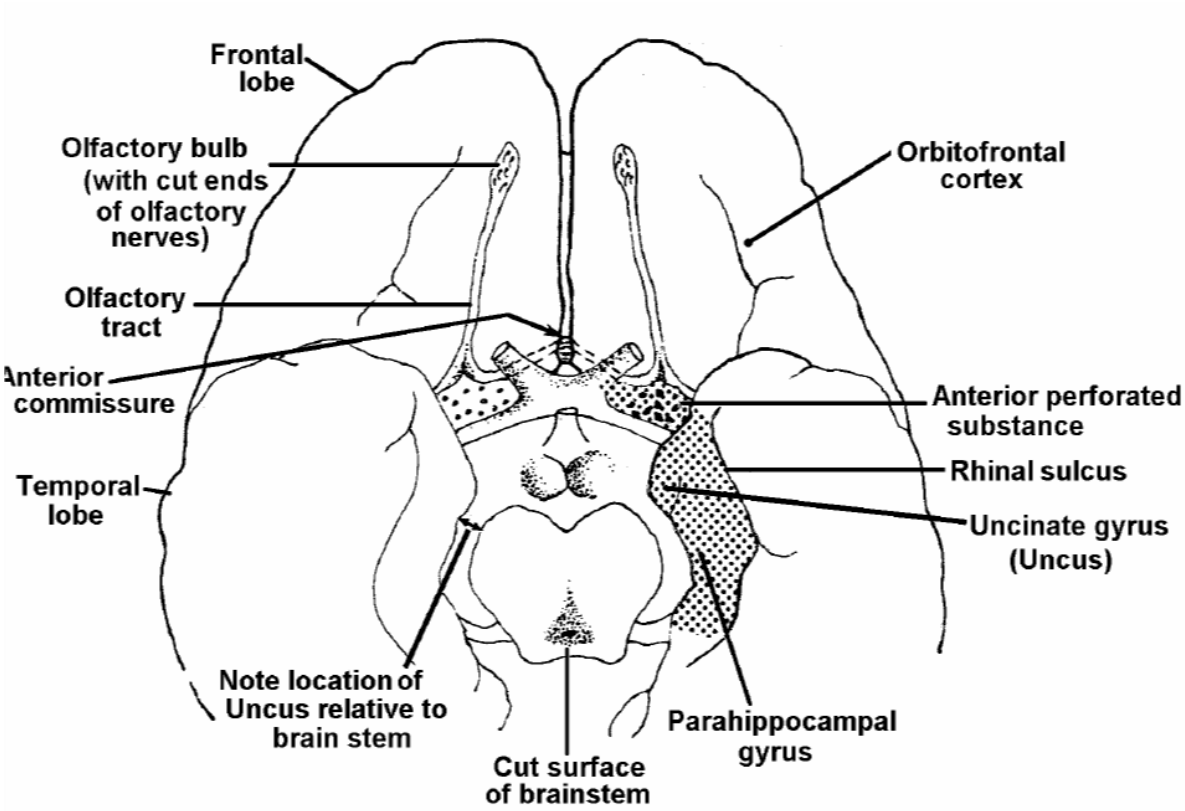


FIG. 2: Ventral view of brain illustrating olfactory structures. Three-layered olfactory cortex has been indicated by stippling. Note the anterior perforated substance where the striate arteries enter the brain.

i. somatosensory cortex of the human brain consists of: Brodmann areas 3, 1 and 2. Brodmann area, a section of the cerebral cortex (named after a famous German neurologist called Korbinian Brodmann)

(i) Brodmann area (BA) 3 is made up of two areas; 3a and 3b

(ii) somatosensory cortex is housed in an area just behind and parallel to the motor cortex at the back of the frontal lobe, in the postcentral gyrus

(iii) The primary somatosensory cortex is commonly referred to as BA 3 because neurons in this area of the cortex are very sensitive primarily to somatosensory stimuli and receive the bulk of the somatosensory input from the thalamus

(iv) in the primary somatosensory cortex, tactile representation is orderly arranged (in an inverted fashion) from the toe (at the top of the cerebral hemisphere) to mouth (at the bottom). However, some body parts may be controlled by partially overlapping regions of cortex→ organized somatotopically, having the pattern of a homunculus, similar to the motor cortex; somatosensory cortex also illustrates principles of contralateral control as in motor cortex and other areas of sensory cortex

2. Realize that the brain systems responsible for a given function may be different in children than in adults.

a. see previous sections discussing synaptic pruning, neuroplasticity, and impact of experience on the developing brain

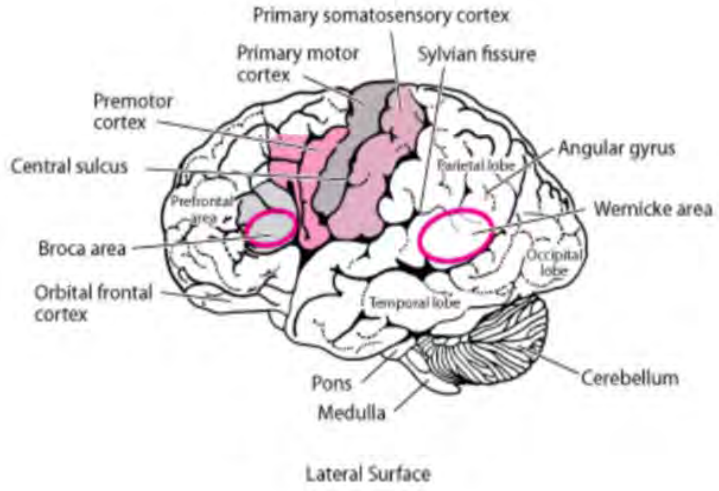
b. despite age-related differences in function, there have been multiple studies and recent meta-analyses looking at studies comparing child and adult brains using fMRI imaging to assess commonalities and many have been found regardless of age in performance of many activities (2016 study)

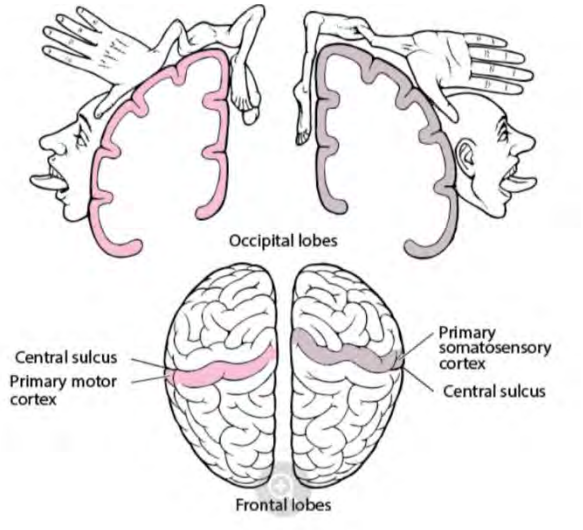
3. Know the range of functions that are served by the frontal lobes

The frontal lobes are anterior to the central sulcus. They are essential for planning and executing learned and purposeful behaviors; they are also the site of many inhibitory functions. There are several functionally distinct areas in the frontal lobes:

a. The primary motor cortex is the most posterior part of the precentral gyrus. The primary motor cortex on one side controls all moving parts on the contralateral side of the body (shown on a spatial map called a homunculus- 90% of motor fibers from each hemisphere cross the midline in the brain stem; thus, damage to the motor cortex of one hemisphere causes weakness or paralysis mainly on the contralateral side of the body.
b. The medial frontal cortex (sometimes called the medial prefrontal area) is important in arousal and motivation. If lesions in this area are large and extend to the most anterior part of the cortex (frontal pole), patients sometimes become abulic (apathetic, inattentive, and markedly slow to respond).
c. The orbital frontal cortex (sometimes called the orbital prefrontal area) helps modulate social behavior
    - patients with orbital frontal lesions can become emotionally labile, indifferent to the implications of their actions, or both (Phineas Gage)
    - They may be alternately euphoric, facetious, vulgar, and indifferent to social nuances
    - bilateral acute trauma to this area may make patients boisterously talkative, restless, and socially intrusive
    - the disinhibition and abnormal behaviors that can occur with aging and in many types of dementia probably result from degeneration of the frontal lobe, particularly the orbital frontal cortex.
d. The left posteroinferior frontal cortex (sometimes called the Broca area or posteroinferior prefrontal area) controls expressive language function. Lesions in this area cause expressive aphasia (impaired expression of words).
e. The dorsolateral frontal cortex (sometimes called the dorsolateral prefrontal area) manipulates very recently acquired information—a function called working memory
    - Lesions in this area can impair the ability to retain information and process it in real time (e.g., to spell words backwards or to alternate between letters and numbers

sequentially)





Homunculus

3/3

Specific parts of the cortex control specific motor and sensory functions on the contralateral side of the body. The amount of cortical space given to a body part varies; eg, the area of the cortex that controls the hand is larger than the area that controls the shoulder. The map of these parts is called the homunculus ("little person").

3. Know the range of functions that are served by the parietal lobes

Several areas in the parietal lobes have specific functions.

a. The primary somatosensory cortex

i. located in the postrolandic area (postcentral gyrus) in the anterior parietal lobes, integrates somesthetic stimuli for recognition and recall of form, texture, and weigh
ii. The primary somatosensory cortex on one side receives all somatosensory input from the contralateral side of the body (see homunculus figure)
iii. Lesions of the anterior parietal lobe can cause difficulty recognizing objects by touch (astereognosis).

b. Areas posterolateral to the postcentral gyrus generate visual-spatial relationships and integrate these perceptions with other sensations to create awareness of trajectories of moving objects→These areas also mediate proprioception (awareness of the position of body parts in space).
c. Parts of the midparietal lobe of the dominant hemisphere are involved in abilities such as calculation, writing, left-right orientation, and finger recognition; lesions in the angular gyrus can cause deficits in writing, calculating, left-right disorientation, and finger-naming (Gerstmann syndrome).
d. The nondominant parietal lobe integrates the contralateral side of the body with its environment, enabling people to be aware of this environmental space, and is important for abilities such as drawing
i. acute injury to the nondominant parietal lobe may cause neglect of the contralateral side (usually the left), resulting in decreased awareness of that part of the body, its environment, and any associated injury to that side (anosognosia)
ii. patients with large right parietal lesions may deny the existence of left-sided paralysis
iii. patients with smaller lesions may lose the ability to do learned motor tasks (eg, dressing, other well-learned activities)—a spatial-manual deficit called apraxia.

4. Know the range of functions that are served by the temporal lobes

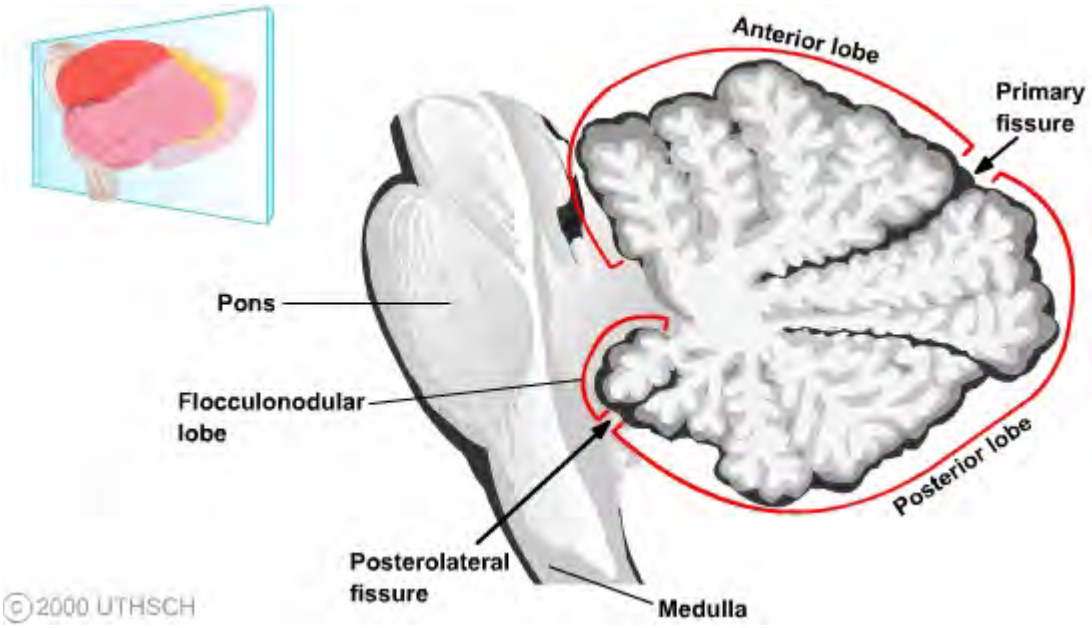
a. The temporal lobes are integral to auditory perception, receptive components of language, visual memory, declarative (factual) memory, and emotion
b. Patients with right temporal lobe lesions commonly lose the ability to interpret nonverbal auditory stimuli (eg, music). Left temporal lobe lesions interfere greatly with the recognition, memory, and formation of language.
5. Know the range of functions that are served by the occipital lobes
a. The occipital lobes contain the primary visual cortex and visual association areas
b. role of this lobe is visual processing and interpretation
c. Typically based on the function and structure, the visual cortex is divided into five areas (v1-v5)
d. The primary visual cortex (v1, BA 17) is the first area that receives the visual information from the thalamus, and its located around the calcarine sulcus
a. The visual cortex receives, processes, and interprets visual information and then this processed information is sent to the other regions of the brain to be further analyzed (example: inferior temporal lobe)
b. this visual information helps us to determine, recognize, and compare the objects to each other

e. Lesions in the primary visual cortex lead to a form of cortical blindness; in one form, called Anton syndrome, patients become unable to recognize objects by sight and are generally unaware of their deficits, often confabulating descriptions of what they see
f. Seizures involving the occipital lobe can cause visual hallucinations, often consisting of lines or meshes of color superimposed on the contralateral visual field

6. Know the range of functions that are served by the cerebellum

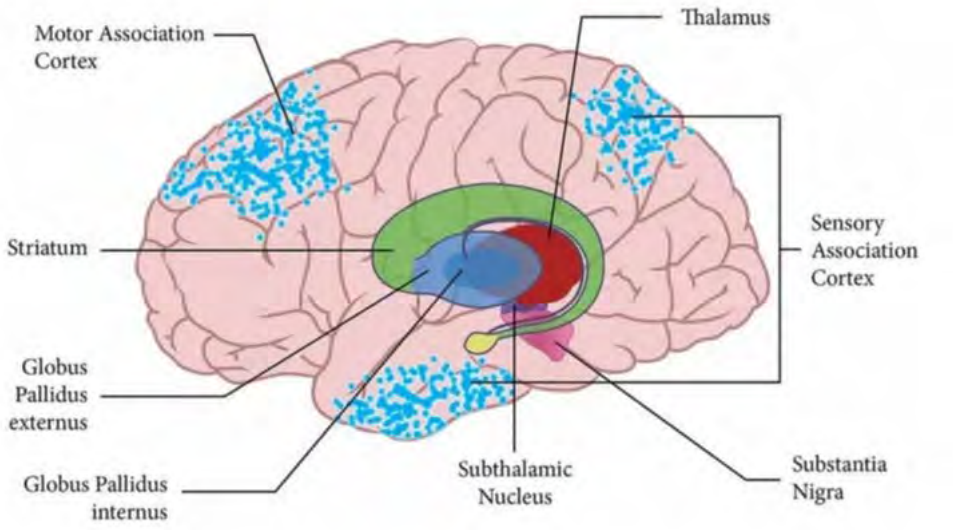
    a. 3 Functional regions of the cerebellum that correspond to its anatomical divisions:

- Vestibulocerebellum- comprised of the **flocculonodular lobe** and its connections with the **lateral vestibular nuclei** →is involved in vestibular reflexes (such as the vestibuloocular reflex; see below) and in postural maintenance
- Spinocerebellum- comprised of the **vermis** and the **intermediate zones** of the cerebellar cortex, as well as the **fastigial** and **interposed nuclei**receives major inputs from the spinocerebellar tract; its output projects to rubrospinal, vestibulospinal, and reticulospinal tracts; is involved in the integration of sensory input with motor commands to produce adaptive motor coordination
- Cerebrocerebellum- largest functional subdivision of the human cerebellum, comprising the **lateral hemispheres** and the **dentate nuclei;** has extensive connections with the cerebral cortex, via the pontine nuclei (afferents) and the VL thalamus (efferents); is involved in the planning and timing of movements and in the cognitive functions of the cerebellum.



7. Know the range of functions that are served by the basal ganglia.

a. group of subcortical nuclei responsible primarily for motor control, as well as other roles such as motor learning, executive functions and behaviors, and emotions
b. term *basal ganglia* in the strictest sense refers to nuclei embedded deep in the brain hemispheres (striatum or caudate-putamen and globus pallidus), whereas *related nuclei* consist of structures located in the diencephalon (subthalamic nucleus), mesencephalon (substantia nigra), and pons (pedunculopontine nucleus)
c. Ideas and concepts regarding the functions of the basal ganglia were strongly influenced by clinical observations during the 20th century, which showed that lesions of the lenticular nucleus (putamen and globus pallidus) and the subthalamic nucleus (STN) were associated with parkinsonian signs, dystonia, and hemiballismus
d. The basal ganglia and related nuclei can be broadly categorized as (1) input nuclei, (2) output nuclei, and (3) intrinsic nuclei
    i. Input nuclei are those structures receiving incoming information from different sources, mainly cortical, thalamic, and nigral in origin
    ii. The caudate nucleus (CN), the putamen (Put), and the accumbens nucleus (Acb) are all considered input nuclei
    iii. the output nuclei are those structures that send basal ganglia information to the thalamus and consist of the internal segment of the globus pallidus (GPi) and the substantia nigra pars reticulata (SNr)
    iv. Finally, intrinsic nuclei such as the external segment of the globus pallidus (GPe), the STN and the substantia nigra pars compacta (SNc) are located between the input and output nuclei in the relay of information
    v. Cortical and thalamic efferent information enters the striatum (CN, Put, and Acb) to be processed further within the basal ganglia system. The output nuclei (GPi and SNr) project mainly to the thalamus (ventral nuclei), which, in turn, project back to the cerebral cortex (mainly frontal lobe)
    vi. the appropriate functioning of the basal ganglia system requires dopamine to be released at the input nuclei→ Dopamine dysfunction is associated with several basal ganglia movement disorders such as the parkinsonian syndrome (i.e., Parkinson's disease), dystonia, chorea, and tics



8. Explain the theory of dual cortical streams for visual-spatial processing.

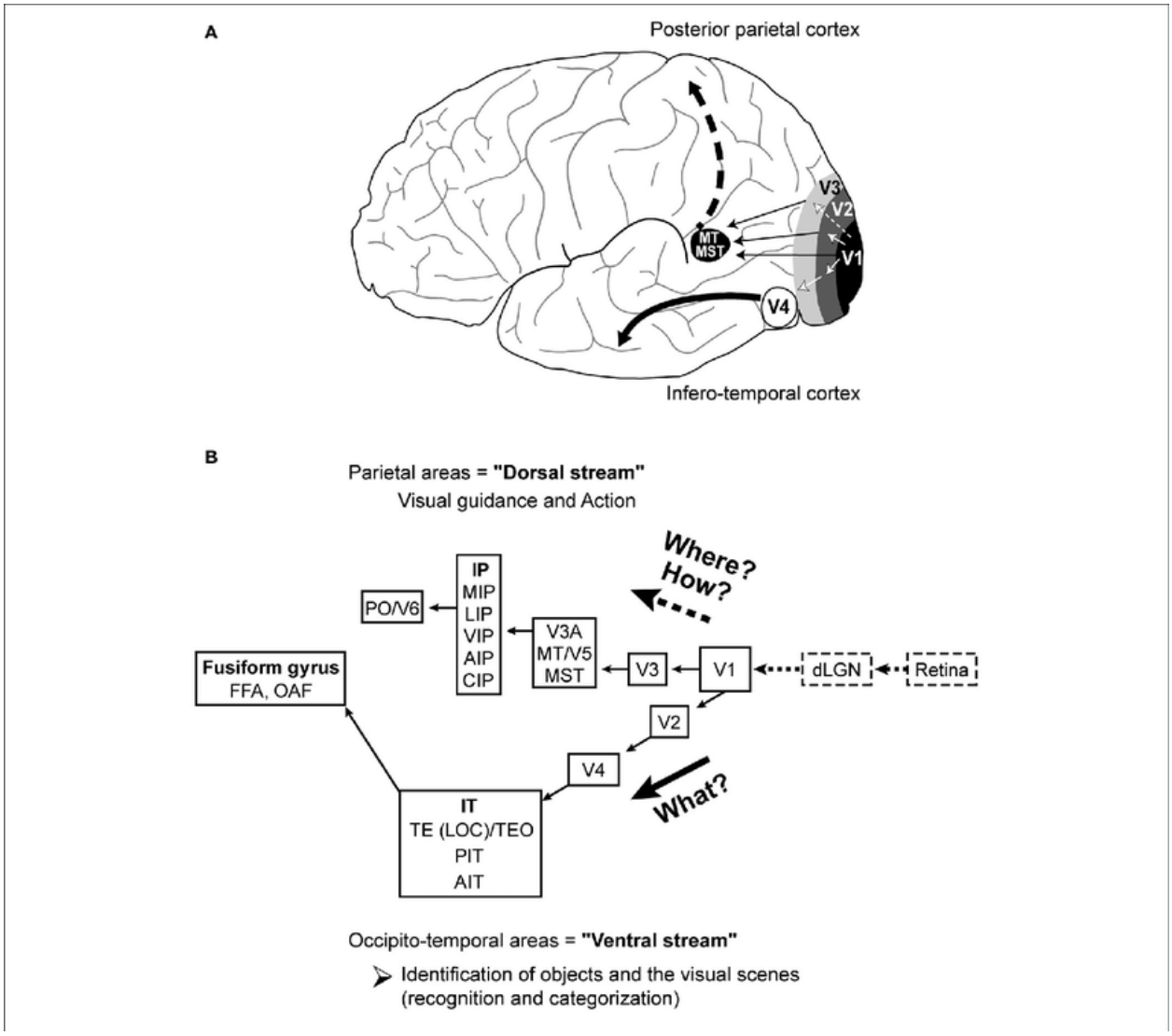
a." two streams hypothesis"- originally characterized by Milner and Goodale (1992) based on their work in patients with visual agnosia

b. outlines that cortical visual processing is divided into two broad networks or "streams" composed of highly interconnected areas : ventral and dorsal

(i) the ventral stream (the "what" or perception), passing from primary visual cortex (V1) through to inferior parts of the temporal lobe, is considered to mediate the transformation of the contents of the visual signal into the mental furniture that guides memory, recognition and conscious perception

(ii) In contrast the dorsal stream (the "where" or action), passing from V1 through to various areas in the posterior parietal lobe, is generally considered to mediate the visual guidance of action, primarily in real time.

c. The brain, however, does not work through mutually insulated subsystems, and indeed there are well-documented interconnections between the two streams
d. Evidence for contributions from ventral stream systems to the dorsal stream comes from human neuropsychological and neuroimaging research, and indicates a crucial role in mediating complex and flexible visual motor skills
e. Same dual cortical stream theorized for hearing as well



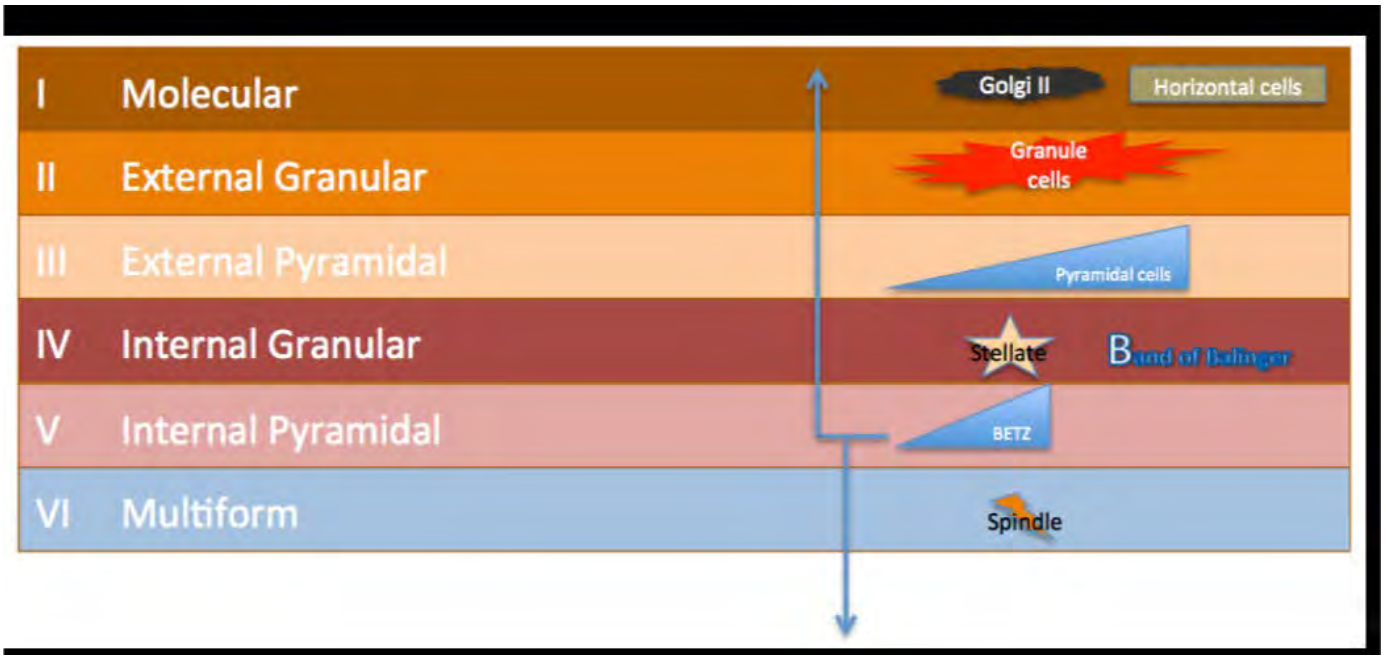
Dorsal and ventral visual streams. (A) Brain localization in humans. Starting with V 1 , the dorsal stream goes to posterior parietal cortex while the ventral stream goes to infero-temporal cortex. (B) The various areas included in the respective streams. Dorsal stream: V 3 ; V 3A ; MT/V 5 , Middle Temporal area; MST, Medial Superior Temporal area; IP, intra-parietal area; MIP, medial intra-parietal area; LIP, lateral intra-parietal area; VIP, ventral intra-parietal area; AIP, anterior intra-parietal area; CIP, caudal part of the intra-parietal area; PO/V 6 , parieto-occipital area; Ventral stream: IT, infero-temporal cortex; TE (=LOC)/TEO, Inferior Temporal Areas; PIT, posterior infero-temporal cortex; AIT, Anterior infero-temporal cortex; FFA, fusiform face area, OFA, occipital face area. Interaction of both streams with each other not represented here.

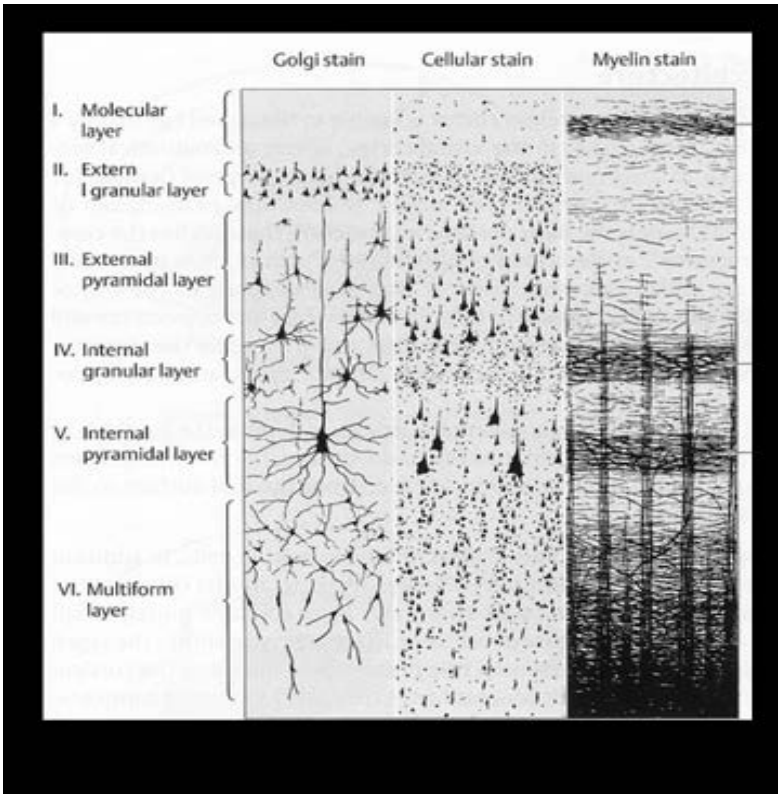
9. Understand the layered and columnar organization of the normal neocortex.
    a. Most of the cerebral cortex is neocortex
    b. However, there are phylogenetically older areas of cortex termed the allocortex

1. These more primitive areas are located in the medial temporal lobes and are involved with olfaction and survival functions such as visceral and emotional reactions
2. In turn, the allocortex has two components: the paleocortex and archicortex
3. the paleocortex includes the piriform lobe, specialized for olfaction, and the entorhinal cortex
4. the archicortex consists of the hippocampus, which is a three-layered cortex dealing with encoding declarative memory and spatial functions

c. The neocortex represents the great majority of the cerebral cortex. It has six layers and contains between 10 and 14 billion neurons. The six layers of this part of the cortex are numbered with Roman numerals from superficial to deep.
    1. Layer I is the molecular layer, which contains very few neurons;
    2. layer II the external granular layer;
    3. layer III the external pyramidal layer;
    4. layer IV the internal granular layer;
    5. layer V the internal pyramidal layer; and
    6. layer VI the multiform, or fusiform layer
d. Each cortical layer contains different neuronal shapes, sizes and density as well as different organizations of nerve fibers.
e. Functionally, the layers of the cerebral cortex can be divided into three parts.
    1. The supragranular layers consist of layers I to III→supragranular layers are the primary origin and termination of intracortical connections, which are either associational (i.e., with other areas of the same hemisphere), or commissural (i.e., connections to the opposite hemisphere, primarily through the corpus callosum); the supragranular portion of the cortex is highly developed in humans and permits communication between one portion of the cortex and other regions.
    2. The internal granular layer, layer IV, receives thalamocortical connections, especially from the specific thalamic nuclei; this is most prominent in the primary sensory cortices
    3. The infragranular layers, layers V and VI, primarily connect the cerebral cortex with subcortical regions; these layers are most developed in motor cortical areas. he motor areas have extremely small or non-existent granular layers and are often called "agranular cortex". Layer V gives rise to all of the principal cortical efferent projections to basal ganglia, brain stem and spinal cord. Layer VI, the multiform or fusiform layer, projects primarily to the thalamus

Layers	Components	Schematic	Afferents		Efferents
I – Molecular	Axons and Dendrites (Cell processes)		From other regions of Cortex and Brainstem	Epomedicine.com	To other regions of cortex (Intra-cortical Association functions)
II - External granular	Densely packed Stellate cells + Small pyramidal cells				
III – External pyramidal	Loosely packed Stellate cells + Medium pyramidal cells				
IV – Internal granular	Densely packed Stellate cells only			+ From Thalamus	
V – Internal pyramidal	Large pyramidal cells only (few stellate cells) – Giant Pyramidal cells of Betz			+ From Brain stem	To Brain stem & Spinal cord (Projection fibers)
VI - Multiform	Multiple sized pyramidal cells + Loosely packed stellate cells				To Thalamus




Golgi stain
Cellular stain
Myelin stain
I. Molecular layer
II. Extern l granular layer
III. External pyramidal layer
IV. Internal granular layer
V. Internal pyramidal layer
VI. Multiform layer

Content Category 2- Biological Mechanisms in Development and Behavior- Sections D & E

ABP Content Specs for Developmental-Behavioral Pediatrics Fellow Collaboration Project

Prepared by: Devina Savant, MD, Brown/Hasbro Children's- DBP Fellow, Dalal Elsori- DBP Fellow, Starrina Gianelloni- DBP Fellow, & Stephanie Klees- DBP Fellow, Rene Bartos- DBP Fellow

Reviewed by: Carrie Kelly, MD, MPH, Brown/Hasbro Children's – Staff/Faculty DBP

D. Genetics
1.Know how to characterize the different Mendelian modes of genetic inheritance
2.Understand genomic imprinting and its implications for genetic inheritance
3.Understand the pattern of inheritance associated with mitochondrial genetic disease
4.Understand the phenomenon of anticipation, associated with triplet repeat geneticdisease
5.Understand the concept of contiguous gene deletion syndromes
6.Understand the continuing interaction between genes and environment that influencesdevelopment and behavior
7.Understand the limitations in interpreting heritability parameter (h2) that is used inpopulation genetic studies
8.Differentiate between malformations and deformations
9.Know genetic mechanisms that contribute to the etiology of developmental andbehavioral disorders
10.Understand how to interpret a pedigree for genetic risk
11.Understand the concept of behavioral phenotypes
12.Understand the concept of uniparental disomy and its role in genetic disorders (eg,Prader-Willi and Angelman syndromes)
13.Recognize developmental-behavioral disorders associated with single gene mutations(Fragile X syndrome, Rett syndrome, Lesch-Nyhan syndrome, etc.)
14.Understand the epigenetic phenomenon of methylation and its implications fordevelopmental-behavioral disorders
15.Understand the concept of mutations or polymorphisms/copy number variations

E. Biological risk factors to neurobiological development
1.Understand how biomedical risk factors (eg, infection, trauma, hypoxia-ischemia,toxins, nutritional deficiencies) jeopardize neurobiological development
2.Recognize factors that may mitigate biomedical risks to neurobiological development(eg, other biological factors, genetic factors)
3.Recognize the phenotypic features of 22q.11.2 deletion syndrome

2. BIOLOGICAL MECHANISMS IN DEVELOPMENT AND BEHAVIOR- 5%

PART D. GENETICS

1. *Know how to characterize the different Mendelian modes of genetic inheritance*

Mendelian inheritance

Basic modes of inheritance for single-gene diseases: autosomal dominant, autosomal recessive, X-linked dominant, X-linked recessive.

Inheritance Pattern	Characteristics	Disease Examples
Autosomal Dominant	Each affected person usually has an affected parent; occurs in every generation	Huntington's disease, neurofibromatosis, achondroplasia, familial hypercholesterolemia
Autosomal Recessive	Both parents of an affected person are carriers; not typically seen in every generation	Tay-Sachs disease, sickle cell anemia, cystic fibrosis, phenylketonuria (PKU)

X-linked Dominant	Females are more frequently affected because all daughters and no sons of an affected man will be affected; can have affected males and females in same generation if the mother is affected	Hypophosphatemic rickets (vitamin D resistant rickets), ornithine transcarbamylase deficiency
X-linked Recessive	Males are more frequently affected; affected males often present in each generation	Hemophilia A, Duchenne muscular dystrophy

Understanding Genetics: A New York, Mid-Atlantic Guide for Patients and Health Professionals. Washington (DC): Genetic Alliance; 2009 Jul 8. [Table, Inheritance Patterns]. Available from: https://www.ncbi.nlm.nih.gov/books/NBK115561/table/appe.T.nc_inheritance_patterncharacteris/

2. *Understand genomic imprinting and its implications for genetic inheritance*

Even though both parents contribute equally to the genetic content of their offspring, genomic imprinting sometimes leads to the exclusive expression of specific genes from only one parent.

Imprinting = the process by which maternally and paternally derived chromosomes are uniquely chemically modified (usually by methylation), leading to different expression of a certain gene or genes on those chromosomes depending on their parental origin.

Parental imprinting is thought to arise through epigenetic modification of DNA that takes place in the ova and sperm during gametogenesis (before fertilization). Genes that are inactivated (silenced) in the female line are said to be maternally imprinted (only the paternally derived allele is expressed and active); genes that are inactivated (silenced) in the male line are said to be paternally imprinted (only the maternally derived allele is expressed and active)

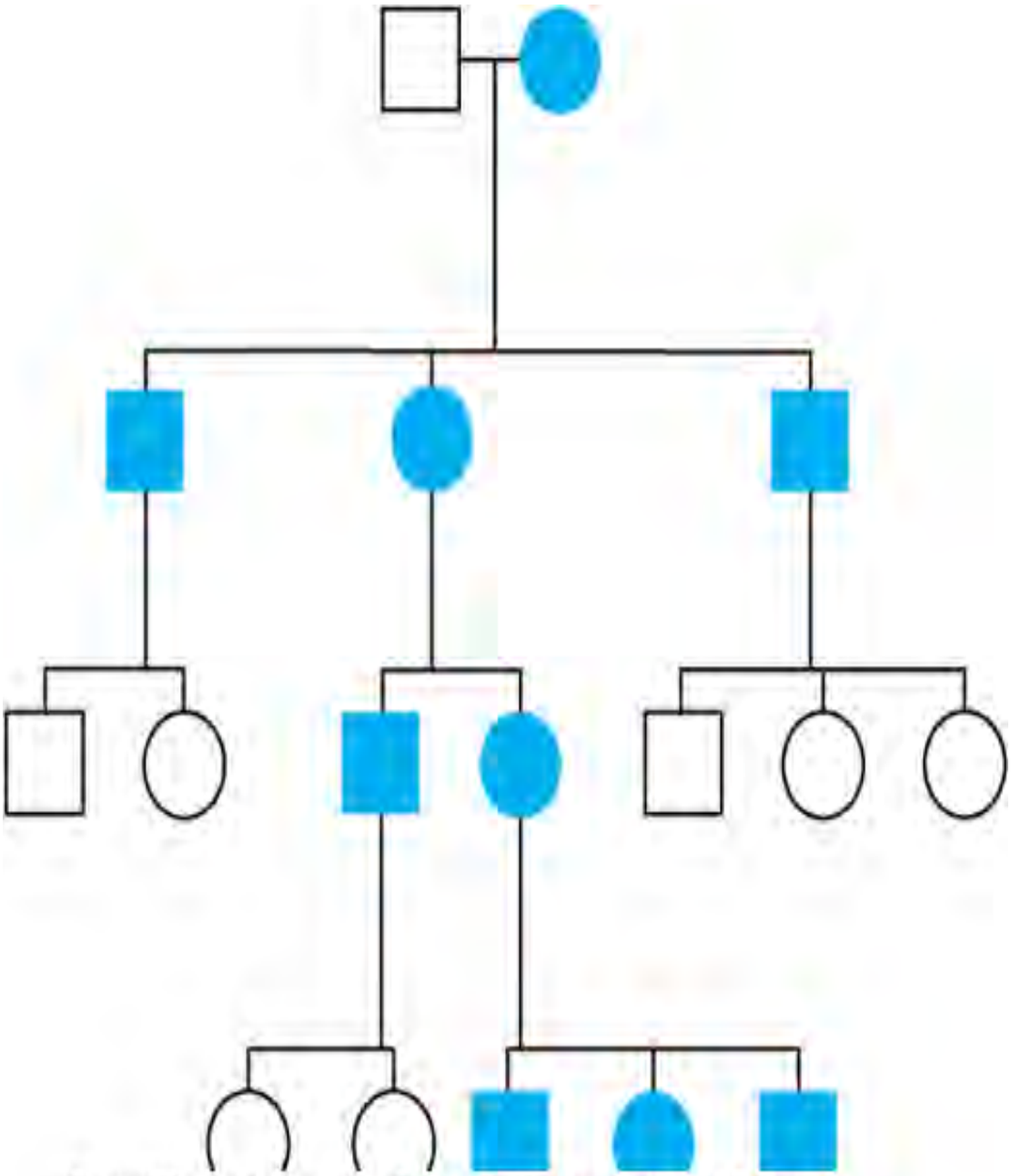
A well-characterized imprinted locus is the 15q11 gene cluster.
Prader-Willi Syndrome (PWS) arises due to the loss of the paternal copy of the PWS "critical region" on chromosome 15q11.2-13
Angelman syndrome results from the loss of the maternal copy of 15q11.2-13.3

3. *Understand the pattern of inheritance associated with mitochondrial genetic disease*
Mitochondrial inheritance refers to traits that are due to genetic variation in the mitochondrial DNA rather than the nuclear genome.
Mitochondria possess a strand of DNA that is completely separate from the cell's nuclear genome. Mitochondrial DNA contains 37 genes that encode proteins involved in oxidative phosphorylation, transfer RNAs, and ribosomal RNAs, all contained in a 16.6 kb circular DNA fragment that replicates autonomously.

Pathogenic variants in mitochondrial genes are responsible for several recognized syndromes and are always maternally inherited because mitochondria are transmitted by the ova, not the sperm.

Conditions caused by a mutation in the mitochondrial DNA have an unusual inheritance pattern:

- both males and females are affected
- the condition is transmitted through the female to her offspring
- if a male has the trait and his spouse doesn’t, their offspring won’t have the trait



Source: Maxine A. Papadakis, Stephen J. McPhee, Michael W. Rabow: Current Medical Diagnosis and Treatment 2020

Because a person's mitochondria derive almost entirely from the ovum, the inheritance pattern is distinct from that of mendelian disorders and is termed "maternal" or, more appropriately, "mitochondrial." An affected woman can pass the defective mitochondrial chromosome to all of her offspring, whereas an affected man has little risk of passing his mutation to a child.

4. *Understand the phenomenon of anticipation, associated with triplet repeat genetic disease*

Anticipation = The tendency in certain genetic disorders for individuals in successive generations to present at an earlier age and/or with more severe manifestations; often observed in disorders resulting from the expression of a nucleotide repeat expansion that tends to increase in size and have a more significant effect when passed from one generation to the next.

Anticipation is a pattern of inheritance in which symptoms become manifest at earlier ages and with increasing severity as traits are passed to subsequent generations. Associated repeat sequences of DNA at disease loci are not stable when passed through meiosis. Repeated DNA sequences, in particular triplets (eg, CGG and CAG), tend to increase their copy number. As these runs of triplets expand, they eventually affect the expression of genes and produce symptoms. Disorders undergoing triplet repeat expansion detected thus far produce primarily neurologic symptoms. Most conditions are progressive. The size of the triplet expansion is roughly correlated with the timing and severity of symptoms.

The most common X-linked disorder demonstrating triplet repeat instability and expansion is Fragile X syndrome.

Other examples of diseases showing anticipation include:

- Huntington Disease
- Myotonic Dystrophy
- Friedreich's Ataxia

5. *Understand the concept of contiguous gene deletion syndromes*

Contiguous gene deletion syndromes: A constellation of clinical findings caused by deletion of a chromosome segment that encompasses two or more adjacent genes

Three common contiguous gene disorders diagnosed by microarray are Williams syndrome, Smith-Magenis syndrome, and 22q11.2 deletion syndrome.

Williams syndrome is a contiguous gene disorder that deletes the gene for elastin and other neighboring genes at 7q11.23

- Characterized by:
    - coarse, elfin-like facies with upturned nose, full lips
    - short stature
    - congenital heart disease (supravalvular aortic stenosis)
    - hypercalcemia

- global developmental delay, most have ID despite strong, superficial language
- ‘cocktail party personality’
- attention problems, anxiety

Smith-Magenis syndrome is associated with a deletion on chromosome 17p11

- Characterized by:
  - prominent forehead, deep-set eyes, cupid-shaped upper lip
  - ID
  - self-mutilating behavior (e.g. head banging, wrist biting, onychotillomania (pulling out fingernails and toenails), and polyembolokoilamania (insertion of foreign bodies into body orifices)
  - sleep disturbance

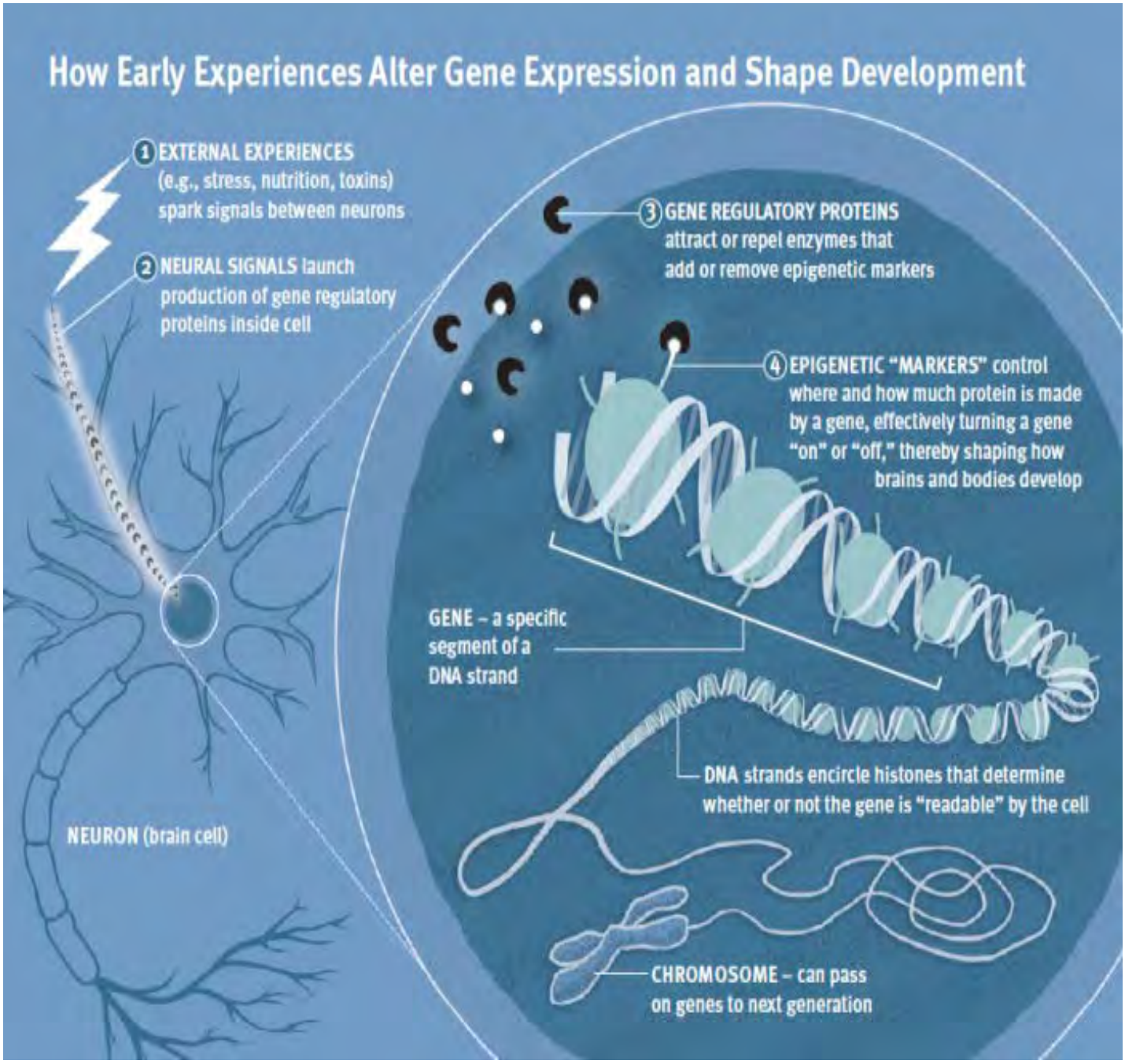
22q11.2 Deletion Syndrome (aka Velocardiofacial Syndrome or DiGeorge Syndrome)

- Characterized by:
  - Characteristic facial features: hooded eyelids, ear anomalies, prominent nasal bridge, bulbous nose, micrognathia, asymmetric crying facies
  - Cleft palate
  - Heart defects (conotruncal cardiac defects such as tetralogy of Fallot)
  - Thymic hypoplasia leading to immunodeficiency
  - Parathyroid hypoplasia leading to hypocalcemia

6. *Understand the continuing interaction between genes and environment that influences development and behavior*

The long-standing debate about the importance of nature *versus* nurture, considered as independent influences, is overly simplistic and scientifically obsolete. Scientists have shifted their focus to take account of the fact that genetic and environmental influences work together in dynamic ways over the course of development. At any time, both are sources of human potential and growth as well as risk and dysfunction. Both genetically determined characteristics and those that are highly affected by experience are open to intervention. The most important questions now concern how environments influence the expression of genes and how genetic makeup, combined with children's previous experiences, affects their ongoing interactions with their environments during the early years and beyond.

Science tells us that the interactions between genes and environment shape human development. Despite the misconception that genes are “set in stone,” research shows that early experiences can determine how genes are turned on and off — and even whether some are expressed at all. The healthy development of all organs, including the brain, depends on how much and when certain genes are activated to do certain tasks. The experiences that children have early in life, therefore, play a crucial role in the development of brain architecture. Ensuring that children have appropriate, growth-promoting early experiences is an investment in their ability to become healthy, productive members of society


How Early Experiences Alter Gene Expression and Shape Development
1 EXTERNAL EXPERIENCES (e.g., stress, nutrition, toxins) spark signals between neurons
2 NEURAL SIGNALS launch production of gene regulatory proteins inside cell
3 GENE REGULATORY PROTEINS attract or repel enzymes that add or remove epigenetic markers
4 EPIGENETIC "MARKERS" control where and how much protein is made by a gene, effectively turning a gene "on" or "off," thereby shaping how brains and bodies develop
GENE – a specific segment of a DNA strand
DNA strands encircle histones that determine whether or not the gene is "readable" by the cell
NEURON (brain cell)
CHROMOSOME – can pass on genes to next generation

National Scientific Council on the Developing Child (2010). *Early Experiences Can Alter Gene Expression and Affect Long-Term Development: Working Paper No. 10*. Retrieved from www.developingchild.harvard.edu.

7. *Understand the limitations in interpreting heritability parameter (h2) that is used in population genetic studies*

Heritability is a measure of how well differences in people's genes account for differences in their traits

Heritability is a statistical concept (represented as h^2) that describes how much of the variation in a given trait can be attributed to genetic variation. An estimate of the heritability of a trait is specific to one population in one environment, and it can change over time as circumstances change.

Heritability estimates range from 0 to 1.

- A heritability close to 0 indicates that almost all of the variability in a trait among people is due to environmental factors, with very little influence from genetic differences
- A heritability close to 1 indicates that almost all of the variability in a trait comes from genetic differences, with very little contribution from environmental factors.
- Many disorders that are caused by mutations in single genes, such as phenylketonuria (PKU), have high heritability. Most complex traits in people, such as intelligence and multifactorial diseases, have a heritability somewhere in

the middle, suggesting that their variability is due to a combination of genetic and environmental factors

Heritability has historically been estimated from studies of twins. Identical twins have almost no differences in their DNA, while fraternal twins share, on average, 50 percent of their DNA. If a trait appears to be more similar in identical twins than in fraternal twins (when they were raised together in the same environment), genetic factors likely play an important role in determining that trait. By comparing a trait in identical twins versus fraternal twins, researchers can calculate an estimate of its heritability.

Some misconceptions about what heritability can and cannot tell us about a given trait:

- Heritability does not indicate what proportion of a trait is determined by genes and what proportion is determined by environment. So, a heritability of 0.7 does not mean that a trait is 70% caused by genetic factors; it means than 70% of the variability in the trait in a population is due to genetic differences among people.
- Knowing the heritability of a trait does not provide information about which genes or environmental influences are involved, or how important they are in determining the trait.
- Heritable is not the same as familial. A trait is described as familial if it is shared by members of a family. Traits can appear in families for many reasons in addition to genetics, such as similarities in lifestyle and environment. For example, the language that is spoken tends to be shared in families, but it has no genetic contribution and so is not heritability.

8. *Differentiate between malformations and deformations*

- Malformation = Morphological abnormality due to innate genetic defect (eg. cleft lip).
- Deformation = Distortion of bodily structure(s) due external physical force (eg. oligohydramnios leading to micrognathia, talipes)
- Disruption = Destruction of bodily structure(s) due to external factor (e.g. amniotic bands leading to digit or limb amputation)

9. *Know genetic mechanisms that contribute to the etiology of developmental and behavioral disorders*

The genetic mechanisms of developmental/behavioral disorders are varied. Mechanisms include:

- the addition of an entire chromosome in each cell (e.g., Down syndrome)
- the loss of an entire chromosome in each cell (e.g., Turner syndrome)
- the loss or deletion of a significant portion of a chromosome (e.g., Cri-du-chat syndrome)
- a microdeletion of a number of closely spaced or contiguous genes within a chromosome (e.g., chromosome 22q11.2 deletion syndrome)
- Mutations within a single gene (e.g., phenylketonuria)
- Imprinting (e.g. Prader-Willi, Angelman syndrome)

Neurodevelopmental Disorders and their Genetic Aetiologies

Group	Disorder	Genetic Aetiology
I (Aneuploidy)	Down's syndrome	Trisomy of chromosome 21 (OMIM #190685).
II (Micro-deletion)	Prader-Willi syndrome / Angelman syndrome	~4 Mb deletion (~7 genes) of chromosome 15q11-q13 (OMIM #176270 and #105830).
	Smith-Magenis syndrome	Deletion (3.7 Mb) of chromosome 17p11.2 (OMIM #182290).
	DiGeorge/velo-cardio-facial syndrome	Hemizygous deletion (1.5 to 3.0-Mb) of chromosome 22q11.2 (OMIM #188400 and

		#192430).
	William's-Beuren syndrome	Deletion of chromosomal region 7q11.2 (OMIM #194050).
III (Single-gene defect)	ATR-X syndrome	Mutations in the ATR-X gene on the X-chromosome (OMIM #301040)
	Barth syndrome (X-linked cardioskeletal myopathy and neutropenia)	Mitochondrial functional impairments due to the tafazzin (TAZ) gene on chromosome Xq28 (OMIM #302060).
	Fragile-X syndrome	CCG repeat expansion of the FMR1 gene (OMIM #300624).

	ICF syndrome	Mutations in the DNA methyltransferase 3B (DNMT3B) gene on chromosome 20 (OMIM #242860).
	Neurofibromatosis	Mutations or deletion (~1.5 Mb) in the neurofibromin gene on chromosome 17q11.2 (OMIM +162200).
	Rett syndrome	Mutations in the MeCP2 gene on the X-chromosome (OMIM #312750).
	Smith-Lemli-Opitz syndrome	Mutations in the gene encoding sterol delta-7-reductase (DHCR7) on chromosome

		11q12-q13 (OMIM #270400).
IV (Multifactorial)	Addictive disorders	Multiple genes (?)
	Attention deficit (hyperactivity) disorders	
	Anxiety disorders	
	Asperger’s disorder	
	Autistic disorders	
	Depressive illness	
	Dyslexia (reading disability)	

	Eating disorders	
	Epilepsy (seizure disorder)	
	Fetal alcohol syndrome	
	Hydrocephalus	
	Manic depressive illness (bipolar disorder)	
	Mental retardation	
	Schizophrenia	
	Spina bifida	

	Tourette’s syndrome	

Table 1. van Loo KM, Martens GJ. Genetic and environmental factors in complex neurodevelopmental disorders. *Curr Genomics*. 2007;8(7):429-444. doi:10.2174/138920207783591717

***Note: This table is a bit out of date (published in 2007--e.g. still using MR and Asperger terminology). However, I included it since it summarizes the genetics of some of the developmental/behavioral disorders we need to know as DBPs and also demonstrates how many of the disorders we see are multifactorial--most likely resulting from interplay of multiple genes + environment.

The Genetics of Autism

Genetic Cause	% of ASD	Type of Genetic Testing
Chromosomal Abnormalities	Up to 5%	Chromosome Analysis or Karyotype
Single Genes	Up to 5%	Testing of individual genes like the Fragile X gene
Copy Number Variants (CNVs)	Up to 11%	Chromosomal Microarray Analysis (CMA)

CAR Autism Roadmap (Center for Autism Research at The Children's Hospital of Philadelphia)

Last updated June 17,2020

Available from : https://www.carautismroadmap.org/the-genetics-of-asd/

Chromosomal disorders

(I included some chromosomal disorders here to review since they weren't included elsewhere in the content specs)

Trisomy 13

- Associated with AMA
- Midline defects including cleft palate, holoprosencephaly
- Cutis aplasia
- Microcephaly
- Microophthalmia, ear anomalies
- Hea
- Severe growth (IUGR)/developmental impairment
- 45% die in 1st month, 85% in first year

Trisomy 18

- Associated with AMA
- Rocker bottom feet

- Hypoplastic nails, clenched fists, overlapping fingers
- Microcephaly, prominent occiput
- Microophthalmia, micrognathia, ear anomalies
- Heart malformations, horseshoe kidneys
- Severe growth (IUGR)/developmental impairment
- 50% die in 1st week, 95% in first year

Down Syndrome

Common dysmorphic features that are commonly seen in individuals with

Down syndrome:

HEENT

- Small head circumference
- Brachcephaly
- low set, folded or dysplastic ears
- Upslanting palpebral fissures
- Epicanthal folds
- Brushfield spots (Speckled iris)
- Flat nasal bridge
- Small mouth, protruding tongue
- Excess skin at nape of neck
- Short neck

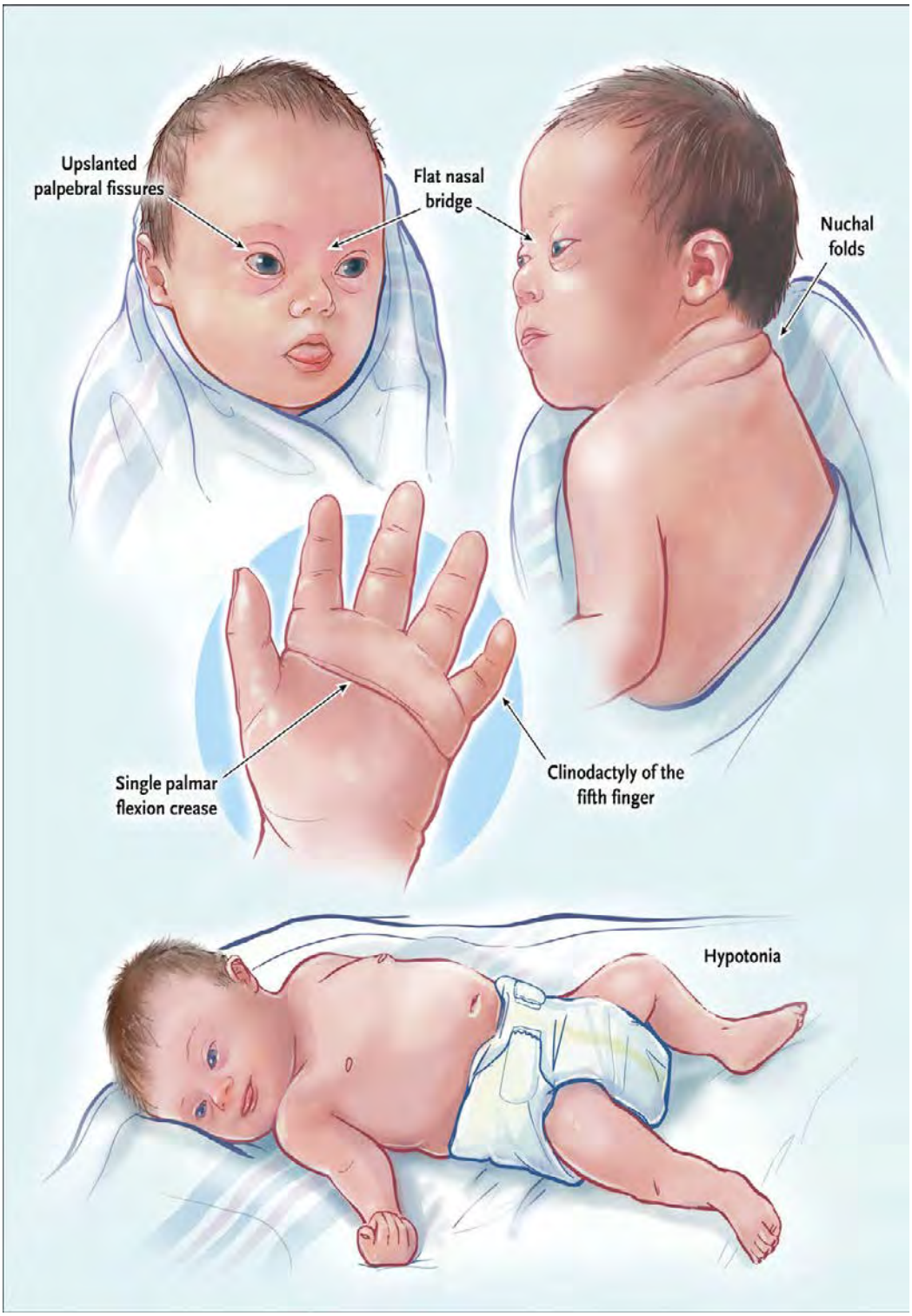
Extremities

- Single transverse palmar crease

- Fifth finger clinodactyly with hypoplastic mid phalanx
- Wide space between first and second toes (sandal gap)
- Hyperextensible joints
- Short stature

Neurology

- Hypotonia
- Abnormal Moro response


Upslanted palpebral fissures
Flat nasal bridge
Nuchal folds
Single palmar flexion crease
Clinodactyly of the fifth finger
Hypotonia

Gastrointestinal:

- Evaluate for feeding problems
- Maintain high index of suspicion for malformations, including duodenal atresia and Hirschsprung disease

Ophthalmology: formal ophthalmology evaluation by 6 months of age for strabismus, cataracts, and nystagmus.

Cardiology:

- 40-50% of individuals with DS have congenital heart defect.
- Most common defect is AV canal (37%)
- Other defects include VSD (31%), ASD (15%), TOF (5%), and PDA (4%).

Hematology: CBC for leukemoid reactions and polycythemia (18%) and rare leukemia (1%)

Audiology:

- Monitor for serous otitis media (50-70%).
- Check hearing every 6 months up to 3 years of age if tympanic membrane cannot be visualized. Consider referral to ENT.

  Audiology evaluation annually for school-aged children.

Sleep: Monitor for signs and symptoms of obstructive sleep apnea.

Endo: Hypothroidism is present 20-40% of individuals with DS.

- Hormone screen at 6 months, 12 months, and then annually

Neck: Risk from atlantoaxial instability is controversial.

- Most important screening is regular and careful history and neurology exam
- Symptoms include new onset gait problems, bowel or bladder dysfunction, and change in reflexes.
- Radiographs for atlantoaxial instability no longer recommended, but could consider for children in contact sports or if symptomatic.
- If radiographs are concerning, then follow up neck MRI is indicated.
- Recommend discouraging high-risk activities if evidence of instability.
- Surgical correction is also controversial, especially if patient is asymptomatic.

Developmental-Behavioral Concerns

- Most individuals with DS have mild to moderate ID, with IQ in the 50 to 70 or 35 to 50 range, respectively, although some are severely impaired with an IQ of 20 to 35
- Language: Expressive more delayed than receptive
- Visuospatial skills: delayed acquisition compared to cognition
- Autism is a common comorbidity, affecting as many as 7 percent of children with DS

- Behavioral and psychiatric disorders are more common in DS than typical children but less common than in those with other causes of intellectual disability
    - Disruptive behavioral disorders, such as attention-deficit hyperactivity disorder, conduct/oppositional disorder, or aggressive behavior, are most common
- Mental health: higher rates of depression and anxiety
- Associated neuro conditions: stereotypies, bradykinesia, ataxia, seizures, early onset Alzheimer disease

Genetics:

- Diagnosis confirmed by karyotype analysis
- Genetic counseling is essential regarding recurrence risk

Genetic causes of Down syndrome and their respective recurrence risks:

1. 95% have nonfamilial trisomy 21
    - Recurrence risk for women who do NOT have advanced maternal age is greater than for other women of same age who have not had a child with Down syndrome. The basis for this increased risk is unknown.
    - Recurrence risk for woman of advanced maternal age is not increased above risk for advanced maternal age.

2. 2-4% have unbalanced translocation between chromosome 21 and another acrocentric chromosome, usually chromosome 14. (Robertsonian translocation)

- Recurrence risk depends on whether one of the parents is a carrier of a balanced rearrangement.
    - If neither parent is a carrier, then the recurrence risk is the same as the population risk.
    - If the father is a carrier, then the recurrence risk is 3-5%.
    - If the mother is a carrier, then the recurrence risk is 10-15%.
    - If one of the parents is a carrier of a 21;21 translocation (isochromosome), then the recurrence risk is 100%

3. 1-2% have mosaic trisomy 21
    - May have phenotypically less severe manifestations
    - The recurrence risk is no greater than the general population risk unless one of the parents is mosaic for trisomy 21.

<u>Klinefelter Syndrome</u>

- 47 XXY
- Most common cause of primary hypogonadism
    - Specifically: hypergonadotropic hypogonadism; tx with androgen replacement
- Clinical features include: small testes, sparse facial hair, gynecomastia
- Most individuals are sterile (accounts for 5-15% of sterility in males)
- Learning and developmental disabilities are common (>50 percent)
- Difficulties with expressive language, auditory processing, social skills, low avg/avg IQ

- Increased risk of CA of breast/mediastinal tumors[15]

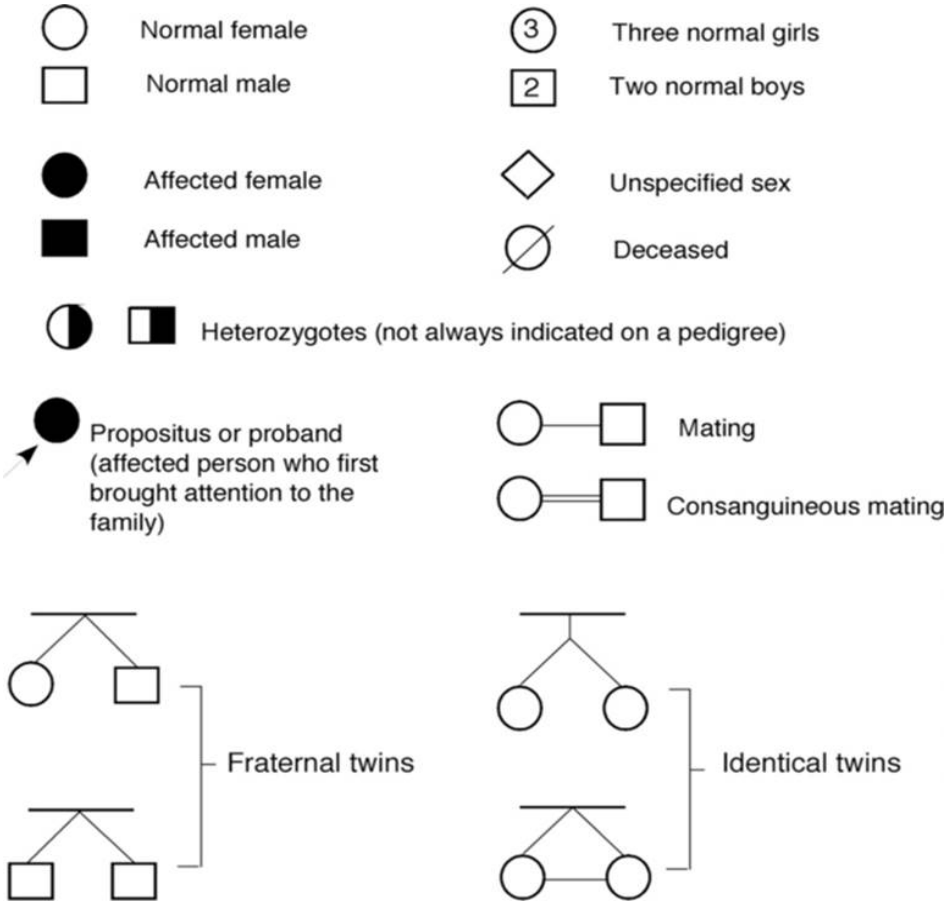
Turner Syndrome

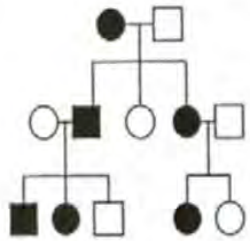
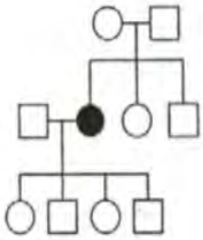
- 1 in /2000-2500 live female births
- Caused by loss of part or all of an X chromosome
    - ~45%: 45,X (monosomy X)
    - ~50%: 45,X with mosaicism (45, X/46XX or 45,X/47,XXX)
    - Several types of anomalies in the X chromosome can cause Turner syndrome, with or without mosaicism (e.g. ring X or isoX)
- Clinical features: short stature, webbed neck (in 30%), low hairline at back of neck

  'shield' chest, cubitus valgus, Madelung deformity of the forearm and wrist
- 'Streak' ovaries (consists of small amounts of connective tissue and no follicles or only a few atretic follicles) primary hypogonadism, premature ovarian failure
- Congenital lymphedema of the hands and feet
- Presenting features in adolescence: short stature, minimal or no breast development, primary amenorrhea
- Cardiac abnormalities in up to 50%: bicuspid aortic valve, coarctation of the aorta
- Renal abnormalities in 30-40% e.g. horseshoe kidney
- Increased risk of autoimmune disorders e.g. hypothyroidism (Hashimoto's thyroiditis), celiac disease, and inflammatory bowel disease (IBD)
- Most have normal IQ.

- Increased risks for selective impairment in nonverbal skills which may include difficulty with nonverbal, problem-solving tasks such as mathematics; psychomotor deficits, such as clumsiness; and problems with visual-spatial organization

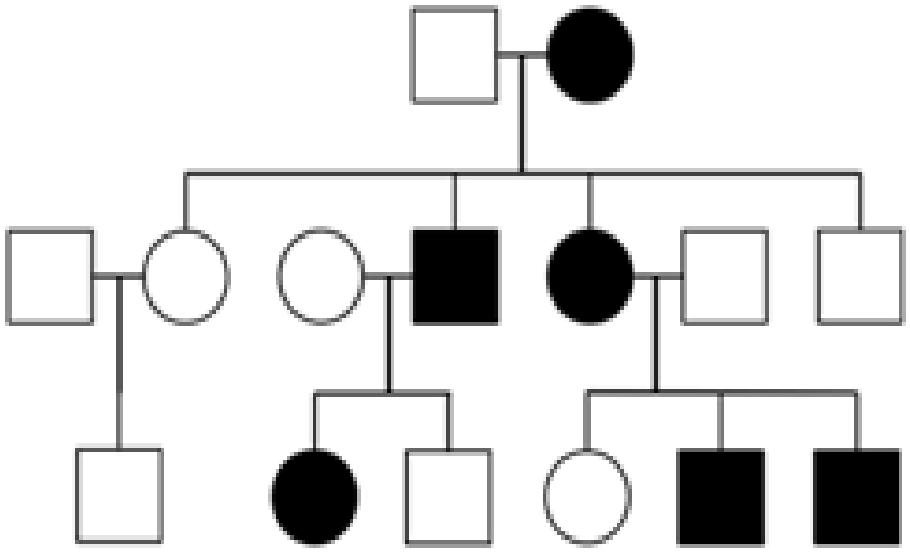
10. *Understand how to interpret a pedigree for genetic risk*



TYPE OF INHERITANCE	CHARACTERISTICS	EXAMPLES
Autosomal dominant 	▪ Both sexes equally affected ▪ Vertical transmission ▪ Father-to-son transmission ▪ Affected individuals transmit trait to ~50% offspring	▪ Huntington disease ▪ Achondroplasia ▪ NF type 1 ▪ Marfan syndrome ▪ Familial hypercholesterolemia
Autosomal recessive 	▪ Both sexes equally affected ▪ Usually no prior family history ▪ Consanguinity ▪ Mating between two carriers transmits trait to ~25% offspring	▪ Hurler syndrome ▪ Hereditary hemochromatosis ▪ Cystic fibrosis ▪ Sickle-cell anemia ▪ Phenylketonuria (PKU) ▪ β-thalassemia ▪ Tay-Sachs

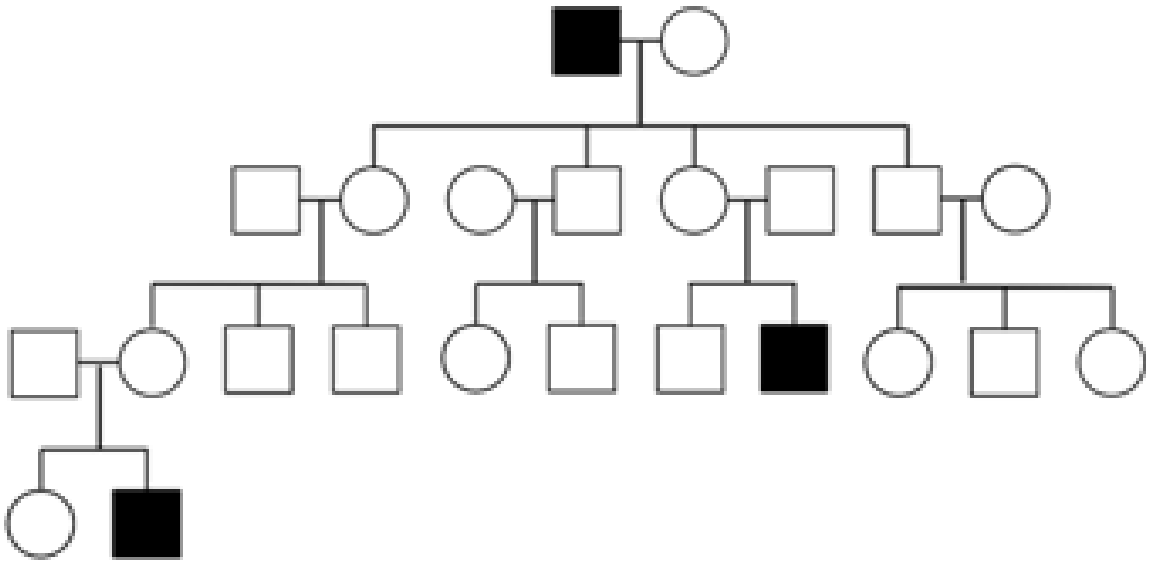
X-linked dominant

In X-linked dominant inheritance, the gene responsible for the disease is located on the X-chromosome, and the allele that causes the disease is dominant to the normal allele in females. Because females have twice as many X-chromosomes as males, females tend to be more frequently affected than males in the population. However, not all pedigrees provide sufficient information to distinguish XD and AD. One definitive indication that a trait is inherited as AD, and not XD, is that an affected father passes the disease to a son; this type of transmission is not possible with XD, since males inherit their X chromosome from their mothers.



X-linked recessive

Because males have only one X-chromosome, any male that inherits an X-linked recessive disease allele will be affected by it (assuming complete penetrance). Therefore, in XR modes of inheritance, males tend to be affected more frequently than females in a population. This is in contrast to AR and AD, where both sexes tend to be affected equally, and XD, in which females are affected more frequently. Note, however, in the small sample sizes typical of human families, it is usually not possible to accurately determine whether one sex is affected more frequently than others. On the other hand, one feature of a pedigree that can be used to definitively establish that an inheritance pattern is <u>not</u> XR is the presence of an affected daughter from unaffected parents; because she would have had to inherit one X-chromosome from her father, he would also have been affected in XR.



11. *Understand the concept of behavioral phenotypes*

Nyhan introduced the term behavioral phenotype in 1972 to describe outwardly observable behavior so characteristic of children with genetic disorders that its presence suggests the underlying genetic condition.

Another definition: behavioral phenotype = a characteristic pattern of motor, cognitive, linguistic and social abnormalities that is consistently associated with a biological/neurodevelopmental disorder.

This does not mean that the behavior is present in all instances but that the probability of its occurrence is increased.

Examples:

- Lesch-Nyhan: characteristic self-mutilation of fingers and lips
- Prader-Willi Syndrome: hyperphagia and compulsive eating
- Williams Syndrome: the superficial sociability, hyperlalia, and language disorder

12. *Understand the concept of uniparental disomy and its role in genetic disorders (eg, Prader-Willi and Angelman syndromes)*

Uniparental disomy (UPD) = The situation in which both copies of a chromosome pair (or chromosome pair segment) are from one parent (i.e., no copy is from the other parent).

UPD can occur as a random event during the formation of egg or sperm cells or may happen in early fetal development.

In many cases, UPD likely has no effect on health or development. Because most genes are not imprinted, it doesn't matter if a person inherits both copies from one parent instead of one copy from each parent. In some cases, however, it does make a difference whether a gene is inherited from a person's mother or father. A person with UPD may lack any active copies of essential genes that undergo genomic imprinting. This loss of gene function can lead to developmental delay, intellectual disability, or other health problems.

Several genetic disorders can result from UPD or a disruption of normal genomic imprinting. The most well-known conditions include Prader-Willi syndrome and Angelman syndrome. Both of these disorders can be caused by UPD or other errors in imprinting involving genes on the long arm of chromosome 15.

Prader-Willi Syndrome:

- 20-30% of cases caused by uniparental disomy (in which paternal copy of gene is replaced by a second maternal copy)

        - 65-75% caused by deletions within the PWS-critical chromosome region
        - Remaining cases are caused by defects in the imprinting process or chromosomal translocations

Angelman Syndrome:

- 2-5 percent cases are caused by uniparental disomy
- Deletions account for > 70 percent of cases

13. *Recognize developmental-behavioral disorders associated with single gene mutations* (Fragile X syndrome, Rett syndrome, Lesch-Nyhan syndrome, etc.)

<u>Fragile X syndrome</u>

- caused by expansion in the number of CGG repeats within the FMR1 gene.
- Expansion of CGG repeats to >200 allows hypermethylation of FMR1, resulting in impaired transcription and reduced production of FMRP, which may adversely impact prenatal and postnatal brain development.
- X-linked disorder
- Most common known genetic cause of *inherited* ID (2nd most common cause of genetic ID after Down syndrome)
- Most common known single-gene cause of ASD
- As repeat size increases, stability decreases, leading to increases in number of repeats in the FMR1 region during gametogenesis. The lower and upper boundaries of repeat length are variably defined, but generally described as:
    - Normal – 5 to 44 CGG repeats

- Intermediate expansion – 45 to 54 CGG repeats
- Premutation – 55 to 200 CGG repeats
- Full mutation – >200 CGG repeats

Physical features

- Prominent ears
- Long and narrow face
- Prominent jaw and forehead
- Macroorchidism (usually post puberty)
- Hyperextensible joints, flat feet
- Strabismus

Cognitive function

- Developmental delay (later attainment of language and motor milestones), ID, LD are common
- Most males have mild to moderate ID

Behavioral features

- ASD, ADHD, anxiety are common features
- ASD in 50-70%

Addl clinical features

- Seizures in 10-20%

Fragile X in girls

Full mutation: females usually less affected than males due to X inactivation

- Phenotype varies from typical to severely affected
- Normal intellect in ~50%, borderline to mild ID also
- Cognitive impairment appears to correlate with the activation ratio of the fragile X chromosome rather than the size of the amplification
- Associated behavioral/emotional difficulties--inattention/ADHD, shyness/social anxiety. ~20% girls with ASD, ~5% seizures

Premutation can be associated with:

- Premature ovarian insufficiency (POI) in women
- Fragile X-associated tremor-ataxia syndrome (FXTAS) later in life
- Neurocognitive deficits

**Fragile X testing is always indicated for girls with GDD/ID

Rett Syndrome

• Mutations in MECP2 on X chromosome

• 1 in 9/10,000 live birth

• X-linked dominant pattern

• Acquired microcephaly

• Developmental regression: 6 to 18 months of apparently normal development before developing severe impairments in communication, cognitive, fine motor skills, coordination

• Stereotypic hand movements. Affected girls lose purposeful use of their hands and begin making repeated hand wringing, washing, or clapping motions

• Autistic-like behaviors and hyperventilation episodes

• Ataxia

• Seizures

• Autonomic nervous system dysfunction

• Sleep disturbances

• Rett syndrome is part of a spectrum of disorders with the same genetic cause. Other disorders on the spectrum include PPM-X syndrome, *MECP2* duplication syndrome, and *MECP2*-related severe neonatal encephalopathy. These other conditions can affect males

Lesch-Nyhan

- Caused by mutations in the *HPRT1* gene that result in a deficiency or complete absence of enzyme hypoxanthine phosphoribosyltransferase 1
- X-linked recessive inheritance pattern
- Severity of the disease have been associated with the degree of enzyme deficiency
- Due to enzyme deficiency-->purines are broken down but not recycled, producing abnormally high levels of uric acid, also associated with low levels of dopamine

which may play a role in the movement problems and other features of this disorder.

- Associated movement disorders include dystonia, chorea, ballismus.
- Self-injury (including biting and head banging)

Neurofibromatosis Type 1 (NF-1)

- Autosomal dominant
- Caused by pathogenic variants in the NF1 gene, located at chromosome 17q11.2 which results in loss of production or reduced function of protein, neurofibronin
- In ~50% cases, caused by spontaneous mutation
- Penetrence is complete but expression is highly variable
- NIH Diagnostic criteria —2 or more must be present to make the diagnosis of NF1:
    - > Six or more café-au-lait macules (>5 mm in prepubertal, >15 mm in in postpubertal individuals)
    - Two or more neurofibromas of any type or one plexiform neurofibroma
    - Freckling in the axillary or inguinal regions
    - Optic glioma
    - Two or more Lisch nodules (iris hamartomas).
    - A distinctive bony lesion, such as sphenoid dysplasia or thickening of the long bone cortex with or without pseudoarthrosis.
    - A first-degree relative (parent, sibling, or offspring) with NF1 based upon the above criteria.

- The typical order of appearance of clinical manifestations is café-au-lait macules, axillary and/or inguinal freckling, Lisch nodules (iris hamartomas), and neurofibromas
- Can be associated with pheo or renal artery stenosis which may manifest with Htn.
- Genetic testing is not required to make the diagnosis but can be helpful in confirming the diagnosis for children who do not meet diagnostic criteria or only demonstrate café-au-lait macules and axillary freckling
- 50% with LD

Tuberous Sclerosis

- Autosomal dominant
- Caused by a mutation in either the TSC1 gene or the TSC2 gene
- Characterized by the development of variety of benign tumors in multiple organs, including the brain, heart, skin, eyes, kidney, lung, and liver
- Diagnosis based upon genetic testing results and/or clinical findings
    - Definite TSC requires two major features or one major and two or more minor features
- Major clinical features:
    - Hypomelanotic macules (≥3, at least 5 mm diameter) (Ash leaf spots)
    - Angiofibromas (≥3) or fibrous cephalic plaque
    - Ungual fibromas (≥2)
    - Shagreen patch (cobblestone/orange peel appearing skin)

    - Multiple retinal hamartomas
    - Cortical dysplasias (includes tubers) (may present as seizures)
    - Subependymal nodules
    - Subependymal giant cell astrocytoma (SEGAs)
    - Cardiac rhabdomyoma
    - Lymphangioleiomyomatosis (LAM)
    - Angiomyolipomas (≥2)
- Minor clinical features:
    - "Confetti" skin lesions (1 to 2 mm hypomelanotic macules)
    - Dental enamel pits (≥3)
    - Intraoral fibromas (≥2)
    - Retinal achromic patch
    - Multiple renal cysts
    - Nonrenal hamartoms
- Developmental and behavioral manifestations:
    - ID/LD in 45-65% patients
    - Autism and ASD-like behaviors, hyperactivity, inattention, and self-injurious behavior are common
- Associated conditions: seizures in 79-90% patients, increased risk for neuroendocrine tumors

Smith-Lemli-Opitz Syndrome

- caused by mutations in the DHCR7 gene on chromosome 11 which leads to deficiency of C7-reductase (necessary for cholesterol production), decrease in cholesterol levels and an accumulation of cholesterol precursor molecules (7-DHC & 8-DHC)
- Autosomal recessive
- Clinical features include: microcephaly, characteristic facial features (e.g. short-upturned nose, high arched palate, low set ears, micrognathia), growth impairment, moderate to severe ID, and multiple major and minor malformations such as cleft palate, heart defects, underdeveloped external genitalia in males, postaxial polydactyly, and 2-3 syndactyly of the toes
- The clinical spectrum is wide: some individuals with SLO have typical development and only minor malformations

Phenylketonuria (PKU)

- Autosomal recessive
- PKU is caused by deficiency of phenylalanine hydroxylase (PAH) which leads to elevated blood and urine concentrations of phenylalanine and its metabolites
- The gene encoding PAH is found on chromosome 12q24.1. More than 1000 mutations have been identified.
- Usually diagnosed by elevation of phenylalanine on newborn screen
- If untreated: progressive ID, behavior problems (irritability, hyperactivity), microcephaly, seizures, may lead to white matter changes, spasticity, hypertonia and movement disorders

- Fair complexion is common
- Can be associated with "mousy" odor (due to the increased concentration of phenylacetic acid)

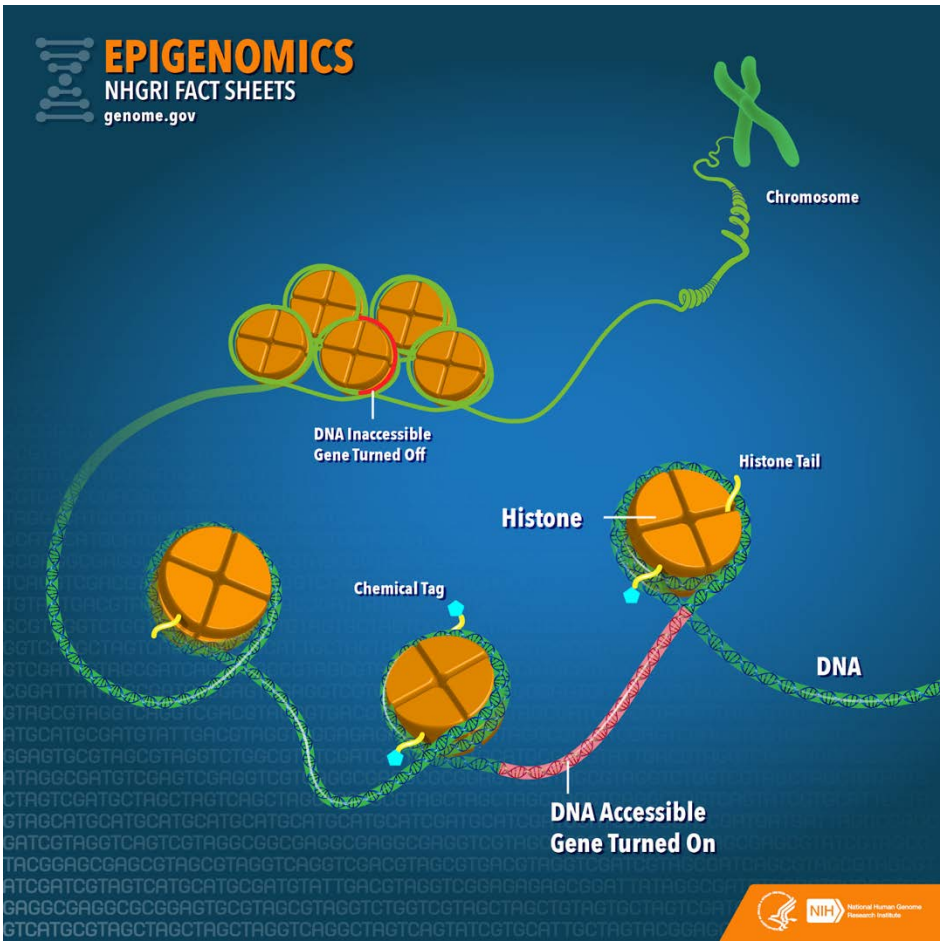
14. *Understand the epigenetic phenomenon of methylation and its implications for developmental-behavioral disorders*

Epigenetics- refers to chemical alterations to DNA nucleotides or proteins that control gene expression but do not alter the DNA sequence

Epigenetic modifications remain as cells divide and in some cases can be inherited through the generations. Environmental influences, such as a person's diet and exposure to pollutants, can also impact the epigenome.

DNA methylation is a common type of epigenetic modification. DNA methylation involves attaching small molecules called methyl groups, each consisting of one carbon atom and three hydrogen atoms, to segments of DNA. When methyl groups are added to a particular gene, that gene is turned off or silenced, and no protein is produced from that gene.

Because errors in the epigenetic process, such as modifying the wrong gene or failing to add a compound to a gene, can lead to abnormal gene activity or inactivity, they can cause genetic disorders.



15. *Understand the concept of mutations or polymorphisms/copy number variations*

A gene mutation is a permanent alteration in the DNA sequence that makes up a gene, such that the sequence differs from what is found in most people. Mutations range in size; they can affect anywhere from a single DNA base pair to a large segment of a chromosome that includes multiple genes.

Most disease-causing gene mutations are uncommon in the general population. However, other genetic changes occur more frequently. Polymorphisms are genetic alterations that occur in more than 1 percent of the population. They are common enough to be considered a normal variation in the DNA. Polymorphisms are responsible for many of the normal differences between people such as eye color, hair color, and blood type. Although many polymorphisms have no negative effects on a person's health, some of these variations may influence the risk of developing certain disorders

Polymorphism = A natural variation in a gene, DNA sequence, protein, or chromosome
Polymorphism involves one of two or more variants of a particular DNA sequence. The most common type of polymorphism involves variation at a single base pair. Polymorphisms can also be much larger in size and involve long stretches of DNA. Called a single nucleotide polymorphism, or SNP (p, scientists are studying how SNPs in the human genome correlate with disease, drug response, and other phenotypes

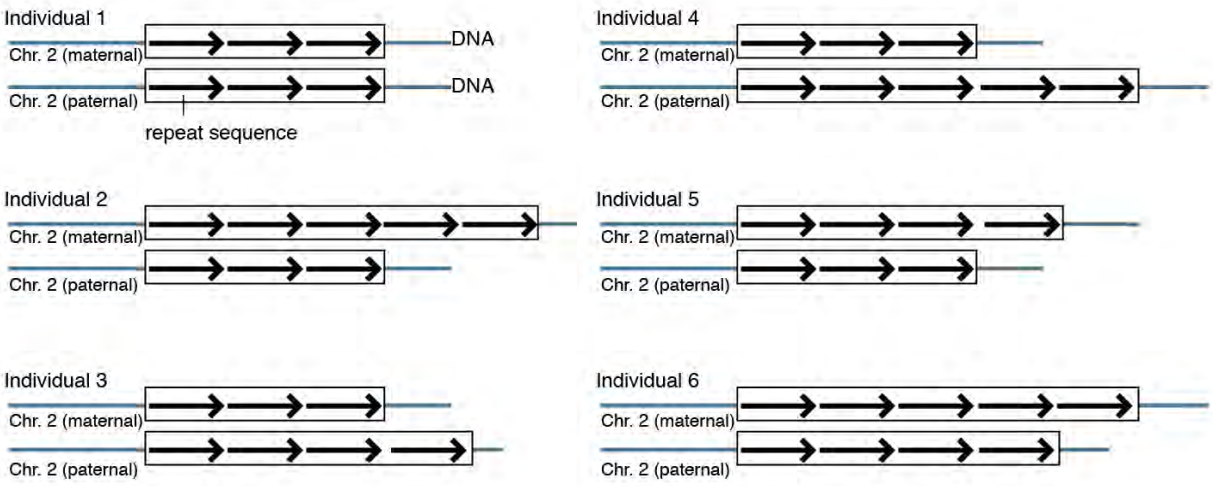
Copy number variation (CNV) = Duplication or deletion of a section of DNA. CNVs can be benign (normal), pathogenic, or of uncertain clinical significance.

People have two copies of most genes, one copy inherited from each parent. In some cases, however, the number of copies varies—meaning that a person can be born with one, three, or more copies of particular genes. Less commonly, one or more genes may be

entirely missing. This type of genetic difference is known as copy number variation (CNV)

Copy number variation results from insertions, deletions, and duplications of large segments of DNA. These segments are big enough to include whole genes. Variation in gene copy number can influence the activity of genes and ultimately affect many body functions.

Copy number variation accounts for a significant amount of genetic difference between people. More than 10 percent of human DNA appears to contain these differences in gene copy number. While much of this variation does not affect health or development, some differences likely influence a person's risk of disease and response to certain drugs. Future research will focus on the consequences of copy number variation in different parts of the genome and study the contribution of these variations to many types of disease.



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Content Category 3: Family and Societal Factors
ABP DBP Board Content Specifications (2014)

ABP Content Specs for Developmental-Behavioral Pediatrics Fellow Collaboration Project

Contributing DBP Fellows & Pediatric Residents: DBP Fellow- Anisha Srinivasan, Seattle Children's/UW, Rebecca Christi, Madigan Army Medical Center; Pediatric Residents- Shailly Gaur, Alexa Ernst, Anna Rees- Atrium Health/Levine Children's Hospital

Staff/Faculty Reviewer: Yasmin Senturias, Developmental Behavioral Pediatrics of the Carolinas- Charlotte; Eric Flake, Madigan Army Medical Center

1. Family and Societal Factors

A. Structural factors

1. Recognize common challenges to the psychosocial development of children in blended families (eg, insecurity associated with loss of a parent, difficulties with attachment to a new parent, reactions to new siblings, inconsistent discipline practices, etc)
2. Understand the typical profile of children whose parents are gay or lesbian compared to children whose parents are heterosexual with respect to their academic achievement, psychological adjustment, and psychosexual development
3. Understand typical reactions that children whose parents are separated or divorced have to their families at various developmental stages
4. Recognize possible effects of new relationships of either parent, after divorce or separation, on children's behavior (eg, bedwetting, separation difficulties, attentional difficulties, depression, hostility or jealousy toward the new adult and/or childre
5. Know how to counsel parents who are separating in order to minimize the adverse effects on their children
6. Understand the ways a single parent can best support her/his child(ren)
7. Recognize typical responses of children at different developmental stages to the loss of a parent by death or disappearance
8. Recognize common symptoms of attachment disorders at various ages
9. Understand the benefits and potential problems of kinship versus non-kinship adoption
10. Understand issues related to children's development and behavior as a result of adoption at different ages
11. Recognize ways adopting parents can assist the adjustment of children from an ethnic/national group different from their own
12. Know the developmental periods that are particularly challenging for children who have been adopted and for their parents
13. Understand the risks of repeated or prolonged foster care arrangements
14. Understand the risks to child development and behavior of group care in large institutions
15. Recognize the challenges to parent-child interactions that may be associated with using modern forms of assisted reproduction
16. Understand the effects of sibling number and order on children's developmental and behavioral challenges
17. Recognize the influences of parental divorce on child development and behavior at different ages
18. Know child factors that affect adjustment to foster care
19. Understand the challenges faced by homosexual parents and their children and be able to counsel about how to manage those challenges
20. Recognize the special challenges to children's development and behavior

of parenting without a partner as a result of choice, death, or divorce
21. Know the typical developmental and behavioral problems that occur commonly among international adoptees
22. Understand the potential problems and benefits pertaining to child behavior and development of multigenerational households

B. Functional factors
1. Know the impact of parental depression on young children (eg, childhood depression, delayed language development)
2. Know the genetic and familial risk of various psychiatric disorders in children whose parent(s) have the disorder (eg, schizophrenia, obsessive compulsive disorder)
3. Know the impact of parental intellectual ability and educational attainment on child development and behavior
4. Understand the developmental and educational challenges posed by being from an immigrant family
5. Understand the influence of religious affiliation and values on children and families
6. Understand the effects on children's development and behavior of a chronic physical illness or disability in a parent or grandparent
7. Recognize the protective factors associated with the integrity and functioning of the nuclear and extended family
8. Recognize children's loyalty to and protection of their parents even in the face of neglect or abuse
9. Understand the vulnerabilities of siblings in a family in which a child has a chronic health condition or disability
10. Distinguish among permissive, restrictive, and authoritative parenting
11. Understand the mechanisms by which parental conflict affects children's behavior and development
12. Recognize the effects of temperament variations on the relationships among siblings
13. Identify predictors of successful adolescent functioning
14. Understand the effects on children of parental substance and/or alcohol use disorders

C. Socioeconomic diversity
1. Recognize the influences of poverty on the epidemiology of developmental and behavioral disorders
2. Know the range of recommendations that might be made to help families with limited resources facilitate their child's development
3. Know the additive disadvantage for children with disability, minority group membership, poverty, and social isolation

D. Societal factors
1. Geographic
   a. Understand common stresses on families living in urban areas

b. Understand common stresses on families living in rural/isolated areas

2. Education
   a. Understand the significance of school-community relationships in the educational success of children
   b. Understand how community values influence educational processes in schools
   c. Understand the impact of parental involvement in schools for educational success of children
   d. Understand issues associated with each of the various school arrangements that are available to families (eg, home schooling, charter schools, private and parochial schools)
   e. Understand how the structure and administrative characteristics of schools can be associated with school behavior and learning problems
   f. Know the effects of full-day kindergarten for children's development
   g. Know the benefits of early childhood education programs (eg, Head Start) on children's development
   h. Know the benefits of Early Intervention Programs on children's development
   i. Understand the impact of different types of childcare on child development
   j. Know how to advise a school about developing programs to minimize bullying and to handle any incident of bullying for both the victim and the bully
   k. Understand the child, school, and family factors which contribute to school refusal
   l. Know how to work collaboratively with schools and families to design an intervention plan for a youngster with school refusal
3. Media
   a. Know the health problems associated with TV or video watching
   b. Understand the risks of excessive media coverage of student suicide
   c. Understand the influences of media exposure on child development and behavior
   d. Know the mechanisms by which TV or video watching contributes to health problems
4. Discrimination
   a. Recognize the effects of stigmatization and discrimination on children and families who are in some way different from their community
   b. Know the range of recommendations for parents to help children to recognize, confront, and protect themselves from the damaging effects of social discrimination
   c. Know the range of recommendations for schools to minimize

isolation and stigmatization of children with individual or family differences

5. Violence
   a. Know the detrimental effects on children of being a witness, victim, or perpetrator of domestic or community violence
   b. Understand the co-occurrence of spouse or partner violence and violence against children
   c. Know how to recognize signs of a child being the victim of a bully
   d. Know how to counsel a family about helping their child cope with a bully either at school or in the community
   e. Know the factors that contribute to a child becoming a bully
   f. Know the impact of relational aggression and ostracism on a child
6. Housing
   a. Recognize the influences of homelessness on child development and behavior
7. Exposure to disasters
   a. Know recommendations for helping children and families in the face of natural disasters (eg, flood, hurricane, fire)
   b. Know recommendations for helping children and families cope with the occurrence of human disasters (eg, mass shootings, automobile or bus accidents)
   c. Understand common behavioral symptoms of functional and dysfunctional coping in the face of a disaster
   d. Know recommendations for helping community groups (eg, schools) cope with the occurrence of disasters
8. Access to health care
   a. Understand the risks to child development of inadequate access to health care
   b. Understand the potential contribution of the "medical home" concept to promoting optimal child development and behavior

3. FAMILY AND SOCIETAL FACTORS

3. A. Structural Factors

1. **Recognize common challenges to the psychosocial development of children in blended families (eg, insecurity associated with loss of a parent, difficulties with attachment to a new parent, reactions to new siblings, inconsistent discipline practices, etc)**

Five common challenges a blended family will face

1. **Blending different family traditions**

When two households blend, everyone will be coming with different traditions. Before you talk to your kids, be sure to talk to your partner about what's important and what can be compromised upon. Never assume the feelings of your partner, your kids, or your partner's kids.

Kids especially may have different expectations around holidays and birthdays. Without proper introduction or preparation, they may feel resentful about having to follow someone else's way of celebrating.

Try to compromise, split time equally between parents, and create new traditions together as a blended family.

2. Helping kids adjust to change

Too many changes at one time can be unsettling. Children thrive off of routine, so set a schedule and stick to it as much as possible. Having clear expectations and outlining what their school weeks will look like — Monday you'll be with your mom, Tuesday dad will pick you up, for example — will help your kids adjust.

Changes	Possible adjustments
New space or home	Make sure kids have their own independent space they feel safe in, such as personal room, play space, or personalized nook.
Moving	Allow kids to have a permanent space for things,

between two homes	even when they aren’t there so they don’t feel like a visitor.
New school	If possible, allow them time to adjust to the new family routine before starting school again.
New schedules	Have a conversation with children a few days before the new schedule starts. Be sure to plan individual attention time, if needed.

For older children, set up conversations before making decisions so they feel they have agency or input in what’s going on.

3. Sibling rivalry

Some kids will be excited about having stepsiblings, while others may initially resent it. Jealousy and conflict may arise quickly in the transition to living together.

You can help ease the transition by:

- setting expectations and rules about respecting each member of the family
- posting house rules that apply to all family members somewhere everyone can see them
- making sure everyone has their own space where they can be alone when they need some space
- displaying pictures of all the kids around your home
- planning activities like a beach or theme park outing everyone will enjoy

It might also be a good idea to trial what living together will be like by going on vacation. A camping trip is a great way to see how siblings interact with each other.

4. Compromising with parent discipline styles

You and your partner may have different discipline styles. The rules in your house might also not match those at your ex-partner's. It's important to get on the same page and follow the same rules before you marry and live under one roof.

The following steps may help:

- prioritize being civil and respectful
- let the main parent remain the prime discipliner until the stepparent has solid bonds with their stepchildren
- avoid ultimatums or disciplining when your partner isn't around
- a stepparent can serve as more of a friend or counselor instead of a disciplinarian
- list and post family rules and be consistent about following them
- make clear that rules in your house may be different than at your ex-partner's home and that's OK
- limit expectations from your partner

5. Managing age differences

Family members of different ages and stages will have different needs. They may also adjust differently to the new family dynamic.

Understanding frustrations and honoring differences can go a long way in a blended family. For example, don't make assumptions or place expectations on older children to look after the younger ones right away. Let them adjust to the new family dynamics first, and ask if that's something they're interested in.

Age differences to be aware of

- **Under 10.** They may adjust more easily, need more attention from parents, and have more basic daily needs.
- **Ages 10 to 14.** They may be more sensitive to feelings, need more reassurance than little ones, and need more time to bond.

- **Ages 15 and older.** They may be less involved in family time, may not openly express as easily, and need a sense of agency and equal respect.

  Spending time individually with your kids, when possible, to listen to their concerns may also help. If you're living with your partner's children for the first time, plan to spend time getting to know them individually, too.

  Avoid traditional thinking or using your background as a blueprint. These expectations can set your blended family up for more challenges. It's important to recognize that you or your partner isn't replacing anyone but setting forth new relationships of trust and communication.

  For example, if your stepchild is accustomed to a stay-at-home mom, they may need more attention and guidance from a parental figure when first moving in.

  Learning to understand racial and cultural differences can make a huge difference when it comes to bonding with your partner and their children. For people of color in the United States, representative role models in their life are particularly important. This could mean finding a family doctor, after-school coaches, extracurricular instructors, or even play groups that match their background

https://www.healthline.com/health/parenting/blended-family-tips#common-challenges

2. Understand the typical profile of children whose parents are gay or lesbian compared to children whose parents are heterosexual with respect to their academic achievement, psychological adjustment, and psychosexual development

Historically there have been three major concerns about the influence of lesbian and gay parents on children.

1. The first of these fears is that development of sexual identity will be impaired among children of lesbian and gay parents.
2. A second category of concerns involves aspects of children's personal development other than sexual identity
3. A third category of specific fears expressed by the courts is that children of lesbian and gay parents may experience difficulty in social relationships

Sexual Identity

Three aspects of sexual identity are considered in the research: gender identity, which concerns a person's self-identification as male or female; gender-role behavior, which concerns the extent to which a person's activities, occupations, and the like are regarded by the culture as masculine, feminine, or both; and sexual orientation, which refers to a person's choice of sexual partners, who may be homosexual, heterosexual, or bisexual

Gender Identity. In studies of children ranging in age from 5 to 14, results of projective testing and related interview procedures have revealed that development of gender identity among children of lesbian mothers follows the expected pattern There was no evidence in any of the studies of gender identity of any difficulties among children of lesbian mothers. No data have been reported in this area for children of gay fathers.

Gender-Role Behavior. A number of studies have reported that gender-role behavior among children of lesbian mothers fell within typical limits for conventional sex roles For instance, Kirkpatrick and her colleagues (1981) found no differences between children of lesbian versus heterosexual mothers in toy preferences, activities, interests, or occupational choices.

Lesbian mothers were no more and no less likely than heterosexual mothers to report that their children often played with "feminine" toys such as dolls. In both family types, however, children's sex-role behavior was seen as falling within the expected range.

The research suggests that children of lesbian mothers develop patterns of gender-role behavior that are much like those of other children. No data are available regarding gender-role behavior for children of gay fathers.

Sexual Orientation. The great majority of offspring of both lesbian mothers and gay fathers described themselves as heterosexual. Taken together, the data do not suggest elevated rates of homosexuality among the offspring of lesbian or gay parents. relationship.

Other Aspects of Personal Development

Studies of other aspects of personal development among children of lesbian and gay parents have assessed a broad array of characteristics. As was the case for sexual identity, studies of these aspects of personal development have revealed no major differences between children of lesbian versus heterosexual mothers. One statistically significant difference in self-concept emerged in Patterson's (1994a) study: Children of lesbian mothers reported greater symptoms of stress but also a greater overall sense of well-being than did children in a comparison group Overall, the belief that children of lesbian and gay parents suffer deficits in personal development has no empirical foundation.

Social Relationships

The most common focus of attention has been on peer relations, but some information about children's relationships with adults has also been collected. Reports by both parents and children suggest typical patterns of development of peer relationships. For example, as would be expected, most school-aged children reported same-sex best friends. The number and quality of adolescents' and young adults' romantic relationships has also been found to be unrelated to maternal sexual orientation (Tasker & Golombok, 1997; Wainright et al., 2004). No data on the children of gay fathers have been reported in this

area.

Summary

Results of research to date suggest that children of lesbian and gay parents have positive relationships with peers and that their relationships with adults of both sexes are also satisfactory. Fears about children of lesbians and gay men being sexually abused by adults, ostracized by peers, or isolated in single-sex lesbian or gay communities have received no support from the results of existing research.

3. Understand typical reactions that children whose parents are separated or divorced have to their families at various developmental stages

Children who have experienced their parents' divorce display a range of emotional and behavioral reactions in the months following the event. Following their parents' separation, children may regress, display anxiety and depressive symptoms, appear more irritable, demanding and noncompliant, and experience problems in social relationships and school performance (5). Parents often feel troubled by and unprepared for their children's reactions to a separation and divorce.

Children need to know that they are not responsible for the separation, that they are loved by both parents, and that their needs will be met. Children's expression of distress differs from that of adults.

Compared with adults, children may be more open to communication through books, workbooks, stories, play and drawings (6). Parents must achieve a balance between acknowledging and accepting the expression of negative feelings on the one hand, and providing clear, consistent rules and structure on the other.

he research suggests that children's responses to their parents' divorce and separation vary widely. Indeed, some children may become happier and less distressed when their parents separate (Amato 1994).

Nonetheless, studies have identified general pathways of children's reactions in the first two years after parental separation and divorce, based on gender and stage of development (age) (see citations in Hodges 1991; Amato 1994). Almost no research exists on infants' or college-aged children's responses. For children between these ages, the responses can be summarized as follows.

- **Preschoolers** (2 to 5 years). These children are too young to grasp the meaning of divorce, and so are likely to become confused and fearful of losing their other parent too. They tend to blame themselves for their parents' divorce. Many regress developmentally, becoming aggressive and throwing tantrums, especially boys.
- **Younger elementary school-aged children** (5 to 8 years old). These children can understand the meaning of divorce enough to become depressed (Kelly 1988,

cited in Di Bias 1996; Hodges 1991), grief-stricken and sad over the loss of family. Many continue to wish for parental reconciliation. They may also feel profound conflict of loyalties (Peterson and Zill 1986, and Brady et al. 1986, cited in Fischer 1997). They are egocentric enough to see divorce as a personal rejection, but may be mature enough to place the blame elsewhere, usually on a parent. Studies show that children at this age may suffer in school and in their social relationships (Demo and Adcock 1988, and Bloom and Dawson 1991, cited in Di Bias 1996). Half of their teachers in one study reported behavior changes (see citation in Hodges 1991).

- **Older elementary school-age children** (9 to 12 years old). These children may also be depressed, sad and grief-stricken, but are also more likely to blame and be angry with one or both parents. Children at this age can also see the world from the parents' point of view, however, and may start to parent a struggling parent or younger siblings.
- **Adolescents** (12 to 16 years old) are less dependent on the family, and therefore divorce would seem to be less significant to them. Still, self-esteem drops for many teenagers (but more so for children) during parental divorce. It may lead adolescents to question their own future ability to maintain a long-term relationship with a partner, and many feel considerable anger towards one or both parents. Also, divorce may trigger delayed or accelerated entry into adolescence.

4. Recognize possible effects of new relationships of either parent, after divorce or separation, on children's behavior (eg, bedwetting, separation difficulties, attentional difficulties, depression, hostility or jealousy toward the new adult and/or children)

Children’s relationships with their parents often worsen after a divorce. Marital disruption creates distance between parents and children which can result in regression of childhood behaviors.
Each year, over a million American children suffer the divorce of their parents.
The stress of divorce damages the parent-child relationship for as many as 40 percent of divorced mothers. The support they receive from home is rated much lower by children of divorced parents than by children from intact homes, and these negative ratings become more pronounced by the time children are in high school and college.
Researchers have found that preschoolers are the children most vulnerable to divorce, due to their lack of cognitive ability to fully understand the divorce process (Wallerstein and Kelly 1979). Although there is limited study, research has shown that the short-term effects often include a high level of regression, acute separation anxiety, and abandonment issues. Children at such a young age often feel as though they are the blame for the parental dissolution. Yet it is CHILDREN OF DIVORCE 9 hard to predict what long term effects divorce has on preschoolers due to their age.

Other studies have shown that the effects may not be a significant as once thought

Li, J-C. A. 2007. "The Kids are OK: Divorce and Children's Behavior Problems." RAND Labor and Population Working Paper, WR-489. Santa Monica, CA.

5. Know how to counsel parents who are separating in order to minimize the adverse effects on their children

The key feature of **co-parenting relationships** is that they focus on what's best for the children. There are many kinds of co-parenting relationships. Explain to them to determine the nature of your co-parenting relationship which will depend on many factors, including how well you and the other parent get along.

- You and the other parent can only expect each other to do what is agreed to verbally or in writing.
- Meetings between you and the other parent are relatively formal—they take place in a neutral location (for example, a coffee shop) at specific times, and you usually have a list of issues to discuss.
- You and the other parent are not personally or emotional involved with one another.
- You and the other parent share little personal information with each other, unless it is related to parenting

As you learn to co-parent, remember to

- work to put aside your anger and cooperate to put your children's needs first
- be polite and treat the other parent with respect
- avoid sarcasm, rudeness and insults

The focus of the discussions should be on what your children need and what's best for your children. How can you meet your children's needs? You also need to be practical and realistic.

- The children's routines of school, extracurricular activities, contact with family and friends, discipline, and responsibilities ideally should remain as normal and unchanged as possible
- Parents should avoid overburdening their children with their own unhappiness/irritability
- Parents should continue to provide support, consistency, and love towards their children throughout the divorce process
    - This can be achieved by parents reaching out for support
- It is best to maintain relationships with both parents following separation/divorce
    - Exception to this would be severe martial conflict and/or abuse
- Parents should attempt to set aside their own negative feelings and develop a collaborative business-type relationship with their ex
    - Attempt to come to a shared parenting agreement via mediation/assessment services

- Assure the child that they are not responsible for the separation and that they are loved by both parents
- Joint Physical Custody is the most successful **IF** the parents have respect for one another, can communicate appropriately, and live in somewhat close proximity
    - This custody arrangement will not work with parents that are in frequent conflict
- Support groups for the child have been show to only be moderately effective; if the parent is struggling, the child will perceive this and tend to struggle as well
- Keep rules as similar as possible between the 2 homes

4. Recognize typical responses of children at different developmental stages to the loss of a parent by death or disappearance

A number of studies have been conducted in recent years (e.g., Anthony, [5] Bluebond-Langer, [23] Gibney, [58] Kane, [79] Koocher, [84] MenigPeterson and McCabe, [99] Piaget, [107] Pitcher and Prelinger, [108] Spinetta, [132] Tallmer et al. [135]) to determine how children at various ages comprehend death.

A fairly standard view was put forth by Nagy [104] in 1948. Analyzing the words and drawings of a relatively large sample (378) of Hungarian children who had been exposed to considerable trauma and death in the preceding few years, she conceptualized a three-stage model of awareness and linked the stages to approximate chronological ages.

Prior to about three years of age, children's cognitive and language development is too immature for them to have any concept of death. According to Nagy's:

STAGE 1 (roughly ages 3-5), death is seen as reversible; the dead are simply considered "less alive," in a state analogous to sleep. Young children functioning at what Piaget [107] termed the "preoperational" level of development will not generally recognize the irreversibility of death. [84,86,95]

STAGE2 (ages 5-9), children begin to comprehend the finality of death, but believe that it happens only to other people.

STAGE 3 (after age 10), the causes of death can be understood, and death is perceived as final, inevitable, and associated with the cessation of bodily activities. As is true in all child development, there is considerable age variation in attainment of the different stages and children may regress when emotionally threatened.

Prior to about six months of age, infants fail to respond to separation from their mothers because they have not yet developed the capacity for memory of a specific personal relationship. [33] The development of stranger anxiety, occurring at about six to eight months, signifies that an infant has established a true object relationship with its mother or primary caretaking figure. This reaction suggests that an infant is developmentally capable of retaining memory traces of his mother and is capable of responding to her absence with displeasure [133] and depression. [40] However, it is not until three or four years

of age that a child has a coherent mental representation of important attachment figures and has achieved object constancy.

Observational studies of children between about four years of age and adolescence have led to conflicting conclusions about the nature of children's grieving and about their ability to achieve a healthy outcome.

Some think that it isn't until adolescence that children have the capacity to tolerate the strong painful affects necessary for completing the separation process and that children are more likely to use immature defense mechanisms, such as denial, that interfere with adequate resolution of loss.

Others believe that after object constancy has been achieved (at three to four years of age), bereavement need not necessarily lead to enduring psychopathology. Increasingly, it is being recognized [27,55,81] that if the child has a consistent adult who reliably satisfies reality needs and encourages the expression of feelings about the loss, healthy adjustment can occur.

Bowlby, [24] emphasize the similarities between adults' and children's responses to loss and see an evolutionary basis for them. In Bowlby's view, the argument about children's capacity for "mourning" is in large part terminological, with many psychoanalysts restricting the use of "mourning" to psychological processes with a single outcome—detachment.

A number of clinicians and clinical researchers (e.g., Alexander and Adlerstein, [2] Bowlby, [27] Elizur and Kaffman, [46] McConville et al., [98] Rutter, [126] Van Eerdewegh et al. [l37]) report that the impact of relationship loss will be greater when it occurs at certain ages or stages than at others.

Rutter [126] and Bowlby [27] have found that bereaved children under the age of five are more susceptible than older children to pathologic outcomes. He speculates that children under the age of one or two are less distressed than bereaved older children because there has been less time to develop ties.

Early adolescence also appears to be a vulnerable time in terms of significant relationship loss. [21] Rutter [126], Van Eerdewegh et al., [137] and Wolfenstein [l42] found that the severely depressed children in their studies mostly seemed to be adolescent boys who had lost their fathers. In contrast, Hilgard et al., [70] using retrospective data, noted a number of outstandingly good adjustments among adults whose parental loss came between the ages of 10 and 15, preceded by a satisfactory home life.

Elizur and Kaffman's data [45,46,78] also suggest that although normal children are at risk following bereavement, preexisting emotional difficulties, in combination with other antecedent variables, may exacerbate symptoms during the early months following loss.

Clarification is needed on the kinds of emotional disturbances and troubled family relationships that place children at greater risk.

https://www.ncbi.nlm.nih.gov/books/NBK217849/

6. Understand the ways a single parent can best support her/his child(ren)

- **Show your love.** Remember to praise your child. Give him or her your unconditional love and support. Set aside time each day to play, read or simply sit with your child.
- **Create a routine.** Structure — such as regularly scheduled meals and bedtimes — helps your child know what to expect.
- **Find quality child care.** If you need regular child care, look for a qualified caregiver who can provide stimulation in a safe environment. Don't rely on an older child as your only baby sitter. Be careful about asking a new friend or partner to watch your child.
- **Set limits.** Explain house rules and expectations to your child — such as speaking respectfully — and enforce them. Work with other caregivers in your child's life to provide consistent discipline. Consider re-evaluating certain limits, such as your child's screen time, when he or she shows the ability to accept more responsibility.
- **Don't feel guilty.** Don't blame yourself or spoil your child to make up for being a single parent.
- **Take care of yourself.** Include physical activity in your daily routine, eat a healthy diet and get plenty of sleep. Arrange time to do activities you enjoy alone or with friends. Give yourself a "timeout" by arranging for child care at least a few hours a week.
- **Lean on others.** Work out a carpool schedule with other parents. Join a support group for single parents or seek social services. Call on loved ones, friends and neighbors for help. Faith communities can be helpful resources, too.
- **Stay positive.** It's OK to be honest with your child if you're having a difficult time, but remind him or her that things will get better. Give your child an age-appropriate level of responsibility rather than expecting him or her to behave like a "little adult." Keep your sense of humor when dealing with everyday challenges.

Be aware that some research has shown that teens in single-parent households have a higher risk of depression and lower self-esteem. https://www.mayoclinic.org/healthy-lifestyle/childrens-health/in-depth/single-parent/art-20046774

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8. Recognize common symptoms of attachment disorders at various ages

Signs and symptoms of (reactive) attachment disorder

Attachment disorder can negatively affect all areas of a child or adolescent’s life and development. There are two main types of reactive attachment disorder: inhibited and disinhibited. Not much research has been done on the signs and symptoms of this disorder beyond early childhood, however as children grow older they may develop either inhibited or disinhibited behavior patterns.

In some cases an adolescent will display symptoms of both types. Some common signs and symptoms may include:

Inhibited type:

- Detached
- Unresponsive or resistant to comforting
- Withdrawn
- Avoidant
- Shuns relationships with everyone

Disinhibited type:

- Indiscriminate sociability
- Inappropriately familiar or selective in choice in attachment figures
- Seeks attention from anyone
- Displays inappropriate childish behavior
- Frequently asks for help doing things
- Violates social boundaries

Additional symptoms:

Relationships: In relationships, a person who has RAD may be bossy, untrusting, manipulative, and controlling. They may have challenges giving or receiving genuine love and affection. Their unstable peer relationships are tenuous at best, as children and teens with RAD blame others for their mistakes or challenges.

Behavioral: Destructive, irresponsible, impulsive, and defiant behaviors. Children or teens with RAD may steal, lie, abuse others, start fires, behave cruelly to animals, or act in a self-destructive manner. They also may avoid physical contact with others, and engage in drug or alcohol abuse.

Moral: Teens with RAD may lack faith, compassion, and remorse for their actions.

Emotional: Children who have RAD may feel sad, moody, fearful, anxious, depressed, and hopeless. These children may display inappropriate emotional reactions.

Thoughts: Children and teens who have RAD may have negative beliefs about themselves, life, and other relationships. These children and teens are unable to understand the concept of cause and effect. Additionally, they may experience inattention and challenges with learning.

9. Understand the benefits and potential problems of kinship versus non-kinship adoption

-There is evidence of the wide use of kinship care as the preferred placement for foster children. By the year 2000, it was estimated that one-third of all foster placements were with relatives. (Urban Institute, 2000)

-Many studies show that relatives express stronger feelings than non-relative caregivers over their responsibility and willingness to do the following:

1) facilitate the child's relationship with birth family
2) assist with children's social/emotional development

3) help children deal with issues of separation and loss
4) parent the children
5) partner with the child welfare agency
6) discuss the child's behavior and adjustment in the home with parents
7) teach children to deal with future relationships with family members
-Research has shown that children in kinship care are more likely than those placed in non-kin care to preserve and continue their family relationships, culture and connections. (Pecora et al, 1999)
-Webster et al 2000 found that children placed in kinship care when initially removed from parents had fewer placement changes compared to children in non-kin care.

Children placed in guardianship care or adopted by relatives can experience a multitude of benefits, including placement stability and emotional wellbeing. Children in temporary care, whether residing with kin or non-kin, may be placed in multiple homes, but there is a greater possibility of multiple moves when a child resides with a non-relative. For instance, in Illinois, 66% of children who lived with non-relatives experienced one move or no moves at all during their first full year of care, compared to 85% of children living with relatives.

Despite the numerous benefits of kin guardianship and adoption, opponents believe that placing children with relatives can be harmful. For some opponents, this concern is further magnified by the fact that relatives do not undergo the same detailed screening as non-relatives in the kin adoption or guardianship process. Children who reside with kin are more likely to have contact with their biological parents than children in non-kin care, and that contact is less likely to be supervised
Recommendation that child welfare agencies should present kin guardianship and adoption as viable options for permanency as soon as children are placed into kinship care. Summary - relative caregivers clearly offer benefits of both placement stability and emotional wellbeing for children. As families pursue kin guardianship or adoption, they often encounter issues involving changes in family dynamics, financial hardship, and insufficient leave.

10. Understand issues related to children's development and behavior as a result of adoption at different ages

Infancy and early childhood
-At this stage of development, children do not understand reproduction or grasp the concept of time and space. Although the child may not completely understand the idea of having a birth mother or what it means to be adopted, this is still a good time to talk with the child about their birth story. This is an opportunity for parents to become comfortable using adoption language.

School Age

-At this stage, a child can understand the adoption process and that it makes them different from other children their age. The child may start to feel anger or sadness when thinking about being adopted. It is normal to see aggression, angry behavior, withdrawal or sadness and self-image problems for adopted children at this age.
-Control may be an issue as the child may feel that they had no control over their life as an adopted child. Fear of abandonment may lead to difficulty with life transitions such as starting school or after-school activities.

Adolescence
-The primary task of an adolescent is to establish an identity. The adopted adolescent may want to know more about their genetics and birth family history. If the adopted adolescent tries searching for more information about themselves and their past, this is normal and should not alarm the adopted parents.

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2804559/

As children grow up, they develop a positive sense of their identity, a sense of psychosocial well-being (1). They gradually develop a self-concept (how they see themselves) and self-esteem (how much they like what they see) (2). Ultimately, they learn to be comfortable with themselves. Adoption may make normal childhood issues of attachment, loss and self-image (2) even more complex. Adopted children must come to terms with and integrate both their birth and adoptive families.

Children who were adopted as infants are affected by the adoption throughout their lives. Children adopted later in life come to understand adoption during a different developmental stage. Those who have experienced trauma or neglect may remember such experiences, which further

Adoptive parents can facilitate and assist this natural grieving process by being comfortable with using adoption language (eg, birth parents and birth family) and discussing adoption issues (5).

INFANCY AND EARLY CHILDHOOD
During infancy and early childhood, a child attaches to and bonds with the primary care-giver. Prenatal issues, such as the length of gestation, the mother's use of drugs or alcohol, and genetic vulnerabilities, may, ultimately, affect a child's ability to adjust. The temperament of everyone involved also plays a role.
As a child approaches preschool age, he or she develops magical thinking, that is, the world of fantasy is used to explain that which he or she cannot comprehend. The child does not understand reproduction, and must first understand that he or she had a birth mother and was born the same way as other children (2,5).

Even though a child as young as three years of age may repeat his or her adoption story, the child does not comprehend it (3,5). The child must first grasp the concept of time and

space, which usually occurs at age four to five years, to see that some events occurred in the past, even though he or she does not remember them. The child must understand that places and people exist outside of his or her immediate environment.
Telling a child his or her adoption story at this early age may help parents to become comfortable with the language of adoption and the child's birth story.

Children need to know that they were adopted. Parents' openness and degree of comfort create an environment that is conducive to a child asking questions about his or her adoption (3).

SCHOOL-AGED CHILDREN
Operational thinking, causality and logical planning begin to emerge in the school-aged child. The child is trying to understand and to master the world in which he or she lives. The child is a problem solver. He or she realizes that most other children are living with at least one other biological relative (6). I

t is the first time that the child sees himself or herself as being different from other children. The child may struggle with the meaning of being adopted, and may experience feelings of loss and sadness (1,7). He or she begins to see the flip side of the adoption story and may wonder what was wrong with him or her; why did the birth mother place him or her up for adoption?

The child may exhibit feelings of being abandoned and angry (1,2). It is normal to see aggression, angry behavior, withdrawal or sadness and self-image problems (1,8) among adopted children at this age. The child attempts to reformulate the parts of his or her story that are hard to understand and to compensate for emotions that are painful (2). As a result, daydreaming is very common among adopted children who are working through complex identity issues (5,7).

Control may be an issue. A child may believe that he or she has had no control over losing one family and being placed with another. The child may need to have reassurance about day to day activities or may require repeated explanations about simple changes in the family's routine (5). Transitions may be particularly difficult. The child may have an outright fear of abandonment, difficulty falling asleep and, even, kidnapping nightmares (1).

It is helpful to explain that the birth mother made a loving choice by placing the child up for adoption, that she had a plan for his or her future. The child may need to hear this statement repeatedly. There is some similarity between the symptoms of grief and symptoms associated with attention deficit/hyperactivity disorder; care givers must be wary not to label a child with attention deficit/hyperactivity disorder when, in fact, the child's behavior is consistent with a normal grieving process (9).

A parent's patience and understanding are crucial at this point of an adopted child's life. Parents may be pro-active by educating school personnel about the natural grieving issues related to adoption that their child is experiencing.

ADOLESCENCE

The adolescent's primary developmental task is to establish an identity while actively seeking independence and separation from family (2). The adopted adolescent needs to make sense of both sets of parents, and this may cause a sense of divided loyalties and conflict (7).

In early adolescence, the loss of childhood itself is a significant issue. The adopted adolescent has already experienced loss, making the transition to adolescence even more complicated (1,7). This period of development may be difficult and confusing. Adolescents may experience shame and loss of self-esteem, particularly because society's image of birth parents is often negative (2).

Adopted adolescents will want to know details about their genetic history and how they are unique. They will reflect on themselves and their adoptive family to determine similarities and differences. They will attempt to ascertain where they belong and where they came from (7). All adolescents may have a natural reticence about talking to their parents, and adopted adolescents may not share questions about their origins with their parents. They may keep their reflections to themselves. Adopted adolescents' search for information about themselves is very normal, and parents should not see this as a threat. Instead, parents' willingness to accept their child's dual heritage of biology and environment will help their child to accept that reality (7).

Children's interest in adoption varies throughout the developmental stages of childhood and adolescence. As children progress from one stage to another, they gain new cognitive abilities and psychosocial structures. They look at adoption differently and, often, have more concerns or questions. Their questions may diminish until a new cognitive and psychosocial level is reached. Parents can facilitate this developmental process by being knowledgeable and supportive, and by continuing to retell their child his or her adoption story. The grief that their child experiences is real and should not be denied or avoided. Support from knowledgeable health care providers is invaluable in helping adoptive parents and their child. Although this statement has addressed common issues that relate to a child's perception of adoption, a psychological or psychiatric referral is indicated if the child suffers from depression, or has symptoms that affect his or her day-to-day functioning. Pediatricians and other professionals who care for children should provide anticipatory guidance by counselling parents of adopted children about relevant issues that concern their child's understanding of his or her adoption.

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2804559/#:~:text=Adoption%20may%20make%20normal%20childhood,the%20adoption%20throughout%20their%20lives.

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11. Recognize ways adopting parents can assist the adjustment of children from an ethnic/national group different from their own

Four cultural socialization strategies

1) Cultural assimilation

-Cultural assimilation is the process in which a minority group or culture comes to resemble a dominant group or assume the values, behaviors, and beliefs of another group. This may occur with little to no effort from the parent because the child is constantly exposed to the majority culture.

2) Enculturation

-Enculturation is the gradual acquisition of the characteristics and norms of a culture or group by a person. In this strategy, parents make an effort to teach their child about their birth heritage and culture through educational, social and cultural opportunities.

3) Racial inculcation

-In this strategy, parents teach their child coping skills for encountering racism and discrimination.

4) Child's choice

-Child's choice is when the parent provides the child with cultural opportunities initially but overtime allows the child to decide if this is something they would like to continue to pursue.

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2366972/

12. Know the developmental periods that are particularly challenging for children who have been adopted and for their parents

Late Elementary school age children demonstrate common problems include hyperactivity, poor school performance, low self-esteem, aggression, defiance, stubbornness, troubled relationships with brothers and sisters, friends, and parents, lack of confidence, fearfulness, sadness, depression, and loneliness. Adoptive parents wonder whether and how much these problems are caused or influenced by adoption or a history of faulty attachment.

During the elementary school-age years, children's identity comes from a combination of their genetic heritage, their experience with their families, and what happens to them as they try to find their place in the wider world. They want to be like their peers and their families.

Adolescence is a time of trying on and choosing in all aspects of life. Two major aspects of adult identity formation will be choice of work and choice of a partner to love. Teenagers look for and imitate role models. They critically examine their family members and have identity concerns and authority struggles with their parents or other adults

Adolescents often express their reactions to loss by rebelling against parental standards. Knowing that they have a different origin contributes to their need to define themselves autonomously.

Adopted adolescents have the same trouble searching for a comfortable identity as do nonadoptees. Problems involving aggression, sexual activities and pregnancy, delinquency and substance abuse, social isolation and depression are the most common ones faced by teenagers and their families.

13. Understand the risks of repeated or prolonged foster care arrangements

- Studies of former foster children revealed that their level of education is below average. The younger the age of the child at placement, the fewer the years of schooling obtained. This is concerning because education level is related to employment and socioeconomic status as an adult. Also of concern, studies have shown that a disproportionate number of the homeless population are individuals who have spent time in the foster care system. It has been found that children in foster care who have had fewer placements, function better as adults.

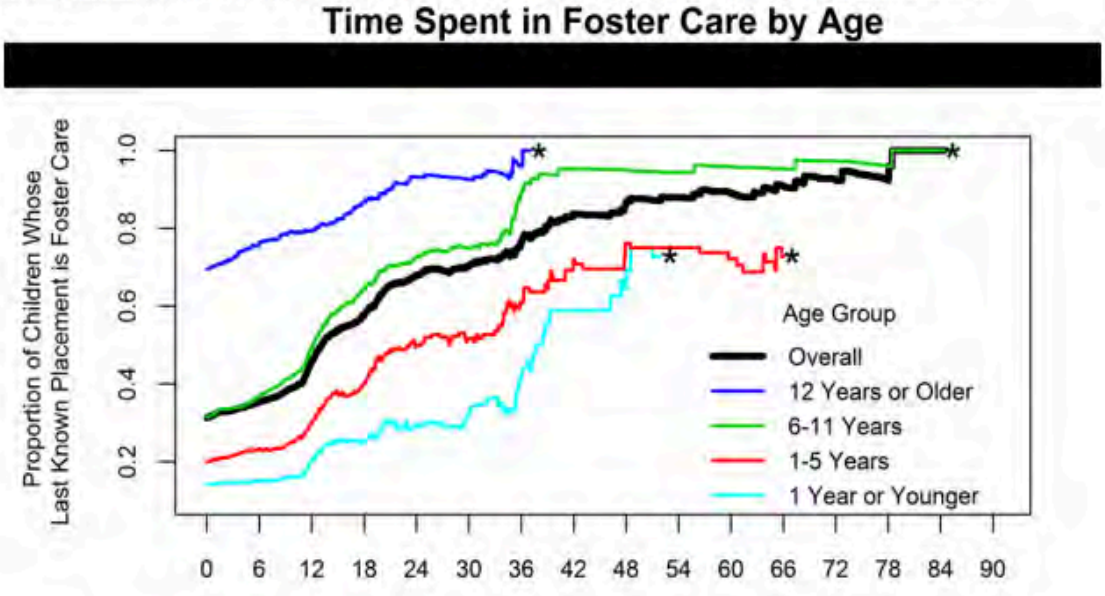
- https://www.irp.wisc.edu/publications/focus/pdfs/foc142g.pdf
- Pecora, P., LeProhn, N., Nasuti, J., (1999). In Hegar, R & Scannapieco, M. (Eds), Kinship Foster
- care (pp. 154-178). NY: Oxford University Press.
- Urban Institute (2000). Report to the Congress on Kinship Foster Care: Part I and II. From the US
- Department of Health and Human Services.

-
- Webster, D., Barth, R., Needell, B. (2000). Placement stability for children in out-of-home care: A longitudinal analysis. Child Welfare, 79 (5), 615-632.
-

Children who are adopted when they are older are more likely to have experienced prior abuse and/or neglect, strong predictors of behavioral and emotional problems (Dance, Rushton, & Quinton, 2002; Garland et al., 2001; McMillen et al., 2005; Simmel, 2007).

- Older children are also more likely to have experienced multiple placements, which are associated with poor adjustment into the adoptive placement (Berry & Barth, 1989), increased internalizing and externalizing problems (Newton, Litrownik, & Landsverk, 2000; Rosenthal & Groze, 1994), and greater mental health service utilization (Rubin et al., 2004).
- Longer periods of time in foster care are associated with greater risk for remaining in foster care instead of achieving permanency.
- Children 12 years or older placed in foster care after a child maltreatment investigation are at particularly high risk for living in long-term foster care.
- Permanency planning efforts are needed to target children at risk for long-term foster care placements. These efforts are particularly critical for children who are placed in foster care as teenagers.
- After spending 12 to 18 continuous months in foster care, children's chances of leaving foster care rapidly decreased. After 36 to 42 months of continuous time spent in foster care, a child's chances of leaving foster care are incredibly low.

Figure 1. Proportion of children whose last known placement is foster care by time spent in care and age



https://www.acf.hhs.gov/sites/default/files/opre/nscaw_ltfc_research_brief_19_revised_for_acf_9_12_13_edit_clean.pdf

14. Understand the risks to child development and behavior of group care in large institutions

- Institutional affect: Children exposed to institutional care often suffer from "structural neglect" which may include minimum physical resources, unfavorable and unstable staffing patterns, and social-emotionally inadequate caregiver-child interactions.
- Developmental Delays and Deviance: Children raised in institutions often suffer from dramatic developmental delays and may follow deviant developmental pathways.
- Hormonal Axis Changes: Atypical patterns of diurnal cortisol activity for children living in institutions were first reported by Carlson and Earls (1997).
- Challenges with Cognitive Development: Recent research continues to show the delayed cognitive performance of children in residential care
- Unstable Attachment: Institution-reared children all experience separation from or loss of their birth parents and other caregivers. In a famous report on institutions for the World Health Organization, Bowlby (1952) concluded that children suffered from the effects of institutional care, even when their physical needs (food, clothes, etc.) were adequately met.

- The evidence for and against the existence of a distinctive set of co-occurring developmental problems in institutionalized children is weighed and found to not yet convincingly demonstrate a "post-institutional syndrome"
- Children exposed to institutional care do not receive the type of nurturing and stimulating environment needed for normal growth and healthy psychological development.
- institutions into four levels, based on the quality of care they provide:
    - (1) institutions characterized by global deprivation of the child's health, nutrition, stimulation, and relationship needs;
    - (2) institutions with adequate health and nutrition support, but deprivation of the child's stimulation and relationship needs
    - (3) institutions that meet all needs except for stable, long-term relationships with consistent caregivers.
    - (4) institutional environment that provides for stable and consistent caregiving, and only deprives children of a regular family life embedded in a regular social environment.

Despite the variability in care that can be found among institutions, it is possible to put together a composite description of what is typical. T

- Group sizes tend to be large (typically 9–16 children per ward, although in extreme cases the number may approach 70). The number of children per caregiver is large (approximately 8:1 to 31:1, although a few institutions have fewer children per caregiver).
- Groups tend to be homogeneous with respect to ages and disability status. Children are periodically "graduated" from one age group to another perhaps as many as two or three times in the first two or three years of life.

- Caregivers for any single child tend to change constantly because there may be a high staff turnover; caregivers may work long shifts (e.g., 24 hours) and be off three days; caregivers may not be consistently assigned to the same group; and caregivers may get up to two months vacation. The result is that a child may see anywhere from 50 to 100 different caregivers in the first 19 months of life.
- Other adults tend to come and go in children's lives, including medical and behavioral specialists, prospective adoptive parents, and volunteers who may visit for only a week or a few months.
- Caregivers typicallyreceive little training, and the training they do receive is more focused on health issues than on social interaction. They spend the vast majority of their hours feeding, changing, bathing, cleaning children and the room, and preparing food rather than interacting with the children. Caregivers are invariably female, so children rarely see men.
- When caregivers perform their caregiving duties, it is likely to be in a business-like manner with little warmth, sensitivity, or responsiveness to individual children's emotional needs or exploratory initiatives.

15. Recognize the challenges to parent-child interactions that may be associated with using modern forms of assisted reproduction

Previous studies on the impact of assisted reproductive technologies (ART) are divided in three sections: (1) cognitive, motor, and language developments, (2) behavior problems and socio-emotional development, and (3) parent-child relationship.

Gibson et al. (1998) indicated that IVF mothers tended to report more children's behavioral problems.

ART parents expected their children, more than naturally conceived children's parents, to adjust to their own and others' expectations (Colpin and Soenen, 2002)

ART suggested that firstly the inability to conceive children was experienced as a stressful situation by individuals and couples, and secondly, parents who conceived using ART experienced additional stress and anxiety during the assisted conception procedure (

Overall the vast majority of the data above suggested that ART parents, especially ART mothers, showed positive parental attitudes and more protection to their children throughout their development.

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3829644/

16. Understand the effects of sibling number and order on children's developmental and behavioral challenges

Large Scandinavian register datasets that became available to researchers beginning in the late 1990s have enabled birth order research, as they contain population data on both family structure and a variety of child outcomes.

Average educational attainment was lower in larger families largely because later-born children had lower average education, rather than because firstborns had lower education in large families than in small families.

Later-born children have lower IQs, on average

Firstborn children are significantly more likely to be employed and to work as top managers, while later-born children are more likely to be self-employed.

The effects of birth order on health are less straightforward than other outcomes we have examined

https://www.nber.org/reporter/2017number4/black.html

17. Recognize the influences of parental divorce on child development and behavior at different ages

Children who have experienced their parents' divorce display a range of emotional and behavioral reactions in the months following the event. Following their parents' separation, children may regress, display anxiety and depressive symptoms, appear more irritable, demanding and noncompliant, and experience problems in social relationships and school performance (5). Parents often feel troubled by and unprepared for their children's reactions to a separation and divorce.

Children need to know that they are not responsible for the separation, that they are loved by both parents, and that their needs will be met. Children's expression of distress differs from that of adults.

Compared with adults, children may be more open to communication through books, workbooks, stories, play and drawings (6). Parents must achieve a balance between acknowledging and accepting the expression of negative feelings on the one hand, and providing clear, consistent rules and structure on the other.

he research suggests that children's responses to their parents' divorce and separation vary widely. Indeed, some children may become happier and less distressed when their parents separate (Amato 1994).

Nonetheless, studies have identified general pathways of children's reactions in the first two years after parental separation and divorce, based on gender and stage of development (age) (see citations in Hodges 1991; Amato 1994). Almost no research exists on infants' or college-aged children's responses. For children between these ages, the responses can be summarized as follows.

- **Preschoolers** (2 to 5 years). These children are too young to grasp the meaning of divorce, and so are likely to become confused and fearful of losing their other parent too. They tend to blame themselves for their parents' divorce. Many regress developmentally, becoming aggressive and throwing tantrums, especially boys.
- **Younger elementary school-aged children** (5 to 8 years old). These children can understand the meaning of divorce enough to become depressed (Kelly 1988, cited in Di Bias 1996; Hodges 1991), grief-stricken and sad over the loss of family. Many continue to wish for parental reconciliation. They may also feel profound conflict of loyalties (Peterson and Zill 1986, and Brady et al. 1986, cited in Fischer 1997). They are egocentric enough to see divorce as a personal rejection, but may be mature enough to place the blame elsewhere, usually on a parent. Studies show that children at this age may suffer in school and in their social relationships (Demo and Adcock 1988, and Bloom and Dawson 1991, cited in Di Bias 1996). Half of their teachers in one study reported behavior changes (see citation in Hodges 1991).
- **Older elementary school-age children** (9 to 12 years old). These children may also be depressed, sad and grief-stricken, but are also more likely to blame and be angry with one or both parents. Children at this age can also see the world from the parents' point of view, however, and may start to parent a struggling parent or younger siblings.
- **Adolescents** (12 to 16 years old) are less dependent on the family, and therefore divorce would seem to be less significant to them. Still, self-esteem drops for many teenagers (but more so for children) during parental divorce. It may lead adolescents to question their own future ability to maintain a long-term relationship with a partner, and many feel considerable anger towards one or both parents. Also, divorce may trigger delayed or accelerated entry into adolescence.

18. Know child factors that affect adjustment to foster care

Problems in the foster child's prior history, particularly attachment disorders and the experience of replacements, affect the extent of adjustment to the foster family.

In-home visits by the child and the absence of parental permission to stay with the foster family are two factors related to the parent-child relationship that impeded adjustment. In general, parental problem factors did not affect adjustment

https://psycnet.apa.org/record/2009-18738-016

19. Understand the challenges faced by homosexual parents and their children and be able to counsel about how to manage those challenges
(from Linville D, O'Neil, Maya. https://www.aamft.org/Consumer_Updates/Same-sex_Parents_and_Their_Children.aspx. Accessed 10/420)

Most research studies show that children with two moms or two dads fare just as well as children with heterosexual parents. In fact, one comprehensive study of children raised by lesbian mothers or gay fathers concluded that children raised by same-sex parents did not differ from other children in terms of emotional functioning, sexual orientation, stigmatization, gender role behavior, behavioral adjustment, gender identity, learning and grade point averages. Where research differences have been found, they have sometimes favored same-sex parents.

For example, adolescents with same-sex parents reported feeling more connected at school. Another study reported that children in gay and lesbian households are more likely to talk about emotionally difficult topics, and they are often more resilient, compassionate and tolerant. The same concerns that face many heterosexual parents when they are deciding to have children also face same-sex parents including time, money, and responsibilities of parenthood. Likewise, many of the parenting tasks faced by same-sex parents are similar to those faced by heterosexual parents, such as providing appropriate structure for children, while also being warm and accepting, setting limits, teaching open and honest communication, healthy conflict resolution, and monitoring of child's peer network and extracurricular activities.

Some differences: adapting to different types of family forms, the impact of social stigma on the family, and dealing with extended family members who may not be supportive of same-sex parenting.

Challenges:

1.Living in a culture that supports heterosexist and homophobic attitudes and beliefs

2. These families are usually part of a blended family and include children from previous heterosexual marriages. Some of these families may deal with disagreement from other family members about the authenticity and validity of their family patterns.

3. Lack of support from a previous heterosexual partner or the other biological parent can cause major conflict and distress within the family system.

4. Lesbian and gay parented families may have concerns about discrimination in parenting and custody arrangements.

5. A parent's minority sexual orientation and/or gender identity status may be brought up in custody disputes as a reason to restrict or deny custody by the children's other parent and/or by the courts.

5. Relationships and problems with non-biological parent figures are common among lesbian and gay parented families simply due to the biological complexities involved with conceiving children when parents are the same sex.
6. In same-sex relationships, it is common for extended family to acknowledge intimate relationships differently from heterosexual relationships; this discriminatory treatment can be confounded by parenting relationships as well. Extended family may see parenting as a necessary step in validating a relationship for same-sex couples or they may view parenting with similar biased and discriminatory views, even denying one parent's relationship to the children.
7. Explaining relationship status and family make-up to school professionals, medical professionals, children's friends/parents, as well as explaining relationship status and family make-up to children, can be uniquely complex for same-sex parents. Though many family relationships may be complex, explaining family relationships is uniquely complex for lesbian and gay parented families because of the lack of societal norms and relevant examples in media, stereotyped notions about such relationships that are common, and the fear of discrimination faced by these families.
8. Competent parenting may be influenced by gay and lesbian parents' ability to accept and acknowledge their identity and how they are able to negotiate living in a heterosexist, homophobic, or otherwise discriminatory society, while rearing their children in a family unit that is not socially sanctioned
9. . Therapists acknowledge the prevalence both of homophobia that is experienced by the family as a result of the actions of others, as well as the existence of internalized homophobia and how this may impact families. Internalized homophobia is defined as a set of negative attitudes and affects toward homosexuality in other persons and toward homosexual features in oneself. Therapists will help illustrate to the family how homophobia could be impacting them.
10. Both internalized homophobia and experiences of outside discrimination may mean that families need more time in therapy to build rapport with the therapist and to feel comfortable disclosing personal and family-related concerns.

See previous responses in # 2

20. Recognize the special challenges to children's development and behavior of parenting without a partner as a result of choice, death, or divorce

See previous responses in # 3-5

- Understand typical reactions that children whose parents are separated or divorced have to their families at various developmental stages.

- There is no “perfect” age for a child’s parents to get divorced; their reactions are expressed in way consistent with their developmental stage
- Many children show behavior changes in the first year of parent separation
    - Most adjustment problems resolve 2- 3 years after the separation, the child’s sense of loss may last for years, especially on holidays, birthdays, and other special events
- **Infants:**
    - Due to change in routine, may be fussier, irritable, or listless; may have sleep and feeding disturbances.
    - May see increases in normal separation and stranger anxiety at about 6 months
- **Toddlers:**
    - Increases in separation anxiety and some regression in behavior
        - Example: loss of toileting and language skills as well as development of eating/sleep disorders
- **Pre-school aged children:**
    - Do not understand what is going on and often repeatedly ask for the separated parent
    - Often test and manipulate differences in limit setting between the separated parents
    - For children ~4-5 years old, they may blame themselves for the separation, begin acting out, have nightmares, have more reluctance to separate, and fear that they may be abandoned
- **School-aged children:**
    - Self-blame and asking/fantasizing about the reunion of the parents
    - Mood and behavior changes (withdrawal and anger) are frequent, school performance may decline, and the child may feel abandoned by the parent no longer living in the home
- **Adolescents:**
    - Aggressive delinquent behavior, withdrawal, substance abuse, inappropriate sexual behavior, and poor school performance
    - Increase in suicidal behavior
        - Junior high school–aged boys and girls living with divorced fathers
- Children are observant and quick to pick up on conflict between parents
    - Also quick to pick up on resolution of conflict
        - Children that have the opportunity to witness the resolution of problems learn importance of problem-solving skills
- Following parents’ separation, children may regress, display anxiety/depressive symptoms, appear more irritable, demanding and noncompliant, and experience problems in social relationships and school performance
    - Allow the child to express their feelings through alternative forms of communication: books, stories, drawings, etc.
- More trouble with dating, more cohabitation, greater likelihood of divorce, higher expectations of divorce later in life, and a decreased desire to have children
    - However, according to many studies negative effects of divorce were greatly mitigated when positive relationships with both parents were maintained
- Children who end up living in nonnuclear families are more likely to have a higher incidence of poor health, learning difficulties, attention-

deficit/hyperactivity disorder, emotional and behavioral difficulties, and emergency department visits

- Interventions such as counseling can help ameliorate these effects

21. Know the typical developmental and behavioral problems that occur commonly among international adoptees

See responses above in multi-racial adoptions

Hawk, B., & McCall, R. B. (2010). CBCL behavior problems of post-institutionalized international adoptees. Clinical child and family psychology review, 13(2), 199–211. https://doi.org/10.1007/s10567-010-0068-x

The major factor contributing to extreme behaviors is age at adoption, with those adopted after 6/18 months having more behavior problems, especially Internalizing, Externalizing, and Attention problems. Generally, samples of post-institutional children have more problems than samples of mixed or non-institutional internationally adopted children, and some problems are more likely to be manifest in adolescence, suggesting the effects of deficient early experiences are not simply the persistence of learned behavior but more general dispositions that become more noticeable or severe during adolescence. Findings are discussed in terms of early deficient social–emotional caregiver–child interactions that characterize most institutional environments as a possible major cause of later difficulties in post-institutionalized children.

22. Understand the potential problems and benefits pertaining to child behavior and development of multigenerational households

Children do well in married two biological parent households (e.g. McLanahan and Sandefur 1994)

Approximately 8+% of children live in a three-generation family household

Urban children born to single mothers, 60% live in three-generation family households during childhood

Multi-generational coresidence can be short duration and frequent transitions

Studies for **young children** are very mixed. Studies have found positive, negative and null outcomes for behavior (East and Felice 1996; Foster and Kalil 2007; Leadbeater and Bishop 1994; Pope et al 1993), and improved, worse, or no effect on cognitive outcomes (Kellam et al. 1977; Foster and Kalil 2007; Mollborn et al. 2011; Unger and Cooley 1992).

Studies of children in **middle childhood** have also found both positive and negative associations with child wellbeing and three-generation family coresidence (Pittman and Boswell 2008; Barbarin and Soler 1993; Sonuga Barke and Mistry 2000)

Living in an extended household has also been associated with lower rates of deviant behavior among teens (Dornbusch et al. 11 1984; Stolba and Amato 1993), improved educational outcomes and decreased likelihood of early labor force participation and independence (Aquilino 1996).

Developmentally verbal ability is better but behavioral skills are worse and it is associated with increased likelihood of being overweight

Low-income and minority children are more likely to live in three-generation family households and these are also the groups of children for whom gaps in school readiness are largest.

B. Functional factors

1. Know the impact of parental depression on young children (eg, childhood depression, delayed language development)

a. Eckshtain et al 2019
    i. Parental depression significantly related to higher levels of internalizing symptoms in children including impaired functioning and depression
        1. Children of parents with lifetime history of depression have 3x rate for MDD
    ii. Treatment and resolution of parental depression allows for greater success in intervention for the child
        1. When parents are currently exhibiting symptoms of depression, CBT unsuccessful for the child
b. Quevedo 2013
    i. Prolonged exposure to maternal depression (positive screen at 30-90 days and at 12 months post-partum) leads to lower averages on the Bayley language scale at 12mon old, as compared to initial depression only (30-90 days post-partum)

2. Know the genetic and familial risk of various psychiatric disorders in children whoseparent(s) have the disorder (eg, schizophrenia, obsessive compulsive disorder)

1. Know the genetic and familial risk of various psychiatric disorders in children whose parent(s) have the disorder (eg. schizophrenia, obsessive compulsive disorder)
    a. Asarnow and Forsyth 2013
        i. Childhood-onset schizophrenia has a greater aggregation of schizophrenia within first degree relatives compared to adult-onset schizophrenia
        ii. Multiple common and rare allelic mutations must combine to increase risk of schizophrenia
        iii. Childhood-onset schizophrenia shares genetic overlap with autism
    b. Hanna et al 2005
        i. Significantly increased rate of lifetime obsessions and compulsions if present in first-degree relative, specifically with ordering compulsions
        ii. Lifetime prevalence of OCD higher if first-degree relative with tic history
    c. Do Rosario-Campos et al 2005
        i. Childhood onset OCD with earlier onsets correlated to higher genetic loading and shared vulnerability with chronic tic disorders

3. Know the impact of parental intellectual ability and educational attainment on child development and behavior

Dubow, E. F., Boxer, P., & Huesmann, L. R. (2009). Long-term Effects of Parents' Education on Children's Educational and Occupational Success: Mediation by Family Interactions, Child Aggression, and Teenage Aspirations. Merrill-Palmer quarterly (Wayne State University. Press), 55(3), 224–249. https://doi.org/10.1353/mpq.0.0030

The benefits of parental educational level when the child is young are not limited to academic achievement throughout the school years, but have long-term implications for positive outcomes into middle adulthood (i.e., higher educational level, more prestigious

occupations). The positive effects of parental education are independent of other indices of parental SES (i.e., father's occupation, value of housing) and family process variables (i.e., negative family interactions), the positive effects of higher IQ, and the negative effects of child aggressiveness. The long-term positive effects of parent education appear to be indirect – mediated through adolescent aspirations and educational attainment – in contrast to the direct long-term effects of the child personal variables (IQ and aggressiveness).

4. Understand the developmental and educational challenges posed by being from an immigrant family

a. Linton and Green 2019 (AAP Policy Statement)
    i. "The Immigrant Paradox" – may arrive to the US healthier than native-born peers
        1. Resilience built from family structure, cultural background; therefore important to provide culturally competent care → including recognizing barriers to Americanized medicine vs. traditional treatments
    ii. Often have limited access to quality care with wrap-around services given language barriers and poor communication between the office and the family
        1. Especially worse for patients with special needs or complex care
    iii. Specific questions about migration to the US must be evaluated to determine if specific screenings may be necessary, if health records are available or need translating, family history or exposures, etc.
        1. Evaluate toxic stress given increased risk, nutritional needs/access to culturally relevant foods, special diets
    iv. Dual-language learners are less likely to be enrolled in high-quality early child care compared to peers, provide testing and resources to assess kindergarten-readiness

5. Understand the influence of religious affiliation and values on children and families

Pederson T. In https://psychcentral.com/news/2019/02/09/how-does-religion-impact-child-development/142770.html. Accessed 10.4.2020

Growing up in a religious household can have positive and negative effects for childhood development. Children raised in religious families tend to have better social and psychological skills but may perform less well academically, compared to their non-religious peers as found by, researchers from The University of Texas at San Antonio (UTSA) who analyzed data from the Early Childhood Longitudinal Study (ECLS)-Kindergarten Cohort. They looked at the effects of parents' religious attendance and how the religious environment in the household (frequency of parent-child religious discussions and spousal conflicts over religion) influenced a nationally representative sample of third-graders.They also reviewed the children's psychological adjustment, interpersonal skills, problem behaviors, and performance on standardized tests in reading, math, and science.

The findings show that third-graders' psychological adjustment and social competence were positively correlated with religion.. However, students' performance on reading, math, and science tests were negatively tied to several forms of parental religiosity and especially math and science.

Religious solidarity among parents and communication between parent and child were linked to positive development characteristics while religious conflict among spouses was connected to negative outcomes.

A major takeaway from the study is that religion can be an important influence, generally for good and sometimes for ill, as children navigate their way through the grade school years, said Bartkowski.

6. Understand the effects on children's development and behavior of a chronic physical illness or disability in a parent or grandparent

Sieh, D. S., Meijer, A. M., Oort, F. J., Visser-Meily, J. M., & Van der Leij, D. A. (2010). Problem behavior in children of chronically ill parents: a meta-analysis. Clinical child and family psychology review, 13(4), 384–397. https://doi.org/10.1007/s10567-010-0074-z

Children with parents who have chronic medical conditions appear to be at increased risk for internalizing and externalizing problem behavior. In view of the high prevalence of parental CMC in the population, the number of children at risk for depressive, anxious and somatic symptoms may be large. Health care practitioners should be aware of this and refer children with clinical levels of problem behavior to professionals offering interventions (e.g., support groups, psychological counseling, psycho-education and family therapy). Medical doctors are recommended to receive education about how illness can impact on families and how to treat undesirable behaviors and emotions of family members (Gorter et al. 2010). Counselors should also take notice of therapeutic landscapes specific to the treatment of a certain diagnosis and illness stages, for example, the need for psycho-education at the onset of muscle disease (Sperry 2009). Specific treatments for clinically elevated levels of problem behavior in children with parental

CMC are required and should be evaluated in randomized control studies. Most importantly, internalizing problems are prevalent among children with parental chronic illness. Young families, those dealing with an ill parent with long illness duration and families with few financial resources may have an increased need for support. Standard screening of children in the target group may consist of assessing demographic risk factors and paying special attention to risks for specific problem behaviors, for instance, single parenthood seems to pose a risk for externalizing problem behavior. Screening children soon after the parent has been diagnosed may be an important step for the prevention of persistent developmental problems. This may be achieved if professionals in contact with the target group (e.g., general practitioners, teachers, school doctors and counselors) are alert for potential problem behavior. Asking a few questions about children's adjustment during a consultation can be an important step to initiate help. Therefore, there should be a family-centered approach instead of focusing exclusively on parents (Visser-Meily et al. 2005a).

Important to -pay attention to children during the rehabilitation of the ill parent by informing them about the disease and assisting them in their needs.

7. Recognize the protective factors associated with the integrity and functioning of the nuclear and extended family

Jæger MM. The Extended Family and Children's Educational Success. American Sociological Review. 2012;77(6):903-922. doi:10.1177/0003122412464040

Analyses using the Wisconsin Longitudinal Study show that, net of family factors shared by siblings from the same immediate family, and factors shared by first cousins account for a nontrivial part of the total variance in children's educational success. Results also show that grandparents', aunts', and uncles' socioeconomic characteristics have few direct effects on educational success.

Resources in the extended family compensate for lacking resources in low-SES families, which then promotes children's educational success. Overall effect of family in educational success originates in the immediate family, the extended family, and in interactions between these two family environments.

Living in an extended household has also been associated with lower rates of deviant behavior among teens (Dornbusch et al. 11 1984; Stolba and Amato 1993), improved educational outcomes and decreased likelihood of early labor force participation and independence (Aquilino 1996).

8. Recognize children's loyalty to and protection of their parents even in the face of neglect or abuse

"Emotional Abuse & Young Children", Florida Center for Parent Involvement (website:

http://lumpy.fmhi.usf.edu/cfsroot/dares/fcpi/vioTOC.html)

Children suffering from emotional abuse are often extremely loyal to the parent, afraid of being punished if they report abuse, or think that this type of abuse is a normal way of life.

9. Understand the vulnerabilities of siblings in the family in which a child has a chronic health condition or disability

Milevsky A. In https://www.psychologytoday.com/us/blog/band-brothers-and-sisters/201406/siblings-children-disabilities. Accessed 10/5/2020

a. Greater risk than average of developing emotional issues, anxiety, and stress and may be an attempt by these siblings to hide their problems. They may want to be well-behaved or protect their already overburdened parents. Become overly responsible and independent.
b. Peer problems, as well as a lack of engagement in extracurricular activities and academic issues as a result of limited time and money.
c. Considering the attention given to the child with the disability, siblings may neglect their own issues.
d. In some cases, siblings experience parentification where they are expected to have many responsibilities for themselves and their sibling, developing duties similar to those of a parent and overlooking their need to act like children.
e. Siblings may feel neglected by parents as attention is taken away from them. Time spent on medical and therapy appointments and parental emotional energy for the child with the disability limits the amount of time and emotional energy parents can spend with the other siblings, resulting in their feeling neglected.
f. Siblings may have similar questions about the sibling with the disability as do parents but have little information or resources available to them. They may have many unanswered questions about their sibling, including whether their disability can be transmitted and what will be in the future. With little or no information, siblings may develop their own ideas about what is happening which may be worse than what is.
g. They may feel guilty wondering if they caused the disability of their sibling, or they may feel guilt about why the disability did not happen to them.
h. They may feel fear about the health of their sibling or about what may happen to their sibling in the future.
i. Siblings may also experience resentment, anger, or jealousy towards their sibling, considering the attention and resources expensed on their sibling.

j. There may be embarrassment as a result of the behaviors and appearance of their sibling. In some cases, the embarrassment may be so great that they disassociate from the sibling with the disability. They may claim to be an only child or may not invite over friends so that they do not have to answer questions about their sibling.
k. POSITIVE-Siblings often develop certain positive characteristics such as self-control, cooperation, empathy, tolerance, altruism, maturity, and responsibility . They may have loyalty and a protective attitude towards their sibling. In some cases, these siblings use someone's attitude about special needs as a test for screening friends and mates. Their involvement with their sibling may even lead them to choose jobs in the helping professions.

10. Distinguish between permissive, restrictive and authoritative parenting
https://www.apa.org/act/resources/fact-sheets/parenting-styles Accessed 10.5.20

Turner, E.A., Chandler, M., & Heffer, R.W. (2009). The Influence of Parenting Styles, Achievement Motivation, and Self-Efficacy on Academic Performance in College Students. *Journal of College Student Development 50*(3), 337-346. doi:10.1353/csd.0.0073.

Authoritative

In this parenting style, the parents are nurturing, responsive, and supportive, yet set firm limits for their children. They attempt to control children's behavior by explaining rules, discussing, and reasoning. They listen to a child's viewpoint but don't always accept it.

Children raised with this style tend to be friendly, energetic, cheerful, self-reliant, self-controlled, curious, cooperative and achievement-oriented.

This style is associated with positively linked to academic performance; and authoritarian and permissive parenting were negatively associated with grades.

Permissive

In this parenting style, parents are warm, but lax. They fail to set firm limits, to monitor children's activities closely or to require appropriately mature behavior of their children.

Children raised with this parenting style tend to be impulsive, rebellious, aimless, domineering, aggressive and low in self-reliance, self-control and achievement.

Restrictive or Authoritarian

The parent is demanding but not responsive. Authoritarian parenting is a restrictive, punishment-heavy parenting style in which parents make their children follow their

directions with little to no explanation or feedback and focus on the child's and family's perception and status

11. **Understand the mechanism by which parental conflict affects children's behavior and development.**

Jekielek, S. (1998). Parental Conflict, Marital Disruption and Children's Emotional Well-Being. Social Forces, 76(3), 905-936. doi:10.2307/3005698
Children remaining in high conflict environments generally exhibit lower levels of well-being than children who have experienced high levels of parental conflict but whose parents divorce or separate. These results support the possibility that marital disruption, following high conflict, may actually improve the emotional well-being of children relative to a high conflict family status.

Sutherland A. In https://ifstudies.org/blog/how-parental-conflict-hurts-kids Accessed 10.5.20

Parents in high-conflict relationships tend to engage in more criticism, aggression, making threats, shouting, and hitting. High-conflict relationships can also produce lax and inconsistent parenting: parents who simply don't pay much attention to their children. In either case, children may fail to form a secure attachment to parents as a result.

Beyond this, in the struggle to understand their parents' conflict, children can come to blame themselves or find harmful ways of coping with the conflict. In addition, on top of their negative emotions, children experience physiological reactions related to stress that may harm their brain development.

Some variables shape the impact of parental conflict: the age, sex, and temperament of the child; the child's coping strategies; and the child's physiological reaction to stress. Also family characteristics matter, too: sibling relationships, attachment to parents, parents' mental health and substance use, and socioeconomic pressure.While socioeconomic pressure tends to worsen parents' mental health and increase parental conflict, the link between conflict and child outcomes remains significant when socioeconomic pressure is accounted for. But conflict is a predictor of outcomes beyond SES.

12. **Recognize the effect of temperament variations on the relationships among siblings.** Saudino, K. J., Wertz, A. E., Gagne, J. R., & Chawla, S. (2004). Night and day: are siblings as different in temperament as parents say they are?. Journal of personality and social psychology, 87(5), 698–706. https://doi.org/10.1037/0022-3514.87.5.698

Highly active children tend to have more conflicts with their siblings and they both give and receive more antagonistic behaviors within the sibling relationship. Also, siblings who differ in activity experience high levels of conflict and negativity and less warmth in the sibling relationship. Shyness :When older siblings are shy, sibling relationships are

less controlling and less competitive and the sibling who is shyer experiences less closeness and less caretaking within the sibling relationship.\

Brody, Stoneman, and Burke (1988) found that parents' perceptions of differences between siblings' activity levels were a more important predictor of adjustment than the absolute activity level of the child. These authors suggest that parents may label one child as maladjusted on the basis of perceptions of behavioral differences between siblings that are not objectively valid.

Brody, G. H., Stoneman, Z., & Burke, M. (1987). Child temperaments, maternal differential behavior, and sibling relationships. Developmental Psychology, 23(3), 354–362. https://doi.org/10.1037/0012-1649.23.3.354

High activity, high emotional intensity, and low persistence levels in both older and younger children were associated with increased agonism between sisters, whereas high activity and low persistence levels for younger brothers were associated with more agonistic behavior among brothers. An imbalance of maternal behavior that favored the younger child was generally associated with lower rates of verbalizations and prosocial and agonistic behavior directed by siblings to one another. The observations of the mother–sibling triadic and sibling dyadic interactions also revealed consistency in the within-family environments.

13. Identify predictors of successful adolescent functioning.
Gall M and Stixrud W. In https://www.nais.org/magazine/independent-school/summer-2008/the-4-s-s-of-adolescent-success/ Accessed 10/5/20.

a. Secure maternal attachment
b. Self-Regulation

Emotional intelligence means that you control your emotions instead of the other way around. Self-regulation, emotional awareness, empathy, and pro-social behavior are all hallmarks of emotional intelligence. Self-discipline enables a teen to postpone gratification, get up in the morning, honor commitments, tolerate frustration, and manage anger responsibly. It takes a lot of emotional maturity to control our impulses, which is why integrity and virtue pivot on self-restraint. The ability to pause and think before we act enables ethical choices. A lack of self-control puts us at the mercy of our urges, appetites, and unbridled passions. Irresponsible or insensitive acts committed in the heat of the moment can have long-lasting, irreversible consequences.

c. Self-Knowledge

It is important to have an understanding what one is good at and acting on that knowledge contributes immeasurably to job satisfaction.

d. Stress Management

Mercer, N., Farrington, D. P., Ttofi, M. M., Keijsers, L., Branje, S., & Meeus, W. (2016). Childhood Predictors and Adult Life Success of Adolescent Delinquency Abstainers. *Journal of abnormal child psychology*, *44*(3), 613–624. https://doi.org/10.1007/s10802-015-0061-4

Why aren't all adolescents delinquent? Overall, this is primarily predicted by individual characteristics of the child, whereas being convicted by age 18 is predicted by both individual and environmental risk factors. High honesty, high conformity, and high family income predicted abstention (from delinquency), low honesty, low conformity, and low family income predicted convicted delinquency.

14. Understand the effects on children of parental substance abuse and/or alcohol use disorders.

Studies show the elevated risk that children of substance abusing parents face in general for poorer academic functioning; emotional, behavioral, and social problems; and an earlier onset of substance use, faster acceleration in substance use patterns, and higher rates of alcohol and drug use disorders. There is also contextual risk factors for children of substance abusing parents, including parenting deficits (less warmth, responsiveness, and physical and verbal engagement as well as harsher and more over-involved interaction styles), greater risk for child maltreatment, and less secure attachment patterns. (from: Solis, J. M., Shadur, J. M., Burns, A. R., & Hussong, A. M. (2012). Understanding the diverse needs of children whose parents abuse substances. Current drug abuse reviews, 5(2), 135–147. https://doi.org/10.2174/1874473711205020135

In addition, there could have been the teratogenic effect of exposure to alcohol such as Fetal Alcohol Spectrum Disorders that could affect developmental and behavioral functioning apart from the physical effects. There can be poor self regulation, neurocognition and adaptive skills in those exposed to alcohol in utero.

C. Socioeconomic Diversity

1**. Recognize the influences of poverty on the epidemiology of developmental and behavioral disorders**

- Poverty is associated with regular experiences of material hardship, decreased parental capacity, and therefore decreased developmental inputs that promote positive developmental outcomes.
- Developmental outcomes are worse for children who experience poverty in early childhood, persistently throughout the course of childhood or those children living is very disadvantaged communities.
- Children who experience at least 1 year of poverty have worse outcomes than those who never experience poverty.
- Children who grow up in poverty as infants and toddlers are 30% less likely to complete high school.

- Children who experience persistent poverty are more likely to not graduate from high school, have lower earnings as adults, be poor as adults, and have poorer health as adults.
- Children living in poverty are 50% more likely to require special education or early intervention services compared children who do not.
- Prevalence of learning disabilities is doubled in children growing up in poverty compared to those who do not.
- Prevalence of ADHD is 40% higher in children growing up in poverty compared to those who do not.
- There is also a higher risk of a variety of other conditions in children who grow up in poverty including lead poisoning, lower birth weight, parental mental health challenges, child abuse, depression, and substance abuse.

2. **Know the range of recommendations that might be made to help families with limited resources facilitate their child's development**
    - The most effective way to promote child development is at the structural level, by ensuring economic stability for the family in the form of higher wages.
    - Other structural changes known to promote child development are access to high quality child care and education, access to quality health care, and parent mental health support.
    - At an individual/family level, the following interventions can help facilitate development:
        i. Connecting families with food pantries or other community food programs.
        ii. Connecting families with free tax preparation programs, which can help families access tax credits they are otherwise unaware of. The Earned Income Tax Credit helps a significant number of children and families live out of poverty.
        iii. Implementing Reach and Read programs and counseling families about the benefits of reading.
        iv. Connecting families with parenting programs such as Healthy Steps for Young Children, Incredible Years, and Triple P.
        v. Implementing programs like the Video Interaction Project (VIP) helps promote cognitive and social development.
        vi. Connecting families with a social worker (if available) to assist with finding stable food sources, housing, employment, parenting, and mental health resources in the community.
3. **Know the additive disadvantage for children with disability, minority group membership, poverty, and social isolation**
    - For many children living in poverty, there is often the cumulative disadvantage of multigenerational legacies of racism, ableism, segregation, and poor access to health care contributing to further downward socioeconomic mobility.
    - Black children living in poverty experience more downward economic mobility from one generation to the next compared to their white peers.

- Our school systems remains highly segregated with students of color, particularly Black and Latinx children attending majority student of color schools, which tend to be under-resourced compared to schools that are majority white students. These students are multiply disadvantaged due to systemic discrimination and a lower quality educational environment which stifles upward socioeconomic mobility.
- Children with disabilities from rural settings also tend to face additional disadvantages of under resourced school systems, decreased access to highly qualified educators, and higher rates of poverty affecting their communities.

D. Societal Factors

2. Education

a. Understand the significance of school-community relationships in the educational success of children

- School-community partnerships consists of collaboration between school and community (public or private) organizations working with a shared sense of responsibility and accountability, aligning resources towards a goal of educational success for every child.
- Different models for school-community partnerships exists across the country, based on the need of a particular community. One of the basic tenets is to promote educational enrichment by reducing the burden on schools to overcome socioeconomic barriers to learning for children and families.
- School-community partnerships have proven to be effective in:
    - Increasing students' performance on state math and reading tests and,
    - Decreasing school dropout rates compared to non-partnered school-community relationships.

b. Understand how community values influence educational processes in schools

- Informing educational processes based on community values can help validate diverse communities' cultures, build an environment of mutual respect and responsiveness between schools and families, and provide a more holistic education.
- A famous example of community values influencing educational processes is Hull House, an early 20th century community hub for European immigrants in Illinois. In addition to providing classes, Hull House served as a connector to community organizations like health centers, colleges, shelters, gymnasiums, etc. Their aim in providing education was to begin

with children's prior life experiences in informing schooling and community resources to build an educational process for social improvement but also for civic engagement.

c. Understand the impact of parental involvement in schools for educational success of children

- Parental involvement is defined as "parental participation in the educational processes and experiences of their children" (Jeynes, 2005). Given this definition, parent involvement includes home-based parental involvement (listening to children read, supervise homework, etc) as well as school-based parental involvement (attending parent-teacher meetings, education workshops, etc).
- Several systematic reviews and meta-analysis of the education literature has demonstrated that parental involvement (effect size ranging from 0.51-0.74) has a greater effect on children's academic achievement than direct educational interventions (effect size 0.4).
- Parental involvement is mutually beneficial for teachers, parents, and children. Effective parental involvement is linked with improved parent-teacher relationships, increased teacher morale, increased parental confidence and satisfaction with parenting.
- It should be noted that effective parental involvement refers to activities that foster greater partnerships between parents and teachers not simply activities carried out by parents meant to primarily benefit the school (fundraising, etc). It is also important to recognize that a majority of the rhetoric surrounding increasing parent involvement accounts for typical disadvantages experienced by white middle-class families, and ignores the barriers faced by families from diverse backgrounds.
- Family circumstances (single parent, large families), psychological resources (physical or mental health conditions, lack of supportive social network), and inflexible jobs are a few barriers to parent involvement in schools.
- Within preschool programs like HeadStart that promote parent involvement in schools, increased parental involvement is correlated with decreased parent controlling behavior, decreased spanking, and increased cognitive stimulation directed at the child.
- Effective parental involvement moderates some of the effects of socioeconomic stressors on cognitive and socio-emotional skills necessary for educational success. The positive correlation between parent involvement in schools and educational success is stronger in more affluent neighborhoods, suggesting that neighborhood enrichment also play a role in educational success.

- There is also a growing body of literature showing that in addition to parental involvement, racial/ethnic/cultural socialization among children of color is also related to cognitive and academic outcomes.

d. Understand issues associated with each of the various school arrangements that are available to families (eg, home schooling, charter schools, private and parochial schools)

- Charter schools are independent public schools, funded by the public and private sectors. There is no tuition responsibility for the family. Charter schools are required to follow some federal and state education laws but have some leeway on federal and state mandates due to their private funding. Quality and mission of charter schools can be highly variable.
- Charter schools are required to evaluate for and provide special education services just like public schools however, charter schools are generally under resourced in terms of special education services (specialists, therapists, ability to make accommodations).
- Private schools are run by private organizations and because they receive no government funding, they are not regulated by state or federal educational standards. Parochial schools are private schools supported by a religious organization (ie, catholic schools). In general, private schools tend to be significantly less socioeconomically and racially diverse compared to other school arrangements. There is usually a significant tuition responsibility for the family.
- Private schools are not required to evaluate for or provide special education services as outlined in the IDEA (Individuals with Disabilities Act). There are some private schools that specialize in serving kids with specific disabilities (ie, ABA-based schools for children with autism).
- Public school districts are required by law to evaluate and provide services for children in private schools. It is the family's decision whether they want to pursue these services.
- Families may elect homeschooling for a variety of reasons including academic, religious, or safety concerns (ie, bullying or racism).
- The biggest drawback for homeschooling is the lack of social interaction with other children and adults. However, many families engage in other activities with other children such as field trips, church groups, community events, sports.
- There is limited reliable data on academic and socioemotional skills of homeschooled children because much of the data is collected by organizations that lobby for homeschooling and there is a strong selection bias (study population tends to be mostly White, mostly Christian, high-income households).
- Some states allow homeschooled children access to public school classes and activities. Homeschooled children can also receive an evaluation and

services for special education through the public school but will likely need to be enrolled part-time in the public school.

e. Understand how the structure and administrative characteristics of schools can be associated with school behavior and learning problems

- A systematic approach (not just classroom-based) to reducing school behavior and learning problems is essential. Evidence has shown that the best way to have a structured approach to reducing school behavior problems is through school improvement teams.
- School improvement teams should consist of administrators, teachers from each grade level, a representative from the school support staff, a behavioral expert (such as a school psychologist or counselor), and students (especially in high school).
- The school improvement team's responsibilities include ongoing needs assessments to address specific behavioral problems, implementation of school wide discipline programs that are positively-based with high behavioral expectations, and ongoing monitoring of the approach.
- School improvement teams should collect information on hot spots of behavioral problems at school- such as when (before or after school, recess, lunch) and where (hallways, bathrooms, playgrounds, cafeterias) via input from teacher surveys, staff meetings, students and school personal in charge of common areas.
- Schools may consider adopting packaged intervention plans such as *Second Step* and *Promoting Alternative Thinking Strategies* to address behavioral issues, but these should be used keeping the school's capacity and specific objectives in mind.
- A school wide targeted approach to mainstreaming students with learning disabilities has the potential to increase student social relationships.

f. Know the effects of full-day kindergarten for children's development

- Full-day kindergarten has been shown to have a positive effect on:
  - Reading and math achievement test scores at the beginning of first grade
  - Socio-emotional development, particularly in the skills of working and playing with others
- Full-day kindergarten also allows parents more time for work, increasing the family income. Higher family income is associated with more positive child development outcomes.
- Full-day kindergarten *may* allow for easier and earlier identification of developmental problems, and improved nutrition.
- There is concern that full-day kindergarten may put undue pressure on children before they are developmentally ready (ie, do not have the emotional regulation skills or social maturity for 6+ hours of schooling).

- Full-day kindergarten may increase some childrens' irritability and increase their risk of behavior problems.

g. Know the benefits of early childhood education programs (eg, Head Start) on children's development

- Early childhood education programs (both formal and informal preschool programs) have been associated with better cognitive and academic outcomes, particularly in children from socioeconomically disadvantaged backgrounds.
- There is some evidence that early childhood education programs such as Head Start improve childrens' ability to sustain attention, enhance receptive language skills, and decrease behavior problems.
- As for long-term outcomes, at age 15 years, quality early childhood education programs are associated with slightly
    - Better cognitive and academic outcomes for children from diverse socioeconomic backgrounds.
    - Increased risk taking and impulsivity, particularly for children who spent longer hours in more center-type care.

h. Know the benefits of Early Intervention Programs on children's development

- Early intervention programs have been shown to decrease the gap in scores of cognitive development measures between children from low-resource families and high-resource families.
- Early intervention programs that include parent psychosocial support and parent education have been associated with decreased maternal anxiety, depressive symptoms, and self-efficacy.
- Of note, there are more studies showing no effect on developmental outcomes from early intervention programs than those showing effect. It is hypothesized that this maybe due to the heterogeneity of early intervention programs and the populations served. Early intervention programs have shown positive effects particularly when they begin in infancy, are delivered more regularly by well-trained professionals, and have specific outcome goals.

i. Understand the impact of different types of childcare on child development

- In general, parental childcare in the early stages of development has a more positive impact on developmental outcomes than nonparental childcare. This is the reason why policies like extended paid parental leave are essential.
- In terms of non-parental childcare, high-quality formal childcare is positively associated with developmental outcomes. High quality formal childcare generally refers to center-based childcare with low child to adult

ratios, sensitive and responsive caregivers, high levels of language stimulation, a positive emotional climate, and opportunities for children to explore their environment.

- Children who attend center-based preschool programs score higher on language and math skills at 5 years of age compared to children who were in parent care or in informal home settings.
- Academic/cognitive benefits from center-based child care are seen more from participation during the preschool years (as opposed to the infancy/toddler years), and are more pronounced for children from socioeconomically disadvantaged families. It is theorized that for these children, a high-quality child care center may be a source of material (learning materials) and psychosocial (stimulation, responsive caregivers) investments. To this point, it must be noted that parents' ability to provide materially and psychosocially for their children is by and large a consequence of societal policies on how parents are supported in caregiving for children rather than an inherent individual inability to do so.
- Overall, there are mixed findings about the impact of center-based childcare on later behavioral development. There is some persuasive evidence that children who spend many hours in nonparental care (particularly center-based care) in early childhood are perceived as having more externalizing behavior problems (aggression, lack of self-control, teacher-child conflict) by their teachers in preschool and early school years. These findings are stronger in White, non-Hispanic children from higher income households.
- With regards to social development, again the studies are mixed but there is some persuasive evidence that high-quality nonparental care are correlated with better social skills later in childhood.
- There are no consistent gender differences in the relationship of maternal employment or child care hours to problem behavior.

j. Know how to advise a school about developing programs to minimize bullying and to handle any incident of bullying for both the victim and the bully

- The best way to minimize bullying is through prevention. Bullying is not a conflict, it is a form of victimization that involves a power differential.
- Bullying prevention efforts can be sustained over time by training school staff and students on how to prevent and address bullying. This training should specify what bullying is, what the school's policies are related to bullying, and how to enforce these policies.
- There are packaged evidence-based anti-bullying curricula/programs designed for schools, but schools should select a program based on its demographics, capacity, and resources.
- The following strategies should be avoided because they are ineffective or reinforce bullying behavior:

        - Zero-tolerance policies: These policies suspend or expel children who bully. However, the threat of suspension/expulsion may discourage adults and children from reporting bullying.
        - Conflict resolution and peer mediation: These strategies are used for conflict management however, bullying is a form or victimization **not** conflict. Using conflict mediation sends the wrong message to the victim that "you are partly wrong and right" instead of "bullying is inappropriate and no one deserves to be bullied."
        - Group therapy for children who bully: These groups may reinforce anti-social or bullying behavior among children who bully.
    - When responding to an incident of bullying, adults must
        - Intervene immediately and not ignore the incident.
        - Separate the youth involved, make sure everyone is safe, and not try to sort out the facts immediately.
        - Not question or talk to youth about the incident in front of other youth.
        - Not make youth apologize or try to mediate on the spot.
        - Model respectful behavior when intervening.
        - Seek medical attention immediately if there is serious physical injury, bodily harm, or sexual abuse.

k. Understand the child, school, and family factors which contribute to school refusal

- School refusal is a behavior, not a diagnosable condition. It is often associated with an underlying anxiety disorder such as separation anxiety, social anxiety, or panic disorder.
- When not associated with an underlying anxiety disorder, a precipitating incident such as being scolded at school, peer conflict, bullying, discrimination, family conflict, or academic difficulties maybe the cause.
- Once school refusal behavior begins, it is self-perpetuating since the more time a child spends away from school, the more anxious they may become about attending school.
- There are 3 peak stages in development when school refusal behavior is more likely to occur:
    - At school entry (age 5 or 6)- when it is often related to separation anxiety.
    - At entry into middle school (ages 11-13)- when it often involves underlying anxiety or affect dysregulation.
    - During high school (ages 14 and older)- during these ages, this behavior is associated with truancy (which is a more volitional behavior). Whereas both truancy and school refusal are types of school avoidance, youth with school refusal often have severe emotional

distress with having to go to school and their parents' have full knowledge that they have remained at home. During high school, school refusal behavior can be associated with serious psychiatric illness.

l. Know how to work collaboratively with schools and families to design an intervention plan for a youngster with school refusal

- The intervention plan should be tailored to the needs of each individual child and family. A comprehensive intervention plan can include environmental modifications, psychotherapy, psychoeducation and potentially pharmacotherapy. Prior to school reentry, a plan must be developed with the parents and youth that includes a hierarchy of steps towards full-day entry.
- In most cases with mild or acute school refusal behavior, an immediate return to school should be emphasized to reduce the reinforcement of being at home. If the child needs to remain at home due to a physical illness, the home environment should be as unentertaining as possible and the child should not be allowed to watch television, play video games, read books or interact with family members.
- For youth with chronic school refusal, return to school should be phased and active involvement of parents and school personnel is crucial.
- Parent should avoid reinforcing school refusal behaviors such as escorting child to school and limiting access to pleasurable activities at home.
- Anxiety management training should be initiated prior to school reentry. These include relaxation training, mindfulness, cognitive behavioral therapy and social skills training. CBT has shown to be an efficacious treatment for children with school refusal behavior.
- School-based interventions may include a 504 modification plan and support groups (for bullying, safety, absenteeism, or teen pregnancy) for youth.
- School-based accommodation strategies may include:
    - Initially allowing youth to spend the whole school day in one part of the school (such as the lobby) rather than return home.
    - Initially allowing for a shortened school day with goal of increasing time to the full school day.
    - Allowing the anxiety-management breaks incorporated into a 504 plan.
    - Rewarding the youth with a phone call to parents later in the day for engaging in school-based activities.
    - If test/performance anxiety in involved, youth may be allowed to take tests in a quiet, private environment with increased time.

- When psychotherapy and/or home and school based accommodations do not result in adequate response, pharmacological treatment with SSRIs can be considered.

4. Discrimination

a. Recognize the effects of stigmatization and discrimination on children and families who are in some way different from their community

- Stigmatization and discrimination for marginalized children and families exists on a structural level and impacts families through laws, policies, regulations, and practices that lead to disproportionate access to resources, services, and opportunities in the health, education and community settings.
- Children and families can be marginalized on the basis of race, gender, sexuality, disability, language, religion, socioeconomic status, nationality, immigrant status, age or the intersectionality of these identities.
- Up until now, most of the pediatric research has focused on disparate outcomes for marginalized populations. Recently, there has been a more specific focus on the negative impact of structural racism on child and adolescent health and development
- There is a strong relationship between self-reported experiences of racial discrimination with poor emotional outcomes, stress, anxiety, and feelings of powerlessness.
- Experiences of racism are also strong predictors of delinquent behaviors, including violent and aggressive behaviors.
- Racism and discrimination are toxic stressors, and negatively impact health outcomes for marginalized individuals over their lifetime from birth outcomes to mental health.
- There is some evidence that higher implicit biases in medical providers negatively affects how they communicate with children and families of color.
- Children and families of color experience significant discrimination and stigmatization in the educational and justice systems.
- Children of color particularly Black, Latinx, and Indigenous children, are at disproportionately identified, placed and disciplined in special education.
- Children of color with disabilities are at much higher risk of being placed in more restrictive educational settings and being suspended and expelled from school.
- In the criminal justice system, children of color with or without disabilities are more likely to be arrested and receive longer sentences.

b. **Know the range of recommendations for parents to help children to recognize, confront, and protect themselves from the damaging effects of social discrimination**
    - Positive racial identity mediates the negative effects of racism and can lead to better developmental outcomes for all children. Having a positive racial identity means having positive attitudes and beliefs about one's racial group.
    - Parents can help children develop positive racial identity by talking with children about race, racism, and other forms of discrimination. Avoiding discussions about racism and other forms of discrimination does not prevent children from developing implicit and explicit biases.
    - Increasing awareness about discrimination improves children's self-esteem and is associated with lower levels of bias in young children.
    - Although research on the effects of social discrimination other than racial discrimination is sparse, similar recommendations about fostering positive relationships with one's in-group and having open, developmentally-appropriate conversations with children about discrimination apply.

c. **Know the range of recommendations for schools to minimize isolation and stigmatization of children with individual or family differences**
    - Schools can minimize isolation and discrimination by eliminating curricula, books, instructional materials, and activities that promote prejudiced and discriminatory attitudes. Schools can instead use nondiscriminatory and nonstereotypical language, resources, practices, and resources.
    - Schools should incorporate roles and contributions of all groups throughout history in their curriculum, particularly groups that have been historically underrepresented.
    - Schools can eliminate explicit or implicit practices that favor the education of some students over others on the basis of race, gender, sexual orientation, disability, immigration status, religion, etc.
    - Schools should establish policies that prevent and prohibit bullying and harassment of all children.
    - All teachers and staff must be trained on working with children from different backgrounds, and recognizing discriminatory practices and policies.

6. Housing

a. Recognize the influences of homelessness on child development and behavior
    - Family homelessness rates have been rising since the 1980s, and positively correlate with income inequality since that decade.
    - Some precursors of homelessness include unstable housing, moving in with family or friends, and domestic violence.

- There is a higher rate of underimmunization, lead exposure, and nutritional deficiencies (including iron deficiency anemia) in homeless children.
- Homeless children are less likely receive preventive care services like immunizations and routine screenings.
- Homeless children and adolescents have significantly higher rates of developmental, behavioral, and psychiatric disorders.
- 19% of homeless infants or toddlers < 36 months have a developmental disorder versus 9% of non-homeless infants and toddlers.
- Child development and behavior in homeless children is likely the cumulative effect of multiple toxic stressors such as poverty, parent mental health disorders, domestic violence, etc.

8. Access to health care

a. Understand the risks to child development of inadequate access to health care

- Since the passage of the Affordable Care Act, about 4.5% of children in the United States are uninsured, decreased from ~10% prior to the passage of the ACA.
- More than lack of insurance coverage, most children and families have inadequate access to healthcare due to the following barriers:
    - Financial (high costs of copays, costs of deductibles, prescription drug coverage),
    - Geographic (transportation, living in region with short supply of health professionals), and
    - Information (health literacy, English proficiency).
- Evidence shows that insurance coverage (public or private) results in improved prenatal care (reduced infant mortality and low birth weight), decreased hospitalizations, and increased immunization rates. Some studies have even shown improved children's performance on standardized reading tests.

b. Understand the potential contribution of the "medical home" concept to promoting optimal child development and behavior

- Children with and without disabilities who have a medical home have access to a usual source of care that is trusted and provides coordinated care.
- Evidence thus far shows that children with a medical home have:
    - Lower rate of emergency department visits,
    - Fewer hospitalizations,
    - Higher rates of immunizations,
    - Increased dental service use,
    - Fewer missed school days,
    - Decreased parental stress,

    - Decreased financial costs to the family.
  - For children with special health care needs, having a medical home is associated with decreased time burden on the family coordinating care and decreased likelihood of having unmet specialty care needs.

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Content Category 4- Elements of Assessment and Management (except 4C)
ABP Content Specs for Developmental-Behavioral Pediatrics Fellow Collaboration Project

Section A and Section B 1&2 - Prepared by Carrie Cuffman, UH Rainbow Babies and Children's-Cleveland DBP Fellow

Reviewed by Shanna Kralovic, MD, UH Rainbow Babies and Children's-Cleveland - DBP Fellowship Director
Section B 3-6 and Section C- Prepared by Rebecca Christi, Kira Belzer, and Joshua Strait- DBP Fellows- Madigan Army Medical Center

Reviewed by Eric Flake, MD, Staff Developmental Behavioral Pediatrics & Bonnie Jordan, Program Director- Developmental Behavioral Pediatrics Fellowship, Madigan Army Medical Center

4. Elements of Assessment and Management

A. Assessment

1. Know the principles of developmental surveillance and screening
2. Understand the characteristics of a good screening test
3. Distinguish between developmental surveillance, developmental screening, and developmental assessment
4. Know the psychometric properties and proper use of common parent-completed questionnaires available for developmental surveillance and screening
5. Know the psychometric properties and proper use of common directly administered developmental screening tools
6. Know the psychometric properties and proper use of common parent-completed behavioral and emotional screening tools
7. Know the standardized measures for assessing the home environment
8. Differentiate global from domain-specific developmental and behavioral screening instruments
9. Understand the issues important to hearing screening for infants in the newborn period and beyond and describe strategies to reduce loss to follow-up
10. Know the methods for assessing hearing at different developmental stages including physiologic measures, behavioral audiometry, conditioned response audiology, play audiometry, and soundfield audiometry
11. Know the methods for assessing vision at different developmental stages
12. Be able to identify common dysmorphic features
13. Know appropriate assessments to evaluate a child's motor coordination
14. Know the procedures to evaluate a child's mental status
15. Know interview and screening methods to assess a child's or adolescent's emotions and moods
16. Understand the important historical information required to assess the possible etiologies for developmental delay/disabilities
17. Know the appropriate laboratory evaluation for developmental delay
18. Know the indications for different types of molecular genetic tests
19. Know the indications for cytogenetic testing
20. Know the indications for biochemical/metabolic testing
21. Know the indications for neuroimaging
22. Understand the importance of identifying child and family strengths in the process of planning intervention
23. Know how to interpret results of infant cognitive assessments
24. Know how to interpret results of verbal and non-verbal cognitive assessments in children
25. Know how to interpret results of academic achievement testing
26. Know how to interpret results of motor assessments

27. Understand the implications of neuromaturational delays for a child with school problems
28. Recognize the impact of cultural differences on developmental testing performance
29. Know how to obtain and interpret a genogram for the understanding of family function
30. Know the principles of behavioral screening
31. Know the psychometric properties and proper use of common child or adolescent self- report behavioral and emotional screening measures
32. Know methods to assess a child's or adolescent's attention span, impulsivity, and hyperactivity
33. Know methods to assess the significance of a child's or adolescent's defiant or aggressive behaviors
34. Know methods to assess an adolescent's substance use
35. Know methods to assess an adolescent's sexual behaviors
36. Know methods to assess family functioning
37. Know methods to assess a child's or adolescent's peer relationships
38. Know methods to assess for social stressors
39. Know interviewing techniques appropriate for children of various ages and developmental levels
40. Know how to interpret results of adaptive skill assessments
41. Know how to interpret results of speech and language assessments
42. Understand current classifications of behavioral health disorders in infants and toddlers
43. Know the indications for fluorescence in situ hybridization (FISH) testing
44. Know the indications for neuroelectrophysiological studies (e.g., electroencephalography)
45. Know methods to assess for autism spectrum disorders
46. Know methods to assess executive functions

B. Management
   1. Communication skills
      a. Understand how to inform parents of a diagnosis of a developmental disability, behavioral/mental health disorder, or life-threatening condition in their child
      b. Understand how to effectively elicit parents' opinions and concerns for their child's development
   2. Anticipatory guidance/health promotion
      a. Know how to counsel families on the potential negative effects of physical punishment
      b. Know specific recommendations for parents to limit the influence of the media on their child's development and behavior
      c. Know how to advise a family of a toddler on knowing when the child is ready to initiate toilet training

d. Know how to advise parents to address resistance to toilet training
e. Know how to advise parents on sleep hygiene to avoid problematic sleep associations and bedtime resistance
f. Know how to advise parents to approach discussions with their child regarding high-risk behaviors at different ages and developmental levels
g. Know how to advise parents concerned about their child's sexual behaviors at different ages and developmental levels and in different settings
h. Know recommendations that could be given to parents to help them facilitate their child's adjustment to a new sibling at different ages and development levels
i. Know how to advise parents on key features of quality child care programs
j. Understand the importance of sexuality education for teenagers with developmental disabilities and describe key features of effective education
k. Know the developmental and behavioral advantages of breast-feeding
l. Know how to advise parents on disciplinary strategies appropriate for children of different ages and developmental levels
m. Know how to advise parents on habits and repetitive behaviors at different ages and developmental levels
n. Know how to advise parents whose child is having temper tantrums or breath holding spells
o. Know how to advise parents of children engaging in aggressive behavior at different ages and developmental levels
p. Know how to advise parents on strategies they can use to promote the development and academic success of their children
q. Know how to advise parents on strategies to promote their child's self-esteem at different ages and developmental levels
r. Know how to advise parents who are concerned about their infant's crying
s. Know how to advise parents concerned about sibling rivalry at different ages and developmental levels
t. Know how to advise parents about the use of transitional objects at different ages and developmental levels
u. Know how to advise parents about fears or anxieties in their child at different ages and developmental levels
v. Know how to advise parents concerned about their child's feeding behaviors at different ages and developmental levels
w. Know how to advise parents on promoting toilet training for a preschool-age child
x. Know how to advise parents on promoting toilet training for a developmentally delayed school-age child

y. Identify strategies to guide families in building coping and resilience and avoiding overprotection in a child with a developmental disability

3. Counseling
   a. Understand how reframing can be used within counseling as a therapeutic maneuver
   b. Understand the important role providing information about a condition can play in therapy
   c. Understand how strategic family therapy can be used to challenge the family with particular tasks to improve existing maladaptive patterns of problem solving or communication
   d. Understand the key constructs of family systems theory
   e. Understand how cognitive restructuring and attribution retraining can be employed in cognitive-behavioral counseling to address disorders of mood or emotion
   f. Know the benefits of group therapy for enhancing social skills and behaviors
   g. Appreciate conditions for which group therapy is most appropriate
   h. Understand the components of and indications for cognitive behavior therapy
4. Behavioral interventions
   a. Understand conditions in which classical conditioning is utilized for treatment
   b. Differentiate among schedules of reinforcement most appropriate for a behavior management plan (eg, fixed and variable; ratio and interval)
   c. Understand that reinforcing incompatible behavior is an alternative to planned ignoring of the target behavior
   d. Know the various forms of punishment, including natural consequences, logical consequences, behavioral penalties, physical punishment, and time out
   e. Understand the importance of identifying the function of a behavior in selecting a behavior management technique
   f. Know the important components of a successful time-out procedure
   g. Know how to initiate a token economy within a home or school
   h. Identify situations in which planned ignoring or time-out would not be an appropriate recommendation to decrease a problem behavior
   i. Know how to initiate a school-home note as a behavioral intervention
   j. Know the important components of a successful extinction procedure
5. Basic pharmacotherapy
   a. Understand the physiologic activity of various neurotransmitters (eg, dopamine, norepinephrine, serotonin, gamma-aminobutyric acid, acetylcholine)

b. Know how to apply the principles of pharmacodynamics
c. Know how to apply the principles of pharmacokinetics
d. Know how common psychotherapeutic agents are metabolized and excreted
e. Understand the issues related to the combined use of psychopharmacologic agents
f. Understand the pharmacodynamic and pharmacokinetic properties of stimulant medications
g. Know indications for the use of stimulant medications
h. Understand the clinical use of stimulant medications
i. Know the side effects and appropriate monitoring of stimulant medications
j. Know indications for the use of selective serotonin reuptake inhibitors
k. Understand the clinical use of selective serotonin reuptake inhibitors
l. Know the side effects and appropriate monitoring of selective serotonin reuptake inhibitors
m. Know indications for the use of mood stabilizers
n. Understand the clinical use of common mood stabilizing medications
o. Know the side effects of common mood stabilizing medications
p. Know indications for the use of tricyclic antidepressants
q. Understand the clinical use of tricyclic antidepressants
r. Know the side effects and appropriate monitoring of tricyclic antidepressants
s. Know indications for alpha-adrenergic agonists
t. Understand the clinical use of alpha-adrenergic agonists
u. Understand the side effects and appropriate monitoring of alpha-adrenergic agonists
v. Know indications for the use of anti-anxiety medications
w. Understand the clinical use of common anti-anxiety medications
x. Know the side effects and appropriate monitoring of common anti-anxiety medications
y. Know indications for the use of antipsychotic medications for problems seen in developmental and behavioral pediatrics
z. Understand the clinical use of antipsychotic medications for problems seen in developmental and behavioral pediatrics
aa. Know the side effects and appropriate monitoring of antipsychotic medications bb. Understand the clinical use of norepinephrine re-uptake inhibitors
cc. Know the side effects and appropriate monitoring of norepinephrine re-uptake inhibitors
dd. Know the clinical use of atypical antidepressants (eg, bupropion, clomipramine, etc)
ee. Know the side effects and appropriate monitoring of atypical antidepressants (eg, bupropion, clomipramine, etc)

ff. Know how to monitor for side effects of antipsychotic medications
gg. Understand the basic principals of pharmacogenomics

6. Other strategies
   a. Understand the importance of adequate basal treatment of pain
   b. Understand how to formulate a pain management plan
   c. Understand the principles and techniques of hypnosis
   d. Understand the principles and techniques of biofeedback
   e. Know appropriate advice for a school on the appropriate timing and nature of memorialization activities after the death of a student
   f. Understand principles in providing post-suicide intervention services in a high school
   g. Know how to evaluate the utility of non-standard therapies for developmental and behavioral disorders
   h. Know the risks of special diets, supplements, and other common alternative treatments that are often recommended for children with developmental disabilities
   i. Know how to counsel families who are utilizing non-standard (alternative) therapies

C. Team processes
   1. Principles of collaboration
      a. Know the importance and process of identifying the underlying agenda for consultation requests from community sites
      b. Know the steps in establishing a collaborative relationship with community organizations and agencies
      c. Know how developmental-behavioral pediatricians can serve as consultants to schools
      d. Know the steps in conducting an evaluation of a child as a school consultant
      e. Know how to conduct an evaluation of a child as a consultant to a child care center
      f. Understand the benefits and challenges of multidisciplinary evaluations within a collaborative team model
      g. Understand how the role of consultant to a school system differs in quality and scope to the role of the physician seeing a patient in the office
      h. Understand issues of confidentiality as they relate to the role of consultant to a school system
   2. Knowledge of other health professionals' roles and methods
      a. Be familiar with the elements of a psychological and educational

evaluation conducted as part of a multidisciplinary team evaluation for a child with learning and/or behavior problems

b. Understand common behavioral assessment techniques
c. Understand the elements of a neuropsychological evaluation of a child
d. Recognize the indications for neurological consultation in a child with a developmental or behavioral problem
e. Know the role of genetic consultation and counseling in a child with developmental disabilities
f. Know the indications for speech and language evaluation and treatment in a child with developmental or behavioral disorders
g. Know how speech and language testing evaluates pragmatics, semantics, and syntax
h. Differentiate between the evaluations and services provided by physical therapy and occupational therapy
i. Recognize the role of occupational therapy in evaluation and treatment of children with developmental disorders
j. Know controversial therapies (eg, sensory integration, optometric training, facilitated communication, etc) and their current status (eg, unproven, questionable, unsafe, proven ineffective, etc)
k. Recognize indications for psychiatric consultation in a child with a developmental or behavioral problem
l. Recognize the role of physical therapy in evaluation and treatment of children with developmental disorders
m. Differentiate among the evaluations and services provided by a play therapist, behavioral counselor, behavior analyst, psychodynamic therapist, and family therapist
n. Know the role of a vision specialist and orientation and mobility specialist in the treatment of a child with visual impairment
o. Know the role of a teacher of the deaf in the treatment of a child with permanent hearing loss

4. **Elements of Assessment and Management**

A. Assessment

1. Know the principles of developmental surveillance and screening

Developmental surveillance is a "flexible, longitudinal, continuous and cumulative process in which knowledgeable health care professionals identify children who may have developmental problems" (Lipkin 2020). It is also useful for allowing for provision of appropriate referrals, providing education to support healthy development and monitoring effects of early intervention/therapy on development (Lipkin 2020). Developmental surveillance should occur at all well child visits from birth through age 5, with particular attention to school readiness 4-5 year old visit to identify issues prior to kindergarten entry (Lipkin 2020).

6 components of developmental surveillance: (Lipkin 2020)

1) Eliciting and attending to parents' concerns about development
2) Obtain, documenting, and maintaining a developmental history
3) Making accurate and informed observations of the child
4) Identifying risks, strengths, and protective factors
5) Maintaining an accurate record of the process and findings
6) Sharing and obtaining opinions and findings with other professionals (i.e. child care providers, home visitors, preschool teachers, developmental therapists)

Developmental screening: use of a validated screening tool to further identify developmental concerns. Developmental screening is meant to identify children whose development differs from same-aged norms and does not result in a diagnosis (Lipkin 2020). Per the AAP recommendations, a validated developmental screening test (global screener) should be administered at the 9-, 18- and 30-month visits, with ASD-specific screening performed at the 18- and 24-month visits.

Lipkin PH, Macias MM, and Council on Children with Disabilities, Section on Developmental and Behavioral Pediatrics. Promoting optimal development: identifying infants and young children with developmental disorders through developmental surveillance and screening. *Pediatrics.* 2020; 145(1): e20193449.

2. Understand the characteristics of a good screening test

- Reliable (able to produce consistent results)
- Valid (able to discriminate between a child at a determined level of risk for delay from the rest of the population)
- Good sensitivity (accuracy in identifying delayed development) to minimize under-referrals (>70%)
- Good specificity (accuracy in identifying children who are not delayed) to minimize over-referrals (>70%)
- Standardized based on a large, representative national sample

- Current (should restandarize norms every 10 years)
- Items with good content and clarity
- PPV range of 30%-50% typical
- Feasible: low cost, reasonable time burden for parent and clinical staff, readability $\leq 5^{th}$ grade level, directions easy to understand
- Culturally and linguistically appropriate

Lipkin PH, Macias MM, and Council on Children with Disabilities, Section on Developmental and Behavioral Pediatrics. Promoting optimal development: identifying infants and young children with developmental disorders through developmental surveillance and screening. *Pediatrics.* 2020; 145(1): e20193449.

Marks KP, LaRosa AC. Understanding developmental-behavioral screening measures. *Pediatrics in Review.* 2012; 33(10):448-456.

3. Distinguish between developmental surveillance, developmental screening, and developmental assessment

Developmental surveillance is the informal and flexible process in which a knowledgeable individual gathers information about the development of the child. Developmental screening uses a standardized and norm-based tool to compare the child's development to other children's development at the same age, but does not result in a diagnosis. Developmental assessment is the means by which a person skilled in evaluating development makes a diagnosis of developmental disorder/ delay. (Lipkin 2020)

Lipkin PH, Macias MM, and Council on Children with Disabilities, Section on Developmental and Behavioral Pediatrics. Promoting optimal development: identifying infants and young children with developmental disorders through developmental surveillance and screening. *Pediatrics.* 2020; 145(1): e20193449.

4. Know the psychometric properties and proper use of common parent-completed questionnaires available for developmental surveillance and screening [Not an exhaustive list]

Parents Evaluation of Developmental Status (PEDS)

- Parent interview form.
    - Designed to screen for developmental and behavioral problems needing further evaluation.
    - Single response form used for all ages. May be useful as a surveillance tool
- Ages 0-8y
- Administration time 2-5 min
- Sensitivity 96%

- Specificity 83%
- Available in multiple languages

Ages and Stages Questionnaire-3 (ASQ-3)

- Parent-completed questionnaire
    - Series of 21 questions screening communication, gross motor, fine motor, problem solving, and personal adaptive skills. Results in pass, monitor, or fail score for domains.
- Ages 2-60 months
- Administration time 10-15 minutes
- Sensitivity 70-90%
- Specificity 76-91%
- Available in multiple languages

SWYC: Milestones

- 12 age-specific forms, keyed to pediatric periodicity schedule.
- Includes cognitive, language, and motor skills
- Ages 1-65 months
- Administration time ~5min
- Sensitivity: Average across ages: 75.8%
- Specificity: Average across ages: 78.3%
- Available in multiple languages

Lipkin PH, Macias MM, and Council on Children with Disabilities, Section on Developmental and Behavioral Pediatrics. Promoting optimal development: identifying infants and young children with developmental disorders through developmental surveillance and screening. *Pediatrics.* 2020; 145(1): e20193449.

5. Know the psychometric properties and proper use of common directly administered developmental screening tools [Not an exhaustive list]

Brigance Early Childhood Screens III

- Practitioner elicited via parent-report/interview
- Ages 0 through 4-8y
- Normed on national, diverse and general/naturalistic sample
- Administration time 10-20min
- Sensitivity ~85-95% across all ages
- Specificity ~80%-93% across all ages

Battelle Developmental Inventory Screening Test-II

- Practitioner elicited via combination of interview, direct administration and observation
- Age range 0 through 7-11 years
- Normed on national, diverse and general/naturalistic sample
- Administration time 10-30min

- Sensitivity 72-93%
- Specificity 79-86%

Bayley Infant Neurodevelopment Screener (BINS)
- Normed on a large national and diverse sample (low birth weight sample)
- Published validation study available
- Age range 3-24 months
- Administration time 5-10 min
- Comprehensive, user-friendly manual
- Sensitivity 75-86%
- Specificity 75-86%

CAT/CLAMS
- User-friendly manual
- Large standardization sample
- Multiple published validation studies available
- Good for identification of language and cognitive delay
- Age range 2-36 months
- 15-20 min administration time
- Sensitivity 21-67% low risk population, 5-88% high risk population
- Specification 95-100% low risk & 82-98% high risk

Drotar D, Stancin T, Dworkin H, Sices L, Wood S. Selecting developmental surveillance and screening tools. *Pediatrics in Review.* 2008; 29(10):e52-e58.

Marks KP, LaRosa AC. Understanding developmental-behavioral screening measures. *Pediatrics in Review.* 2012; 33(10):448-456.

6. Know the psychometric properties and proper use of common parent-completed behavioral and emotional screening tools [Not an exhaustive list]

Ages and Stages Questionnaire: Social-Emotional-2 (ASQ-SE)
- Screening and surveillance of milestones in social-emotional and mental health
- Items focus on self-regulation, compliance, communication, adaptive functioning, autonomy, affect and interaction with people
- Ages 1-72 months
- Administration time 10-15 min
- Sensitivity 78%
- Specificity 95%
- Available in multiple languages

Pediatric Symptom Checklist (PSC)
PSC 17b (17 items)

- General psychosocial screening and functional assessment in the domains of attention, externalizing, and internalizing symptoms
- Ages 4-16y
- < 5 min administration time
- Subscales have obtained reasonable agreement with validated and accepted parent-report instruments
- Cronbach alpha was high for each subscale

PSC 35b (35 items)

- Youth self-report ≥11 yo
- Sensitivity 80-95%
- Specificity 68-100%

Strengths and Difficulties Questionnaire (SDQ)

- Resilience and psychosocial risk for mental health and social-emotional, behavioral skills.
- Generates indicators for conduct problems, hyperactivity, emotional symptoms, peer problems, and prosocial behavior.
- parent and teacher report 4–17; 3- to 4-y-old versions available.
- Youth selfreport:11–16 y
- Administration time 5-10 min
- Reliable and valid in various population for a number of general mental health conditions
- Sensitivity 63-94%
- Specificity 88-98%
- Cross-cultural research and translations

Lipkin PH, Macias MM, and Council on Children with Disabilities, Section on Developmental and Behavioral Pediatrics. Promoting optimal development: identifying infants and young children with developmental disorders through developmental surveillance and screening. *Pediatrics.* 2020; 145(1): e20193449.

7. Know the standardized measures for assessing the home environment

Home Observation for Measurement of the Environment Inventory (HOME)

- 45-90 minute home visit with the primary caregiver and child present
- Observations made of parent/child interaction and discussions with the parent about objects, events, transactions with the goal to measure the quality and quantity of stimulation and support available to a child in the home environment
- Different versions for ages 0-3y, 3-6y, 6-10y, 10-14y and 16-21y

https://thesanfordschool.asu.edu/home-inventory

Home Screening Questionnaire (HSQ)

- Consists of a form that can be completed by parents without a home visit. Adapts segments of the HOME to a questionnaire format

- Administration time 15-20 minutes
- Scoring 5 minutes
- Different versions for ages 0-3y and 3-6y

Frankenburg WK, Coons CE. Home screening questionnaire: its validity in assessing the home environment. *Journal of Pediatrics.* 1986; 108(4): 624-626.

Stim Q Cognitive Home Environment

- Easy-to-administer instrument designed to measure cognitive home environment (no home visit required)
- Based on a questionnaire that is administered to the child's primary caregiver by a trained interviewer
- Available in 3 different forms: 5-12mo, 12-36 mo, 36-72 mo
- 4 subscales: Availability of Learning Materials, Reading, Parental Involvement in Developmental Advance, and Parental Verbal Responsivity
- Available in English and Spanish
- 15-20 minutes to administer, 2-3 minutes to score

https://med.nyu.edu/pediatrics/developmental/research/belle-project/stimq-cognitive-home-environment

8. Differentiate global from domain-specific developmental and behavioral screening instruments

Global developmental screens evaluate all areas of development. Domain-specific screens evaluate one to a few specific areas of development. Condition-specific developmental screens are made to screen for individual conditions. Behavioral screening instruments focus on behavioral concerns and screen for disorders such as ADHD, ODD, CD.

Marks KP, LaRosa AC. Understanding developmental-behavioral screening measures. *Pediatrics in Review.* 2012; 33(10):448-456.

9. Understand the issues important to hearing screening for infants in the newborn period and beyond and describe strategies to reduce loss to follow-up

Newborn hearing screening and re-screening protocols:

- All infants screened by 1 month of age
- Comprehensive evaluation by 3 months of age for those who did not pass screening
- Appropriate intervention by 6 months of age if confirmed hearing loss

- Infants admitted to NICU for >5 days should have ABR as part of screening to avoid missing sensorineural hearing loss
- For rescreening – complete screening on both ears even if only 1 ear failed initial screening
- Repeat hearing screen after readmission in the first month of life for infants with conditions associated with potential hearing loss (eg, hyperbilirubinemia, culture-positive sepsis)

Screening Method options:

- OAE only (may miss sensorineural hearing loss)
- ABR only
- 2 step protocol: Combined OAE + ABR. Decreases failure rate at discharge.

Strategies to reduce loss to follow-up:

- Improve communication with families:
    - Standardize or "script" the message given to the parents when an infant does not pass the initial screening test.
    - Get a second point of contact for the family (e.g., telephone number of a relative or friend).
    - Clear communication with the family about the results of the screening and appropriate follow up and resource information
    - Provide information through an interpreter for those who need it
- Improve infrastructure/tracking:
    - Standardize the process for collecting additional contact information for infants who do not pass their screening.
    - States should implement data-management and-tracking systems as part of an integrated child health information system to monitor the quality of EHDI services and provide recommendations for improving systems of care.
    - Information systems should be designed and implemented to interface with electronic health charts and should be used to measure outcomes and report the effectiveness of EHDI services at the patient, practice, community, state, and federal levels.
- Improve audiology appointment follow-up rates:
    - Schedule a follow-up appointment (rescreening or diagnostic evaluation appointment) at the time that the infant does not pass the screening—before the family leaves the hospital
    - Call the family before the diagnostic audiology appointment to verify the appointment time and place and include the reasons why the appointment is important. Offer assistance to get to the appointment, if necessary (e.g., transport vouchers).
    - Make two audiology appointments when scheduling diagnostic evaluations, so that the infant who cannot be completely evaluated at the first appointment is scheduled to return within a reasonable timeframe. Cancel the second appointment, if not needed.

- Improve communication between providers:
    - Verify the identity of the PCP or clinic responsible for follow-up with both the parent and assigned provider at the time the infant is screened before the family leaves the hospital.
    - Use a fax-back form at the time of diagnostic evaluation to alert the PCP of the results and need for follow-up. Use fax-back forms between all parts of the care continuum (audiology, PCP, specialists, early intervention).
    - Obtain consent from parents for release of information at first contact with early intervention, so that information can be shared between early intervention, the PCP, and the state EHDI database.
    - Provide PCPs with early intervention reports with clinically useful and timely information for providers.

Joint Committee on Infant Hearing. Year 2007 Position Statement: Principles and Guidelines for Early Hearing Detection and Intervention Programs. *Pediatrics.* 2007;120(4):898- 921.

Winston-Gerson R, Hoffman J. Chapter 3: Tracking, Reporting & Follow-Up. *The NCHAM eBook: A Resource Guide for Early Hearing Detection and Intervention.* 2020. Accessed at infanthearing.org/ehdi-ebook/

10. Know the methods for assessing hearing at different developmental stages including physiologic measures, behavioral audiometry, conditioned response audiology, play audiometry,
and sound field audiometry

Physiologic measures (typically used 0-6mo)

- Otoacoustic emissions (OAE)
    - Measurement of normally produced sound responses generated by hair cells in the cochlea → measured by placing small probe w/ soft rubber tip in the ear and providing sound stimulation
    - Presence of OEAs indicates middle and inner ear functioning appropriately
    - Limitations: doesn't provide info about degree/severity of hearing loss and may not detect minimal or slight hearing loss or sensorineural hearing loss
- ABR (auditory evoked response)
    - Measures the auditory nerve's response to sound
    - 3-4 electrodes placed on child's head near the ears → provide stimuli (clicks) through headphones)→ presence or absence of waveforms at specific sound levels and frequencies can confirm/describe hearing loss
    - Child must be completely still – require sedation ages 6m-7y
    - Different types used for screening (automated ABR) vs diagnosis

Behavioral audiometry

- Sounds played and child's response monitored and recorded

- Behavioral observation audiometry: 0-5mo
    - Audiologist observes/records response to sounds (quieting, eye widening, startle)
- Visual reinforcement audiometry: 6m-2y
    - Observes and records when child turns to the sound stimulus and gives a visual reinforcement or reward that is timed to the response (toy or puppet lights up/moves)
- Conditioned orientation reflex: same as VRA but includes more than one sound source and puppet reinforce used, i.e. one on left and one on right
- Conditioned play audiometry: 2-5y
    - Audiologist establishes a "listening game" by using toys to maintain the child's attention and focus on the listening task (i.e. drop a block in the bucket when sound is heard)
- Conventional audiometry (>5y)
    - Child raises hand or provides verbal response to presentation of various sounds
- Soundfield audiometry
    - Sounds presented via speakers rather than headphones (used when child cannot tolerate headphones)
    - Cannot test ears separately, results reflect hearing in stronger ear

(https://www.asha.org/public/hearing/Pure-Tone-Testing/)

Foust T, Hoffman J. Chapter5: An Introduction to Audiology for Nonaudiologists. *The NCHAM eBook: A Resource Guide for Early Hearing Detection and Intervention.* 2020. Accessed at infanthearing.org/ehdi-ebook/

11. Know the methods for assessing vision at different developmental stages

At all ages:

- Assess visual system history: family history of eye disorders, parent observations/concerns re: vision, strabismus, ptosis, eye injuries
- Perform ocular examination:
    - External examination of ocular structures (eyelids, conjunctiva, sclera, cornea, iris) evaluating for ptosis, nonresolving conjunctivitis, cloudy/enlarged cornia, photophia, eye discharge (particularly <1 yo → nasolacrimal duct obstruction)
    - Red reflex testing evaluates for
        - Asymmetric brightness – unequal refractive error
        - Absent – opacity i.e. cataract
        - Temporal displacement – strabismus
        - White – consider retinoblastoma
    - Pupil examination: asymmetry >1mm often attributable to ocular injury/disease or neurologic disorder, <1mm generally benign unless associated w/ ptosis or ocular motility deficit
    - Ocular alignment and motility

        - Corneal light reflex test (i.e. Hirschberg test) - abnormal if light reflex is displaced
        - Cover test: child fixates on small target, each eye alternately uncovered; shift in eye's alignment as it assumes fixation on the target indicates possible strabismus
    - Ophthalmoscopy: visualize structures in the back of the eye in older, cooperative children
- Visual acuity:
    - Preverbal children/<3 yo: check child's ability to fixate & follow an object held in front
    - Starting at age 3-4 yo: use age-appropriate optotype:
        - LEA symbols (circle, square, apple, house) or HOTV (letters – H, O, T, V) charts initially, Snellen eye chart when knows letters
    - Instrument-based screening:
        - Can attempt starting at 12 months
        - Photoscreener: identify optical characteristics of the eyes to estimate refractive error, media clarity, ocular alignment, and eyelid position

Donahue SP, Baker CN, Committee on Practice and Ambulatory Medication, Section on Ophthalmology, American Association of Certified Orthoptists, American Association for Pediatric Ophthalmology and Stabismus and American Academy of Ophthalmology. Procedures for the Evaluation of the Visual System by Pediatricians. *Pediatrics.* 2016;137(1):e20153597.

12. Be able to identify common dysmorphic features
Definitions and pictures:
https://elementsofmorphology.nih.gov/index.cgi?lid=fcb5dcb0a3c3dae5

Cranium:
Brachycephaly: shortened anteroposterior length of the head compared to width
Dolichocephaly: apparently increased anteroposterior length of head compared to width
Trigonocephaly: wedge-shaped head (apex of triangle at the midline of the forehead)
Turricephaly: tall head relative to width and length

Forehead:
Frontal bossing: bilateral bulging of the lateral frontal bone prominences with relative sparing of the midline
Prominent glabella: forward protrusion of the glabella (area of the forehead in midline between supraorbital ridges)

Maxilla/Midface/Mandible:
Midface retrusion: Posterior positioning and/or vertical shortening of the infraorbital and perialar regions, or increased concavity of the face and/or reduced nasolabial angle
Retrognathia: posteriorly positioned lower jaw
Micrognathia: apparently reduced length and width of the jaw when viewed from the front

Eyes:
Palpebral fissure, downslanted
Palpebral fissure, upslanted
Epicanthal fold: A fold of skin starting above the medial aspect of the upper eyelid and arching downward to cover, pass in front of and lateral to the medial canthus
Hypertelorism: widely spaced eyes
Hypotelorism: closely spaced eyes
Synophrys: meeting of the medial eyebrows in the midline

Nose:
Wide nasal bridge: increased breadth of the nasal bridge

Philtrum:
Smooth philtrum: flat skin surface, with no ridge formation in the central region of the upper lip between the nasal base and upper vermillion border

Oral Cavity:
Macrodontia: large teeth
Macroglossia: large tongue
High palate: height of the palate more than 2 SD above the mean (height at the first permanent molar more than twice the height of the teeth)
Cleft uvula: uvula separated into two parts most easily seen at the tip
Hand/Foot:
Clinodactyly: a digit that is laterally curved in the plane of the palm
Cutaneous syndactyly: soft tissue continuity in the A/P axis between two fingers that etends distally to at least the level of the PIPJ
Preaxial polydactyly: duplication of all or part of the first ray (thumb side)
Split hand/foot (i.e. lobster claw deformity): longitudinal deficiency of a digital ray of the hand except rays 1 and 5
Postaxial polydactyly: presence of supernumerary digit that is not a thumb/great toe
Sandal gap: a widely spaced gap between the great toe and the second toe

13. Know appropriate assessments to evaluate a child's motor coordination [not an exhaustive list]
Alberta Infant Motor Skills

- Ages 0-18months
- 58 item, performance-based, norm-referenced, observational tool.
- Good for assessing for delays and following motor development over time

https://libguides.lib.umanitoba.ca/c.php?g=297419&p=1993829

Bayley-III

- Assessment motor development of children 1-42 months of age
- Identify children with developmental delays and provide information for intervention planning

https://libguides.lib.umanitoba.ca/c.php?g=297419&p=1993817

Beery VMI

- Assessment designed to measure the integration of visual perception and motor behavior
- Paper and pencil
- 2-18 yo

https://libguides.lib.umanitoba.ca/c.php?g=297419&p=1993819

Movement Assessment Battery for Children -2

- Standardized test (administered to child) + checklist (completed by caregiver)
- Focus on identification and description of impairments of motor function in children
- 3-17 y

https://libguides.lib.umanitoba.ca/c.php?g=297419&p=1993877

Bruininks-Oseretsky Test of Motor Proficiency 2nd Ed (BOT-2)

- Discriminative, norm-referenced test
- Ages 4-21 years
- Evaluates balance and coordination, gross and fine motor performance

https://libguides.lib.umanitoba.ca/c.php?g=297419&p=1993877

14. Know the procedures to evaluate a child's mental status

Evaluate the following components:

- Appearance: weight, height, nutritional status, precocious or delayed physical maturation, personal hygiene, style/appropriateness of dress
- Motor behavior: general level of physical activity, abnormalities of gait, balance, posture, tone, fine/gross motor coordination, abnormal movements
- Voice, Speech and Language: pitch, tone, volume, phonation, prosody, tempo, abnormal articulation, unusual or inappropriate use of words, echolalia, abnormal syntax
- Interaction with examiner: eye contact, friendly/cooperative, resistant, oppositional, shy, withdrawn, reliable informant
- Mood and affect: persistent abnormality of mood or poor emotional regulation, emotional lability, range of affect
- Cognitive functions: test attention, orientation, memory, judgment, abstraction and intelligence
- Thought processes: tempo of thoughts (flight of ideas, acceleration, slowing, poverty), stream of thought (goal direction, incoherence, tangential thinking, derailment, clang associations), perseveration, logical/metaphorical thinking
- Thought content: i.e. anxiety, separation anxiety, school refusal, panic attacks, phobias, obsessions, compulsions, impulsions, delusions, hallucinations, ideas of reference, ideas of influence, thought alienation, thought-broadcasting, depersonalization, déjà vu, derealization, suicidal ideation, homicidal ideation
- Fantasy: elicited through play, drawing, story telling
- Insight: is child aware that he has a problem

Nurcombe B, Tramontana M, LaBarbera JD. Chapter 7. Diagnostic Evaluation for Children and Adolescents. *Current Diagnosis and Treatment: Psychiatry, 2nd edition.* Ebook accessed at:

https://accessmedicine.mhmedical.com/content.aspx?bookid=336&sectionid=39717878#3281523

15. Know interview and screening methods to assess a child's or adolescent's emotions and moods

PHQ-2 and PHQ-9

- Depression screen if + 2 question screen → 9 question screen
- incorporates DSM-IV depression criteria with other leading major depressive symptoms into a brief self-report instrument

https://www.apa.org/pi/about/publications/caregivers/practice-settings/assessment/tools/patient-health

SCARED

- 41 items – youth and parent versions
- Obtain total score + subscales (separation anxiety, GAD, school avoidance, panic disorder/somatization, social anxiety)
- Ages 8-18y

https://www.pediatricbipolar.pitt.edu/resources/instruments

SDQ (see above)

Children's Depression Inventory

- Completed by children 7-17 yo
- 27 or 10 items

https://www.sciencedirect.com/topics/medicine-and-dentistry/childrens-depression-inventory

16. Understand the important historical information required to assess the possible etiologies for developmental delay/disabilities

Family History:

- 3 generations, maternal and paternal
- Consanguinity
- Previous pregnancy outcomes: miscarriages, stillbirths, neonatal or childhood deaths
- Family history of birth defects, childhood deaths, ID, developmental delays, autism, known genetic conditions, learning disabilities
- Ethnic background

Prenatal history:

- Potential teratogen exposure, including substances, infections, maternal conditions (i.e. diabetes, PKU)
- Fetal movements
- Results of prenatal tests

Perinatal history:

- Gestation, mode of delivery, Apar scores, resuscitation
- Birthweight, length, head circumference
- Feeding, muscle tone, other problems

Postnatal history:

- Milestones, school performance
- Regression?
- Unusual behavior, personality
- Coordination, seizures, unusual movements, increased/decreased tone
- Growth (height, weight, head circumference)
- Previous illnesses
- Hearing, vision

Meschino WS. The child with developmental delay: an approach to etiology. *Paediatr Child Health.* 2003;8(1):16-19.

17. Know the appropriate laboratory evaluation for developmental delay

After completing medical history, 3 generation family history and physical, dysmorphologic, and neurologic examinations:

- If specific diagnosis is suspected, arrange for appropriate diagnostic studies to confirm
- If diagnosis is unknown/no clinic diagnosis suspected, follow stepwise process:
    1. All: Chromosomal microarray, Fragile X testing

    Consider metabolic testing: serum total homocysteine, acyl-carnitine profile, amino acids; and urine organic acids, glycosaminoglycans, oligosaccharides, purines, pyrimidines, GAA/creatine metabolites

    2. If no dx found:
        - For male and family history suggestive of X-linkage: XLID panel that contains genes causal of nonsyndromic XLID and complete high density X-CMA. Consider X-inactivation skewing in the mother of the proband
        - Female: MECP2 deletion, duplication, and sequencing study

    3. If microcephaly, macrocephaly or abnormal neuro exam: brain MRI

    4. If still no dx: consider referrals to other specialists, signs of inborn errors of metabolism (which haven't been screened for)

Moeschler JB, Shevell M, Committee on Genetics. Comprehensive Evaluation of the Child with Intellectual Disability or Global Developmental Delays. *Pediatrics.* 2014, 134(3):e903-e918. Reaffirmed October 2019.

18. Know the indications for different types of molecular genetic tests

In autism:
Genetic Testing: Recommended and offered to all families. Dysmorphic features or ID increases likelihood of finding genetic abnormality.

- CMA analysis

- Fragile X-analysis
- Consider MECP2 testing & evaluation for Rett syndrome if female patient
- Other genetic/metabolic testing guided by family history and physical exam
- If no etiology from above testing, can consider whole exome testing
- Referral to genetics/neurology is appropriate if specific syndrome or metabolic disorder suspected (i.e. male w/ marked macrocephaly, pigmented macules on penis – evaluate for PTEN)

In GDD/ID:

- See content spec above (#17)

Identification, Evaluation, and Management of Children With Autism Spectrum Disorder. Susan L. Hyman, Susan E. Levy, Scott M. Myers, COUNCIL ON CHILDREN WITH DISABILITIES, SECTION ON DEVELOPMENTAL AND BEHAVIORAL PEDIATRICS Pediatrics Jan 2020, 145 (1) e20193447; **DOI:** 10.1542/peds.2019-3447

19. Know the indications for cytogenetic testing
See above indications for cytogenetic testing in autism and GDD/ID.

Other (more general) indications for cytogenetic testing: advanced maternal age (>35 years), multiple abnormalities on fetal ultrasound, multiple congenital anomalies, unexplained growth retardation in the fetus, ambiguous genitalia, primary amenorrhea or infertility, recurrent miscarriages/hx of stillbirths/neonatal deaths, clinical findings consistent with a known anomaly, some malignancies and chromosome breakage syndromes (Bloom syndrome, Fanconi anemia)

Bacino CA, Lee B. Cytogenetics. In: Kliegman RM et al, eds. *Nelson Textbook of Pediatrics*. 19th ed. Elsevier Saunders; 2011,394:413.

20. Know the indications for biochemical/metabolic testing
In the neonate:

- Lethargy, poor feeding, seizures, hypotonia, vomiting should prompt consideration of metabolic testing; hepatomegaly important associated finding, sometimes peculiar odor (i.e. maple syrup urine disease, PKU)

In older children:

- Intellectual disability or autism and presence of other symptoms: motor deficits, neurological deterioration or regression, seizures, myopathy, recurrent emesis, cardiomyopathy; can sometimes have an episodic or intermittent pattern of symptoms of vomiting/acidosis/mental deterioration/coma or cyclical behavioral changes; enlargement of liver or spleen, family history suggestive

Rezvani I, Rezvani G. An approach to inborn errors of metabolism. In: Kliegman RM et al, eds. *Nelson Textbook of Pediatrics*. 19th ed. Elsevier Saunders; 2011,394:413

Mew NA, MacLeod E, Batshaw ML. Inborn Errors of Metabolism. *Children with Disabilities, 8th ed.* Batshaw ML, Roizen NJ, Pellegrino L, ed. 2019: Baltimore, Brookes Publishing Co.

21. Know the indications for neuroimaging
In GDD/ID: MRI if microcephaly, macrocephaly, or abnormal findings on neurologic examination (focal motor findings, pyramidal signs, extrapyramidal signs, intractable epilepsy, or focal seizures)

Moeschler JB, Shevell M, Committee on Genetics. Comprehensive Evaluation of the Child with Intellectual Disability or Global Developmental Delays. *Pediatrics.* 2014, 134(3):e903-e918. Reaffirmed October 2019.

In autism:
Need for MRI directed by history and physical exam

- Atypical regression
- Micro or macrocephaly
- Seizures
- Intracranial manifestations of genetic disorders
- Abnormal neurologic exam

Identification, Evaluation, and Management of Children With Autism Spectrum Disorder. Susan L. Hyman, Susan E. Levy, Scott M. Myers, COUNCIL ON CHILDREN WITH DISABILITIES, SECTION ON DEVELOPMENTAL AND BEHAVIORAL PEDIATRICS Pediatrics Jan 2020, 145 (1) e20193447; **DOI:** 10.1542/peds.2019-3447

In hearing loss:

- CT if conductive hearing loss
- MRI if sensorineural hearing loss

Shekdar KV, Bilaniuk LT. Imaging of pediatric hearing loss. *Neuroimaging Clinics of North America*. 2019; 29(1): 103-115.

In microcephaly:

- Congenital microcephaly: if disproportionate and no other features to suggest another condition → MRI, if proportionate + neurologic symptoms or family hx of neuro disease → MRI, if proportionate and no symptoms, no family history → can CONSIDER MRI if new neuro signs/sx or worsening microcephaly
- Postnatal microcephaly: if disproportionate and no other features to suggest another condition AND severe (<3 SD) OR neuro signs/symptoms→ MRI, if proportionate + neurologic symptoms or family hx of neuro disease → MRI, if proportionate and no symptoms, no family history → can CONSIDER MRI if new neuro signs/sx or worsening microcephaly

American Academy of Neurology. Practice Parameter: Evaluation of the Child with Microcephaly. AAN Summary of Evidence-based Guideline for CLINICIANS. 2009.

22. Understand the importance of identifying child and family strengths in the process of planning intervention
→ mostly self-explanatory – it is important
- Notably, early intervention/IFSP development is often heavily strength-focused
- Evidence suggests that programs designed to emphasize and support the strengths of families rather than focusing on children alone is more beneficial in facilitating child development
- Important benefits of recognizing and incorporating the child's strengths into interventions include maximizing independence and improving self-image.

Kral MC. Interpreting psychoeducational testing reports, individualized family service plans , and individualized education program plans. In: Voight RG, Macias MM, Myers SM, Tapia CD, editors. American Academy of Pediatrics Developmental and Behavioral Pediatrics. 2nd ed. 2018. American Academy of Pediatrics. Itasca, IL.

Rose L, Herzig LD, Hussey-Gardner B. Early Intervention and the Role of Pediatricians. *Pediatrics in Review.* 2014;35(1) e1-e10.

23. Know how to interpret results of infant cognitive assessments
Common developmental/intelligence tests used in infancy:

- Mullen Scales – ages 0-68 months, visual receptive/expressive and language receptive/expressive
- Bayley Scales of Infant and Toddler Development-III: ages 16 days to 42 months; cognitive, language (expressive/receptive), motor (fine/gross), social-emotional, adaptive

Limited ability to predict school-age functioning exists until age 18-24 months.
Tests are developmental in a nature and therefore IQ per se cannot be measured until older than age 2 (preferably 3 or older).
Direct effects of early events (i.e. pre-term birth, hypoxic-ischemic encephalopathy) have larger effects; environmental effects become more influential as child ages.
At 3-4 years, IQ, executive function, pre-academic readiness, visual motor integrative skills and verbal/nonverbal discrepancies can be assessed; weak test floors at early ages = inflation of scores at low end, refusals are problematic.

Aylward GP. Issues in neurodevelopmental testing of infants born prematurely: the Bayley Scales of Infant Development Third Edition and other tools. *Follow-up for NICU graduates.* Needelman H, Jackson BJ, ed. 2018, Springer: Cham, Switzerland.

24. Know how to interpret results of verbal and non-verbal cognitive assessments in children
General principles of interpreting neuropsych tests (can apply to most of the specifications asking about "interpreting results"):

Norm-referenced testing gives standardized scores that can be presented in multiple ways – percentile score, T score, standard scores or scale scores. See figure below (from Batshaw Chapter 13).

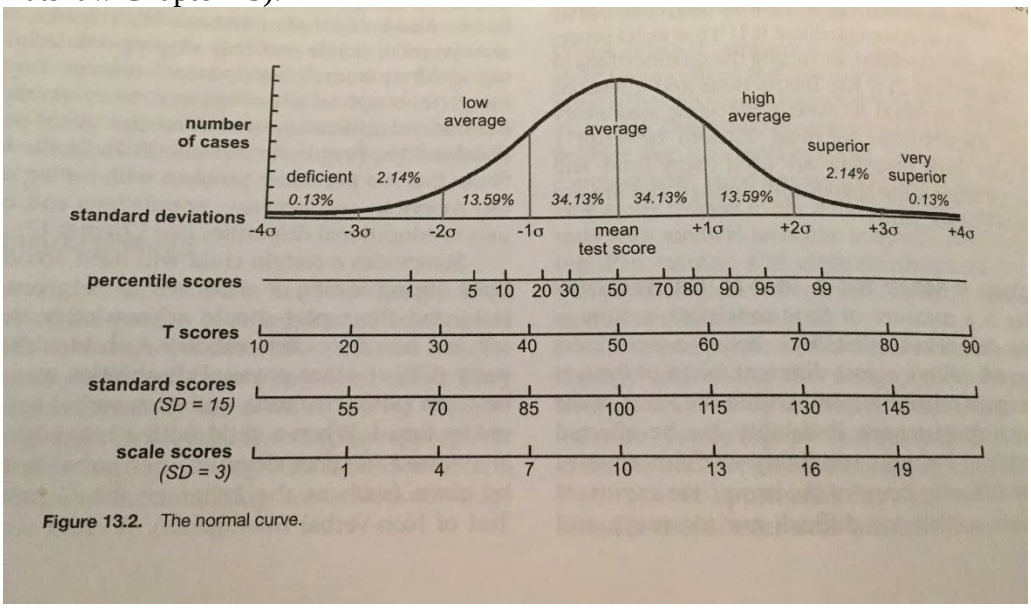


Figure 13.2. The normal curve.

Test scores can also be expressed as age or grade equivalents: indicate that performance is typical of a specific age group or grade level in the normative sample. These are easily understood by parents and teachers but should not be used for making diagnostic/placement decisions due to low reliability and validity. In the specific case of academic achievement testing, they also do not indicate that the student has acquire all of the knowledge of a child that grade or age.

Specific points related to cognitive testing:

- IQ scores capture only a fraction of the many abilities that govern a person's performance in the real world
- Verbal knowledge measures are affected by the home and school environment
- IQ scores are unstable in young children and not necessarily predictive of later performance on intelligence tests

Verbal vs nonverbal cognitive testing:

- verbal testing not appropriate in individuals that cannot be assessed via verbal interaction (i.e. a speech or language disorder, hearing impairments, culturally different background, neurologic trauma, selective mutism)
    - Examples of nonverbal measures: TONI-4, UNIT (Universal Nonverbal Intelligence Test), WASI and WISC-IV have good nonverbal subtests
    - (caveat – some nonverbal measures require more verbal instructions than others do; some tests simply have nonverbal components; TONI and UNIT best at being nearly entirely nonverbal)
- Large difference between verbal and nonverbal performance (15 points or more) might suggest a significant problem that is deserving of further investigation (learning disability, language impairment, etc. If difference is large full scale IQ can be misleading as their profile is more complex that one number can indicate.

For an intellectual disability diagnosis via DSM-V: requires deficits in intellectual and adaptive functioning. Regarding the IQ scores generally accepted as indicating deficit in intellectual function: scores of approximately 2SD or more below the population mean including margin for measurement error (70±5)

Kenworth L, Gutermuth Anthony L. Neuropsychological Assessment. *Children with Disabilities, 8th ed.* Batshaw ML, Roizen NJ, Pellegrino L, ed. 2019: Baltimore, Brookes Publishing Co.

McCallum RS. Context for Nonverbal Assessment. *Handbook of Nonverbal Assessment.* 2017: Knoxville, Springer International Publishing.

American Psychiatric Association. Diagnostic and Statistical Manual of Mental Disorders. 5th ed. Arlington, VA: American Psychiatric Association; 2013

25. Know how to interpret results of academic achievement testing

See content spec 24 for basic interpretation concepts.

For diagnosis of learning disability: academic achievement testing is one component of the evaluation.
Per DSM V: "Low achievement scores on one or more standardized tests or subtests within an academic domain (i.e., at least 1.5 standard deviations [SD] below the population mean for age, which translates to a standard score of 78 or less, which is below the 7th percentile) are needed for the greatest diagnostic certainty. However, precise scores will vary according to the particular standardized tests that are used. On the basis of clinical judgment, a more lenient threshold may be used (e.g., 1.0–2.5 SD below the population mean for age), when learning difficulties are supported by converging evidence from clinical assessment, academic history, school reports, or test scores. If intellectual disability is present, specific learning disorder can be diagnosed only when the learning difficulties are in excess of those usually associated with the intellectual disability."

American Psychiatric Association. Diagnostic and Statistical Manual of Mental Disorders. 5th ed. Arlington, VA: American Psychiatric Association; 2013

26. Know how to interpret results of motor assessments
See content spec 24 for basic interpretation concepts.

Three common standardized tests used for detailed developmental evaluation of a child's motor development are the Bayley Scales of Infant Development, Peabody Developmental Motor Scales and the Toddler Infant Motor Evaluation. Results for these are given as standard scores, percentile ranking or age-equivalent performance.

The Pediatric Evaluation of Disability Inventory and Gross Motor Function Measure were developed for measurement of motor function, intervention planning, developmental progress, and motor outcome in children with CP and related disabilities. PEDI scores includes standard scores and scaled scores and reflects functional skills attained. The GMFM is designed for measurement of change and uses a percentage score and an interval-level score.

Lipkin PH. Motor Development and Dysfunction. *Developmental-Behavioral Pediatrics.* 4th edition. Carey WB, Crocker AC, Coleman WL, Elias ER, Feldman HM eds. 2009: Saunders-Elsevier. Philadelphia, PA. pp643-652.

27. Understand the implications of neuromaturational delays for a child with school problems

In relation to ADHD:

- cortical development in ADHD lags behind typically developing children by years but follows normal sequence of brain development
- Cortical delay is most prominent in the lateral prefrontal cortex (area responsible for suppressing inappropriate responses, executive control of attention, evaluation of reward contingencies, higher-order motor control, working memory)

Reiff MI, Stein MT. Attention-Deficit/Hyperactivity Disorder. American Academy of Pediatrics Developmental and Behavioral Pediatrics. 2nd ed. Voight RG, Macias MM, Myers SM, Tapia CD, editors. 2018. American Academy of Pediatrics. Itasca, IL.

28. Recognize the impact of cultural differences on developmental testing performance

Cultural differences can have a large impact on developmental testing performance for many reasons:

- People from different cultures have varying beliefs about normal development as well as varying child-rearing practices (i.e. some place more emphasis on toy play vs family play, encourage more or less independence, more emphasis on quiet play, etc.)
- Linguistics – even if translated into the child's language, words may have slightly different meanings in other language and affect the outcome of the test
- Differing values/beliefs from the dominant culture
- Differing exposures due to environment (i.e. children from New York City may rarely know the answer to "how many wheels does a wheelbarrow have?")

Pacter L, Dworkin P. Maternal Expectations about Normal Child Development in 4 Cultural Groups. *Arch Pediatr Adolesc Med*. 1997;151:1144-1150.

Mushquash C, Bova DL. Cross-cultural assessment and measurement issues. *Journal on Developmental Disabilities.* 2007;13(1):53-65.

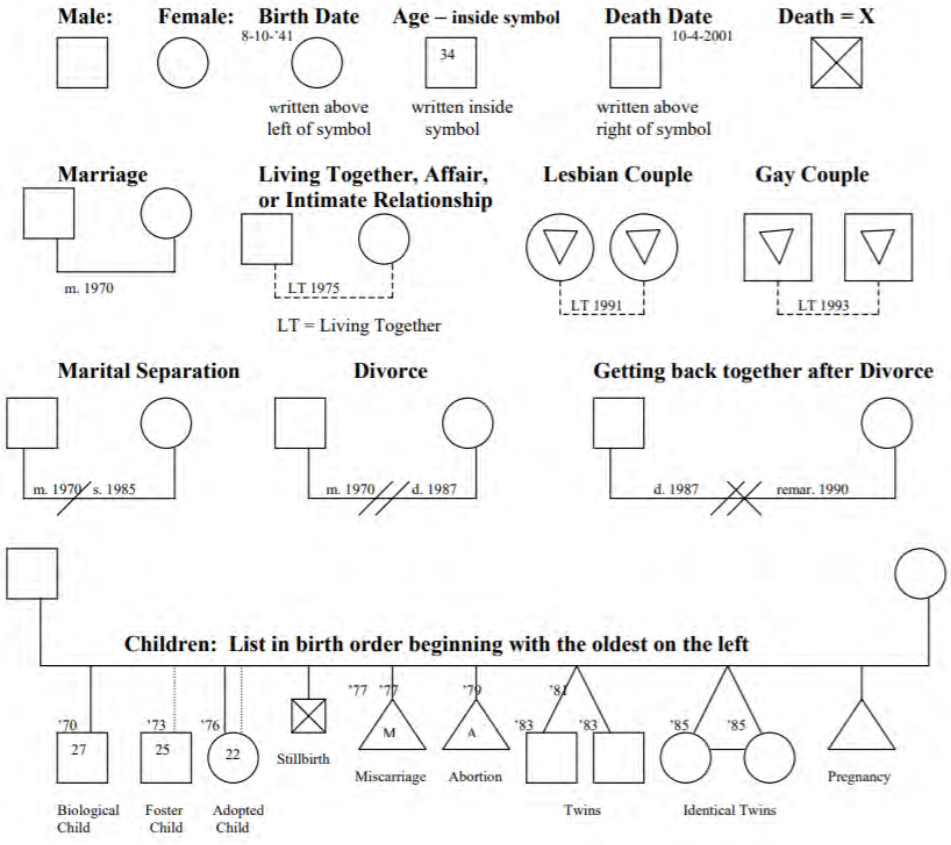
29. Know how to obtain and interpret a genogram for the understanding of family function

A genogram is a pictorial display of a person's family relationships and medical history.

- Allows clinician to have an informed understanding of the client's family situation and his/her primary social context
- Details of family history are taken and presented in a standard format (see symbols below)
- Names, ages, occupations, relationships, geographic locations are recorded
- Other information including miscarriages, adoptions, separation, remarriages, infertility, causes of death, legal problems, etc can also be included
- Clinician should be aware of any unusual configurations or family groupings and note significant developmental stages and events alongside accompanying crises or difficulties
- Recent stressful events and life events which appear to have occurred at unusual times are also relevant
- Use of the genogram over time enables the clinician to apply knowledge of a family's behavior patterns, key family events and of individual family members to the care of a family over time
- Patterns in the family's development can frequently be observed by the family in the genogram and process of discussing the chart can be an interactive process between clinician and patient

Stanion P, Papadopoulos L. Genograms in counselling practice: constructing a genogram (part 2). *Counselling Psychology Quarterly.* 1997; 10 (2):

Standard Genogram symbols:



https://calswec.berkeley.edu/sites/default/files/genogram_symbols_063018.pdf

30. Know the principles of behavioral screening

- The US health care system does not reliably identify or treat children's behavioral or emotional concerns (<1/8 children with B/E concerns receive treatment).
- Clinicians' ability to identify developmental and behavioral problems in primary care, on the basis of clinical judgment alone has been shown to have low sensitivity, ranging from 14% to 54% and a specificity ranging from 69% to 100%. Providers are less likely to identify problems in minority or non–English-speaking children and adolescents.
- Implementing behavioral screening can improve identification of B/E concerns.

Weitzman C, Wegner L and the Section on Developmental and Behavioral Pediatrics, Committee on Psychosocial Aspects of Child And Family Health, Council on Early Childhood, and Society For Developmental and Behavioral Pediatrics. *Pediatrics.* 2015;135 (2): 384-395; DOI: https://doi.org/10.1542/peds.2014-3716

31. Know the psychometric properties and proper use of common child or adolescent self-report behavioral and emotional screening measures [not an exhaustive list]
Baby Pediatric Symptom Checklist:
- 2-17 mo, 12 items, parent completed
- Psychometrics: retest and internal reliability

Preschool Pediatric Symptom Checklist:
- 18-60 mo, 18 items, parent completed
- No psychometrics available

Strengths and Difficulties Questionnaire:
- Ages 3-17 years, 25 items
- Parent/teacher 3(4)-y-old; parent/teacher 4–10-y-old; parent/teacher follow-up forms available
- Psychometrics: Variable across cultural groups; sensitivity: 63%–94%, specificity: 88%–96%; available in >70 languages

Pediatric Symptom Checklist (17 or 35 items)
- Ages 4-16yo
- Parent completed; youth self-report >10 y; pictorial version available
- 35: Sensitivity: 80%–95%, specificity: 68%–100%; available in multiple languages

Ages & Stages Social-Emotional (ASQ:SE-2)
- 1-72 months old
- 22-36 items depending on age (10-15 min)
- English, Spanish and French

Brief Infant-Toddler SE Assessment- BITSEA
- 12-36 months
- 46 items, 2 scales
- Sliding scale of cost depending on use

Weitzman C, Wegner L and the Section on Developmental and Behavioral Pediatrics, Committee on Psychosocial Aspects of Child And Family Health, Council on Early Childhood, and Society For Developmental and Behavioral Pediatrics. *Pediatrics.* 2015;135 (2): 384-395; DOI: https://doi.org/10.1542/peds.2014-3716

32. Know methods to assess a child's or adolescent's attention span, impulsivity, and hyperactivity
Child Behavior Checklist (CBCL/6-18)

- Identifies problem behavior in youths ages 6-18 years, including possible disorders and internalizing or externalizing problems
- Checklist of 120-questions
- Scored on a 3-point scale that ranges from 0 (not true) to 2 (very true/often true)

Conners' Rating Scales

- Conners' Parent Rating Scale-Revised for parents/caregivers
- Conners' Teacher Rating Scale-Revised for teachers
    - Consist of 27/28 questions (short versions of the scale) divided into 4 subscales: 1)oppositional problems, 2) cognitive problems,3) hyperactivity and 4) an ADHD index
    - Scoring is based on a 4-point scale
- Conners-Wells' Adolescent Self-Report Scale for teenagers

Vanderbilt ADHD Rating Scales

- Vanderbilt ADHD Parent Rating Scalf
    - Provides information on a parent's perception of social functioning and school performance
    - Consists of 45 questions
    - Assess for oppositional/conduct/anxiety symptoms as well
- Teacher Rating Scale (VADTRS)
    - Provides information on school performance and ADHD symptoms
    - Consists of 43 questions

ADHD Rating Scale-IV (ADHD-RS-IV)

- Includes separate forms for parents/caregivers and teachers
- Based on an 18-item scale divided into subscales for hyperactivity/impulsivity and inattentiveness
- Scored on a 4-point frequency scale ranging from 0 = never/rarely to 3 = very often

https://chadd.org/for-professionals/clinical-practice-tools/

33. Know methods to assess the significance of a child's or adolescent's defiant or aggressive behaviors

"Broad" rating scales often are administered first that assess for symptoms associated with a variety of disorders, such as depression, anxiety, aggression, withdrawal, inattention, hyperactivity, and delinquent behavior: Youth Self Report, Child Behavior Checklist (Parent Report Form) and Teacher's Report Form, the Behavior Assessment System for Children, and Conners' Parent and Teacher Rating Scales – Revised

The Modified Overt Aggression Scale can also be useful.

- Assess 4 types of aggressive behavior: verbal aggression, aggression against property, autoagression, physical aggression
- Behaviors rated over the past week.
- https://depts.washington.edu/dbpeds/Screening%20Tools/Modified-Overt-Aggression-Scale-MOAS.pdf

Zahrt DM, Melzer-Lange MD. Aggressive Behavior in Children and Adolescents. *Pediatrics in Review.* 2011;32(8):325-332.

Harstad EB, Barbaresi WJ. Disruptive Behavior Disorders. American Academy of Pediatrics Developmental and Behavioral Pediatrics. 2nd ed. Voight RG, Macias MM, Myers SM, Tapia CD, editors. 2018. American Academy of Pediatrics. Itasca, IL.

34. Know methods to assess an adolescent's substance use

Screening: goal is to define the adolescent's substance use along a spectrum ranging from abstinence to addiction so that this information can be used to guide the next steps/intervention

Screening tools:

- S2BI (Screening to Brief Intervention): single frequency-of-use question per substance, includes tobacco, marijuana, and other/illicit drug use, discriminates among no use, no SUD, moderate SUD, and Severe SUD
- BSTAD (Brief Screener for Tobacco, Alcohol and Other Drugs): identifies problematic tobacco, alcohol and marijuana use
- NIAAA Youth Alcohol Screen: 2-question alcohol screen (screens for friends' and personal use)
- CRAFFT: designed to identify substance use, substance-related riding/driving risk and substance use disorder in ages 12-21.

  Have you ever ridden in a CAR driven by someone (including yourself) who was high or had been using alcohol or drugs?

  Do you ever use alcohol or drugs to RELAX, feel better about yourself or fit in?

  Do you ever FORGET things you did while using alcohol or drugs?

  Do you FAMILY and FRIENDS ever tell you that you should cut down on your drinking?

  Have you ever gotten into TROUBLE while you were using alcohol or drugs?

  2 or more positive indicates need for further assessment
- CAGE: Cut down/Annoyed/Guilty/Eye-opener

Levy SJL, Williams JF, Committee on Substance Use and Prevention. Substance Use Screening, Brief Intervention, and Referral to Treatment. *Pediatrics.* 2016; 138(1) e 20161211.

35. Know methods to assess an adolescent's sexual behaviors

Introduce Confidentiality/Time Alone:

- Review office confidentiality policies and office visit structure that includes time alone with the adolescent patient, with both patient and parent present (starting ~11yo)

Interview Techniques to Consider:

- Consider use of a questionnaire, as some adolescents may be more comfortable w/ written questionnaire on sensitive subjects
- Use open-ended questions that avoid yes/no response
- Reflection responses that mirror patient's feelings
- Restatement of the patient's feelings or summarizing the interview
- Clarification of a statement
- Use of questions that may give the provider insight into the patient
- Offering reassuring statements and supportive statements
- Use of gender-neutral language
- Make a normative statement first before jumping in

5 P's Framework:

- Partners (men/women/both, # partners in bast 2 and 12 months)
- Prevention of pregnancy
- Protection from STDs
- Practices (recent, vaginal/anal/oral)
- Past history of STDs
- Also, consider discussion of sexual assault/abuse, reproductive life plan, sexual problems (if sexually active)

Marcell AV, Burstein R, and Committee on Adolescence. Sexual and reproductive health care services in the pediatric setting. *Pediatrics.* 2017; 140(5):e20172858.

36. Know methods to assess family functioning

SWYC

• *Family factors (10 items): parental depression, tension in couple's relationship, substance use, smoking, food insecurity and shared reading

https://www.floatinghospital.org/the-survey-of-wellbeing-of-young-children/parts-of-the-swyc/family-questions

Parenting Stress Index:

- Construct: Identify stressful aspects of parent-child interaction
- Screening and triage measure for evaluating the parenting system and identifying issues that may lead to problems in the child's or parent's behavior. Focuses on three major domains of stress: child characteristics, parent characteristics and situational/demographic life stress.
- for use with parents of children ranging in age from 1 month to 12 years
- short form (10 min) and long form (20 min) available
- https://www.apa.org/pi/about/publications/caregivers/practice-settings/assessment/tools/parenting-stress

37. Know methods to assess a child's or adolescent's peer relationships

PROMIS Peer Relationship measures

(Patient Reported Outcome Measures/NIH)

• 8 item short forms

• Parent Proxy (5-17 yo), Youth (8-17)
• https://www.assessmentcenter.net/promisforms.aspx

* SDQ – Peer subscale (see further info above)
• 4-16 yo
• 5 questions

- Self-report: appropriate for middle childhood and adolescence. Potential for bias, must consider the child's level of cognitive and social understanding. Aggressive children, in particular are unreliable in self-reporting (tend to be more positive and discrepant from teacher's reports and playground observation).
- Peer assessments: typically measure friendships and general acceptance within the peer group. Often obtained from a class of children, so similar to having multiple informants. May be more accurate than adult reports as more likely to be present during both prosocial and antisocial interactions. May be difficult to obtain. Appropriate for all age levels (may need to be adapted for early childhood).
- Adult reports: Parent/teacher perceptions generally assessed with a checklist. May be systematically biased – mothers who are depressed rate children more negatively, some parents may be positively biased. Teachers reports limited to observed behavior (aggressive behavior/bullying may not be done in front of teachers), expectations influence perceptions and ratings of children's behavior, more effective in preschool and elementary school (younger kids have 1-2 teachers all day).
- Observation: Useful for research, less practical for clinical practice.

Pepler DJ, Craig WM. Assessing children's peer relationships. *Child Psychology & Psychiatry Review*. 1998: 3(4). 176-182.

38. Know methods to assess for social stressors
Safe Environment for Every Kid - (SEEK)
• 20 yes/no items
• maternal depression, alcohol and substance abuse, domestic violence, harsh parenting, major parental stress, food insecurity as well as, fire alarm and poison control #
https://Seekwellbeing.org

Adverse Childhood Experiences Questionnaire (ACE Q)
• Lists 10 ACEs (physical abuse, verbal abuse, sexual abuse, physical neglect, and emotional neglect, parent who's an alcoholic, a mother who's a victim of domestic violence, a family member in jail, a family member diagnosed with a mental illness, and the disappearance of a parent through divorce, death or abandonment) and asked to count exposure
• Lists 7 more ACEs and again asks for count, not specifics
• Parent (birth – 19 yo) and youth (13-19 yo) versions
https://elcentro.sonhs.miami.edu/research/measures-library/aces/index.html

39. Know interviewing techniques appropriate for children of various ages and developmental levels

Infancy: child held by caregiver, use soft tone of voice and gentle handling. Narrating to infant what will happen in the visit can make the infant and caregiver feel more comfortable

Toddler (1-3y): Toddler may seek more control and active participation in the visit. Buid rapport with toddler by allowing and encouraging exploration, being sensitive to toddler's needs for "emotional refuling" from the cargiver (caregiver = secure base and safe haven), indulge toddler's desire for autonomy and control (i.e. letting toddler listen to their own heart)

Preschool (3-6y): Egocentric and magical thinking = may view illness as punishment for certain behaviors or as something caused by magic. Can be helpful to reassure that illness is not his or her fault or a result of bad behavior. Probe for understanding of why he or she doesn't feel well; can engage with child in finding a solution.

School age (7-12y): Facilitate rapport by inquiring about school, hobbies, friends. Can be actively engaged in interview and directly queried about feelings/concerns/goals. Invite to assume greater responsibility in the treatment process

Adolescence (13-21y): Include time with parent and adolescent alone. Acknowledge confidentiality and its limitations before parent leaves the room. Identify mutually agreed-upon therapeutic goals to build an alliance with caregiver and adolescent.

Shah PE, Ribaudo J. Interviewing and Counseling Children and Families. American Academy of Pediatrics Developmental and Behavioral Pediatrics. 2nd ed. Voight RG, Macias MM, Myers SM, Tapia CD, editors. 2018. American Academy of Pediatrics. Itasca, IL.

40. Know how to interpret results of adaptive skill assessments

Limitations in adaptive behavior are defined: performance that is at least 2SD below the mean in any type of adaptive behavior (conceptual, social or practical) or an overall score on a standardized measure of conceptual, social and practical skills (American Association for Individuals with Developmental Disabilities)

Conceptual skills: receptive/expressive language, reading, writing, money conepts, self-directions
Social skills: interpersonal, responsibility, gullibility, naivete, follow rules/obeys laws, avoids victimization
Practical skills: ADLs, occupational skills, maintaining a safe environment

Vineland Adaptive Behavior Scales- 3rd edition

- Norm-referenced test, provides standard score (mean 100, SD 15)
- Interview, parent/caregiver and teacher forms
- Ages 0-90+ (3-18 for teacher forms)

Pearsonassessments.com

ABAS-III

- norm-referenced assessment of adaptive skills, provides standard score (mean 100, SD 15)
- measures adaptive behavior by reporting a General Adaptive Composite, which provides an overall level estimate of adaptive behavior, as well as in three adaptive domains (conceptual, social, and practical), and in individual adaptive skill areas (communication, community use, functional academics/pre-academics, home/school living, health and safety, leisure, self-care, self-direction, social, and motor).
- Ages birth – 89years

WPSpublishing

Problems of adaptive measures:

- Limited usefulness in planning interventions
- Reliance on parent report
- Scored based on typical performance rather than peak performance, which might be a better reflection of child's potential

Sturner R. General Principles of Psychological Testing. In *Developmental-Behavioral Pediatrics*, 4th ed. Ed by Carey WB, Crocker AC, Coleman WL, Elias ER, Feldman HM. 2009 Saunders Elsevier Philadelphia PA.

41. Know how to interpret results of speech and language assessments

Speech and Language assessments typically include evaluations of:

- Phonology (sound system of a language), including phonological awareness (metalinguistic awareness of all levels of speech sound system including word boundaries, stress syllables, phonemes)
- Semantics (meaning of language, vocabulary and word relationships)
- Morphology (understanding of morphemes – smallest units of language that have meaning)
- Syntax (structure/sentence structure/grammar)
- Pragmatics (rules and conventions for using language/gestures in social contexts), including discourse-level language skills (conversation, narrative, expository).
- Literacy assessment (due to well-established connection between spoken and written language)
- Speech sound assessment (articulation/phonology)

The following procedures and data sources may be utilized in the comprehensive assessment for spoken language disorders (SLD):

- Standardized Assessments—empirically developed evaluation tools with established reliability and validity. (few standardized assessments for individuals who speak languages other than English).
- Language Sampling—techniques to elicit spontaneous language in various communication contexts (e.g., free play, conversation/dialogue, narration, expository speech) and derive measures (e.g., Mean Length of Utterance [MLU], Type-Token Ratio [TTR], Developmental Sentence Scoring [DSS], clausal density, use of subordinate clauses) to complement data obtained from standardized language assessments.

- Dynamic Assessment—a language assessment method in which an individual is tested, skills are addressed, and then the individual is re-tested to determine treatment outcome (i.e., test-teach and re-test). Can help distinguish between language disorder and language difference
- Systematic Observation/Contextual Analysis—observation in the classroom and in various other contexts to describe communication and identify specific problem areas.
- Ethnographic Interviewing—a technique for obtaining information from the student and the student's family/caregiver and teachers that avoids the use of leading questions and "why" questions and uses open-ended questions, restatement, and summarizing for clarification.
- Parent/Teacher/Child Report Measures—checklists and/or questionnaires completed by the family member(s)/caregiver, teacher, and/or student.

Ekelman BL, Lewis BA. Speech and Language Disorders. *Children with Disabilities, 8th ed.* Batshaw ML, Roizen NJ, Pellegrino L, ed. 2019: Baltimore, Brookes Publishing Co.

American Speech-Language-Hearing Association. Spoken Language Disorders. Accessed at: https://www.asha.org/Practice-Portal/Clinical-Topics/Spoken-Language-Disorders/. Accessed July 8, 2020.

42. Understand current classifications of behavioral health disorders in infants and toddlers

DSM-5 (American Psychiatric Association)

- Some changes made from DSM-IVTR → V:
    - Diagnoses most common in infancy/childhood at start of each diagnostic chapter, rather than in individual chapter
    - Social communication disorder and disruptive mood dysregulation disorder added
    - Added more precise criteria to other diagnoses (ADHD, ASD, PTSD, etc)
    - See fact sheet for summarized changes:https://www.psychiatry.org/psychiatrists/practice/dsm/educationalresources/dsm-5-fact-sheets

American Psychiatric Association. Diagnostic and Statistical Manual of Mental Disorders. 5th ed. Arlington, VA: American Psychiatric Association; 2013

DC:0-5TM (previously DC:0-3R)

- Published by Zero to Three
- Includes disorders occurring in children from birth through 5 years old
- Criteria extended to younger ages when appropriate, including, in some cases, to the first year of life
- Introduces discussion of several new disorders, including:
    - Relationship-Specific Disorder of Early Childhood
    - Dysregulated Anger and Aggression Disorder of Early Childhood
    - Atypical Social-Communication Emergent Neurodevelopmental Disorder
- Retains the multi-axial system, allowing optimal consideration of context in assessment and diagnosis; most axes revised substantially

- Crosswalk between DC:0-5 and DSM-V/ICD 10 on Zero to Three Website: https://www.zerotothree.org/resources/1540-crosswalk-from-dc-0-5-to-dsm-5-and-icd-10

Zero to Three. DC 0-5 Manual and Training. Accessed at: https://www.zerotothree.org/resources/2221-dc-0-5-manual-and-training. Accessed July 12, 2020.

43. Know the indications for fluorescence in situ hybridization (FISH) testing

FISH testing: appropriate if specific diagnoses suspected, i.e. Velo-cardiofacial syndrome, FISH also appropriate if unable to obtain microarray in patient with GDD/ID of unknown etiology

American Academy of Neurology. AAN Summary of Evidence-Based Report for Clinicians: Genetic and Metabolic testing in Children with Developmental Delay. 2011.

44. Know the indications for neuroelectrophysiological studies (e.g., electroencephalography)

- EEG Indications:
    - Altered states, such as coma
    - Evaluation of brain death
    - Clinical spells or paroxysmal events
    - Classification of seizures
    - Localized versus generalized
    - Specific patterns associated with epilepsy syndromes
    - Epilepsy diagnosis and prognosis
    - Decisions about weaning anticonvulsant(s)
    - Epilepsy monitoring
    - Monitoring of sleep states (polysomnography)
    - Monitoring the depth of general anesthesia
    - Monitoring status epilepticus during neuromuscular blockade or medication-induced coma
    - Monitoring nonconvulsive status epilepticus
    - Intensive care monitoring for cerebral ischemia and infarction
- Evoked potentials: assess integrity of neural pathways
    - Visual:
        - Assessment of visual pathways from the retina to the occipital cortex
        - Evaluation of visual function, visual loss, and optic nerve disease
    - Auditory (brainstem auditory evoked potential)
        - Assessment of sequential activation of the subcortical auditory pathway from the inner hair cell of the cochlea to the midbrain

        - Physiologic hearing assessment (with or without other modalities); evaluation of brain stem function
    - Somatosensory
        - Assessment of sensory pathways at multiple levels (ie, peripheral, spinal, subcortical, cortical)
        - Evaluation of spinal cord lesions; preoperative and intraoperative monitoring during spine surgery and certain types of upper and lower extremity procedures
    - Motor-evoked potentials
        - Assessment of corticospinal tract pathways
        - Evaluation of motor neuron disease, spinal cord lesions, and monitoring during spinal cord surgery

EMG (Electromyography)

- Identify types of myopathic or neuromuscular conditions

Nerve conduction studies

- Indications: acute nerve injury, chronic nerve disorders, detect neuropathic disorders in individuals who have hereditary risk prior to sx, identify unaffected nerves to distinguish a mononeuropathy from a polyneuropathy, follow the progression of a neuropathic condition

Indications for EMG and nerve conduction studies together:

- Assessment of: myopathies, disorders of the neuromuscular junction, and neuropathies caused by axonal degeneration, demyelination, and conduction blocks
- Serial studies: follow the effectiveness of treatment, determining prognosis on the basis of disease progression

Gooch CL, Weimer LH. The electrodiagnosis of neuropathy: basic principles and common pitfalls. *Neurol Clin*. 2007;25(1):1-28. doi:10.1016/j.ncl.2007.01.011.

Hirtz D, Ashwal S, Berg A, et al. Practice parameter: evaluating a first nonfebrile seizure in children: report of the quality standards subcommittee of the American Academy of Neurology, The Child Neurology Society, and The American Epilepsy Society. *Neurology*. 2000;55(5):616-623. PMID: 10980722.

Lascano AM, Lalive PH, Hardmeier M, Fuhr P, Seeck M. Clinical evoked potentials in neurology: a review of techniques and indications. *J Neurol Neurosurg Psychiatry.* 2017;88(8):688-696. doi:10.1136/jnnp-2016-314791.

45. Know methods to assess for autism spectrum disorders

Structured observation tools:

ADOS-2

- Different modules specific for use across the age span; examiner provides a series of presses to the patient
- Requires intensive training to administer and score
- 30-45 min to administer
- Used in conjunction with detailed history to determine if DSM-5 criteria are met

CARS2

- Clinician completes a 15-point scale based on history and observation

Other tools:

Social Responsiveness Scale (SRS)

- 65-item questionnaire used to measure autistic traits on a continuum

Autism Diagnostic Inventory-Revised (ADI-R)

- Length, semistructured parent interview
- Supports a knowledgeable clinician in applying diagnostic criteria of ASD

Social Communication Questionnaire (SCQ)

- Designed to elicit similar information to the ADI-R in an abbreviated questionnaire format

Identification, Evaluation, and Management of Children With Autism Spectrum Disorder. Susan L. Hyman, Susan E. Levy, Scott M. Myers, COUNCIL ON CHILDREN WITH DISABILITIES, SECTION ON DEVELOPMENTAL AND BEHAVIORAL PEDIATRICS
Pediatrics Jan 2020, 145 (1) e20193447; **DOI:** 10.1542/peds.2019-3447

46. Know methods to assess executive functions

Behavior Rating Inventory of Executive Functioning (BRIEF-2)

- Ages 5-18 years
- Parent and teacher forms, 10-15 min to complete
- Global executive composite + comprehensive coverage of subdomais of executive functioning

Rey Complex Figure Test

- Copy strategy: patient asked to copy complicated line drawing first by drawing freehand then from memory after a delay
- Ages 6-89 years
- Takes 45, including 30 min delayed interval
- Developmental scoring norms capture problem-solving strategy, which is a key correlate of executive function that is not often addressed
- Scoring system is complex and prone to error

Wisconsin Card Sorting Test

- Client's task is to sort stimulus cards according to different principles—by color, form, or number of shapes shown. Shifts in the sorting principle require the client to quickly alter his or her approach
- Ages 6.5 years to 89 years 11 mo,
- Takes 20-30 min
- Difficult to reliably score if not using computer administration, complex relationship between scales and executive function
- https://www.wpspublish.com/wcst-wisconsin-card-sorting-test

Kenworth L, Gutermuth Anthony L. Neuropsychological Assessment. *Children with Disabilities, 8th ed.* Batshaw ML, Roizen NJ, Pellegrino L, ed. 2019: Baltimore, Brookes Publishing Co.

B. Management
1. Communication skills

a. Understand how to inform parents of a diagnosis of a developmental disability, behavioral/mental health disorder, or life-threatening condition in their child
The SPIKES model is one commonly used for sharing bad news:

- Setting: privacy, limit interruptions/silence electronics, involves others (i.e. family members) if parent/child desires
- Perception: determine parent's understanding of the condition, correct misinformation and misunderstandings
- Invitation: determine how much info the parent desires, ask permission to give the results/share the diagnosis, if parent declines, offer to meet again in the future when ready
- Knowledge: Briefly summarize events leading up to this point, provide a warning statement, use nonmedical terms and avoid jargon
- Emotions: address emotions as they arise, use empathic statements to recognize emotion, validate responses to help the parent realize their feelings are important
- Strategy and summary: Summarize news to ensure understanding, plan for follow up, offer a means of contact if additional questions arise

Berkey FJ, Wiedemer JP, Vithalani ND. Delivering Bad or Life-Altering News. *American Family Physician.* 2018; 98(2): 99-104.

Additional strategies for breaking bad news skillfully:

- Do not disclose bad news over the telephone
- Use trained translators as needed
- Avoid telling a lone parent without his or her spouse and/or a preferred support person present (if possible)
- Recognize that parents are primarily responsible for their child

- Show caring, compassion, and a sense of connection to the patient and the family
- Pace the discussion to the parents' emotional state; do not overwhelm them with information
- Do not use jargon
- Elicit parents' ideas of the cause of the problem; ensure they do not blame themselves or others
- Name the illness and write it down for the parents
- Ask the parents to use their own words to explain what you have just told them to confirm effective transmission of information
- Address the implications for the child's future
- Acknowledge their emotions and be prepared for tears and a need for time; it is helpful to bring a social worker and/or chaplain to the meeting
- Be willing to show your own emotion; aloofness or detachment is offensive
- Give parents time to be alone to absorb the information, react, and formulate additional questions
- Be able to recommend relevant community-based resources
- Provide contacts with other willing families with a similarly affected child
- Provide a follow-up plan and make an appointment for the next conversation

Technical Report: Communicating with Children and Families: From Everyday Interactions to Skill in Conveying Distressing Information. Pediatrics. 2011;121(5):e1441-e1460. http://pediatrics.aappublications.org/content/121/5/e1441. Reaffirmed December 2016.

b. Understand how to effectively elicit parents' opinions and concerns for their child's development

The SHARE framework can be used:
Set the tone/**Support** the parent and child through a therapeutic alliance

- Create a safe space and open environment to share the details important for child development/family functioning
- Use developmental knowledge to guide how to build rapport with child and caregiver (i.e. indulge toddler's desire for autonomy and control by allowing them to listen to their heart)

Hear the parent's concerns about the child's behavior and development and the effects on family functioning through the use of guided questions

Address specific risk factors for child development/family functioning & **Allow** parents to reflect how cultural traditions contribute to their expectations of child behavior and development

Reflect with the parent on their experience of the child/ **Reframe** the child's behavior and development in terms of the child's developmental level/**Revisit** the therapeutic goals set

Empower the parent and child by formulating an action plan to address the concerns voiced in the visit

Shah PE, Ribaudo J. Chapter 5: Interviewing and Counseling Children and Families. American Academy of Pediatrics Developmental and Behavioral Pediatrics. 2nd ed. Voight RG, Macias MM, Myers SM, Tapia CD, editors. 2018. American Academy of Pediatrics. Itasca, IL.

2. Anticipatory guidance/health promotion

a. Know how to counsel families on the potential negative effects of physical punishment
Ineffectiveness of corporal punishment:

- Effects are transient – in one study within 10 minutes most (73%) of children had resumed the behavior for which they had been punished
- No benefit from physical punishment in the short-term (2016 meta-analysis)

Cycle of corporal punishment and aggressive child behavior:

- Fragile Families and Child Wellbeing study: children who were spanked more than 2x per month at age 3 were more aggressive at age 5; correlations found between spanking at age 5 and higher levels of externalizing behavior and lower receptive vocabulary scores at age 9. Subsequent study looked at same data and found increased spanking → increased externalizing behaviors → more spanking.

Corporal punishment and adverse outcomes. Corporal punishment:

- Of children <18mo increases likelihood of physical injury
- Repeated use can lead to aggressive behavior and altercations between parent and child and negatively affect parent-child relationship
- Associated with increased aggression preschool and school aged-children
- Makes it more likely that children will be defiant and aggressive in the future
- Increased risk of mental health disorders and cognition problems
- Spanking alone is associated with adverse outcomes (similar to those in children who experience physical abuse)

Guidelines for anticipatory guidance:

- Direct discussion advising against any form of corporal punishment
- Council that spanking is not an appropriate or effect disciplinary strategy: Spanking is ineffective in the long term, increases aggression and anger instead of teaching responsibility and self-control
- Combine this advice with teaching new strategies to replace corporal punishment

Sege RD, Siegel BS; Council on Child Abuse and Neglect; Committee on Psychosocial Aspects of Child and Family Health. Effective Discipline to Raise Healthy Children. *Pediatrics.* 2018;142(6):e20183112.

b. Know specific recommendations for parents to limit the influence of the media on their child's development and behavior

Young Children:

- Hands-on, unstructured and social play is important for brain development in young children
- Children <18 months: No screen media use except for video chatting
- Children 18-24 months: If desire to introduce digital media, choose high-quality programming apps and use them with children (this is how children learn best)
- Good resources for finding quality products: Common Sense Media, PBS Kids, Sesame Workshop. (Avoid fast-paced programs, apps with lots of distracting content and violent content)
- Children 2-5y: Limit media to 1 hour or less of high-quality programming. Shared use between parent and child promotes learning, interaction and limit setting.
- No screens during meals and for 1 hour before bedtime
- Avoid using media to calm child as this could lead to problems with limit setting and with ability of child to develop their own emotional regulation

Council on Communications and Media. Media and Young Minds. *Pediatrics.* 2017;138(5)e20162591.

School age children and adolescents:

- Encourage families to create and follow a Family Media Use Plan (available on healthchildren.org)
    - Enforce consistent limits on hours per day of media as well as types of media used
    - Children/adolescents should get min 1 hour/day active time and adequate sleep (8-12 hours, depending on age)
    - Children should not sleep with devices in their bedrooms; avoid screens for 1 hour before bedtime
- Parents should have ongoing conversations with children about online citizenship and safety, including treating others with respect online and offline, avoiding cyberbullying and sexting, being wary of online solicitation, and avoiding communications that can compromise personal privacy and safety.
- Parents should actively develop a network of trusted adults (eg, aunts, uncles, coaches, etc) who can engage with children through social media and to whom children can turn when they encounter challenges.

Council on Communications and Media. Media use in school-aged children and adolescents. *Pediatrics.* 2016;138(5)e20162592.

c. Know how to advise a family of a toddler on knowing when the child is ready to initiate toilet training

There is no “right age” but in general starting before age 24 months is not recommended. Signs of readiness for toilet training include:

- Child can imitate parent’s behavior
- Child begins to put things where they belong

- Child demonstrates independence by saying “no”
- Child expresses interest
- Child can walk and sit on the toilet
- Child can indicate when he is “going” and when he needs to “go”
- Child able to pull pants/underwear on and off

Toilet Training Guidelines: Parents – The Role of the Parent in Toilet Training. *Pediatrics.* 1999; 103(Supplement 3): 1362-1363.

d. Know how to advise parents to address resistance to toilet training

- Parents should use a child-oriented approach to toilet training and begin/continue the process at a pace determined by their child
- Setbacks during the process tend to occur or escalate if the child is pushed too hard or too fast of if a significant, stressful family event occurs
- Regression is normal and is not failure. Parents should be accepting of the setback and gently reinforce expected toileting behavior
- Address medical conditions that may influence success of toileting (such as constipation) prior to starting toilet training, if possible. Continue to treat to achieve soft, not-painful bowel movements during toilet training to avoid stool with-holding.

Stadtler A, Gorski PA, Brazelton TB. Toilet Training Methods, Clinical Interventions and Recommendations. *Pediatrics.* 1999;103(Supplement 3): 1359-1361.

Fleisher DR. Understanding Toilet Training Difficulties. *Pediatrics.* 2004;113(6): 1809-1810.

e. Know how to advise parents on sleep hygiene to avoid problematic sleep associations and bedtime resistance

To avoid problematic sleep associations:

- Have a consistent bedtime routine
- Put the child to bed drowsy but awake at bedtime to encourage self-soothing skills
- Avoid using methods to get the child to sleep that require parent involvement (rocking, nursing, having a parent present etc)

Mindell JA, Owens JA. A Clinical Guide to Pediatric Sleep: Diagnosis and Management of Sleep Problems, 3rd ed. Chapter 8: Night wakings in young children: sleep associations. 2015: Walters Kluwer, Philadelphia.

To avoid bedtime resistance:

- Establish a set bedtime that coincides with child’s natural sleep onset time
- Set and enforce a consistent bedtime and wake time
- Establish a regular and consistent routine
- Avoid evening direct light exposure and increase light exposure in the morning
- Establish an age-appropriate napping schedule

- Limit activities that promote wakefulness whil in bed (texting, watching TV, etc)
- Don't use the bed for punishment
- Avoid sleeping in environments other than the bedroom
- Stick to firm bedtime limits
- Ignore complaints or protests about bedtime

Mindell JA, Owens JA. A Clinical Guide to Pediatric Sleep: Diagnosis and Management of Sleep Problems, 3rd ed. Chapter 7 Bedtime problems in young children. 2015: Walters Kluwer, Philadelphia.

f. Know how to advise parents to approach discussions with their child regarding high-risk behaviors at different ages and developmental levels

For childhood:

- Discussion of topics such as bike/water safety, car safety/wearing seat belt, traffic safety, how to be safe with adults, what to do in emergencies
- Be a good role model
- Younger children need frequent repetition of rules/guidelines for safety

For teens: In general, take an open, honest and non-judgmental approach

- Help your child make a plan to resist peer pressures; be there for him/her when they need support or help
- Talk about issues when they arise in the media at school or with friends
- Support your child's own problem-solving and decision making skills by helping/prompting them to solve problems without parental interference
- Teens are more likely to make good choices if they feel connected to their families and parents set clear limits and boundaries
- Topics: sex, drugs/alcohol, driving, firearm safety, water safety, internet safety/cyber bullying

Hagan JF, Shaw JS, Duncan PM, eds. Bright Futures: Guidelines for Health Supervision of Infants, Children, and Adolescents. 4th ed. Elk Grove Village, IL: American Academy of Pediatrics; 2017

g. Know how to advise parents concerned about their child's sexual behaviors at different ages and developmental levels and in different settings

Characteristics of normal sexual behaviors in children:

- Frequently see in children younger than 6 yo and between children of similar age and development
- Self-stimulation, personal space intrusiveness, interest in language or images of a sexual nature, exhibitionism and mutual curiosity in peers genitals
- Transient and responsive to parental redirection and admonishment
- Diminishes if parents divert the child's behavior without emotional response

- New siblings or new caregiving situations with additional children may trigger sexual behaviors
- Children residing in homes in which nudity and/or sexuality are more open and acceptable may demonstrate more sexual behaviors

Characteristics of abnormal sexual behaviors in children:

- Occurring between children of different development and/or age (>4 years) requires assessment and possible reporting to child protective services
- Behavior problems that are coercive, persistently intrusive, injurious and frequent require assessment and treatment beyond parent redirection
- Exposure to sexual acts or materials, if persistent or disturbing to the child can result in problematic sexual behavior; careful assessment for abuse and supervisional neglect is appropriate

Other considerations:

- Life stresses, especially interparental violence, are strongly associated with sexual behavior problems in children
- Children exposed to physical abuse, sexual abuse, or neglect are more likely to have sexual behavior problems. Children with frequent, persistently intrusive or abusive sexual behaviors should be assessed for possible abuse or neglect

Kellog ND, Committee on Child Abuse and Neglect. Clinical Report – The Evaluation of Sexual Behaviors in Children. *Pediatrics.* 2009;124(3):992-998. Reaffirmed October 2018.

h. Know recommendations that could be given to parents to help them facilitate their child's adjustment to a new sibling at different ages and development levels

Toddlers: Age 1-2 years

- Look at picture books about a new baby
- When the baby arrives, do something special for your older child (special gift, extra time with dad/other special adult, take somewhere special)

Preschoolers: Ages 2-4 years

- Wait a while before telling your preschooler about the baby (like when buying nursery furniture/baby clothes).
- Picture books and sibling classes can be helpful
- Involve the preschooler in planning for the baby
- Time major changes in your child’s routine (finish toilet training, switching to a bed before baby arrives, if possible)
- Prepare your child for when you are in the hospital
- Set aside special time for your child
- Ask family and friends to spend a little time with your older child when they come to see the new baby
- Have your older child spend time with dad

School-aged Children: Ages 5 and up

- Tell the child what is happening in language she can understand
- Have your child help get things ready for the new baby
- If possible, have your older child come to the hospital soon after the baby is born
- When you bring the new baby home, make your older child feel that she has a role in caring for the baby
- Do not overlook your older child's needs and activities

Dixon SD. The Prenatal Visit: Making an Alliance with a Family. *Encounters with Children: Pediatric Behavior and Development.* St. Louis Mosby 1992.

i. Know how to advise parents on key features of quality child care programs

Indicators of High-Quality Child Care:

- Immunization of staff and children
- Infection control practices: hand washing w/ soap and running water after diapering, before handling food, children wash hands after toileting and before eating, routine cleaning of facility, toys, equipment
- Nutrition: safe food storage, sanitary food preparation, healthy meals and snacks
- Environment: clean air, smoke free, pest control
- Oral health: teeth brushing
- Physical activity: active play, limited screen time
- Staff ratios and supervision: small group sizes and high staff to child ratios
- Staff qualifications: consistent caregiving, college degrees in ECE, child development associate's credential, ongoing in-service training, low turnover rate, strong background checks
- Policies for children with special health care needs: medication administration, child care health consultation, care plans completed at the medical home
- Emergency procedures: cardiopulmonary resuscitation and first aid training, written policies, disaster planning procedures, staff +children familiar with procedures, up-to-date parent contact lists
- Injury prevention: safe play equipment, safe sleep practices, developmentally appropriate toys, toxins out of reach, child abuse prevention training, policies on discipline and restraint, sunscreen/insect repellant use policies, water play safety, facility safety (fire and CO detectors, etc)

Donoghue EA, Council on Early Childhood. Quality Early Education and Child Care from Birth to Kindergarten. *Pediatrics.* 2017;140(2):e20171488.

j. Understand the importance of sexuality education for teenagers with developmental disabilities and describe key features of effective education

- Have a right to same information as their peers but it may need to be modified:
    - Simplifying information
    - Teaching in a special needs rather than regular education setting
    - Using special teaching materials (anatomically correct dolls, role playing)

    - Frequent review and reincement of information
- Should include the following topics:
    - Body parts, pubertal changes, personal care and hygiene, medical exams, social skills, sexual expression, contraception strategies and the rights and responsibilities of sexual behavior
- IEPs should include provision of sexual education
- Pediatricians should promote open discussion of sexuality with children and their families to facilitate education

Murphy NA, Elias ER, and the Council on Children with Disabilities. Sexuality of Children and adolescents with developmental disabilities. *Pediatrics.* 2006;118(1):398-403.

k. Know the developmental and behavioral advantages of breast-feeding

- Consistent differences in neurodevelopmental outcomes found for breast fed vs formula fed infants, but many confounders (parental education, home environment, SES)
    - Evidence that adjusted outcomes of intelligence scores and teacher's ratings significantly greater in breast fed infants
    - Significant positive effects of human milk feeding on longer-term neurodevelopment in preterm infants

AAP Section on Breastfeeding. Breastfeeding and the Use of Human Milk. *Pediatrics.* 2012;129(3):e827-e841.

- Regarding behavior, evidence less clear. Some studies support a like between lack of behavior problems and breastfeeding, and others have not found this association.

Kramer MS, Fombonne E, Igumnov S, et al. Effects of prolonged and exclusive breastfeeding on child behavior and maternal adjustment: evidence from a large, randomized trial. *Pediatrics.* 2008; 121(3): e435-e440.

Heikkila K, Sacker A, Kelly Y, et al. Breast feeding and child behavior in the Millenium Cohort Study. *Arch Dis Child.* 2011;96(7):635-42.

l. Know how to advise parents on disciplinary strategies appropriate for children of different ages and developmental levels

Basic behavioral management principles:

- Identify antecedents and consequences
- Look for opportunities to modify antecedents and manipulate consequences
    - Examples of modifying antecendents: if trigger is restricted access, remove forbidden items from the environment, if trigger is desire for attention, play with child with frequent positive attention, if trigger is fear, avoid adverse stimulus and expose gradually

    - Examples of manipulating consequences: reward behaviors that you want to continue with attention; withdraw attention (ignore vs time out) for behaviors that are undesired
- The goal is to encourage desired behavior while decreasing undesired behavior
- Identify any exposure to inappropriate behavior and stop exposure if possible
- Consequences most effective if they occur immediately after the behavior, particularly for young children
- Positive parenting is recommended for all ages. Principles of positive parenting include:
    - "time in" - child directed play in which parents give a play by play about what the child is doing – good for modeling appropropriate behaviors and stimulating language, pros-ocial skills
    - Praise
    - Giving effective commands

Parenting for Different Ages

- Toddlers: Goal is to teach expectations through sustained effort. Meltdowns are common due to limited means of communication. Child should removed from the situation or redirected
- Early Childhood: Focus on appropriate modeling and reinforcing positive behaviors with praise. Selective use of ignoring, consequences, or time out (for high-intensity behaviors like aggression).
- Elemnatry school: Parents should focus on helping learn how to solve problems.
- At every level, positive reinforcement and consistency are the mainstays of parenting

Blum NJ, Pipan ME. Chapter 7: Basics of Child Behavior and Primary Care Management of Common Behavioral Problems. In: Voight RG, Macias MM, Myers SM, Tapia CD, editors. American Academy of Pediatrics Developmental and Behavioral Pediatrics. 2nd ed. 2018. American Academy of Pediatrics. Itasca, IL.

Bauer NS, Childers DO, Curtin M. Principles of positive parenting can be shared during pediatric visits. AAP News, December 13, 2016.
https://www.aappublications.org/news/2016/12/13/PositiveParenting121316

m. Know how to advise parents on habits and repetitive behaviors at different ages and developmental levels

Thumb sucking:

- Normal/adaptive in infants and toddlers, can delay onset of boredom and be soothing
- Harmless in infants and toddlers
- If persists beyond age 4-6 years, can be harmful: anterior open bite, alveolar bone growth, mucosal trauma, altered growth of facial bones, psychological sequalae in elementary school years (bullying, parental nagging)
- Stopping parental reaction to thumb sucking in older children may be successful if sucking has been reinforced by parental attention

- Treatment should be considered for children >4 years who suck their thumb in multiple settings; more successful in child is a willing partner

Banging:

- Rhythmic banging of hand/object occurs in typical development of infants
- Head banging: occasionally associated with onset of illness (i.e. headache, otitis media)
- Head banging when child is relaxed is not a sign of a developmental or emotional disorder and typically does not cause injuries to the brain (although can cause callus formation/abrasions)
- Typically developing children who head bang during tantrums typically don't injure themselves (or if they make a mistake and do, it typically doesn't happen a second time)
- Tx: reassure parent head banging alone is not a sign of a disorder, injury unlikely, consider padding the surface they are head banging against, ignore if occurring during tantrums
- In children with developmental disabilities, head banging can be severe and cause intracranial or retinal injury and require use of helmets or medication

Nail biting

- If related to stressors, focus on helping child cope with stressors
- Habit reversal behavioral treatment in older children

Blum NJ. Repetitive Behaviors and Tics. In Developmental-Behavioral Pediatrics, 4th ed. Ed by Carey WB, Crocker AC, Coleman WL, Elias ER, Feldman HM. 2009 Saunders Elsevier Philadelphia PA.

n. Know how to advise parents whose child is having temper tantrums or breath holding spell

Breath holding spells:

- Provoking event causes anger, fear, frustration or mild injury → crying → crying stops at end-expiration when child becomes apneic and cyanotic or pale → event may end or child may lose consciousness and fall to the ground
- 3 subtypes: cyanotic, pallid, and mixed (if pallid, at increased risk for vasovagal syncope later in life)
- Anemia may increase frequency of BHS, treatment with ferrous sulfate 5mg/kg/day decreases frequency
- Tx: reassure that spells don't harm child, education about the events, reassure that if child loses conscious, he begins breathing at that time; no need to restrict activities or avoid anger/fear as this can lead to difficulty setting limits and the child may lead to trigger BHS during tantrums to get to desired response from parent

Blum NJ. Repetitive Behaviors and Tics. In Developmental-Behavioral Pediatrics, 4th ed. Ed by Carey WB, Crocker AC, Coleman WL, Elias ER, Feldman HM. 2009 Saunders Elsevier Philadelphia PA.

o. Know how to advise parents of children engaging in aggressive behavior at different ages and developmental levels

- Aggressive behavior can become a problem during preschool years (developmental themes = emotions, sense of self, identification with powerful figures)
- Aggressive acts to get a desired object can begin around age 2
- By age 3, verbal aggression becomes more common
- By age 4-6, hostile aggression (aggressive act designed to hurt another person without any object being involved); girls particularly may exhibit relational aggression
- Preschool: aggressive challenges may become frequent
- By grade school: children have learned to restrict targets of aggressive challenges to kids with whom they have a chance of winning
- Some aggressive acts inevitable and provide a learning opportunity
- Unless conflicts are excessive, adults should stay out the of the middle because children need to learn to self regulate

Worrisome signs:

- Being kicked out of preschool for aggression (preschools usually comfortable with normal, mild aggression)
- Chronically angry or anxious-seeming children
- Chronic aggressors may = depression or anxiety
- Biting behavior beyond age 2.5

Dixon SD. Four Years: Clearer Sense of Self. *Encounters with Children: Pediatric Behavior and Development.* St. Louis Mosby 1992.

p. Know how to advise parents on strategies they can use to promote the development and academic success of their children

- Promote early literacy – read, read, read to your children. Encourage reading in older children
- Good school time routine – enough time for sleep, eating, homework, etc.
- Consistent communication with the school and involvement in child's schooling
- Be interested and positive in and about homework
- Do activities outside or school which promote learning (i.e. visits to museums, zoos, etc., talking about magazines/books/current events)

Klass P. Promoting early literacy. In Augustyn M, Zuckerman B, Caronna EB, eds. The Zuckerman-Parker Handbook of Developmental and Behavioral Pediatrics for Primary Care, 3rd Ed. 2011: Lippincott Williams and Wilkins. Philadelphia.

Academic Development Institute. A Guide for Parents: Helping your child succeed in school. 2006. Academic Development Institute: Lincoln, Il.

Wegner LW. School Achievement and Underachievement. In Developmental-Behavioral Pediatrics, 4th ed. Ed by Carey WB, Crocker AC, Coleman WL, Elias ER, Feldman HM. 2009 Saunders Elsevier Philadelphia PA.

q. Know how to advise parents on strategies to promote their child's self-esteem at different ages and developmental levels

- Help your child develop a sense of ownership and commitment by providing them with opportunities for assuming responsibilities (i.e. chores, participating in charity drives, etc)
- Provide opportunities for making choices and decisions and solving problems
- Offer encouragement and positive feedback and help child to feel special (i.e. special time with parent, brief note commending the child for an accomplishment, etc.)
- Help child establish self-discipline by providing realistic expectations, clear-cut rules, and logical consequences
- Teach children to deal with mistakes and failure (avoid over-reacting to mistakes, share what parent personally learned from mistakes made)

Brooks RB. Self Esteem and resilience. *The Zuckerman Parker Handbook of Developmental and Behavioral Pediatrics for Primary Care*, 3rd ed. Ed. Augustyn M, Zuckerman B, Caronna E, editors. 2011. Lippincott Williams & Wilkins. Philadelphia.

r. Know how to advise parents who are concerned about their infant's crying

- Typical infant periods of fussiness begins about 2 weeks of age, peaks between the first and second month and disappears by about 3-4 months
- Fussy behavior begins in late afternoon and resolves in early evening
- Usually resolves with holding, positioning, gently rocking, feeding
- Infant colic= periods longer or more intense (total more than 3 hours/day on more than 3 days/week)

Stein MG. Getting on Track: months one and two. *Encounters with Children: Pediatric Behavior and Development.* St. Louis Mosby 1992.

s. Know how to advise parents concerned about sibling rivalry at different ages and developmental levels

- Parents should spend separate time with each child, structured around that child’s needs and interests
- Consider family meetings to deal with controversies/conflicts
- Avoid comparisons and contrasts between siblings
- Encourage children to seek out different spheres of interest, activity and achievement

- Do not consistently sacrifice one child's interests, activities, free time to the achievements of a sibling
- Establish and follow codes of behavior and "fairness" so that arguments can be settled with rules that apply to all children (i.e. rotating some tasks by turns, respecting privacy of siblings' rooms)
- Be wary of making a fetish of absolute equal treatment (don't promise exactly the same attention, gifts, etc)
- Set appropriate limits for sibling behavior
- Younger siblings may need to be protected from physical harm
- Older siblings need to know that they (and their belongings) will also be properly protected
- Allow siblings some reasonable latitude to work out conflicts
- Reassure that sibling rivalry is not unusual and is not a reflection of poor parenting

Klass P. Chapter 11: Brothers and Sisters. In Developmental-Behavioral Pediatrics, 4th ed. Ed by Carey WB, Crocker AC, Coleman WL, Elias ER, Feldman HM. 2009 Saunders Elsevier Philadelphia PA.

t. Know how to advise parents about the use of transitional objects at different ages and developmental levels

- Transitional objects used effectively by children to shut out environmental stimuli and to calm themselves when upset
- Selection of a transitional object may occur as early as 7-8 months but usually occurs between 1-2 yo
- 2/3 of children in the US have a transitional object; other cultural groups have a lower rate
- Most children discard their transitional object around age 3-4
- Some children retain the attachment into the school years without evidence of psychopathology
- May be useful to have a duplicate for washing/replacement

Dixon SD. Eight to Nine Months: Exploration and Discovery. *Encounters with Children: Pediatric Behavior and Development.* St. Louis Mosby 1992.

u. Know how to advise parents about fears or anxieties in their child at different ages and developmental levels

Typical childhood fears:
0-7 months: change in stimulus level, loss of support, loud, sudden noises
8-18 months: separation, strangers, loud events, sudden movements toward, touching, physical restraints, large crowds, water, being bathed
2 yrs: loud sounds, dark colors, large objects, large moving things, hats/mittens, going down the drain or toilet, wind and rain, animals

2.5 years: movement, familiar objects movemed, moving objects, unexpected events linked
3 years: visual fears, masks, old people, people with scars/deformities, the dark, parents going out at night, animals, burglars
4 years: auditory fears, the dark, wild animals, mother's departure, imaginary creatures, recalled past events, aggressive actions, threats
5 years: decrease in fears, injury, falls, dogs
6 years: fearful age; supernatural events, hidden people, being left or lost, small bodily injuries, being left or lost, death of loved ones, fire, thunder
7 years: cellars, shadows, ideas suggested by TV/movies, being late for school, missing answers in school
8 -9 years: school failure, personal failure, ridicule by peers, disease, unanticipated events
10-11 years: wild animals, high places, criminals, older kids, loss of possessions, paretnal anger, remote possibilities of catastrophe, school failure, pollution
12-17 years: physical changes in body, isolation, sexual fears, loss of face, world events

Dealing with fears:
Overall goals: support for adaptive coping and protection from unreasonable fears

- Stranger anxiety: new adults should approach slowly and be prepared to back off
- Preschooler's imaginary fears: acknowledge fearful feeling without adding credence to imaginary creature; reassurance of support and safety (attempting to argue child out of fantasy is futile)
- School age children: information typically reduces fears; reassurance of normalcy of fears is helpful; parents can help by sharing their own fears and how they coped

Dixon SD. Three years: emergence of magic. *Encounters with Children: Pediatric Behavior and Development.* St. Louis Mosby 1992.

v. Know how to advise parents concerned about their child's feeding behaviors at different ages and developmental levels

Newborn: Feeding every 1.5-2 hours for first few days; typically stabilizes when mom's milk comes in to around every 2-3 hours

3-4 months: drive for social interactions may lead to distraction during feeding. Balance social needs w/ feeding needs. At least a few feeds a day should be in a quiet room without distractions.

Around 8 months: self feeding should start as it is an important opportunity for child to assert some independence and explore the feel and texture of food. Self-feeding is a great opportunity for improvement of fine motor skills.

15-18 months: feeding is an important time for expression of control and independence

- Appetite often erratic at this age

- Toddler will choose adequate nutrients over time if nutritious foods available
- Avoid forced feeding or battles over food
- Messy feeding expected and appropriate
- Bulk of diet should be foods the family eats
- Throwing food/screaming mean child is done eating

Encounters with Children: Pediatric Behavior and Development. Dixon SD, Stein MT, editors. St. Louis Mosby 1992.

w. Know how to advise parents on promoting toilet training for a preschool-age child

- Narrate the toileting process while child is watching siblings or parent on the toilet
- Be positive toward toileting behavior
- Praise the child for undressing self; allow child to undress self
- Keep potty seat in a regular place in the bathroom and let the child know they can sit on it and what it is used for
- Use training pants during the day
- Praise the child for knowing what the potty seat is for and encourage sitting on the seat
- Encourage use of the potty after a meal or at a time the child usually has a BM
- Expect some regressions without explanation
- Whole process typically takes months

Stein MT. 15 to 18 months: Asserting Independence and Pushing the Limits. *Encounters with Children: Pediatric Behavior and Development.* St. Louis Mosby 1992.

x. Know how to advise parents on promoting toilet training for a developmentally delayed school-age child

Often requires:

- More support and training from experts
- More time, instruction and persistence in comparison to typically developing peers

May need to consider starting the process prior to typical prerequisite skills cited (interest, knowledge of full bladder, etc)

Similar process and intervention options exist for developmentally delayed child. Consider more use of tools such as timed voiding, visual schedules, frequent reminders, “first…then” boards, picture cards for communication.

For children in school and not yet toilet trained, include in the IEP.

Cocchiola M.A., Redpath C.C. (2017) Special Populations: Toilet Training Children with Disabilities. In: Matson J. (eds) Clinical Guide to Toilet Training Children. Autism and Child Psychopathology Series. Springer, Cham. https://doi.org/10.1007/978-3-319-62725-0_13

y. Identify strategies to guide families in building coping and resilience and avoiding overprotection in a child with a developmental disability

- Promote self-understanding and acceptance in the child
    - Help the child set realistic goals that are possible to achieve
    - Help child seek help when needed and find opportunities that incorporate their strengths
- Advocate for the child (school, services, etc) and provide emotional support
- Acknowledge and accept the child's strengths and limitations
- Maintain appropriately high expectations for future education and independence
- Support the child's extracurricular activities
- Teach persistence at home
- Teach social skills/social awareness
- Help build a strong support system for the child including more than just parents
- Acknowledge disabilities outside the child's control while supporting the development of a growth mindset, encouraging the child to put effort into developing skills in domains in which effort and practice will improve skills

Ofiesh N., Mather N. (2013) Resilience and the Child with Learning Disabilities. In: Goldstein S., Brooks R. (eds) Handbook of Resilience in Children. Springer, Boston, MA. https://doi.org/10.1007/978-1-4614-3661-4_19

Dweck CS, Master A. Self-Concept. *Developmental-Behavioral Pediatrics.* 4th edition. Carey WB, Crocker AC, Coleman WL, Elias ER, Feldman HM eds. 2009: Saunders-Elsevier. Philadelphia, PA. p427-435.

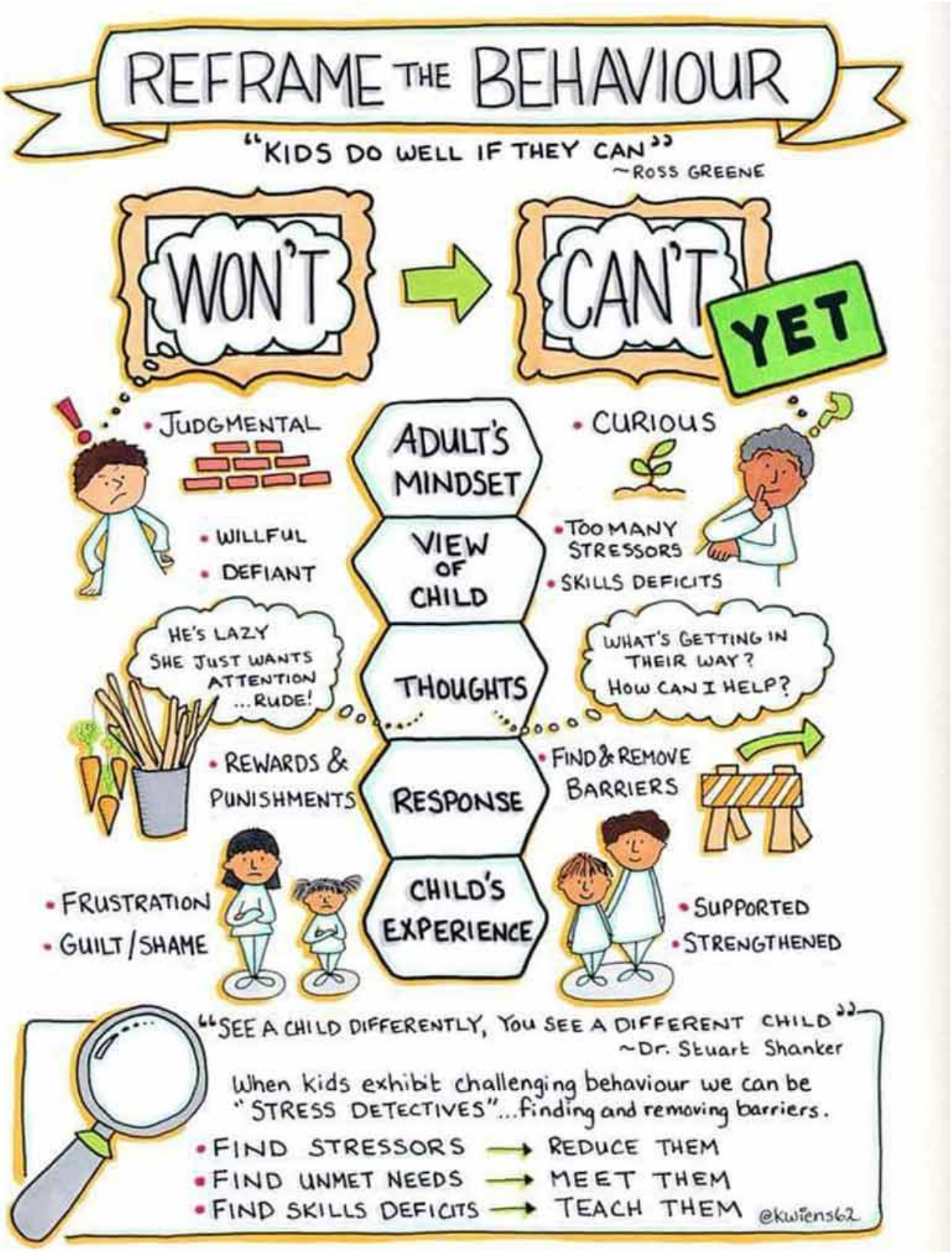
3. Counseling

a. Understand how reframing can be used within counseling as a therapeutic maneuver.

- **Reframing** is a psychotherapeutic technique used in therapy to help create a different way of looking at a situation, person, or relationship by changing its meaning.
- Also referred to as cognitive **reframing**, it's a strategy therapists often used to help patients look at situations from a slightly different perspective
- Technique used frequently in family therapy and individual cognitive behavioral therapy for children and teens
- Reframing involves three stages. (using a clinical example of an adolescent boy and his father being at odds with each other)
    - First, the therapist validates the perspectives of the family members.
        - For example, the therapist might indicate to the boy, "I understand why you think your dad doesn't like you. He often criticizes the

way you dress, acts disgusted by the music you listen to, and thinks your friends are losers." To the father, the therapist might say, "You've tried to give him a proper sense of moral values and respect for authority. But the way he dresses and behaves drives you crazy. In fact, sometimes you think that he is intentionally trying to push your buttons."

- During the second stage, the therapist provides an alternative perspective that puts the behavior in a benign or even positive light.
    - For example, the therapist might say to the boy, "I wonder if your dad's nagging, negative comments and restrictions are a sign of his love for you. If he didn't care for you so much, he might just let you dress and act however you wanted." To the father, the therapist might say, "Your son's lifestyle choices might just be his way of gaining independence. I suggest we focus on the big issues, like not getting rearrested, and let him have his little rebellions."
- In the final stage of reframing, the MST therapist checks with family members to assess their agreement with the reframe.
    - The end result should be a mutually agreed-upon alternative for certain types of negative interactions. With this alternative explanation in place, family members can move forward on interventions that require all participants to collaborate.


REFRAME THE BEHAVIOUR
"KIDS DO WELL IF THEY CAN"
~ROSS GREENE
WON'T
CAN'T
YET
ADULT'S MINDSET
• JUDGMENTAL
• WILLFUL
• DEFIANT
• CURIOUS
VIEW OF CHILD
• TOO MANY STRESSORS
• SKILLS DEFICITS
HE'S LAZY
SHE JUST WANTS ATTENTION
...RUDE!
WHAT'S GETTING IN THEIR WAY?
HOW CAN I HELP?
THOUGHTS
• REWARDS & PUNISHMENTS
RESPONSE
• FIND & REMOVE BARRIERS
• FRUSTRATION
• GUILT/SHAME
CHILD'S EXPERIENCE
• SUPPORTED
• STRENGTHENED
"SEE A CHILD DIFFERENTLY, YOU SEE A DIFFERENT CHILD"
~Dr. Stuart Shanker
When kids exhibit challenging behaviour we can be "STRESS DETECTIVES"...finding and removing barriers.
• FIND STRESSORS → REDUCE THEM
• FIND UNMET NEEDS → MEET THEM
• FIND SKILLS DEFICITS → TEACH THEM
@kwiens62

b. Understand the important role providing information about a condition can play in therapy.

- Providing information about a condition in therapy can help with building an effective physician-patient communication and ultimately good therapeutic trust and a strong therapeutic alliance

c. Understand how strategic family therapy can be used to challenge the family with particular tasks to improve existing maladaptive patterns of problem solving or communication

- Brief Strategic Family Therapy (BSFT) is specifically designed to provide families with the tools to overcome adolescent behavior problems and the family dysfunction that often accompanies these adolescent problems through: 1) focused interventions to correct maladaptive patterns of family interaction, and 2) skills building strategies to strengthen families.
- Families of youth with behavior problems such as drug and alcohol use, delinquency, affiliation with antisocial peers, and unsafe sexual activity tend to interact in ways that permit or promote these problems
    - The goal of the BSFT approach, therefore, is to change the patterns of family interactions that allow or encourage problematic adolescent behavior.
    - By working with families, the BSFT intervention not only decreases youth problems, but also creates better functioning families
    - Because therapists bring about changes in family patterns of interactions, these changes in family functioning are more likely to last after treatment has ended because multiple family members have changed the way they behave with each other.

d. Understand the key constructs of family systems theory

- Murray Bowen was an American psychiatrist and a professor in psychiatry at Georgetown University.
    - Bowen was among the pioneers of family therapy and a noted founder of systemic therapy. Beginning in the 1950s he developed a systems theory of the family.
    - In the early days of family systems development, most psychiatrists utilized Freudian psychoanalytic theory in treatment, and the family was not involved other than to provide information.
    - Only the “designated Patient” was seen by the Physician.
    - The idea of seeing family members also was viewed as a radical departure from common practice
- Bowen family systems theory is a theory of human behavior that views the family as an emotional unit and uses systems thinking to describe the complex interactions in the unit. It is the nature of a family that its members are intensely connected emotionally.
    - There are two main variables in the Bowen Theory:
      1) Degree of anxiety.
      2) Degree of integration of self.
    - According to Bowen, all organisms are reasonably adaptable to acute anxiety.

        - When anxiety is chronic, the organism develops tension, either within the organism or in the relationship system.
        - The tension produced by enduring anxiety precipitates symptoms, dysfunction or sickness.
        - The symptoms are manifested by physical illness, by emotional dysfunction, social illness characterized by impulsiveness, withdrawal or social misbehavior in a spouse; or by emotional or behavioral dysfunction in a child.

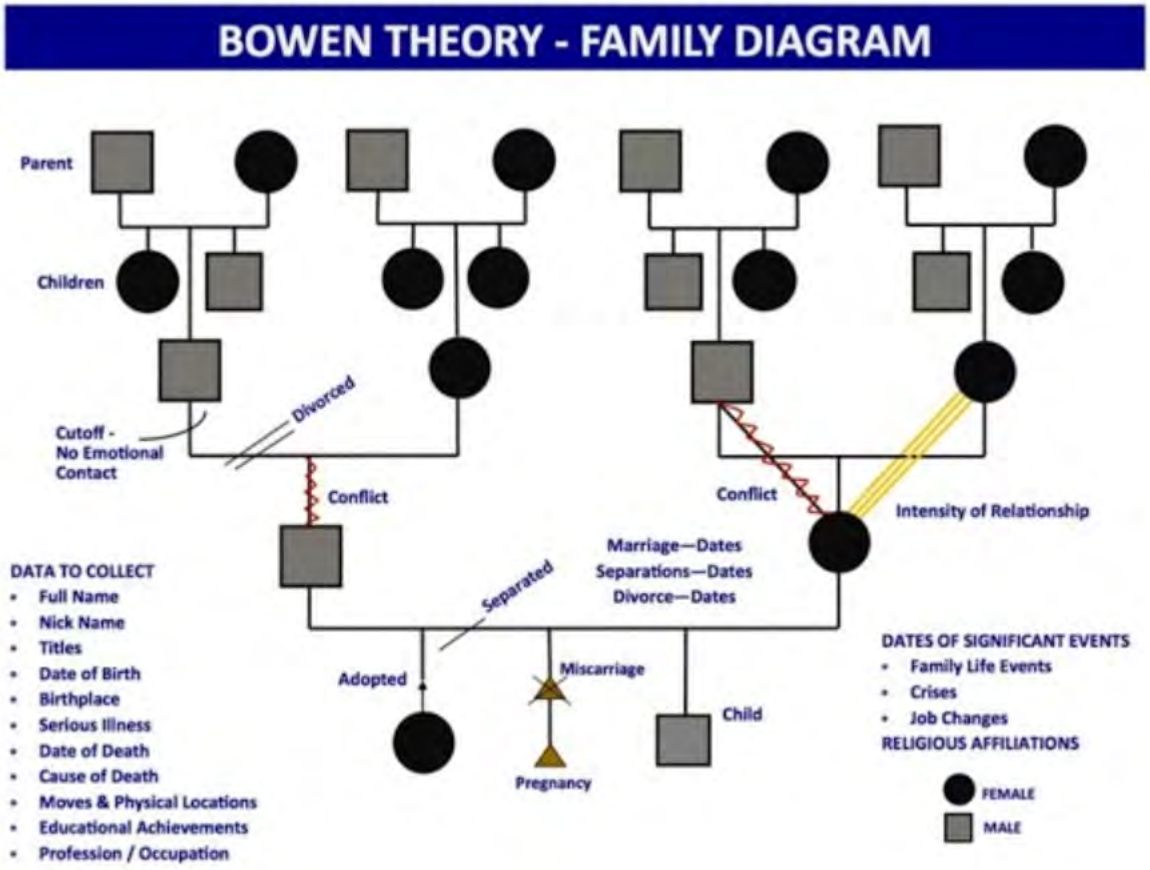
- Eight major theoretical concepts form the foundation of the Bowenian approach. These concepts are interconnected, and a thorough understanding of each may be necessary in order to understand the others.
- These theoretical constructions include, in no particular order:
    - Differentiation of self, the core concept of Bowen's approach, refers to the manner in which a person is able to separate thoughts and feelings, respond to anxiety, and cope with the variables of life while pursuing personal goals.
        - It defines people according to the degree of "fusion" between the emotional and intellectual functioning of the individual.
        - An individual with a high level of differentiation may be better able to maintain individuality while still maintaining emotional contact with the group.
        - A person with a low level of differentiation may experience emotional fusion, feeling what the group feels, due to insufficient interpersonal boundaries between members of the family.
        - Highly differentiated people may be more likely to achieve contentment through their own efforts, while those with a less-developed self may seek validation from other people.
    - An emotional triangle represents the smallest stable network of human relationship systems (larger relationship systems can be perceived as a network of interlocking triangles).
        - A two-person dyad may exist for a time but may become unstable as anxiety is introduced.
        - A three-person system, however, may provide more resources toward managing and reducing overall anxiety within the group.
        - Despite the potential for increased stability, many triangles establish their own rules and exist with two sides in harmony and one side in conflict—a situation which may lead to difficulty. It is common for children to become triangulated within their parents' relationship.
    - The family projection process, or the transmission of a parent's anxiety, relationship difficulties, and emotional concerns to the child within the emotional triangle, may contribute to the development of emotional issues and other concerns in the child.
        - The parent(s) may first focus anxiety or worry onto the child and, when the child reacts to this by experiencing worry or anxiety in

turn, may either try to "fix" these concerns or seek professional help.
        - However, this may often have further negative impact as the child begins to be further affected by the concern and may become dependent on the parent to "fix" it.
        - What typically leads to the most improvement in the child is management, on the part of the parent(s), of their own concerns.
- The multigenerational transmission process, according to Bowen, depicts the way that individuals seek out partners with a similar level of differentiation, potentially leading certain behaviors and conditions to be passed on through generations.
    - A couple where each partner has a low level of differentiation may have children who have even lower levels of differentiation.
    - These children may eventually have children with even lower levels of differentiation.
    - When individuals increase their levels of differentiation, according to Bowen, they may be able to break this pattern, achieve relief from their symptoms of low differentiation, and prevent symptoms from returning or occurring in other family members.
- An emotional cutoff describes a situation where a person decides to best manage emotional difficulties or other concerns within the family system by emotionally distancing themselves from other members of the family.
    - Cutting emotional connections may serve as an attempt to reduce tension and stress in the relationship and handle unresolved interpersonal issues, but the end result is often an increase in anxiety and tension, although the relationship may be less fraught with readily apparent conflict.
    - Bowen believed emotional cutoff would lead people to place more importance on new relationships, which would add stress to those relationships, in turn.
- Sibling position describes the tendency of the oldest, middle, and youngest children to assume specific roles within the family due to differences in expectation, parental discipline, and other factors.
    - For example, older children may be expected to act as miniature adults within the family setting. These roles may be influenced by the sibling position of parents and relatives.
- The societal emotional process illustrates how principles affecting the emotional system of the family also affect the emotional system of society.
    - Individuals in society may experience greater anxiety and instability during periods of regression, and parallels can be noted between societal and familial emotional function.
    - Factors such as overpopulation, the availability of natural resources, the health of the economy, and so on can influence these regressive periods.
- The nuclear family emotional process reflects Bowen's belief that the nuclear family tends to experience issues in four main areas: intimate

partner conflict, problematic behaviors or concerns in one partner, emotional distance, and impaired functionality in children.

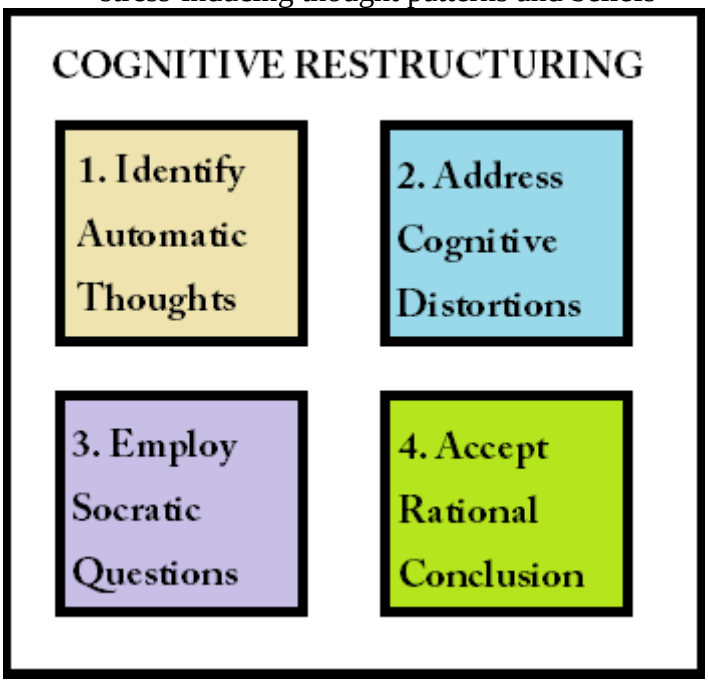
- Anxiety may lead to fights, arguments, criticism, under- or over-performance of responsibilities, and/or distancing behavior.
- Though a person's particular belief system and attitude toward relationships may impact the development of issues according to relationship patterns, Bowen held them to be primarily a result of the family emotional system.

- Bowen Family Diagram: the family diagram, developed by Murray Bowen, symbolizes a living organism, the multigenerational family emotional system.
  - More than any other symbol, the diagram announces the necessity to shift paradigms, to move beyond an individual cause-and-effect model to a multiperson systems model in understanding human behavior. The diagram represents much more than genealogy; it represents the profound emotional connections between the generations. People are born and die, but a family's past lives in the present
  - Diagrams are read chronologically from left to right: the oldest child in a family appears furthest to the left. Males are represented by squares, females by circles. When information about people's lives is collected, added to the basic diagram, and thought about, one's own life takes on a new understanding and meaning.

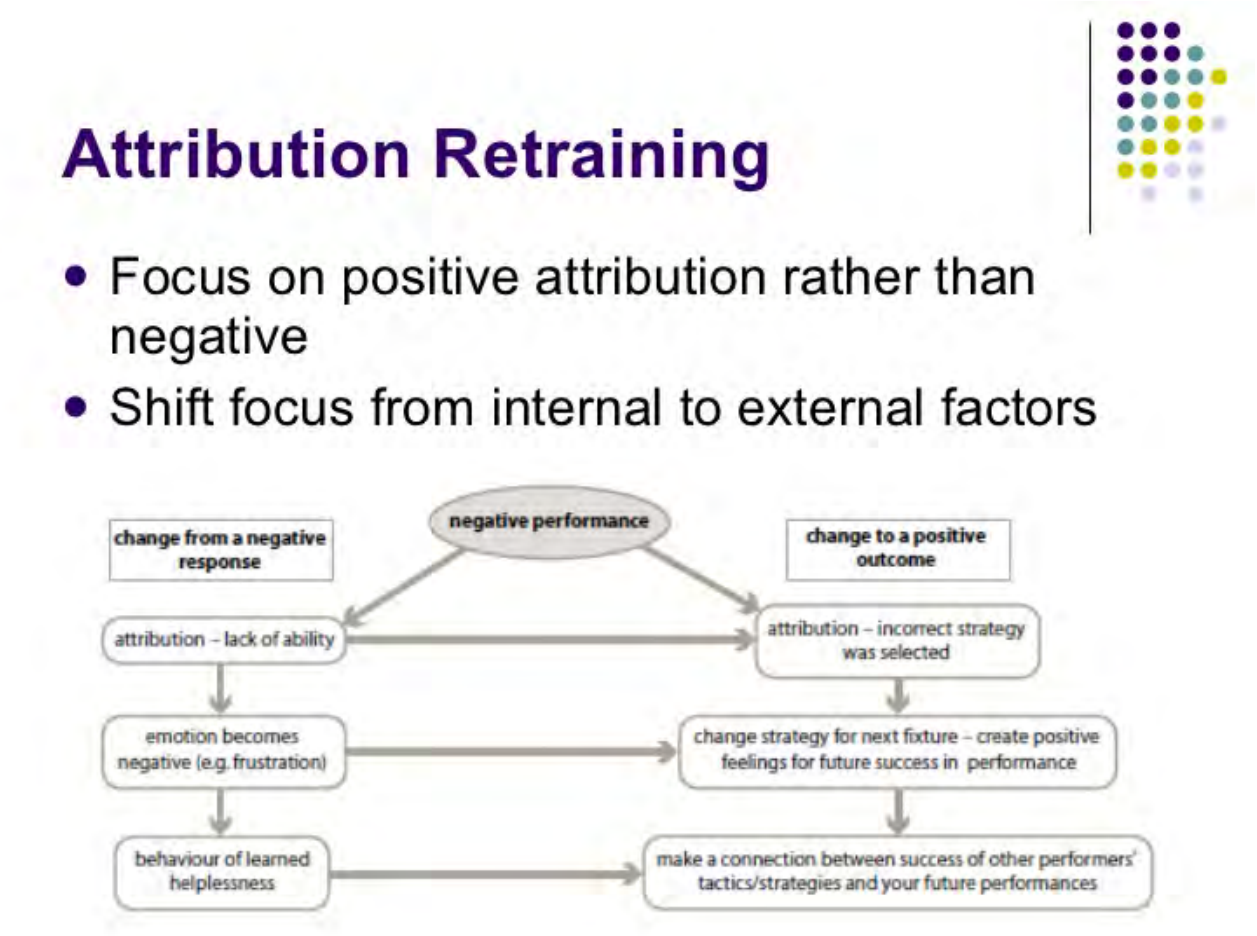


e. Understand how cognitive restructuring and attribution retraining can be employed in cognitive-behavioral counseling to address disorders of mood or emotion.

- **Cognitive restructuring,** also known as **cognitive** reframing, is a technique drawn from cognitive therapy that can help people identify, challenge and alter stress-inducing thought patterns and beliefs



- Attribution retraining (AR) is one of the therapeutic approaches within the larger group of cognitive behavior therapy. It is a therapy that treats clients' maladjusted emotions and behaviors by changing their explanations for problems and symptoms



f. Know the benefits of group therapy for enhancing social skills and behaviors

- Group therapy can be extremely beneficial for many neurodevelopmental and psychiatric diagnosis in pediatric population
- Development of social skills and behaviors is important adaptive skill for building self-reliance and independence
- Different forms of group therapy for pediatric populations:
    - **Dialectical behavior therapy (DBT)**: This emphasizes a combination of individual psychotherapy and group skills training classes to help kids learn and use new strategies to improve their emotional functioning. Skills learned include mindfulness, emotion regulation, distress tolerance, and interpersonal effectiveness. Dialectical behavior therapy is an offshoot of the more common practice of cognitive behavioral therapy (CBT). DBT was initially developed to help individuals with borderline personality disorder
    - **Process groups**: These groups help kids work through psychosocial stressors in a safe and supportive environment. Grief groups, social groups to work through bullying, eating disorders groups, play therapy groups, and empowerment groups are examples of process groups. Process group therapy is a powerful tool in helping kids learn how to trust others, how to share experiences that they might otherwise keep bottled up, and how to provide and accept supportive feedback to and from peers.

- **Social skills groups**: These groups include skill-building exercises and practice with age-appropriate peers. Social skill groups are typically aimed towards developing social skills in younger children, approximately ages two to eight. Social skill group therapy sessions can involve role-playing practices to help kids better understand how to start conversations as well as appropriate body language.
- **Cognitive behavioral therapy (CBT) groups**: These groups help kids identify inaccurate thinking that reinforces negative behaviors and are generally focused on a specific issue. CBT groups are particularly effective with anxiety disorders.

g. Appreciate conditions for which group therapy is most appropriate

- Not much research has been done regarding which conditions that group therapy is most appropriate for in children and adolescents
- Most evidence available to support its use in anxiety disorders- especially social anxiety disorder

h. Understand the components of and indications for cognitive behavior therapy

- Anxiety disorders
  - Cognitive-behavioral therapy (CBT) focuses on the interplay between cognitions, behaviors and emotions, helping patients to recognize and modify maladaptive anxiety-provoking thoughts and to change patterns of avoidance. The content of CBT programs can vary but typically includes psychoeducation and exposure to anxiety producing stimuli and situations, couched within an active and collaborative patient-therapist relationship, and reinforced by the use of patient-centered homework assignments.
  - Exposure treatment is central to all efficacious CBT for pediatric anxiety disorders; this involves the child gradually but repeatedly experiencing the feared situation with the intent of reducing the associated anxiety, or learning to tolerate and manage normal, expected levels of anxiety.
  - There are multiple semi-structured, manualized CBT programs for pediatric anxiety disorders that have been found to be efficacious when delivered by a trained clinician
  - CBT is indicated for all of the childhood anxiety disorders, including separation anxiety disorder, generalized anxiety disorder, social anxiety disorder, and specific phobia in adolescents and children age seven and older
  - Children younger than seven typically do not possess the developmental abilities needed to understand and apply cognitive-behavioral strategies to their symptoms, but CBT has been adapted for delivery to parents of children with anxiety disorders, or to parents and children together
- PTSD
  - Trauma-focused cognitive-behavioral therapy (TF-CBT) for children and adolescents with PTSD is a parallel child and parent (or primary caregiver) treatment model that incorporates cognitive-behavioral, developmental, neurobiological, attachment, family, and empowerment principles.

- Goals include helping children and parents gain resiliency and coping skills, master learned and over-generalized avoidance of feared trauma memories, make more adaptive meaning of traumatic experiences, and resume optimal developmental trajectories.
- Children with trauma-related symptoms can benefit from trauma-focused psychotherapy whether or not they meet diagnostic criteria for PTSD.
- Phases and components — TF-CBT is comprised of multiple components and is provided in three phases. Within each treatment session, therapy is provided in individual, parallel sessions to the child and the parent (or caregiver, hereafter "parent"), and in conjoint child-parent sessions as described below.
  - **Stabilization phase** – Includes psychoeducation, parenting skills, relaxation skills, affect modulation skills, and cognitive processing skills.
  - **Trauma narration and processing phase** – Includes trauma narration and processing.
  - **Integration and consolidation phase –** Includes in vivo mastery, conjoint child-parent sessions, and enhancing safety.

- OCD
  - The basic approach and components of effective CBT programs for children with OCD are similar [7]. The framework and administration described here are from a widely-used and well-studied program [8]. CBT can be customized to the developmental limitations of the child and has been used to good result used in children as young as five years of age. Parents need to be critically involved when CBT is administered to pediatric OCD patients.
  - The program is delivered in five phases of treatment:
    - Psychoeducation
    - Cognitive training
    - Mapping OCD
    - Graded exposure and response prevention (ERP)
    - Relapse prevention and generalization training
- Unipolar Depression
  - Both the cognitive-focused and the behavior-focused approaches have empirical support for efficacy in treatment of depressed adolescents
  - Regardless of emphasis, the efficacious forms of CBT for depression in adolescents share the following methods or treatment components
    - Increasing involvement in pleasurable, mood-enhancing activities
    - Increasing quantity and improving quality of social interactions
    - Improving conflict resolution and social problem-solving skills
    - Reducing physiological tension
    - Modifying or restructuring depressogenic thoughts

- Schizophrenia

- CBT for children/adolescents with schizophrenia (and in some cases family members) consists of:
    - Cognitive approaches to address maladaptive and delusional beliefs
    - Behavioral approaches, including social and vocational skills training
- Treatment goals of CBT include reducing the intensity of delusions and hallucinations (and related distress), improving social functioning, and promoting the active engagement of children and families in treatment.
- Clinical trials have found mixed evidence for the efficacy of CBT in children/adolescents with schizophrenia

- Substance Use Disorder
    - CBT for SUD in adolescents focuses on:
        - Enhancing adolescent coping, problem solving, and decision-making skills related to substance use;
        - Teaching skills to help adolescents cope with cravings and overcome temptations to use substances (eg, drug refusal skills, avoiding high-risk situations);
        - Improving interpersonal relationships (eg, communication, anger management, and mood regulation skills);
        - Reducing risky behaviors associated with substance use (eg, HIV/sexual risk behaviors, riding with or driving while intoxicated).
    - CBT for SUD may be delivered individually to adolescents or in a group format

4. Behavioral interventions

a. Understand conditions in which classical conditioning is utilized for treatment

The goal of behavioral intervention utilizing classical conditioning (CC) is teaching new skills using reinforcement, or eliminating certain behaviors using extinction. For example, when a parent says “bottle” every time they present a bottle to their toddler, the child will begin to understand the word “bottle” represents the object. CC occurs when two different events occur in such a way that one of the events begins to signal or elicit the other event (which has been used to explain the development of certain behaviors, i.e. connect *frightening experiences such as a nightmare with darkness, sharp pain with bowel movements, bullying/ridicule through interaction with peers at school, failing grade with test-taking*, etc.).

Reinforcement occurs when the relationship between the conditioned stimulus (eg, *darkness*) and the unconditioned response (eg, *fear*) becomes strengthened through repeated association. Conversely, extinction is said to occur when the relationship is weakened such that the stimulus no longer elicits the response (eg, *repeatedly going to school without ridicule, forming friends or taking test/submitting assignment with passing score*, etc.), in which case the conditioned response is likely to disappear or become extinguished. Also applicable for children with ASD, ABA focuses on the principles that

explain how learning takes place. Positive reinforcement is one such principle. When a behavior is followed by some sort of reward (verbal phrase, piece of candy, etc.), the behavior is more likely to be repeated.

The mode of CC relates to an automatic stimulus that is reinforced after a certain event/action. For example, CC is derived from experiments with Pavlov "Pavlov's Dogs" who would automatically salivate every time they heard a bell ring, as they had been repeatedly provided with a treat after a bell was rung. So too with children, who will likewise form associations using reinforcement, or eliminate these associations using extinction.

b. Differentiate among schedules of reinforcement most appropriate for a behavior management plan (eg, fixed and variable; ratio and interval)

Interval Schedule
Interval schedules involve reinforcing a behavior after a period of time has passed.

Fixed Schedule
In a fixed schedule the number of responses or amount of time between reinforcements is set and unchanging. The schedule is predictable.

- **Fixed Interval**

  In operant conditioning, a fixed interval schedule is when reinforcement is given to a desired response after specific (predictable) amount of time has passed. An example of a fixed-interval schedule would be a teacher giving students a weekly quiz every Monday. Over the weekend, there is suddenly a flurry of studying for the quiz. On Monday, the students take the quiz and are reinforced for studying (positive reinforcement: receive a good grade; negative reinforcement: do not fail the quiz). For the next few days, they are likely to relax after finishing the stressful experience until the next quiz date draws too near for them to ignore.

Variable Schedule
In a variable schedule the number of responses or amount of time between reinforcements change randomly. The schedule is unpredictable.

- **Variable Interval**

  In operant conditioning, a variable interval schedule is when the reinforcement is provided after a random (unpredictable) amount of time has passes and following a specific behavior being performed.

Ratio Schedule
In a ratio schedule reinforcement occurs after a certain number of responses have been emitted.

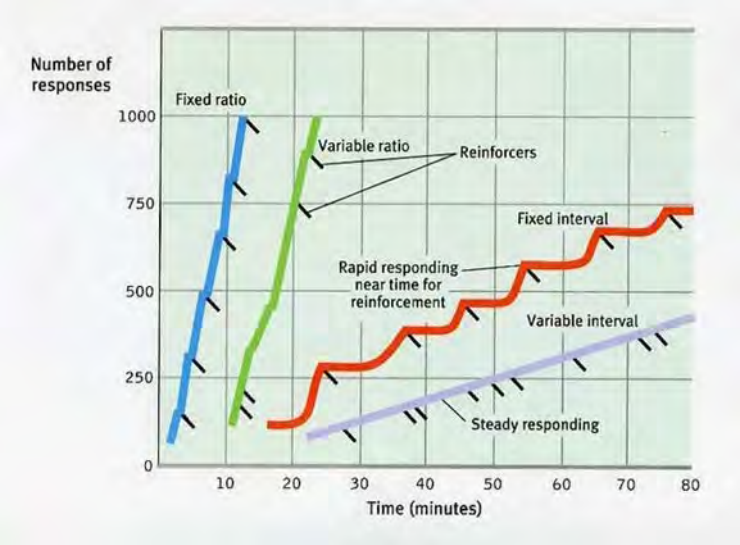
- **Fixed-Ratio**

  In operant conditioning, a fixed-ratio schedule reinforces behavior after a specified number of correct responses. A variable ratio schedule is a schedule of reinforcement where a behavior is reinforced after a random number of responses.

An example of a fixed-ratio schedule would be a child being given a candy for every 3-10 pages of a book they read. For example, they are given a candy after reading 5 pages, then 3 pages, then 7 pages, then 8 pages, etc. The unpredictable reinforcement motivates them to keep reading, even if they are not immediately reinforced after reading one page.

- **Variable Ratio**

A variable ratio schedule is a schedule of reinforcement where a behavior is reinforced after a random number of responses. This kind of schedule results in high, steady rates of responding. Subjects are persistent in responding because of the hope that the next response might be one needed to receive reinforcement. This schedule is utilized in lottery games.



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c. Understand that reinforcing incompatible behavior is an alternative to planned ignoring of the target behavior

One way to address a problem behavior is to substitute and then reinforce another behavior.

Differential reinforcement of incompatible behavior (DRI) is a procedure in which the teacher would identify a behavior that's incompatible with, or cannot occur at the same time as, the problem behavior. The focus is on replacing negative behaviors with positive behaviors. For example, a student cannot at the same time:

- Sit in a desk *and* wander around the classroom
- Be verbally aggressive toward a peer *and* give that peer a compliment

In these instances, sitting at the desk and complimenting a peer are examples of positive, incompatible behaviors that can replace the inappropriate behaviors of wandering around the classroom or being verbally aggressive.

Steps for Using DRI

1. Identify a problem behavior that is occurring at a fairly frequent rate and collect baseline data if necessary.
2. Brainstorm alternative (incompatible) behaviors that would keep the student from engaging in the problem behavior.
3. Determine and deliver reinforcement when the student is engaging in the alternate/ incompatible behavior.
4. Deliver appropriate consequence if the student engages in negative behavior.

d. Know the various forms of punishment, including natural consequences, logical consequences, behavioral penalties, physical punishment, and time out

Natural consequences: As long as your child isn't doing something dangerous and gets plenty of attention for good behavior, ignoring bad behavior can be an effective way of stopping it. Ignoring bad behavior can also teach children natural consequences of their actions. For example, if your child keeps dropping her cookies on purpose, she will soon have no more cookies left to eat. If she throws and breaks her toy, she will not be able to play with it. It will not be long before she learns not to drop her cookies and to play carefully with her toys.

Logical consequences: are logically linked to the misbehavior. Most logical consequences are simply the temporary removal of a possession or privilege. Examples are: taking away toys or crayons that are not handled properly, not replacing a lost toy, not repairing a broken toy, sending your child to school partially dressed if the child won't dress himself, having your child clean up milk he has spilled, cleaning the floor your child has tracked mud on, and turning off the TV if children are fighting about it.

Behavioral penalties: Punishments applied as a consequence to certain undesirable behaviors (eg, if she does not pick up her toys, you will put them away for the rest of the day, no screen time, no hanging out with friends, loss of privileges, etc.)

Physical punishment: a.k.a. corporal punishment. The Global Initiative to End all Corporal Punishment of Children provided a comprehensive definition of spanking and corporal punishment: “The definition of corporal or physical punishment adopted by the Committee on the Rights of the Child in its *General Comment No. 8* (2006) has the key reference point, ‘any punishment in which physical force issued and intended to cause some degree of pain or discomfort, however light.’ According to the committee, this mostly involves hitting (“smacking,” “slapping,” or “spanking”) children with the hand or with an implement (a whip, stick, belt, shoe, wooden spoon, or similar), but it can also involve, for example, kicking, shaking, or throwing children; scratching, pinching, biting, pulling hair, or boxing ears; forcing children to stay in uncomfortable positions; burning, scalding, or forced ingestion (for example, washing a child’s mouth out with soap or forcing them to swallow hot spices). The AAP strongly supports the need for adults to avoid physical punishment and verbal abuse of children.

Time out: Time-out from reinforcement ("time-out") is a procedure in which a child is placed in a different, less-rewarding situation or setting whenever he or she engages in undesirable or inappropriate behaviors. Typically, time-out is used in tandem with positive discipline techniques. For example, time-out might be employed to reduce the frequency of a student's negative behaviors (e.g., loud confrontations with teaching staff) while an individualized reward system might be put in place to increase the frequency of appropriate student behaviors (e.g., quickly and courteously complying with teacher requests).

e. Understand the importance of identifying the function of a behavior in selecting a behavior management technique

The four functions of behavior are sensory stimulation, escape, access to attention and access to tangibles. BCBA Megan Graves explains the four functions with a description and example for each function.

- **Sensory Stimulation**: "A person's own movements/actions feel good to that individual. For example, a child twirls his or her hair as they sit for an extended amount of time. If twirling hair gives that individual the sensory input they are seeking, then hair twirling will continue."
- **Escape**: "Something is (or signals) an undesirable situation and the person wants to get away from it. For example, a therapist says, 'Wash your hands,' and the learner runs out of the bathroom."
- **Access to Attention**: "Someone desires for access to social interaction(s). For example, the child screams, 'Look at me!' If screaming gets access to attention, then screaming will continue."
- **Access to Tangibles**: "Someone wants access to a specific item or activity. For example, Michelle takes the iPad away from Aaron, so Aaron pinches her. If pinching gets access to the iPad, then pinching will continue."

Identifying the function of behavior helps us to prevent problem behavior, teach our kids better ways to have their needs met and ensure consistency across all environments. We observe your child in his or her environments, describe what is going on before and after the problem behavior occurs, identify the function, teach a replacement behavior that still meets the same need as the problem behavior and reinforce the replacement behavior. Understanding the function of the behavior helps us to decrease the problem behaviors and increase appropriate or desired behaviors

f. Know the important components of a successful time-out procedure

Step 1: Decide whether a particular student/child would benefit from time-out. While time-out generally is effective in reducing problem behaviors, some children will not respond well to a time-out procedure … If you suspect a skill deficit, you should first be sure that the student has learned the appropriate skill(s) before you select time-out as a behavioral consequence.

Step 2: Select the type of time-out to be used. Teachers can choose from several time-out options that differ in the degree to which they exclude children from the instructional and/or social setting (Yell, 1994).

- 1. Non-Exclusionary Time Out. The child remains in the instructional setting but is temporarily prevented from engaging in reinforcing activities. Examples include planned ignoring, and removal of reinforcing objects or activities.
- 2. Exclusionary Time Out: Contingent Observation. The student is removed from the instructional setting to another part of the classroom. The student is instructed to continue to watch the instructional activities but cannot otherwise participate in them.
- 3. Exclusionary Time Out: Exclusion. The student is removed from the instructional setting to another part of the classroom. The student is prevented from watching or otherwise participating in group activities. (NOTE: An adult must supervise the student at all times during exclusion time out).
- 4. Exclusionary Time Out: Isolation/Seclusion. The student is removed from the instructional setting to a separate time-out room. (NOTE: An adult must supervise the student at all times during isolation/seclusion time out).

When choosing a form of time-out, you should try to pick the option that is least restrictive (i.e., keeps the child within the classroom and engaged in learning) whenever possible (Yell, 1994).

Step 3: Decide on other elements of the time-out program. When putting together a time-out plan, you must decide:

- how long each time-out period will last. Generally, a short (3-5 minute) time-out period is a good interval to start with, as there is no research to suggest that longer time-outs are any more effective than shorter ones.
- if the student is to receive a single warning before being sent to time-out. A teacher-delivered warning allows the child an opportunity to improve his or her behaviors and thus avoid being timed out. Warnings can take the form of verbal statements or non-verbal signals (e.g., eye contact with the student, a checkmark on the blackboard, etc.).
- what activities the student will engage in while in time-out. While you have considerable latitude in selecting what the student will do in time-out, keep in mind that time-out activities should never be more rewarding than what is going on in the classroom. Appropriate time-out activities might include completing class assignments, copying classroom rules, or writing a brief account of both the problem behavior that resulted in the time-out and more appropriate behaviors that would have helped the student to avoid time-out.
- how to judge that the student is ready to rejoin the class after time-out. In most cases, the child will behave appropriately in time-out and simply return to the classroom activity when the time-out period is over. However, if the student continues to be disruptive during time-out, you can simply reset the timer to zero and tell the student that he or she must act appropriately for a set interval of time

(e.g., 5 minutes) before the student can return to the class activity. The timer is reset at each additional outburst--until the child complies.

Step 4: Train the student/child in the time-out procedures. Prior to putting the time-out program into effect, sit down with the student and review the time-out procedures. They should:

- know what type(s) of inappropriate behaviors will earn him or her a time-out;
- have a clear understanding of the steps in the time-out process, including the use of a teacher warning (if selected), the agreed-upon signal that the student must go to time-out, the location of the time-out site, appropriate student behavior expected during time-out, and the length of time that time-out will last.
- understand how to reenter the classroom appropriately after time-out.

You will probably also want to walk them through a typical time-out sequence to ensure that the child clearly understands the process.

g. Know how to initiate a token economy within a home or school

Token economies (earning rewards and privileges contingent on performing desired behaviors) help children develop an understanding of cause and effect for behavior.

- Measure occurrence of appropriate behavior. (eg, voiding in toilet, reading page in book)
- Allow for visual feedback on progress for child. (eg, stickers on chart, coin/token in jar)
- Reminds adults to reinforce appropriate behavior.
- Provides motivation for child to see his/her progress.

Using token economies effectively

- Select durable tokens
- Consider student's/children's interest in token boards
- Deliver tokens immediately
- Use powerful reinforcers

h. Identify situations in which planned ignoring or time-out would not be an appropriate recommendation to decrease a problem behavior

1) Planned Ignoring

Ignoring isn't always the best option. Before deciding to ignore behavior, it's a good idea to check a few things.

Is the behavior rewarded by someone else's attention?

If the behavior is rewarded by someone else's attention – for example, siblings or friends – it won't make any difference if you ignore it. In this case, you might need to look at another behavior tool – for example, changing your child's environment .

Should you ignore the behavior?
Some behavior might be rewarded by your attention, but you might not be able or willing to ignore it. You can't ignore dangerous behavior or behavior that hurts others or damages property – for example, biting, hitting, pulling on the curtains or throwing things. In this case, a behavior tool like consequences or time-out might be appropriate.

Can you ignore the behavior if it gets worse?
Sometimes you might start ignoring behavior, but it gets worse and you end up giving it attention. For example, your child is tapping a block on a wooden floor, which you ignore. But then your child starts banging the block. If you criticize your child for banging the block, you run the risk of rewarding that behavior. This makes it more likely to happen again. In this situation, you could try simple breathing exercises while the banging is happening. But if you feel that you can't ignore the behavior if it gets worse, it's better not to try ignoring it in the first place.

Can you ignore the behavior wherever it happens?
If you ignore the behavior in one place but not another, you'll get more of the behavior in the place you don't want it. For example, if you ignore your child yelling at home but not at the supermarket, you might get more yelling at the supermarket. You could try planning ahead for the behavior at the supermarket.

Can you ignore the behavior whenever it happens?
This is crucial. If you ignore sometimes and not at other times, you can make it harder to change your child's behavior. Rewarding your child's behavior some of the time strengthens the behavior more than if the behavior is rewarded every time it happens. Planning ahead for your child's behavior and stress management for you can help.

Will other people ignore the behavior?
If you have managed to successfully ignore a behavior, but your partner, friend or relative suddenly comes in and pays attention, your good work will be undone. Backing each other up is an important part of managing your children's behavior, so it's good if you and your partner can talk and agree on what behavior you'll ignore. Sometimes others will find it difficult to understand your use of planned ignoring and might not be able to do it. If you're in this situation it's better not to use ignoring.

2) Time-Out

Increased Maladaptive Behaviors: If the child perceives time-out as a punishment, time-out can have serious side effects that are commonly associated with punishment, including increases of other maladaptive behaviors and withdrawal from or avoidance of the adults administering time-out. In addition, given their social inexperience, young children tend to internalize negative labels, see themselves as they are labeled, and react accordingly. If timeout is functioning to heighten opportunities to practice maladaptive behaviors—either withdrawal or aggression—its use should be reconsidered

Escape and Avoidance: Timeout may actually be reinforcing to a student when it functions as a consequence that allows him/her to escape from doing disliked tasks or complying with an adult demand. For example, work that is poorly suited to the child—too difficult, unclear, boring or tedious—may invoke misbehavior as the timeout appears more inviting to the student than struggling with the assigned work. If not careful, timeout may provide the student with a convenient way to escape having to be compliant, to get out of dealing with the teacher's correction or doing as told. In this case, timeout may actually function to strengthen the acting- out behavior.

Practice of Undesirable Behaviors: One of the drawbacks to using timeout is that it removes the child from classroom activities and deprives him/her from the opportunity to engage in appropriate or productive behaviors. Timeout should not be used with students who engage in withdrawal or self-stimulation. Once again, the importance of the functional behavior assessment cannot be overstressed. Timeout can also provide repetitive practice of aggressive behaviors. The student who argues continuously or talks back when instructed to go to timeout, or who screams, swears, and kicks while in timeout, is given the opportunity to practice highly ineffective responses to problems, rather than to learn more acceptable and productive alternative behaviors, such as following directions, accepting correction or a consequence, or sharing disagreements calmly

Negative Reactions of Others: When looking at the effectiveness of timeout, it is also important to consider the reactions of others. The student who challenges the adult by refusing to go to timeout, leaving or disrupting the classroom with shouting or profane language during timeout, may garner reinforcing laughter, comments or increased status from peers. These reactions not only will likely strengthen future occurrences of such acting-out behavior from the targeted student, but also may encourage, through modeling, similar behaviors from other students. Such intense misbehaviors as screaming, swearing, and kicking can also result in increased anxiety or fear among student witnesses who may be concerned about the safety of their classroom environment or question the effectiveness of the teacher in keeping order.

- time-out is more rewarding than the classroom setting. In some cases, teachers discover that time-out is in fact more diverting and rewarding for a student than is the classroom. For example, a student who is timed out in a neighboring classroom may enjoy the social opportunities available in that room and continue to act out to return to it as often as possible. If the time-out situation appears to be too reinforcing, take steps to move the location or change the activities to make it less inviting.
- the student lacks the skills to engage in the appropriate behavior. Time-out should be stopped and the student should be taught the needed behavior skill(s).
- the student is actually using misbehavior to escape the classroom setting. Time-out should be replaced with other behavior management strategies that do not allow the child to flee the classroom. If possible, you should also take steps to make the classroom more inviting for the student.

i. Know how to initiate a school-home note as a behavioral intervention

Schools seek effective but workable classroom interventions to address the problem behaviors of younger students. School-home notes are one strategy that holds promise for the primary classroom: the teacher sends home a daily note rating the student's school behaviors (Jurbergs, Palcic, & Kelley, 2007). Based on the teacher report, the parent provides or withholds a home reward. School-home notes have the advantages of both strengthening communication between teacher and parents and including the parent in the intervention as dispenser of praise and home rewards.

Preparation. Here are the steps to setting up a school-home note:

1. *Select target behaviors.* The teacher and parent decide on 2-4 behaviors to track through the school-home note. Behaviors listed on the note should be phrased as desired 'replacement' behaviors (that is, positive behaviors to replace the student's current challenging behaviors). For example, a behavior target for a non-compliant child might be "The *student followed teacher requests*."
2. *Design a school-home note*. The teacher and parent design a note incorporating target behaviors. While any rating format may be used, a simple version may be best--e.g., Yes (2 pts)...So-So (1 pt).....No (0 pts). See the attached school-home note for a generic example. A free application is also available on Intervention Central to create Behavior Report Cards, which can be used as school-home notes: http://www.interventioncentral.org/teacher-resources/behavior-rating-scales-report-card-maker
3. *Decide on the cut-point for an acceptable daily school-home note rating*. The parent and teacher decide on the minimum daily points that the child must earn on the school-home note to be eligible to earn a reward. For example, a teacher and parent create a school-home note that has 4 behavior-rating items, with a maximum of 2 points to be earned per item. The maximum points that can be earned per day on the school-home note therefore is 8 (4 items times 2 points per item). The teacher and parent initially decide that the student must earn a minimum of 5 points to earn a daily reward.
4. *Develop a reinforcer menu*. Based on a knowledge of the child, the parent develops a reinforcer ('reward') menu containing 4-8 reward choices. Whenever the student attains a positive rating on the school-home note, he or she can select a reward from this menu.

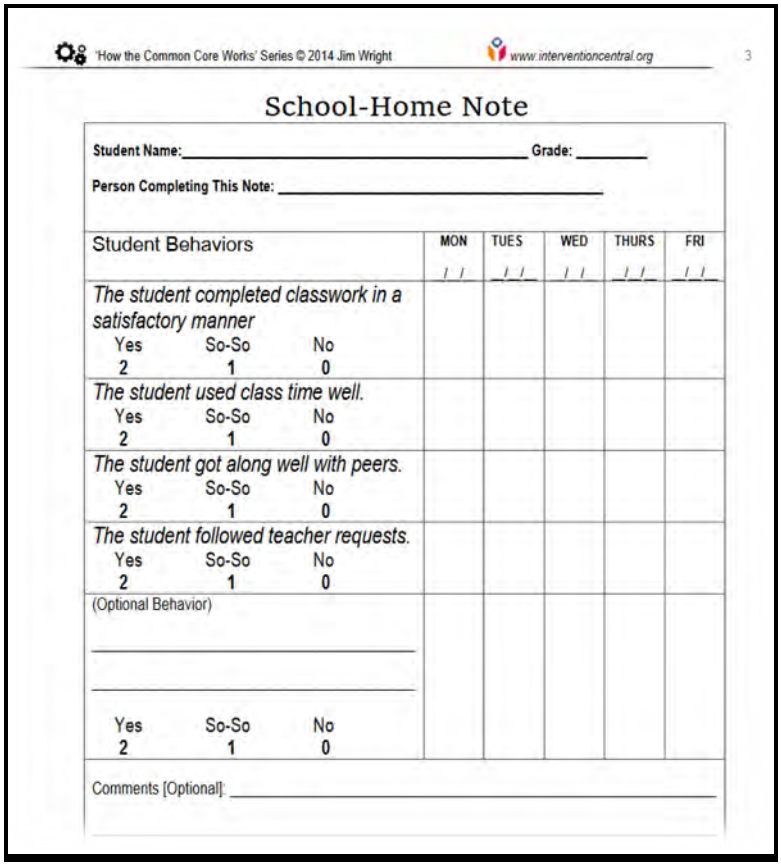
Implementation. Here are the daily steps for using school-home notes:

1. *Rate the student's school behavior.* At the conclusion of the school day, the teacher rates the student's behavior on the school-home note. The teacher meets briefly with the student to share feedback about the ratings and offers praise (if the ratings are positive) or encouragement (if the ratings are below expectations).
2. *Send the completed school-home note to the parent.* The teacher communicates the school-home note results with the parent in a manner agreed upon in advance, e.g., in the student's backpack, via email or a voicemail report.

3. *Provide the home reward*. The parent reviews the most recent school-home note with the child. If the child attained the minimum rating, the parent provides praise and allows the student to select a reward from the reinforcer menu. If the student failed to reach the rating goal, the parent withholds the reward but offers encouragement.

Maintenance. These are two items that are periodically updated to maintain the school-home note program:

1. *Refresh the reinforcer menu.* Every 2 to 3 weeks, the parent should update the reinforcer menu with the child to ensure that the reward choices continue to motivate.
2. *Raise the school-home note goal*. Whenever the student has attained success on the school-home note on most or all days for a full 2 weeks, the teacher and parent should consider raising the student point goal incrementally.



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School-Home Note

Student Name:_____ Grade: _____

Person Completing This Note: _____

Student Behaviors	MON / /	TUES / /	WED / /	THURS / /	FRI / /
The student completed classwork in a satisfactory manner Yes 2 So-So 1 No 0					
The student used class time well. Yes 2 So-So 1 No 0					
The student got along well with peers. Yes 2 So-So 1 No 0					
The student followed teacher requests. Yes 2 So-So 1 No 0					
(Optional Behavior) _____ _____ Yes 2 So-So 1 No 0					

Comments [Optional]: _____

* Intervention Central

j. Know the important components of a successful extinction procedure

Extinction refers to a procedure used in Applied Behavioral Analysis (ABA) in which reinforcement that is provided for problem behavior (often unintentionally) is

discontinued in order to decrease or eliminate occurrences of these types of negative (or problem) behaviors. While this procedure is most commonly used in children with Autism and Down Syndrome, it can also be used very successfully to address a broader array of problem behaviors, including those exhibited by individuals ***without*** developmental disabilities.

Extinction procedures often take three different forms depending upon the functions of the behavior (i.e. What is causing the behavior). One of the forms is to **use extinction with behaviors maintained by positive reinforcement.**

Example: Dannie tries to get mom's attention by dropping her toy on the floor. Her mom smiles at Dannie, picks up the toy and hands it back to her. This series of actions reinforces Dannie's negative behavior because she is getting the attention that she is seeking. As a result, she will continue to engage in this type of behavior in order to receive the positive reinforcement that her mom provides. To address this problem, Dannie's mom should ignore Dannie when she drops the toy; if she consistently ignores this problem behavior, it is highly likely that Dannie will reduce engaging in this behavior as her actions no longer produce the effect that she is seeking.

Another form of this procedure is **extinction on behaviors maintained by negative reinforcement**. This is commonly referred to as "**escape extinction**."

Example: Dannie throws a tantrum when she doesn't want to eat her food. Her mom responds by sending her to a 'corner' for time out. Because Dannie is able to avoid eating the food that she doesn't want to eat, it is highly likely that she will engage in the same behavior in the future. To correct this, Dannie's mom should let Dannie throw tantrums (regardless of how long it takes), while continuing to insist that Dannie eat her food. Initially, these tantrums will increase as Dannie becomes more and more frustrated, but eventually her tantrums will decrease as long as her actions do not provide her with the desired outcome.

The third form of this procedure is **extinction on behaviors maintained by automatic reinforcement**. This is commonly referred to as "**sensory extinction**."

Example: Dannie likes to turn the light switch on and off because she is visually stimulated by the fan starting and stopping. In order to address this behavior, Dannie's mom should disable the fan so that when Dannie flips on the light switch, she no longer gets the visual stimulation from the fan starting and stopping. Over time, Dannie will decrease engaging in this behavior of flipping the light switch because it no longer provides the automatic reinforcement she is seeking.

More facts about extinction procedure:

• An i**ncrease** in the negative behavior will likely be observed shortly after extinction procedures are implemented: this is referred to as an **extinction burst**. It is very

important for the person administrating therapy to maintain consistency and continue with the procedure, regarding of the child's reaction.

Typically, extinction bursts will increase initially and the child will engage in this negative behavior more frequently before the behavior goes away or decreases to an appropriate level. Extinction bursts can also happen after a long period during which the child does not engage in problem behavior. This is referred to as Spontaneous Recovery. It is very important to be mindful of this possibility in order to be prepared to deal with it in the same way the behavior was dealt with initially.

• All three forms of extinction procedures decrease the occurrence of problem behavior over time.

• Very simply, extinction equates to lack of reinforcement. Instead of getting something good to strengthen the behavior, or having something added or taken away to suppress the behavior, nothing happens. From the perspective of the child, the behavior no longer works to get the desired reinforcement any more.

• All forms of extinction procedures can be frustrating for the learner. Their level of frustration varies from learner to learner in each specific situation.

• Extinction procedures can also be frustrating for parents and caregivers because the reduction in positive behavior (behavior change) can be slow. This is generally tolerable if the behavior is mildly protesting or attention seeking, such as whining or crying. However, if the behavior involves self-injury or direct aggression to siblings, parents or caregivers, and it intensifies during the "extinction burst" period, parents may find the procedure to be impossible to maintain.

In these situations, implementing an additional procedure to increase the desired behavior, e.g., giving a reward of allowing a child to take smaller bites (for a selective eater), rewarding a child when he plays with his sibling to reduce aggression toward them, or increasing privileges for carrying out previously agreed-upon chores (a "behavioral contract") to increase compliance with directions, and other similar techniques can be quite helpful.

Table 7.1. Examples of Antecedent and Consequence Modification for Different Triggers

Trigger	Antecedent Modification	Consequence Modification
Restricted access (being told "no," "you can't," "stop," "don't," etc)	Remove forbidden items from environment (eg, childproofing). Clearly state rules for access (eg, first you do "X," then you get to do "Y"). Distract the child to another activity prior to the child gaining access to what is forbidden.	Consistently enforce rules. Allow access for appropriate behavior. Remove child from situation. Ignore misbehavior while persisting in denying access.
Need or desire for attention	Play with child with frequent positive comments on their play. Spend time with child without distractions from work, phone, computer, TV, etc. Plan family activities. Have someone available to engage child when caregiver is busy.	Increase frequency of attention for appropriate behavior. Ignore misbehavior meant to get attention. Institute time-out for certain attention-seeking misbehavior. Avoid inadvertent attention to minor misbehavior.
Task demands (that a child does not want to do)	After gaining the child's attention, clearly explain to the child, or show them, what is expected. Clearly state what will happen when the task is accomplished (first . . . then . . .). Clearly state what will happen if the child does not comply (eg, use a single warning).	Follow through on task demands. Carry out warnings that are made when task not completed.
Difficulty with task demands or communication	Alter task demand if difficulty is too high; provide breaks. Help child before frustration occurs. Model the behavior desired. Provide low-stress time for practicing skill. Show the child in pictures the sequence of tasks expected. Augment communication with sign language, pictures, etc.	Reward efforts for successful task completion. Follow nonpreferred tasks with preferred tasks. Provide help when request for help is made appropriately.
Transitions, change in routine, or change in what child expects	Create visual schedules. Prepare the child for changes and transitions. Establish routines. Teach skills related to flexibility.	Calmly persist with change while ignoring inappropriate behavior. Praise successful transitions. Schedule nonpreferred activities before preferred activities.
Provocation by sibling or peer	Monitor interactions. Teach appropriate interactions. Find activities both enjoy.	Pay attention when siblings are interacting well. Institute time-out or other punishment (eg, toy is removed) for both participants.
Fear or aversive sensory stimulus	Avoid fearful or adverse stimulus. Expose gradually.	Reward successful adaptation to exposure. Respond to appropriate requests for breaks or escape.

*AAP Developmental and Behavioral Pediatrics, 2nd Edition Page 97

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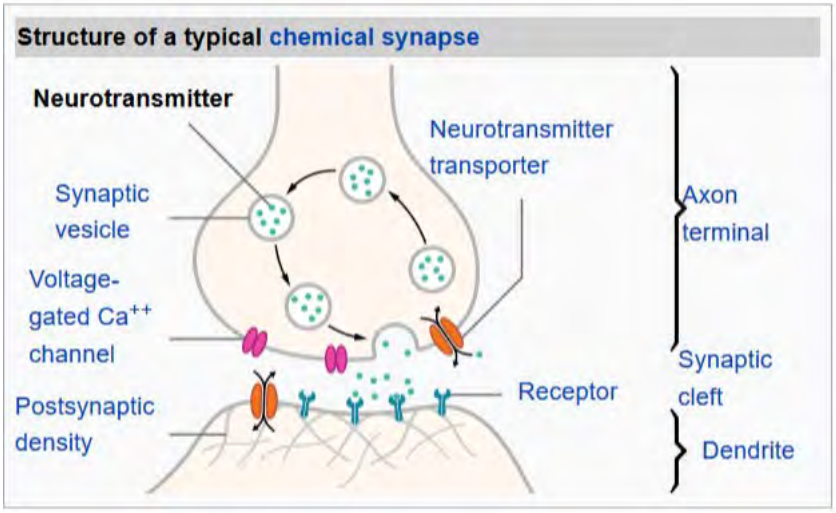
5. Basic pharmacotherapy
a. Understand the physiologic activity of various neurotransmitters (e.g., dopamine, norepinephrine, serotonin, gamma-aminobutyric acid, acetylcholine)

- Neurotransmitters are endogenous chemicals acting as signaling molecules that enable neurotransmission
    - They are a type of chemical messenger which transmits signals across a chemical synapse from one neuron (nerve cell) to another 'target' neuron, to a muscle cell, or to a gland cell
    - Neurotransmitters are released from synaptic vesicles in synapses into the synaptic cleft, where they are received by neurotransmitter receptors on the target cell
    - Many neurotransmitters are synthesized from simple and plentiful precursors such as amino acids, which are readily available and only require a small number of biosynthetic steps for conversion
    - Neurotransmitters are essential to the function of complex neural systems
    - The exact number of unique neurotransmitters in humans is unknown, but more than 200 have been identified



- Dopamine
    - Dopamine is made up of a benzene ring with two hydroxyl side groups attached to one amine group via an ethyl group
    - It is produced by dopaminergic neurons in the brain from tyrosine via the addition of a hydroxyl group which transforms it to L-DOPA (or Levo-DOPA) and subsequent removal of a carboxylic acid group from the ethyl side chain linked to the amine group resulting in Dopamine
    - The dopaminergic neurons that produce this signaling molecule are located in the brain at the substantia nigra and ventral tegmental area

which are both located in the midbrain as well as the arcuate nucleus of the hypothalamus

- After production of dopamine, the neurotransmitter is packaged into a synaptic vesicle, vesicular monoamine transporter 2 (VMAT2) and stored until action potentials induce the release of dopamine into the synaptic cleft and cause binding to dopamine receptors on the postsynaptic neuron.
- Dopamine neurotransmitters bind to five subtypes of dopamine receptors: D1, D2, D3, D4, and D5, which are members of the G-protein coupled receptor family that are divided into two major subclasses: D-1-like and D-2-like.
- The binding of dopamine to these receptors initiates cascades of signaling responsible for activating functions in the associated areas of the brain where each receptor type is most prevalent
- D1-like receptors are more prevalent than D2-like receptors→understanding how dopamine functions in the human brain as a neurotransmitter requires looking at the effect of dopamine binding to D1-like and D2-like receptor types to exert their effects via second messenger systems
- The binding of dopamine to D1-like receptors (D1 and D5) results in excitation via the opening of Na+ channels or inhibition via the opening of K+ channels.
- D1-like receptor stimulation induces the activation of adenylate cyclase, the enzyme that converts ATP to cAMP, thereby increasing cAMP levels leading to the disinhibition of protein kinase A (PKA) which phosphorylates downstream targets such as cAMP regulatory element binding protein (CREB). The translocation of CREB to the nucleus and CREB-dependent transcription of numerous genes is responsible for the synaptic plasticity necessary for learning and memory formation. In contrast, D-2 like receptor binding (D2, D3, and D4) lead to inhibition of the target neuron by exerting an opposite effect of inhibiting adenylate cyclase through coupling to G proteins Gi/o which decreases the production of cAMP. Whether dopamine is excitatory or inhibitory is a matter of which type of effect on a target neuron is exerted which is based on which types of receptors are on the membrane surface of the neuron and how the neuron responds to increases or decreases in cAMP concentration.
- Dopamine plays important roles in executive function, motor control, motivation, arousal, reinforcement, and reward through signaling cascades that are exerted via binding to dopaminergic receptors at the projections found in the substantia nigra, ventral tegmental area, and arcuate nucleus of the hypothalamus of the human brain.
    - In the substantia nigra, the nigro-striatal pathway projects dopaminergic neurons from the input area (known as the pars compacta) to the dorsal striatum and plays a primary role in the control of motor function and learning motor skills

- If the dopaminergic neurons in the nigro-striatal pathway degenerate, this causes a dysregulation of motor control, a hallmark of Parkinson's Disease
- In the ventral tegmental area (VTA), the mesolimbic pathway projects from the prefrontal cortex to the nucleus accumbens of the amygdala, cingulate gyrus, hippocampus, and pyriform complex of the olfactory bulb.
    - The dopaminergic projections in the amygdala and cingulate gyrus are responsible for emotion formation and processing.
    - In the hippocampus, the presence of dopaminergic neurons is associated with learning, working memory, and long-term memory formation.
    - Lastly, the pyriform complex of the olfactory bulb is responsible for providing humans with the sense of smell.
    - In the mesolimbic pathway, dopamine is released during pleasurable situations, causing arousal and influences behavior (motivations) to seek out the pleasurable activity or occupation and bind to dopaminergic receptors present in the nucleus accumbens and prefrontal cortex.
    - Increased activity in the projections to the nucleus accumbens play a major role in reinforcement and in more extreme cases with addictions.
    - In the arcuate nucleus of the hypothalamus, dopamine neurons make up the tuberoinfundibular pathway which projects to the pituitary gland and inhibits the secretion of the hormone prolactin.
        - Dopamine produced by neurons in the arcuate nucleus are released in the hypothalamo-hypophysial blood vessels which supply the pituitary gland with dopamine to inhibit the production of prolactin

Table 1.

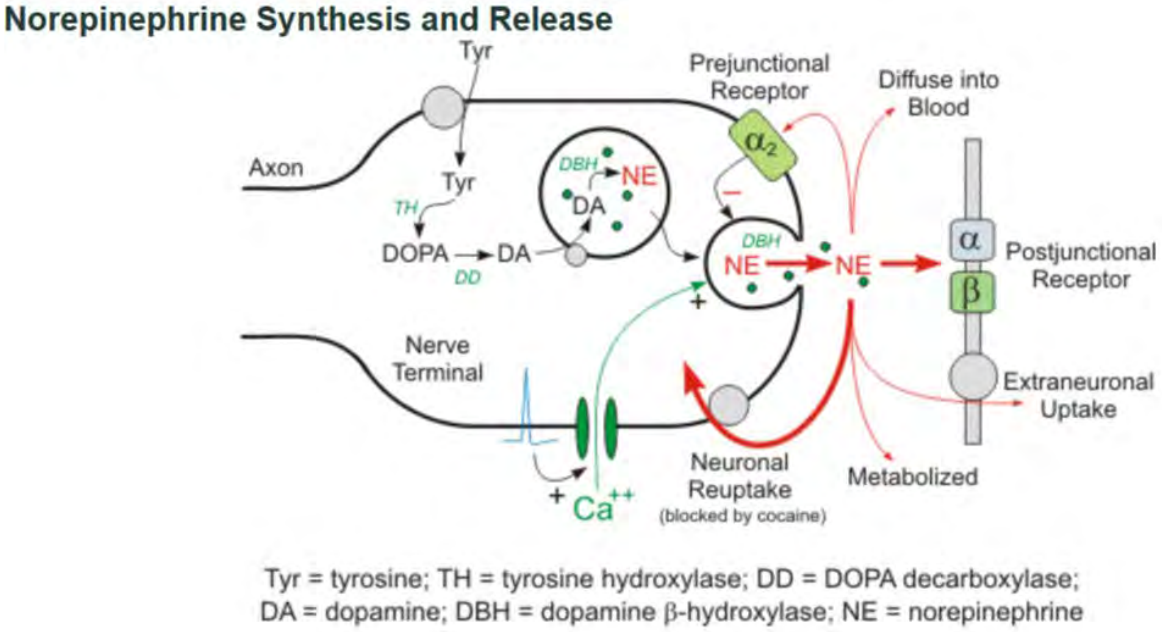
Dopamine receptor classification, localization, and their functions.

Receptors	D1	D5	D2	D3	D4
Location	Striatum, nucleus accumbens. Olfactory bulb, amygdala hippocampus, substantia nigra Hypothalamus, frontal cortex	Cortex, substantia nigra, hypothalamus	Striatum, VTA Olfactory bulb, cerebral cortex	Striatum, islands of Calleja, cortex	Frontal cortex, amygdala, hypothalamus, nucleus accumbens
Type	Gs-coupled	Gs-coupled	Gi-coupled	Gi-coupled	Gi-coupled
Mechanism	Increased intracellular level of cAMP by activated adenylate cyclase	Adenylate cyclase↑	Increased intracellular level of cAMP by activate adenylate cyclase	Adenylate cyclase↓	Adenylate cyclase↓
Function	Locomotion, learning and memory, attention, impulse control, sleep, regulation of renal function	Cognition, attention, decision making, motor learning, renin secretion	Locomotion, learning and memory, attention, sleep, reproductive behaviour	Locomotion, cognition, attention, impulse control, sleep, regulation of food intake	Cognition, impulse control, attention, sleep, reproductive behavior
Selective agonist	SKF-38393 SKF-81297 Fenoldopam (SKF-82526)	—	Bromocriptine Pergolide Cabergoline Ropinirole	7-OH-DPAT Pramipexole Rotigotine PD-128907	A-412997 ABT-670 PD-168077
Selective antagonist	SCH-23390 SCH-39166 SKF-83566	—	Haloperidol Raclopride Sulpiride Spiperone Risperidone	Nafadotride GR-103691 GR-218231 SB-277011A NGB-2904 PG-01037 ABT-127	A-381393 FAUC213 L-745870 L-750667

- Norepinephrine
    - also known as noradrenaline, is a neurotransmitter of the brain that plays an essential role in the regulation of arousal, attention, cognitive function, and stress reactions
    - primary neurotransmitter at postganglionic sympathetic adrenergic nerves as it also functions as a hormone peripherally as part of the sympathetic nervous system in the "fight or flight" response
    - The noradrenergic system has been implicated in the pathogenesis of some significant neuropsychiatric disorders and has been an important pharmacologic target in various psychiatric, neurologic, and cardiopulmonary disorders
    - Production of NE at the cellular level:
        - Tyrosine is an aromatic amino acid that can cross the blood-brain barrier and be transported into neurons in the presynaptic terminal, where it serves as a precursor for catecholamine synthesis
        - Elevated levels of tyrosine in the central nervous system (CNS) triggers the production of norepinephrine and other catecholamines
        - The steps for norepinephrine synthesis are as follows:
            - Tyrosine is first hydroxylated to dihydroxyphenylalanine (DOPA) by tyrosine hydroxylase (rate-limiting step)
            - DOPA is then decarboxylated by L-amino acid decarboxylase to produce dopamine
            - Dopamine gets transported into a vesicle through vesicular monoamine transporter (VMAT) where it can be converted to norepinephrine by neurons that contain an additional enzyme, dopamine beta-hydroxylase
            - Norepinephrine can then be released from the presynaptic terminal to the synaptic cleft via exocytosis or convert to

epinephrine in neurons that contain the enzyme phenylethanolamine-N-methyl transferase.

- Phenylalanine is another amino acid that can also be used for catecholamine synthesis after it is converted to tyrosine by phenylalanine hydroxylase
- In the peripheral nervous system, chromaffin cells in the adrenal medulla (following the same steps as mentioned above) synthesize norepinephrine
- Once synthesized, they get stored in chromaffin granules.
- Norepinephrine can be released into the bloodstream or be converted to epinephrine by phenylethanolamine-N-methyl transferase
- The release of these hormones into the bloodstream is stimulated by acetylcholine (released from preganglionic splanchnic fibers) which binds nicotinic receptors located in the adrenal medulla



- **Central Nervous System**

  - The central noradrenergic system is composed of two primary ascending projections that originate from the brainstem: The dorsal noradrenergic bundle (DNB), and the ventral noradrenergic bundle (VNB)
  - The DNB originates from A6 locus coeruleus, located in the dorsal pons, and is composed of primarily noradrenergic neurons.
  - It functions as the predominant site of norepinephrine production in the central nervous system.

        - It sends projections to innervate the cerebral cortex, hippocampus, and cerebellum exclusively and has projections that overlap with projections from the VNB to innervate areas of the amygdala, hypothalamus, and spinal cord
        - The VNB originates from nuclei in the pons and medulla and sends projections to innervate the amygdala, hypothalamus, and areas of the midbrain and medulla.

- **Peripheral Nervous System**

        - The sympathetic nervous system and neuroendocrine chromaffin cells (located in the adrenal medulla) are primarily responsible for the synthesis and exocytosis of norepinephrine and other catecholamines into the blood circulation.
        - The hormones act on alpha- and beta-adrenergic receptors of smooth muscle cells and adipose tissue located throughout the body.

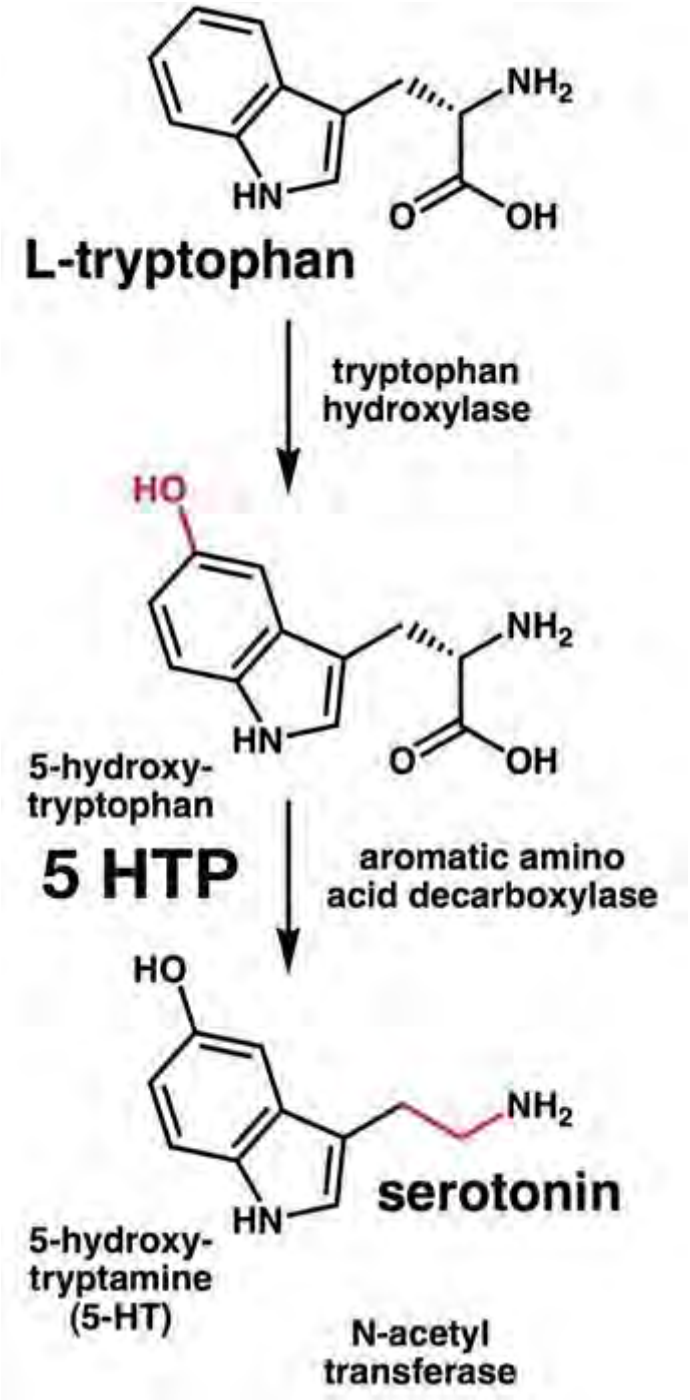
- the noradrenergic system has various functions throughout the central and peripheral nervous systems
    - One major role it is involved in is the body's "fight or flight" response.
    - During states of stress or anxiety, norepinephrine and epinephrine are released and bind to adrenergic receptors throughout the body which exert effects such as dilating pupils and bronchioles, increasing heart rate and constricting blood vessels, increasing renin secretion from the kidneys, and inhibiting peristalsis.

        - Norepinephrine has also shown to play a role in metabolic effects such as stimulating glycogenolysis and gluconeogenesis (while reducing glucose clearance) and inducing ketogenesis and lipolysis.
        - In the CNS, the noradrenergic system classically operates in its roles in promoting wakefulness and arousal and facilitating sensory signal detection.
        - Further studies have shown that it may also influence some areas of cognition and behavior such as attention, working memory, long-term mnemonic processing, and behavioral flexibility
        - Specifically, alpha-1 and alpha-2 receptors have been found to influence cognitive functions such as working memory, attention, and fear and spatial learning
        - Beta-1 and beta-2 receptors have been found to function in auditory fear, spatial reference and fear memory, and memory retrieval
        - Stimulation or inhibition of these functions is dependent on agonism or antagonism of the adrenergic receptors

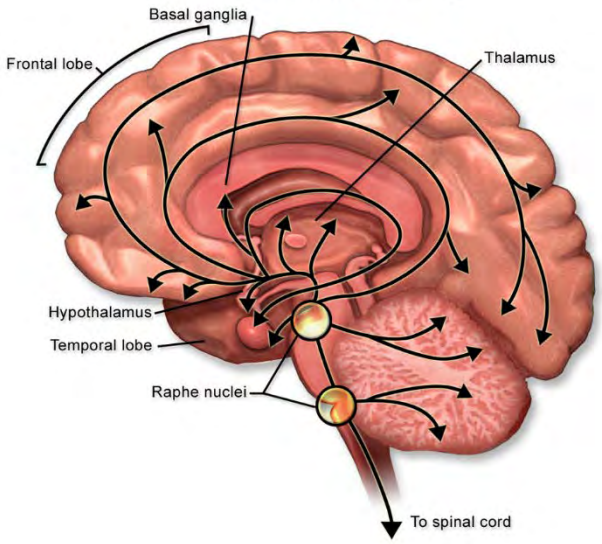
        - In general, alpha-1 and beta receptors enhance neurotransmission and plasticity, promoting stimulatory effects on the CNS, while alpha-2 receptors have inhibitory effects in the CNS such as reducing norepinephrine release and decreasing neuronal excitability.

- Serotonin
    - Serotonin or 5-hydroxytryptamine (5-HT) and serotonin receptors are important in the regulation of virtually all brain functions, and dysregulation of the serotonergic system has been implicated in the pathogenesis of many psychiatric and neurological disorders
    - Serotonin is a small molecule that functions both as a neurotransmitter in the central nervous system and as a hormone in the periphery
        - Serotonin is synthesized through a multistep pathway in which L-tryptophan is converted into L-5OH-tryptophan by an enzyme called tryptophan hydroxylase (Tph).
        - L-5OH-tryptophan is then converted to serotonin by an aromatic L-amino acid decarboxylase
        - There are two Tph genes: *Tph1* and *Tph2*
        - *Tph1* is expressed mostly in enterochromaffin cells of the gut and is responsible for most of the serotonin present in the blood
        - On the other hand, the gene *Tph2* is expressed exclusively in serotonergic neurons of the brainstem and is responsible for the production of serotonin in the brain
        - Brain-derived serotonin (BDS) acts as a neurotransmitter, while gut-derived serotonin (GDS) acts as a hormone and regulates a wide variety of processes
        - These two pools of serotonin, one in the blood and the other in the brain, should be viewed from a functional point of view as two distinct molecules
    - As stated above, in the central nervous system (CNS), serotonin is almost exclusively produced in neurons originating in the raphe nuclei located in the midline of the brainstem.
        - These serotonin-producing neurons form the largest and most complex efferent system in the human brain.
        - The most caudal raphe innervate the spinal cord, while the more rostral raphe, the dorsal raphe nucleus and the medial raphe nucleus, innervate much of the rest of the CNS by diffuse projections.
        - Indeed, virtually every cell in the brain is close to a serotonergic fiber, and nearly all behaviors as well as many other brain functions are regulated by serotonin.
        - Not surprisingly, serotonin receptors and transporters are a major focus of CNS drug development, and many current medications modulate serotonin neurotransmission including: SSRI's, TCA's, MAOI's (among other)

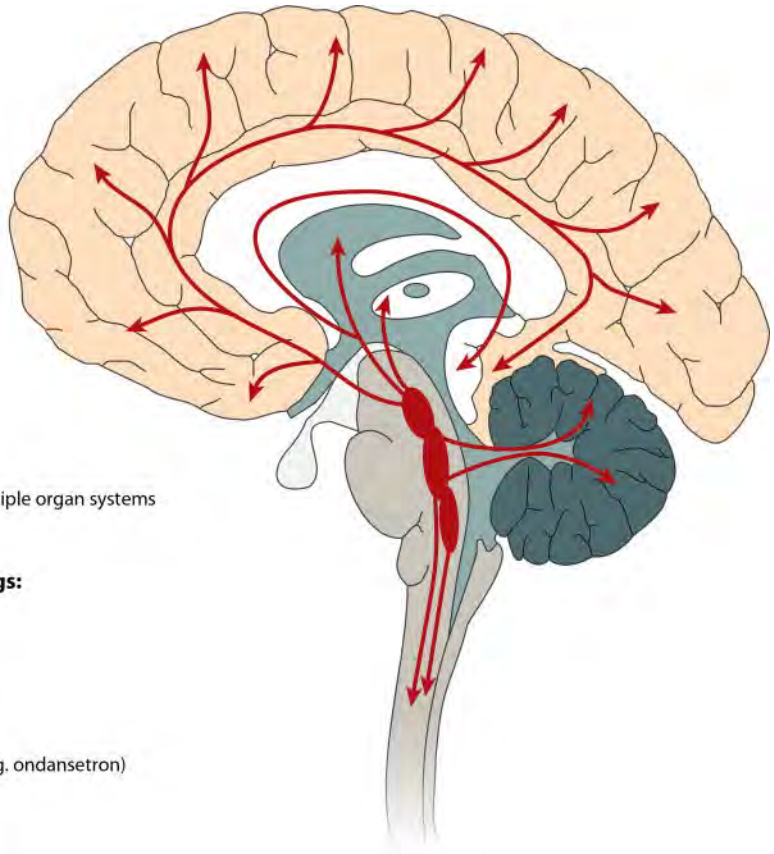
- Research in recent years, however, has brought to light the complexity of serotonin and the interplay b/t central and peripheral effects→release of 5-HT in the synaptic cleft, where it majorly has a receptor-dependent effect through one pathway will cause adrenergic responses (e.g., mydriasis, increased heart, and respiratory rates, etc.), which in observation are adrenergic outcomes→Thus, different serotonergic and adrenergic receptor subtypes might show distinctive molecular affinities and actions
- There are 15 known types of serotonin receptors (also known as 5-HT receptors, after the chemical name for serotonin, 5-hydroxytryptamine)
    - Mediate both excitatory and inhibitory neurotransmission
    - The serotonin receptors are activated by the neurotransmitter serotonin, which acts as their natural ligand.
    - The serotonin receptors modulate the release of many neurotransmitters, including glutamate, GABA, dopamine, epinephrine / norepinephrine, and acetylcholine, as well as many hormones, including oxytocin, prolactin, vasopressin, cortisol, corticotropin, and substance P, among others.
    - The serotonin receptors influence various biological and neurological processes such as aggression, anxiety, appetite, cognition, learning, memory, mood, nausea, sleep, and thermoregulation.


NH2
HN
O
OH
L-tryptophan
tryptophan
hydroxylase
HO
NH2
HN
O
OH
5-hydroxy-
tryptophan
5 HTP
aromatic amino
acid decarboxylase
HO
NH2
serotonin
HN
5-hydroxy-
tryptamine
(5-HT)
N-acetyl
transferase

Serotonin Pathway

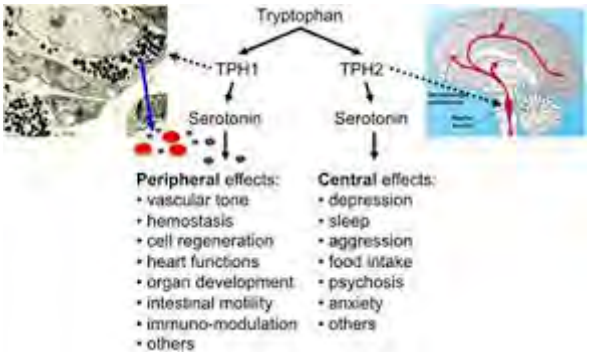


Behavioral effects:
Mood
Perception
Memory
Anger
Aggression
Fear
Stress responses
Appetite
Addiction
Sexuality



Other CNS effects:
Motor control
Cerebellar regulation
Sleep/circadian rhythms
CNS vascular tone
Emesis
Respiratory drive
Body temperature
Descending regulation of multiple organ systems

Central serotonergic drugs:
SSRIs
Tricyclic antidepressants
MAOIs
Other antidepressants
Buspirone
Atypical antipsychotics
Triptans
5-HT_3 receptor antagonists (e.g. ondansetron)
Fenfluramine
Ergotamine/methysergide
Hallucinogens



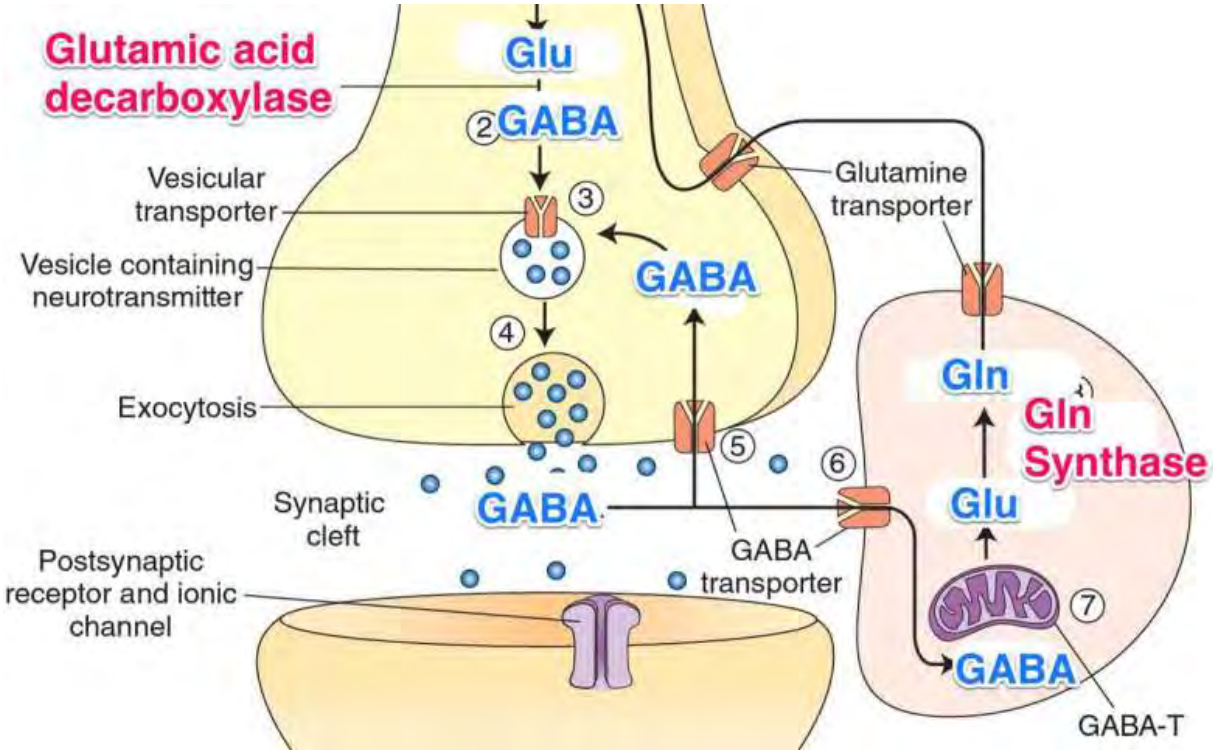
- GABA ***gamma*-Aminobutyric acid**, or **γ-aminobutyric acid**
    - Is an amino acid that serves as the chief inhibitory neurotransmitter in the brain and a major inhibitory neurotransmitter in the spinal cord
    - Its principal role is reducing neuronal excitability throughout the nervous system.
    - It exerts its primary function in the synapse between neurons by binding to post-synaptic GABA receptors which modulate ion channels, hyperpolarizing the cell and inhibiting the transmission of an action potential.
    - Disorder in GABA signaling is implicated in a multitude of neurologic and psychiatric conditions- including anxiety disorders, schizophrenia, bipolar disorder, major depressive disorder
    - Modulation of GABA signaling is the basis of many pharmacologic treatments in neurology, psychiatry, and anesthesia
    - There are numerous uses for drugs that modulate GABA signaling
        - Benzodiazepines are a drug class that exerts its effects by binding to the GABA-A receptor, resulting in increased chloride ion permeability by changing the frequency with which the chloride channels open→used in surgical anesthesia, the treatment of epilepsy, REM-sleep disorders, alcohol withdrawal, essential tremor, and muscle spasticity
        - Ethanol, one of the oldest and most widely-used psychoactive substances, also exerts effects on the GABA-A receptor→Alcohol withdrawal is treated with GABA modulating drugs, such as benzodiazepines
        - Furthermore, ethanol and benzodiazepines exhibit cross-tolerance with one another due to their similar mechanism of action
        - Overdosing or taking multiple GABA modulating drugs can result in respiratory depression due to increased GABA signaling in the medulla of the brain stem
        - Many other drugs modulate GABA signaling, including the following:
            - Barbiturates, sedative drugs which increase the duration at which the chloride channel is open when GABA binds the GABA-A receptor
            - Vigabatrin, an antiepileptic inhibitor of GABA transaminase

- Propofol, a sedative commonly used in general anesthesia and allosteric modulator and agonist of the GABA-A receptor
- Flumazenil, a benzodiazepine antagonist which binds to the GABA-A receptor and can reverse benzodiazepine intoxication and improve mental status in hepatic encephalopathy
- Baclofen, a muscle relaxant and GABA-B agonist
- Valproic acid, a mood stabilizer and anti-epileptic that is hypothesized to have an inhibitory effect on GABA uptake
- Zolpidem, a sedative-hypnotic, exerts its effects on the GABA-A receptor
- Gabapentin, commonly prescribed to treat neuropathic pain, partially exerts its effects by increasing GABA synthesis via modulation of glutamate dehydrogenase

- Because GABA is the fundamental neurotransmitter for inhibiting neuronal firing, its function is determined by the neural circuit that it is inhibiting.
  - It is involved in complex circuits throughout the central nervous system.
  - For example, GABA is released by striatal neurons in both the direct and indirect pathways projecting to the globus pallidus, which in turn extends GABA neurons to other brain areas, inhibiting unwanted motor signals.
  - Another example is that GABA signaling in the medulla is involved in the maintenance of respiratory rate→Increased GABA signaling reduces the respiratory rate.
  - A third example is found in the spinal cord, where GABA serves in the inhibitory interneurons→These neurons help to integrate excitatory proprioceptive signals, allowing for the spinal cord to integrate sensory information and create smooth movements.

- GABA is involved in several disease states:
  - Pyridoxine deficiency is a rare disease in which the vitamin is not available for the synthesis of GABA. It usually presents as frequent seizures during infancy that are resistant to treatment with anticonvulsants but responds very well to vitamin supplementation.
  - The clinical features of hepatic encephalopathy are thought to be due to elevated ammonia levels binding to the GABA-A/GABA complex and increasing chloride ion permeability.
  - The symptoms of Huntington disease are partially caused by a lack of GABA in the striatal projections to the globus pallidus.
  - Dystonia and spasticity are believed to be related to a deficiency in GABA signaling.

- GABA is synthesized in the cytoplasm of the presynaptic neuron from the precursor glutamate by the enzyme glutamate decarboxylase, an enzyme which uses vitamin B6 (pyridoxine) as a cofactor
    - After synthesis, it is loaded into synaptic vesicles by the vesicular inhibitory amino acid transporter.
    - SNARE (soluble N-ethylmaleimide-sensitive fusion protein attachment protein receptors) complexes help dock the vesicles into the plasma membrane of the cell.
    - When an action potential reaches the presynaptic cell, voltage-gated calcium channels open and calcium binds to synaptobrevin, which results in the fusion of the vesicle with the plasma membrane and releases GABA into the synaptic cleft where it can bind with GABA receptors.
    - GABA can then be degraded extracellularly or taken back up into glia or the presynaptic cell
    - It is degraded by GABA-transaminase into succinate semialdehyde which then enters the citric acid cycle.
- GABA binds to two major post-synaptic receptors, the GABA-A and GABA-B receptors.
    - The GABA-A receptor is an ionotropic receptor that increases chloride ion conductance into the cell in the presence of GABA.
    - The extracellular concentration of chloride is normally much higher than the intracellular concentration.
    - Consequently, the influx of negatively charged chloride ions hyperpolarizes the cell, inhibiting the creation of an action potential.
    - The GABA-B receptor functions via a metabotropic G-protein coupled receptor which increases postsynaptic potassium conductance and decreases presynaptic calcium conductance, which consequently hyperpolarizes the postsynaptic cell and prevents the conduction of an action potential in the presynaptic cell.
    - Consequently, regardless of binding to GABA-A or GABA-B receptors, GABA serves an inhibitory function.
    - One exception would be in the fetal and neonatal brain-→Due to extracellular concentrations of chloride being lower than intracellular levels in the developing brain, GABA has an excitatory role in the fetal and neonatal brain.
        - When GABA-A receptors open chloride channels in the developing brain, the cell becomes hypopolarized and thus more likely to fire an action potential.
        - Consequently, drugs that increase GABA signaling have been reported to be of limited efficacy in the treatment of seizures in preterm neonates



- Acetylcholine
  - Acetylcholine is a neurochemical that has a wide variety of functions in the brain and other organ systems of the body
  - Specifically, it is a neurotransmitter that acts as a chemical message that is released by neurons and allows them to communicate with one another as well as other specialized cells such as myocytes and cells found in glandular tissues
  - The name "acetylcholine" is derived from its chemical structure, as it is an ester of acetic acid and choline
  - Tissues of the body that use this chemical messenger or are responsive to it are referred to as cholinergic
  - There is a class of chemicals called anticholinergics that interfere with acetylcholine's action on tissues as well
  - While ACh operates as a neurotransmitter in many parts of the body, it is most commonly associated with the neuromuscular junction
  - Acetylcholine also functions as a neurotransmitter in the autonomic nervous system, acting both as the neurotransmitter between preganglionic and postganglionic neurons as well as being the final release product from parasympathetic postganglionic neurons
  - Acetylcholine intervenes in numerous physiological functions, such as the regulation of cardiac contractions and blood pressure, intestinal peristalsis, glandular secretion, etc
  - Typically, acetylcholine is an excitatory mediator
  - Acetylcholine ensures rapid but generally fleeting neurotransmission due to the prompt inactivation of the mediator in the synaptic cleft by the enzyme acetylcholinesterase

- Acetylcholine receptors subdivide into two types: nicotinic - ion channels for sodium and calcium, and muscarinic -coupled with G proteins.
- Acetylcholine is also involved in the immune system as T lymphocytes secrete it
- Acetylcholine performs its actions by binding the cholinergic receptors (muscarinic and nicotinic)
    - Acetylcholine performs various functions through cholinergic muscarinic receptors
        - In the cardiovascular system, it determines generalized vasodilation; decrease in heart rate (negative chronotropic effect); reduction of cardiac contraction force (negative inotropic effect), at the ventricular level this effect is less pronounced; decrease in the speed of conduction in the specialized tissue of the sinoatrial and atrioventricular nodes (negative dromotropic effect).

        - In the gastrointestinal system, through stimulation of the vagus nerve, the tone, the amplitude of contractions, and the secretory activity of the stomach and intestine increase, sphincters are released.
        - In the respiratory system, it determines bronchoconstriction and stimulation of the chemoreceptors of the aortic and carotid glomus, with consequent reflex hyperpnea.
        - In the urinary system, through parasympathetic sacral stimulation, it causes contraction of the detrusor muscle of the bladder, increasing the emptying pressure and ureteral peristalsis, the release of sphincters.
        - In the exocrine glands, it stimulates the secretion of all the exocrine glands that receive a parasympathetic innervation, including the lacrimal, tracheobronchial, salivary, digestive glands and the exocrine sweat glands.
        - In the eye, it determines miosis and accommodation of the lens in close vision, inducing the contraction of the sphincter muscle of the pupil and the ciliary muscle.
        - Involved in erectile function in the male reproductive system

    - Through the nicotinic cholinergic receptors, however, acetylcholine allows for skeletal muscle contraction; in the adrenal glands, the release of adrenaline and norepinephrine; and in the peripheral sympathetic ganglia, activation of the sympathetic system with the release of norepinephrine

- The system of cholinergic nerve fibers that release acetylcholine at their endings is widespread in both the central and peripheral nervous systems
    - In the periphery, all the preganglionic fibers are cholinergic, sympathetic, and parasympathetic, the parasympathetic

postganglionic, and the motor fibers that innervate the voluntary skeletal muscle

- In the somatic nervous system, acetylcholine is used at the neuromuscular junctions, triggering the firing of motor neurons and affecting voluntary movements
- Within both the sympathetic and parasympathetic systems, acetylcholine is utilized by presynaptic neurons of the intermediate horn of the spinal cord to communicate with post-synaptic neurons
- Within the parasympathetic nervous system alone, the postganglionic neuron releases acetylcholine as its primary neurotransmitter
- Within the sympathetic nervous system, the only postganglionic neurons that release acetylcholine as their primary neurotransmitter are those found innervating the sudoriferous (sweat) glands and some blood vessels of non-apical skin.

▪ In the central nervous system, the cholinergic system has extensive branches in the spinal cord, thalamus, limbic system, and cortex

- Within the brain, acetylcholine has involvement in memory, motivation, arousal, and attention
- Acetylcholine originates from two major places in the brain: 1) basal forebrain and 2) the mesopontine tegmentum area
- Acetylcholine originates in the basal forebrain from both the basal nucleus of Meynert and the medial septal nucleus→ The basal nucleus of Meynert works on the M1 receptors within the neocortex and the medial septal nucleus functions in the hippocampus as well as parts of the cerebral cortex at the M1 receptors
- The mesopontine tegmentum is in the brain stem, and acetylcholine comes from its pedunculopontine nucleus and laterodorsal tegmental nucleus→the mesopontine tegmentum mainly activates the M1 receptors in the brainstem. The M1 receptors in the brainstem are present in the raphe nucleus, lateral reticular nucleus, deep cerebellar nuclei, pontine nuclei, locus coeruleus, and the inferior olive. However, the mesopontine tegmentum also projects to the basal ganglia, thalamus, basal forebrain, and tectum.

o Acetylcholine (ACh) is clinically significant in many disease processes, the most commonly seen of which include Alzheimer disease (AD), Lambert-Eaton myasthenic syndrome (LEMS), and myasthenia gravis (MG)

        - Patients with AD have reduced cerebral content of choline acetyltransferase, which leads to a decrease in acetylcholine synthesis and impaired cortical cholinergic function→Cholinesterase inhibitors (donepezil, rivastigmine, and galantamine) increase cholinergic transmission by inhibiting cholinesterase at the synaptic cleft and provide modest symptomatic benefit in some patients with dementia
        - MG is an autoimmune disorder that is recognized by the rapid weakening of the skeletal muscles after repeated use→due to an antibody-mediated process in which antibodies are produced that have a tropism for acetylcholine receptors or their associated proteins, located in the postsynaptic membrane of the neuromuscular junction
        - LEMS is a disorder of reduced Ach release from the presynaptic nerve terminals, despite normal ACh vesicle number, normal ACh presynaptic concentration, and normal postsynaptic acetylcholine receptors→This condition occurs when there is autoimmunity (production of autoantibodies) to the voltage-gated calcium channels found on presynaptic neurons' axon terminum

b. Know how to apply the principles of pharmacodynamics (see schematic below for comparison of pharmacodynamics versus pharmacokinetics)

- Pharmacodynamics is the study of a drug's molecular, biochemical and physiologic effects or actions→ In a general sense, pharmacodynamics is the study of dose-response relationships; takes into account the complex interactions between the drug, the human body, and then the pathogen that might be causing an infection in the patient
    - comes from the Greek words "pharmakon" meaning "drug" and "dynamikos" meaning "power."
    - All drugs produce their effects by interacting with biological structures or targets at the molecular level to induce a change in how the target molecule functions in regards to subsequent intermolecular interactions.
    - These interactions include receptor binding, postreceptor effects, and chemical interactions.
    - Examples of these types of interactions include (1) drugs binding to an active site of an enzyme, (2) drugs that interact with cell surface signaling proteins to disrupt downstream signaling, and (3) drugs that act by binding molecules like tumor necrosis factor (TNF)
    - subsequent to the drug-target interaction occurring downstream, effects are elicited which can be measured by biochemical or clinical means.
- Pharmacodynamic Concepts
    - There are a few key concepts and terms used in the description of pharmacodynamics that describe the extent and duration of a drug's action:
        - Emax is the maximal effect of a drug on a parameter that is measured. For example, this could be a measure of platelet

inhibition as an *ex-vivo* test or the maximum lowering of blood pressure
        - EC50 is the concentration of the drug at steady state that produces the half of the maximum effect
        - Hill coefficient is the slope of the relationship between drug concentration and drug effect. Hill coefficients above 2 indicate a steep relationship (i.e., small changes in concentration produce large changes in effect), and hill coefficients above 3 indicate an almost instantaneous "all or none" effect.
- Drugs produce their effects by interacting with biologic targets, but the time course of the pharmacodynamic effect is dependent on the mechanism and biochemical pathway of the target
    - Effects can be classified as direct or indirect and immediate or delayed
    - Direct effects are usually the result of drugs interacting with a receptor or enzyme that is central to the pathway of the effect
        - For example, Beta-blockers inhibit receptors that directly modulate cAMP levels in smooth muscle cells in the vasculature
    - Indirect effects are the result of drugs interacting with receptors, proteins of other biologic structures that significantly upstream from the end biochemical process that produces the drug effect
        - For example, Corticosteroids bind to nuclear transcription factors in the cell cytosol which translocate to the nucleus and inhibit transcription of DNA to mRNA encoding for several inflammatory proteins
    - Immediate effects are usually secondary to direct drug effects
        - For example, Neuromuscular blocking agents such as succinylcholine, which consists of two acetylcholine (ACh) molecules linked end to end by their acetyl groups, interact with the nicotinic acetylcholine receptor (nAChR) on skeletal muscle cells and leave the channel in an open state, resulting in membrane depolarization and generation of an action potential, muscle contraction and then paralysis within 60 seconds after administration
    - Delayed effects can be secondary to direct drug effects
        - For example, Chemotherapy agents which interfere with DNA synthesis, like cytosine arabinoside which is used in acute myeloid leukemia, produce bone marrow suppression that occurs several days after administration.
- Several important dosing principles that are based upon pharmacodynamics:
    - Kd: The pharmacologic response depends on the drug binding to its target as well as the concentration of the drug at the receptor site
        - Kd measures how tightly a drug binds to its receptor. Kd is defined as the ratio of rate constants for association (kon) and dissociation (koff) of the drug to and from the receptors

        - At equilibrium, the rate of receptor-drug complex formation is equal to the rate of dissociation into its components receptor + drug
        - The measurement of the reaction rate constants can be used to define an equilibrium or affinity constant (1/Kd)
        - The smaller the Kd value, the greater the affinity of the antibody for its target
        - For example, albuterol has a Kd of 100 nanomolar (nM) for the beta-2 receptor while erlotinib has a Kd of 0.35 nM for the estimated glomerular filtration rate (EGFR) receptor indicating that erlotinib has approximately 300 times the receptor interaction than albuterol
    - Receptor Occupancy: From the law of mass action the more receptors that are occupied by the drug, the greater the pharmacodynamic response; but all receptors do not need to be occupied in order to get a maximal response
        - This is the concept of spare receptors and occurs commonly to include muscarinic and nicotinic acetylcholine receptors, steroid receptors, and catecholamine receptors
        - Maximal effects are obtained by less than maximal receptor occupancy by signal amplification.
    - Receptor Up- and Downregulation: Chronic exposure of a receptor to an antagonist typically leads to upregulation, or an increased number of receptors, while chronic exposure of a receptor to an agonist causes downregulation, or a decreased number of receptors
        - Other mechanisms involving alteration of downstream receptor signaling may also be involved in up- or downmodulation without altering the receptor number on the cell membrane
        - For example, the insulin receptor undergoes downregulation to chronic exposure to insulin. The number of surface receptors for insulin is gradually reduced by receptor internalization and degradation brought about by increased hormonal binding
        - One exception to this rule is the receptor for nicotine that demonstrates upregulation in receptor numbers upon extended exposure to nicotine, despite nicotine being an agonist, which explains some of its addictive properties.
    - Effect compartment and indirect pharmacodynamics: A delay between the appearance of drug in the plasma and its intended effect may be due to multiple factors to include transfer into the tissue or cell compartment in the body or a requirement for the inhibition or stimulation of a signal to be cascaded through intracellular pathways
        - These effects can be described by either using an effect compartment or using indirect pharmacodynamic response models, which describe the effect of the drug through indirect mechanisms such as inhibition or stimulation of the production or elimination of endogenous cellular components that control the effect pathway

c. Know how to apply the principles of pharmacokinetics

- The main processes involved in pharmacokinetics are absorption, distribution, and the two routes of drug elimination, metabolism and excretion→Together they are sometimes known by the acronym 'ADME'→ study of how the body PROCESSES a drug
- A fundamental concept in pharmacokinetics is drug clearance, that is, elimination of drugs from the body, analogous to the concept of creatinine clearance. In clinical practice, clearance of a drug is rarely measured directly but is calculated as either of the following:

$$\text{clearance} = \text{dose}/\text{AUC} \quad \text{(equation 1)}$$

or

$$\text{clearance} = \text{infusion rate}/C_{ss} \quad \text{(equation 2)}.$$

  - AUC, the area under the curve, represents the total drug exposure integrated over time and is an important parameter for both pharmacokinetic and pharmacodynamic analyses. As indicated in equation 1, the clearance is simply the ratio of the dose to the AUC, so that the higher the AUC for a given dose, the lower the clearance. If a drug is administered by continuous infusion and a steady state is achieved, the clearance can be estimated from a single measurement of the plasma drug concentration (C_{ss}) as in equation 2.
  - Clearance can conceptually be considered to be a function of both distribution and elimination. In the simplest pharmacokinetic model,

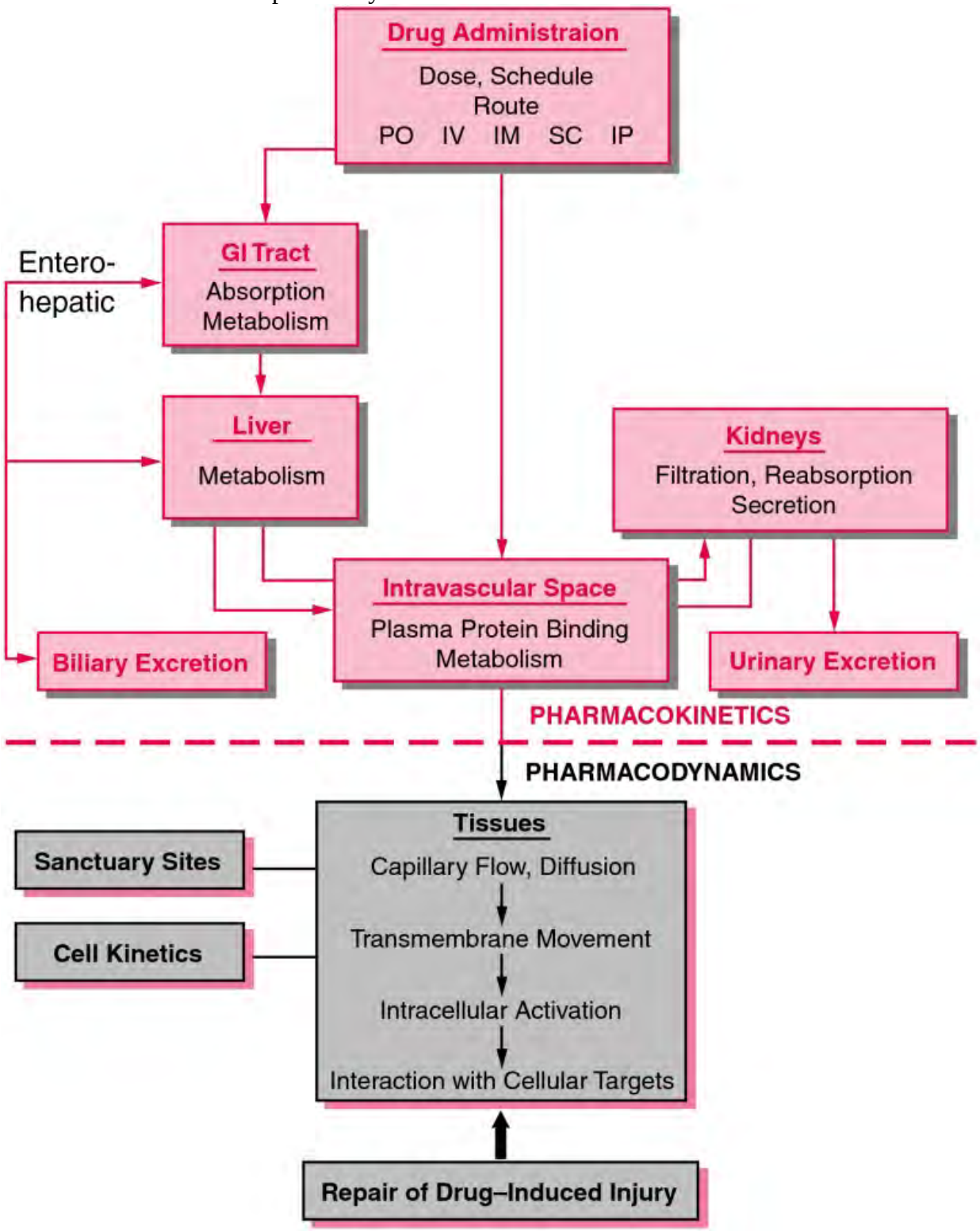
$$\text{clearance} = V\,K \quad \text{(equation 3)}.$$

  - V is the volume of distribution, and K is the elimination constant. V is the volume of fluid in which the dose is initially diluted, and thus the higher the V, the lower the initial concentration. K is the elimination constant, which is inversely proportional to the half-life, the period of time that must elapse to reach a 50% decrease in plasma concentration. When the half-life is short, K is high and plasma concentrations decline rapidly. Thus both a high V and a high K result in relatively low plasma concentrations and a high clearance.
- Although pharmacokinetic analysis can be conducted without specifying any mathematical models, it is helpful to use such models as guides in therapeutic decision making- linear versus nonlinear
  - Linear pharmacokinetic models
    - Characteristics of drugs with linear pharmacokinetics:

1. Half-life is independent of concentration
2. Clearance is independent of dose
3. Clearance is independent of schedule

  - Nonlinear pharmacokinetic models imply that some aspect of the pharmacokinetic behavior of the drug is saturable.
    - In contrast to the administration schedule of drugs with linear pharmacokinetics, alteration of the administration schedule of

drugs that display nonlinear kinetics may markedly affect the AUC and potentially alter clinical effects



d. Know how common psychotherapeutic agents are metabolized and excreted.

- Psychotropic drugs are removed from the body by two processes: metabolism, which occurs mainly in the liver, and excretion, the elimination of the drug from the body, which may be from the liver into bile and then the gut, or from the kidney
    - Lithium carbonate, being highly water- soluble, is a rare example of a psychotropic drug excreted unchanged by the kidneys.
    - Lorazepam excretion is mostly via the kidney, which makes it the benzodiazepine of choice in patients with liver impairment.
    - However, most psychotropic agents need to be converted to metabolites with more polar structures to facilitate excretion via the liver or kidney.
- Metabolism consists of two distinct phases:
    - Phase I involving oxidation, reduction or hydrolysis in the liver, and phase II conjugation, commonly with sulfate or glucuronic acid in the liver or gut wall.
    - Liver metabolism
        - Some drugs are metabolized in phase I to other known psychotropic agents (e.g. amitriptyline to nortriptyline, clomipramine to imipramine), while others have active metabolites which, although not prescribed in their own right, contribute to the therapeutic effect
        - Some compounds, such as the benzodiazepine clorazepate and the analgaesic agent tramadol, require liver metabolism in order to be effective, as the metabolite is the active compound, rather than the administered drug, which is referred to as a 'pro-drug'
        - Cytochrome P450 enzymes: phase I metabolism, which is effected by a series of hepatic enzymes known as cytochrome P450 (CYP) isozymes, is of considerable clinical relevance in psychotropic drug use
            - Drugs may be substrates, inhibitors or inducers of one or more P450 enzymes. Introducing or stopping a drug which is metabolized by, or otherwise influences the activity of, a particular enzyme, may have ramifications for the plasma concentrations of existing drugs.
            - Additionally, existing drugs may influence the eventual concentration of a newly introduced drug, with implications for efficacy, adverse effects and toxicity.
        - Substrates – a substrate is something that is changed by an enzyme. Many antidepressants, anxiolytics, hypnotics and antipsychotics are substrates of CYP2D6 (Table 1) and CYP3A4 (Table 2).
            - Other CYP enzymes metabolize psychotropic agents, notably CYP1A2 (olanzapine, clozapine) and CYP2C19 (involved in the metabolism of various anxiolytics and antidepressants including amitriptyline and diazepam)
            - Some drugs can be substrates for several CYP enzymes; for example, the demethylation of sertraline to its metabolite can be effected by six different CYP enzymes.
        - Inhibitors – an inhibitor interferes with the ability of an enzyme to effect metabolism, retarding the breakdown of co-prescribed drugs and thus potentially increasing their plasma levels
            - Paroxetine and fluoxetine are relatively strong inhibitors of CYP2D6
            - Duloxetine may also act as a moderately potent CYP2D6 inhibitor

- Fluoxetine also inhibits CYP3A4 and fluvoxamine inhibits CYP1A2
- When several substrates of the same enzyme are prescribed together, the metabolism of one or more drugs may also be inhibited, resulting in elevated plasma concentrations and possibly increased side effects.
- Plasma concentrations of some substrate drugs, such as aripiprazole, can be raised both by inhibitors of CYP2D6 and by CYP3A4 inhibitors.

▪ Inducers – an inducer speeds up enzyme activity, usually by causing the synthesis of greater amounts of enzyme, such that any co-prescribed drug metabolized by the same CYP enzyme will be broken down more rapidly
  - Carbamazepine, an antiepileptic drug with mood-stabilizing properties, is sometimes prescribed with antipsychotic agents when treating bipolar affective disorder
  - As a CYP3A4 inducer, carbamazapine can cause plasma concentration, and therefore the clinical effect, of existing antipsychotics to be reduced.
  - In epilepsy, a common comorbid condition in patients with learning disabilities who have psychiatric illness, the combination of CYP3A4-inducing antiepileptic agents (carbamazepine, phenytoin) with CYP3A4-inhibiting SSRI antidepressants (i.e. fluoxetine), may make plasma concentrations of both drugs, and therefore epileptic control, difficult to predict, especially as SSRIs can reduce seizure threshold.
  - Care should be taken in starting CYP3A4 inducers for patients taking oral contraceptive drugs which are CYP3A4 substrates – for example, the popular herbal remedy St John's wort has CYP3A4-inducing effects and may therefore compromise contraceptive efficacy.

▪ Genetic variations in metabolizer status:
  - a further point to note when considering CYP2D6 interactions is that 7% of the Caucasian population lack this important enzyme and are referred to as poor metabolizers→Such individuals may find standard tricyclic antidepressant doses intolerable but respond well to very low doses
  - In contrast, 'extensive metabolizer' status has been reported in up to 5 out of 100 people
  - CYP2C19 also has a genetic polymorphism of clinical significance, with the frequency of poor metabolizers among certain Asian populations being reported at up to 25%

- **Renal excretion**
  - Lithium is eliminated only by the kidneys. Like sodium, it is filtered by the glomerulus and 80% is reabsorbed by the proximal tubule, but it is not reabsorbed by the distal tubule. The intake of sodium and water are the principal determinants of its elimination. Any reduction in the rate of clearance of lithium can have profound clinical implications, since the range of plasma concentrations for therapeutic effect is relatively small but

concentrations slightly higher are associated with toxicity; thus it has a narrow therapeutic window.

- Reduced renal function or co-prescription of diuretics, especially thiazides, which induce sodium deficiency, can reduce lithium excretion significantly and thus precipitate toxicity.
- ACE inhibitors, angiotensin II-antagonists, and non-steroidal anti-inflammatory analgesics may also raise plasma lithium by interfering with excretion.

Psychotropic (and selected other) drugs known to be CYP3A4 substrates, inhibitors and inducers

CYP3A4 inhibitors

Antidepressants
Nefazodone
Fluoxetine

Other drugs
Cimetidine
Erythromycin
Ketoconazole (and grapefruit juice)

CYP3A4 substrates

Antidepressants
Fluoxetine
Sertraline
Amitriptyline
Imipramine
Nortriptyline
Trazodone

Anxiolytics, hypnotics and antipsychotics
Alprazolam
Aripiprazole
Buspirone
Diazepam
Midazolam
Triazolam
Zoplicone
Haloperidol
Quetiapine
Sertindole

Miscellaneous
Buprenorphine
Carbamazepine
Cortisol
Dexamethasone
Methodone
Testosterone
Calcium channel blockers
- Diltiazem
- Nifedipine
- Amlodipine

Other drugs
- Amiodarone
- Omeprazole
- Oral contraceptives
- Simvastatin

CYP3A4 inducers

Antidepressants
St John's wort

Miscellaneous
Carbamazepine
Phenobarbitone
Phenytoin

Psychotropic (and selected other) drugs known to be CYP2D6 substrates, inhibitors and inducers

CYP2D6 inhibitors

Antidepressants

Paroxetine
Fluoxetine
Duloxetine

CYP2D6 substrates

Antidepressants

Paroxetine
Fluoxetine
Citalopram
Sertraline
Venlafaxine
Amitriptyline
Clomipramine
Desipramine
Imipramine
Nortriptyline

Antipsychotics

Aripiprazole
Chlorpromazine
Haloperidol
Thioridazine
Zuclopenthixol
Perphenazine
Risperidone

Miscellaneous

Buproprion
β-blockers
- Propanolol
- Metoprolol
- Timolol
- Bufaralol

Dexfenfluramine
Ecstasy
Opioids
- Codeine
- Hydrocodone
- Dihydrocodeine
- Tramadolol
- Ethyl morphine

e. Understand the issues related to the combined use of psychopharmacologic agents.

- *Psychiatric polypharmacy refers to the prescription of two or more psychiatric medications concurrently to a patient.*
- *It can be categorized as same-class, multi-class, adjunctive, augmentation and total polypharmacy.*
- Despite the increasing use of polypharmacy in children, there is not yet a uniform definition of polypharmacy in pediatric patients
- polypharmacy is increasingly acknowledged as a common concern in pediatric patients including both potential benefits such as control of complex or multiple disease conditions and harms such as adverse drug effects, drug-to-drug interaction, hospitalization, poor medication adherence, mortality, resource wastage, burden of medical care, and high cost of healthcare

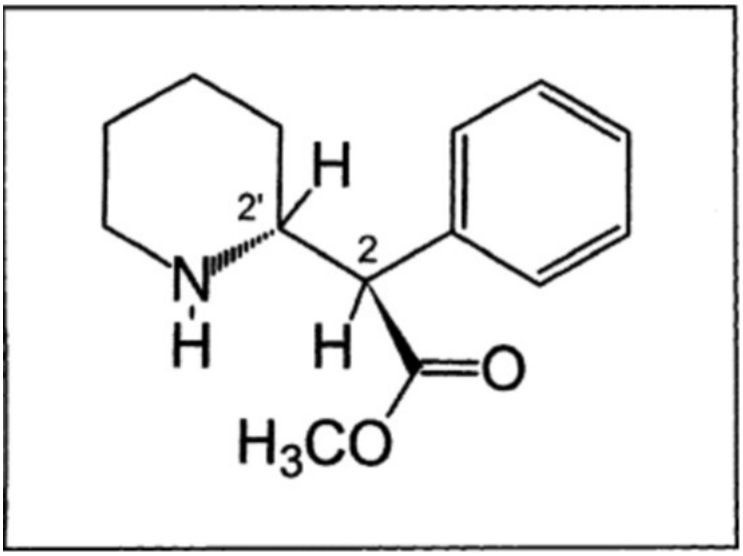
- There is limited information about the actual rates of psychotropic polypharmacy in the pediatric population. However, the data that are available demonstrate that this practice is on the rise.
- *Dealing with polypharmacy requires an understanding of its associated factors.*
- In patients with multiple diagnoses or one diagnosis refractory to monotherapy, polypharmacy may be warranted.
- *Education, guidelines and algorithms for the appropriate management of various conditions are effective ways to avoid irrational polypharmacy*.

f. Understand the pharmacodynamic and pharmacokinetic properties of stimulant medications.

- Simplest way to break this down is by considering the two most common types of stimulant medications utilized in pediatric populations- methylphenidate and amphetamines with focus primarily on long-acting stimulant formulations
- A number of long-acting stimulant formulations have been developed with the aim of providing once-daily dosing, employing various means to extend duration of action, including a transdermal delivery system, an osmotic-release oral system, capsules with a mixture of immediate- and delayed-release beads, and prodrug technology.
- Coefficients of variance of pharmacokinetic measures can estimate the levels of pharmacokinetic variability based on the measurable variance between different individuals receiving the same dose of stimulant (interindividual variability) and within the same individual over multiple administrations (intraindividual variability).
- Differences in formulation clearly impact pharmacokinetic profiles.
- Many medications exhibit wide interindividual variability in clinical response. Stimulants with low levels of inter- and intraindividual variability may be better suited to provide consistent levels of medication to patients.
- The pharmacokinetic profile of stimulants using pH-dependent bead technology can vary depending on food consumption or concomitant administration of medications that alter gastric pH.
- While delivery of methylphenidate with the transdermal delivery system would be unaffected by gastrointestinal factors, intersubject variability is nonetheless substantial.
- Unlike the beaded formulations and, to some extent (when considering total exposure) the osmotic release formulation, systemic exposure to amphetamine with the prodrug stimulant lisdexamfetamine dimesylate appears largely unaffected by such factors, likely owing to its dependence on systemic enzymatic cleavage of the precursor molecule, which occurs primarily in the blood involving red blood cells.
    - The high capacity but as yet unidentified enzymatic system for conversion of lisdexamfetamine dimesylate may contribute to its consistent pharmacokinetic profile.
- The reasons underlying observed differential responses to stimulants are likely to be multifactorial, including pharmacodynamic factors.
- While the use of stimulants with low inter- and intrapatient pharmacokinetic variability does not obviate the need to titrate stimulant doses, stimulants with

low intraindividual variation in pharmacokinetic parameters may reduce the likelihood of patients falling into subtherapeutic drug concentrations or reaching drug concentrations at which the risk of adverse events increases.

- Methylphenidate
  - Methylphenidate is a piperidine-derived central nervous system stimulant. The methylphenidate molecule possesses 2 centers of chirality; thus, a total of 4 enantiomers of this drug exist



  - Methylphenidate increases brain dopamine and norepinephrine levels by blocking the presynaptic reuptake transporters
  - Methylphenidate inhibits the dopamine (DA) and norepinephrine (NE) transporters (DAT and NET), thereby elevating DA and NE levels in the brain
  - Due to pharmacokinetic differences, many products are not bioequivalent and not interchangeable on a mg per mg basis; when available, dosing conversion information is provided→dose titration should be optimized for efficacy and tolerability
  - Short-acting methylphenidate (e.g., Ritalin, Methylin) is available as a tablet, chewable tablet, or liquid
    - Behavioral effects may be seen within 30 minutes
    - The duration of action is three to five hours and the half-life is two to three hours
  - Long-acting methylphenidate is available in tablets, chewable tablets, orally disintegrating tablets, capsules, oral suspension, and a patch
    - The onset and duration of action vary depending upon the formulation
      - Single pulse wax matrix sustained release tablets (e.g., Metadate-ER) have onset of action within 20 to 60 minutes and last up to 8 hours.
      - Osmotic release methylphenidate tablets (Concerta) are coated with immediate release methylphenidate (for initial dosing) and use an osmotic pump to gradually release

methylphenidate to approximate a three-times-per-day dosing schedule. Osmotic release methylphenidate usually has onset of action within 20 to 60 minutes and lasts up to 12 hours. Osmotic release methylphenidate tablets must be swallowed whole.

- Extended release orally disintegrating tablets (Cotempla XR-ODT) contain a mixture of immediate release and extended release methylphenidate
    - They have onset of action within one hour and last up to 12 hours
    - Although the prescribing information indicates that extended release orally disintegrating tablets should be taken consistently with or without food, we suggest that they be taken consistently with food – given the general stimulant effect of appetite suppression
- Sustained release capsules (e.g., Aptensio XR, Focalin-XR, Metadate CD, Ritalin-LA, Adhansia XR) contain a mixture (usually one-to-one) of immediate release and enteric-coated delayed release beads (or pearls) and generally approximate a twice-per-day dosing schedule
    - They typically have onset of action within 20 to 60 minutes and may last up to 12 hours (up to 16 hours for Adhansia XR)
    - Extended release methylphenidate capsules (Jornay PM) contain beads within two film coatings: an outer coating that delays release for approximately 10 hours and an inner coating that controls release throughout the day
    - Sustained release stimulant capsules can be opened and sprinkled on soft foods.
- The oral suspension (Quillivant XR) and chewable tablet (Quillichew ER) are a racemic mixture of the d- and l-isomers of methylphenidate; the d-isomer is more CNS active. It has onset of action within one hour and lasts as long as 8 hours (Quillichew ER) or 12 hours (Quillivant XR)
- The methylphenidate patch (Daytrana) delivers methylphenidate transdermally
    - The methylphenidate patch has onset of action of 60 minutes. The duration of action may be as long as 12 hours but can be controlled by early removal of the patch (the effects last approximately two to three hours after the patch is removed)
    - The methylphenidate patch should not be cut; if a lower dose is needed a smaller patch should be prescribed.
- The dose-response rate of methylphenidate is highly variable, and careful dose titration is necessary within the limitations of available formulations

        - We do not suggest weight-based dosing, but as a general guide, significant reduction in core symptoms (ie, hyperactivity, impulsivity, and inattention) typically occurs at doses between 0.3 and 0.6 mg/kg
        - Dexmethylphenidate (the d-enantiomer of methylphenidate) is generally efficacious at doses approximately one-half of those needed for methylphenidate
- Amphetamines
    - can be prescribed as a single salt (dextroamphetamine, amphetamine sulfate) or as mixed dextroamphetamine-amphetamine salts.
    - Lisdexamfetamine is a prodrug of dextroamphetamine that is pharmacologically activated after oral ingestion and was designed to discourage drug misuse
    - Amphetamine medications for ADHD are available in immediate and sustained release preparations
    - The immediate release preparation of dextroamphetamine-amphetamine (Adderall [brand name]) has an onset of action of 20 to 60 minutes and a reported duration of up to 6 hours
    - Longer-acting preparations (6 to 13 hours) of amphetamine and dextroamphetamine-amphetamine are available as capsules, a liquid suspension, orally disintegrating tablets, or chewable tablets
        - The onset and duration of action vary depending upon the formulation, typically ranging from 20 to 60 minutes for onset of action and at least 10 hours' duration
    - Sustained release capsules (e.g., Adderall XR, Mydayis) contain a mixture of immediate release and enteric-coated delayed release beads (or pearls) and approximate a twice-per-day or three-times per day dosing schedule →they typically have onset of action within 20 to 60 minutes and last up to 10 hours (Adderall XR and generic) or 16 hours (Mydayis). Sustained release amphetamine capsules can be opened and sprinkled on soft foods.
    - Lisdexamfetamine (Vyvanse) capsules or chewable tablets have onset of action within 60 minutes and lasts up to 10 hours.
    - Note on liquid suspension formulations of amphetamines
        - The liquid suspension of Dyanavel extended release amphetamine has an onset of action within 60 minutes and lasts up to 13 hours
        - The pharmacokinetics of the liquid suspension of Adzenys extended release amphetamine suggest an onset of action within 60 minutes and duration of action of approximately 10 hours →these liquid suspensions should not be mixed with food or other liquids before administration.
    - The pharmacokinetics of extended release orally disintegrating amphetamine (Adzenys XR-ODT) suggest an onset of action within 60 minutes and duration of approximately 10 hours.

g. Know indications for the use of stimulant medications

- Stimulants are recommended as first-line treatment when considering initiation of pharmacologic therapy for children and adolescents with diagnosis of ADHD
    - Dosage should be individualized; a discontinuation trial after 6 months of therapy is also recommended to reassess underlying psychopathology (AACAP [Pliszka 2007]; AAP 2011; Cortese 2018; NICE 2018)
    - Also recommended to discontinue medication if no improvement is seen after appropriate dosage adjustment and compliance over a 1-month period of time
- Symptomatic management of narcolepsy and daytime sleepiness- approved as labeled indications for certain formulations of methylphenidate (Methylin, Metadate ER, Ritalin) and for immediate release formulations of dextroamphetamine and amphetamine (Adderall) and dextroamphetamine (dexedrine)

h.Understand the clinical use of stimulant medications.- See g. above

i. Know the side effects and appropriate monitoring of stimulant medications.

- General adverse effects — Many of the side effects of stimulants are mild, of short duration, and reversible with adjustments to the dose or dosing interval
    - The frequency of most side effects is similar with methylphenidate and amphetamines. However, treatment with mixed dextroamphetamine-amphetamine salts may be associated with greater decrease in weight over time and increased risk of irritability than treatment with methylphenidate
    - Side effects may occur more frequently in preschool children than in older children
- Relatively common side effects include anorexia, poor growth or weight loss, sleep disturbance, jitteriness, and emotional lability (e.g., social withdrawal)
- Deceleration of linear growth may occur but appears to attenuate over time;
    - cessation of treatment may result in normalization of growth, and adult height does not appear to be affected
    - Growth should be regularly monitored during treatment with stimulants

- Less common side effects include increased heart rate and blood pressure, headache, dizziness, gastrointestinal symptoms, priapism, and peripheral vasculopathy, including Raynaud phenomenon
    - Blood pressure and heart rate should be monitored before and during treatment with stimulants
- Patients treated with the methylphenidate patch may develop contact sensitization if the patch is worn in the same location every day
    - The methylphenidate patch has also been associated with permanent loss of skin color (chemical leukoderma)

- Priapism —
    - Priapism is a rare complication of methylphenidate stimulants
        - Most of the cases occurred in boys <18 years (median age 12.5 years, range 8 to 33 years); two cases required surgical treatment.

        - Priapism occurred in a variety of settings including increased dose, longer than typical dosing interval, and temporary or permanent discontinuation of methylphenidate stimulants.
    - Priapism has also been reported among four patients taking amphetamine stimulants for ADHD
        - However, the correlation between amphetamine stimulants and priapism is uncertain because these patients were also taking other medications associated with priapism.

- Psychiatric effects
    - **Psychosis** – Children and adolescents treated with stimulant medications **rarely** may develop psychotic symptoms (e.g., hallucinations, delusional thinking, or mania), but causality has not been established
        - In pooled analysis of data from 49 randomized trials of medications used to treat ADHD (35 of which evaluated stimulants), psychotic symptoms developed in 11 children during 743 person-years of follow-up (incidence rate of 1.48 episodes per 100 person-years)
        - In subsequent analysis of two commercial insurance claims databases that evaluated psychosis requiring treatment with antipsychotic medication after initiation of stimulant therapy in 221,846 adolescents and young adults (age 13 to 25 years), 343 episodes occurred in 143,286 person-years of follow-up (incidence rate of 2.4 episodes per 1000 person-years)
        - The median time between initiation of stimulant and the psychotic episode was 128 days
        - The absolute risk was higher with amphetamines than with methylphenidate (2.83 versus 1.78 episodes per 1000 person-years, hazard ratio 1.7, 95% CI 1.3-2.1)
        - Although it is not possible to predict which patients will develop psychotic symptoms after initiation of stimulants, the risk may be increased in children with a family history of mental illness (e.g., major depressive disorder, bipolar disorder)
    - **Suicidal thinking** – Rare cases of suicidal thinking have been reported among individuals taking stimulants for ADHD, but causality has not been established
- Tics — Stimulant medications have been reported to cause new onset of tics or worsening of tics in children with tic disorders
    - Tics or a family history of tics are listed as a contraindication to some forms of methylphenidate and a significant adverse effect of both methylphenidate and amphetamines.
    - Nonetheless, stimulant medications often improve attention and behavior without worsening tics in children who have chronic tics or Tourette syndrome
    - ADHD and tic disorders frequently coexist: Approximately 20 percent of children with ADHD develop chronic tic disorders and approximately 50

percent of children with chronic tics or Tourette syndrome have comorbid ADHD.
    - Given the frequency of comorbid tic disorders and ADHD and the typical waxing and waning pattern of tics, new or worsening tics in children who receive stimulant medications may be coincidentally rather than causally associated
    - Most up to date evidence out there has found that risk of new or worsening tics was not associated with stimulant formulation, dose, duration of treatment, or patient age
        - Have been a few meta-analyses that found that dextroamphetamine specifically, however, was associated with exacerbation of tics when prescribed at higher than usual recommended doses→could suggest avoidance of higher-than-usual doses of dextroamphetamine
    - For children in whom stimulant medications were discontinued because of new or worsening tics, rechallenge with stimulants may be warranted, particularly if the behavioral response to nonstimulant medications was inferior to that with stimulants.
- Diversion and misuse —
    - Stimulant diversion consists of the transfer of medication from the patient for whom it was prescribed to another individual
    - Stimulant misuse consists of taking higher doses of medication than prescribed to achieve euphoria or combining stimulant medications with illicit drugs or alcohol
    - A systematic review of studies related to diversion and misuse of ADHD medications indicated that 5 to 9 percent of grade- and high-school-age students and 5 to 35 percent of college-age individuals reported nonprescribed stimulant use in the year before the study
        - The proportion of students with stimulant prescriptions who were ever asked to give, sell, or trade their medications ranged from 16 to 29 percent.
        - Diversion and misuse were more common among whites, members of fraternities and sororities, students with lower grade point averages, and students who report ADHD symptoms.
        - Diversion and misuse also was more common with immediate than extended release preparations.
        - The most commonly reported reasons for stimulant diversion and misuse included studying, staying awake, improved alertness, experimenting, and "getting high."
        - One survey found that ADHD subjects who diverted or misused their medication were more likely to have a comorbid conduct disorder or substance use disorder
- Contraindications to stimulants — Contraindications to stimulant medications may include:
    - Symptomatic cardiovascular disease
    - Moderate to severe hypertension

- Hyperthyroidism
- Known hypersensitivity or idiosyncrasy to sympathomimetic amines
- Motor tics or Tourette syndrome
- Glaucoma
- Agitated states
- Anxiety
- History of drug abuse
- Concurrent use or use within 14 days of the administration of monoamine oxidase inhibitors

j. Know indications for the use of selective serotonin reuptake inhibitors

- Labeled indications for use of SSRI's in pediatric populations includes depression, anxiety, and OCD
- For children and adolescents with acute depressive disorders, first line pharmacotherapy is fluoxetine.
  - There is more consistent, high quality evidence for the efficacy of fluoxetine than other antidepressants
- Acute pediatric depressive episodes do not remit with fluoxetine in approximately 30 percent of patients
  - Sertraline could be considered next best option based upon efficacy in medical literature, but escitalopram or citalopram are reasonable based upon current evidence from randomized trials on efficacy in pediatric populations as well
  - Another reasonable alternative is venlafaxine (SNRI) which appears to be comparable to SSRIs in treatment resistant patients
  - Paroxetine less of an option for pediatric depression because of its lack of demonstrated efficacy in randomized trials

k. Understand the clinical use of selective serotonin reuptake inhibitors –

- Major Depression (see above) – CBT plus SSRI considered first line treatment for children/adolescents with concurrent major depression and anxiety disorder
- Generalized anxiety disorder
- Social anxiety disorder
- Selective mutism
- Panic disorder — Specific phobias

l. Know the side effects and appropriate monitoring of selective serotonin reuptake inhibitors

- The risks associated with SSRIs for pediatric anxiety and depression should be carefully weighed against their potential benefits whenever the use of these medications is considered.
- Risks and benefits should be discussed with both the parents and child before initiating treatment.
- SSRIs have been associated with psychiatric adverse events, such as disinhibition, agitation, and worsening of anxiety symptoms.

- Physical side effects most commonly include headaches, gastric distress, and sleep disturbance
- Antidepressant medications are associated with an increased risk of suicidal thinking and behavior in children and adolescents
    - The US Food and Drug Administration issued a "black box" warning in 2004, stating that children and adolescents taking antidepressant medication, including SSRIs and TCAs, are at increased risk for suicidal thinking or behavior

- Some children receiving venlafaxine have been reported to experience weight gain, elevated cholesterol, and hypertension

Selective serotonin reuptake inhibitors (SSRIs)*				
Fluoxetine	Children: 5 to 10 mg Adolescents: 10 mg	After 7 days increase daily dose to 20 mg; then after 4 and 8 weeks increase daily dose by 20 mg, if needed	10 to 80 mg	▪ Prolonged half-life. ▪ Metabolized by and inhibits CYP2D6.
Fluvoxamine	25 to 50 mg at bedtime	Increase daily dose by 25 mg (child) or 25 to 50 mg (adolescent) after a minimum of 7 days, if needed	50 to 300 mg	▪ Girls generally require lower maintenance doses than boys. ▪ Give with meals and bedtime in divided doses to minimize side effects. ▪ Metabolized by CYP1A2 and 2D6. ▪ Inhibits CYP1A2 and 2C19.
Sertraline	12.5 to 25 mg	Increase daily dose by 12.5 mg (child) or 25 to 50 mg (adolescent) after a minimum of 7 days, if needed	50 to 200 mg	▪ Diarrhea more frequent than other SSRIs. ▪ Metabolized by CYP2D6. ▪ Inhibits CYP2D6 with larger doses.
Paroxetine	5 to 10 mg	Increase daily dose by 5 mg (child) or 10 mg (adolescent) after a minimum of 7 days, if needed	10 to 60 mg	▪ Short half-life. ▪ Mild anticholinergic side effects. ▪ Metabolized by and inhibits CYP2D6. ▪ Weight gain.

m. Know indications for the use of mood stabilizers.

- The oldest and most studied of mood stabilizers is lithium
- However, many drugs that were first developed as anticonvulsants to treat epilepsy also act as mood stabilizers
    - These include carbamazepine, oxcarbazepine, divalproex and lamotrigine.
    - Anticonvulsants (e.g., valproate, gabapentin, lamotrigine, oxcarbazepine) may be indicated for mood stabilization in children with ASD, for example, if there is evidence of mood instability and a mood disorder (e.g., bipolar disorder) despite educational and behavioral interventions and +/- other pharmacologic medication trials
- While atypical antipsychotics are not often considered "classic" mood stabilizing medications, they are commonly used in the DBP patient for this primary purpose or as an adjunct with other mood stabilizers (i.e. with lithium cases of refractory mania)

- Indications for use of atypical antipsychotics in pediatric populations:
    - Autism- treatment of associated irritability (including aggression, deliberate self-injurious behavior, temper tantrums, and quickly changing moods)
        - Risperidone and aripiprazole are the only psychotropic medications approved by the FDA specifically for treatment of individuals with ASD [11]. However, many other medications are

used off-label. Parents and caregivers should be informed if the medication is being used off-label.
    - Bipolar I disorder (acute manic or mixed episodes)
    - Disruptive behavior disorders (e.g., conduct disorder, oppositional defiant disorder); aggression
    - Schizophrenia (adolescents)
    - Tourette syndrome, tic disorders (>6 yo)
    - ADHD (listed as labeled indication for Abilify specifically- limited data available on efficacy)

n. Understand the clinical use of common mood stabilizing medications.

- ASD is one of the common clinical uses for mood stabilizing medications in the DBP world
- Some of the most common clinical uses for mood stabilizing medications specifically in children with ASD can include:
    - Maladaptive/problem behaviors —
        - Maladaptive behaviors in children with ASD include irritability, aggression, explosive outbursts (tantrums), and self-injury.
        - These behaviors may occur in response to anxiety or frustration, which should be the first targets of management
        - Maladaptive behaviors also can be due to anxiety, mood disorders, or impulse control problems; if one of these conditions is identified as a cause for the behavior, medications that target that symptom should be used of course and this can be a difficult clinical management question to answer
        - Potential agents
            - The atypical antipsychotic agents, risperidone and aripiprazole, are the only medications approved by the US Food and Drug Administration (FDA) to treat irritability and self-injurious and aggressive behaviors in children with ASD
            - Risperidone — Risperidone is the most commonly used antipsychotic for the treatment of maladaptive behaviors in children with ASD → approved by the FDA for the treatment of irritability presenting with aggression, tantrums, and/or deliberate self-injury in children (≥5 years) with ASD
            - Aripiprazole is approved by the FDA for the treatment of irritability in children (aged 6 to 17 years) with ASD
            - Other atypical antipsychotics can be used, but are technically prescribed used off-label (olanzapine, clozapine, quetiapine, ziprasidone)
                - Olanzapine — Several small prospective studies (only one of which was blinded) demonstrated clinical improvement in disruptive behaviors in

children with ASD who were treated with olanzapine (an atypical antipsychotic)→However, weight gain and other side effects, such as sedation, often prohibit continued use of olanzapine; the risk of extrapyramidal side effects with olanzapine appears to be low

- Strong evidence that risperidone and aripiprazole are beneficial for disruptive behaviors and only marginal evidence for benefit with other atypical antipsychotics

- Repetitive behaviors and rigidity — Repetitive behaviors, stereotypies, and rigidity in children with ASD often interfere with function

  - Repetitive behaviors and rigidity are core symptoms of ASD.
  - They also can be exacerbated by anxiety or other conditions.
  - Medication may be warranted if the behaviors interfere with function and have not responded adequately to non-pharmacologic interventions (e.g., behavior therapy).
  - Pharmacologic treatment involves a tradeoff between benefits and risks
  - The literature is limited by the lack of tools that specifically measure stereotypies and rigidity. Many studies measure obsessive-compulsive behaviors, which may not be the best proxy for stereotypies and rigidity in children with ASD.

o. Know the side effects of common mood stabilizing medications.

- The use of antipsychotic agents often is limited by their side effect profile. Side effects of atypical antipsychotics may include, but are not limited to:
  - Increased appetite
  - Weight gain
  - Elevated blood sugar secondary to insulin resistance
  - Dyslipidemia
  - Blood pressure changes
  - Electrocardiogram (EKG) changes, such as prolongation of QTc (more commonly seen with ziprasidone)
  - Fatigue and drowsiness
  - Dizziness
  - Drooling
  - Liver function abnormalities
  - Increase in prolactin (of unknown clinical significance)
  - Gynecomastia
  - Less common, but serious, side effects include dystonic reactions, tardive dyskinesia, akathisia (subjective sense of restlessness, often accompanied

by voluntary movements of the limbs or trunk), neuroleptic malignant syndrome, and agranulocytosis (with clozapine)

- Children who are treated with atypical antipsychotic agents should be monitored regularly
    - At baseline, the child's weight and height should be measured and body mass calculated as well
    - Given the cardiometabolic side effects, it is important to take a thorough patient and family history, asking specifically about obesity, diabetes, dyslipidemia, hypertension, and cardiovascular disease.
    - A baseline EKG should be obtained; the EKG should be repeated when the patient has reached the steady state (particularly in patients treated with ziprasidone).
- Follow-up visits should include measurement of weight and blood pressure and evaluation for extrapyramidal findings with the use of a scale, such as the Abnormal Involuntary Movement Scale (see below)
    - Baseline and follow-up laboratory studies may include fasting plasma glucose, fasting lipids, complete blood count, liver function tests, thyroid stimulating hormone (TSH), prolactin, and electrolytes.
    - The optimal frequency of laboratory monitoring is not clear
        - Suggested from literature is follow-up at three months after starting medication and every six months thereafter for most of the studies listed above and when doses are increased.
        - Prolactin and TSH may be monitored less frequently unless symptoms of abnormalities arise.
        - More frequent testing may be necessary if metabolic abnormalities are identified

ABNORMAL INVOLUNTARY MOVEMENT SCALE (AIMS)

Public Health Service
Alcohol, Drug Abuse, and Mental Health Administration
National Institute of Mental Health

NAME: _____
DATE: _____
Prescribing Practitioner: _____

CODE 0=None
1=Minimal, may be extreme normal
2=Mild
3=Moderate
4-Severe

INSTRUCTIONS:
Complete Examination procedure (attachment d.)
Before making ratings

MOVEMENT RATINGS: Rate highest severity observed. Rate movements that occur upon activation one less than those observed spontaneously. Select movement as well as code number that applies.		RATER Date	RATER Date	RATER Date	RATER Date
Facial and Oral Movements	**1. Muscles of Facial Expression** e.g. movements of forehead, eyebrows, periorbital area, cheeks, including frowning, blinking, smiling, grimacing	0 1 2 3 4	0 1 2 3 4	0 1 2 3 4	0 1 2 3 4
	2. Lips and Perioral Area e.g., puckering, pouting, smacking	0 1 2 3 4	0 1 2 3 4	0 1 2 3 4	0 1 2 3 4
	3. Jaw e.g. biting, clenching, chewing, mouth opening, lateral movement	0 1 2 3 4	0 1 2 3 4	0 1 2 3 4	0 1 2 3 4
	4. Tongue Rate only increases in movement both in and out of mouth. NOT inability to sustain movement. Darting in and out of mouth.	0 1 2 3 4	0 1 2 3 4	0 1 2 3 4	0 1 2 3 4
Extremity Movements	**5. Upper (arms, wrists,, hands, fingers)** Include choreic movements (i.e., rapid, objectively purposeless, irregular, spontaneous) athetoid movements (i.e., slow, irregular, complex, serpentine). DO NOT INCLUDE TREMOR (i.e., repetitive, regular, rhythmic)	0 1 2 3 4	0 1 2 3 4	0 1 2 3 4	0 1 2 3 4
	6. Lower (legs, knees, ankles, toes) e.g., lateral knee movement, foot tapping, heel dropping, foot squirming, inversion and eversion of foot.	0 1 2 3 4	0 1 2 3 4	0 1 2 3 4	0 1 2 3 4
Trunk Movements	**7. Neck, shoulders, hips** e.g., rocking, twisting, squirming, pelvic gyrations	0 1 2 3 4	0 1 2 3 4	0 1 2 3 4	0 1 2 3 4
Global Judgments	**8. Severity of abnormal movements overall**	0 1 2 3 4	0 1 2 3 4	0 1 2 3 4	0 1 2 3 4
	9. Incapacitation due to abnormal movements	0 1 2 3 4	0 1 2 3 4	0 1 2 3 4	0 1 2 3 4
	10. Patient's awareness of abnormal movements Rate only patient's report No awareness 0 Aware, no distress 1 Aware, mild distress 2 Aware, moderate distress 3 Aware, severe distress 4	0 1 2 3 4	0 1 2 3 4	0 1 2 3 4	0 1 2 3 4
Dental Status	**11. Current problems with teeth and/or dentures?**	No Yes	No Yes	No Yes	No Yes
	12. Are dentures usually worn?	No Yes	No Yes	No Yes	No Yes
	13. Edentia?	No Yes	No Yes	No Yes	No Yes
	14. Do movements disappear in sleep?	No Yes	No Yes	No Yes	No Yes

p. Know indications for the use of tricyclic antidepressants.

- Tricyclic antidepressants inhibit the reuptake of norepinephrine and serotonin→its these actions that dictate the potential clinical uses and indications for TCA's in the DBP patient
- Major depressive disorder
    - Tricyclics are rarely indicated for depressed youth because of a frequent lack of efficacy, an unfavorable side effect profile, high lethality in overdose, and the availability of SSRIs
    - However, it is reasonable to administer tricyclics in patients who do not respond to multiple trials of SSRIs, serotonin-norepinephrine reuptake inhibitors, and other antidepressants
- Anxiety disorders- TCAs not considered in first- or second-line treatment of anxiety disorders in children, because of the limited support from clinical trials and side effect profile that is typically less well-tolerated than SSRIs
- Chronic pain management- limited data available for efficacy- amitriptyline
- Migraine prophylaxis- amitriptyline
- Obsessive-compulsive disorder (OCD), treatment- clomipramine
- ADHD- specifically imipramine, nortriptyline - tricyclic antidepressants inhibit the reuptake of norepinephrine and serotonin. With the availability additional nonstimulant medications, tricyclic antidepressants usually are reserved for children and adolescents who respond poorly to a trial of stimulants, atomoxetine, or alpha-2-adrenergic agonists; have unacceptable side effects with these medications; or have significant comorbid conditions
- Enuresis- imipramine, nortriptyline
- Neuropathic pain (limited data)- nortriptyline
- Insomnia- most TCAs are sedating; the most sedating drugs are amitriptyline, doxepin, and trimipramine.
    - TCAs decrease SOL and arousals and have been used to treat insomnia in adults who have underlying depression.
    - Most of these medications have not been studied for insomnia in patients without underlying depression

q. Understand the clinical use of tricyclic antidepressants. – see p. above

r. Know the side effects and appropriate monitoring of tricyclic antidepressants.

- Cyclic antidepressants tend to have dose-related side effects at therapeutic doses
- These antidepressants are "broad spectrum" in that they interact with many neurotransmitter systems, which is the basis for both their antidepressant efficacy and their side effects
    - Tricyclics block muscarinic M1, histamine H1, and alpha-adrenergic receptors, and commonly cause cardiac effects, anticholinergic effects, antihistaminic effects, decreased seizure threshold, sexual dysfunction, diaphoresis, and tremor.
    - In addition, cyclic antidepressants are dangerous in overdose.
    - The side effects of tricyclic and tetracyclic antidepressants generally make them less tolerable compared with selective serotonin reuptake inhibitors (SSRIs) and other newer antidepressants

- Cardiac — All of the cyclic antidepressants are potentially cardiotoxic and should be avoided in susceptible individuals with history of heart disease
    - At therapeutic serum levels, tricyclic antidepressants can cause orthostatic hypotension, which is one of the most common reasons for discontinuing them
        - Orthostasis is most likely to occur in patients with preexisting postural hypotension, is aggravated by concurrent antihypertensive medications and dehydration, and often occurs at low tricyclic serum levels.
        - Blockade of alpha-adrenergic receptors is the primary cause.
    - Tricyclic antidepressants can cause electrocardiogram changes, benign or otherwise
        - The primary concern is prolongation of the QT interval, particularly when tricyclics are co-administered with other medications (e.g., other psychotropic medications, certain antimicrobials, and antiarrhythmic drugs) that can prolong the QT interval
        - Patients with acquired long QT syndrome are at risk for ventricular arrhythmias, most notably polymorphic ventricular tachycardia, which may result in sudden cardiac arrest.
- Seizures — All of the cyclic antidepressants can lower seizure threshold. Seizures are directly related to dose and serum level and are thus more likely to occur at higher doses (and in overdoses)
- Bone fractures — Observational studies have found an association between tricyclic use and bone fractures
- Anticholinergic — The tricyclics block muscarinic acetylcholine receptors and cause anticholinergic effects such as blurred vision, constipation, dry mouth (which may lead to dental caries), and urinary retention
    - In addition, these anticholinergic effects can cause tachycardia, ocular crisis in patients with narrow-angle glaucoma, and confusion and delirium.
- Antihistaminic — The cyclic antidepressants block histamine receptors and cause sedation, increased appetite leading to weight gain, confusion, and delirium
    - The sedative properties are sometimes harnessed for patients with insomnia, but more benign options are available.
- Other side effects — The cyclic antidepressants can cause a number of other side effects at therapeutic doses, most of which are dose dependent:
    - Sexual dysfunction including impaired arousal (especially in men) and orgasm→ occurs less often with tricyclics compared with SSRIs
    - Diaphoresis appears to be related to noradrenergic effects.
    - Tremor.
    - Acute hepatitis appears to be an allergic reaction that is uncommon but dangerous and potentially fatal
        - Elevated liver function tests should be followed with further tests for a few days
        - Hepatitis is reversible if the medication is discontinued.

s. Know indications for alpha-adrenergic agonists.

- ADHD:
    - Alpha-2-adrenergic agonists (e.g., extended release clonidine or guanfacine) usually are reserved for children and adolescents who respond poorly to a trial of stimulants or atomoxetine, have unacceptable side effects with stimulants or atomoxetine, or have significant comorbid conditions.
    - Alpha-2-adrenergic agonists may take up to two weeks for initial response (compared with 20 minutes to a few hours for stimulants) and there are fewer data regarding their efficacy than for stimulants and atomoxetine
    - Clonidine may be useful in overaroused, easily frustrated, highly active, or aggressive individuals
    - Extended release guanfacine (Intuniv) can be used for once-daily treatment of ADHD or as an adjunct to stimulant therapy in children and adolescents aged 6 to 17 years→Guanfacine has a longer half-life and fewer side effects than does clonidine
- Comorbid ASD and ADHD
- ODD and/or CD with or without comorbid ADHD
- Tic Disorders/Tourette's
    - The alpha adrenergic agonists guanfacine and clonidine are effective for treating the symptoms of ADHD and may be helpful in patients with TS who have ADHD or predominant behavioral symptoms, particularly impulse control problems and rage attacks
- Insomnia:
    - Alpha-adrenergic agonists (especially clonidine) are commonly prescribed to treat childhood insomnia.
    - Anecdotal clinical experience suggests that these drugs are generally effective and well tolerated in children with ADHD and sleep-onset insomnia, although little empirical evidence exists to justify the high level of clinician preference for these medications. The pros and cons of using this class of drugs for sleep are summarized below:

Considerations regarding alpha agonists for sleep in children

Pros	Cons
▪ Pharmacokinetics: Rapid absorption, onset action within 1 hour, peak effects 2 to 4 hours ▪ Generally well tolerated ▪ Widespread usage/acceptability ▪ Short half-life (clonidine) creates potential for middle-of-the-night dosing	▪ Little empirical evidence for efficacy, tolerability ▪ Effects on sleep architecture: Increased SWS, reduced REM ▪ May cause mid-sleep wakening ▪ Side effects: • Hypotension • Anticholinergic • Irritability, dysphoria • Rebound hypertension on discontinuation • Exacerbation • Parasomnias ▪ Tolerance often develops ▪ Narrow therapeutic index; risk of overdose
Bottom line – Little data to support current level of clinician preference, but clinical experience suggests generally effective and well tolerated in ADHD.	

SWS: slow-wave sleep; REM: rapid eye movement; ADHD: attention deficit hyperactivity disorder.

- Clonidine — central alpha-2-adrenergic agonist that decreases adrenergic tone
    - It is one of the most widely used medications for insomnia by both pediatric and mental health practitioners, particularly in children with sleep-onset delay and ADHD
    - Despite its widespread use, data regarding safety and efficacy in children with ADHD and sleep problems are limited to several descriptive or retrospective studies, which report adequate clinical response and a relatively low side effect profile
    - The immediate release form of the drug is rapidly absorbed with an onset of action within one hour and peak effects at two to four hours.
    - Tolerance often develops, which may necessitate dose escalation.
    - Mid-sleep awakening may also occur as blood levels drop during the night.
    - Effects on sleep architecture are fairly minimal but may include increased SWS and decreased rapid eye movement (REM) sleep
        - Thus, direct effects may include an increase in SWS partial-arousal parasomnias (e.g., sleep terrors) and REM sleep rebound later in the night with a related increase in nightmares
- Guanfacine is a selective alpha-2A adrenergic receptor agonist used primarily to treat ADHD
    - Similar to clonidine, off-label use in children with insomnia takes advantage of one of the drug's common side effects: mild drowsiness.
    - Guanfacine is generally less sedating than clonidine
    - Its long half-life (17 hours) suggests that it may be more appropriate than clonidine for sleep-maintenance insomnia, but evidence from trials to date have not supported that reasoning

t. Understand the clinical use of alpha-adrenergic agonists – see s. above

u. Understand the side effects and appropriate monitoring of alpha-adrenergic agonists

- Clonidine:
    - Side effects of clonidine include sedation, depression, bradycardia, headache, and possible hypotension
    - Discontinuation of clonidine requires tapering to prevent a rebound increase in blood pressure.
    - Oral clonidine may cause generalized rash, urticaria, or angioedema in patients with a history of hypersensitivity reactions to transdermal clonidine
- Guanfacine
    - Discontinuation of guanfacine requires tapering by ≤1 mg every three to seven days to prevent a rebound increase in blood pressure →During the taper, blood pressure and heart rate should be monitored
    - Adverse effects include headache, fatigue, abdominal pain, constipation, and sedation

v. Know indications for the use of anti-anxiety medications

- SSRI's, SNRI"s, atypical antidepressants already covered in other sections; will focus here solely on benzodiazepines and their use in the DBP patient
- Benzodiazepines
    - limited role in the treatment of pediatric anxiety disorders
    - They have a rapid onset of anxiolysis compared with antidepressants (minutes to hours versus days to weeks)
    - Benzodiazepines are, however, associated with significant adverse effects and their use in this population should be limited
    - SSRIs are generally preferred to benzodiazepines for long-term treatment of a pediatric anxiety disorder, though benzodiazepines can be useful in these patients to treat disabling anxiety while waiting for an antidepressant to take effect, or to treat SSRI-induced jitteriness

w. Understand the clinical use of common anti-anxiety medications – see other sections regarding SSRI's, SNRI's, atypical antidepressants, benzodiazepines; summary chart of options below:

Pharmacotherapy for anxiety disorders in children and adolescents

Agent	Initial daily dose	Suggested dose titration based upon response	Maintenance daily dose range	Selected characteristics*
Selective serotonin reuptake inhibitors (SSRIs)*				
Fluoxetine	Children: 5 to 10 mg Adolescents: 10 mg	After 7 days increase daily dose to 20 mg; then after 4 and 8 weeks increase daily dose by 20 mg, if needed	10 to 80 mg	▪ Prolonged half-life. ▪ Metabolized by and inhibits CYP2D6.
Fluvoxamine	25 to 50 mg at bedtime	Increase daily dose by 25 mg (child) or 25 to 50 mg (adolescent) after a minimum of 7 days, if needed	50 to 300 mg	▪ Girls generally require lower maintenance doses than boys. ▪ Give with meals and bedtime in divided doses to minimize side effects. ▪ Metabolized by CYP1A2 and 2D6. ▪ Inhibits CYP1A2 and 2C19.
Sertraline	12.5 to 25 mg	Increase daily dose by 12.5 mg (child) or 25 to 50 mg (adolescent) after a minimum of 7 days, if needed	50 to 200 mg	▪ Diarrhea more frequent than other SSRIs. ▪ Metabolized by CYP2D6. ▪ Inhibits CYP2D6 with larger doses.
Paroxetine	5 to 10 mg	Increase daily dose by 5 mg (child) or 10 mg (adolescent) after a minimum of 7 days, if needed	10 to 60 mg	▪ Short half-life. ▪ Mild anticholinergic side effects. ▪ Metabolized by and inhibits CYP2D6. ▪ Weight gain.
Serotonin norepinephrine reuptake inhibitor (SNRI)*				
Venlafaxine extended-release (ER)	37.5 mg	Increase daily dose by 37.5 mg (child) or 75 mg (adolescent) after a minimum of 7 days, if needed	75 to 225 mg	▪ Dose-related increase in diastolic blood pressure and/or heart rate may be seen. ▪ Some children may experience weight loss. ▪ Metabolized by CYPs 2D6 and 3A4. ▪ Prolongation of Qt interval.

Duloxetine	30 mg	Increase daily dose by 30 mg after a minimum of 14 days, if needed	30 to 60 mg Some patients may benefit from a higher daily dose, increased by 30 mg increments every 2 to 4 weeks, to maximum of 120 mg per day	▪ Some children may experience dose-related gastrointestinal-related adverse effects (eg, nausea and abdominal pain), and weight loss. Palpitations and increased pulse were observed more frequently than with placebo in a pediatric GAD trial. ▪ Use with strong inhibitors (eg, fluvoxamine) or inducers (eg, carbamazepine, rifampin) of CYP1A2 should in general be avoided. ▪ Strong CYP2D6 inhibitors (eg, fluoxetine, paroxetine, tipranavir) can increase duloxetine concentrations by up to 60%.
Tricyclic antidepressants (TCA)				
Clomipramine	Children ≥10 years old and adolescents: 25 mg	Increase daily dose by 25 mg after a minimum of 7 days, if needed; give in divided doses with meals and bedtime	25 to 250 mg (2 to 6¶ mg/kg per day); doses >2.5 mg/kg per day should be used cautiously¶	Applies to clomipramine and imipramine: ▪ Cardiovascular screening including ECG recommended prior to initiating treatment.¶ ▪ Anticholinergic side effects may limit usefulness in children. ▪ Drowsiness, irritability and vomiting may be seen. ▪ Give in divided doses with meals and at bedtime to minimize side effects. ▪ Metabolized by CYPs 1A2, 2C19 and 3A4.
Imipramine	10 to 25 mg	Increase daily dose by 25 mg after a minimum of 7 days, if needed; give in divided doses with meals and bedtime	10 to 300 mg (2 to 6¶ mg/kg per day); doses >2.5 mg/kg per day should be used cautiously¶	

Benzodiazepines				
Clonazepam	0.25 to 0.5 mg		1 to 6 mg	Applies to clonazepam and lorazepam: ▪ Drowsiness, irritability and oppositional behavior may be seen ▪ Subject to abuse, addiction and diversion
Lorazepam	0.25 to 0.5 mg		0.25 to 8 mg	

x. Know the side effects and appropriate monitoring of common anti-anxiety medications

- Common adverse effects of benzodiazepines include drowsiness, irritability and oppositional behavior
- Benzodiazepines can be subject to abuse, addiction, and diversion

y. Know indications for the use of antipsychotic medications for problems seen in developmental and behavioral pediatrics – covered in sections on mood stabilizing agents above

z. Understand the clinical use of antipsychotic medications for problems seen in developmental and behavioral pediatrics – covered in sections on mood stabilizing agents above

aa. Know the side effects and appropriate monitoring of antipsychotic medications. – covered in sections on mood stabilizing agents above

bb. Understand the clinical use of norepinephrine re-uptake inhibitors

- The serotonin-norepinephrine reuptake inhibitors include venlafaxine, desvenlafaxine (the active, major metabolite of venlafaxine), duloxetine, milnacipran, and levomilnacipran
- The only SNRI that has shown any potential for efficacy of treatment of pediatric unipolar depression is venlafaxine→can be considered an option in pediatric patients who are unresponsive to initial treatment with SSRIs

cc. Know the side effects and appropriate monitoring of norepinephrine re-uptake inhibitors

- Side effects of venlafaxine from trials compared to placebo:
    - Nausea – 24 versus 16 percent
    - Dizziness – 13 versus 9 percent
    - Dry mouth – 12 versus 10 percent
    - Insomnia – 11 versus 11 percent
    - Diaphoresis – 10 versus 5 percent
    - Constipation – 9 versus 9 percent
- Risk of increased blood pressure as well with venlafaxine treatment, especially at higher doses→blood pressure should be checked at baseline, and regularly monitored for patients at high doses of venlafaxine (e.g., every two to six months)

dd. Know the clinical use of atypical antidepressants (e.g., bupropion, clomipramine, etc)

- Tricyclic antidepressants are covered in other sections above
- Other atypical depressants studied for possible efficacy in pediatric populations:
    - Bupriopion
        - antidepressant that blocks the reuptake of norepinephrine and dopamine
        - limited data available in children- most studies looking at efficacy have been in adolescent patients
        - ADHD- bupriopion has more stimulant properties than the tricyclic antidepressants and is of modest efficacy in decreasing hyperactivity and aggressive behavior
            - In treating attention deficit hyperactivity disorder in adolescents with or at risk for SUD, the clinician should consider alternative agents to psychostimulants such as bupriopion
        - Depression- can be utilized as an option in depression refractory to SSRI's/SNRI's; may be most beneficial in patients with comorbid ADHD, conduct disorder, or adolescents with substance abuse problems or who want to quit smoking
    - Mirtazapine (Remeron)- not labeled by FDA for use in pediatric populations, but is used for treatment of insomnia, especially in patients with comorbid psychiatric diagnoses
        - Lower doses used for treatment of insomnia; higher doses used typically for treatment of anxiety/depression
        - Not good evidence to support efficacy, however, in pediatric populations over placebo
    - Trazodone- Serotonin Reuptake Inhibitor/Antagonist used in treatment of insomnia and migraine prophylaxis in pediatric populations
        - FDA approved down to 18 months for treatment of insomnia in pediatric populations; noted to have efficacy especially in patients with comorbid psychiatric diagnoses

ee. Know the side effects and appropriate monitoring of atypical antidepressants (e.g., bupropion, clomipramine, etc)- side effects of atypical antidepressants comparable to other antidepressants

- Side effects of Bupriopion

- Dry mouth – 21 percent
- Nausea – 13 percent
- Insomnia – 12 percent- due to mildly stimulating effects?
- Dizziness – 10 percent
- Anxiety – 6 percent- due to mildly stimulating effects?
- Dyspepsia – 6 percent
- Sinusitis – 5 percent
- Tremor – 5 percent
- Weight loss
- *Risk for seizures- Seizures may occur with bupropion and the incidence appears to be correlated with dose; risk especially increased in patients with anorexia nervosa and bulimia →contraindicated for this reason in these patients

ff. Know how to monitor for side effects of antipsychotic medications – see sections above covering this content in mood stabilizing agents

gg. Understand the basic principles of pharmacogenomics

- Pharmacogenomics refers to the role of various components of the genome on response to a drug
  - Pharmacogenetics is a subcategory of pharmacogenomics that refers to the role of genetic variation on response to a drug.
    - Pharmacogenetics generally is used to refer to a specific DNA polymorphism or coding variant rather than epigenetic or transcriptomic changes across the genome.
    - In practice, pharmacogenetics and pharmacogenomics are often used interchangeably.
  - Among the most commonly studied are genetic sequence variants, structural changes in chromosomes (e.g., translocations), epigenetic variants (e.g., changes in gene methylation), and variation in the expression profile of genes (changes in messenger RNA [mRNA] levels) or noncoding RNA (e.g., changes in microRNA).
    - Genetic variation
      - refers to differences in genetic sequences among individuals in a population
      - single nucleotide polymorphisms (SNPs) refer to variation at a single base pair, typically with a population frequency of at least 1 percent
      - Other forms of variation include insertions, deletions, copy number variants, and short tandem repeats
      - Variants that are seen at much lower prevalence than 1 percent of the population are often referred to as mutations, although this term may also be used to distinguish between variation that is inherited versus variation that arises de novo
      - All forms of variation have the potential to impact phenotype, regardless of their frequency, but the impact

depends on a number of factors including the location of the variation within the genome and the functional consequences of the variation

- Epigenetic changes are those that affect genes without altering the gene sequence.

  - This may occur via changes in gene methylation or histone modification (methylation, acetylation), either of which can influence the rate of transcription or silencing of gene expression.
  - Other epigenetic changes include the alterations in noncoding RNAs and telomere length. These epigenetic changes can be passed on from parents to offspring but can also result from environmental influences on the epigenome.
  - An example of an epigenetic change that affects drug metabolism is reduced sensitivity of a tumor to a chemotherapeutic drug due to gene methylation

  - The genetic variation can be inherited through the germline or acquired (e.g., somatic mutation in a tumor)
  - The availability of high-throughput techniques to interrogate the entire genome has facilitated many pharmacogenomic studies.

- Pharmacogenomic influences on drug response have traditionally been divided into four categories based upon the impact of genetic variability on the pharmacologic properties of a drug:
  - Effect on drug pharmacokinetics; an example is a genetic variant that alters drug metabolism, affecting plasma concentration.
  - Effects on pharmacodynamics; an example is a genetic variation that reduces binding of the drug to its receptor, thereby decreasing therapeutic efficacy.
  - Effects on idiosyncratic reactions, such as the likelihood of a hypersensitivity reaction to a certain drug.
  - Effects on disease pathogenesis or severity and response to specific therapies; these include specific molecular defects related to the pathogenesis of certain malignancies for which specific targeted therapies have been developed.

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6. Other strategies

a**. Understand the importance of adequate basal treatment of pain**

Children with impairment of the central nervous system (ex. CP) are at increased risk for pain in comparison to healthy children, however this population remains vulnerable to under recognition and under treatment of pain. Patient's in this population are at risk for acute pain from surgical interventions as well as chronic pain from a number of sources including GERD, constipation, spasticity, hip subluxation and central neuropathic pain. Pain can often be difficult to identify, particularly in a nonverbal child. Due to complexity of identifying and treating pain in children, pain often goes unrecognized/undertreated. One study in children with CP showed that 90% experienced recurrent pain for more than 1 year, yet only half were receiving any treatment directed at pain. In children with progressive genetic, metabolic, or neurologic conditions with no cure pain is one of the 3 most common symptoms reported by parents with symptoms often not well controlled. It is essential to treat pain as recurrent pain can have significant effects on daily life and quality of life including sleep and family interactions. Pain can lead to distress, anxiety, depression, irritability/aggression, insomnia, fatigue, and negative coping behaviors in the child and family members. It is therefore essential to have adequate basal treatment of pain.[1]

b. Understand how to formulate a pain management plan

Treatment starts with a comprehensive evaluation. Initial goal is to identify and treat the cause of pain when possible. Assess acute causes of pain vs chronic causes of pain. Tailor the pain management plan to each child based on severity, frequency and duration of episodes of pain and the expected outcome after an empirical medication trial.

Additionally, pain management requires close follow up and availability of the provider. Treatment should then follow the below steps:
Step 1- mild pain: use of nonopioid analgesics
Step 2- moderate to severe pain: use of opioid analgesics, starting with a lower dose. Adjunctive therapies can be used at either step. Adjunctive therapies include anticonvulsants and antidepressants which can be used for neuropathic pain in addition to other therapies such as physical therapy, etc.

Other treatment principles guided by the WHO include “by the clock,” “by the mouth,” and “by the child,” which indicates treatment should be schedule when pain is frequent, with rescue doses available PRN (by the clock). Medications should be given by the least-invasive route (by the mouth) and be tailored to the needs of the child and response to treatment (by the child).

Of note, there is no standard approach to treatment of pain for children with significant impairment of the central nervous system. Therefore, pain management plans should be guided by safety of medications and response to therapies.[1]

c. Understand the principles and techniques of hypnosis

Hypnosis is the induction of a deeply relaxed state, with increased suggestibility and suspension of critical faculties. Once in this state, patients are given therapeutic suggestions to encourage change in behavior or relief of symptoms. [2]

Hypnosis is form of mind-body therapy and is a skill set involving interpersonal communication designed to facilitate therapeutic change in maladaptive psycho-physiological reflexes. Hypnotherapy in children is a well-established therapeutic modality. Clinical hypnosis involves establishing a strong rapport with patients and individualizing the therapy to the specific goals and characteristics of the patient. There are no large randomized controlled trials to assess the efficacy of hypnotherapy, but research has suggested benefits for children and adolescents for certain conditions including IBS and pain management with promising evidence for its application in conditions such as enuresis, tics/Tourette syndrome, migraines and anxiety.[3]

d. Understand the principles and techniques of biofeedback

Biofeedback is the use of electronic or electromechanical equipment to measure and then feed-back information about physiologic processes to an individual. These physiologic processes can then be controlled by the individual for therapeutic purposes. Feedback can be provided in auditory, visual, kinesthetic, or multimedia formats and even in video game forms. Biofeedback is a useful tool to help gauge what topics, thoughts and other phenomena trigger mind/body arousal in children and adolescents. The benefit for patients include allowing them to observe the immediate, objective mind-body interactions allowing them to see that a change in the thoughts/feelings can result in a physiologic response. Research shows benefits of peripheral forms of biofeedback in pediatrics particularly for headache, asthma, enuresis and rehabilitation applications.

Additionally, neurofeedback (i.e. EEG biofeedback) has been shown to have benefit for ADHD. There is positive evidence for other indications including insomnia and chronic pain syndromes, but data is not conclusive.[3]

e. Know appropriate advice for a school on the appropriate timing and nature of memorialization activities after the death of a student

Commerative activities and memorialization efforts should not be a focus of the school response in the immediate aftermath of a death. If done too soon, there may be a perception that the school is trying to "close the chapter" on grieving. In general, schools should avoid formal commemorative or memorialization activities or acts to mark the death of a popular student or staff member since failure to respond in the future in a similar manner to the death of a less popular student/staff may raise equity concerns. Instead, less formal but thoughtful commemorative activities developed over time with active student involvement is often much more meaningful and therapeutic to students and staff.[4]

f. Understand principles in providing post-suicide intervention services in a high school

The following principles should be utilized in providing post-suicide intervention services in a high school:

1. Identify the students who are at risk or may be profoundly affected by the suicide (ex. students with history of suicide attempts, friends and siblings of the child who committed suicide, etc.)
2. Develop a response and support plan for all identified students, in collaboration with mental health professionals. This should include an assessment of whether it is appropriate to make contact with the parent/guardians of the identified at risk students, contacting the student, placing referrals to a student support services officer for support, referrals to a mental health professional for a risk assessment, if appropriate and a documented plan of the support to be provided to the student.
3. Monitor staff wellbeing
4. Keep the school community informed of funeral arrangements, changes to previously planned activities, availability of counseling services in the school and any other changes or updates.

Long term, schools should also review curriculum and school-based prevention and intervention programs that are being provided, consider professional learning opportunities for school staff in understanding the impact of good mental health and wellbeing on students learning and the use of effective coping skills. Additionally, schools should consider holding events for students and the school community that foster optimistic thinking and a sense of connectedness.[5]

g. Know how to evaluate the utility of non-standard therapies for developmental and behavioral disorders

Complementary, alternative and integrative therapies can be grouped into 3 general areas: natural products, mind and body practices and other therapies. The National Center for Complementary and Integrative Health maintains a web site in which current information on novel therapies in popular use are reviewed and can be a useful tool in evaluating the utility of non-standard therapies.[6]

h. Know the risks of special diets, supplements, and other common alternative treatments that are often recommended for children with developmental disabilities

- Nutrition: gluten-free and casein-free diets/ vitamin supplementation – generally safe but can be expensive/ difficult for families to maintain.
- Immunomodulation – antifungals and antibiotics in addition to prebiotics and probiotics – popular among families. No supportive data. Pro/prebiotics are likely safe with minimal harm however the use of antifungals/ antibiotics can induce resistance/ have side effects.
- Biochemical and metabolic therapies- use of precursors and coenzyme factors (ex. B12 shots) – no evidence, likely minimal harm.
- Detoxification- theory that heavy metal poisoning can cause symptoms (true in regards to lead, but non stardized testing is often used for other heavy metals). Chelation therapy has been used to "treat this" but has not shown benefit and has resulted in severe side effects including death.
- Manipulative and body- based practices- chiropractic manipulation, craniosacial massage, massage therapy, therapeutic touch- no proven benefit, likely no harm.
- Music and other expressive therapies (ex. Horseback riding therapies, etc.)- conflicting evidence, no harm but can be expensive for families
- Hyperbaric oxygen therapy – no proven benefit, very expensive, some harm including blindness has been reported.[7]

i. Know how to counsel families who are utilizing non-standard (alternative) therapies

Between 28% and 74% of children with ASD are given at least 1, and usually more than 1, complementary therapy. It is therefore critical to ask all families about any complementary treatments they are providing as they may not bring them up otherwise. Providers should work with families to identify the target symptoms they are hoping to address and work in a stepwise fashion to identify any benefits or side effects from complementary medicines. In general, any therapy that has shown benefit and is safe should be recommended to families (ex. Melatonin), any therapy that has limited evidence and is safe should be accepted (ex. Yoga therapy) and any therapy that is unsafe that has no evidence or is known to not be beneficial should be discouraged (ex. Chelation therapy).[7]

Resources:

1. Hauer, Julie, and Amy J. Houtrow. “Pain Assessment and Treatment in Children With Significant Impairment of the Central Nervous System.” *Pediatrics*, vol. 139, no. 6, 2017, doi:10.1542/peds.2017-1002.
2. Vickers, A. “Hypnosis and Relaxation Therapies.” *Western Journal of Medicine*, vol. 175, no. 4, 2001, pp. 269–272., doi:10.1136/ewjm.175.4.269.
3. “Mind-Body Therapies in Children and Youth.” *Pediatrics*, vol. 138, no. 3, 2016, doi:10.1542/peds.2016-1896.
4. *National Center for School Crisis and Bereavement*, 16 July 2020, www.schoolcrisiscenter.org/.
5. “Guidelines to Assist in Responding to Attempted Suicide or ...” *Suicideguidelines.pdf*, www.education.vic.gov.au/Documents/school/principals/health/suicideguidelines.pdf.
6. Lubetsky, Martin J., et al. “Recognition of Autism Spectrum Disorder—Early Identification and Diagnosis.” *Autism Spectrum Disorder*, 2011, pp. 19–37., doi:10.1093/med/9780199753857.003.0016.
7. Hyman, Susan L., et al. “Identification, Evaluation, and Management of Children With Autism Spectrum Disorder.” Pediatrics, vol. 145, no. 1, 2019, doi:10.1542/peds.2019-3447.

4C

1. Principles of collaboration

a. Know the importance and process of identifying the underlying agenda for consultation requests from community sites

- the core functions of consultation likely include: (1) continued training, (2) problem-solving implementation barriers, (3) provider engagement, (4) case support, (5) accountability, (6) mastery skill-building, (7) appropriate treatment adaptation and (8) planning for sustainability

- identified agenda-setting, collaborative goal setting, formulation, planning, discussion (including disagreements, communication of understanding, challenging, explanation), didactic instruction, modeling, role-play, monitoring or observation, review and reflection, summarizing, confidence-building, praise and reinforcement, and feedback as core *processes* that can be used in consultation

Nadeem E, Gleacher A, Beidas RS. Consultation as an implementation strategy for evidence-based practices across multiple contexts: unpacking the black box. Adm Policy Ment Health. 2013 Nov;40(6):439-50. doi: 10.1007/s10488-013-0502-8. PMID: 23716145; PMCID: PMC3795855.

b. Know the steps in establishing a collaborative relationship with community organizations and agencies

1) identify and define strategic partnerships/engage stakeholders (a group of organizations with a common interest who agree to work together toward a common goal)
2) Establish personal relationships, and begin to build trust
3) Clarify the goals and objective each partner wants to accomplish
4) Choose and implement a partnership that is mutually beneficial

5) Establish goverance, procedures, ground rules, and decision-making structure

Snow, John. Engaging Your Community: A Toolkit for Partnership, Collaboration, and Action. Department of Heath and Human Services, OAH, HHSP23320100000550G.

1) Connect with leaders at partner organizations to promote engagement
2) Define and prioritize goals
3) Build new partner relationships and strengthen long standing ones
4) Ensure screening and referral protocols are seamless

www.fiercehealthcare.com >population-health> (Dec 13, 2016)

c. Know how developmental-behavioral pediatricians can serve as consultants to schools
1) Provide education about neurodevelopmental disabilities in general
2) Provide clinical insights about an individual patient (when allowed by parent)
3) serve as members of the IEP evaluation team
4) provide results of developmental or medical evaluation (when allowed by parent)

d. Know the steps in conducting an evaluation of a child as a school consultant
1) parent requests evaluation

Steps for a comprehensive educational evaluation include:
2) Team meeting to determine necessary evaluation (Psychological, Psycho-educational testing, interviews, class room observation, functional behavioral assessment, speech/language, occupational therapy, physical therapy, medical evaluation, hearing/vision)
3) Each evaluation is completed and evaluator writes a report
4) IEP team holds an eligibility meeting (include parents and professionals) to review results and determine eligibility and any necessary services

e. Know how to conduct an evaluation of a child as a consultant to a child care center
Young children in child care centers may be evaluated using screening tools or validated developmental assessments that are age appropriate.

Child care health consultants (CCHCs) are health professionals who know about child health, child development, and health and safety in child care settings. CCHCs and the child care staff work together to promote healthy and safe environments for young children.

Child Care Health Consultants | ECLKC
eclkc.ohs.acf.hhs.gov › article › child-care-health-consult... Jul 27, 2020

American Academy of Pediatrics, American Public Health Association, National Resource Center for Health and Safety in Child Care and Early Education. 2014. *Caring for infants and toddlers in child care and early education*. *Applicable standards from: Caring for our children: National health and safety performance standards; Guidelines for early care and education programs*, 3rd Edition. Elk Grove Village, IL: American Academy of Pediatrics; Washington, DC: American Public Health Association. Available at http://nrckids.org.

f. Understand the benefits and challenges of multidisciplinary evaluations within a collaborative team model
Benefits of multidisciplinary evaluation: access to an entire team of experts, improves service coordination, expedites referral process, creates new avenues for service implementation, allows patients to create goals for themselves.

Challenges include: time pressure in providing services (team discussion), team members have unique backgrounds, requires frequent collaboration to be effective, dependent on resources (manpower, access, timing), incomplete decisions can happen when only part of the evaluation is completed, documentation can be more difficult (who documents what – may see more patients in one day).

https://brandongaille.com/11-multidisciplinary-team-advantages-and-disadvantages/

g. Understand how the role of consultant to a school system differs in quality and scope to the role of the physician seeing a patient in the office

-medical identification is often the first line/ first to identify a problem that relates to learning
-be familiar with local school district/state policy on special education eligibility and early intervention
-educational advocacy for the patient/student based on identified needs
-perform or interpret psychometric testing

Voight, Robert G. et al. AAP Developmental and Behavioral Pediatrics, 2nd Edition Ch.20.

h. Understand issues of confidentiality as they relate to the role of consultant to a school system

-medical confidentiality falls under HIPAA
-educational confidentiality falls under FERPA (Family Education Rights and Privacy Act); clinicians must adhere to both HIPAA and FERPA in the educational system
-FERPA applies to all schools that receive funds from US Dept of Education
-both policies require consent from parent (or eligible student if 18yrs) for release of information (medical or educational)

https://www2.ed.gov/policy/gen/guid/fpco/ferpa/index.html

4C

2. Knowledge of other health professionals' roles and methods

a. Be familiar with the elements of a psychological and educational evaluation conducted as part of a multidisciplinary team evaluation for a child with learning and/or behavior problems

-psychometric tools are performed to understand cognitive function, language function, motor function, academic function, social-emotional function, adaptive and behavioral function of a child
-formal testing may not be needed if screening is normal and input from family/teacher/medical does not indicate a need for further testing
-a Functional Behavioral Assessment (FBA) focusing on the problem behavior, antecedent behaviors and interventions. Results in a Behavioral intervention plan (BIP)

Voight, Robert G. et al. AAP Developmental and Behavioral Pediatrics, 2nd Edition Ch.20
https://teach.com/online-ed/psychology-degrees/online-masters-applied-behavior-analysis/functional-behavior-analysis/

b. Understand common behavioral assessment techniques

-clinical interview (DSM5, DC 0-5) consisting of medical history, behavior history, behavioral observation,
-supplements to clinical interview: ADHD rating scales (NICHQ/Vanderbilt, Connors), Behavioral rating scales (BASC Behavior Assessment System for Children, CBCL Child Behavior Checklist, ECBI Eyberg Child Behavior Inventory, MOAS Modified Overt Aggression Scale)

-understand parent-child dyad or parent-teacher dyad
-recognize risks for behavior disorders (ACES) and comorbid conditions

c. Understand the elements of a neuropsychological evaluation of a child

Psychological evaluations focus on identifying and diagnosing a specific neurodevelopmental disorder (e.g., ADHD, Autism Spectrum Disorder, etc.) or psychiatric disorder (mood or anxiety disorder, conduct disorder, etc.); involve obtaining a detailed history, behavioral observations and a series of standardized parent, teacher, or self-report measures, personality tests, and sometimes tests of general cognitive functioning; performed by licensed clinical psychologists or school psychologists.

Psychoeducational evaluations focus on academic achievement and general cognitive (or intellectual) abilities; conducted by licensed clinical psychologists and school psychologists; used to determine eligibility for special education services and accommodations for a specific learning disability.

Developmental evaluations focus on early childhood development and are typically requested for infant, toddlers, and preschool children to determine whether or not development is occurring at expected rates across domains and if child qualifies for early intervention; performed by a variety of healthcare/therapy/school providers such as licensed clinical psychologists, developmental pediatricians, speech pathologists, occupational therapists, school psychologists.

Neuropsychological evaluations are performed by licensed clinical psychologists who have specialized training in neuropsychology. A neuropsychological evaluation focuses on learning and behavior in relation to an individual's brain, involves the use of standardized, norm-referenced tests and behavioral observations, and incorporates knowledge of brain development, organization, and functioning into the evaluation findings. Neuropsychological evaluations are considered the most comprehensive type of evaluation, and typically include psychological and psychoeducational testing components, but the major difference is that neuropsychological testing goes a step further to understand the relationship between behavioral, cognitive, and functional deficits and underlying brain functions. Neuropsychological evaluations measure cognitive abilities and processes such as attention and executive functions, reasoning, language, memory and learning, and visual-spatial/visual-motor abilities. Thus, by looking at a patient's functioning and abilities comprehensively, one can better understand not just "what" the problem is, but "why" there is a problem. For instance, a child may struggle to accurately answer questions about what he/she just read. Whereas a standard psychoeducational assessment might determine "IF" the child qualifies for special education services for reading comprehension problems, a neuropsychological evaluation would investigate "WHY" the child is having reading comprehension problems. For example, is the child's difficulty due to visual processing difficulties (such as visual-perception or visual attention/visual scanning problems), weakness in working memory, receptive versus expressive language deficits, or even performance anxiety, etc.. A clearer understanding of "why" problems exist and their underlying cognitive abilities or processing deficits, as well as emotional and behavioral factors, is crucial in determining the most effective, individually-tailored intervention plan.

Patients may be referred for neuropsychological evaluations in cases involving known neurological conditions (e.g., seizures, neurofibromatosis, brain tumors) and injuries (e.g., concussions), as well as exposure to toxins (e.g., lead poisoning, alcohol and substance exposure prior to birth) and medical conditions and factors which increase risks for brain injury (e.g., history of complicated pregnancy, delivery, and prematurity). Neuropsychological evaluations are also recommended for assessment of complex neurodevelopment disorders (such as ADHD and learning disorders). For instance, a patient with ADHD may fail to respond to standard treatment efforts, and comprehensive testing may be necessary to determine the severity of deficits so that expectations can be more appropriately tailored, and to determine the presence of co-existing disorders and problems which may be hampering treatment progress. In other cases, very bright individuals with ADHD and/or learning disorders may be presenting unique challenges to parents and educators. These "twice exceptional" students often benefit from comprehensive evaluations to fully understand their range of strengths and weaknesses so that the appropriate combination of enrichment, accommodations, and treatments can be developed.

https://www.cvillemindworks.com/evaluation-descriptions

d. Recognize the indications for neurological consultation in a child with a developmental or behavioral problem
-concern for epilepsy, abnormal neurological examination, regression, genetic condition

e. Know the role of genetic consultation and counseling in a child with developmental disabilities
-aim to identify underlying genetic etiology for developmental disabilities (and thus genetic comorbidities)
-aim to provide accurate recurrence risk information for parents (and for patient once he/she is of child-bearing age)
-aim to understand risks for other family members
-standard genetic evaluation for diagnoses such as ASD, Intellectual Disability, children with progressive deterioration of skills (any domain), dysmorphic features, known genetic change

f. Know the indications for speech and language evaluation and treatment in a child with developmental or behavioral disorders
-delay communication skills or feeding skills
-delay can be in language acquisition (expressive language), articulation, receptive language or pragmatics

g. Know how speech and language testing evaluates pragmatics, semantics, and syntax
- **Syntax** is the study of sentence structure and the rules of grammar. While people can do what they want with language (and many often do), syntax helps common users of a language understand how to organize words so that they make the most sense.
- **Semantics**, on the other hand, is the study of the meaning of sentences.
- **Pragmatics** takes semantics one step further, because it's the study of the meaning of sentences within a certain context. The Social use of language.

Standardized testing and motor examination are used to evaluate

h. Differentiate between the evaluations and services provided by physical therapy and occupational therapy

-PT: Gross motor impairments/delays, evaluation for orthotics of the lower extremities/ambulation, support and equipment for overall mobility, strength

-OT: fine motor impairments/delays, evaluations for equipment to support upper extremities, equipment for adaptive function and activities of daily living, support for sensory/emotional regulation as it relates to adaptive functioning

i. Recognize the role of occupational therapy in evaluation and treatment of children with developmental disorders

-Occupational therapists select interventions for children based upon an analysis of the child's performance of daily life roles, how their performance is affected by their disability, and how their environment supports or constrains their performance

- many occupational therapists have moved away from impairment-based interventions at the body structures and functions level aimed at remediating the child's deficits (known as 'bottom-up' interventions), and instead to focus on improving functional activity performance and participation ('top-down' interventions), as well as partnering with parents to deliver therapy embedded within daily life

- Pediatric occupational therapy helps children gain independence while also strengthening the development of fine motor skills, sensory motor skills, and visual motor skills that children need to function and socialize.

Novak I, Honan I. Effectiveness of paediatric occupational therapy for children with disabilities: A systematic review. *Aust Occup Ther J.* 2019;66(3):258-273. doi:10.1111/1440-1630.12573

- Pediatric occupational therapists address upper extremity function, fine motor skills, visual-motor function, sensory processing skills, and the occupations or tasks that are expected of the child.[22] These tasks are referred to as activities of daily living (daily tasks such as feeding, eating, dressing, or toileting) and instrumental activities of daily living (complex tasks such as cooking, shopping, or using a telephone). Occupational therapists are also involved in identifying the equipment needs a child might have to perform tasks. To address impairments in the child with cerebral palsy, for example, the occupational therapist works on grasping and hand coordination; to help with an activity such as dressing, the occupational therapist works with the child to practice the skill and use an assistive device; and to aid in participation, the occupational therapist provides strategies that the child can use in and out of the classroom such as self-regulation techniques or taking notes on a keyboard versus on paper.

Prescribing Physical, Occupational, and Speech Therapy Services for Children With Disabilities. Amy Houtrow, Nancy Murphy and COUNCIL ON CHILDREN WITH DISABILITIES. Pediatrics April 2019, 143 (4) e20190285; DOI: https://doi.org/10.1542/peds.2019-0285

j. Know controversial therapies (eg, sensory integration, optometric training, facilitated communication, etc) and their current status (eg, unproven, questionable, unsafe, proven ineffective, etc)

-sensory integration therapy is unproven. Sensory-based therapies involve activities that are believed to organize the sensory system by providing vestibular, proprioceptive, auditory, and tactile inputs. Brushes, swings, balls, and other specially designed therapeutic or recreational equipment are used to provide these inputs

Sensory integration therapies for children with developmental and behavioral disorders Section On Complementary And Integrative Medicine; Council on Children with Disabilities; American Academy of Pediatrics; Michelle Zimmer, Larry Desch
Pediatrics. 2012 Jun;129(6):1186-9. doi: 10.1542/peds.2012-0876. Epub 2012 May 28.

-optometric training: Because they are difficult for the public to understand and for educators to treat, learning disabilities have spawned a wide variety of scientifically unsupported vision-based diagnostic and treatment procedures. Scientific evidence does not support the claims that visual training, muscle exercises, ocular pursuit-and-tracking exercises, behavioral/perceptual vision therapy, “training” glasses, prisms, and colored lenses and filters are effective direct or indirect treatments for learning disabilities. There is no valid evidence that children who participate in vision therapy are more responsive to educational instruction than children who do not participate.

Sheryl M. Handler, Walter M. Fierson and the Section on Ophthalmology and Council on Children with Disabilities, American Academy of Ophthalmology, American Association for Pediatric Ophthalmology and Strabismus, and American Association of Certified Orthoptists. Learning Disabilities, Dyslexia, and Vision. Pediatrics March 2011, 127 (3) e818-e856; DOI: https://doi.org/10.1542/peds.2010-3670

-facilitated communication: Facilitated communication is a technique that involves a facilitator physically supporting the hand, wrist or arm of an autistic person while the person spells out words on a keyboard or similar device. It's sometimes called 'assisted typing' or 'supported typing'. It is the position of the American Speech-Language-Hearing Association (ASHA) that Facilitated Communication (FC) is a discredited technique that should not be used. There is no scientific evidence of the validity of FC, and there is extensive scientific evidence—produced over several decades and across several countries—that messages are authored by the "facilitator" rather than the person with a disability. Furthermore, there is extensive evidence of harms related to the use of FC. Information obtained through the use of FC should not be considered as the communication of the person with a disability.

https://www.asha.org/policy/ps2018-00352/
ASHA Position Statement, Facilitated Communication

k. Recognize indications for psychiatric consultation in a child with a developmental or behavioral problem

- **Specific Criteria for Referrals:** The referring practitioner should consider the following criteria when considering the decision to refer.

1. When a child or adolescent demonstrates an emotional or behavioral problem that constitutes a threat to the safety of the child/adolescent or the safety of those around him/her. (e.g. suicidal behavior, severe aggressive behavioral, an eating disorder that is out of control, other self-destructive behavior),
2. When a child or adolescent demonstrates a significant change in his/her emotional or behavioral functioning for which there is no obvious or recognized precipitant. (e.g. the sudden onset of school avoidance, a suicide attempt or gesture in a previously well functioning individual),
3. When a child or adolescent demonstrates emotional or behavioral problems (regardless of severity), and the primary caretaker has serious emotional impairment or substance abuse problem. (e.g. a child with emotional withdrawal, whose parent is significantly depressed, a child with behavioral difficulties whose parents are going through a “hostile” divorce),

4. When a child or adolescent demonstrates an emotional or behavioral problem in which there is evidence of significant disruption in day-to-day functioning or reality contact. (e.g. a child/adolescent who has repeated severe tantrums with no apparent reason, a child reports hallucinatory experiences without an identifiable physical cause),
5. When a child or adolescent is hospitalized for the treatment of a psychiatric illness,
6. When a child or adolescent with behavioral or emotional problems has had a course of treatment intervention for six to eight weeks without meaningful improvement,
7. When a child or adolescent presents with complex diagnostic issues involving cognitive, psychological, and emotional components that may be related to an organic etiology or complex mental health/legal issues,
8. When a child or adolescent has a history of abuse, neglect and/or removal from home, with current significant symptoms as a result of these actions,
9. When a child or adolescent whose symptom picture and family psychiatric history suggests that treatment with psychotropic medication may result in an adverse response. (e.g. the prescription of stimulants for a hyperactive child with a family history of bipolar disorder or schizophrenia),
10. When a child or adolescent has had only a partial response to a course of psychotropic medication or when any child is being treated with more than two psychotropic medications,
11. When a child under the age of five experiences emotional or behavioral disturbances that are sufficiently severe or prolonged as to merit a recommendation for the ongoing use of a psychotropic medication, or
12. When a child or adolescent with a chronic medical condition demonstrates behavior that seriously interferes with the treatment of that condition.

https://www.aacap.org/AACAP/Member_Resources/Practice_Information/When_to_Seek_Referral_or_Consultation_with_a_CAP.aspx

l. Recognize the role of physical therapy in evaluation and treatment of children with developmental disorders

- Physical therapists address gross motor skills, strength building, endurance, and fitness. They also focus on prevention or reduction of impairments to achieve optimal functional mobility and participation. They help children move, often with the use of strategies to prevent the progression of impairments and through the use of adaptive equipment such as orthotics (braces) and various mobility aids such as walkers, wheelchairs, and lifts. For a child with cerebral palsy, for example, the physical therapist addresses impairments related to spasticity, weakness, poor postural control, and lack of coordination. To minimize activity limitations, the physical therapist helps the child with walking skills (among others). To address participation restrictions, the physical therapist helps the child learn to navigate a public space such as the hallways at school.[21]

Prescribing Physical, Occupational, and Speech Therapy Services for Children With Disabilities. Amy Houtrow, Nancy Murphy and COUNCIL ON CHILDREN WITH DISABILITIES. Pediatrics April 2019, 143 (4) e20190285; DOI: https://doi.org/10.1542/peds.2019-0285

m. Differentiate among the evaluations and services provided by a play therapist, behavioral counselor, behavior analyst, psychodynamic therapist, and family therapist

-play therapist: child play therapy is a way of being with the child that honors their unique developmental level and looks for ways of helping in the 'language' of the child – play. Licensed mental health professionals therapeutically use play to help their clients, most often children ages three to 12 years, to better express themselves and resolve their problems. PT therapy, as briefly mentioned, typically involves few rules or limits for the child. How long are the sessions? Most commonly they are about 30-50 minutes long. As opposed to a lot of discussions or verbal interaction, PT simply involves play, in the hope that a child will feel free to both explore their lives and express their thoughts and emotions through toys, art, etc. The overall goal of PT is to teach children (or help them learn) to express themselves and their emotions in healthy ways, learn how to demonstrate respect and empathy, and discover new, healthier ways to solve encountered problems.
-behavioral counselor: A behavioral counselor is a professional in the field of psychology who uses a variety of different therapies to help their patients change certain behaviors. ... In behavioral therapy, a professional will work with an individual patient to try and reinforce desirable behaviors and stop the ones that are not.
-behavior analyst: ABA places a stronger emphasis on the immediate and uses practical conditioning (strength of a behavior is modified by reinforcement) to create positive change. ABA practitioners utilize conditioning to encourage positive development of prosocial behavior. For example, the behavioral interventionist may conduct sessions where they provide a small reward each time the client (often a child diagnosed with autism) is able to perform a certain activity and/or task (completing a self help task like brushing teeth) or practicing a social skill such as turn-taking while playing.
-psychodynamic therapist: Psychodynamic therapy involves the interpretation of mental and emotional processes rather than focusing on behavior (Strupp, Butler, & Rosser, 1988). Psychodynamic therapists attempt to help clients find patterns in their emotions, thoughts, and beliefs in order to gain insight into their current self.
-family therapist: Marriage and family therapists work with individuals, couples, and families. They evaluate family roles and development, to understand how clients' families affect their mental health. They treat the clients' relationships, not just the clients themselves.

n. Know the role of a vision specialist and orientation and mobility specialist in the treatment of a child with visual impairment

- COMS (Certified Orientation and Mobility Specialist) Orientation and mobility instruction is a sequential process in which visually impaired individuals are taught to utilize their remaining senses to determine their position within their environment and to negotiate safe movement from one place to another.
- CLVT(Certified Low Vision Specialist) uses functional vision evaluation instruments to assess visual acuity, visual fields, contrast sensitivity function, color vision, stereopsis, visual perceptual and visual motor functioning, literacy skills in reading and writing, etc. as they relate to vision impairment and disability. The CLVT also evaluates work history, educational performance, ADL and IADL performance, use of technology, quality of life and aspects of psychosocial and cognitive function.
-Low Vision Specialists refers to an ophthalmologist or optometrist who has completed additional training and certification in the area of low vision. An exam conducted by a low vision specialist is similar to the exam conducted by either an ophthalmologist or optometrist.

However, it will have additional components that focus on helping children maximize their usable vision through low vision devices for

- near tasks (closer than 16 inches),
- intermediate tasks (16 inches to 3 feet), and
- distance tasks (beyond 3 feet).

The low vision specialist can prescribe aids such as magnifiers, monoculars, or video magnifiers. A low vision specialist also considers how lighting and nonoptical aids such as a reading stand, bold-lined paper, or nonprescription sun lenses can help children use their vision more efficiently.

-A certified low vision therapist (CLVT) has completed an internship and passed an exam to demonstrate knowledge of low vision. The CLVT conducts a functional vision assessment (FVA) to determine how a child is using his or her vision for education, recreation, and daily living tasks. The low vision therapist may also be a teacher of students with visual impairments (TVI) or an orientation and mobility (O&M) specialist. The CLVT works closely with either an ophthalmologist or optometrist who prescribes low vision devices based on the functional vision assessment. The CLVT also teaches children how to use these aids and other techniques to maximize their functional vision. A CLVT is not a doctor and cannot diagnose an eye disease, prescribe eyeglasses or contact lenses, or prescribe medications.

https://www.acvrep.org/certifications/clvt
https://familyconnect.org/education/expanded-core-curriculum/orientation-and-mobility/

o. Know the role of a teacher of the deaf in the treatment of a child with permanent hearing loss

- Areas of learning most likely to be impacted by hearing loss:

•Vocabulary: Gaps due to decreased ability to overhear incidental language
•Early reading: Phonology/phonemic awareness issues related to not distinctly hearing speech sounds
•Language processing: Issues due to fragmented hearing, vocabulary gaps, syntax, listening rate, etc. Not understanding words in context.
•Syntax: Incomplete understanding of rules (i.e., cannot hear /s/ or /ed/ endings so do not understand plurals, possessives, past tense)
•Listening skills: Can be challenges with simple discrimination of sounds, phrases or comprehension of conversation or verbal instruction in class (they may hear but not process full meaning)
•Understanding intent/emotions of others and nonverbal social cues
•Viewing information from different perspectives; critical thinking
•Social language: Socially awkward due to delays in pragmatic language development, how to interact appropriately with peers
•Attention/distractibility: Periodic inattention due to listening fatigue and gaps in understanding; 'tuning out' when it is hard to understand
• Passive or immature skills in responding when they do not understand what was said; lack of self-advocacy
• Understanding group discussions or participating in small group work due to distance/noise in class and socially

Any of these areas can adversely impact school performance and social behavior. All of these issues can contribute to cumulative challenges in academic performance and school function over time.

- Teachers of the deaf/hard of hearing can recommend assessments appropriate to identifying issues in these vulnerable areas. They can provide insights into team assessment results based on their knowledge of how hearing loss impacts development and learning. Only 1% of IEPs are for children who are deaf or hard of hearing. Viewing assessment results without these insights often causes these low incidence issues to go unrecognized. Teachers can provide inservice, training or consultation to school staff and families. They can assist in determining appropriate modifications and accommodations. While a special education teacher can address academic issues and a speech therapist can address speech/language issues, the teacher of the deaf/hard of hearing can address all of the issues above and support improvement in student performance, using knowledge specific to the learning needs of students with hearing loss.

Content Category 5- Adaption to General Health Problems and Their Treatment

ABP Content Specs for Developmental-Behavioral Pediatrics Fellow Collaboration Project

Prepared by Adam Langenfeld, MD/PhD, University of Minnesota DBP Fellow

Reviewed by Andrew Barnes, MD, MPH, University of Minnesota DBP Fellowship Director

5. **Adaptation to General Health Problems and Their Treatment**
   A. Acute illness
      1. Know the impact of a child's acute illness on the child's behavior and the family's functioning
      2. Understand child and family factors that might moderate the stress reaction during an acute illness
      3. Identify sources of family stress during a child's acute illness
      4. Understand methods of prevention and management of behavioral problems that develop during or immediately after an acute illness
      5. Know the sequence of development of children's concepts of illness causation
   B. Chronic conditions
      1. Understand the different strategies of clinical management that lead from a non- categorical approach to special health care needs and a diagnostically specific approach
      2. Understand the benefits and limitations of using a non-categorical approach to special health care needs in public policy
      3. Know the sources of stress for children who require medical technology and for their families
      4. Understand the functional domains that may be affected in children with chronic illness or disabilities
      5. Understand the international classification systems related to developmental- behavioral pediatrics
      6. Know how various demographic factors are related to the prevalence of disabilities in US children (eg, age, ethnicity, socioeconomic status
      7. Differentiate between major depression versus depressed mood secondary to adjustment to a chronic condition
      8. Know the factors that affect the social relationships of children with special health care needs
      9. Understand the benefits of the inclusion of children with disabilities or chronic illness in mainstream schools and community activities
      10. Know appropriate management to support parents and siblings of children with special health care needs
      11. Know the factors (child, family, and societal) that promote resilience to the stresses of chronic health conditions
      12. Know how to counsel parents on dividing responsibility for day-to-day management of a chronic illness between the parent and the child
      13. Know the impact of painful procedures on children with chronic illnesses
      14. Know how to minimize the impact of painful procedures on children
      15. Understand the difficulties associated with the inclusion of children with disabilities or chronic illnesses in mainstream schools and community activities
      16. Know that the prevalence of behavioral health disorders is increased in children with special health care needs

17. Know the effects of chronic health care conditions on child development
18. Know how to counsel families experiencing stress related to a child's chronic condition

C. Hospitalization
   1. Know how to advise families about behavioral problems that are commonly seen after hospitalization
   2. Know how to prepare a child for a planned hospitalization
   3. Know appropriate management for a child who has behavior problems after hospitalization or during prolonged hospitalization
   4. Know the benefits of therapeutic play programs for children in hospitals
   5. Know how to advise families and health care providers about strategies to help children better cope with hospitalization and/or medical procedures.

D. Terminal illness
   1. Understand differences in children's responses to life-threatening illness at various age levels
   2. Know how to plan appropriate psychosocial support for a child or adolescent with a terminal illness
   3. Know how to plan appropriate psychosocial support for the family of a child with a terminal illness
   4. Differentiate between acute grief and clinical depression

E. Death
   1. Understand the development of children's concepts of death and the psychosocial implications of their perceptions
   2. Understand the factors that may affect children's concepts of death
   3. Know appropriate advice for parents on how to explain to children of various ages the death of a family member
   4. Understand the impact on children of the death of a peer
   5. Know how to advise families on whether a child should attend a funeral
   6. Understand the variations in response of children at various age levels to the death of a family member

F. Adherence
   1. Differentiate between adherence and compliance
   2. Identify child factors that affect likelihood of adherence to a complex medical or behavioral plan
   3. Know how to develop a management plan to promote adherence to therapeutic regimens in children and adolescents at various developmental levels
   4. Identify family factors that affect likelihood of adherence to a complex medical or behavioral plan
   5. Identify community factors that affect likelihood of adherence to a complex medical or behavioral plan

G. Vulnerable child
    1. Know the hallmark features of the vulnerable child syndrome
    2. Be able to provide advice to parents to prevent the development of the vulnerable child syndrome in the aftermath of a child's life- threatening illness
H. Impact of treatments for chronic conditions on development/behavior
    1. Know the behavioral effects of commonly used medications
    2. Recognize behavioral effects of sympathomimetic medications
    3. Recognize the behavioral effects of anti-epileptic drugs
    4. Understand the developmental and behavioral effects of central nervous system ventricular shunts
    5. Understand the developmental and behavioral effects of cranial radiation
    6. Understand the developmental and behavioral effects of the anesthetic and bypass/circulatory arrest procedures used during cardiac surgery in infants
    7. Recognize the behavioral effects of chemotherapeutic drugs
    8. Recognize the long-term developmental and behavioral outcomes of survivors of childhood cancer

Adaptation to General Health Problems and Their Treatment

Acute illness

1. Know the impact of a child's acute illness on the child's behavior and the family's functioning
    a. **Encounters with Children Chapter 26**
    b. "An understanding of a child's perspective on the acute illness visit is crucial for two reasons. First, it generates age-appropriate ways to communicate with children, a core pediatric value. Equally important is the clinician's recognition that *the experience of the office visit itself shapes the way a child learns about the body, physical symptoms and illness and the healing process*." – page 650
    c. Clinicians must recognize that each acute illness is an opportunity to evaluate how an individual family copes with illness and be willing to provide interventions to address these underlying coping mechanisms

Encounters with Children, Chapter 26, Table 26-1

Child's Experience	**Parent's Experience**
Physical discomfort of the illness and treatment	Emotional responses: guilt, fear, anger, depression, lethargy
Fantasies about illness	Loss of social contacts
Restrictions (bed rest, diet)	Decreased or altered sensory input
Change in relationship with parents (indulgent or hostile)	Increased childcare responsibilities
Worry about body integrity	Expense of illness
Fear of serious illness	Interference with employment Sleep deprivation/fatigue
Decreased recreation	Effects on marital relationship
Fear, anger, guilt	Depression Distortions and misconceptions

2. Understand child and family factors that might moderate the stress reaction during an acute illness
    a. **Zuckerman Chapter 53** - Grief, Resiliency, and Coping in Children and Families Facing Stressful Circumstances
    b. "When stress affects individuals, perceptions, expectations, and social context play a role in its effects. For example, a child living in an unstable home may respond differently to a parent's angry outburst than a child living in a more secure and predictable environment. Child temperament and interpersonal factors make the experience of stress more complicated than simply stimulus and response." – Zuckerman page 276
    c. "Resiliency describes the quality in stress-resistant individuals that protects them from vulnerability in the face of environmental risk, including the capacity to successfully and meaningfully adapt to stress. Although sometimes presented as an individual trait, a closer look finds three domains of factors that contribute

to resistance to stress: (1) positive personality dispositions, (2) a supportive and nurturing family milieu, and (3) advantages coming from a thriving social support system." – Zuckerman page 276
        i. Two pathways – the Strengthening/"Steeling" Pathway and the Sensitization Pathway
    d. "Children without a home are more likely to suffer from accompanying conditions of food insecurity, malnutrition, and chronic disease." – Zuckerman pg. 280
    e. "Respecting a child and family's cultural values, beliefs, and attitudes is essential in facilitating communication and cooperation. Ask about cultural interpretations of medical and social issues. 'What do you call this problem? What do you think caused it? How do you treat it? What do you expect the treatment to do?'" – Zuckerman page 7
    f. "The World Health Organization (WHO) defines social determinants of health as the conditions in which people are born, grow, work, live, and age and the wider set of forces and systems shaping the conditions of daily life" – Zuckerman page 50
    g. **Encounters with Children Chapter 3** – cultural viewpoints on illness
    h. **Voight Chapter 3**, page 30
    i. **Zuckerman Chapter 10** – cultural aspects of parenting
3. Identify sources of family stress during a child's acute illness
    a. "Children without a home are more likely to suffer from accompanying conditions of food insecurity, malnutrition, and chronic disease." – Zuckerman pg. 280
    b. "These office visits may reflect a parent's perspective of this child as fragile and vulnerable, or she may be growing up in a family where somatic complaints for minor problems among other family members are frequently brought to medical attention… chronic stress or acute situational stressors may trigger the onset of physical or emotional symptoms that are repeatedly brought to medical attention." – Encounters page 651
4. Understand methods of prevention and management of behavioral problems that develop during or immediately after an acute illness
    a. **Encounters with Children, Chapter 26, Table 26-3** – TEACHER Method
        i. Method for enhancing communication with pediatric patients and their parents – help to communicate about acute illness visit to help prevent further problems in behavior
    b. **Zuckerman Chapter 53** – Pediatrician's Role in Times of Significant Distress
        i. Model and encourage open, honest communication about acute illness
        ii. Coach caregivers in age-appropriate explanations
        iii. Be honest without overwhelming
        iv. Provide anticipatory guidance
        v. Maintain routines
        vi. Expect behavioral changes to occur
        vii. Encourage emotional expression

viii. Connect with community resources
ix. Refer to mental health providers

c. **Voight Chapter 4** – Primary Care Management of Behavioral Problems

5. Know the sequence of development of children's concepts of illness causation

Encounters with Children, Chapter 26, Table 26-2

Age	**Understanding**	**Examples**
4-6 years	Circular, magical, or global responses	"I got a boo-boo on my head because I didn't eat my soup!"
6-8 years	Concrete, rigid responses with a "parrot-like" quality; little comprehension by the child; enumeration of symptoms, actions, or situations associated with illness	"The cough is a bug that tickles my throat and makes me stay home and in bed. The bug can't play with my friends."
8-11 years	Increased generalization, with some indication of the child's contribution to the response; the quality of invariant causation remains	"I get these headaches sometimes because I fight with my sister and won't let her play with my things"
11-13 years	Beginning use of an underlying principle; greater delineation of causal agents of illness	"Kids get sick when germs like a virus get into their body. It's the virus that makes us feel lousy, causes a fever and makes us want to sleep more."
Over 14 years	Organized description of mechanism(s) underlying illness and recovery; abstract principles	"Last year I had mono along with three of my friends. I know it's caused by a virus that makes you sleepy and not want to eat and zaps energy. But my friends got better quickly. I stayed out of school for a month. It seems that the mono virus was harder on my body and tougher to get rid of. My mom thinks it's because we've had a lot of stress in the family. She may be right because I know I get better quicker from a cold when things at home are cool."

<u>Chronic conditions</u> (see **Carey, Chapter 35**)

6. Understand the different strategies of clinical management that lead from a noncategorical approach to special health care needs and a diagnostically specific approach
    a. "Many pediatric tertiary care centers offer either primary care for patients with CYSHCN (children and youth with special healthcare needs) and CMC (children with medical complexity) or secondary care for patients of community-based providers. Studies of families who obtain their care from this specialized patient-centered tertiary medical home are more likely to encounter expanded care teams, including care coordinators, social work support, and community health workers with experience in complex care and community-based services." – Zuckerman pg. 184

b. **Zuckerman Chapter 36** has discussion about managing the care of a patient with complex healthcare needs, including how to efficiently organize an outpatient visit
c. "Providers also identify a number of barriers including inadequate time for patient encounters, metrics for productivity that don't allow for the extra time needed for complex care visits, insufficient reimbursement for the extra administrative time required between visits, and very often, a lack of support services to offer comprehensive care management." – Zuckerman page 184
d. Further, for children with less common chronic conditions, additional factors negatively impact care including unfamiliar diagnoses with a dearth of practice guidelines, fragmented care that may involve large numbers of subspecialists and caregivers, dysfunctional communication systems and poor interoperability between electronic health records, polypharmacy, multiorgan system involvement, and family systems that may be stressed and overwhelmed. – Zuckerman page 184
e. Building efficiency and innovation for complex visits
    i. Organize your system – use quality improvement to pilot innovations
    ii. Special scheduling – alternative iterations or scheduling encounters with longer appointments, integrate with subspecialists
    iii. Use best practices of AAP Patient-Centered Medical Home

Organizing the patient visit (Zuckerman page 184-185)

Previsit

- Create a rough outline of your agenda in an editable Shared Plan of Care (SPoC) based on guidelines and medical research on diagnosis. Store in an easy to access, editable page of your electronic medical record (EMR).
- Include a section for your medical goals, parent/guardian and patient goals as well.
- The SPoC is helpful for subspecialists, covering providers, school nurses, and family. It is also a great way to remind you about medical details and family goals before the next visit.
- Access, review, and update SPoC with interval procedures, subspecialty, and admission notes.
- Determine the best way to communicate or query subspecialists to clarify concerns. Electronic means are often easier than a phone call.
- For the family, consider implementation of a previsit checklist to prompt provider of the current needs and concerns, including symptoms, treatment plan changes, refill and durable medical equipment needs, or communication with other medical, school-based, or community-based team members.

The outpatient visit

- Before the visit, consider a rough outline of your agenda for this visit: Consider templated and reusable plain language, discharge instructions of family needs written in checklist fashion that may outline the next steps of care and/or build in transparency and accountability for all concerned (including the patient!). Ideally, offer an SPoC when appropriate, and update it at least every 6 months.

- Templates: Consider creating or reaching out to others about reusable smart phrase templates for the outpatient visit in the EMR that contain critical information based on diagnosis and can be edited and placed in an easy-to-access part of the EMR. A box format may be easier to navigate than a paragraph.
- Create or borrow action plans, medical orders for life-sustaining treatment forms, guardianship paperwork, medical authorizations for school medication use, and durable medical equipment prescription information on formulas, diapers, shower chairs, or hospital beds.
- Anticipatory guidance: Little is published on appropriate anticipatory guidance for children based on their diagnosis. Modify the anticipatory guidance you offer these families to ensure that it is tangible to their circumstance. Become available with resources in your community and have information and Web sites available for families. Learn about local Special Olympics or programs at your local YMCA.
- Consider safety recommendations: Parking placard for children who may bolt, shoe ID, or MedicAlert applications, police programs that may keep track of high-risk patients, contact with emergency medical service to ensure they understand do not resuscitate, medical needs, home locks for windows and doors. Consider creating an editable safety action plan for seizures, G-tube issues, and shunt issues that can be shared with others.
- Psychosocial needs: With high rates of mental health issues for both patients and parents, have a plan to offer support, connections to others living with same issues, and mental health services to these families.

Post-visit.

- Catalog and use a secure means to track and assign tasks for care management.
- SPoC/after-visit summary: A plan of care is jointly developed and shared among the primary care provider, the CYSHCN, their family, and the broader health care team. It identifies the strengths and needs of the child and family and is routinely evaluated and updated in partnership with the family (at least) every 6 months; and clearly identifies all entities that participate in a child's care coordination activities and the family's shared goals. In the emerging models of accountable care organizations, increasingly patient engagement and family activation is considered a key measure of performance or as a quality metric of a practice. Electronic health record-based plans of care, with shared access with caregivers and eligible adolescents, may be an area where these aspects can be measured and where accountability for following through on next steps of treatment—collaboratively by providers or by families—may be monitored.

Encounters with Children, Chapter 26, Table 26-4

Approach	**Action**
Recognition of a chronic disorder	Help the child and family understand fluctuating symptoms, good days and bad days, amelioration of symptoms (vs. cure)
Treatment that should fit the pattern of the child's and family's daily activities	Use knowledge about family life, family members, schedules, roles, decision-making, who provides care at various times
Assisting the child and family in identifying major concerns, priorities in care and outcome	Child, family, and clinician have shared agenda. Ask "What would you like to see changed?" Initially

	address only three concerns
Valuing and building on small successes	Set small goals, specific measures of progress
Effective use of a clinician's - Skills - Patience - Persistence	 Help child and family articulate problems, strengths, and goals Set realistic goals View the illness as chronic disorder for biological and psychosocial aspects of illness Plan follow-up visits, phone calls, periodic assessment

7. Understand the benefits and limitations of using a non-categorical approach to special health care needs in public policy
    a. "In 1998 the American Academy of Pediatrics (AAP) defined children with special health care needs (CSHCN) as those who have or are at increased risk for a chronic physical, developmental, behavioral, or emotional condition and who also require health and related services of a type or amount beyond that required by children generally. In 2009-2010 the Maternal and Child Health Bureau reported that there were 11 million children or 15% of all children in the United States in the category of children and youth with special health care needs (CYSHCN)." – Zuckerman pg. 183
    b. In 2011, a subcategory, called children with medical complexity (CMC), was identified and defined as patients with chronic conditions, multiple family-identified service needs, functional limitations requiring medical equipment, and extraordinarily high health care use and cost – Zuckerman page 183
    c. The growing number of CYSHCN has outpaced a consistent national strategy for managing their multifaceted needs – this population has been identified as a priority for care innovation and quality improvement
8. Know the sources of stress for children who require medical technology and for their families

Zuckerman Chapter 36, Table 36-1

Table 36-1 • DURABLE MEDICAL EQUIPMENT NEEDS

1. **Cardiovascular/respiratory devices**: oxygen and vital sign monitoring, ventilators, nebulizers, blood pressure monitors, tracheostomy supplies, and suction pump and supplies

2. **Feeding and Nutrition Kitchen accessories:** feeding chair or high chair, special bottles, spoons, G-tube, feeding pump, bags, tubing, special formula, blender

3. **Self-care/hygiene:** shower chair, commode, shower mat, diapers and pull-ups, shower hand bars, latex-free gloves, chux, wipes

4. **Bedding/sleep:** hospital bed, bed side rails, video baby monitor

5. **Positioning/mobility**: wheelchair, stroller, walkers or crutches, gait trainers, standers, braces, orthotics, prosthetics, adaptive car seat, recreational, home and vehicle modifications (ramps, integration of special seating or wheelchair)

6. **Adaptive tech**: communication devices, PECs, adaptive computer or tablet attachments, low-vision or low-hearing adaptive equipment

9. Understand the functional domains that may be affected in children with chronic illness or disabilities
    a. "In 1998 the American Academy of Pediatrics (AAP) defined children with special health care needs (CSHCN) as those who have or are at increased risk for a chronic physical, developmental, behavioral, or emotional condition and who also require health and related services of a type or amount beyond that required by children generally." – Zuckerman pg. 183
    b. **Voight Chapter 7** – Developmental assessment and brief examples of delays across different domains

Zuckerman Chapter 53, Table 53-1

Table 53-1 • IMPACT OF STRESS ON CHILDREN ACROSS AGE RANGES

Age Range	Cognitive Symptoms	Affective Symptoms	Behavioral Symptoms	Physiological Symptoms
Very young children (0-33	Demonstrate poor verbal	Exhibit sadness Act withdrawn	Irritability Engage in attention-	Change in appetite; poor feeding

3 y)	skills Exhibit memory problems Demonstrate developmental delays	Exhibit anxiety/fearfulness; develop new fears or anxieties Increased crying; inconsolable crying Flat or blunted affect Intense/prolonged separation distress	seeking Exhibit regressive behaviors Elevated startle response Exhibit aggressive behaviors Head banging	Low weight gain Digestive problems Sleep disturbances Develop rashes; skin irritability
Preschool-age children (3-6 y)	Difficulty focusing; inattention Exhibit memory problems Compromised social skills	Feelings of guilt or shame Exhibit sadness Act withdrawn Exhibit anxiety Lack self-confidence Exhibit separation distress	Irritability Engage in attention-seeking Exhibit regressive behaviors Elevated startle response Exhibit aggressive behaviors Increased temper tantrums Hyperactivity	Change in appetite Somatic complaints (e.g., stomachaches, headaches) Sleep disturbances Nightmares Enuresis/encopresis (after acquisition of skill)
School-age children (6-12 y)	Difficulty focusing; inattention Exhibit memory	Feelings of guilt or shame Exhibit sadness Act withdrawn Exhibit anxiety	Irritability; moodiness Elevated startle response Exhibit	Change in appetite Somatic complaints (e.g., stomachaches, headaches)

	problems Compromised social skills Decreased school performance	Lack self-confidence Feelings of anger Emotional avoidance; flat affect	aggressive behaviors Hyperactivity School avoidance Increased oppositional behaviors; noncompliance	Sleep disturbance Nightmares
Adolescents (13 y and up)	Difficulty focusing; inattention Exhibit memory problems Decreased school performance	Feelings of guilt or shame Exhibit sadness Act withdrawn Exhibit anxiety Feelings of anger Emotional avoidance; flat affect Exhibit depression Feelings of embarrassment	Irritability; moodiness School avoidance/failure Suicidal ideation Sexual acting out Increased oppositional behavior; noncompliance Engage in high-risk behaviors (e.g., truancy, substance use) Antisocial behaviors Relational difficulties	Change in appetite Somatic complaints (e.g., stomachaches, headaches) Sleep disturbance Nightmares

10. Understand the international classification systems related to developmental-behavioral pediatrics
    a. International Classification of Functioning, Disability, and Health (ICF) and ICF-CY (children and youth)

b. Standard language for children's health, education, and social services; used to record the characteristics of child development, environmental factors affecting child development, and developmental delays
    i. https://pubmed.ncbi.nlm.nih.gov/16100508/

Fig. 1. Interactions between the components of ICF

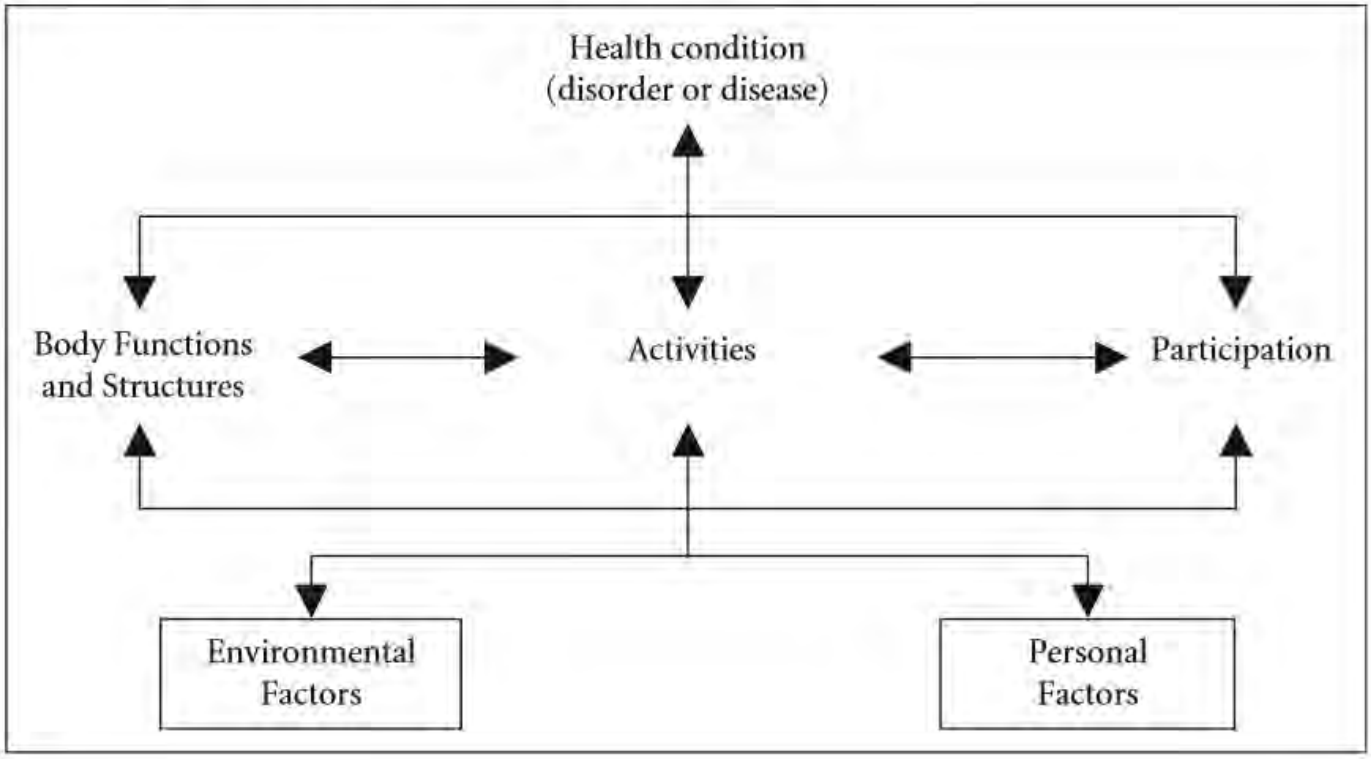

*Image taken from ICF-CY handbook (*https://apps.who.int/iris/handle/10665/43737)

11. Know how various demographic factors are related to the prevalence of disabilities in US children (eg, age, ethnicity, socioeconomic status)
    a. Parents of CMC experienced more difficulty with childcare, employment, and parenting skills. Financial stress in addition to these social determinants of health are particularly challenging with families from lower socioeconomic or underserved communities, known in the literature as "double jeopardy." – Zuckerman pg. 183
    b. "Children without a home are more likely to suffer from accompanying conditions of food insecurity, malnutrition, and chronic disease." – Zuckerman pg. 280
    c. "The effects of (1) traditional cultural beliefs, (2) poverty and access to material goods, and (3) being a part of a racial or ethnic minority are distinct but often interlinked." – Zuckerman page 46
    d. "The World Health Organization (WHO) defines social determinants of health as the conditions in which people are born, grow, work, live, and age and the wider

set of forces and systems shaping the conditions of daily life" – Zuckerman page 50

e. Encounters with Children Chapter 3

f. **Voight Chapter 1**, page 9

g. **Voight Chapter 3**, page 30

12. Differentiate between major depression versus depressed mood secondary to adjustment to a chronic condition
    a. "CYSHCN and CMCs have up to four times higher rates of psychiatric comorbidities, such as anxiety and depression, than their healthy, neurotypical peers. Consequently, screening for depression and emotional health concerns remains a priority for care team members in a patient's medical home, with established pathways to connect children and household members of concern with behavioral health supports." – Zuckerman pg. 183
        i. Cognitive behavioral therapy may be a very helpful approach for most verbal children.
    b. **Voight Chapter 18** – Mood Disorders
    c. **Zuckerman Chapter 40 –** Depression
13. Know the factors that affect the social relationships of children with special health care needs
    a. "Linking children with others via support groups, camps, organizations, and Web sites are helpful ways to ensure they feel connected to others." – Zuckerman pg. 184
    b. Voight Chapter 23
14. Understand the benefits of the inclusion of children with disabilities or chronic illness in mainstream schools and community activities
    a. Voight Chapter 23
15. Know appropriate management to support parents and siblings of children with special health care needs
    a. "In addition to the amount of time involved and the extra care required for families who care for CMC, parents consistently convey the burden and stress they face from inadequate and fragmented care coordination, frequent provider visits, financial difficulties, complex and time-consuming bureaucracy, and household and relationship stress, while compensating for missed days at work or caretakers' inability to work." – Zuckerman pg. 183
    b. "Depression, and/or the so-called medical post-traumatic stress disorder, and other behavioral and emotional disorders have higher prevalence among the adult caretakers of CYSHCN and CMCs, and research suggests that many parents are unsuccessful or unable to seek care for themselves." – Zuckerman pg. 183
    c. "Availabilities of social media, Web sites, chronic condition online forums, and special education Listservs offer newer ways to communicate with providers, peers, and virtual communities to access medical information." – Zuckerman pg. 183
16. Know the factors (child, family, and societal) that promote *resilience* to the stresses of chronic health conditions

a. "Resiliency describes the quality in stress-resistant individuals that protects them from vulnerability in the face of environmental risk, including the capacity to successfully and meaningfully adapt to stress. Although sometimes presented as an individual trait, a closer look finds three domains of factors that contribute to resistance to stress: (1) positive personality dispositions, (2) a supportive and nurturing family milieu, and (3) advantages coming from a thriving social support system." – Zuckerman pg. 276
b. Encounters with Children, Chapter 3
c. **Zuckerman Chapter 53** - Grief, Resiliency, and Coping in Children and Families Facing Stressful Circumstances
d. **Carey Chapter 34, page 341**

17. Know how to counsel parents on dividing responsibility for day-to-day management of a chronic illness between the parent and the child
18. Know the impact of painful procedures on children with chronic illnesses
    a. "Pain is defined by the International Association for the Study of Pain as 'an unpleasant sensory and emotional experience associated with actual or potential tissue damage or described in terms of such damage.' This definition implies that pain has two components—a neuro-physiologically determined sensation that results from stimulation of nociceptors and the interpretation of that stimulus, which is impacted by a host of genetic, personality, cognitive, developmental, experiential, environmental, and emotional factors." – Zuckerman page 79
        i. Untreated pain may inhibit immune function, induce stress hormone release, increase blood pressure, inhibit healing due to immobility, and decrease pain threshold, which subsequently results in hyperalgesia and allodynia
        ii. Inadequate pain control in children with illness may cause worsening procedure anxiety
    b. "For many children with chronic conditions who have frequent doctors' appointments, inadequately treated procedure pain may result in increased anxiety about subsequent medical encounters and reluctance to attend of medical visits." – Zuckerman page 79
19. Know how to minimize the impact of painful procedures on children
    a. "Developmentally appropriate explanations of procedures (e.g., immunizations, venipuncture, operations) are critical, and rehearsal of the sequence of events helps to ease a child's anxieties. The parent's presence during a procedure usually lessens the stress to the child; separation should generally be avoided. Be truthful if a procedure will cause any pain. Offer choices to the child as possible (which arm to draw blood from, what color cast he or she would like)." – Zuckerman page 8
    b. **Zuckerman Chapter 19** – Pain
        i. General principles include preventative approach, interdisciplinary approach (pharmacology, cognitive-behavioral, physical, and integrative approaches)

ii. "Needlesticks are extremely troubling for children and parents, and needle pain should be addressed in all inpatient and outpatient settings. Evidence-based approaches include comfort positioning, use of topical anesthetics, distraction and other mind-body strategies, and breastfeeding or use of sucrose for infants." – Zuckerman page 80
iii. Minor alterations to the environment or basic calming techniques may be the only approaches necessary in procedures with limited pain magnified by anxiety
iv. Behavioral/cognitive approaches
    1. Parental presence (when not anxious or minimizing pain)
    2. Age and developmental-stage-appropriate preparation
    3. Visual imagery/distraction/hypnosis
v. Physical approaches (massage, heat, cold, pressure, vibration)
vi. Pharmacologic approaches
    1. Local anesthetics (EMLA, LMX, LET gel, J-tip, cooling sprays)
    2. Drugs for mild-moderate pain (Acetaminophen, NSAIDS)
    3. Drugs for moderate-severe pain (opioids – tramadol, oxycodone, morphine)
    4. Adjunctive drugs. "Drugs in this category include anticonvulsants (**pregabalin** and **gabapentin**) and antidepressants (**amitriptyline** and **duloxetine**), which have efficacy against the pain of nerve injury and neuropathic pain. This type of pain, which is often opioid resistant, has unique characteristics and is often described as burning, shooting, or electric. Treatment of comorbid anxiety should be considered using medications such as **benzodiazepines** and SSRIs. **Benzodiazepines** can also be useful to treat muscular spasms associated with pain. Management of sleep disorders using **melatonin**, **amitriptyline, trazodone,** or **zolpidem** may also be necessary."

Zuckerman Chapter 19, Table 19-1

Table 19-1 • DOSING DATA FOR ACETAMINOPHEN AND NONSTEROIDAL ANTI-INFLAMMATORY DRUGS (NSAIDS)

Drug	Usual Adult Dose	Usual Pediatric Dose	Comments

Oral NSAIDs			
Acetaminophen	650-1000 mg q 4 h	10-15 mg/kg q 4 h	Acetaminophen lacks the peripheral anti-inflammatory activity of other NSAIDs; available as oral suspension.
Ibuprofen	400-600 mg q 4-6 h	10 mg/kg q 4-6 h	Available as several brand names and as generic; available as oral suspension.
Naproxen	500 mg initial dose followed by 250 mg q 6-8 h or 500 mg q 12 h	For patients >2 y of age 5 mg/kg q 12 h	Dose is expressed as naproxen base; 200 mg naproxen is equivalent to 220 mg naproxen sodium. Available as oral suspension.
Celecoxib	100-200 mg q 12 h	For patients >2 y of age ≥10 kg to ≤25 kg, 50 mg q 12 h >25 kg 100 mg q 12 h	COX-2 inhibitor may cause less bleeding and gastritis. Useful for children with thrombocytopenia.

Table 19-2 • DOSING DATA FOR OPIOID ANALGESICS

Drug	Route	Initial Pediatric Dose	Initial Adult Dose
Morphine	IV/SC PO/SL/PR	0.05-0.1 mg/kg q 2-4 h 0.15-0.3 mg/kg q 2-4 h	5-10 mg q 2-4 h 10-15 mg q 2-4 h
Hydromorphone	IV PO/SL	0.015 mg/kg q 3-4 h 0.05 mg/kg q 3-4 h	0.2-0.6 mg q 2-4 h 1-2 mg q 3-4 h
Fentanyl	IV/IN	1-2 µg/kg q 10-60 min	25-75 µg q 10-60 min
Oxycodone	PO/SL	0.1-0.2 mg/kg q 4-6 h	5-10 mg q 4-6 h
Tramadol	PO/SL		50-100 mg q 6 h

20. Understand the difficulties associated with the inclusion of children with disabilities or chronic illnesses in mainstream schools and community activities
21. Know that the prevalence of behavioral health disorders is increased in children with special health care needs
    a. CDC accumulates data on mental health diagnoses in children (for example, see: https://www.cdc.gov/childrensmentalhealth/data.html)
22. Know the effects of chronic health care conditions on child development
    a. Zuckerman Chapter 36 – Chronic Conditions
        i. Children with chronic problems have diminished family functioning, more school absences, and less participation in community activities compared with healthy peers. – Zuckerman page 183
    b. **Carey Chapter 35** – Chronic Health Conditions
        i. Infants – illness may affect basic growth, cause failure to thrive, bring excessive fatigue; children may be less responsive due to physical or neurocognitive deficits; children may require excessive medical care or procedures, increasing risk of vulnerable child syndrome
        ii. Toddlers – may display slowed development of early independence, leading to diminished resilience, as well as delayed developmental milestones; may limit task mastery

iii. School-age – frequent school absences can interfere with learning and socialization; normal separation may be difficult; children with visible conditions may be stigmatized
iv. Adolescents – forced dependence on caregivers may limit development of individual identity, separation from parents, and sexual development; this may lead to rebellion and denial (e.g. a child with type 1 diabetes who stops taking insulin and develops DKA); sexual development may be affected directly by condition (e.g. myelomeningocele) or fears of inadequacy (e.g. cystic fibrosis)

23. Know how to counsel families experiencing stress related to a child's chronic condition
    a. **Zuckerman Chapter 53** - Grief, Resiliency, and Coping in Children and Families Facing Stressful Circumstances
        i. "When stress affects individuals, perceptions, expectations, and social context play a role in its effects. For example, a child living in an unstable home may respond differently to a parent's angry outburst than a child living in a more secure and predictable environment. Child temperament and interpersonal factors make the experience of stress more complicated than simply stimulus and response." – Zuckerman Chapter 53, page 276
        ii. Section on discussing "Family member diagnosed with life-threatening illness"
    b. "The caregiving and emotional burden of neurodevelopmental disorders including seizures, autism spectrum disorder, cerebral palsy, and global developmental delay also have important implications for family and marital functioning." – Zuckerman page 183
    c. **Carey Chapter 35** – Chronic Health Conditions

<u>Hospitalization</u>

24. Know how to advise families about behavioral problems that are commonly seen after hospitalization
    a. Carey, Chapter 33

Carey, Chapter 33, Table 33-2 – Common Behavioral Symptoms after Hospitalization or Surgery

Regression (thumb sucking, bed-wetting, baby talk)	Sleep disturbances
Conflicts about eating	Aggressiveness, acting out
Increased dependency	Fear of physicians or nurses
School phobia	Depression
Decreased attentiveness; academic failure	Encopresis

b. Encounters with Children, Chapter 26, page 664
i. Infant – few behavioral changes after hospitalization if connection to family is re-established. During hospitalization, a young infant may show behavioral (fussiness, inconsolability, sleep disturbances) or physiological reactions (vomiting, constipation, poor weight gain, poor feeding)

ii. Toddler – behavioral changes in a toddler are more dramatic and require a longer recovery phase. Normal toddlers who experience abrupt or prolonged separation from parents undergo a progression of behavioral patterns
    1. **Protest** reaction – tantrums, refusal to eat, aggressive behavior
    2. **Despair** – a quiet, withdrawn, sad appearance
    3. **Denial** – the previously depressed, despairing toddler now appears outgoing and responsive to everyone indiscriminately and shows no differential response to familiar care providers (indicates severe stress for the child)
    4. **Red flags** – favorite patient of every nurse, no objection to examination, extraordinary effort to engage with every passerby, withdrawn/"too good"
iii. Preschooler – may utilize imagination, verbal questioning, magical thinking to understand and deal with acute stresses of strange environment
    1. Can tolerate longer absences of parents
    2. Magical thinking may cause distortions of causality; may feel personally responsible for illness (**explanatory model** that may be resistant to change despite evidence)
iv. School-aged – can develop peer group friendships and attachments to nurses, physicians, other personnel to buffer feelings around parental separation
    1. Better verbal skills, understanding of causality – more mental control of situation (e.g. understands basic causes of illness and logic of treatment)
    2. Hospital experience may be stressful due to lack of control and feelings of guilt/anger, leading to behavioral manifestations of depression, anxiety, or behavioral regression
    3. Understanding *loss of control* as a model for behaviors may be useful in helping child understand and navigate stressful experience
v. Adolescent – hospitalization may represent quest for personal identity separate from family
    1. May experience loss of idealized self as result of disease, along with abrupt separation from peer group and activities, leading to significant distress
    2. Disease and invasive procedures may distort body image and future plans
    3. Age-appropriate independence may be distorted by need for care, increased dependence, and disruption of social support

c. Children of all age groups may experience loss of developmental milestones, which is both a stress and protective strategy

i. Behavioral alterations may be an adaptive and healthy response to the experiences

d. Intensity and duration of developmental regression are affected by:
    i. Child's social and language developmental level
    ii. Personality and temperament
    iii. Previous styles of coping with new situations
    iv. Family's response to illness
    v. Fears and fantasies

25. Know how to prepare a child for a planned hospitalization
    a. "Anticipatory guidance about diagnostic/therapeutic procedures and an elective hospitalization help both the child and the parent. A preschool child's magical thinking, a school-aged child's concrete thinking, and an older school-aged child's emerging ability to understand causality moderate their response to illness and the clinician's language and content of information." – Zuckerman page 8
    b. "Hospitalization is associated with a sudden environmental change, loss of independence, and separation from primary caretakers. Provide the child with a description of the hospital environment and the procedures encountered. Keep some familiar toys, books, or attachment objects with the child if separation is unavoidable. Engage the assistance of a child life specialist to provide developmentally appropriate support if one is available." – Zuckerman page 8
    c. **Encounters with Children, Chapter 26, page 667**
        i. Provide parents and the child with age-appropriate explanation for hospitalization
        ii. For elective procedures, provide the child with a description of the hospital setting and the procedures that will be encountered before anesthesia
        iii. Encourage a rooming-in policy for parents and encourage family members to be present as much as possible
        iv. Hospitalize children in a pediatric setting that is appropriately designed for children
        v. Encourage parents to bring a few familiar toys and books or a "lovey"
        vi. Be truthful about procedures
        vii. Provide realistic options for the child and age-appropriate opportunities to participate in care
        viii. Identify sensory information, cognitive interventions, and coping strategies
26. Know appropriate management for a child who has behavior problems after hospitalization or during prolonged hospitalization
    a. Prepare parents for responses to hospitalization, including developmental regression (**Encounters with Children, Chapter 26, page 667**)
    b. See similar discussion for behavioral changes to acute illness, as above
    c. **Carey, Chapter 33**
27. Know the benefits of therapeutic play programs for children in hospitals

a. "In association with the emergence of hospitals and wards designed specifically for the needs of children, more attention has been given to the emotional needs and developmental capacities of children in hospitals… Family visiting and involvement, child life programs, school involvement and attention to psychosocial needs are now nearly routine" – Encounters page 662

28. Know how to advise families and health care providers about strategies to help children better cope with hospitalization and/or medical procedures.
    a. Encounters with Children, page 664
        i. Infant – maintain similar schedules, objects, and caretakers to minimize impact of changes in hospital setting; young infants will establish relationships with sensitive, consistent caretakers and build on these very quickly
        ii. Toddler – encourage rooming-in policy for parents, encourage family members to be as present as possible, bring familiar toys or "lovey", monitor for red flags, use age-appropriate language to explain procedures, utilize distraction methods (blowing bubbles, hand holding, singing, etc.), monitor for pain and use safe medications to eliminate or modify pain
        iii. Preschooler – Use age-appropriate language to discuss procedures and be truthful about procedures, encourage rooming-in policy for parents, encourage family members to be as present as possible, bring familiar toys or "lovey", utilize distraction methods, monitor for pain and use safe medications to eliminate or modify pain
        iv. School-aged – encourage participation in decision-making by providing concrete/specific options, encourage ongoing interaction with school/classmates through electronic communication/homework participation, use age-appropriate language to discuss procedures, utilize distraction methods, monitor for pain and use safe medications to eliminate or modify pain
        v. Adolescent – encourage participation in decision-making as much as child is interested/able to participate, encourage ongoing interaction with school/classmates through electronic communication/homework participation, give opportunities to discuss disease/hospital experience and its impact on personal identity, monitor for pain and use safe medications to eliminate or modify pain
    b. **Encounters with Children, Chapter 26, page 667** – general guidelines
        i. Provide parents and the child with age-appropriate explanation for hospitalization
        ii. For elective procedures, provide the child with a description of the hospital setting and the procedures that will be encountered before anesthesia
        iii. Encourage a rooming-in policy for parents and encourage family members to be present as much as possible

iv. Hospitalize children in a pediatric setting that is appropriately designed for children
v. Encourage parents to bring a few familiar toys and books or a "lovey"
vi. Be truthful about procedures
vii. Provide realistic options for the child and age-appropriate opportunities to participate in care
viii. Identify sensory information, cognitive interventions, and coping strategies

c. Provide parents with expectations for child's physical and psychological recovery

Carey, Chapter 33, Table 33-5 – Minimization of Stress

Before Hospitalization
Outpatient procedures, evaluation, and treatment whenever possible
Involvement of parents in decisions about procedures and hospitalization
Television, school, and community programs about illness and hospitals
Prehospitalization tours
Specific information about planned procedures and hospitalizations
Parents present as much as possible
During Hospitalization
Rooming-in for parents
Encourage visiting for siblings, peers, extended family
Child life program (recreation and therapeutic play)
Limited number of nurses and other caregivers
Child's choice of clothes, food, activities
Maximal mobility
Pain control
Procedures
Limited number
Preparation of child
Child's choice of when, where, and with whom
Limited waiting time
Parents accompany and support child
Hospital Structure
Rooming-in facilities
Play space
Waiting space near operating and recovery rooms
Comfortable accommodations for parents
Cheerful, child-oriented décor
Single- or double-bed rooms

Terminal illness

29. Understand differences in children's responses to life-threatening illness at various age levels

a. "Parents often seek advice about what to say and how to talk with their children about serious illness and death. Clinicians can serve as both a model to children and families during pediatric visits and a support to caregivers." – Zuckerman pg. 281

30. Know how to plan appropriate psychosocial support for a *child or adolescent* with a terminal illness
    - **a. Zuckerman Chapter 39**
        - i. Healthcare providers must attend to the immediate physical needs of these children
        - ii. Children at different developmental stages have different conceptual understandings of the meaning of death
        - iii. Many parents and clinicians are uncomfortable when children openly acknowledge an awareness of their impending death
        - iv. To the extent possible, children should be informed about their health status
        - v. Facilitating discussion about children's concerns often involves projective techniques such as play or picture drawing
        - vi. Clinicians are often anxious that they will not know what to say to a child who is dying
        - vii. Children must be allowed, even encouraged, to continue to have hope and to go on with their lives
        - viii. Many children and adolescents feel guilty and ashamed about their illness
        - ix. To the extent possible, children should be informed about and participate in the decisions regarding their healthcare
            1. Older children and adolescents may possess the intellectual and emotional maturity to allow them to play a significant role regarding critical and difficult decisions (e.g., whether to discontinue aggressive treatment).
    - **b. Voight Chapter 9**

Zuckerman Chapter 39, Table 39-1 and 39-2

Table 39-1 • GENERAL PRINCIPLES FOR PRACTITIONERS IN THE CARE OF DYING CHILDREN AND THEIR FAMILIES

Physical Context
Minimize physical discomfort and symptoms
Optimize pain management
Emotional Context
Provide an opportunity for the expression and sharing of personal feelings and concerns for both the children and their families in an accepting atmosphere
Tolerate unpleasant affect (e.g., sadness, anger, despair)
Social Context
Facilitate communication among members of the health care team and the children and their families
Encourage active participation of the children and their families in the treatment decisions and the management of the illness
Personal Context
Treat each child and family member as a unique individual
Form a personal relationship with the child and the family
Acknowledge your own feelings as a health care provider and establish a mechanism(s) to meet your personal needs

Table 39-2 • PRINCIPLES INVOLVED IN INFORMING CHILDREN ABOUT A TERMINAL ILLNESS OR IMPENDING DEATH

Inform the child over time in a series of conversations. During the initial conversation, it is important to convey that the child has a serious illness.
If the child asks directly whether he or she is going to die, initially explore the reason for the question and the child's concerns (e.g., "Are you afraid that you might die?" "What are you worried about?"). Do not provide false reassurances ("No, don't worry, you're going to be okay."), but always try to maintain hope ("Some children with your sickness have died, but we are going to do everything we can to try to help you get better.").
Focus initial discussions on the immediate and near future. Young children have a limited future perspective. Dying "soon" to them may mean minutes, hours, or days and not months or years.
Answer questions directly, but do not overwhelm the child with unnecessary details.
Assess the child's understanding by asking him or her to explain back to you what you have discussed.
Reassure the child of the lack of personal responsibility or guilt. For this reason, avoid using "bad" in the description of the illness (e.g., "You have a bad sickness.").

31. Know how to plan appropriate psychosocial support for the *family* of a child with a terminal illness
    a. **Zuckerman Chapter 53** – section on "family member diagnosed with life-threatening illness"
        i. Encourage clear, simple description of illness
        ii. Prepare children for visits
        iii. Avoid false statements that are offered to console
        iv. Encourage caregivers to model emotional expression and coping
        v. Attune to children's cues
    b. **Zuckerman Chapter 39** – parents
        i. Parents faced with the impending (or actual) death of their child may demonstrate shock, denial, anxiety, depression, or anger.

ii. The response of family members to the death may be affected by the duration of the illness.
iii. Members of the family may proceed with anticipatory grieving at different rates.
iv. As part of anticipatory grieving, family members and health care professionals may wish for the death of a terminally ill child.
v. Family members should be allowed, even encouraged, to continue to have hope and to go on with their lives.
vi. To the extent possible, parents should be informed about and participate actively in decisions regarding their child's care.
1. **The health care providers must provide families with clear professional recommendations and be willing to discuss alternate options when appropriate options exist.**
vii. Sudden or unexpected death requires an immediate recognition of the loss.
viii. Support systems for families of dying children are often hospital-based and frequently withdrawn at the time of the child's death.
ix. At the time of the child's death, parents and other family members should be offered assistance with immediate and pragmatic needs.
x. Families in grief may feel immobilized and incapable of making even simple decisions.

c. Zuckerman Chapter 39 – siblings
i. The needs of siblings are often neglected when a child in the family is dying
ii. The siblings should be included in receiving information about the child's health status and treatment plan and should participate to some extent in the provision of care for the ill child
iii. Siblings respond to the death with the same diversity of emotional responses as seen in adults

32. Differentiate between acute grief and clinical depression
a. Zuckerman Chapter 40 – Depression
b. Acute grief/bereavement – "Bereavement or adjustment disorders can be diagnosed when the onset of symptoms follows a significant life event or loss and do not meet criteria for MDD." – Zuckerman page 203
i. Definition – acute onset of depressive symptoms in response to a significant life event or loss that resolves over a period of time, usually by one year following the event. Grief is the emotional response and bereavement is the period of grief and mourning after a loss.
c. Clinical depression

Table 40-1 • SIGNS AND SYMPTOMS OF DEPRESSION

Depressed or irritable mood, loss of interests, or pleasure most of the day, nearly every day for at least 2 weeks. The symptoms must cause significant distress or impairment in social, academic, or other important areas of functioning and must be accompanied by changes in *at least four* of the following:	Feeling sad, blue, miserable, hopeless, irritable, "blah," cranky, edgy "Dysphoric mood"—sadness along with irritability; exaggerated sense of frustration over minor things Please note that this can either be subjective feeling or observation made by others (e.g., appears tearful all the time, looks sad all the time, nothing makes him/her happy, "not caring for things anymore") In milder forms of depression, functioning may appear normal but requires markedly increased effort
1. Poor appetite or weight loss/gain	In children, this can be seen as failure to make expected weight gain. Appetite loss or appetite increase nearly every day Changes of more than 5% of weight gain (if not dieting/exercising) is always concerning
2. Change in sleep patterns	Insomnia, frequent awakenings, or hypersomnia nearly every day
3. Change in the level of activity	Psychomotor agitation (pacing, moving a lot, cannot sit still, hand wringing) or retardation (slowed speech, slow body movements) Observable by others; not just subjective feelings
4. Fatigue or loss of energy	Feels tired all the time, even the smallest things perceived as needing great effort
5. Feeling of worthlessness or excessive and inappropriate guilt	Feeling as they cannot do anything right, "a failure," exaggerated sense of responsibility for the negative events, feeling as if they are "shame" for the family

6. Change in concentration or indecisiveness	Less able to focus in class, complete assignments, can present as a drop in school grades, cannot complete tasks that was able to in the past
7. Suicidal ideation	Recurrent thoughts or death and dying, thinking life would be better off without him/her, undeserving of life, unable to cope with the pain of depression

Death

33. Understand the development of children's concepts of death and the psychosocial implications of their perceptions
    a. **Zuckerman Chapter 53** – section on "death of a relative, friend, or other close acquaintance"
        i. Infants and toddlers (0-2 yrs): Do not have the cognitive capacity to understand death. Experience death as loss of an attachment figure
        ii. Young children (3-5 yrs): Do not understand finality or universality of death; view death as a changed state.
        iii. Latency-age children (6-11 yrs): Understand finality and universality of death.
        iv. Adolescents (12+ yrs): Demonstrate adult-like, nuanced understanding of death. Capable of wrestling with the abstract and philosophical aspects.
    b. **Carey Chapter 14, page 149**
34. Understand the factors that may affect children's concepts of death
    a. "The impact of a death on a child is dependent on many factors including the child's relationship with the deceased, the child's history of other losses, and the nature of the death. A child's developmental age also plays an essential role in how the child will understand and grieve the loss." – Zuckerman pg. 282
    b. "The primary modulator for any child's experience is his or her conception of death, which relates, in typically developing children, to their age" – Carey, pg. 149
    c. Caregiver ability to support the child following a death provides a tempering effect on the child's experience

Carey Chapter 37, Table 37-1 – Factors Influencing Expressions of Grief

Parents and Siblings	Parents	Brothers and Sisters
The manner of death	Parent-dead child relationship	Developmental status
Family cohesion	"Special meaning" of child	Individual temperament
Mutual support	Age of child	How parents grieve
Responses of friends, extended family	Individual temperament	Surviving sibling-child relationships
Physical and mental health	Quality of marriage	Surviving siblings' relationships
Access to resources	Parents' ability to show affection	Responses at school
Cultural: religious beliefs, traditions	Career demands, response of colleagues	

35. Know appropriate advice for parents on how to explain to children of various ages the death of a family member
    a. **Zuckerman Chapter 53**
        i. Infants and toddlers (0-2 yrs): Benefit from consistent, responsive caregiving during difficult times
        ii. Young children (3-5 yrs): Provide sympathetic reminders that deceased is not coming back. Provide support and reassurance that death is not their fault.
        iii. Latency-age children (6-11 yrs): Be responsive to questions and check in with them, even if they appear "fine" on the surface.
        iv. Adolescents (12+ yrs): Benefit from emotional and psychological availability of caring, supportive adults.
36. Understand the impact on children of the death of a peer

37. Know how to advise families on whether a child should attend a funeral
    a. **Zuckerman Chapter 53**
        i. Provide a clear explanation of the services – developmentally appropriate explanation of what will take place, who will be there, and what to expect about behaviors of others
        ii. Provide choice, if possible – children may be resentful when excluded or overwhelmed if compelled to participate
        iii. Find appropriate alternatives
        iv. Ensure access to a compassionate adult
        v. Find ways to involve children
    b. **Carey Chapter 14**
        i. Limit the amount of time spent at the funeral home for young children
        ii. Allow older children and adolescents to participate in the service in a way that assists the grieving process with a remembrance or reading of personal significance
38. Understand the variations in response of children at various age levels to the death of a family member

a. See Zuckerman pg. 282 for detailed description of concepts of death at different developmental stages
    i. Infants and toddlers (0-2 yrs): May exhibit symptoms of grieving, including changes in sleep/eating, increased irritability, heightened distress during transitions or separations.
    ii. Young children (3-5 yrs): May believe death is their fault (e.g. "Mommy died 'cause I told her 'go away!'") due to egocentrism.
    iii. Latency-age children (6-11 yrs): Seek more detailed information about the deceased, the cause of death, and impact of the death on the family. May appear emotionality unaffected as an attempt to maintain a sense of predictability and control.
    iv. Adolescents (12+ yrs): Prefer to process grief with peers rather than with adults.

Encounters with Children, Chapter 27, Table 27-1

Age	**Understanding**
Infants	Have no cognitive understanding of death. Death is the same as a significant separation.
0-2 yr	Respond to changes in routine, caregivers, emotional chaos of family situation Experience separation anxiety, irritability and regression
Preschoolers (3-5 yr)	Death is temporary, reversible, living under different circumstances Literal thinking about causes Link seemingly unrelated factors to the death (transductive reasoning) Use magical thinking to explain death
School age (6-8 yr)	Death is final, irreversible, but not universal See death as a person or spirit that catches you Death "catches" the elderly, disabled May be contagious Concerned with safety, predictability Need for details
Preadolescent (9-12 yr)	Adult understanding: death is final, irreversible, universal Disconnect between thinking and emotions Understand the biological aspects of death Interested in rituals, roles, ceremonies Understand causality: may feel guilty about contributing to it Intellectualize death: thoughts more available than feelings
Adolescent (13-18 yr)	Adult understanding: death is final, irreversible, universal Engage in high-risk activities (challenge own mortality) Understand existential implications of death Reject adult rituals and support May see some aspects of stigma

<u>Adherence</u>

39. Differentiate between adherence and compliance
    a. Adherence/compliance – degree to which a patient correctly follows medical advice

b. "It is not uncommon to find either poor adherence to a treatment or suboptimal adaptation to an illness after appropriate medications, advice, and education have been tried" – Encounters page 660

40. Identify child factors that affect likelihood of adherence to a complex medical or behavioral plan
41. Know how to develop a management plan to promote adherence to therapeutic regimens in children and adolescents at various developmental levels
    a. "Returning to the patient and parent agenda model, it is useful to discuss with the child and family their perceptions of the illness and their experiences with trying to solve specific problems" – Encounters page 660
    b. By reframing the individual problems of a child with a disease as family problems, a foundation for better compliance with any care plan or therapeutic effort can be developed

Encounters with Children Table 26-5

Issue	**Biomedical**	**Solution Building with Family**
Patient Role	"Sick patient", passive partner	Equal and active participants
Patient focus	Problems, dilemmas	Solutions
Expert	Clinician	Clinician, patient and family as problem solvers and solution builders
Encounter focus	Deficits, pathology of the individual; extended history of the problem	Strengths, competence of family system; brief history of the problem; detailed history of goals and solutions
Therapeutic focus	Clinician's decisions	Clinician, patient and family develop solutions

42. Identify family factors that affect likelihood of adherence to a complex medical or behavioral plan
43. Identify community factors that affect likelihood of adherence to a complex medical or behavioral plan

<u>Vulnerable child</u>

44. Know the hallmark features of the vulnerable child syndrome
    a. "The term *vulnerable child* is used to refer to children who have an increased, atypical, or exaggerated susceptibility to disease or disorder due to medical, socioeconomic, psychological, biological, genetic, and environmental risk factors." – Zuckerman pg. 444
    b. Initially described by Green and Solnit. "In this circumstance, a child with an imagined or real illness in early life is the targeted of altered attachment by her parents" – Encounters page 186
        i. Parents develop sense that child is susceptive to illness, injury, or loss
        ii. Child viewed as fragile, incapable of age-appropriate behavioral expectations
        iii. Perception of vulnerability leads to ongoing intra-familial stress, altered interaction between child and parent, inability to either allow age-appropriate autonomy or set limits
    c. **Zuckerman Chapter 87** – Vulnerable Children

d. **Carey Chapter 34** – Early Health Crises and Vulnerable Children

45. Be able to provide advice to parents to prevent the development of the vulnerable child syndrome in the aftermath of a child's life- threatening illness
    a. **Zuckerman Chapter 87** – Vulnerable Children
    b. **Carey Chapter 34** – Early Health Crises and Vulnerable Children

Carey Chapter 34, Table 34-3 – Prevention and Management of the Vulnerable Child Syndrome

Prevention at the Time of Health Crisis

Understand and discuss parents' belief about even minor problems
Do not use terms, such as allergy and colitis, that suggest a diagnostic entity when there is no real evidence supporting it
Recognize when parents are particularly anxious about an event and specifically deal with their fears

Management

After a thorough evaluation, provide a clear statement of the child's physical health
Help the parents understand the link between the present problem and their past anxieties about their child's health
Support and advise parents about interacting with their child in an appropriate manner
Make a referral for a psychiatric evaluation and treatment, if necessary

Impact of treatments for chronic conditions on development/behavior

46. Know the behavioral effects of commonly used medications (resource: *UpToDate*)
    a. Atomoxetine (Strattera)
        i. Increased risk of suicidal thinking
        ii. Rare psychiatric side effect (hallucinations, delusional thinking, mania)
        iii. New onset motor tics
    b. Tricyclic antidepressants (imipramine, desipramine, nortriptyline) - inhibit reuptake of serotonin and norepinephrine
        i. Primarily anticholinergic effects (dry mouth, constipation, urinary retention), fatigue, cardiac arrhythmias
    c. Dopamine reuptake inhibitors (bupropion) - blocks reuptake of norepinephrine and dopamine
        i. Irritability, insomnia, motor tics, decreased seizure threshold at high doses
    d. Systemic glucocorticoids (e.g. for acute asthma exacerbation)
        i. Most symptoms are mild and reversible for short duration of treatment, may be worsened with chronic use
        ii. Emotional lability, hypomania, mania, depression, psychosis, delirium, confusion
        iii. Sleep disturbance
47. Recognize behavioral effects of sympathomimetic medications (resource: *UpToDate*)
    a. Beta-agonists
        i. Albuterol for asthma
        ii. Excitement/hyperactive behavior, nervousness, tremor, headache, insomnia, anxiety

b. Alpha-agonists
    i. Clonidine
        1. Sedation, depression, bradycardia, headache
    ii. Guanfacine
        1. Headache, fatigue, sedation
c. Stimulants
    i. Including methylphenidates and amphetamines
    ii. Side effect profiles are generally similar across medications
    iii. Psychosis - rarely develop symptoms (hallucinations, delusional thinking, mania), causality has not been established
    iv. Tics - new onset or worsening of tics in children with existing tic disorders
        1. Tics or family history of tics are included as contraindication for some formulations of methylphenidate
        2. ADHD and tic disorders frequently co-occur
    v. Other - Sleep disturbance, jitteriness, emotional lability, including social withdrawal
    vi. Monitor for stimulant diversion (transfer of medication from the patient for whom it was prescribed to another individual)

48. Recognize the behavioral effects of anti-epileptic drugs (reference: *UpToDate - Antiseizure drugs: Mechanism of action, pharmacology, and adverse effects*)
    a. Common side effects of antiseizure drugs table

Drug	**Neurologic side effects**
Carbamazepine	Drowsiness, dizziness, blurred or double vision, lethargy, headache
Clobazam	Somnolence, aggression, irritability, ataxia, insomnia
Ethosuximide	Sleep disturbance, drowsiness, hyperactivity
Felbamate	Insomnia, dizziness, headache, ataxia
Gabapentin	Somnolence, dizziness, ataxia
Lacosamide	Ataxia, dizziness, headache, diplopia
Lamotrigine	Dizziness, tremor, diplopia
Levetiracetam	Somnolence, dizziness, agitation, anxiety, irritability, depression
Oxcarbazepine	Sedation, headache, dizziness, vertigo, ataxia, diplopia
Phenobarbital	Alteration of sleep cycles, sedation, lethargy, behavioral changes, hyperactivity, ataxia, tolerance, dependence

Phenytoin	Confusion, slurred speech, double vision, ataxia
Pregabalin	Dizziness, somnolence, ataxia, tremor
Primidone	Alteration of sleep cycles, sedation, lethargy, behavioral changes, hyperactivity, ataxia, tolerance, dependence
Topiramate	Nervousness, difficulty concentrating, confusion, depression, anorexia, language problems, anxiety, mood problems, tremor
Valproate	Tremor, dizziness
Vigabatrin	Drowsiness, dizziness
Zonisamide	Somnolence, dizziness, ataxia, confusion, difficulty concentrating, depression

49. Understand the developmental and behavioral effects of central nervous system ventricular shunts (resource: *UpToDate - Hydrocephalus in Children: Management and Prognosis*)
    a. Developmental and behavioral effects should be evaluated while considering the underlying cause of hydrocephalus
        i. Factors include degree of prematurity, other CNS malformations, other congenital abnormalities, and epilepsy
    b. Shunt malfunction can lead to increased intracranial pressure (ICP) - symptoms include headache, lethargy, irritability, new focal deficits
    c. Epilepsy can be present prior to shunt placement but can be a complication of shunt malfunction
        i. Seizure is associated with poor cognitive outcomes (<25% of children with seizures have IQ>90 in one study)
    d. Motor, visual, or auditory deficits
    e. Reduced IQ scores with need for special education services
        i. Scores were modulated by underlying cause of hydrocephalus (congenital versus infectious, IVH), degree of prematurity
50. Understand the developmental and behavioral effects of cranial radiation
51. Understand the developmental and behavioral effects of the anesthetic and bypass/circulatory arrest procedures used during cardiac surgery in infants
52. Recognize the behavioral effects of chemotherapeutic drugs
53. Recognize the long-term developmental and behavioral outcomes of survivors of childhood cancer

Resources

Encounters with Children
Suzanne D Dixon; Martin T Stein
Fourth edition. Philadelphia: Mosby Elsevier 2006

Developmental-Behavioral Pediatrics
William B Carey; William L Coleman; Allen C Crocker; Ellen R Elias; Heidi M Feldman
Fourth edition. Philadelphia: Saunders Elsevier 2009

Developmental and behavioral pediatrics
Robert G Voigt (Professor of pediatrics); Michelle M. Macias; Scott M. Myers; Carl D. Tapia; American Academy of Pediatrics. Section on Developmental and Behavioral Pediatrics.
Second edition. Itasca, IL : American Academy of Pediatrics 2018

Zuckerman Parker handbook of developmental and behavioral pediatrics for primary care
Marilyn Augustyn; Barry S. Zuckerman
Fourth edition. Philadelphia : Wolters Kluwer 2019

UpToDate (https://www.uptodate.com/home)

International classification of functioning, disability and health: children and youth version: ICF-CY

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Content Category 6- Developmental-Behavioral Aspects of Chronic Conditions and Treatment

ABP Content Specs for Developmental-Behavioral Pediatrics Fellow Collaboration Project

Prepared by: Meagan Butsch, DO, Madigan Army Medical Center DBP (now staff at Tripler Army Medical Center); Jonathan Chooey, DO, Paul Patterson, MD, & Eric Flake, MD- Madigan Army Medical Center

Reviewed by: Yasmin Senturias, MD, Staff DBP at Developmental Behavioral Pediatrics of the Carolinas- Charlotte & Eric Flake, MD, Staff DBP at Madigan Army Medical Center

6. Developmental-Behavioral Aspects of Chronic Conditions and Treatment

A. Prematurity and adverse prenatal conditions

1. Prematurity and small for gestational age (SGA) infants
    a. Know the major and minor morbidity outcomes associated with prematurity
    b. Know the most prevalent special health care needs affecting children born prematurely
    c. Understand the neonatal medical/surgical conditions that are associated with adverse developmental and behavioral outcomes in children born prematurely
    d. Understand the association of socioeconomic factors and outcome in children born prematurely
    e. Plan appropriate management for children and families in the neonatal intensive care unit to maximize the child's developmental and behavioral outcomes
    f. Understand the pathogenesis of cerebral palsy and associated brain lesions in infants born prematurely
    g. Understand the natural history of increased muscle tone in infants born prematurely
    h. Understand the pathogenesis of visual impairment in infants born prematurely
    i. Understand the pathogenesis of hearing impairment in infants born prematurely
    j. Understand the typical growth patterns of premature infants
    k. Understand the effectiveness of early intervention for children born prematurely
    l. Understand the variations in developmental and behavioral outcomes after premature birth as a function of the child's birth weight and gestational age
    m. Know the developmental and behavioral outcome for children who are small for gestational age
    n. Plan the developmental monitoring of a child born prematurely or with low birth weight
    o. Understand components of effective early intervention programs for children born prematurely
2. Prenatal drug exposure
    a. Recognize specific medications that are known teratogens
    b. Know the developmental and behavioral consequences of prenatal exposure to illicit drugs (eg opioids, cocaine, methamphetamines)
    c. Know the features of fetal alcohol spectrum disorders
    d. Understand the developmental and behavioral characteristics of children with fetal alcohol spectrum disorders
    e. Know the developmental and behavioral consequences of prenatal

exposure to tobacco
    f. Know the developmental and behavioral consequences of prenatal exposure to anticonvulsants
    g. Know the developmental and behavioral consequences of prenatal exposure to selective serotonin reuptake inhibitors
  3. Prenatal infections
    a. Know how to plan the laboratory evaluation of a child with a suspected prenatal infection
    b. Understand the usual route of transmission of common prenatal and perinatal infections (eg, herpes, HIV)
    c. Recognize the features of congenital syphilis
    d. Recognize the features of congenital herpes simplex and varicella zoster infections
    e. Know the epidemiology of cytomegalovirus infection
    f. Recognize the features of congenital cytomegalovirus infection
    g. Recognize the features of congenital rubella infection
    h. Recognize the features of congenital Toxoplasma infection
    i. Know the developmental and behavioral outcomes associated with specific prenatal infections
  4. Maternal adverse effects
    a. Know the effects of maternal toxemia (preeclampsia) on fetal growth and development
    b. Understand the developmental and behavioral consequences for children born of mothers with diabetes mellitus
    c. Know the developmental and behavioral consequences for the child of maternal chronic illness or malnutrition during pregnancy

B. Adverse perinatal conditions
  1. Know the criteria for identifying perinatal (intrapartum) asphyxia as the cause of developmental disabilities
  2. Know the developmental and behavioral outcomes associated with perinatal asphyxia

C. Chromosomal and genetic disorders
  1. Down syndrome
    a. Understand that the phenotypic expression of Down syndrome may be caused by trisomy 21, translocation of chromosome 21, or mosaicism
    b. Know the epidemiology of Down syndrome
    c. Know the typical IQ range and changes over time of children with Down syndrome
    d. Differentiate between language and visual-spatial skills in the development of children with Down syndrome
    e. Identify factors contributing to speech and language delays in children with Down syndrome
    f. Understand how to evaluate a child with Down syndrome who

presents with behavior problems
   g. Understand the reasons for multi-modal language training for toddlers with Down syndrome
   h. Know how to plan appropriate management of a neonate, child, or adolescent with Down syndrome
   i. Know the medical problems commonly associated with Down syndrome
   j. Recognize the typical phenotypic features associated with Down syndrome
   k. Know the evaluation and recommendations for a child with Down syndrome who wants to participate in athletic activities
   l. Know the neurologic complications associated with Down syndrome
2. Fragile X syndrome
   a. Understand the pathogenesis of fragile X syndrome
   b. Know the appropriate laboratory evaluation to establish the diagnosis of fragile X syndrome
   c. Recognize the phenotypic features of boys and men with fragile X syndrome
   d. Know the prevalence of fragile X syndrome
   e. Know the developmental and behavioral characteristics of boys with fragile X syndrome and premutation carriers
   f. Understand the developmental and behavioral characteristics of girls with fragile X mutation
   g. Know the medical problems commonly associated with fragile X syndrome
   h. Understand the wide range of outcomes in children with fragile X syndrome
   i. Understand the importance of evaluating family members of children identified with fragile X syndrome
   j. Know the medical problems in adult fragile X premutation carriers with fragile X tremor ataxia syndrome
3. Sex chromosome abnormalities
   a. Recognize the phenotypic features of Turner (XO) syndrome
   b. Know the developmental and behavioral characteristics of girls with Turner (XO) syndrome
   c. Recognize the phenotypic features of boys with a karyotype of 47, XYY
   d. Know the developmental and behavioral characteristics of boys with a karyotype of 47, XYY
   e. Know the developmental and behavioral characteristics of boys with Klinefelter (XXY) syndrome
   f. Know the developmental and behavioral characteristics of girls with a 47, XXX karyotype
   g. Recognize the phenotypic features of Klinefelter (XXY) syndrome at different ages

h. Know the cytogenetic heterogeneity associated with Turner (XO) syndrome
i. Know the natural history of Turner (XO) syndrome
j. Know the medical problems commonly associated with Turner (XO) syndrome

4. Rett syndrome
   a. Know the developmental and behavioral characteristics of Rett syndrome at different stages
   b. Recognize the physical signs and symptoms of Rett syndrome at different stages
   c. Know the etiology and appropriate laboratory evaluation of Rett syndrome
   d. Know how to plan the management of a child with Rett syndrome
   e. Understand the prognosis for a child with Rett syndrome
   f. Recognize the broader phenotype of children with MECP2 mutations who do not fit the clinical criteria for Rett syndrome
5. Trisomy 13 and Trisomy 18
   a. Recognize the phenotypic features of Trisomy 13
   b. Know the developmental and behavioral characteristics of children with Trisomy 13
   c. Recognize the phenotypic features of Trisomy 18
   d. Know the developmental and behavioral characteristics of children with Trisomy 18
6. Williams syndrome
   a. Know the developmental and behavioral characteristics of children with Williams syndrome
   b. Recognize the phenotypic features of Williams syndrome
   c. Know the medical problems commonly associated with Williams syndrome
   d. Know the etiology and appropriate laboratory investigation for diagnosis of Williams syndrome
7. Prader-Willi syndrome and Angelman syndrome
   a. Know the developmental and behavioral characteristics of children with Prader-
      Willi syndrome
   b. Know the etiology and appropriate laboratory evaluation for diagnosis of Prader-
      Willi syndrome
   c. Understand the natural history of Prader-Willi syndrome
   d. Know how to plan the management for a child with Prader-Willi syndrome
   e. Know the developmental and behavioral characteristics of children with Angelman syndrome
   f. Recognize the phenotypic features of Prader-Willi syndrome

at different developmental stages
g. Recognize the phenotypic features of Angelman syndrome
h. Know the etiology and appropriate laboratory evaluation to establish the diagnosis of Angelman syndrome

8. 22q11.2 deletion syndrome
   a. Know the appropriate laboratory evaluation to establish the diagnosis of 22q11.2 deletion syndrome
   b. Know the developmental and behavioral characteristics of children with 22q11.2 deletion syndrome
   c. Recognize the phenotypic features of 22q11.2 deletion
9. Other genetic disorders
   a. Know the etiology and appropriate laboratory evaluation to establish the diagnosis of Smith-Magenis syndrome
   b. Know the developmental and behavioral characteristics of children with Smith-
      Magenis syndrome
   c. Know the etiology of neurofibromatosis types I and II
   d. Know how to plan the diagnostic evaluation of a child with neurofibromatosis types I and II
   e. Know the medical problems commonly associated with neurofibromatosis types I and II
   f. Know the developmental and behavioral characteristics of children with neurofibromatosis types I and II
   g. Know the etiology and appropriate medical evaluation to establish the diagnosis of tuberous sclerosis
   h. Recognize the phenotypic features of tuberous sclerosis
   i. Know the developmental and behavioral characteristics of children with tuberous sclerosis
   j. Know the medical problems commonly associated with tuberous sclerosis
   k. Recognize the phenotypic features of neurofibromatosis types I and II
   l. Know the phenotypic features of other neurocutaneous syndromes, eg, Sturge-
      Weber, incontinentia pigmenti
   m. Know the developmental and behavioral characteristics of children with other neurocutaneous syndromes

D. Metabolic disorders
1. Phenylketonuria (PKU)
   a. Understand the importance of neonatal screening for PKU
   b. Know the principles of dietary treatment of individuals with PKU
   c. Know how to plan appropriate counseling for a girl with PKU about sexuality, pregnancy, and genetic risks
   d. Know the phenotypic features of PKU
   e. Know the developmental and behavioral characteristics of children with

PKU

f. Know the evaluation of suspected PKU

2. Mucopolysaccharidoses
   a. Understand the usual natural history of developmental skills in children with mucopolysaccharide disorders
   b. Recognize the phenotypic features suggestive of a mucopolysaccharide disorder
   c. Know the etiologies and appropriate laboratory evaluation to establish the diagnosis of the major mucopolysaccharide disorders
   d. Know the developmental and behavioral characteristics of children with mucopolysaccharide disorders
3. Purine metabolism
   a. Know the developmental and behavioral characteristics of children with Lesch- Nyhan syndrome
   b. Know the evaluation of suspected Lesch-Nyhan syndrome
   c. Recognize the phenotypic features of Lesch-Nyhan syndrome
   d. Plan the management of a child with Lesch-Nyhan syndrome

E. Infectious diseases
1. Meningitis and encephalitis
   e. Recognize the neurological, developmental, and behavioral complications of various forms of meningitis
2. HIV/AIDS
   a. Know the neurological, developmental, and behavioral complications of pediatric HIV/AIDS and its treatment
   b. Understand the natural history of pediatric HIV/AIDS
   c. Understand the epidemiology of pediatric HIV/AIDS
3. Chronic otitis media
   a. Know the sensory, developmental, and behavioral consequences of chronic otitis media with effusion

F. Central nervous system (CNS) disorders
1. Seizures and epilepsy
   a. Differentiate between seizures and epilepsy
   b. Understand the international classification of epileptic syndromes
   c. Know how to plan the evaluation of a child with staring spells
   d. Know that the prevalence of seizures and epilepsy is higher in children with developmental disorders than in the general population
   e. Know the appropriate management for a child with epilepsy and attention deficit hyperactivity disorder
   f. Know that developmental and behavioral difficulties are more common in children with epilepsy than in children with other types of chronic physical illnesses

g. Know how to plan the evaluation of a child with suspected seizures
h. Know the etiologies and natural history of infantile spasms

2. Hydrocephalus
   a. Know the causes of hydrocephalus in children
   b. Know the developmental and behavioral characteristics of children with hydrocephalus
3. Stroke
   a. Understand the developmental and behavioral sequelae following a stroke at different ages
   b. Understand the natural history of language development in children following a stroke at different ages
   c. Know the causes of pediatric stroke at different ages
4. Traumatic Brain Injury (TBI)
   a. Differentiate between mild, moderate, and severe head injury on the basis of the Glasgow coma score
   b. Know the long-term management for a child with traumatic brain injury
   c. Understand the pathogenesis of brain injury with penetrating head trauma
   d. Know the developmental, cognitive, and behavioral consequences of penetrating injury to specific brain regions
   e. Understand the pathogenesis of brain injury with closed head trauma
   f. Know the developmental, cognitive, and behavioral consequences of mild, moderate, and severe closed head injury
5. Central nervous system (CNS) tumors
   a. Know the developmental, cognitive, and behavioral consequences in children who have had CNS tumors
   b. Know the behavioral, developmental and neurological symptoms suggestive of a CNS tumor
6. Congenital CNS malformation
   a. Know brain malformations that may be found in individuals with no developmental or behavioral disabilities
   b. Know the brain malformations most commonly found in individuals with developmental disabilities (eg, holoprosencephaly, schizencephaly, lissencephaly)

G. Sensory defects
1. Visual impairments
   a. Know the leading causes of visual impairment in childhood
   b. Understand the importance of a functional vision assessment in determining which literacy media to use with significant visual impairment or blindness (eg, Braille, large print, optical modifications)
   c. Know how to assess development in children who are visually impaired
   d. Know how to plan the management of developmental/behavioral issues in children with a range of visual impairments
   e. Understand the developmental and behavioral problems associated

with severe visual impairment
   f. Understand the developmental and behavioral problems associated with mild to moderate visual impairment
   g. Understand the developmental maturation of visual acuity in early childhood
   h. Understand the meaning of cortical visual impairment, and other forms of visual impairment
2. Hearing loss
   a. Recognize the typical audiogram of a child with conductive or sensorineural hearing loss
   b. Know the effectiveness of teaching sign language to children who have severe to profound hearing loss
   c. Know how to plan the developmental assessment of a child with severe/profound hearing loss
   d. Know the impact of deafness on development, behavior, and academic achievement
   e. Understand the efficacy of cochlear implantation for children with profound hearing loss
   f. Understand the factors that affect decisions on use of cochlear implants
   g. Recognize the importance of early language exposure and instruction for children with hearing impairment
   h. Know the three common educational/communication methods for children with hearing impairments: oral, manual, and total language and reasons for choosing each
   i. Know the leading causes of severe hearing impairment
   j. Know the criteria for amplification with hearing aids
   k. Understand the developmental and behavioral consequences of mild or moderate hearing loss
   l. Understand the developmental and behavioral consequences of unilateral hearing loss
   m. Understand the developmental-behavioral consequences of severe or profound hearing loss

H. Other chronic conditions
1. Understand the reasons for inclusion of thyroid testing in the neonatal screening battery
2. Differentiate between congenital and acquired hypothyroidism in terms of developmental outcomes
3. Know the impact of type I diabetes on development and behavior
4. Understand the association of recurrent diabetic ketoacidosis and problems in family functioning
5. Understand the effects of excess corticosteroids on affect and behavior in children
6. Identify the developmental and behavioral consequences of iron deficiency

at different ages

7. Know the factors that affect the developmental and behavioral outcome of children with congenital heart disease
8. Know the nutritional deficiencies associated with developmental and behavioral problems in children
9. Understand the impact of prolonged caloric malnutrition (failure to thrive) on development and behavior
10. Identify the developmental and behavioral effects of lead exposure
11. Understand the risks for lead exposure in children with developmental disabilities
12. Understand the impact of iodine deficiency and maternal hypothyroidism on the fetus
13. Understand the emotional and family factors that affect symptoms and prognosis in children with chronic illnesses, such as asthma
14. Know the factors affecting the developmental and behavioral outcome of children with hematologic disorders
15. Know the factors affecting the developmental and behavioral outcome of children with oncologic disorders
16. Know the factors affecting the developmental and behavioral outcome of children with rheumatologic disorders
17. Know the factors affecting the developmental and behavioral outcome of children with immunologic disorders
18. Know the factors affecting the developmental and behavioral outcome of children with pulmonary disorders
19. Know the factors affecting the developmental and behavioral outcome of children with renal disorders
20. Know the factors affecting the developmental and behavioral outcome of children with gastrointestinal disorders
21. Know the developmental risks to the fetus and young child of mercury toxicity

6. Developmental Behavioral Aspects of Chronic Conditions and Treatment

A. Prematurity and Adversee prenatal conditions

1. Prematurity and SGA infants.

a) Know the major and minor morbidity outcomes for prematurity:

- VLBW and ELBW infants at higher risk for poor outcomes and Cognitive Scores as well as use of special Education
- Increased a visual motor disturbances (Cerebral Palsy)
- Broncho Pulmonary dysplasia (BPD) most common sequelae of prematurity
- Retinopathy of Prematurity
- IVH - significant mortality.
  - Grade I: uni/bilateral hemorrhage in the germinal matrix (<10%) Ventricular area
  - Grade ll: 10-50% hemorrhage of ventricular area (no ventricular dilation)
  - Grade lll: Ventricular dilation, with at least 50% occupied by hemorrhage. May see periventricular echodensities.
  - Grade IV: hemorrhage extending into parenchyma, likely cystic PVL
- Hydrocephalus: increase risk for spasticity, developmental concerns
  - shunted hydrocephalous: increased risk neurobehavioral issues such as attention and behavioral concerns.
- PVL: The more extensive the damage, specifically in the Frontal-Parietal and frontal-parietal-occipital regions worse the developmental outcomes - Presence of PVL at 40 weeks predictor of poor developmental outcomes
  - -Cerebral Palsy: Increasing risk with decrease in birth weight. Rising Incidence as survival rates increase in the VLBW and ELBW infants.
- **Summary:** 2-5 year developmental outcomes shows 20-60% of

survivors have a disability with 10-20% having significant adverse sequelae.

- 2 year outcomes poorer outcomes in correlation to decreased BW
- 6 to 8 years old outcomes show increased issues w/ school and
- behavioral disorders (VLBW, ELBW, CP, ROP) exposure even w/o severe Neurological disability

b. Know the most prevalent special health care needs:

- Children w/ Prematurity, depending on extent of injury (listed above) need variable support.
- Children with obvious motor or cognitive disability are still likely to require academic and/or behavioral supports
- Children with CP can be eligible for accommodation under section 504 of the Rehabilitation Act of 1973

Section 504 stipulates that students with disabilities have equal access to services and benefits through 'minor adjustments'

c. Med/Surg conditions associated with adverse developmental and behavioral outcomes in children born prematurely

- IVH (see above)
- Hydrocephalous (see above)
- Cerebral Palsy (see above and below) - Physical assistance typical
- BPD (see above) some association with increase Neuro behavioral concerns with use of antenatal steroids (DART) for BPD

d: SES: Socioeconomic Factors and outcomes of children born prematurely

- socioeconomic concerns can increase risk of poor developmental outcomes, lower SES is at higher risk, especially for VLBW, ELBW premature infants

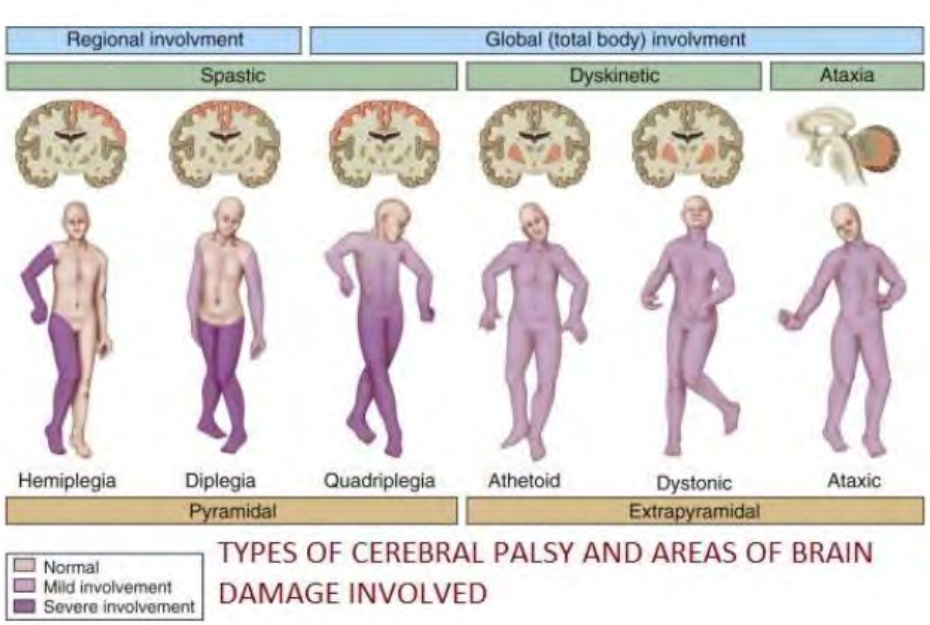
e: Appropriate NICU management includes adequate nutritional

- Supports and consideration for optimal treatment of Medical/Surgical comorbidities
- Parental Education and Presence in the NICU is also important for

overall NICU outcomes

f: Pathogenesis of Cerebral Palsy

- Most cases are from unknown etiology
- CP can be Prenatal in origin, 2° to conditions such as
  1) Congenital brain malformations
  2) Neuronal migration disorders
  3) Vascular disturbances
  4) Genetic conditions
  5) Maternal infections
  6) Other maternal factors
- Perinatal (trauma, asphyxia, infection, hemorrhage)
  - IVH: PVL: Low BW (VL/EL) more significant contributor to severity of cerebral palsy compare to actual Prematurity
  - **HIE: term infants, seizures, MRI Changes, ECG Changes suggest worsening outcome ESPECIALLY for quadriplegic and/or Extrapyramidal (dyskenetic CP)** w/ injury to cerebellum, thalamus, basal ganglia
- As a single group, 50% of children with CP have ID, 28-35% have epilepsy, 12% have some degree of hearing impairment, 4% are deaf, 36% have some form of visual deficit, 24% are nonverbal, 26% have behavioral problems, and 23% have sleep disorders
- Hemiplegia: Epilepsy occurs in about 25% of children with hemiplegia and is much more common in children with cerebral dysgenesis. Severe outcomes infrequent
- Spastic Diplegia: is commonly associated with complications of preterm birth (and LBW). Less likely to have epilepsy, more likely to have intellectual disability. Speech is commonly affected
- Dyskinetic CP: Broad causes – roughly 45% with normal intellect. Higher rates of epilepsy and speech disorders as well as hearing impairment. Deafness is 10% in this population (the highest)
- Quadriplegia: the highest associated deficits. Only about 25% have normal intellect, more than half with epilepsy and visual impairment.



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g: Understand the history of increased tone in infants born prematurely

- Spasticity is velocity dependent vs. Increased tone is sustained
- Lesions in the pyramidal tracts (rotor) cause overall increase in “resting tone”
- Spasticity results from Upper Motor Neuron injury (hyperreflexia, clonus etc.)

h: Understand vision impairment in infants and prematurely.

- Most common issue in Retinopathy of prematurity (ROP)
- Higher risk of amblyopia, strabismus, refractive error, cortical vision impairments is associated with prenatal brain injury
- Later in life, children at increased risk of glaucoma and retinal detachments

i. Understand hearing impairment on children born prematurely

- Increased risk for sensorineural hearing loss
- Hearing loss found in ~20% of infants born < 20 wga (11% in < 25 wga)
- Overlap with certain genetic factors, inversely related to BW, gestational age

j: Understand the typical growth factors of infants born prematurely

- Important that children "follow their curve”

- Catch up growth usually occurs when an infant reaches 5-10th%
- Most significant growth at 36-40 weeks post-natal age.
- Attainment of catch up growth affected by BW, Gestational Age, genetic potential continuing other morbidity.

k: Understand that Early Intervention is effective for children born prematurely

- Multidisciplinary team to outline goals for individualized family service plan (IFSP)
- EIS is covered under part C of IDEA, adopted in 1986.
- Part C allows for provision of services to children from birth to age 3 years with the goals of minimizing a child's need for special education services.
- diagnosis is not required for a referral
- IDEA mandates developmental supports for infants and toddlers with disabilities/delays, the eligibility criteria and fee structure are detained by individual states

(PREP 2019 pipkin ph okamoto j council on children with disabilities; council on school health. The IDEA for children with special education needs. pediatrics. 2015 (Rose L herzig D, Hussey Gardner B early intervention and the role of pediatricians, pediatric rev

l: Understand the variations in developmental and behavioral outcomes after premature birth

- function of BW and gestational age
- Outcomes inversely related to BW/GA - ie the smaller and more premature an infant the more developmental complications expected

m: know the outcomes for SGA infants: (see above)

n: plan the developmental monitoring of a child born prematurely or with low BW

- Early Intervention recommendation / referral
- Bayley Infant Toddler Scales of Development
- Monitor THROUGH school age as children at increased risk for executive function, academic and behavioral disorders

o: Understand components of effective Early Intervention

- includes monitoring, education & parental and therapies as indicated
- EIS will also assist families with transition to IEP after age 3

2. Prenatal Drug Exposure

a. Recognize medications that are known teratogens

- Alcohol (see "c")
- illicit drugs (see "b")
- others
    - Carbamazepine (Tegretol) : FDA catagory "s" increases risk of spina bifida, head/face deformations, heart defects
    - Phenytoin (Dilantin): Fetal hydentoh syndrome (abnormal finger/toes, developmental delay, cleft palate, microcephaly, brain malformations)
    - Valproate (Depakote): spina bifida, neural tube defects, hypospadias, heart defects, limb and genetic defects
    - Warfarin (Coumadin): low BW, ID, microcephaly, deafness, malformed bones
    - Cyclophosphamide (Cytoxan): INGR, cranial/eye malformations, hydrocephaly, microtia, hearing defects, craniosynastosis (some limb detects,)
    - Lead: (decrease cognitive ability, increased behavioral concerns)
    - Lithium: Epsteins anomaly - most also have lower IQ (but general development and growth and behavioral concerns)
    - Thalidomide: Phocomelia – a rare congenital deformity where hands or feet are attached close to the trunk. (NOT FDA approved in USA, no longer used globally)

b. Know the developmental / behavioral consequences of illicit drugs.

- Opioids - NAS, small growth, behavioral complications (some evidence for Cardiac issues)
- Cocaine - Preterm delivery, SGA, ~ decrease in cognitive and increase behavioral issues
- Methamphetamines: SGA, increase risk for "neurodevelopmental concerns”
- Growing up in a home with substance abuse is an ACE and heightens the risk for neglect and abuse.

Table. Short- and Long-term Effects of Fetal Substance Exposure

Effect	Alcohol	Marijuana	Opiates	Cocaine	Methamphetamine
Short-term effects or birth outcome					
Fetal growth	+++	+/–	+	+	+
Anomalies	+++	–	–	–	–
Withdrawal	–	–	+++	-	Unknown
Neurobehavior	+	+	+	+	–
Long-term effects					
Growth	+++	–	–	+/–	Unknown
Behavior	+++	+	+	+	Unknown
Cognition	+++	+	+/–	+	Unknown
Language	+	-	Unknown	+	Unknown
Achievement	+++	+	Unknown	+/–	Unknown

- Adapted with permission from Behnke et al (2013).◦ +++, strong effect; +, effect; +/–, no consensus about effect; –, no known effect.

Reprinted with permission from Smith VC, Wilson CR, AAP COMMITTEE ON SUBSTANCE USE AND PREVENTION. Families Affected by Parental Substance Use. *Pediatrics*. 2016;138(2): e3

c. Know the Features of Fetal Alcohol Syndrome -

- Direct toxic effect of alcohol. Dose dependent. Time during gestation of exposure
- (Greater toxic impact early in gestation) - Later alcohol exposure has an impact on growth factors.
- Prevalence of FAS has been estimated to 6-9 cases per 1000 children.
- No single anomaly unique of or pathogenic for FAS however, combination of features leading to diagnosis:

  1) growth deficiency (-25D of nml), < 3% of growth (pre/post natal)

  2) Characteristic facial appearance (short palpebral fissure, flat philtrum, thinning of upper vermillion lip border)

  3) CNS damage - microcephaly, behavioral/cognitive challenges (any range of recognized structural, neurologic or functional CNS deficit)

- FASD (spectrum disorder) is an umbrella term to describe the range of effects that occur with prenatal alcohol exposure.

d. Understand developmental and behavioral characteristics of children with FAS

- Prenatal alcohol exposure is associated with a higher incidence of ADHD and specific learning disabilities, executive function, visual spatial, impulse control, language, memory, social and problem solving deficits.
- small stature considerations, children should still have typical growth velocity
- Important to ensure other syndromes not present (ie Williams, Aarskog, Opitz-Frias)
- Monitor hearing and Vision, typically abnormalities are minor
- Developmental Delay typical, all motor skills effected and poor athletic performance is typical
- Sleep disturbance common - more often relieved with medication vs therapy
- Most common mental health disorder is ADHD. stimulant medication response varies in FASD due to dopamine levels in the brain, Alcohol can cause under or overproduction of dopamine which produce symptoms for ADHD. If stimulants further exacerbate dopamine levels, FASD ADHD Symptoms will NOT improve
- Past Working memory, understanding of intersocial relationships. (post emotional control)
- Variations of IQ measures:
    1) specific learning disorder
    2) poor memory and judgement
    3) poor intersocial skills stemming from poor language and communication skills,
    4) poor athletic ability / coordination
    5) poor fine motor skills
- FASD children at high risk of high school drop out.

-Kulig JW American academy of pediatrics committee on substance abuse (pediatrics)
-Smith VC wilson CR AAP families affect ed by parental substance use
-WIlliam JF smith VC AAPC FETAL alcohol spectrum disorders
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e. Developmental Behavioral outcomes following exposure to tobacco.

- SGA, increased risk for behavioral concerns regarding attention and hyperactivity increased externalizing behavioral, including conduct disorder

f. Exposure to anticonvulsants- see (a) for anticonvulsants

g. Know the developmental /behavioral consequences for maternal SSRI use.

- Premature delivery, SGA, IUGR, NAS,
- Recent studies suggest statistically significant outcomes of Autism with maternal SSRI use.
- NAS - neonatal abstinence syndromes Withdrawal of infant to substances (typically opioids)
  - Signs: tremors, fussiness, tachypnea, sleep disorder with constant yawning

3. Prenatal infections

a. Know how to plan the lab evaluation of a child with suspected infection

- Consider blood cultures, write eval and other standard labs
- CXR - other imaging as indicated.
- Amp and Gent, consider acyclovir
- Common infection: GBS, Hib, e.coli, Listeria, HSV (viral cause)

b: Understand usual route of transmission of common infections

- GBS; Perinatal infection, prolonged ROM. Late onset could be transmitted during pregnancy or through another avenue (uncertain in most cases)
- HSV: risk of infection increased to 30-50% in late pregnancy. 85% perinatal transmission during in intrapartum period delivery) - Postpartum infection via oral transmission (kissing the baby)
- HIV transmitted during pregnancy, delivery of or breastfeeding. C-sections can reduce transmission is mothers with large viral load. Retroviral medications reduce transmission rate to 1%

c. Recognize congenital syphilis

- Typically appear 3-4 weeks but can present at birth and as late an age 5 years
- Early Symptoms can include fever, low weight, inflammation of umbilical cord, anemia, hepatomegaly, shedding of skin on palms (desquamation), rhinitis (snuffles), choreoretinitis
- Late symptoms: bone pain, retinitis pigmentosa, Hutchinson Triad (upper central incisor, interstitial Keratitis(blurred vision, tearing), eighth nerve deafness) saddle nose, Saber shins

d. Recognize congenital HSV and Varacella Zoster

- HSV: SEM (45%), skin vesicles around mucosal surfaces. Death and nuerodevelopmental disease less likely
- Disseminated: Increased death, (25%) slight long term effects.
- CNS disease (38%) poor NDV disease outcomes
- Infants con present w/ hypothermia, seizures, temporal lobe
- bleeding
- VZV: limb abnormalities, hypertrophic skin, Microphthalmia, pendular nystagmus, brain malformations

e. Know the epidemiology of CMV

- transmitted transplacentally, contaminated birth canal, breast milk,
- Viral latency can cause late progression

f. Recognize the features of congenital CMV

- Microcephaly Retinitis, Periventricular calcifications, SNHL, rash, hepatosplenomegaly

g. Recognize features of congenital Rubella infection.

- IUGR, microcephaly, cardiac issues (PDA cod pulmonary artery stenosis) Hepatosplenomegaly, dermal erythropoiesis (blueberry muffin rash), cataracts / retinopathy, SNHL

i: Know the developmental / behavioral outcomes association with prenatal infections

- Degree varies based on severity, type and age of infection HSV encephalitis example of highly likely to experience developmental sequela.
- CMV typically demonstrate SNHL.
- Bacterial meningitis hearing loss

h: Recognize congenital toxoplasmosis infection.

- Triad (Chorioretinitis, hydrocephalus, intracranial calcification (ring enhancing)
  petechiae and purpura (blueberry muffin)

4. Maternal Adverse Effects

a. Know the effects of toxemia (preeclampsia) of fetal growth and development

- Pre. E can lead to lUGR and oligohydramnios as well as medically indicated preterm birth. Morbidity and mortality are increased
- Oligohydramnios-(too little fluid), can also lead to premature delivery, poor growth and birth defects

b. Understand the developmental and behavioral consequences for children born to mothers with diabetes mellitus

- maternal gestational diabetes seems to be negatively associated with childhood cognitive development, particularly language development
    - The quality of the evidence was poor, as most studies did not adequately control for confounding factors such as pre-pregnancy obesity or maternal socioeconomic status, Preterm delivery etc.
- In a Danish cohort study, adolescents born to mothers with pre-gestational type 1 diabetes had lower scores on cognitive testing compared with controls after adjustment for confounding factors
- Congenital anomalies can occur:
    - Cardiac – Cardiovascular malformations occur in 3 to 9 percent of diabetic pregnancies. Cardiac defects that present more frequently in Infants of mothers with diabetes than in the normal newborn population include transposition of the great arteries (TGA), double outlet right ventricle (DORV), ventricular septal defect (VSD), truncus arteriosus, tricuspid atresia, and patent ductus arteriosus (PDA)
    - CNS – Anencephaly and spina bifida are 13 and 20 times more frequent, respectively, among Infants of mothers with diabetes compared with infants of mothers without diabetes.
        - **The majority of cases of caudal regression syndrome occur in Infants of mothers with diabetes**
        - This syndrome consists of a spectrum of structural defects of the caudal region, including incomplete development of the sacrum and, to a lesser degree, the lumbar vertebrae
    - Other anomalies include:
        - Flexion contracture of the limbs.
        - Vertebral anomalies.
        - Cleft palate.
        - Intestinal anomalies including small left colon syndrome, which occurs primarily in infants of mothers with diabetes

c. Know the developmental and behavioral consequence for the child of maternal chronic illness or malnutrition during pregnancy.

- Outcomes can be specific to type of malnutrition or illness
  - Folic acid – increase risk of NTD
  - Anemia (decreased blood flow) – kidney impairment, cognitive/behavioral outcomes associated with prematurity, low birth weight, brain injury (all potential outcomes for poor blood flow)
  - Risk factors for SGA infants

B. Adverse perinatal Conditions

1. Know the criteria for identifying perinatal (intrapartum) asphyxia as the cause of Developmental Disabilities

- The diagnostic criteria for neonatal hypoxic-ischemic encephalopathy (Asphyxia) are as follows:
  - Metabolic acidosis with pH <7.0 (in umbilical cord or infant blood sample)
  - Base Deficit -12
  - APGAR score = five at 10 minutes with a continued need for resuscitation
  - Presence of multiple organ-system failures
  - Clinical evidence of encephalopathy: hypotonia, abnormal oculomotor or pupillary movements, weak or absent suck, apnea, hyperpnea, or clinical seizures
  - Neurologic findings cannot be attributed to other cause (inborn error of metabolism, a genetic disorder, congenital neurologic disorder, medication effect)

Gillam-Krakauer M, Gowen Jr CW. Birth Asphyxia. [Updated 2019 Nov 8]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2020 Jan-. Available from: https://www.ncbi.nlm.nih.gov/books/NBK430782/

2. Know the developmental and behavioral outcomes associated with perinatal asphyxia

- Most infants with mild encephalopathy develop normally, while infants with moderate to severe encephalopathy are more likely to develop long-term neurologic morbidity
- Severe brain MRI abnormalities are usually associated with marked electroencephalogram (EEG) abnormalities and poor outcome.
- Among survivors, permanent neurologic sequelae of neonatal brain injury can be mild, such as learning difficulties or attention deficit disorder, or may be severe and disabling,

such as cerebral palsy, epilepsy, visual impairment, or severe cognitive and developmental disorders.

- One report of 110 survivors of neonatal encephalopathy found that subnormal intelligence quotient (IQ) scores at six to seven years of age were present in more than 25 percent of children overall; an IQ score <70 among survivors with and without cerebral palsy was found in 96 and 9 percent, respectively
- Cerebral palsy develops in approximately 13 percent
- Although definitions vary, a more severe degree of neonatal encephalopathy, as categorized by modified Sarnat criteria and the presence of seizures are associated with increased risk of adverse outcome.
- Infants with moderate encephalopathy have a 20 to 35 percent risk of later sequelae from the insult, although those whose neurologic examinations are completely normal within one week and whose brain MRI show no evidence of injury have a good likelihood of normal outcome.

Categories (total 6)	Signs of neonatal encephalopathy (NE) in each category			
	Normal	Mild NE	Moderate NE	Severe NE
1. Level of consciousness				
	Alert, responsive to external stimuli (state dependent, e.g. post feeds)	Hyper-alert, has a stare, jitteriness, high-pitched cry, exaggerated responds to minimal stimuli, inconsolable	Lethargic	Stupor/coma
2. Spontaneous activity				
	Changes position when awake	Normal or decreased	Decreased activity	No activity
3. Posture				
	Predominantly flexed when quiet	Mild flexion of distal joints (fingers, wrist usually)	Moderate flexion of distal joint, complete extension	Decerebrate
4. Tone				
	Strong flexor tone in all extremities + strong flexor hip tone	Normal or slightly increased peripheral tone	Hypotonia (focal or general) or hypertonia	Flaccid Rigid
5. Primitive reflexes (circle only the highest level in each sign; the maximum score is only 1 in any one category)				
Suck	Strong, easily illicit	Weak, poor	Weak but has a bite	Absent
Moro	Complete	Partial response, low threshold to illicit	Incomplete	Absent
6. Autonomic system (circle only the highest level in each sign; the maximum score is only 1 in any one category)				
Pupils	In dark: 2.5–4.5 mm; in light: 1.5–2.5 mm	Mydriasis	Constricted	Deviation/dilated/non-reactive to light
Heart rate	100–160 bpm	Tachycardia (HR > 160)	Bradycardia (HR < 100)	Variable HR
Respiration	Regular respirations	Hyperventilation (RR > 60/min)	Periodic breathing	Apnoea or requires ventilator
Total score				

Modified Sarnat scale for clinical encephalopathy staging

C. Chromosomal and Genetic Disorders

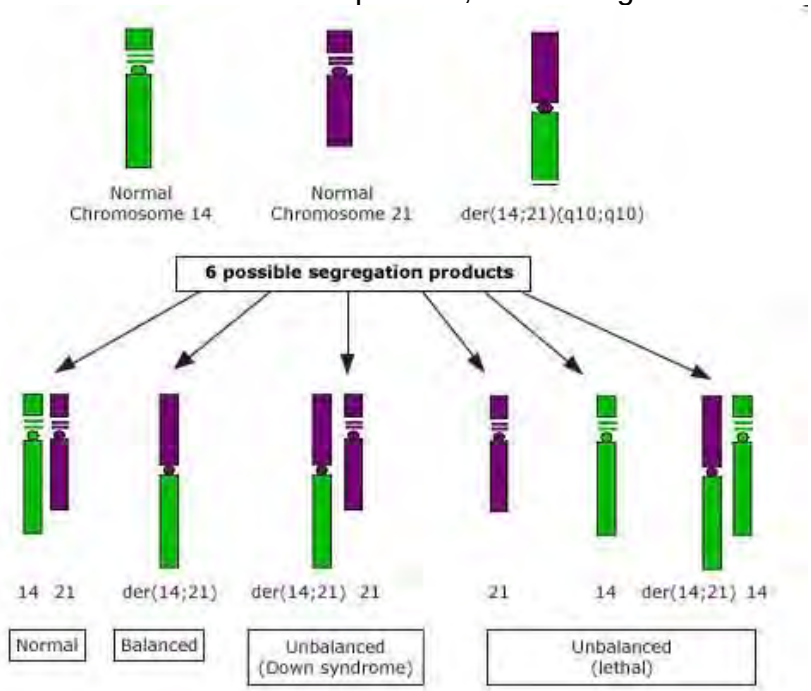
1. Down syndrome

a. <u>Understand the phenotypic expression of Down Syndrome can be caused by Trisomy 21,</u>

<u>Translocation of Chromosome 21 or mosaicism.</u>

- Karyotype at birth/early infancy recommended to confirm diagnosis
- Trisomy 21: ~95% of DS cases
  - Nondisjunction occurring during meiosis I
  - **Advanced maternal age is the most significant risk** factor (8 fold increase in women >40y) this can also factor in meosis II (15 fold increase)
- Translocation (unbalanced): 4-5% if children with DS.
  - Not affected by maternal age

- Inherited or de nova mutation
- Most translocations involve chromosome 14 and 21, either as 14/21 or 21/21 translocation
- If found in infants – both parents should be tested for a **balanced** translocation and if positive, counseling about recurrence risk.



Pediatric Board Exam Studying by **Sheila Kilbane, MD**.

- Mosaicism: Within the trisomy 21 cell line in the post zygotic embryo accounts for no more than 1% of DS.
  - Examined cell lines will have a certain % of 47** and others will have 46**. This is the percent of mosaicism.
  - Babies born with mosaic Down syndrome can have the same features and health problems - it is possible that these babies may have fewer characteristics of the syndrome than those with other types of Down syndrome.

b. Know the epidemiology of Down Syndrome

- Please see above for risk factors/percent occurrence

c. Know the typical IQ range and changes over time of children with Down syndrome

- Almost all individuals with DS have cognitive impairment
- Most are mildly to moderately intellectually disabled, with an intelligence quotient (IQ) in the 50 to 70 or 35 to 50 range, respectively, although some are severely impaired

- IQ reaches a plateau in adolescence (after age ~10) that continues into adulthood
- In general, the average age of sitting (11 months), creeping (17 months), and walking (26 months) is approximately twice the typical age.
- The sequence of language development is the same, although the rate is slower, with the average age for the first word at 18 months.
- cognitive deficits are primarily in morphosyntax, verbal short-term memory, and explicit long-term memory

d. Differentiate between language and visual spatial skills in the development of children with Down Syndrome

- propose that visual-spatial working memory is relatively preserved compared to verbal working memory in DS individuals
- language is severely affected in these individuals, with expressive vocabulary, grammar and syntax being the weakest areas of function
- Children with DS have often been characterized with relative strengths in visuo-spatial processing and poor verbal processing skills
- The relative strength of visuo-spatial skills is supported by strong fine motor skills documented through the use of gestures in early development as well as task completion (block building, drawing line, copying, folding)

George Grouios and Antonia Ypsilanti (August 29th 2011). Language and Visuospatial Abilities in Down Syndrome Phenotype: A Cognitive Neuroscience Perspective, Genetics and Etiology of Down Syndrome, Subrata Dey, IntechOpen, DOI: 10.5772/20483. Available from: https://www.intechopen.com/books/genetics-and-etiology-of-down-syndrome/language-and-visuospatial-abilities-in-down-syndrome-phenotype-a-cognitive-neuroscience-perspective

e. Identify factors contributing to speech and language delays in children with Down syndrome

- Hearing loss - Many children with Down syndrome experience hearing loss in the early years and this has been shown to influence their spoken language development
    - Speech sound discrimination - Speech sound discrimination and production skills at around 7 to 10 months predict later progress in learning to use spoken words. hearing problems are likely to influence the ability to discriminate between sounds, and this will affect a child's ability to learn to understand words as well as to say them. For example, consider "cat", "hat", "mat", "sat", and "bat" - these are all early

words with only one differing sound, but each has a different meaning. Even a mild hearing loss may make it difficult to distinguish between these words when spoken.

- Storing spoken word patterns - more difficulty in learning and storing accurate sound patterns for words (phonological representations)
- Cognitive delay
- Less input - There is some evidence that children with Down syndrome experience less conversation with their parents than other children
- Verbal short-term memory - Most children with Down syndrome have particular difficulties with verbal short term memory. Verbal short term memory is important for holding verbal information and influences word learning

Thiemann-Bourque, K.S., Warren, S.F., Brady, N., Gilkerson, J. & Richards, J.A. (2014) Vocal interaction between children with Down syndrome and their parents. *American Journal of Speech-Language Pathology*, 23(3), 474-485
Jarrold, C., Thorn, A.S.C., & Stephens, E. (2009). The relationships among verbal short-term memory, phonological awareness, and new word learning: Evidence from typical development and Down syndrome. *Journal of Experimental Child Psychology*, 102(2), 196-218
https://www.seeandlearn.org/en-us/language-and-reading/design/language-vocabulary/

f. Understand how to evaluate a child with Down syndrome who presents with behavior problems

- Behavioral and psychiatric disorders are more common in DS than typical children but less common than in those with other causes of intellectual disability
- Attention-deficit hyperactivity disorder, conduct/oppositional disorder, or aggressive behavior, are most common.
- Autism is a common comorbidity of DS, affecting as many as 7 percent of children with DS
- Always consider medical cause – reflux/celiac/GI abnormality/constipation that could be causing pain.

g. Understand the reasons for multi modal language training for toddlers with Down Syndrome

- Because of strength in nonverbal skills, gestures, repetition and visual interpretation, children with DS benefit form multiple methods of communication, not just verbal.
- Vocabulary is often delayed as is overall expressive language
    - Using PECS and/or ACC while also developing verbal vocabulary and skills can support a child with DS capable of learning language but with

relative weakness in expressive skills.

Kathleen Mays MS Global Down Syndrome Foundation 2017 webinar series (https://safe.menlosecurity.com/doc/docview/viewer/docNFD335D04E2C3bc6def6fc71d78628c4a52116f165b7179c6ce008ba200eaf6d28e6bde78588f)

h. Know how to plan appropriate management of a neonate, child or adolescent with Down syndrome

- Newborn:
  - Audiology evaluation – repeat q6 months until age 4-5years.
    - This is the most common medical problem in Down Syndrome (~75%)
  - Echocardiogram – CHD in 40-50%
  - Ophthalmology – looking for cataract (only ~ 15%), strabismus or nystagmus (annual to age 5)
  - Lab evaluation - CBC, Hb (annually, CRP and ferritin if low Hb)
    - Anemia, iron deficiency, TAM (~10%)
  - TSH – May have obtained with newborn screen
- Infant:
  - Swallow Assessment if indicated (FTT).
  - Ophthalmology – if not yet completed by 6mo (annual to age 5)
  - TSH – repeat at 6 mo and 12 mo of life. Then Annually
- Child:
  - Audiology – children aged 3-5 y have serious otitis and can bh=have persistent serous effusion tube dysfunction. May need annual evaluations through adulthood)
  - Ophthalmology – annual to age 5y, then if no disease, can be completed 2years (and then every 3 years at age 13y)
  - TSH – Annually after the age of 2y
  - Celiac – grab a tissue transglutaminase immunoglobulin A (tTG-IgA) and don't forget the total quantitative IgA if symptomatic with persistent behavioral problems, constipation, diarrhea or belly upset. (this is also considered in Adolescent time frame)
  - Sleep Study – recommended to have by age 4y.
  - Sexuality – begin discussion regarding puberty and need for gynecological care for females

- Adolescent
  - Audiology – Annual evaluations through adulthood.
  - Ophthalmology – increased risk for keratoconus and lens opacities. If no disease present can monitor q3 years after age 13y.
  - TSH – annually
  - Hg – annually after the age of 13 (include CRP and ferritin if concerned for iron deficiency)
  - Sexuality – continue discussions – consider birth control for females.

i. Know the medical problems commonly associated with Down Syndrome

- Please see above, and table below:
- Also, not previously mentioned: increased risk for gastrointestinal tract anomalies, which occur in approximately 5 percent of cases.
  - Duodenal atresia or stenosis, sometimes associated with annular pancreas, is the most characteristic lesion, occurring in 2.5 percent
- CBC can evaluate for anemia and TAM, which can increase risk for AML in childhood.

Table. Medical Problems Common in Down Syndrome

Condition	%
Hearing problems	75
Vision problems	60
Cataracts	15
Refractive errors	50
Obstructive sleep apnea	50–75
Otitis media	50–70
Congenital heart disease	40–50
Hypodontia and delayed dental eruption	23
Gastrointestinal atresias	12
Thyroid disease	4–18
Seizures	1–13
Hematologic problems	
Anemia	3
Iron deficiency	10
Transient myeloproliferative disorder	10
Leukemia	1
Celiac disease	5
Atlantoaxial instability	1–2
Autism	1
Hirschsprung disease	<1

Reprinted with permission from Bull MJ, Committee on Genetics. Clinical Report Health supervision for children with Down syndrome. *Pediatrics*. 2011; 128(2):406

j. Recognize the typical phenotypic features associated with Down Syndrome

- Newborn:
    - Flat facial profile, Slanted palpebral fissures, Hypotonia, Transverse palmar (Simian), Excessive skin at nape of the neck, Hyper flexibility of joints, sandle toe gap, Incurved fifth finger, can have CHD (murmur)
- Head Neck Specific:
    - Upslanting palpebral fissures, Epicanthic folds, Flat facial profile/flat nasal bridge, Folded or dysplastic ears, Brushfield spots (grayish/brown **spots** on the periphery of the iris), Protruding tongue, Excessive skin at nape of the neck

k. Know the evaluation and recommendations for a child with Down Syndrome who wants to participate in athletic activities

- Children may have atlantoaxial instability. If this displacement of the atlanto- occipital joint increases it can cause pressure on the spinal cord.
- Participation in some sports (contact) puts child at increased risk.
- Children should be screened for cord symptoms (gait changes, abnormal use of arms and hands, bowel or bladder control changes or significant neck pain, head tilt weakness or sensory changes)
- **Routine radiographic evaluation is not recommended for asymptomatic children**
- If symptoms occur, the appropriate action is a plain cervical radiograph in neutral
    - if no concerns in neutral, obtain flexion and extension

l. Know the neurologic complications associated with Down Syndrome.

- As above, children may have compression of spinal cord
    - (gait changes, abnormal use of arms and hands, bowel or bladder control changes or significant neck pain, head tilt, weakness or sensory changes)

2. Fragile X Syndrome (FXS)

a. Understand the pathogenesis of FXS

- X linked syndrome characterized by expansion of the cytosine-guanine-guanine (CGG) trinucleotide repeat of the fragile X mental retardation 1 region (FMR1) of Xq27.3
- The number of repeats and th amount of methylation affects the phenotype.

- In full mutations, the FMR1 gene is more methylated, silencing the gene with a decrease in fragile X mental retardation protein (FMRP) – this protein is critical for brain development
    - Decrease in FMRP = decreased gene activity = intellectual disability
- **Most commonly inherited cause of intellectual disability**
- Typically, the normal allele has 5-44 CGG repeats**, 55-200 repeats are permutation carriers**
    - As the region expands, it becomes more unstable and likely to expand further when passed between mother and child.
- Those with >200 are full mutation FXS

b. Know the appropriate lab evaluation to establish the diagnosis of FXS

- **The Southern blot analysis test** determines if the gene has a full mutation, its approximate size, whether the gene has been "methylated," and if there is mosaicism of the gene (a mixture of different cell types).

- **The polymerase chain reaction (PCR) analysis** can determine the actual number of CGG repeats (a pattern of DNA) that are present in the Fragile X gene. For various technical reasons, PCR has not been the test of choice to diagnose a full mutation, but is quite accurate in determining premutation and normal gene repeat numbers. However, PCR is less expensive and quicker than Southern blot, and recent advances in technology have increased its ability to identify Fragile X full mutations. PCR may thus be the only test used in the near future

c. Recognize the phenotypic features of boys and men with FXS

- Primarily affects males, however females can be carriers or show milder phenotypes.
- Young boys: behavioral and developmental delay are often first symptoms
- Tall stature with relative macrocephaly
- Later childhood, classic features start to develop
    - Long narrow face with prominent forehead and protuberant ears
    - Macroorchidism is present in 80% adolescents and adults

d. Know the prevalence of FXS

- Affects roughly 1 in 4,000 boys and 1 in 8,000 girls

e. Know the developmental and behavioral characteristics of boys with FXS and permutation carriers

- Boys with the fragile X mutation have moderate to severe intellectual disability, most with IQs less than 55.
- Boys with full mutation noted to have borderline IQ should be tested for mosaicism.
    - Mosaicism correlates with FMRP levels, so the less methylation on the X FMR1 region due to mosaicism, the more functional the individual.

f. Understand the developmental and behavioral characteristics of girls with FXS mutation

- Girls with a full mutation of one of the FRAXA (fragile site, X chromosome, A site) allele can exhibit the same neurodevelopmental sequelae as their male counterparts with FXS
- Girls are described with a milder phenotype but the outcomes largely dependent on FMRP levels, determined by X chromosome inactivation.
- Mean cognitive ability for girls with a full mutation is in the borderline range, but one third are disabled.
- Girls demonstrate a relative strength in receptive language

g. Know the medical problems commonly associated with FXS

- Increased risk for seizures, more often in boys than girls
    - Complex partial are most common and respond well to treatment
- Visual deficits include strabismus refractive errors (hyperopia and astigmatism)
- Joint hypermobility, **pes planus**, hip dysplasia, scoliosis, clubfoot
- Adults may have mitral valve prolapse
- Premature ovarian failure (women who are premutation carriers – full carriers typically have intact fertility)

h. Understand the wide range of outcomes in children with FXS

- Individuals with FXS have a specific profile of strengths and weaknesses, with less variability than other causes of ID
    - Cognitive strengths:
        - Receptive vocabulary, visual memory, experiential l earning, imitations
    - Cognitive weaknesses:

        - Short term memory, working memory, spatial memory, auditory processing, sequential processing, abstract thinking, math, executive function.
    - IQ tends to decrease with age in adulthood.
    - Children with FXS may demonstrate language delays as early as 9m
    - Stronger expressive language skills compared to other children with different forms of ID
    - Difficulty with language pragmatics, estimated 15-50% with comorbid Autism dx.

i. Understand the importance of evaluating family members of children identified with FXS

- When a child with FXS is identified, families often have other children that may be showing symptoms or at risk of carrying premutation/mutation.
- FXTAS presents at 50-60y old, once a child is identified may be able to identify older population in the family
- Female carriers of premutation at risk for ovarian failure
- Adult population at higher risk for mitral valve prolapse

j. Know the medical problems in adult FX permutation carriers with FX tremor ataxia syndrome

- 30-40% of men and 4-8% of women carrying the premutation expansion develop fragile X tremor ataxia syndrome FXTAS
- Symptoms usually appear after age 50-60y and include ataxia, tremor, lower extremity neuropathy mood lability, and cognitive decline.

3. Sex Chromosomal Abnormalities

a. recognize the phenotypic features of Turner (XO) syndrome

b. Know the developmental and behavioral characteristics of girls with Turner (XO) syndrome

c. Recognize the phenotypic feature of boys with a karyotype of 47, XYY

d. Know the developmental and behavioral characteristics of boys with a karyotype of 47, XYY

e. know the developmental and behavioral characteristics of boys with Klinefelter syndrome

f. Know the developmental and behavioral characteristics of girls with a 47, XXX karyotype

g. recognize the phenotypic features of Klinefelter (XXY) at different ages

h. Know the cytogenetic heterogeneity associated with Turner (XO) syndrome
i. know the natural history of Turner (XO) syndrome
j. Know the medical problems commonly associated with Turner (XO) syndrome

4. Rett Syndrome
   a. Know the developmental and behavioral characteristics of Rett syndrome at different stages
   b. recognize the physical signs and symptoms of Rett syndrome at different stages
      - Girls with classic Rett syndrome go through 4 clinical stages
        - **Initial:** normal development for the first 6-18m of life, followed by a brief plateau in skills
        - **Second:** rapid regression in language and motor skills.
          - Purposeful fine motor hand movements are replaced by rhythmic hand wringing
          - Symptoms may include bruxism, apnea and hyperpnea, seizures, gait apraxia or ataxia and tremors
          - Head circumference may start to show a deceleration in growth where it was originally nml.
        - **Third:** latent, plateau during which the neurologic losses stabilize.
          - Begins at age 5-7y and can continue into adulthood
          - Apraxia and motor problems most prominent symptoms
          - Seizures, occurring in up to 90% of individuals with Rett Syndrome, typically begin in this stage.
        - **Final**: late motor deterioration characterized by decreased or lost mobility
          - Can develop dystonia, poor or hand deformities, scoliosis
          - No further decline in cognitive skills, communication or fine motor skills.
   c. Know the etiology and appropriate lab evaluation of Rett Syndrome

- Caused by deletions or mutations of the MECP2 gene.
    - Three types of *MECP2* mutations occur: missense, frameshift, and nonsense. The type of mutation may affect phenotypic expression
    - >99% are de nova mutations
- Blood DNA analysis to identify mutations of *MECP2*

<u>d. Know the plan and management of a child with Rett syndrome (associated issues)</u>

- No specific therapy is available for RTT. Management consists of treating the associated conditions
    - Nutrition/GI: Monitor feeding/growth and support as indicated, often with high caloric dense foods
    - Bone quality: low bone mineral density is common and may lead to fractures
        - Increase Ca, VitD intake. Monitor bones and lateral spine
    - Seizures: Most seizures associated with RTT are easily controlled and respond to standard antiepileptic drugs
        - Consider video EEG to discern behavioral event vs seizure.
    - Breathing: no known treatment for the awake breathing disturbances with alternating hyperventilation and apneic episodes that is often associated with RTT
    - Electrocardiogram (ECG) should be obtained when the diagnosis of RTT is made. If the QTc interval is >0.45, a cardiologist should be consulted
    - Monitor for scoliosis and sleep disturbancese
    - Therapies for motor issues and speech delay.

<u>e. Understand the prognosis for a child with Rett Syndrome</u>

- Girls typically survive into adulthood, but have a higher risk for sudden unexplained death than age matched peers.
    - May be related to increased incidence of long QT interval in girls with Rett Syndrome.

<u>f. Recognize the broader phenotype of children with MECP2 mutation who do not fit the clinical criteria for Rett Syndrome.</u>

- Atypical Rett syndrome ranges in severity.
    - Most severe is with onset before 6 months of age, characterized by hypotonia and infantile spasms
    - A milder form is similar to classic Rett, but with regression occurring later and more gradually

- Another milder variant is with less regression and milder intellectual disability.

- MECP2 is NOT universally lethal in boys
  - MECP2 mutation identified in 1.3-1.7% of males with ID with three phenotypes:
    - First: same MECP2 mutation as girls with Classic Rett. Boys develop severe neonatal encephalopathy and die before 2 years of age
    - Second: mutation that is not seen in girls with Rett. Boys have mild to severe intellectual disability and can survive into adulthood
    - Third: associated with the complete duplication of MECP2, is severe. Boys have infantile hypotonia, recurrent respiratory infections, severe intellectual disability, absence of speech, seizures and spasticity.

5. Trisomy 13 and Trisomy 18
   a. Recognize the phenotypic features of Trisomy 13
      - The classic triad is micro/anophthalmia, cleft lip and/or palate, and postaxial polydactyly
        - but the clinical presentation in patients with trisomy 13 can be quite variable
      - Abnormalities observed in ≥50 percent :
        - Central nervous system (CNS) – **Holoprosencephaly** with incomplete development of forebrain and olfactory and optic nerves, severe intellectual disability, deafness, **cutis aplasia**
        - Craniofacial – Abnormal auricles, microphthalmia/anophthalmia, colobomata, sloping forehead (fissure or cleft of the iris, ciliary body, or choroid)
        - Skin and limbs – Capillary hemangiomata, simian crease, hyperconvex narrow fingernails, polydactyly of hands and sometimes feet, prominent heel
        - Cardiac – **Found in approximately 80 percent of patients**; includes ventricular septal defect (VSD), patent ductus arteriosus (PDA), atrial septal defect (ASD), and dextroposition
        - GI/Genitalia –, **omphalocele,** Cryptorchidism in males; bicornuate uterus in females

b. Know the developmental and behavioral characteristics of children with Trisomy 13
    - The median survival for liveborn children is seven days, and 91% die within the first year, with the majority (approximately 80%) dying within the first month of life
    - Severe/Profound ID, seizures and failure to thrive common for those that survive >1 year.
c. Recognize the phenotypic features of Trisomy 18
    - major phenotypic features include **intrauterine growth restriction (IUGR**), small mouth, micrognathia, pointy ears, horseshoe kidney, and **flexed fingers, with the index finger overlapping the third finger and the fifth finger overlapping the fourth**. Congenital heart disease occurs in greater than 50 percent of affected individuals. Ventricular septal defects and patent duct arteriosus are the most common defects. The gastrointestinal system is involved in approximately 75 percent of cases. Meckel diverticulum and malrotation are the predominant abnormalities. **Rocker bottom feet common**
d. Know the developmental and behavioral characteristic of children with Trisomy 18
    - 50% of affected infants die within the first two weeks of life, and only 5 to 10 % survive the first year
    - Severe/profound ID for those that survive >12m with major medical concerns.

6 . William's Syndrome

a. Know the developmental and behavioral characteristics of children with Williams syndrome
    - Mild-to-moderate intellectual disability is common, with uneven cognitive disabilities.
    - Verbal and memory performance is less impaired than visual-spatial perception
    - Tend to be very social, gregarious, and often overly friendly with strangers.
    - Behavioral abnormalities include anxiety and attention deficit disorder
b. Recognize the phenotypic features of Williams Syndrome
    - Periorobital fullness, wide mouth, full lips, long philtrum, small chin and stature.
    - "Stellate Iris" and 'musical talent', hyperacusis, idiopathic hypercalcemia.
    - **Super aortic valve stenosis**
c. Know the medical problems commonly associated with Williams Syndrome
    - Superaortic valve stenosis in ~ 70% of children

- increased risk for myocardial ischemia, acute hemodynamic deterioration, and sudden death because of their cardiovascular anomalies
- Renal abnormalities may be present due to increased calcium and/or vessel elasticity abnormalities such as renal artery stenosis.
- Constipation can be severe leading to diverticulosis

d. Know the etiology and appropriate lab investigation for diagnosis of Wiliams Syndrome

- Diagnosed by FISH 7q11.23 (this should also be picked up by microarray due to microdeletion of 7q11)
- Region includes the ELN gene which codes for Elastin

7. Prader Willi Syndrome and Angelman Syndrome

a. Know the developmental and behavioral characteristics of children with PWS

- Infants with PWS have significant delayed motor milestones
  - Sitting emerges at an average age of 12months and walking at 24m
- Global developmental delays are present in infancy
  - Cognitive outcomes in PWS are typically in the mild to moderate range of ID (mean IQ of 65)
  - Children have great difficulty with deficits in adaptive skills as well
- Class behavioral phenotype:
  - Extreme rigidity and persistent tantrum behaviors in early childhood
  - Comorbidity of ADHD and Autism
  - Behaviors are often food driven, so careful control of access to food is important in behavioral management
    - Food should NEVER be used as a reward or punishment
  - Older children often develop perseverative speech patterns and compulsive behaviors including skin picking
  - Some adolescents and adults meet the diagnostic criteria for OCD and/or psychosis

b. Know the etiology and appropriate lab evaluation for a diagnosis for PWS

- Loss of paternal gene expression in the 15q11-q13 domain
- Can result from microdeletion in the paternally derived chromosome (65-70% of cases) OR maternal uniparental disomy (30%) OR imprinting errors

including de novo methylation of paternal alleles (~1%)

- A CGH + SNP is sufficient to start with, and will account for 75% of uniparental disomy (by noting the elongated chain of nucleotide polymorphisms at the 15q region) as well as microdeletions.
- methylation studies can unequivocally identify PWS but will not determine type of genetic change (UPD/imprinting error)
- A FISH will directly identify the 15q11 microdeletion but will not distinguish between PWS and Angelman.

Smith, A., & Hung, D. (2017). The dilemma of diagnostic testing for Prader-Willi syndrome. Translational pediatrics, 6(1), 46–56. https://doi.org/10.21037/tp.2016.07.04

c. understand the natural history of Prader Willi Syndrome (PWSH)

TABLE Suggested Criteria for Prompting Molecular Testing for PWS

Age at Assessment	Features Sufficient to Prompt DNA Testing
Birth to 2 y	Significant hypotonia with poor suck and difficulty with weight gain
2–6 y	Congenital hypotonia with history of poor suck; global developmental delay
6–12 y	History of congenital hypotonia with poor suck (hypotonia often persists), global developmental delay, and excessive eating (hyperphagia; obsession with food) with central obesity if uncontrolled
13 y through adulthood	Cognitive impairment, usually mild mental retardation, excessive eating (hyperphagia; obsession with food) with central obesity if uncontrolled, and hypothalamic hypogonadism and/or typical behavior problems (including temper tantrums and obsessive-compulsive features)

Adapted from Gunay-Aygun M, Schwartz S, Heeger S, O'Riordan MA, Cassidy SB. *Pediatrics*. 2001;108(5). Available at: www.pediatrics.org/cgi/content/full/108/5/e92.

Abbreviation: PWS, Prader-Willi syndrome.

Reprinted with permission from McCandless SE, The Committee on Genetics. Clinical report - health supervision for children with Prader-Willi syndrome. *Pediatrics*. 2011;127(1):195-204.

d. Know how to plan the management for a child with PWS

- Hypothalamic insufficiency causes lack of food satiety, poor temp control, central apnea, and endocrinology abnormalities (growth hormone deficiency, hypothyroidism and adrenal insufficiently)
    - Hyperphagia can occur at age 1-6y. We often see a biphasic pattern with FTT in infancy followed by

the hyperphagia and weight gain (obesity) into toddler years.
    - Hyperphagia is notable and includes stealing food, stealing things to get food, eating non-food items
    - Monitoring for obesity related disease is important as well as maintaining a well balanced diet and exercise regimen.
- Hypothyroidism should be monitored/screened for in early childhood
    - Children may experience early adrenarche but puberty is often delayed.
    - Short stature typical
        - Growth hormone treatment has shown benefits including improved linear growth, increased lean body mass and less severe delays in development – however, risk of tonsillar hypertrophy and increased risk of apnea.
- Obstructive sleep apnea occurs in 80% - sleep study screening is recommended in childhood
- Children should be monitored for scoliosis (due to hypotonia)
- Should also receive an annual vision screening due to strabismus and nystagmus concerns.

e. Know the developmental and behavioral characteristics of children with Angelman Syndrome

- Delays apparent from infancy with No regression
- Developmental issues include neonatal hypertonia followed by progressive hypertonia, gait ataxia and severe language impairment.
- Seizures of multiple types occur and can start in infancy
- Children appear happy, including laughing, smilng and excitability.
- Autism disorder can occur and is associated with a mutation in MECP2 that is also responsible for Rett Syndrome
    - When this gene deletion is present, it alters the expression of the UBE3A gene, thus the interaction of MECP2 and UBE3A mutations are responsible for autism in Angelman

f. Recognize the phenotypic features of PWS at different developmental stages

- See above regarding FTT in infancy with obesity into childhood/adult.
- Distinctive facial features such as a narrow forehead, almond-shaped eyes, and a triangular mouth; short stature; and small hand and feet.
- Some people with Prader-Willi syndrome have unusually fair skin and light-colored hair.
- Both affected males and affected females have underdeveloped genitals.

https://ghr.nlm.nih.gov/condition/prader-willi-syndrome

g. Recognize the phenotypic features of Angelman Syndorme
   - Behavioral phenotype of 'happy and excitable" children often flap hands
   - Ataxia
   - Facial features are 'coarse', skin and hair can be fair and scoliosis may be present

h. Know the etiology and appropriate ab evaluation to establish the diagnosis of Angelman syndrome.
   - inactivate or deletion of the maternal copy of the UBE3A gene.
     - Most cases of Angelman syndrome (about 70 percent) occur when a segment of the maternal chromosome 15 containing this gene is deleted.
     - In other cases (about 11 percent), Angelman syndrome is caused by a mutation in the maternal copy of the UBE3A gene.
     - Rarely, Angelman syndrome can also be caused by a chromosomal rearrangement called a translocation, or by a mutation or other defect in the region of DNA that controls activation of the UBE3A gene.
       - These genetic changes can abnormally turn off (inactivate) UBE3A or other genes on the maternal copy of chromosome 15.
   - CGH will identify the microdeletion but not identify PWS from Angelman. **A methylation study will determine if maternal or paternal copy present.**
   - A FISH can also directly identify the microdeletion of 15q11 but not differential paternal versus maternal copy.

8. **8. 22q11.2 deletion syndrome**

a. Know the appropriate laboratory evaluation to establish the diagnosis of 22q11.2 deletion syndrome

- 1 in 5,900 births
- classic triad of features is conotruncal cardiac anomalies, hypoplastic thymus, and hypocalcemia
- If clinical suspicion obtain:
    - Cardiac evaluation and echocardiogram (urgently)
    - Serum calcium and phosphorus levels
    - Complete blood cell count with differential to evaluate for lymphopenia
    - Chest radiograph to evaluate for absence of a thymic shadow
    - Renal ultrasound to assess for structural genitourinary tract abnormalities
    - T and B cell subsets by fluorescence-activated cell sorting (FACS)
    - Immunoglobulin levels and, if appropriate, antibody responses to vaccines
- Genetic testing: FISH or CGH
- the TREC count on NBS is low (if performed)

b. Know the developmental and behavioral characteristics of children with 22q11.2 deletion
syndrome

- Second most common cause of developmental delay after Down syndrome
- Specific learning disabilities
- speech and language impairment
- behavioral abnormalities

c. Recognize the phenotypic features of 22q11.2 deletion

- Cleft palate
- Cardiac anomalies
- Cervical spine anomalies
- Pierre Robin sequence

9. Other genetic disorders

a. Know the etiology and appropriate laboratory evaluation to establish the diagnosis of Smith-Magenis syndrome :

- Smith-Magenis syndrome is caused by a deletion of chromosome band 17p11.2
- occurring in 1 in 25,000.
- Diagnosed with CGH or FISH. (Ref: Nelsons, 108)

b. Know the developmental and behavioral characteristics of children with Smith-Magenis syndrome:

- Common findings with Smith-Magenis include: brachycephaly, midface hypoplasia, prognathism, myopia, cleft palate, short stature, severe behavioral problems and intellectual disability. (Ref: Nelsons 81.3)
- Most affected children have moderate ID.

- The physical features of frontal prominence; hoarse, deep voice; and coarse facial features (eg, heavy brows, synophrys, prognathism) may not manifest until late childhood.
- Children with Smith-Magenis syndrome exhibit unusual behaviors including self-hugging, pulling out fingernails and toenails, and insertion of foreign objects into their body.
- Typically have sleep problems because of circadian rhythm disturbances.
- Sleep problems and self-injurious behaviors increase with age.
- can be hyperactive and aggressive.

c. Know the etiology of neurofibromatosis types I and II: (Ref Nelsons 596.1)

- Autosomal dominant
- Type 1 and Type 2 are two separate entities (clinically and genetically distinct diseases)
- NF1 most prevalent type (1 in 3,000 births) caused by dominant loss of function mutation in NF-1 gene (17q11.2)
- NF2 rarer (1 in 25,000 births) caused by mutation of NF-2 gene (22q1.11)
- NF-1 clinically diagnosed when any 2 of the following 7 features are present
    - 6 or more café-au-lait macules larger than 5mm in greatest diameter in prepubertal individuals and larger than 15mm in greatest diameter in pospubertal individuals
    - Axillary or inguinal freckling
    - 2 or more iris Lisch nodules
    - 2 or more neurofibromas or 1 plexiform neurofibroma
    - Distinctive osseous lesion (sphenoid dysplasia or cortical thining of long bones
    - Optic gliomas
    - A first-degree relative with NF-1 whose diagnosis was based on aforementioned criteria
- NF-2 diagnosed when 1 of the following 4 features is present
    - Bilateral vestibular schwannomas
    - Parent, sibling, or child with NF-2 and either a unilateral vestibular schwannoma or any two of the following: meningioma, schwannoma, glioma, neurofibroma, posterior subscapular lenticular opacities
    - unilateral vestibular schwannoma and any two of the following: meningioma, schwannoma, glioma, neurofibroma, posterior subscapular lenticular opacities
    - multiple meningiomas (2 or more) and unilateral vestibular schwannoma or any two of the following: schwannoma, glioma, neurofibroma, cataract.

d. Know how to plan the diagnostic evaluation of a child with neurofibromatosis types I and II

- NF-1
    - Clinical diagnosis as above
    - Should have clinical exam, neurologic assessment, and ophthalmologic exam annually
    - BP monitoring annually

    - Scoliosis evaluation
    - Neuropsychologic and education testing as indicated
    - Genetic testing not necessary as this is a clinical diagnosis, consider if child only has 1 of the 7 criteria as above.
    - MRI only in individuals who are symptomatic
- NF-2
    - Clinical diagnosis as above
    - Initial diagnosis: Ophthalmologic exam, MRI brain, audiology exam

e. Know the medical problems commonly associated with neurofibromatosis types I and II

- NF-1
    - Learning disabilities (~30%)
    - Seizures (~8%)
    - Moyamoya syndrome
    - Precocious puberty
    - Malignant neoplasm (~3%) with incidence of pheochromocytoma, rhabdomyosarcoma, leukemia, and Wilms tumor higher than in general population
    - Scoliosis (~10%)
    - Also at risk for hypertension from renal vascular stenosis or pheochromocytoma
- NF-2:
    - Tinnitus, hearing loss, facial weakness, headache, unsteadiness may appear during childhood though signs of a cerebellopontine angle mass are more commonly present in 2nd and 3rd decades of life.
    - Neurologic lesions as above in diagnostic criteria
    - Ophthalmologic lesions to include cataract, retinal hamartomas, epiretinal membranes
    - Cutaneous lesions: skin tumors, skin plaques, subcutaneous tumors, intradermal tumors (rare)

f. *Know the developmental and behavioral characteristics of children with neurofibromatosis types I and II*

- NF-1
    - intellectual disability (6%-7%)
    - learning disorders (50%-75%)
    - attention-deficit/hyperactivity disorder
    - executive function deficits
    - autism spectrum disorder
    - developmental coordination disorder
    - seizures
    - sleep disruption
- NF-2
    - There is no association between NF2 and neurodevelopmental and neurobehavioral disorders

g. Know the etiology and appropriate medical evaluation to establish the diagnosis of tuberous sclerosis

- Autosomal dominant, variable expression
- Prevalence 1 in 6000 newborns
- TSC1(9q34) and TSC2(16p13) genes; TSC1 encodes hamartin, TSC2 encodes tuberin
- Diagnosed clinically when at least 2 of the major criteria or one major plus 2 minor criteria are present
- Major criteria
    - Cortical tuber
    - Subependymal nodule
    - Subependymal giant cell astrocytoma
    - Facial angiofibroma or forehead plaque
    - Ungual or periungal fibroma (non-traumatic)
    - Hypomelanotic macules (>3)
    - Shagreen patch
    - Multiple retinal hamartomas
    - Cardiac rhabdomyoma
    - Renal angiomyolipoma
    - Pulmonary lymphangioleiomymatosis
- Minor criteria
    - Cerebral white matter migration lines
    - Multiple dental pits
    - Gingival fibromas
    - Bone cysts
    - Retinal achromatic patch
    - Confetti skin lesions
    - Nonrenal hamartomas
    - Multiple renal cysts
    - Hamartomatous rectal polyps
- MRI q1-3 years
- Consider genetic testing if high suspicion but not all criteria met.

h. Recognize the phenotypic features of tuberous sclerosis

- Extremely heterogeneous disease with wide clinical spectrum
- Phenotypic features as above in the diagnostic criteria
- Hallmark of the disease is involvement of the central nervous system
- >90% of patients show typical hypomelanotic macules that have been likened to an ash leaf on the trunk and extremities

i. Know the developmental and behavioral characteristics of children with tuberous sclerosis

- ADHD (~50%)
- Aggressive behaviors

- Cognitive impairment (can be severe, follows bimodal distribution 70% normal IQ, 30% with IQ<20)
- Autism spectrum disorder
- Specific learning disabilities, attention deficits, executive control impairments, language deficits, memory problems, visual-spatial problems, self-injurious behaviors, aggressive outbursts, difficult temper tantrums, and chronic sleep problems have all been reported to be relatively more common among children who have TSC than those who do not.

j. Know the medical problems commonly associated with tuberous sclerosis

- Epilepsy
- May present in infancy with infantile spasms and hypsarrhythmic EEG pattern
- Retinal hamartomas are present in 90% of cases
- Multiple dental pits of permanent teeth and gingival fibromas (fleshy growths on gums) are also common findings.
- CNS manifestations vary, depending on the size, location, and growth of cortical tubers, subependymal nodules (SENs), and subependymal giant cell astrocytomas (SEGAs). Obstruction of foramen of Monro can lead to obstructive hydrocephalus. Other complications include cranial nerve palsies, motor and sensory deficits, cerebellar dysfunction, and abnormal gait.
- Renal complications are the most common cause of mortality in patients who have TSC. Life-threatening retroperitoneal hemorrhage from rupture of aneurysmal blood vessels that feed angiomyolipomas has been reported. Polycystic kidney disease can cause hypertension and progressive renal failure. Patients with renal lesions also are predisposed to urinary tract infections and nephrolithiasis.
- Cardiac rhabdomyomas can involve the conducting pathways resulting in arrhythmias. Dilated cardiomyopathy, nonimmune hydrops fetalis, and fetal or neonatal death have been reported resulting from diffuse infiltration of the myocardium by rhabdomyoma.
- Vascular aneurysms have been described in abdominal, axillary, basilar, and pulmonary arteries, although the exact prevalence is not known.

k. Recognize the phenotypic features of neurofibromatosis types I and II

- NF-1
    - café-au-lait macules
    - Axillary or inguinal freckling
    - iris Lisch nodules
    - neurofibromas or 1 plexiform neurofibroma
    - Distinctive osseous lesion (sphenoid dysplasia or cortical thining of long bones
    - Optic gliomas
- NF-2 diagnosed when 1 of the following 4 features is present
    - Bilateral vestibular schwannomas
    - unilateral vestibular schwannoma
    - meningioma

- schwannoma
- glioma
- neurofibroma
- posterior subscapular lenticular opacities
- cataract.

l. Know the phenotypic features of other neurocutaneous syndromes, eg, Sturge-Weber, incontinentia pigmenti

- Sturge-Weber: sporadic vascular disorder, GNAQ gene mutation
  - Phenotypic features
    - Port wine stain present at birth
    - Glaucoma
- Incontinentia pigmenti: IKBKG gene. X-inactivation of an X-linked dominant gene which is lethal in males.
  - Disease has 4 phases
    - 1st: birth or shortly after- erythematous linear streaks, resolves around 4mo. Lesions may be confused for herpes simplex, vesicles present
    - 2nd: Blisters resolve and become verrucous plaques, involute around 6mo
    - 3rd: Pigmentary stage, hyperpigmentation in macular whorls, fade in adolescence
    - 4th: hairless, anhirotic, hypopigmented patches or streaks
  - Dermatologic abnormalities (as above)
  - Dental abnormalities (late dentition, hypodontia, conical teeth)
  - Ocular abnormalities (neovascularization, microphthalmos, optic nerve atrophy, cataracts
  - Seizures

m. Know the developmental and behavioral characteristics of children with other neurocutaneous syndromes

- Sturge-Weber
  - Intellectual disability
  - Severe learning disabilities
  - Intractable epilepsy
- Incontinetia pigmenti
  - Intellectual disability

D. Metabolic disorders

1. Phenylketonuria (PKU)

a) Definition: PKU is an inbo rn error of metabolism caused by a mutation of the phenyla]anine hydroxylase (PAR) gene, resulting in a reduction of the rate of

conversion of phenylalanni e to tyrosine. Elevated blood phenylalanine levels are toxic to the brain.

b) Types
   i. Classic PKU: PAH severely reduced or absent
   ii. Variant PKU: some enzyme activity
   iii. Non-PKU hyperphenylalaninemia

c) Genetics
   - Autosomal recessive
   - Various mutations of the PAH gene cause different type

a. Understand the importance of neonatal screening for PKU

1. Infants born to mothers with untreated PKU are at high risk for intellectual disability,
low birth weight, poor growth, heart defects, microcephaly, and behavior problems.
2. Girls with PKU should be counseled regarding sexuality, pregnancy, and importance of dietary control

b. *Know the principles of dietary treatment of individuals with PKU*

1. Low-phenylalanine diet

a) The degree of intellectual disabilities, attention deficits, and psychiatric illnesses are related to control of blood phenylalanine levels and timing of onset of treatmen.t

b) Life-long treatment is recommended although some dietary relaxation as a patient grows older may be possible (except in pregnancy).

c) Dietary control is important prior to onset of pregnancy

c. Know how to plan appropriate counseling for a girl with PKU about sexuality, pregnancy, and genetic risks

Pregnancy and Sexuality Risks

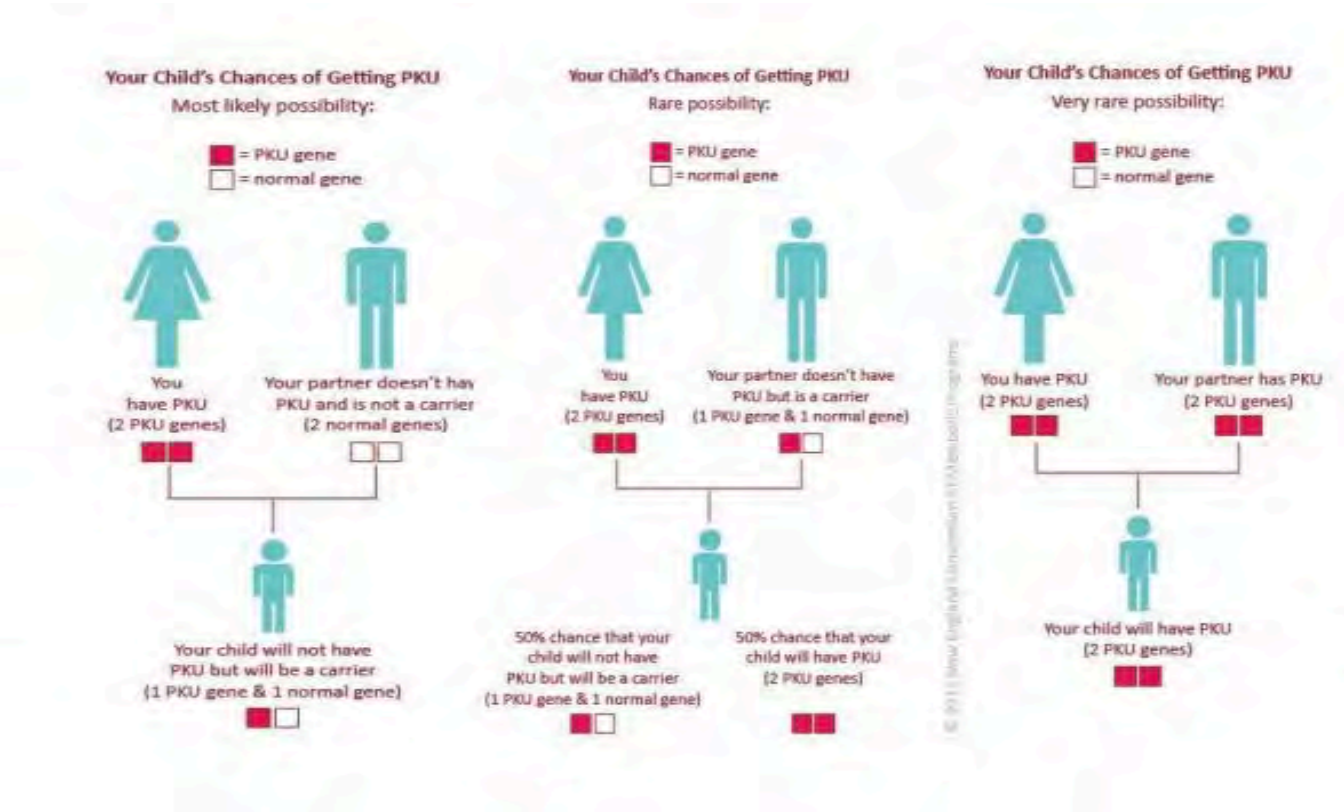
- Any woman with PKU who is planning to have a baby needs to be very careful to control her phenylalanine levels. Babies born to mothers who have high levels of phenylalanine are at risk for having a very small head, intellectual disability, growth problems, and heart problems.
- During Pregnancy there needs to be the following nutritional goals.
- Controlling blood Phenylalanine (Phe) concentrations within the recommended range for PKU pregnancy of 150-250 mmol/l
- Maintaining adequate nutrition
- Ensuring appropriate pregnancy weight gain
- Supplementation of Tyrosine
- DHA Supplementation (essential fats)

Genetic Risks

- For a child to inherit PKU, both the mother and father must have and pass on the defective gene. This pattern of inheritance is called autosomal recessive.

- It's possible for a parent to be a carrier — to have the defective gene that causes PKU, but not have the disease. If only one parent has the defective gene, there's no risk of passing PKU to a child, but it's possible for the child to be a carrier.

- Most often, PKU is passed to children by two parents who are carriers of the disorder, but don't know it.



d. *Know the phenotypic features of PKU*

1. Progressive brain damage and intellectual disability
2. Musty or mouse-like odor
3. Light skin and hair
4. Skin disorders (e.g. eczema)

e. *Know the developmental and behavioral characteristics of children with PKU*

- Historically untreated PKU exhibited severe behavioral disturbances to include psychotic, autistic and aggressive disorders.
- Behavioral, emotional and social problems in older children and adults
- Many studies converge upon a behavioral pattern that resembles ADHD symptoms.
    - Why people with PKU may have ADHD:
        - 2 main reasons why psychologists and researchers have focused on ADHD as a potential problem area in people with PKU:
            - 1. ADHD has been linked to lower levels of the neurotransmitter dopamine, and there is evidence suggesting that dopamine levels may be lower in individuals with PKU.
            - 2. Clinicians have noticed that there are certain behavioral and learning problems in diet-treated PKU patients despite having normal-range intelligence. Studies have shown that many of these symptoms are similar to those found in individuals diagnosed with ADHD.

- The research focus has shifted away from IQ to determining if more subtle psychological/emotional changes occur in PKU patients adhering to diet therapy.
- Some of the many characteristics of people with EF difficulties include disorganization, being easily frustrated, and poor judgment.
- Processing speed and can be measured by a variety of tasks with time limits. People who suffer from problems in information processing speed might take longer to start/complete complex tasks and appear to struggle.
- One strong piece of evidence suggests that if one stays within the current blood Phe range for their age group then information processing will not be affected.
- To date, it appears that math skills may be affected in some individuals with PKU.
- Psychiatric disorders occur more frequently in PKU showed no significantly different overall rate than the general population.

f. Know the evaluation of suspected PKU

- Newborn metabolic screening (most cases detected)
    - Early detection and treatment critical to prevention of brain damage
- Serum phenylalanine levels
- Urine ferric chloride test
- False negative screening can occur rarely (usually through mishandling of the specimen or transmission of results). If the following are present, obtain a quantitative serum amino acid assay: plateau in development or regression, suggestive physical features, consanguinity, emerging neurological abnormalities)

2. Mucopolysaccharidoses

a. *Understand the usual natural history of developmental skills in children with mucopolysaccharide disorders*

- Mucopolysaccharidoses (MPS) are a group of rare, inherited lysosomal storage disorders, caused by mutations in lysosomal enzymes involved in the degradation of glycosaminoglycans (GAGs).
- They often present with significant neurological signs and symptoms, including impaired cognitive abilities, difficulties in language and speech, and/or behavioral and sleep problems.
- patients can show autistic-like behavior
- Patients with MPS I and MPS IH/S (the intermediate phenotype) generally show milder or no cognitive involvement, milder somatic symptoms, and slower disease progression
- MPS I patients also typically show an early delay in attainment of motor skills, considered to be more related to the patients' limited range of joint motion, progressive orthopedic manifestations, and peripheral neuropathy, rather than to brain abnormalities
- MPS I patients are perceived as social, compliant, and somewhat fearful [3], [40]. When they become older, treated MPS IH patients can present with attention problems
- MPS II - develop severe speech and language delay [50], [51], which mirrors the cognitive decline. Hearing loss is almost universal among MPS II patients and is generally diagnosed at around 2 years of age
    - MPS II - frequently observed behavioral problems in neuronopathic patients are hyperactivity, challenging behaviors, frustration, and impulsivity. In addition, approximately half of these patients develop perseverative chewing behavior
    - Many MPS II patients have sleeping problems
- MPS III, or Sanfilippo syndrome, is associated with severe neurological manifestations and, unlike the other MPS disorders, only mild somatic involvement
    - Around 3 years of age and declines rapidly thereafter, resulting in cognitive skills at less than a two-year-old level after the age of 6 years
    - Speech and language delay are the most frequent initial symptoms of MPS IIIA and IIIB. Language delay may be apparent by the age of 2 years, before cognitive decline starts [6], [57]. Only 50% of these patients acquire the capacity to associate two words before the age of 3 years [7], [54], [55]. Hearing loss, which generally develops in some children with MPS III around 2–3 years of age [6], [7], when children acquire most language, may also contribute to the speech and language delay.
    - Patients with rapidly progressing MPS IIIA can usually acquire adaptive skills up to approximately 4 years of age, after which these are lost [57]. Fine motor skills reach a plateau at around 2 to 3 years of age, mirroring cognitive decline [6]. As MPS III patients have minimal musculoskeletal

problems, development of gross motor skills tend to be preserved until 5 to 6 years of age.

Developmental and behavioral aspects of mucopolysaccharidoses with brain manifestations — Neurological signs and symptoms

Elsa G.Shapiro[ab]Simon A.Jones[c]Maria L.Escolar[d]

b. Recognize the phenotypic features suggestive of a mucopolysaccharide disorder

Common features

1. Coarse facial features
2. Short stature
3. Skeletal abnormalities
4. Thickened skin
5. Enlarged liver and spleen
6. Excessive body hair
7. Developmental delay/intellectual disability (depending on type)
8. Visual impairment
9. Hearing impairment

c. Know the etiologies and appropriate laboratory evaluation to establish the diagnosis of the major mucopolysaccharide disorders

Mucopolysaccharidoses (MPSs) are a group of lysosomal storage diseases, each of which is produced by an inherited deficiency or malfunctioning of an enzyme involved in the degradation of mucopolysaccharides, now called glycosaminoglycans (GAGs)

- GAGs are long chains of sugar carbohydrates that help build bone, cartilage, tendons, corneas, skin, and connective tissue. GAGs are also found in the fluid that lubricates the joints.
- GAGs (e.g. chondroitin, heparan, dermatan, keratan sulfates) accumulate in the lysosomes leading to permanent, progressive cellular damage, affecting appearance, physical abilities, organ functioning, often mental development

d. Know the developmental and behavioral characteristics of children with mucopolysaccharide disorders

- Think of MPS (or other lysosomal storage disease) if a child's development plateaus or regresses.

	MPS IH [15], [23], [24], [26], [33], [40], [41]	**MPS II, rapidly progressing [26], [42], [50], [75]**	**MPS IIIA and B [6], [7], [9], [10], [26], [57], [76]**
Neurocognition	Plateaus at ± 3 years of	Plateaus at 4 to 4.5 years	Plateaus at 2 to 3 years of

	age, then progressive decline	of age, then progressive decline	age, then progressive decline (skills of a 6-month-old at 6 years)
Speech & language	Limited skills develop until 2 years of age, then decline Probably affected by hearing loss	Mirrors cognitive delay and decline Affected by hearing loss	Decline at ± 2 years of age (before start of cognitive impairment) Affected by hearing loss
Motor skills	Decline from ± 2 years of age Gross motor skills affected before fine motor skills	Plateaus at 4 years Gross motor skills affected ± 1 year of age before fine motor skills	Mirrors decline in cognition Loss of skills after 4 years of age
Behavior	Young: somewhat fearful Older children: attention problems Adolescents: depression and social withdrawal	Hyperactive & aggressive at ± 4 years of age Sleeping problems	Aggressive behavior at 3 to 4 years of age Decreased attention Lack of fear, acquired autistic deficits Sleeping problems
Brain abnormalities[a]	Most are evident at < 2 years of age: •Hydrocephalus •↑ ICP •Ventriculomegaly •↑ PVS •Atrophy •↓ FA corpus callosum	Variable age of detection (median age 6 years): •Hydrocephalus •Ventriculomegaly •↑ PVS •Atrophy •Seizures/epilepsy	Variable age of detection (median age 5 years): •Ventriculomegaly •↑ PVS[b] •Atrophy •Seizures/epilepsy

Developmental and behavioral aspects of mucopolysaccharidoses with brain manifestations — Neurological signs and symptoms

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3. Purine metabolism

A. Definition: rare, inherited disorder caused by a deficiency of the enzyme hypoxanthineguanine phosphoribosyltranferase (HPRT), responsible for re-cycling purines
   - Lack of HPRT causes build-up of uric acid in all body fluids

B. Genetics
   - X-linked

a. Know the developmental and behavioral characteristics of children with Lesch-Nyhan syndrome

b. Know the evaluation of suspected Lesch-Nyhan syndrome

1. Elevated serum and urine uric acid levels (variable)
2. Macrocytic anemia (Vit B12, folate, and iron typically normal)
3. HPRT enzyme activity in blood or tissue
4. ID of genetic mutation in HPRT gene

c. Recognize the phenotypic features of Lesch-Nyhan syndrome

1. Dystonia/spasticity
2. Growth failure
3. Moderate developmental delay/intellectual disability (avg. IQ==60)
    i) 3-12 months: delayed motor development with hypotonia
    ii) 6-18 months: abnormal involuntary movements and spasticity
4. Aggressive, impulsive behavior
5. Self-mutilation
6. Arthritis
7. Renal failure

d. Plan the management of a child with Lesch-Nyhan syndrome

- A striking feature of LNS is self-mutilating behaviors – characterized by lip and finger biting – that begin in the second year of life. Abnormally high uric acid levels can cause sodium urate crystals to form in the joints, kidneys, central nervous system, and other tissues of the body, leading to gout-like swelling in the joints and severe kidney problems.

- Treatment for LNS is symptomatic. Gout can be treated with allopurinol to control excessive amounts of uric acid. Kidney stones may be treated with lithotripsy, a technique for breaking up kidney stones using shock waves or laser beams. There is no standard treatment for the neurological symptoms of LNS. Some may be relieved with the drugs carbidopa/levodopa, diazepam, phenobarbital, or haloperidol.

- The prognosis for individuals with LNS is poor. Death is usually due to renal failure in the first or second decade of life.
- Behavior treatment includes:
    a) Operant conditioning/desensitization/extinction
    b) Medications: benzodiazepines, gabapentin, neuroleptics

E. Infectious diseases

1. Meningitis and encephalitis

a. Recognize the neurological, developmental, and behavioral complications of various forms of meningitis

A. Neurological, developmental, and behavior consequences of meningitis

1. Hearing impairment
    a) S. pneumoniae: 25-35%
    b) H. influenzae and N. meningitidis: 5-10%
2. Neuromotor, LO, speech, and behavior problems: 10%

B. Association between neurological outcomes and CSF findings
1. Increased CSF protein, decreased CSF glucose; worse outcome
2.. Delayed sterilization of CSF increases risk of adverse neurological and hearing outcomes
3. Focal neurological signs and/or seizures worse outcome

2. HIV/AIDS

a. Know the neurological, developmental, and behavioral complications of pediatric HIV/AIDS and its treatment

- Acute encephalopathy, developmental delay or regression, mild to moderate cortical atrophy, learning disabilities, ADHD
- ZDV (zidovudine) or other ARV (ant iretroviral) agents may pose long term risks of neoplasia; organ system toxicities, or mitochondrial dysfunction.

b. Understand the natural history of pediatric HIV/AIDS

- Children progress to moderate symptoms in the 2nd year-of-life, remaining moderately sympotmatic for more than half their expected lives (Barnhart et al, 1996)

d. Understand the epidemiology of pediatric HIV/AIDS

- 6000 pregnant women with HIV give birth per year
    a) With treatment and preventive strategies (including no breast feeding), transmission to fetus/newborn 1-2%
    b) Perinatal transmission of HIV accounts for 90% of pediatric AIDS cases and almost all new HIV infections in children.

3. Chronic otitis media

a. Know the sensory, developmental, and behavioral consequences of chronic otitis media with effusion

A. Sensory, developmental, and behavioral consequences of chronic otitis media with effusion
1. Otitis media with effusion (OME) does not lead to long tenn adverse developmental outcomes. (Feldman, 2006)
2. Tympanostomy tubes in otherwise healthy children who have persistent middle-ear effusion do not improve developmental outcomes. (Paradise et al, 2007)
3. Conductive hearing loss secondary to OME may warrant early tympanostomy tube insertion

B. Factors that moderate impact of otitis media on development

1. Risk factors prompting lower threshold for monitoring hearing, speech, and language and for surgical intervention AAP Clinical Practice Guideline, 2004)

a) Permanent hearing loss independent of OME
b) Suspected or diagnosed speech and language delay or disorder
c) Autism Spectrum Disorder
d) Syndromes (e.g. Down) or craniofacial disorders that include cognitive, speech, and language disorders
e) Blindness or uncorrectable visual impairment
f) Cleft palate with or without associated syndromes
g) Developmental delay

F. Central nervous system (CNS) disorders

1. Seizures and epilepsy a. Differentiate between seizures and epilepsy
   b. Understand the international classification of epileptic syndromes
   c. Know how to plan the evaluation of a child with staring spells
   d. Know that the prevalence of seizures and epilepsy is higher in children with developmental disorders than in the general population
   e. Know the appropriate management for a child with epilepsy and attention deficit hyperactivity disorder
   f. Know that developmental and behavioral difficulties are more common in children with epilepsy than in children with other types of chronic physical illnesses
   g. Know how to plan the evaluation of a child with suspected seizures
   h. Know the etiologies and natural history of infantile spasms

2. Hydrocephalus
   a. Know the causes of hydrocephalus in children
   b. Know the developmental and behavioral characteristics of children with hydrocephalus

3. Stroke
   a. Understand the developmental and behavioral sequelae following a stroke at different ages
   b. Understand the natural history of language development in children following a stroke at different ages
   c. Know the causes of pediatric stroke at different ages

4. Traumatic Brain Injury (TBI)
   a. Differentiate between mild, moderate, and severe head injury on the basis of the Glasgow coma score
   b. Know the long-term management for a child with traumatic brain injury
   c. Understand the pathogenesis of brain injury with penetrating head trauma
   d. Know the developmental, cognitive, and behavioral consequences of penetrating injury to specific brain regions
   e. Understand the pathogenesis of brain injury with closed head trauma

f. Know the developmental, cognitive, and behavioral consequences of mild, moderate, and severe closed head injury

5. Central nervous system (CNS) tumors
   a. Know the developmental, cognitive, and behavioral consequences in children who have had CNS tumors
   b. Know the behavioral, developmental and neurological symptoms suggestive of a CNS tumor

6. Congenital CNS malformation
   a. Know brain malformations that may be found in individuals with no developmental or behavioral disabilities
   b. Know the brain malformations most commonly found in individuals with developmental disabilities (eg, holoprosencephaly, schizencephaly, lissencephaly)

G. Sensory defects

1. Visual impairments

- Visual impairment: significant limitation of visual capability resulting from disease, trauma, or congenital or degenerative conditions that cannot be corrected by conventional means, such as refractive correction, medication, or surgery.
- Social blindness: not looking at someone or acknowledging their presence
- Virtual blindness: visual impairment severe enough to result in functional blindness
- Total blindness: inability to distinguish light from darkness
  - Legal blindness visual acuity 20/200

a. Know the leading causes of visual impairment in childhood

a. Refractive errors
b. Strabismus/amblyopia
c. Structural defects of the eyes
d. Glaucoma
e. Nystagmus
f. Optic nerve atrophy/hypoplasia
g. Cataracts
h. Retinopathy of prematurity
i. Cortical abnormalities

b. Understand the importance of a functional vision assessment in determining which literacy media to use with significant visual impairment or blindness (eg, Braille, large print, optical modifications)

- A functional vision assessment (FVA) is an evaluation of the day-to-day visual skills of an individual who is visually impaired. A Functional Vision Evaluation is a legally required document.
    - The purpose is to supplement the results of the clinical eye exam with descriptions of the student's observable behaviors that may relate to vision.
- Functional Vision Assessment (FVA) should be done first, in order to determine what the student is able to see and how he or she is using his or her vision. This assessments to help to guide the team decision about the best instructional medium for a given student, such as braille, print, dual media (both print and braille), auditory, tactile or some combination.
- FVE identifies the student's range of visual function; determines how much usable vision a student has to perform visual tasks; and identifies priorities and strategies for intervention.
    - For students who have no vision, and functional vision reports are required for them, as well. The purpose of the report confirms the absence of vision and presents recommendations on how to modify instruction for the student.
    - Results of the functional vision assessment identify factors that help or hinder student performance and provide recommendations to the educational team about how to increase and enhance the student's visual efficiency.
    - Results will identify a student's visual skills with and without optical devices in a variety of environments and with a variety of levels of visual difficulty. Note ways in which visual fatigue or discomfort can be alleviated or minimized.
- Children taught Braille have higher literacy and employment rates as adults

c. Know how to assess development in children who are visually impaired

a. Medical
    i. General health
    ii. Associated health problems
b. Developmental
    i. Cognition. motor, speech/language, social, emotional
    ii. Only measurement instruments standardized on individuals with visual impairment should be used.
    iii. Assessors should have specialized training

d. Know how to plan the management of developmental/behavioral issues in children with a range of visual impairments

Medical Treatment of cause of impairment

a. Optimal correction of residual vision
b. Provision of compensatory experiences (examples)
    - Use of sound to stimulate reaching, crawling, exploration of the environment
    - Exploitation of tactile sense to stimulate exploration of the environment
        - Guidance in maintenance of proper

posture from early childhood
- Teaching generally accepted gestures
  - Teaching daily living skills in preschool years
  - Teaching usual early-childhood activities such as running, jumping, skipping, swimming, riding a tricycle
  - Individualized Education Plan (IEP) for adaptive school services
  -

c. Role of vision specialist: Assist parents, teachers, therapists with appropriate adaptive strategies

- A Teacher of Students with Visual Impairments (also called a Teacher of the Visually Impaired, a vision specialist, VI teacher, vision itinerant teacher, etc.) is typically a licensed special education teacher who has received certification and specialized training, in meeting the educational needs of students who are blind or have visual impairments ages birth through 21

- The role of the Teacher of Students with Visual Impairments (TVI) is to provide direct and/or consultative special education services specific to vision loss. The TVI provides support to students, teachers, and parents and acts as a liaison with community services. The TVI works with the educational team by advising the team about ways of enhancing the student's learning by adapting activities and materials to the student's abilities. Although the TVI is not an academic tutor, they may spend some time ensuring that the student understands concepts introduced in academic courses.

- A TVI will conduct Functional Vision Assessments - This evaluation is updated at a minimum, every three years to determine ongoing eligibility and need for school based vision services.

- As a result of the Functional Vision Assessment, the TVI may recommend other specialized evaluations as needed, particularly in low vision, orientation and mobility, and adaptive physical education.

- Other responsibilities of the TVI include:
  - ***Actively Participate in the Individualized Education Program (IEP)***
  - ***Recommend Educational & Instructional Strategies***
  - ***Ongoing Observations***
  - ***Use of Natural Environments to Address Goals***
  - ***Communication with Caregivers and Classroom Teachers***
  - Direct Instruction in the Expanded Core Curriculum
  - Choose appropriate educational materials,
  - Create a classroom environment that encourages independence, academic success,

- Ultimately prepare the student to be the most productive member of society they can be.

e. Understand the developmental and behavioral problems associated with severe visual impairment

c. Motor development
   i. Readiness-to-move milestones achieved on time (e.g. stand)
   ii. Lack of visual information leads to unwillingness to move
      1. Poor muscle and coordination development

d. Speech
   i. Speech milestones achieved on time
   ii. Differences in use and content
      a. Language patterns focused on self and less on actions or events outside of self
      b. Unaware of others' experiences

f. Understand the developmental and behavioral problems associated with mild to moderate visual impairment

- May need adaptations for school work
- Problems more like due to co-morbidities

g. Understand the developmental maturation of visual acuity in early childhood

Birth - Visual fixation present and Visual acuity at one mo: 20/400
6-9 wk - Fixation well-developed
3 mo - Visual following
4 mo - Accommodation
4 mo - Stereopsis

a. Timing and method of measuring visual acuity and abilities
   i. Newborn to 3 mo
      1. Red reflex
      2. Inspection
   b) 6 mos to I year
      1. Fix and follow with each eye
      2. Alternate occlusion
      3. Corneal light reflex
      4. Red reflex
      5. Inspection
   c) 3 years (approx)
      1. Visual acuity
      2. Corneal light reflex
      3. Stereoacuity
      4. Red reflex
      5. Inspection
   d) Older than *5* years

1. Visual acuity
2. Corneal light reflex/cover uncover
3. Stereoacuity
4. Red reflex inspection

Vision testing methods

a. Acuity

- Up to age 3
  1. Recognizable cartoon pictures (Allen test) or symbols of readily identifiable shapes (LH test)
- Up to age *5*
  1. Choosing which of four letter shapes are presented (HOTV test) or which direction a letter E is pointing (tumbling E test)
  2. *5* years and older
     a. Snellen letters or numbers

h. Understand the meaning of cortical visual impairment, and other forms of visual impairment

Cortical Visual Impairment (CVI) is a temporary or permanent visual impairment caused by the disturbance of the posterior visual pathways and/or the occipital lobes of the brain.

- The degree of vision impairment can range from severe visual impairment to total blindness.
- The degree of neurological damage and visual impairment depends upon the time of onset, as well as the location and intensity of the insult.
- It is a condition that indicates that the visual systems of the brain do not consistently understand or interpret what the eyes see. The presence of CVI is not an indicator of the child's cognitive ability.

Cause

The major causes of CVI are asphyxia, perinatal hypoxia ischemia ("hypoxia": a lack of sufficient oxygen in the body cells or blood; "ischemia": not enough blood supply to the brain), developmental brain defects, head injury, hydrocephalus, and infections of the central nervous system, such as meningitis, and encephalitis.

Characteristics

- Initially, children with CVI appear blind. However, vision tends to improve.
- Cortical Visual Impairment is a more appropriate term than Cortical Blindness.

- A lot of neurological disorders can cause CVI, and CVI often coexists with ocular visual loss, so the child should be seen by both a pediatric neurologist and a pediatric ophthalmologist.
- Cortical Visual Impairment is a difficult diagnosis.
  - It is diagnosed when a child has poor or no visual response and yet has normal pupillary reactions and a normal eye examination.
  - The child's eye movements are most often normal.
  - The result of an MRI (Magnetic Resonance Imaging) in combination with an evaluation of how the child is functioning visually, provide the basis for diagnosis.

Behavioral/Visual Characteristics

Children with CVI have different abilities and needs.

- Some children have good language skills and others do not.
- Spatial confusion is common in children with CVI because of the closeness of the occipital and parietal lobes of the brain.
- Habilitation should be carefully planned.

Common characteristics of visual function demonstrated by children with CVI:

- Vision appears to be variable: sometimes on, sometimes off; changing minute by minute, day by day.
- Many children with CVI may be able to use their peripheral vision more effectively than their central vision.
- One third of children with CVI are photophobic, others are compulsive light gazers.
- Color vision is generally preserved in children with CVI (color perception is represented bilaterally in the brain, and is less susceptible to complete elimination).
- The vision of children with CVI has be described much like looking through a piece of Swiss Cheese.
- Children may exhibit poor depth perception, influencing their ability to reach for a target.
- Vision may be better when either the visual target or the child is moving.

The behaviors of children with CVI reflect their adaptive response to the characteristics of their condition:

- Children with CVI may experience a "crowding phenomenon" when looking at a picture: difficulty differentiating between background and foreground visual information.
- Close viewing is common, to magnify the object or to reduce crowding.
- Rapid horizontal head shaking or eye pressing is not common among children with CVI.

- Overstimulation can result in fading behavior by the child, or in short visual attention span.
- The ability of children with CVI to navigate through cluttered environments without bumping into anything could be attributed to "blindsight", a brain stem visual system.
- Children are often able to see better when told what to look for ahead of time.
- Children with CVI may use their peripheral vision when presented with a visual stimulus, appearing as if they are looking away from the target.
- Some children look at an object momentarily and turn away as they reach for it.

Myths

The following statements are not true, according to current knowledge in the field:

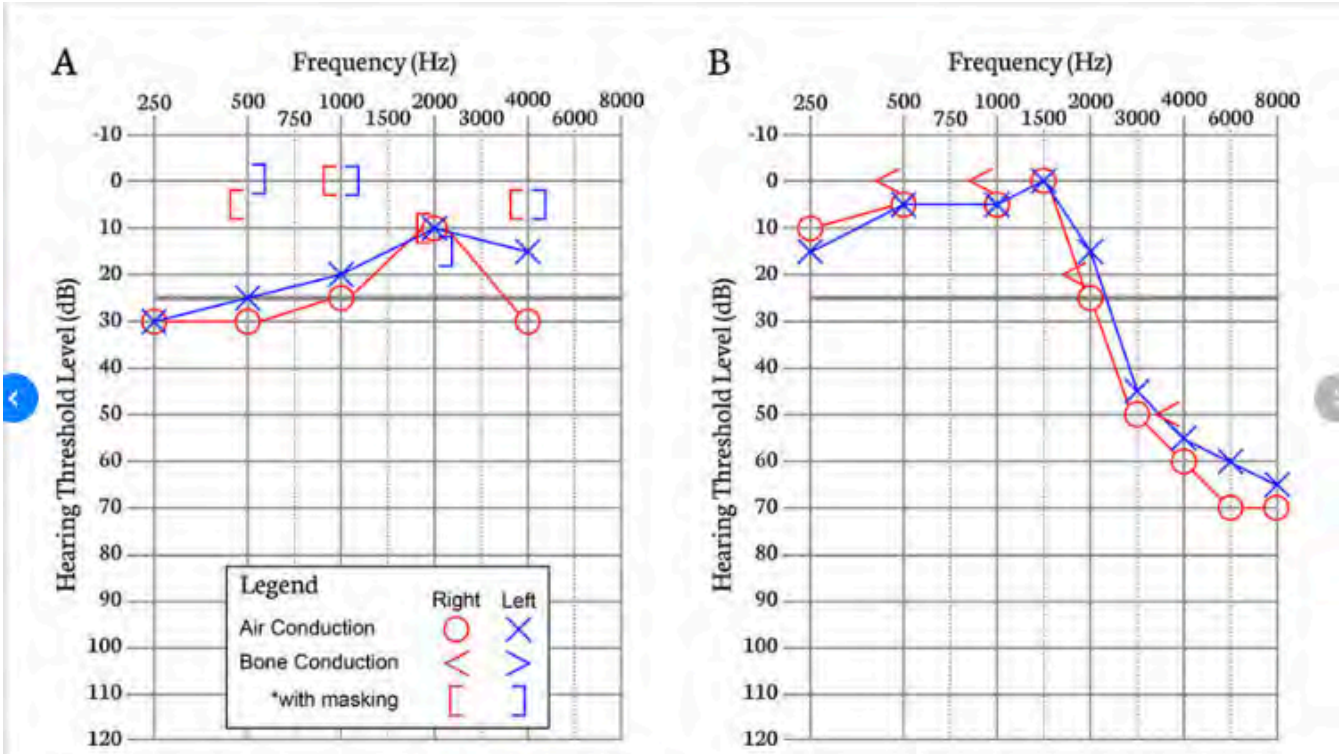
- Children with CVI are visually inattentive and poorly motivated.
- All children with CVI will have cognitive deficits.
- CVI is not a true visual impairment.
- Children with CVI are totally blind.
- Children whose visual cortex is damaged are Cortically Blind.

2. Hearing loss

I. Types of hearing loss
   a. Conductive
   b. Sensory
      i. Cochlear dysfunction
   c. Neural
      i. 8th nerve or brainstem pathway dysfunction
   d. Central processing disorder
   e. Mixed

a. Recognize the typical audiogram of a child with conductive or sensorineural hearing loss

- Conductive loss
- Sensorineural loss



(A) Audiogram illustrating a bilateral conductive hearing loss, which is diagnosed when air conduction thresholds are >10 dBHL worse than bone conduction thresholds and bone conduction thresholds fall within the normal hearing range but air conduction thresholds do not. In cases of conductive hearing losses, the etiology of the hearing loss lies in the outer or middle ear while the inner ear remains intact. *As bone conduction is perceived in both ears, masking noise is presented to the nontest ear to evaluate bone conduction only in the test ear. (B) Audiogram demonstrating a bilateral high-frequency sensorineural hearing loss. In the absence of a conductive impairment, the difference between air and bone conduction thresholds is <10 dBHL. This audiogram suggests damage to the inner ear in the presence of a normally functioning outer and middle ear.

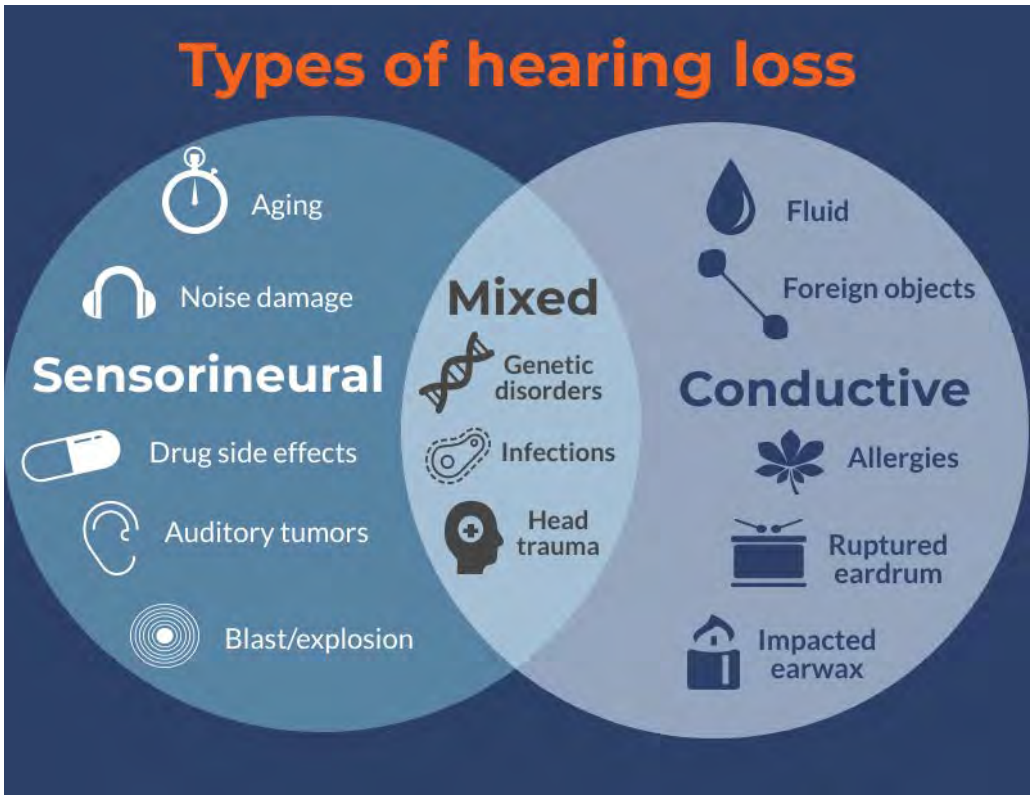


Table 1. **Causes of Hearing Loss**

Conductive Hearing Loss	Sensorineural Hearing Loss
Congenital • Microtia/atresia • Tympanic membrane abnormalities • Ossicular malformations	• Genetic disorders (syndromic, connexin 26, mitochondrial) • In utero infections (cytomegalovirus, measles, mumps, rubella, varicella, syphilis) • Anatomic abnormalities of the cochlea or temporal bone • Exposure to ototoxic drugs during pregnancy (alcohol, isotretinoin, cisplatinum) • Hyperbilirubinemia
Acquired • Infection (acute otitis media, otitis externa, ossicular erosion) • Otitis media with effusion • Foreign body (including cerumen) • Cholesteatoma • Trauma (ossicular disruption, tympanic membrane perforation)	• Infections (bacterial meningitis, measles, mumps, rubella, Lyme disease) • Trauma (physical or acoustic) • Radiation therapy for head and neck tumors • Neurodegenerative or demyelinating disorders (Alport, Cogan syndromes)

b. *Know the effectiveness of teaching sign language to children who have severe to profound hearing loss*

- If implemented at an early age and utilized as a "language" with proficient users (e.g. signing parents who are also deaf, it is as effective as any language
- Typically, hearing parents do not know sign language well enough to help their child learn it proficiently.
    i. Many children of hearing parents have very inadequate language of any type by the mid-school years

c. *Know how to plan the developmental assessment of a child with severe/profound hearing loss*

- Comprehensive assessment of cognitive, speech/language, social, and emotional ski11s at various developmental stages (infant, toddler, preschooler, school age, adolescence)
- Assessors require specialized skills

d. Know the impact of deafness on development, behavior, and academic achievement

- Deaf infants coo, begin to babble, but do not develop expressive babbling/early jargon. Without reinforcement, cooing and babbling drop out.
- Children whose hearing impairment is detected and have interventions before 6 months of-age, fare far better in all developmental, social and behavioral domains than children whose hearing impairment is detected after 6 months-of-age.
- Mean reading level of a deaf high school graduating senior is at the 4th grade level.
- Hearing impaired children have difficulty with attention, higher rates of behavior problems, especially externalizing behaviors.

e. Understand the efficacy of cochlear implantation for children with profound hearing loss

- Unilateral cochlear implantation is safe and effective for children and likely to be cost effective in profoundly and prelingually deaf children . Bilateral implants may provide additional benefit.
- A recent systematic review indicated all studies reported that unilateral cochlear implants improved scores on all outcome measures (Bond et al, 2009)
- Auditory, speech, language, and reading ski1Is achieved 4-6 years after cochlear implantation were most strongly associated with nonverbal IQ, implant functioning, and use of an oral communication mode

f. Understand the factors that affect decisions on use of cochlear implants

a. Criteria for use
    ii. Chi1dren 2 years and older with severe-to-profound deafness (i.e., pure tone average thresholds of 70 dB HL or greater)
    iii. Children 12 to 23 months of age with profound deafness (i.e., pure tone average thresholds of90 dBHL or greater.)
    iv. Whenever possible, outcomes from word and sentence recognition testing are also used to determine candidacy.
b. Disparities in use
    v. White and Asian children with severe to profound SNHL have higher proportionate rates of cochlear implantation than black and Hispanic children.
    vi. Implanted children are more likely to live in areas with higher median incomes.

g. Recognize the importance of early language exposure and instruction for children with hearing impairment

- Without appropriate intervention, children with mild hearing Joss function one to four grade levels below peers; children with severe-to-profound hearing loss function no higher than 3rd or 4th grade.
- The key is learning a language (e.g. signing, English, etc.) and starting early

h. *Know the three common educational/communication methods for children with hearing impairments: oral, manual, and total language and reasons for choosing each*

a. Oral: speaking/lip reading (combined with hearing aids or cochlear implants)
b. Manual: signing
c. Total communication: signing and speaking/lip reading

i. Know the leading causes of severe hearing impairment

1) Sensorineural
    i) Genetic (30-59% of all chi1dhood SNHL)
        (1) Mutations of connexin 26 gene
        (2) Syndromes: e.g. Pendred, Usher
        (3) X-linked: Alport syndrome
    ii) Acquired
        (1) Congenital infections (TORCH, meningitis)
        (2) Hyperbilirubinemia
        (3) Complications of prematurity
        (4) Ototoxic drugs
        (5) Head or acoustic trauma
    iii) Malformative
        (1) Malformations
        (2) Syndromes (e.g. Goldenhar, Treacher Collins)

2) Conductive
    i) Otitis media with effusion
    ii) Ossicular injury

j. Know the criteria for amplification with hearing aids

- Permanent, bilateral hearing loss with thresholds greater than 25 dB HL in 1000-4000 Hz frequency (speech) range (Minn. Department of Health guidelines)
- In unilateral hearing loss with measurable hearing in the affected ear, amplification may be beneficial.
- Middle ear pathology should be ruled out
- Amplification should be implemented by one month after detection

k. Understand the developmental and behavioral consequences of mild or moderate hearing loss

- Children with mild hearing loss miss 25-50% of speech in the classroom and may be inappropriately labeled as having a behavior problem.
- One third to one half of children with mild degrees of hearing loss have academic, social and behavioral difficulties

l. Understand the developmental and behavioral consequences of unilateral hearing loss

- School-aged children with unilateral hearing loss appear to have increased rates of grade failures, need for additional educational assistance, and perceived behavioral issues in the classroom
- Children with unilateral hearing loss (in one ear) are ten times as likely to be held back at least one grade compared to children with normal hearing.

m. *Understand the developmental-behavioral consequences of severe or profound hearing loss*

- At risk for significant learning. behavioral, and social problems
- One or more additional disabilities are common, such as cerebral palsy.

H. Other Chronic Conditions

1. Understand the reasons for inclusion of thyroid testing in the neonatal screening battery

Newborn screening (NS) for congenital hypothyroidism (CH) is one of the major achievements in preventive medicine. Most neonates born with CH have normal appearance and no detectable physical signs.

- Hypothyroidism in the newborn period is almost always overlooked, and delayed diagnosis leads to the most severe outcome of CH, mental retardation, emphasizing the importance of NS.
- Blood spot thyroid stimulating hormone (TSH) or thyroxine (T4) or both can be used for CH screening.
    - T4 is more sensitive but not cost-effective, so screening by TSH or T4 is used in different programs around the world

Patients who are not identified and treated promptly have mental retardation and variable degrees of growth failure, deafness, and neurologic abnormalities, as well as classic hypometabolic symptoms of hypothyroidism.

- Chromosomal: Has multiple causes, most of which are nongenetic.
- Incidence. One in 3600 to 1 in 5000 in the United States from screening; 1 in 3000 in Europe; 1 in 6600 to 1 in 7300 in Sweden by clinical diagnosis; and 1 in 5700 in Japan. Inheritance. Usually sporadic.
- Disorders of thyroid hormonogenesis may be inherited as autosomal recessive traits. Thyroid hormone transport defects may be X linked.
- Sex ratio: 3:1, female to male (New England states), 2:l female to male by clinical diagnosis.
- Racial and Ethnic Variability. Considerably less in African-American populations (1 in 17 000 in Georgia and 1 in 10 000 in Texas); more prevalent in Hispanic populations (1 in 2700) and Native Americans (1 in 700).

2. Differentiate between congenital and acquired hypothyroidism in terms of developmental outcomes usually identified by neonatal screening

Neonatal hypothyroidism

- Thyroid gland - function to synthesize T4 and T3
- Dietary intake of iodine required for this synthesis
- By 10-12 wks gestation. fetal hypothalamic-pituitary-thyroid system can concentrate Iodine and synthesis iodothyronine
- About 1/3 of maternal T4 crosses placenta
    - Low levels may affect fetal development, especially brain development

- Normal levels may partially protect hypothyroid fetus until birth

Congenital hypothyroidism

- Prevalence: 1 in 4,000infants
- Lower in African-Americans (1 in 20,000)
- Higher in Hispanics and Native Americans (1 In 2,000)
- 90% of Infants with hypothyroidism have developmental defects, usually thyroid dysgenesis
- Occurs twice as often in girls
- Most Infants with congenital hypothyroidism are asymptomatic at birth (due to transplacental passage of maternal T4)
- Early symptoms/signs
- Increased head size (due to myxedema of brain)
- Prolonged physiologic icterus
- Weak cry
- Excessive sleepiness
- Constipation
- Low temp. low heart rate
- Often cardiomegaly and/or asymptomatic pericardia! effusion
- Large abdomen often with umbilical hernia
- Feeding difficulties include, sluggishness, lack of interest. choking during nursing
- Respiratory difficulties - due to large tongue
- Cold. mottled skin
- Edema of genitals, hands and feet
- Anemia unresponsive to treatment

Later symptoms after 3 months of age

Physical Signs:

- Short extremities
- Wide open fontanels
- Eyes far apart**,** narrow palpebral fissures, swollen eye lids
- Depressed nasal bridge
- Open mouth with thick, broad tongue
- Short, thick neck
- Broad hands and short fingers
- Dry, scaly skin with little perspiration
- Myxedema - esp. of eyelids, backs of hands and genitalia
- Yellowish skin but white sclera
- Hair - minimal but coarse & brittle with hairline that extends down forehead
- Later symptoms - delayed mental development
- Lethargic

- Slow to sit and stand
- Hoarse voice
- Delayed language development
- Usually hypotonic (rarely have generalized muscular hypertrophy)
- Acquired hypothyroidism
- Most common cause, lymphocytic thyroiditis
- Usually in adolescents but can occur as early as age 2
- First clinical sx is deceleration of growth
- Other symptoms:
- Decreased energy
- Myxedematous changes in skin
- Constipation
- Cold Intolerance
- Increased need for sleep
- Delayed osseous maturation
- Does not produce cognitive changes or delays
- Symptoms reversible with treatment

3. Know the impact of type I diabetes on development and behavior

- Girls with recurrent ketoacidosis have more behavior problems (but also higher family conflict and lower family cohesion)
- McCarthy et al (2002)
  - Children with type I diabetes performed better than siblings in math and core scores and better than matched classmates in reading.
  - Poorer academic performance tended to occur in those with poorer control but same pattern In sibling scores so likely related to parenting factors and not disease factors
  - Significantly more school absences than sibs
  - More behavior problems than sibs- compliance, mood variability, and fatigue

Type I Diabetes

- In adults, having diabetes (both type I and type II) doubles the risk of co-morbid depression
- Depression associated with poorer metabolic control and poor adherence to medication and diet (which caused which?)
- In children. poorer metabolic control of type I diabetes associated with lower reading scores and lower grade point average - may be due to a common third variable (such as family factors or mood) rather than metabolic factors directly

Effects of hypoglycemia in children

- Question about possible persistent decrease In cognitive function from severe hypoglycemic episodes In young children

- Short term significant Impact on frontal and less so on temporal lobe and basal ganglia functions (including attention and memory)
- Early deficits mainly resolved over a period of 6-12 months, even when coma or seizures had occurred
- Some persistence of spatial long term memory deficits If severe hypoglycemic episodes began before age 5 yrs.

4. Understand the association of recurrent diabetic ketoacidosis and problems in family functioning

- Families with lower rates of parental warmth and higher rates of parental negativity are associated with higher occurrence of DKA episodes. Examination of these two variables alone correctly classifies a child's likelihood of having experienced a DKA episode or not 78% of the time.

- Specifically, changes of only one unit in these variables (as measured by diabetes specific measures) leads to a significant change in the odds that a child will fall into the group that has experienced a DKA episode.

- Targeting these family characteristics may improve compliance and metabolic control, and decrease the probability that the child will experience future diabetes-related complications, such as DKA.

Liss et al., 1998, Steinhausen, 1982; Stevenson et al., 1991; Weissberg Benchell et al., 1995; Wysocki et al., 1996

5. Understand the effects of excess corticosteroids on affect and behavior in children

Children and adolescents treated with oral, inhaled, and intravenous corticosteroids (CS) may experience adverse psychological side effects (APSE), including psychotic symptoms. These can occur at any point during treatment, including withdrawal.

Behaviors can range from psychotic symptoms to mild changes in mood and cognition
Rates of psychiatric disturbance (predominantly depression and anxiety) in children with Cushing syndrome show rates of 44%, with compulsive behaviors predominating.

https://adc.bmj.com/content/archdischild/90/5/500.full.pdf

6. Identify the developmental and behavioral consequences of iron deficiency at different ages

- Case-controlled study that examined social-emotional behavior found differences in iron-deficient anemic infants (e.g., more wary, hesitant, solemn, unhappy, kept closer to their mothers). Two of three randomized trials to prevent iron deficiency that assessed this domain reported social-emotional differences as well.

- Studies positively associated with overall developmental, motor, and social quotients at 2 years and with overall developmental quotient at 4 years agreeing in increased concerns about anxiety/depression, social problems, and attention problems.

- children with iron deficiency anemia or some other indication of chronic, severe iron deficiency in infancy had poorer performance on tests of some specific cognitive functions.

7. Know the factors that affect the developmental and behavioral outcome of children with congenital heart disease

- Vast majority have normal outcomes with IQ In average range and school performance as expected for cognitive ability
- Increased rate of neurodevelopmental problems in comparison to healthy peers
- Often mild problems with cognition, expressive language, attention, subtle neuro-motor function, visual-motor Integration, & executive function

- Factors affecting outcome
  - Key cardiac and CNS structures develop at same time prenatally - intrauterine factors may affect both
  - Role of chronic hypoxia (impairment In cognitive function increases with age for those with cyanotic heart disease)
  - Possible role for nutritional deficiencies related to feeding difficulties for babies with cyanotic heart disease
  - Deficits for children with cyanotic cardiac disease improve after surgery but not as much as hoped for
  - No change In cognitive function or academic achievement after surgery for children with acyanotic heart disease In most studies
  - Persistence in poor concentration and learning difficulties
  - Possible role of frequent absence from school or altered parental expectations
  - Complications of surgery including seizures, strokes, etc. may affect development and/or behavior long term as well

8. Know the nutritional deficiencies associated with developmental and behavioral problems in children

Vitamin	Food Sources	Symptoms caused by deficiency	Diseases related - usually by causing impaired absorption or metabolism	Overdose Symptoms
Vitamin A	Breast milk, cow's milk, vegetables, fruits, eggs, butter, liver	Ocuta rlesio ns Apath y Retardation of mental and physical growth	Malabsorption in celiac disease, pancreatic or liver diseases, iron deficiency anemia, chronic infectious diseases, chronic ingestion of mineral oil	Acute- Nausea, vomiting, drowsiness, cranial nerve palsies, diplopia, papillcdema Chronic - poor weight gain, irritability, tender swelling of bones, hepatospenomegaly, increase intracranial pressure In preg women - may lead to severe congenital rnalformanations
B1 - thiamine	Breast milk or cow's milk., vegetables, fruits, eggs, cereals	Fatigue, mental apathy, irritability, depression. drowsiness, poor concentration, nausea. Peripheral neuritis with tingling, burning, and paresthesias of toes and feet. Later heart failure, ptosis, atrophy of optic nerve, atrophv, ataxia, eventuallv coma	Leigh - pyruvic lactic acidosis Thiamine responsive anemia Maple Sugar Urine Disease	Generally nontoxic In severe excess can cause hypotension, cardiac arrhythmias, headache, anaphylaxis, weakness, convulsions
B2 - Riboflavin	Liver, kidney, brewer's yeast, milk, cheese, eggs, leafy vegetable	Glossitis, anemia. eye and skin changes	Faulty absorb with biliary atresia or hepatitis Impaired metabolism with pyruvate kinase deficiency or glutaric academia Il	Generally nontoxic Tums urine is orange

B3-Niacin	Liver, pork., salmon poultryred meat good sources (milk and eggs produce tryptophao which can substitute)	early symptoms - anorexia, lassitude, burning sensations, weakness, numbness later symptoms - dermatitis, diarrhea, and dementia (incl depression, disorientation, insomnia and delirium)	Children with parasites at high risk Hartnup	Flushing, wheezing, increased intracranial blood flow, headache Abn liver function with signs of liver toxicity

Vitamin	Food Sources	**Symptoms caused by** deficiency	Diseases related - usually by causing impaired absorption or metabolism	Overdose Symptoms
B6 Pyridoxine	Cows milk or cereal	In infants - irritability and convulsions {often hypsarrhythmic) In older children and adults - peripheral neuritis. dermatitis and anemia	Malabsorption with celiac disease Importantto add B6 if getting high protein diet Isoniazid treatment of tuberculosis increases requirements for vitamin as does pregnancy Cystathioninuria, Homocystinuria, Xanthurcnic aciduria. Oxaluria	Neuropathies - burning pains, parastesias, ataxia, clumsiness, paralysis, perioraJ numbness, impairment in position and vibration sense
Biotin	Deficiency is rare	Somnolence, hallucinations, hyperesthesia	Avitdin is a biotin agooist found in raw egg white) Propionic academia. Biotinase deficiency, holocarboxylasedeficiency	
Folic acid	Green vegetables, fruits, animal organs (kidney and liver) Also present in human and cow's milk	McgalobJastic anemia{incidence peaks at4-7 mo of age) Irritable, failure to gain weight. chronic diarrhea Folate deficiency in pregnant women associated with neural tube defects in **babies**	May result from malabsorption in celiac disease, chronic infectious enteritis and enteroenteric fistulas May result from treatment with anticonvulsants (phenytoin, primidone, Phenobarbital) Congenital folate malabsorption- autosomal recessive - leads lo severe	Generally non. toxic

			anemia, convulsions, MR	
Vitamin		Symptoms caused by deficiency	Diseases related - usually by causing impaired absorption or metabolism	Overdose Symptoms
Bl2	Derived from food in animal sources Humans cannot synthesize it Present in Jots of foods so dietary deficiency rare except withsevere vegan diets	Macrocytic anemia if from autosomal recessive disorder affecting absorption and processing- appears in infants likely to present around 9 mo to 11 yearsdepending on stores acquired from mother **Weakness,** anorexia. irritability, listlessness, Tongue which is smooth, red and painful Neurologic - ataxia. paresthesias, hyporcflexia. clonus, coma	May be caused by surgery involving stomach or terminal ileum, lack of secretion of intrinsic factor by stomach, abo of receptor sites io terminal ilium	

Vitamin	Food Sources	Symptoms caused by deficiency	Diseases related - usually by causing impaired absorption or **metabolism**	Overdose Symptoms
C-Ascorbic acid	Citrus fruits, tomatoes, breast milk, dark green vegetables	Scurvy hritability, Bleeding of gums, tenderness in legs, delayed wound healing, anemia	Note- vitamin C enhances absorption of iron	Renal colic, diarrhea, hemolysis if have G6PD deficiency, increased estrogen
D	Direct exposure to ultraviolet rays of sun, Commonly added to cow's milk and some orange	Rickets, anterior fontanel larger than normal and late in closing, large head, defects in teeth enamel and inc caries, bone changes, poor muscle development with poor tone Late symptoms- tetany, carpopedal spasm, laryngospasm, and seizures (7 2ndary to low serum calcium)	Impaired absorption with celiac disease, cystic fibrosis, or pancreatitis Anticonvulsants (phenatoins or phenobarbital) interfere with metabolism of vitamin D	Hypotonia, anorexia, irritability, constipation, hypercalcemia and hypercalciuria, Over time can lead to aortic valvular stenosis, hypertension, retinopathy, and

	juices			clouding of the cornea, renal damage and metastatic calcification
Vitamin	Food Sources	Symptoms caused by deficiency	Diseases related - usually by causing impaired absorption or metabolism	Overdose Symptoms
E		Anemia, increased platelet adhesiveness, role in retinopathy of prematurity Neurologic syndrome with cerebellar ataxia, peripheral neuropathy,· posterior column abn.	Worse in cystic fibrosis and acanthocytosis; Diets high in unsaturated fatty acid increase vit E requirement in premature babies who absorb vitamin E poorly Excess iron administration exaggerates signs of vitamin E dcficiencv	Headache, fatigue, easy bruising and bleeding, diplopia, muscle weakness, cretinuria In newborns - jaundice in newborns, hemolytic anemia
K	Soy beans, alfalfa, spinach, tomatoes, kale, cow's mild	Hypothrombinemia and decreased hepatic synthesis or pro convertin leading to bleeding especialy in newborns	Newborn status puts at risk Also results from decreased absorption or utilization of fat or to factors limiting its synthesis in the intestine (prolonged use of antibiotics or diarrhea in infants, particularly breast fed ones) Irradiation of food may produce vit K deficiency	

9. Understand the impact of prolonged caloric malnutrition (failure to thrive) on development and behavior

Undernutrition is one of the most important public health problems, affecting more than 900 million individuals around the World.

Marked changes in the function of the autonomic nervous system have been described in undernourished experimental animals.

Some of these effects seem to be epigenetic, passing on to the next generation.

Undernutrition in children has been linked to poor mental development and school achievement as well as behavioral abnormalities.

A much less clear picture is related to the effects of undernutrition and mental health. On one hand, difficulties in achieving reliable and valid measurements of mental development and behavior in poor children and allowing for the confounding and possibly interacting effects of social background have largely been addressed in the literature. On the other hand, there are very relevant studies showing that children in Peru who recovered from early stunting demonstrated normal levels of cognition in comparison to non-stunted counterparts.

Crookston BT, Penny ME, Alder SC, Dickerson TT, Merrill RM, Stanford JB, Porucznik CA, Dearden KA. Children who recover from early stunting and children who are not stunted demonstrate similar levels of cognition. J. Nutr. 2010;140:1996–2001

10. Identify the developmental and behavioral effects of lead exposure

- Lead exposure is one of the most common preventable poisonings of childhood. Data from the Center for Disease Control (CDC) show that 6% of all children ages 1-2 years and 11% of black (non-Hispanic) children ages 1-5 years have blood lead levels in the toxic range.

- Exposure to lead can have a wide range of effects on a child's development and behavior. Even when exposed to small amounts of lead levels, children may appear inattentive, hyperactive, and irritable. Children with greater lead levels may also have problems with learning and reading, delayed growth, and hearing loss. At high levels, lead can cause permanent brain damage and even death.

11. Understand the risks for lead exposure in children with developmental disabilities

Children with developmental disabilities are at higher risk for lead exposures because:

- They may have motor delays and spend more time on the ground
- They may have a higher sensory seeking need resulting in mouthing lead containing substances
- They may have more nutritional deficiencies leading to increase in pica

12. Understand the impact of iodine deficiency and maternal hypothyroidism on the fetus

Severe iodine deficiency during development results in maternal and fetal hypothyroidism and associated serious adverse health effects, including cretinism and growth retardation.

Dietary iodine is an intrinsic component of thyroid hormone, and adequate thyroid hormone is necessary for normal development.

Iodine supplementation of pregnant women is recommended in many regions where mild to moderate maternal iodine deficiency is prevalent

Although not better than iodine salt supplementation, in regions of moderate to severe iodine deficiency without effective salt iodization, lactating women who receive 1 dose of 400 mg I as oral iodized oil soon after delivery can provide adequate iodine to their infants through breast milk for ≥6 mo, enabling the infants to achieve euthyroidism.

It is not clear to what extent mild maternal iodine deficiency during pregnancy influences child neurobehavioral development (30). Although 2 observational studies have reported that mild

maternal iodine deficiency during pregnancy is associated with either lower child intelligence quotient (31) or educational assessment (32) scores, there is conflicting information from observational studies with regard to the effects of maternal iodine supplementation on child neurobehavioral development in areas of mild iodine deficiency.

Zimmermann MB. Nutrition: are mild maternal iodine deficiency and child IQ linked? *Nat Rev Endocrinol* 2013;9505–6.

Hynes KL, Otahal P, Hay I, Burgess JR. Mild iodine deficiency during pregnancy is associated with reduced educational outcomes in the offspring: 9-year follow-up of the gestational iodine cohort. J Clin Endocrinol Metab 2013;98:1954–62

13. Understand the emotional and family factors that affect symptoms and prognosis in children with chronic illnesses, such as asthma

- Direct medical effects of an illness as well as emotional & behavioral responses (Indirect or reactive) to the illness may last hours to days to months to years

- Physical illnesses may impinge on a child's health related quality of life:
    - disease state
    - functional status
    - psychological functioning
    - social functioning

- Physical Illnesses also impact on the family
    - Parents (or significant caretakers) play central roles in the day-to-day management of these illnesses and as a result, are not spared the stress of responding to & living with these health conditions.
    - Siblings, relatives, friends, teachers, & other significant people are affected to varying degrees experience the Impact of physical illnesses.

14. Know the factors affecting the developmental and behavioral outcome of children with hematologic disorders

- Sickle Cell Disease
    - Children with sickle cell disease (SCD) are at an elevated risk for neurocognitive deficits that impact quality of life, particularly in terms of school functioning.
    - The risks for neurocognitive deficits vary substantially by SCD genotype.
    - CVA is related to further increased risk of cognitive impairment
- Hemophilia
    - Overall males with hemophilia perform within age expectation on standardized cognitive testing
    - Adolescents/young adults appear to be at risk for difficulties with attention, executive function, and independence skills in their daily lives.
    - Increased risk of ADHD

15. Know the factors affecting the developmental and behavioral outcome of children with oncologic disorders

- Changes at the level of structural and functional neurobiology have been linked to increased risk of behavior problems and cognitive dysfunction
- The treatment modality matters; radiation vs chemo
- Learning problems usually show up within a few years of treatment.
- Lower IQ scores, which can vary depending on the intensity of the treatment
- Lower academic achievement test scores
- Problems with memory and attention
- Poor hand-eye coordination
- Behavior problems
- Non-verbal skills like math are more likely to be affected than language skills like reading or spelling, but nearly any area of brain development can be affected.
- Other late effects that may show up, depending on the type of treatment used, include things like seizures and frequent headaches.

16. Know the factors affecting the developmental and behavioral outcome of children with rheumatologic disorders

- Higher rate of internalizing behaviors vs the general population; predominantly anxiety and depression
- Higher rate of externalizing behaviors vs the general population (rule breaking and aggression)

17. Know the factors affecting the developmental and behavioral outcome of children with immunologic disorders

- HIV
    - Varying degree of cognitive impairment in HIV+ children
    - Children presenting with more severe HIV-1 disease and immune compromise have significantly more abnormal neurological signs and developmental delays than children presenting with milder HIV-1 symptomatology.
    - Early antiretroviral treatment results in lower risk neurodevelopmental impairment
    - Language impairment

18. Know the factors affecting the developmental and behavioral outcome of children with pulmonary disorders

- Studies performed in infants with bronchopulmonary dysplasia (BPD) revealed that they have more adverse motor function, worse cognitive development and poorer academic progress than infants without BPD.

Cheong JLY, Doyle LW. An update on pulmonary and neurodevelopmental outcomes of bronchopulmonary dysplasia. Semin Perinatol. 2018 Nov;42(7):478-484. doi: 10.1053/j.semperi.2018.09.013. Epub 2018 Oct 2. PMID: 30401478.

- The National Survey of Children's Health (NSCH) in 2003, revealed that children with asthma are at a higher risk of developmental, emotional, and behavioral problems. ADHD, depression, anxiety, learning problems, and behavioral problems was more common in asthmatics. It is speculated that it may be due to the malaise of chronic illness, acute exacerbations, missed school days, poor self-image, difficulty with school adjustments, and iatrogenic effects of medications, but no causal relationship has been identified.

Blackman JA, Gurka MJ. Developmental and behavioral comorbidities of asthma in children. J Dev Behav Pediatr. 2007 Apr;28(2):92-9. doi: 10.1097/01.DBP.0000267557.80834.e5. PMID: 17435459.

19. Know the factors affecting the developmental and behavioral outcome of children with renal disorders

- Chronic kidney disease in childhood is associated with neurocognitive deficits to include lower performance on cognitive tests compared to unaffected siblings, reduced attention and executive function

Mendley SR, Matheson MB, Shinnar S, et al. Duration of chronic kidney disease reduces attention and executive function in pediatric patients. *Kidney Int*. 2015;87(4):800-806. doi:10.1038/ki.2014.323

20. Know the factors affecting the developmental and behavioral outcome of children with gastrointestinal disorders

- Necrotizing enterocolitis is associated with increased risk for poor neurodevelopmental outcomes because it often occurs during a period of rapid and dynamic neurologic development when the brain is particularly vulnerable to insults and nutrient deficits. Pathogenesis is likely multi-factorial to include nutritional and non-nutritional factors.

Hickey M, Georgieff M, Ramel S. Neurodevelopmental outcomes following necrotizing enterocolitis. Semin Fetal Neonatal Med. 2018 Dec;23(6):426-432. doi: 10.1016/j.siny.2018.08.005. Epub 2018 Aug 17. PMID: 30145060.

- Inflammatory bowel disease has a strong association with various neurologic manifestations to include peripheral neuropathy, demyelinative disorders, and cerebrovascular disease, but also ADHD and tics.

Ben-Or O, Zelnik N, Shaoul R, Pacht A, Lerner A. The neurologic profile of children and adolescents with inflammatory bowel disease. J Child Neurol. 2015 Apr;30(5):551-7. doi: 10.1177/0883073814521296. Epub 2014 Apr 2. PMID: 24700662.

- Hypotonia and developmental delay are associated with infantile-onset celiac disease, which is likely due to nutritional deficits and toxic effects of severe malabsorption. ADHD and learning disorders are also associated with celiac disease.

Zelnik N, Pacht A, Obeid R, Lerner A. Range of neurologic disorders in patients with celiac disease. Pediatrics. 2004 Jun;113(6):1672-6. doi: 10.1542/peds.113.6.1672. PMID: 15173490.

21. Know the developmental risks to the fetus and young child of mercury toxicity

Acute or chronic mercury exposure can cause adverse effects during any period of development. Mercury is a highly toxic element; there is no known safe level of exposure. Ideally, neither children nor adults should have any mercury in their bodies because it provides no physiological benefit.

Mercury exposure can cause acute and chronic intoxication at low levels of exposure. Mercury is neuro-, nephro-, and immunotoxic.

Of special interest is the development of the central nervous system. With the formation of neuronal cells and the subsequent stages of development, the central nervous system is created.[55] Damage of the nervous system caused by mercury is likely to be permanent.[56,57] Neurotoxic effects can result from prenatal or early postnatal exposure.[58]

Prenatal mercury exposure and the neurological development of the children were demonstrated. Outcomes associated with prenatal mercury exposure included the loss of IQ points, and decreased performance of tests, including memory, attention, language, and spatial cognition. Prenatal mercury exposure was measured as mercury concentration in maternal hair, cord blood, or children's hair.

Neurodevelopmental effects in the fetus are associated with maternal exposure. Mercury can also cause neurocognitive deficits and neuromotor disabilities.

A cohort of 1022 children born 1986-1987 was exposed to methylmercury. The mothers episodically ate pilot whale meat, which is potentially high in methylmercury, and continuously ate fish with a comparably lower methylmercury concentration. At age of 7 and 14, neuropsychological tests were performed, showing neuropsychological dysfunctions mainly for language, attention, and memory, and less for visuospatial and motor functions. Neurophysiologic tests showed delayed brainstem auditory-evoked potentials,[57] decreased autonomic heart rate variability, both attributed to prenatal exposure.

Content Category 7-Congitive & Adaptive Diabilities

ABP Content Specs for Developmental-Behavioral Pediatrics Fellow Collaboration Project

Subsection A- Prepared by Meagan Butsch, DO, Madigan Army Medical Center (now staff)

Reviewed by Eric Flake, MD, Madigan Army Medical Center

Subsection B and C- Prepared by Melissa Harada, MD, UCLA Developmental Behavioral Pediatrics (now staff)

Reviewed by Irene Koolwijk, MD, UCLA DBP Fellowship Director

7. Cognitive/Adaptive Disabilities

A. Etiology and Evaluation

1. Understand current psychological theories of intelligence
2. Understand the levels of severity of intellectual disability
3. Know the diagnostic criteria for intellectual disability
4. Understand the interaction of environment and biology in the etiology of intellectual disability
5. Know the risk factors related to the causes of intellectual disability
6. Know the conditions that may affect the validity of assessments of intellectual ability
7. Know the differential diagnosis of intellectual disability
8. Know the common etiologies of intellectual disability
9. Plan the medical evaluation of a child with intellectual disability
10. Interpret the psycho-educational assessment of a child with intellectual disability
11. Know the co-morbid conditions associated with intellectual disability
12. Know the epidemiology of intellectual disability
13. Understand the concept of levels of support required by individuals with intellectual disability
14. Understand the special health concerns in adolescents with intellectual disability
15. Know the assessments of adaptive behavior

B. Intervention

1. Understand the specific behavioral, educational, and social challenges associated with each developmental stage for a child with intellectual disability
2. Know the educational interventions available for children with intellectual disability
3. Understand the effects of early intervention for children with or at risk for developmental delays
4. Understand the appropriate educational interventions and accommodations for a child with intellectual disability
5. Plan the treatment of common behavioral and emotional problems associated with intellectual disability
6. Understand the principles of planning for the transition to adulthood for youth with intellectual disability
7. Know the pharmacological interventions for behavioral disorders in children with intellectual disability

C. Outcome

1. Understand the academic potential of individuals with the different levels of severity of intellectual disability
2. Understand the occupational potential of individuals with the different levels of intellectual disability

3. Understand the independent living potential of individuals with the different levels of intellectual disability
4. Know the factors that mediate or moderate the outcome for individuals with intellectual disability
5. Understand the impact of different levels of intellectual disability on social relationships

7. Cognitive/Adaptive Disabilities

A. Etiology and Evaluation

1. Understand current psychological theories of intelligence

There are 4 major theory types of intelligence:

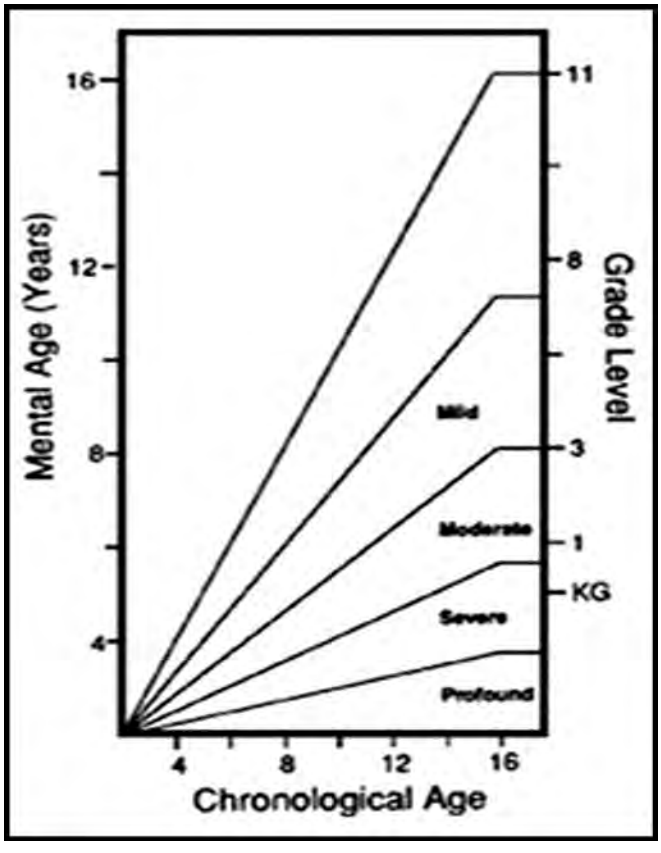
(1) psychometric theories - Psychometric theories derive from studying individual differences in test performance on cognitive tests. Questions about the structure of human intelligence, including the importance of general intelligence, have dominated the psychometric theories.

(2) cognitive theories - Cognitive theories derive from studying the processes involved in intelligent performance. These processes range from the very simple (e.g., inspection time) to the fairly complex (e.g., working memory). Different theorists have focused on different processes (or aspects of these processes, such as processing speed)

(3) cognitive-contextual theories - Cognitive-contextual theories emphasize processes that demonstrate intelligence within a particular context (such as a cultural environment). Major theories include Sternberg's triarchic theory, Gardner's theory of multiple intelligences, and Piaget's theory of development.

(4) biological theories - Biological theories emphasize the relationship between intelligence, and the brain and its functions. Numerous relationships have been found, but none have been elaborated into a detailed theory of the neuropsychology of intelligence.

2. Understand the levels of severity of intellectual disability



Profound ID

- Learns at less than ¼ usual rate
- No reading skills
- Often with other medical conditions
- Self-stim and Self Abusing behaviors

Mild ID

- Learns at ½ to ¾ normal rate
- 3rd-7th grade reading level
- Vocational/occupational education
- Live independently (24-34%)
- May marry, have children
- Competitive employment (work habits and social skills) (37-55%)

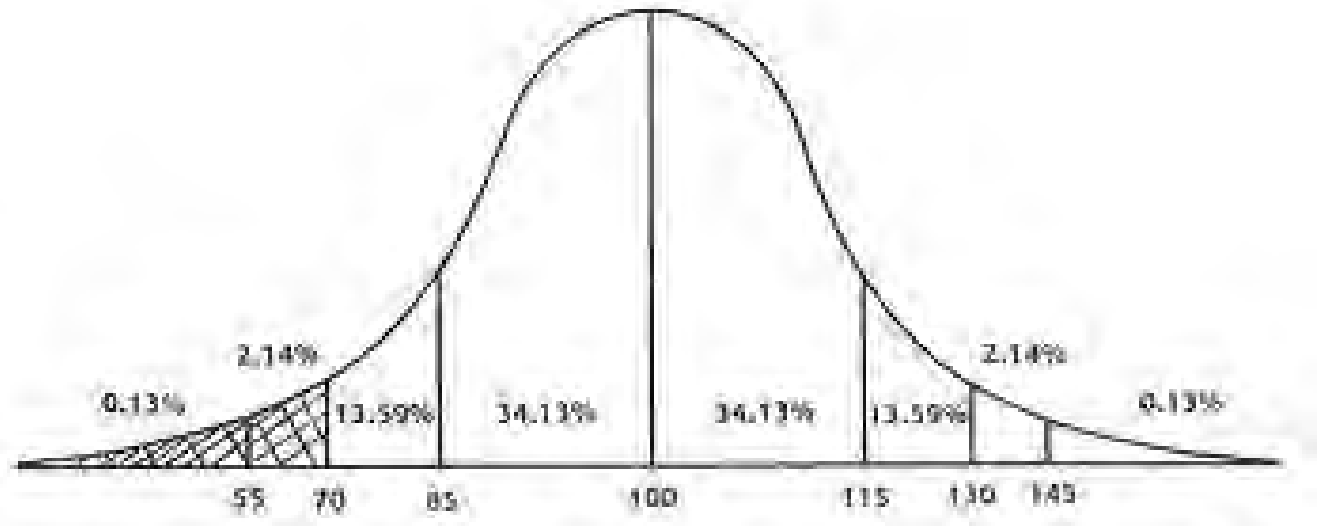
Moderate ID

- Learns at 1/3 to 1/2 to usual rate
- 1st-4th grade reading level
- Vocational/life skills education
- ADL's formally taught
- Supervised group home
- Rarely marry/parent
- Supportive/sheltered environment

3. Know the diagnostic criteria for intellectual disability

The DSM Criteria for ID are:

- The following 3 criteria must be met:
  - *A. Deficits in general mental abilities such as*
    - reasoning,
    - problem-solving,
    - planning,
    - abstract thinking,
    - judgment,
    - academic learning
    - learning from experience.
  - *B. Impairment in adaptive functioning for the individual's age and sociocultural background. The limitations result in the need for ongoing support at school, work, or independent life.*
  - *C. All symptoms must have an onset during the developmental period.*



Statistical prevalence 2-3%
Actual – varies according to study
M:F 1.4:1

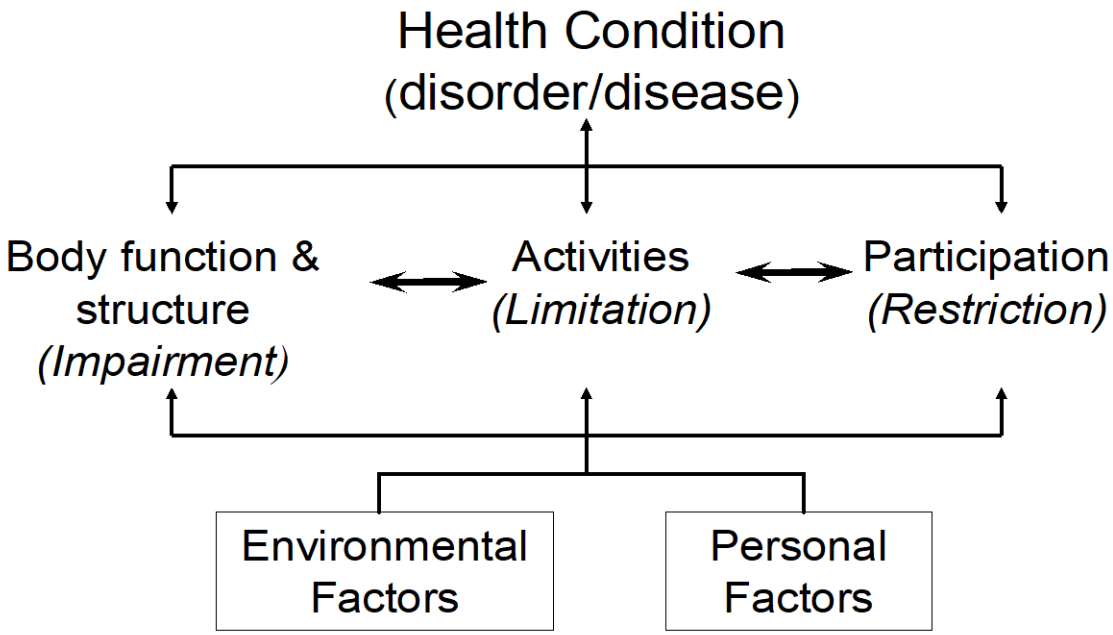
IQ	<1960's	1970-1980's	1980-2000's MR	AAIID (support) ID

55-69	Moron	Educable	Mild	Intermittent
40-54	Imbecile	Trainable	Moderate	Limited
25-39	Idiot	Custodial	Severe	Extensive
<24	Idiot		Profound	Pervasive

4. Understand the interaction of environment and biology in the etiology of intellectual disability

Environmental Influences on Intelligence: Intelligence is also strongly influenced by the environment. Factors related to a child's home environment and parenting, education and availability of learning resources, and nutrition, among others, all contribute to intelligence. A person's environment and genes influence each other, and it can be challenging to tease apart the effects of the environment from those of genetics.

International Classification of Function



Etiology:

Timing	Severe ID	Mild ID
Prenatal	70%	51%
Perinatal	4%	5%
Postnatal	5%	1%
Unknown	21%	43%

Prenatal Causes

- Infection
- Genetic
    - Trisomies
    - Klinefelter syndrome
    - Fragile X syndrome
    - Prader-Willi Syndrome
- Toxins and teratogens
- Congenital hypothyroidism
- Inborn Errors of Metabolism
- Brain malformation
- Hypoxia
- Complications of prematurity
- Intracranial hemorrhage
- Perinatal CNS infection

Post-Natal Causes

- Acquired brain injury
- CNS hemorrhage, infection or malignancy

- Severe environmental deprivation
- Severe malnutrition
- Toxins

- **Most common inherited disorder Fragile X – IQ (25-60)**
- **Most common chromosomal abnormality in live-born infants Trisomy 21 IQ (40-60)**
- **Most common teratogen - Fetal Alcohol Syndrome IQ 20-120 (mean of 65)**
    - **Social skills often peak at age 6yo level**
    - **Executive functioning skills**

5. *Know the risk factors related to the causes of intellectual disability*

- Infections (present at birth or occurring after birth
- Chromosomal abnormalities (such as Down syndrome)
- Environmental
- Metabolic (such as hyperbilirubinemia, or very high bilirubin levels in babies)
- Nutritional (such as malnutrition)
- Toxic (intrauterine exposure to alcohol, cocaine, amphetamines, and other drugs)
- Trauma (before and after birth)
- Unexplained (doctors do not know the reason for the person's intellectual disability)

6. *Know the conditions that may affect the validity of assessments of intellectual ability*

Tests of cognitive/intellectual functioning, commonly referred to as intelligence tests, are widely accepted and used in a variety of fields, including education and neuropsychology. Prominent examples include the Wechsler Adult Intelligence Scale, fourth edition (WAIS-IV; Wechsler, 2008) and the Wechsler Intelligence Scale for Children, fourth edition (WISC-IV; Wechsler, 2003).

Poor performance on a set of cognitive or neuropsychological measures does not always mean that an individual is truly impaired in that area of functioning. Additionally, poor performance on a set of cognitive or neuropsychological measures does not directly equate to functional disability.

In instances of inconsistent or unexpected profiles of performance, a thorough interpretation of the psychometric data requires use of additional information. The evaluator must consider the validity and reliability of the data acquired, such as whether or not there were errors in administration that rendered the data invalid, emotional or psychiatric factors that affected the individual's performance, or sufficient effort put forth by the individual on all measures.

To answer the latter question, administration of performance validity tests (PVTs) as part of the cognitive or neuropsychological evaluation battery can be helpful. Interpretation of PVT data must be undertaken carefully.

If an individual has not given his or her best effort in taking the test, the results will not provide an accurate picture of the person's neuropsychological or cognitive functioning. Performance validity indicators, which include PVTs, analysis of internal data consistency, and other corroborative

evidence, help the evaluator to interpret the validity of an individual's neuropsychological or cognitive test results.

7. *Know the differential diagnosis of intellectual disability*

Diagnostic Considerations
CNS trauma

Environmental deprivation

Major sensory deficits (eg, deafness, blindness)

Malnutrition

Mitochondrial cytopathies

Neurodegenerative disorders

Differential Diagnoses
- Autism Spectrum Disorder
- Borderline intellectual functioning
- Child Abuse & Neglect, Posttraumatic Stress Disorder
- Childhood Disintegrative Disorder
- Cognitive Deficits
- Learning Disorder, Reading
- Learning Disorder, Written Expression
- Mathematics Learning Disorder
- Pediatric Depression
- Pervasive Developmental Disorder
- Rett Syndrome
- Severe communication/language disorders

8. *Know the common etiologies of intellectual disability*

See # 5

9. *Plan the medical evaluation of a child with intellectual disability*

- History
    - Developmental
    - Family History
- Physical
    - Observation
    - Soft neurological
    - Major and minor dsymorphologies

- Chromosomal analysis
    - High resolution chromosomal analysis
    - Comparative Genomic Hybridization
- Fragile X
- Thyroid studies, lead, metabolic workup
- MRI?
    - Regression
    - Macro/microcephaly
    - Focal neuro/seizures
    - Skin

10. *Interpret the psycho-educational assessment of a child with intellectual disability*

Under the IDEA, special education and related services are provided for children from 3 to 21 years of age. All children with disabilities are mandated to receive a comprehensive education in the least restrictive environment, with priority given to the most severely impaired children. IDEA requires the states to identify and evaluate eligible children and generate an Individualized Education Plan (IEP) annually, with measurable and appropriate goals.

The legislation includes due process so that parents may participate actively in their child's education. Requirements for the participation of physicians and the extent of their participation vary among states. When it is not required by the state, parents may choose to obtain a physician's evaluation and assessment for developmental and other associated conditions and to identify medical needs and interventions. It is often helpful for the child and family to have a physician advocate who can act as an intermediary with state agencies.

Special education services offered by the school may include training in social skills, assistance in self-help, vocational training, behavioral assistance, and academic program modifications. Children may be placed in a special education class or provided resource classes or an aide. Principles of least restrictive environment and inclusion must be applied to enable a child with ID to participate in home or community settings among children who do not have a disability. Not all children can participate in regular academic classes, but, depending upon the extent of disability, they may be included in many other activities such as home room, physical education, lunch, recess, art, or music. Assistive technology options can reduce the effect of the disability and improve functioning in communication, daily living, mobility, communication, education, and vocational preparation.
Other educational settings are available for children who are unable to attend regular classes. These include a self-contained special education class with adaptive, self-help life skills training, a specialized day school, or, in rare cases, a residential institutional placement. The state is responsible for reimbursement, and the extent of the school's responsibility for related services, such as nursing, is controversial [40].

According to the IDEA Improvement Act of 2004, after 16 years of age, an Individualized Transition Plan must be included in the student's annual IEP. This includes vocational planning and provisions for including the patient in decision-making, when possible

10. Know the co-morbid conditions associated with intellectual disability

- Behavioral/Mental Illness 30-60%
- Seizures 10-40%
- Sensory Impairments 7-20%
- Cerebral Palsy 10-20%
- Sleep Disorders 80% in SMR
- Recurrent Emesis 15% in SMR
- Obesity 30-50%
- Autism 10-30%
- Etiologic specific medical complications

11. *Know the epidemiology of intellectual disability*

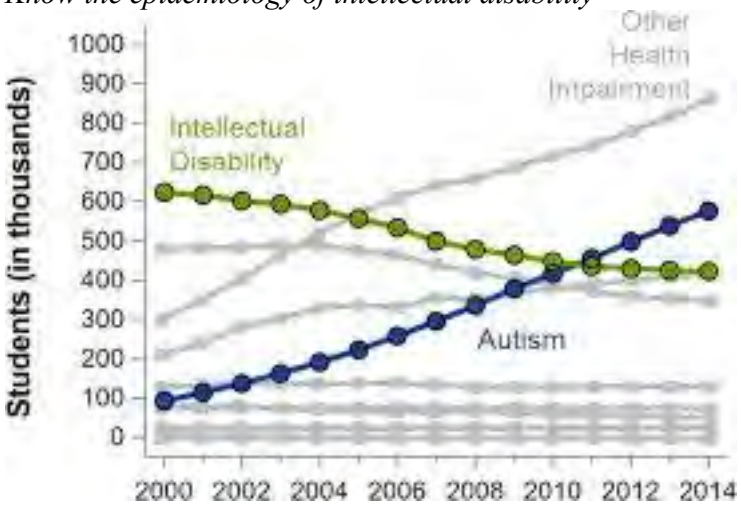
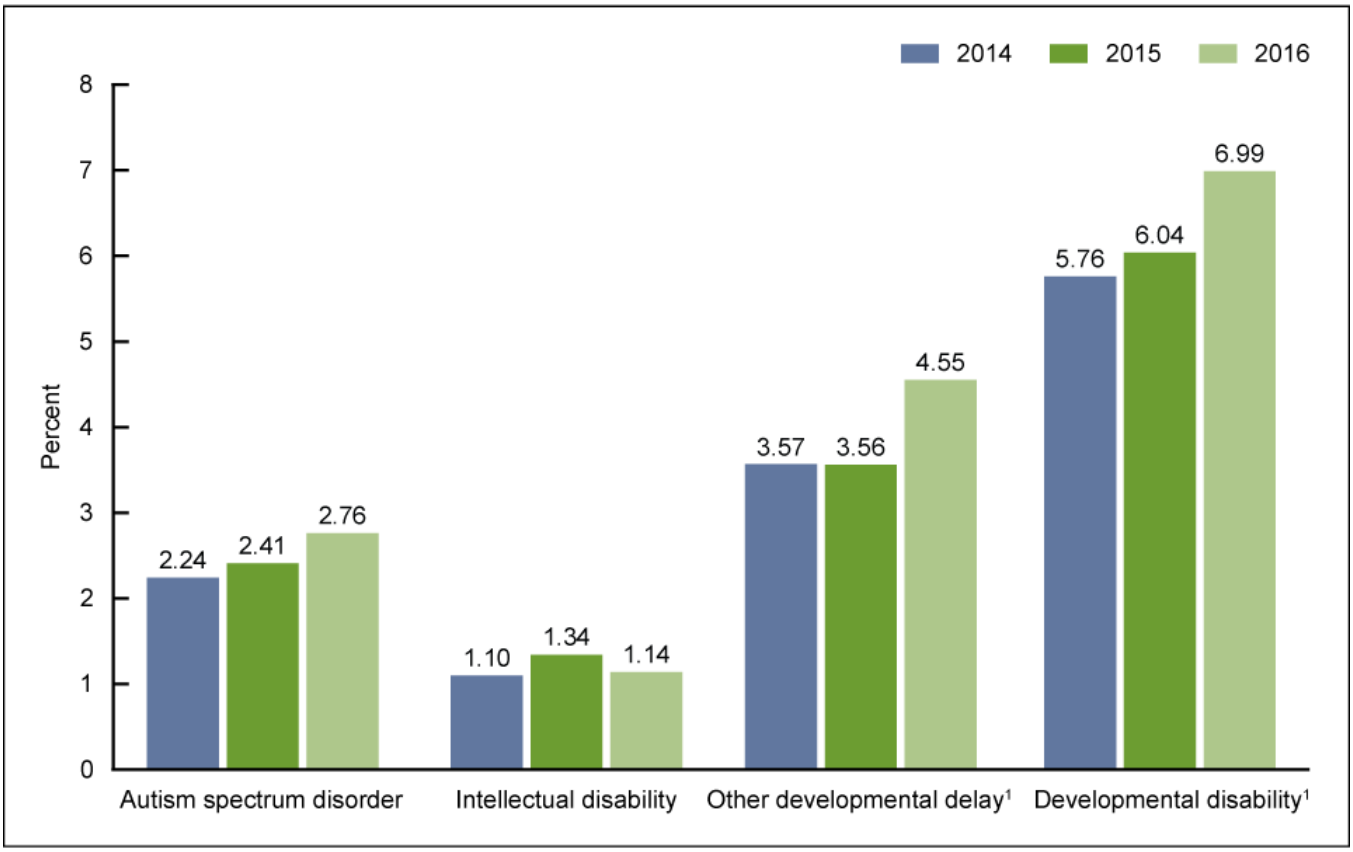


Figure 1. Prevalence of children aged 3–17 years ever diagnosed with selected developmental disabilites, by year: United States, 2014–2016



[1]Linear increase from 2014 to 2016 is statistically significant ($p < 0.05$).
NOTES: Developmental disability includes autism spectrum disorder, intellectual disability, and any other developmental delay. Access data table for Figure 1 at: https://www.cdc.gov/nchs/data/databriefs/db291_table.pdf#1.
SOURCE: NCHS, National Health Interview Survey, 2014–2016.

12. *Understand the concept of levels of support required by individuals with intellectual disability*
    - **Intermittent: PRN support**
    - **Limited: support on a constant but not permanent basis**
    - **Extensive: support constant, throughout life-span but not in ALL environments**
    - **Pervasive: support constant, throughout life-span and needed in ALL environments**

Dimension	Support	Activities	Time Span	Intensity
Intellectual Abilities				
OJT @DQ Money Concepts	Job Coach Trainer	Job Tasks Balance Checkbook	Time-Limited On-Going	Limited Limited
Adaptive Behavior				
Make Breakfast	House Parent	Prep / Clean	Time Limited	Limited
Health (Physical, Mental, Etiologic)				
Pat. dislocation Depression	Health Counseling Med Prompt	Orth. Surgery Clinic Visits Daily Meds	Time-Limited On-going	Extensive Extensive
Context				
Group Home	Houseparent	Homemaking	On Going	Limited
Social Roles, Participation, Interactions				
Interpersonal Relationships	Peer Befriending	Social Outings	On Going	Intermittent

- **4 levels of support Educational, Medical, Social, Political/Legal**

  Natural
  - family
  - neighbors
  - friends

  Informal –
  - support groups
  - respite
  - community agencies

  Formal Entitlements
  - public agencies
  - SSI
  - respite waiver programs

Educational - IDEA
- For children with disabilities ages 3-21yo
- Free and appropriate education (FAPE)

- "designed to meet each child's unique needs and prepare them for employment and independent living"

- Realistic goals-appropriate placement:
- Mild MR: formal education
- Severe MR: life skills classes
- LRE (Least Restrictive Environment)
- Inclusion vs. Segregation
- Behavior management:
- FAB: Functional Analysis of Behavior
- BIP: Behavior Intervention Plan
- Related Services (RN, PT, OT, ST, IHP)

Medical - See above #9

Social - Social support has been identified as a major protective factor in preventing mental health problems and also as a major contributor to quality of life. People with intellectual disabilities (ID) have been identified as having limited social support structures. Interventions have been focused on promoting their social presence and integration. However, previous studies have shown that this does not always lead to the formation of social relationships. To date few studies have looked at how having an ID leads to impoverished social networks.

Political / Legal -

- Social Security Administration –

a) The child must have a full scale IQ score of 70 or below, or a full scale IQ score of 71-75 with a verbal or performance score of 70 or below, and
b) The child must have an extreme limitation in one of the following areas, or a severe limitation in two of the following areas:
    - understanding, remembering, or applying information (ability to learn term and concepts, follow instructions, solve problems
    - interacting with others (ability to understand social cues, cooperate, make and maintain friendships, handle conflicts)
    - concentrating on tasks and maintaining pace (ability to complete tasks in a timely manner, ignore or avoid distractions, work close to others without distracting them), and
    - managing oneself (ability to protect self from harm, regulate emotions, control behavior, maintain personal hygiene).

14. Understand the special health concerns in adolescents with intellectual disability

Transition planning to facilitate the transition from childhood to adulthood, including adult health services, should begin at or by age 12 years in youth with ID [34,35]. Early individualized discussions may address issues of vocation, independent living/functioning, independent decision-making or guardianship, care coordination, sexuality, reproduction, life expectancy, health insurance, eligibility for adult community-based services, medical comanagement by different specialists, advance directives, and communication preferences of the child or youth with ID. The child or youth with ID should be involved as much as possible in the decision-making process, and his or her level of understanding should be assessed and documented. Youth with ID and autism spectrum disorder

are more likely to have identified needs for support after school and less likely to take an active leadership role, as compared with youth with other disabilities [36].

A transition plan should be documented by age 14 years and reviewed annually thereafter [34]. A transition plan may include the assessment of transition readiness, realistic goal setting, focused interventions, and anticipated timelines; the transition plan is used to implement and monitor the transition process until 18 years of age or the time of actual transition. Transition-related resources for professionals include educational guide books, evaluation tools, and checklists.

Transition includes assistance with adjustment from school to work or other adult activity. According to the Individuals with Disabilities Education Act (IDEA), the Individualized Education Plan (IEP) for students must include a transition plan by 16 years of age. The student should be involved in the development of the plan and student advocacy and self-determination should be encouraged. When the youth actually transitions to the adult service provider, a health record and medications summary with an individualized collaborative care plan should accompany him or her. Environmental factors such as the family system, provision of post-school services, and access to transport often affect transition outcomes

15. Know the assessments of adaptive behavior

Adaptive Functioning

Adaptive functioning is affected by three basic skill sets:

Conceptual

This includes reading, numbers, money, time, and communication skills.

Social
These skills help us to get along well with others. These skills include understanding and following social rules and customs; obeying laws, and detecting the motivations of others in order to avoid victimization and deception.

Practical Life Skills

These are the skills needed to perform the activities of daily living. This includes feeding, bathing, dressing, occupational skills, and navigational skills.

The assessment of adaptive behavior became a formal part of the diagnostic nomenclature for mental retardation with the publication of the 1959 manual of the American Association of Mental Deficiency (Heber, 1959, distributed in 1961). The 1961 manual (Heber, 1961) discussed adaptive behavior with respect to maturation, learning, and social adjustment. This framework, reiterated in 1983, described adaptive behavior limitations consisting of “significant limitations in an individual’s

effectiveness in meeting the standards of maturation, learning, personal independence, or social maturity that are expected for his or her age level and cultural group, as determined by clinical assessment and, usually, standardized scales" (Grossman, 1983, p. 11).

The 1983 manual characterized the tasks or activities encompassed by adaptive behavior (and, plausibly social competence) as:

In infancy and early childhood: sensorimotor development, communication skills, self-help skills, socialization, and interaction with others;
In childhood and early adolescence: application of basic academic skills in daily life activities, application of appropriate reasoning and judgment in mastery of the environment, and social skills—participation in group activities and interpersonal relations; and

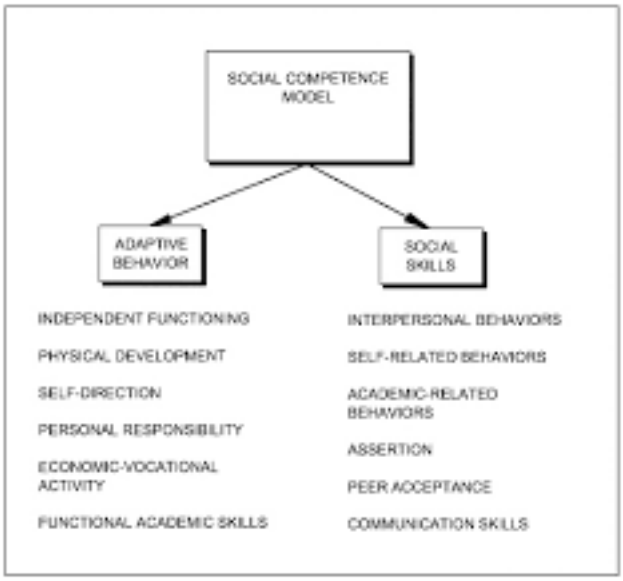
In adolescence and adult life: vocational and social responsibilities.

During the 1960s, a wider variety of adaptive behavior measures was developed and disseminated (e.g., Allen et al., 1970; Balthazar & English, 1969; Leland et al., 1967). Indeed, by the late 1970s, the number of available adaptive behavior measures, largely interview or observational in format, had burgeoned, including checklists pertaining to vocational behaviors (Walls & Werner, 1977). Measures developed in the 1960s have typically been updated in subsequent editions with enhanced psychometric characteristics and scoring (e.g., Sparrow & Cicchetti, 1985).

Over the past 25 years there has also been further refinement of the parameters and structure of tests of adaptive behavior and social competence. This refinement was based on large samples of research participants and data from service registries (McGrew & Bruininks, 1990; Siperstein & Leffert, 1997; Widaman et al., 1987, 1993). Novel frameworks for conceptualization of adaptive behavior have been proposed (American Association on Mental Retardation, 1992), and conventional frameworks have been endorsed for application in differential diagnosis and classification practices (Jacobson & Mulick, 1996). Finally, the difficulties and complexities of differentiating mild mental retardation from its absence or from other disabling conditions (e.g., Gresham et al., 1995; MacMillan, Gresham, et al., 1996; MacMillan, Siperstein, & Gresham, 1996) have remained an enduring concern in both professional practice and policy formulation.

Most Common Adaptive Tests

- Vineland Adaptive Behavior Scale (VABS)
- Adaptive Behavior Scales (ABS)
- Woodcock-Johnson Scales of Independent Behavior



B. Intervention

1. Understand the specific behavioral, educational, and social challenges associated with each developmental stage for a child with intellectual disability (ID)

Mild ID:

- Difficulties may manifest in late preschool or early school-age
- Anticipated to gain skills at one-half to two-thirds the rate of children without ID
- Difficulties in academics with early reading, writing, arithmetic, time, money, grasping complex information
- Socially immature compared to other children
- Communication and thinking more concrete and less mature
- Planning, problem solving and money management need long term support
- Greater gullibility, less mature judgement, vulnerability to manipulation

Moderate ID:

- Present earlier than those with mild ID, manifesting with: learning and language difficulties in the preschool years and social and communication challenges, which require limited although possibly substantial support
- Usually develop at approximately one-third to one-half the rate of children without ID
- Developmental gains are generally slow, conceptual skills lag well behind peers, substantial help or support is needed for conceptual tasks

Severe ID:

- Gain skills at one-quarter to one-third the rate of children without ID and one-half the pace of children with mild ID
- Academics: little understanding of written language or numbers, time or money concepts
- Caregivers provide extensive help in problem solving
- Spoken language is very limited and they may have limited understanding of speech-language and gestural communication

Profound ID:

- Conceptual skills often do not go beyond the concrete
- Abilities involve manipulation of objects
- Limited understanding of symbolic language, but may be able to understand basic instructions
- May express nonverbally and understand a simple instruction, gesture or emotional cue
- Concurrent sensory and physical limitations are common, and coexisting medical conditions may need additional equipment

2. Know the educational interventions available for children with intellectual disability (ID)

- Individuals with Disabilities Education Act (IDEA)
    - Under IDEA, children with ID qualify for special education and related services
    - Right to free and appropriate public education (FAPE)
    - Educational conditions should adhere to least restrictive environment (LRE) = whenever possible, children with disabilities will receive education in the regular classroom and pull out services or a self-contained classroom are provided only when that is impossible
    - Organizations:
        - Early Intervention (0-3 years)
        - Committee on Preschool Special Education (3-5 years)
        - Committee on Special Education (5-21 years)
    - Goals and services specified in:
        - Individualized Family Service Plan (IFSP) for children <3 years old
        - Individualized Education Program (IEP) for 3-21 years
        - Should reflect appropriate goals - educational, vocational and/or adaptive and take into consideration strengths and weaknesses
    - Transition process should begin at 16 years of age
        - Individualized Transition Plan
        - Continued support beyond the educational realm: employment, adult living skills and recreation
- Section 504 of the Rehabilitation Act
    - Federal law that protects individuals with disabilities from discrimination in various settings, including public school system
    - Children with ID may receive classroom modifications or accommodations
    - Accommodations = how a child accesses information or demonstrates mastery of a learning objective is adapted
    - Modifications = the learning material provided is modified for the child
- See Content Specification #4 below for additional details

3. Understand the effects of early intervention (EI) for children with or at risk for developmental delays

- Children who are suspected of having ID/GDD or other neurodevelopmental disorders should be referred to the state early intervention program (for children 0–3 years old)
- <u>What are long term effects of EI?</u>
    - Beneficial effects on cognition, language, academics (reading and math) and youth behavior
    - May minimize decline of intellectual functioning
    - Helps to maximize a child's functional potential
    - Prevents secondary social, emotional or behavioral problems at later stages of development
    - Provides support to families in caring for their children at home

- EI has been shown to be quantitatively most successful in:
    - Children with mild versus severe delays
    - Children at environmental risk (ie: low socioeconomic status, barriers to developmental stimulation at home or child care, toxic stress) versus neurobiological risk (ie: genetic syndromes, structural brain anomalies) for developmental delay
    - Children at risk for developmental disability versus children with known disability
- What type of EI works best?
    - Intervention that is well planned
    - EI that is given as early as possible can improve the functional outcome and quality of life for all children with developmental delays or those at risk
    - EI programs that focus on the family unit are more effective than those that just focus on the child
- Meta-analysis of 31 studies of EI for children with disabilities <3 years of age:
    - Children who are "developmentally delayed" make greater cognitive gains than those with more severe cognitive disabilities
    - Children with motor disabilities made the least gains
    - Well defined EI curricula have greater effects on child development as opposed to those with less structure
    - High level of parent involvement and interventions that pair child and parent are most effective
- EI for children with severe developmental disabilities:
    - Fosters more comfortable and developmentally appropriate parent-child interactions (preventing secondary social, emotional or behavioral impairments)
    - Enhances the ability of parents to care for children at home
    - Improves functional outcomes
    - Can be a critical component of long-term acceptance
- National Early Intervention Longitudinal Study (NEILS):
    - Families who participate in EI reported positive family outcomes, felt competent in roles as parents and in collaborative roles with professionals
    - Challenges: parents feeling less competent in handing their child's behavior versus other basic needs, less likely to participate in community activities

4. Understand the appropriate educational interventions and accommodations for a child with intellectual disability

- See Content Specification #2 above for additional details
- Classroom options:
    - Inclusion or collaborative team teaching classroom – children with special needs participate in the same classroom as typically developing children with 1 main teacher and 1 or more special education teachers
    - For children with more severe impairment (moderate to profound ID) a self-contained classroom may be needed with small student to teacher ratio and provision of individual or group paraprofessionals
- Related Services:
    - Speech-language therapy
        - Enhancing communication with a picture exchange communication system or augmentative communication device may be helpful
    - Occupational therapy

    - Physical therapy
    - Counseling
- Additional Considerations:
    - Compared with typically developing children, children with ID learn at a slower pace and may require more frequent repetitions before mastering a skill
    - The gap between typically developing children and children with ID may become increasingly wider as the years go by

5. Plan the treatment of common behavioral and emotional problems associated with intellectual disability

- 30% of individuals with ID may have comorbid mental health conditions
- May include: ADHD, depression, mood disorders, aggressive behaviors and self-injurious behavior
- Increased behavioral and emotional problems in children with ID may not meet standard diagnostic criteria
- Behavior interventions may be aimed at addressing difficult behaviors or specific comorbid conditions, and should be implemented in home or school with carryover to other settings
- Interventions can improve behavior-related functioning, performance and participation in everyday activities
- Self Injurious Behavior: may present in stress, depression, anxiety or as a medication side effect
- Repetitive or Self-Stimulating Behaviors: more common in severely impaired children with ID
- Suicidial Ideation and Substance Abuse: some youth with ID are at increased risk compared to typically developing peers
- Depression: important to identify because it can persisit and impact functioning. Occurs more frequently in ID due to Down Syndrome than in ID due to other causes
- Interventions:
    - Family counseling
    - Education
    - Behavior interventions for specific disorders (ie: ADHD, ASD)
    - Psychopharmacologic therapy
    - Interventions should be appropriate to the child's developmental level of functioning and diagnosis
- Behavior interventions
    - Children may benefit from interventions to improve socialization skills and behavior
    - For interventions, determine:
        - Antecedent triggers for behavior
        - Function of a problem behavior
        - Consequences of behavior/reactions of others to the behavior
    - Positively reinforce desirable behavior
    - Ignore misbheaviors (provided behaviors are not dangerous)
    - Applied behavioral analysis may be helpful

6. Understand the principles of planning for the transition to adulthood for youth with intellectual disability

- Transition planning should begin by 12 years of age
- May involve: vocation, independent living/functioning, independent decision-making, care coordination, sexuality, reproduction, life expectancy, health insurance, eligibility for adult

community based services, medical management by different specialists, advance directives, communication preferences

- A transition plan focuses on:
    - Assessment of transition readiness
    - Setting realistic goals
    - Planning anticipated timelines
    - Interventions needed to attain successful transition
- Intervention supports may include: a job coach, assistance with transportation, legal advocacy, supported employment, social security disability benefits and income, assisted living
- Transition and post-transition functioning may be impacted by factors such as:
    - Availability and access to services
    - Opportunities for housing and employment
    - Access to transportation
    - Individual's family system
- When planning transition, common concerns are issues related to:
    - Self-determination
    - Supported decision-making
- Guardianship: legal process to appoint a guardian for an individual deemed to have incapacitated decision-making capacity
    - All persons are deemed capable before the law until it can be shown in court that the person requires some supervision
    - Assess competency to make medical decisions
    - Other skill areas reviewed:
        - General supervision and safety
        - Finding housing
        - Employment
        - Education
        - Buying and selling property
        - Making contracts
        - Controlling one's finances
    - Alternatives to full guardianship (eg, limited guardianship, representative payee, power of attorney) should be considered to guarantee that the individual retain as much autonomy as they are able
    - Need to plan for guardianship determination by 18 years of age
        - If not determined by 18 years of age, parents may be blocked from participating in their child's care until a court determination of guardianship
- Maintain respect for personal autonomy
    - Important that health care professionals presume that their young adult patients possess decision-making capacity
    - Parents must balance goals of wanting to empower their children to achieve maximal independence while promoting their children's safety

7. Know the pharmacological interventions for behavioral disorders in children with intellectual disability

- Psychopharmacologic therapy is more likely to be used in children with a comorbid neurodevelopmental disorder or whose ID is severe
- Medication can improve:

    - Mental health
    - Participation
    - Functioning
    - Reduce burden of care
- Medications are generally started only after initial adequate appropriate behavior intervention, to minimize risk and unnecessary use of medication and may be reserved for individuals who:
    - Pose a risk of injury to self or others
    - Have severe impulsivity
    - Are at risk for losing access to an important service (ie: foster home, school, residential placement)
    - And if other treatments have failed
- The use of medications should be part of a clear, comprehensive psychosocial treatment plan that includes:
    - Behavioral baseline functioning
    - Identified target behaviors and goals
    - Evidence-based intervention appropriate to the diagnosis and target behaviors
- Prior to initiating pharmacotherapy:
    - The clinician and caregiver should agree upon measurable goals and target behaviors, obtain baseline assessments of target symptoms
    - Patients with irritability, aggression, self-injury, or disruptive behavior should be evaluated for other possible causes (including medical or psychiatric disorders or abuse/environmental factors)
- Weigh potential adverse effects against possible benefits
    - Medication for behavior can have negative effects on attention, concentration, learning and quality of life
    - Medications can cause temporary or persistent drug-related movement disorders
- Children with ID may be more sensitive to side effects
    - Start low, go slow: Use the safest, most effective treatment at the lowest possible dose and slowly increase
    - Regime should be simple
    - Use the fewest possible doses
    - Aim for monotherapy when possible
    - Use frequent and close monitoring
- Idiosyncratic medication responses may happen
    - Patients with ID may have increased sensitivity to disinhibiting effects of sedative-hypnotics
    - Children with Down Syndrome may be especially sensitive to anticholinergic medication
- There are no medications that are FDA approved specifically for use in individuals with ID
- Behavioral/emotional problems in children with ID may not meet standard diagnostic criteria
    - If formal psychiatric diagnosis is uncertain or not possible, consider targeting symptom clusters (see Table 1 below)

TABLE 1 Targeting medication to symptom clusters

Symptom cluster	Probable diagnostic categories	Order of applicable drug class	Examples
Aggression, self-harm, explosive temper	Conduct disorder, intermittent explosive disorder	1. Atypical antipsychotic 2. Anti-epileptic mood stabiliser 3. α-adrenergic agonist	Risperidone, olanzapine, aripiprazole Carbamazepine, valproic acid Clonidine
Hyperactivity, impulsivity, inattention	Attention-deficit hyperactivity disorder	1. Stimulant 2. Noradrenaline reuptake inhibitor 3. Mood stabiliser 4. α-adrenergic agonist 5. Atypical antipsychotic	Methylphenidate, dexamfetamine Atomoxetine Carbamazepine Clonidine Risperidone
Severe anxiety	Generalised anxiety disorder, panic disorder, specific phobias	1. SSRI 2. Benzodiazepine	Fluoxetine, sertraline Lorazepam
Depressive phenotype (withdrawal, irritability, sadness, reduced energy, sleep disturbance, change in appetite, loss of usual interests or preoccupations)	Major depressive disorder	SSRI	Fluoxetine, sertraline
Insomnia	Sleep-onset association disorder	1. Sedative-hypnotic 2. α-adrenergic agonist	Melatonin, 'z-drugs' (zolpidem, zopiclone, zaleplon) Clonidine
Handicapping obsessions and compulsions, extremely rigid or ritualistic behaviour	Obsessive–compulsive disorder Autistic stereotypy	1. SSRI 2. Atypical antipsychotic	Fluoxetine, sertraline Risperidone, olanzapine, aripiprazole
Bipolar phenotype (behavioural cycling associated with rages, aggressiveness, irritability, self-injury and euphoria; extreme hyperactivity or restlessness, diminished need for food or sleep, over-talkative)	Bipolar disorder	1. Mood stabiliser 2. Atypical antipsychotic 3. Lithium salts	Carbamazepine, valproic acid Risperidone, olanzapine, aripiprazole Lithium carbonate

SSRI, selective serotonin reuptake inhibitor.

Source: Bramble, D. Psychopharmacology in children with intellectual disability. *Advances in Psychiatric Treatment.* 2011;17(1):32-40.

C. Outcome

1. Understand the <u>academic</u> potential of individuals with the different levels of severity of intellectual disability

<u>Mild ID</u>:

- 6th grade level
- Most need sustained academic supports to learn and acquire age-typical conceptual skills (like reading)
- Difficulties with early reading, writing, arithmetic, time, money and grasping complex information

<u>Moderate ID:</u>

- 2nd grade level
- Substantial support needed for conceptual tasks
- Academic skills generally at the elementary level

<u>Severe ID</u>:

- Preschool/Pre-K level
- May learn to speak, master sight words (ie: "stop" or "exit")
- Little understanding of written language or numbers, time or money concepts

<u>Profound ID</u>:

- Individuals with profound ID do not learn to read
- Conceptual skills often do not go beyond the concrete

See Table 1 under Content Specification #2 below

2. Understand the occupational potential of individuals with the different levels of intellectual disability

Mild ID:
- Vocational skills with intermittent support
    - Some may attain vocational positions with straightforward occupational skills or paid employment in a competitive workplace with appropriate support
- Function appropriately in personal care

Moderate ID:
- Limited support
- Work in supervised settings
    - Can be trained to do unskilled or semiskilled work
    - May be employed in jobs that require minimal communication, conceptual demands and cognitive skills
    - Most need sheltered, supported, noncompetitive vocations
- Perform basic personal care (dress, toilet, eat independently)
    - May need significant amounts of support time and teaching
    - Can participate in all household tasks with ongoing support and teaching
- May travel independently in familiar settings

Severe ID:
- Require extensive supports
- Adults without prohibitive coexisting conditions may be able to participate in sheltered vocational setting
- May learn some self-care skills
    - Require training in social and self-help daily living skills in high school
    - Can learn some basic daily living activities, while requiring significant ongoing support and supervision

Profound ID:
- Pervasive supports and supervision
- May learn some simple self-help skills (ie: feeding, dressing), but usually dependent in all aspects of personal care and daily living
    - Some benefit from training in self-help skills, though gains are slow
    - May use objects in goal-directed fashion in self-care or simple recreation
- Outcomes are variable, and can vary from total dependence to some simple expression and social responsiveness

Table 1.

Levels of Severity of Intellectual Disability (ID)

LEVEL OF ID (% CHILDREN WITH ID)	LEVEL OF SUPPORT (IN CONCEPTUAL, SOCIAL, PRACTICAL DOMAINS)	ASSOCIATED ESTIMATED IQ SCORE	PROJECTED ULTIMATE ACADEMIC ACHIEVEMENT
Mild (85%)	Intermittent	55–70	Up to sixth-grade level
Moderate (10%)	Limited	40–55	Up to second-grade level
Severe (3%–4%)	Extensive	25–40	Preschool level
Profound (1%–2%)	Pervasive	<25	--

Note: The level of severity is based on the level of adaptive functioning and support. (1)(6)

Source: Purugganan O. Intellectual Disabilities. *Pediatrics in Review.* 2018;39(6):299-309.

3. Understand the independent living potential of individuals with the different levels of intellectual disability

Mild ID:

- May live independently or in a supervised setting
- Provided supports may include decision making for health, nutrition, finances or raising a family
- May need support intermittently, particularly in complex daily living situations (ie: social or financial stress)

Moderate ID:

- Usually live in supervised settings – group home in a community setting with support
- Most learn daily living activities and independent self-care with sufficient teaching and support
- Complex daily living tasks (ie: money management) need considerable support

Severe ID:

- Typically live in a group home (if not behaviorally disruptive) or with their family
- Substantial supports with daily living
- Associated with reduced lifespan

Profound ID:

- Dependent in all aspects of daily living
- Comprehensive assistance often needed
- Medical needs, coexisting conditions and behavioral characteristics impact placement decisions
- Associated with reduced life expectancy

4. Know the factors that mediate or moderate the outcome for individuals with intellectual disability

- Intelligence quotient (IQ) score is not an accurate measure of long term potential

- Combination of adaptive and intellectual functioning is more predictive of outcomes than IQ measure alone
- Quality of life has improved in settings where institutionalization is uncommon and support services and employment opportunities are available
- Outcomes for children with ID depend on multiple factors:
    - Severity Level:
        - Individuals with mild ID have greater capacity to respond to interventions
        - Children with mild ID usually attain more learning, speech and language, reading, interpersonal communication and vocational skills than those with severe ID
    - Cause of ID:
        - Intellectual deficits may vary by cause of ID
        - Down Syndrome: more difficulties in executive functioning and verbal working memory, but better reaction times
        - Most causes of ID do not result in progressive deterioration in function, but some genetic and metabolic causes may result in progressive worsening in function
    - Coexisting Conditions:
        - Comorbid conditions (eg, other developmental disabilities, mental health disorders, vision/hearing impairment, motor deficits, seizure disorder), may impair progress and functional outcomes
        - Behavior difficulties, substance abuse, obesity, depression, suicidal ideation, limited understanding of risk and accidents, neglect, abuse or harm by others may impact course and functioning
    - Environmental and social factors:
        - Family socioeconomic and psychosocial factors may impact a child's course
        - Social opportunities, caregiver support, expectations, attitudes and motivation
        - Children with ID are more likely to be affected by poverty than typically developing children
    - Access to services:
        - Availability and provision of individualized supports has considerable impact on outcomes
        - Deinstitutionalization has improved the lives of many individuals with ID
        - Access, availability, provision of needed interventions, quality and extent of transition and post-school services, necessary expertise in care, residential options, occupational opportunities, transport, socioeconomic and family factors all impact success of transitioning into adulthood

5. Understand the impact of different levels of intellectual disability on social relationships

- Compared with typically developing peers, young children with ID tend to have less frequent play with other children, fewer friends, poorer quality of friendships, and reduced levels of social participation

- Mild ID:
    - Better social skills, communication and social judgement than children with more severe ID
    - Usually can relate to others, communicate in friendship, display social play and extracurricular interests

- Interests and skill levels may be similar to younger children
- Social adaptive and communication outcomes vary by coexisting conditions (ie: Autism)
- Adults with mild ID may marry and become parents, but raising children is challenging

- Moderate ID:
  - Simple spoken language and social friendships are possible, but significantly limited by social and communication deficits
  - Social cues, social judgement and social decisions often require regular help and support
  - Few are able to marry or raise children

- Severe ID:
  - Benefit from socially supportive interactions with familiar people who relate at the level of the individual's development
  - Speech-language development may be delayed until 4-5 years of age, although some do not development speech-language
  - Individuals may use basic single word or phrase or gesture to communicate
  - Substantial assistance with social skills
  - In general, severely affected individuals do not marry or raise children

- Profound ID:
  - Limited understanding of symbolic language, but may be able to understand basic instructions
  - May express nonverbally and understand a simple instruction, gesture or emotional cue
  - Outcomes are variable, and can vary from total dependence to some simple expression and social responsiveness

- Interventions:
  - Social supports may be provided in educational and other settings
  - Community interventions can improve friendship development and inclusion in recreation with typical peers
  - Participation can be enhanced by selecting activities appropriate to the ability and interest of the child, and including individualized mentoring and peer interactions

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Content Category 8- Specific Learning Disorders- Sections A, B, & E

ABP Content Specs for Developmental-Behavioral Pediatrics Fellow Collaboration Project

Prepared by Jonathan Chooey, DO, Madigan Army Medical Center DBP Fellow

Reviewed by Abigail Angulo, MD, Colorado Children's DBP Fellowship Director

8. Specific Learning Disorders

A. General

3. Understand the discrepancy definition of learning disabilities
4. Understand the low achievement definition of learning disabilities
5. Know how to plan the evaluation of a child for a learning disability
6. Understand the structure of educational interventions attempting to provide the least restrictive environment (eg, classroom aide, resource room)
7. Understand the overlap between intellectual disability and learning disorders
8. Know the epidemiology of learning disabilities
9. Know the natural history of learning disabilities
10. Understand the issues related to differentiating learning disabilities from normal variations in academic skill acquisition
11. Understand the concept of learning disability as a failure to respond to validated intervention

B. Specific learning disorder with impairment in reading

1. Understand the cognitive and adaptive skills that are necessary for the typical development of reading abilities
2. Understand the relationship between early language delays and later reading disorders
3. Understand current concepts of the genetics of reading disorders
4. Understand the current concepts regarding the underlying neuropsychological deficits in reading disorders
5. Understand the appropriate educational interventions and accommodations for children with a reading disorder
6. Understand the range of prognoses for children with a reading disorder
7. Know the specific CNS localization of deficits related to reading disorder
8. Know the conditions commonly associated with reading disorder
9. Understand that reading disorders may result in academic failures in other subject areas
10. Know the diagnostic criteria for reading disorder
11. Recognize the symptoms of reading disorder
12. Know the differential diagnosis for low achievement in reading
13. Know subtypes of reading disorder (eg, word reading; reading comprehension)

E. Nonverbal learning disorder

1. Recognize the signs and symptoms of a non-verbal learning disorder
2. Understand the natural history of non-verbal learning disorders
3. Know the conditions commonly associated with non-verbal learning disorders
4. Know the appropriate educational interventions and accommodations for children with non-verbal learning disorder

8. **Specific Learning Disorders**

A. General

1. Understand the discrepancy definition of learning disabilities

   Discrepancy refers to a significant difference between academic achievement and cognitive ability, typically defined as a 2- standard deviations or more difference in academic achievement in reading (fluency or comprehension), writing (spelling or written expression), or math (number sense or mathematical reasoning) and full-scale IQ.

   A student with a learning disability may, in general terms, seem quite capable of learning but have unexpected difficulty in one or more of the academic areas.

   Voigt RG, Macias MM, Myers SM, Tapia CD. *Developmental and Behavioral Pediatrics.* Itasca, IL: American Academy of Pediatrics; 2018.

2. Understand the low achievement definition of learning disabilities

   Academic skills that are substantially and quantifiably below those expected for an individual's chronological age, and lack of response to intervention (see #9 below)

   This definition allows children with a low-average IQ to receive special education services without having to meet a far lower academic achievement score, which would have to occur if a discrepancy definition were used. For example, a child with an IQ of 80-90 (low average) would have to have an academic achievement score of 50-60, whereas a child with an IQ of 100 (average) would only have to have an academic achievement score of 70 to receive services.

   Voigt RG, Macias MM, Myers SM, Tapia CD. *Developmental and Behavioral Pediatrics.* Itasca, IL: American Academy of Pediatrics; 2018.

   Kline M. *Rudolphs Pediatrics.* New York; McGraw-Hill Education; 2018.

3. Know how to plan the evaluation of a child for a learning disability

   If children are not already identified with concerns for LD through school standardized testing or classroom concerns, parents can provide a formal written request for a LD evaluation. Ultimately it is the responsibility of the school to determine whether or not a child qualifies for special education services.

   Voigt RG, Macias MM, Myers SM, Tapia CD. *Developmental and Behavioral Pediatrics.* Itasca, IL: American Academy of Pediatrics; 2018.

4. Understand the structure of educational interventions attempting to provide the least restrictive environment (eg, classroom aide, resource room)

   Part B of the Individuals with Disabilities Education Act (IDEA) requires that educational programs be provided in the least restrictive environment (LRE). This means that, to the extent possible, children in both public or private schools, will be educated with their non-disabled peers, and the school system will provide additional supports and modifications to make this happen. It allows children with special needs opportunities to interact with typical

peers whenever possible and still have support throughout the day. “Special classes, separate schooling, or other removal of children with disabilities from the regular educational environment occurs only if the nature or severity of the disability is such as education is regular classes with the use of supplementary aids and services cannot be achieved satisfactorily.” Placement decisions are made on an individual basis by a child’s IEP team. For a child with a LD, the LRE may be a regular classroom with minimal extra help or a separate education classroom for a certain period of the day or for certain academic subjects, whereas for children with severe disabilities, a self-contained classroom with an aide and classmates that are also disabled may be the LRE.

Voigt RG, Macias MM, Myers SM, Tapia CD. *Developmental and Behavioral Pediatrics.* Itasca, IL: American Academy of Pediatrics; 2018.

5. Understand the overlap between intellectual disability and learning disorders

In the United States, learning disability is not the same as intellectual disability, a diagnosis defined by deficient performance (at or below 2 standard deviation from the mean) on measures of both intellectual skill and functional skills in self-help and adaptive behavior. In contrast, children who have LD perform overall within the normal range of intelligence and adaptive functioning but show isolated specific academic skills deficits.

Rimrodt SL, Lipkin PH. Learning Disabilities and School Failure. *Pediatrics in Review*. 2011;32(8): 315-324

6. Know the epidemiology of learning disabilities

Prevalence estimates: 5-15% across all LD types among school-age children across different languages and cultures.
Incidence (likelihood of developing LD during childhood):
- Reading disorders range from 5.3-11.8%
- Mathematics disorders 5.9-13.8%

Boys are 2 to 3 times more likely than girls to manifest any type of LD
Risk factors:
- Prematurity especially if <32 week gestational age, and with perinatal and postnatal complications (prolonged ventilation, intracranial hemorrhage, sepsis, seizures, prolonged acidosis, or hypoglycemia)
- Cyanotic congenital heart disease

Genetic disorder associated with LD:
- Klinefelter syndrome
- Turner syndrome (risk of problems with visuospatial cognitive skills and math achievement)
- Velocardiofacial syndrome
- Spina bifida with shunted hydrocephalus (risk of problems with visuospatial cognitive skills and math achievement)

Voigt RG, Macias MM, Myers SM, Tapia CD. *Developmental and Behavioral Pediatrics.* Itasca, IL: American Academy of Pediatrics; 2018.

7. Know the natural history of learning disabilities

Onset, recognition, and diagnosis of specific learning disorder generally occurs in elementary school as children are required to begin learning reading, writing and math skills. However,

there can be signs that appear prior to that time. For example, preschool children may not show an interest in learning rhymes or playing games that involve repetition. They may also use baby talk, mispronounce words, and have trouble remembering names of letters (including those in their own name), numbers, or days of the week. Preschool and kindergarten children may also have trouble associating the letters with the sounds they make, in breaking words down into syllables, and recognizing words that rhyme.

Elementary school children typically display marked difficulty learning letter-sound correspondence, fluent word decoding, spelling, or math facts; reading aloud is slow, inaccurate, and effortful, and some children struggle to understand the magnitude that spoken or written number represents. In math areas, they may have problems learning basic math facts and procedures for adding and subtracting. Children with specific learning disorder may complain frequently that reading or math are too hard or try to avoid doing work in those subjects.

As they progress into middle school, they may mispronounce or skip parts of long, multi-syllable words and confuse words that sound alike. They may have trouble remembering dates, names, and telephone numbers and may have trouble completing homework or tests on time. They may write using poor spelling and ignore common grammar rules. They may express fear of reading aloud or refuse to read aloud.

Adolescents may continue to show these problems around comprehension, spelling and grammar rules, and be unable to master math facts. Although they may have mastered word decoding, reading remains slow and effortful, and they are likely to show marked problems in reading comprehension and written expression, and poor mastery of math facts or mathematical problem solving. They may have issues with drawing conclusions from what is read and try to avoid reading in school or as a leisure activity.
Diagnostic and Statistical Manual of Mental Disorders: DSM-5. Washington, DC; American Psychiatric Publishing; 2013.

8. Understand the issues related to differentiating learning disabilities from normal variations in academic skill acquisition

   Specific learning disorder is distinguished from normal variations in academic attainment due to external factors (e.g., lack of educational opportunity, consistently poor instruction, learning in a second language), because the learning difficulties persist in the presence of adequate educational opportunity and exposure to the same instruction as the peer group, and competency in the language of instruction, even when it is different from one's primary spoken language.

   Diagnostic and Statistical Manual of Mental Disorders: DSM-5. Washington, DC; American Psychiatric Publishing; 2013.

9. Understand the concept of learning disability as a failure to respond to validated intervention

"The DSM-5 characterizes "specific learning disorders" as being evidenced by academic skills that are substantially and quantifiably below those expected for an individual's chronologic age, and that are not due to intellectual disability, uncorrected vision or heart deficits, other mental or neurological disorders, psychosocial adversity, lack of proficiency in the language of academic instruction, or inadequate instruction. Thus, child with specific learning disorders must now show a lack of response to intervention before being considered to have a learning disability by many school districts."

Follows the low achievement definition above.

Response to intervention is when schools provide additional support for struggling students, even before a diagnosis is established.

Kline M. *Rudolphs Pediatrics.* New York; McGraw-Hill Education; 2018.

Rimrodt SL, Lipkin PH. Learning Disabilities and School Failure. *Pediatrics in Review*. 2011;32(8): 315-324

B. Specific learning disorder with impairment in reading
    1. Understand the cognitive and adaptive skills that are necessary for the typical development of reading abilities

    The skills needed for normal reading encompass five domains:

    - Phonemic awareness - A critical language skill in the development of reading. It is the ability to recognize and manipulate individual sounds of language (phonemes) to form words. This skill progresses from primitive manifestations (rhyming) to more sophisticated forms, including the ability to blend component sounds into a word (/f/ /ah/ /t/ is blended into fat), to segment sounds of a word (fat is segmented into /f/ /ah/ /t/), and to delete a sound from an intact word (deleting /f/ from fat leaves /at/).
    - Phonics – Knowledge of relationships between letters and sounds and spelling-sound correspondence, which allows decoding of words. The English language is particularly difficulty for beginning readers because it contains inconsistent mappings between letters and sounds.
    - Sight word acquisition – automaticity of reading words by sight without having to decode sound by sound; sight word acquisition facilitates reading efficiency and allows interpretation of homonyms.
    - Vocabulary – the knowledge of word meanings
    - Comprehension – An active, intentional process where meaning is extracted from text. Reading comprehension is influenced by the ability to decode and sight read, vocabulary, oral comprehension, and executive functioning. When reading passages, children construct meaning from sentences, which they must remember as reading progresses; at the same time they must link the information that is read to their background knowledge to anticipate what comes next.

    Grizzle KL, Simms MD. Early Language Development and Language Learning Disabilities. *Pediatrics in Review.* 2005;26(8): 274-283
    UptoDate

    2. Understand the relationship between early language delays and later reading disorders

Oral language skills, including phonology, semantics, grammar, and pragmatics are the foundation for reading. Dyslexia is therefore characterized by reading recoding problems with core problems based in phonological (speech) processing problems. However broader, language skills, including vocabulary and comprehension processes, are involved and can modify the impact of phonological difficulties. Children with more diffuse language problems typically are at higher risk for reading comprehension deficits. Children with persistent SLI (speech language impairment) at 8.5 years of age have been shown to have pervasive problems with spelling, word-level reading, and reading comprehension at 15 years of age.

Voigt RG, Macias MM, Myers SM, Tapia CD. *Developmental and Behavioral Pediatrics.* Itasca, IL: American Academy of Pediatrics; 2018.

3. Understand current concepts of the genetics of reading disorders

   Family and twin studies suggest that 50% of the problems in performance can be accounted for by heritable factors, with environmental influences greater in children who have lower IQ scores. Genetic linkage analyses suggest loci on chromosome 2, 3, 6, 15, and 18, with four candidate susceptibility genes (DYX1C1, KIAA0319, DCDC2, ROBO1) identified. These genes are involved in neuronal migration and axon growth.

   Rimrodt SL, Lipkin PH. Learning Disabilities and School Failure. *Pediatrics in Review*. 2011;32(8): 315-324

4. Understand the current concepts regarding the underlying neuropsychological deficits in reading disorders

   The majority (80%) of individuals who have an RD have difficulty with single-word decoding, a condition referred to as dyslexia. Decoding refers to the ability to sound out words and requires that a child understand the “alphabetic principle”; that is, each letter has a corresponding sound, and these sounds can be combined to make words. Not only do poor decoding skills impair efficient, quick, and automatic reading, they also can have a pronounced effect on comprehension of text. The more cognitive effort that is devoted to reading word-by-word, the less is available to understanding what is being read. Although slow, dysfluent reading is typical in young developing readers, continuing this somewhat arduous process without the expected increase in reading vocabulary and the automatic recognition of words can have dramatic effects on what a child is able to comprehend and the amount of material that he or she is able to read.

   As previously noted, the primary language skill involved in the development of word reading is phonologic awareness. Failing to make progress in the acquisition of PA places a child at much greater risk to experience reading difficulties. Deficits at the phonologic level of language affects a child’s ability to recognize that words she or he hears can be broken into individual sounds and, in turn, results in the child finding it difficult to perform the most basic of reading skills, decoding words. Strong reading has a “boot strapping” effect over time; that is, good readers tend to read more frequently and, in turn, improve their reading, language, vocabulary, and academic skills in general. Poor readers, on the other hand, fall further behind their peers in reading skills and in other academic areas, vocabulary, and general cognitive skills. Children experiencing reading difficulties who do not receive the benefit of effective early instruction are not likely to catch up to same-age/grade peers. Evidence suggests that nearly 90% of children identified as poor

readers at the end of first grade remain poor readers at the end of fourth grade. In fact, as many as 75% of children determined to have RDs early in their school careers continue to struggle with reading throughout their lives. Conversely, only 5% to 10% of children who develop satisfactory reading skills early in their school experience struggle with reading later in life.

Grizzle KL, Simms MD. Early Language Development and Language Learning Disabilities. *Pediatrics in Review.* 2005;26(8): 274-283

5. Understand the appropriate educational interventions and accommodations for children with a reading disorder

   Multiple studies have clearly demonstrated that reading curricula that included explicit teaching of phonics are more effective. This is not surprising because phonological awareness has emerged as a critically important prerequisite for the development of good, basic reading skills.

   For children who qualify for special educational services, the IEP must list specific learning goals, as well as the nature and intensity of services to be provided. Some children may require direct, individual, or small-group instruction with a special education teacher in a special education resource room. In other cases, support from the regular or special education teacher, or a paraprofessional assistant, may be provided in the regular classroom. According to federal law, interventions must be provided in the LRE while still meeting the child's educational needs. In addition to direct instructional services, IEP's can include accommodations, such as shortened assignments, increased time to complete tests, or oral administration of tests for children with reading problems. The key is to ensure that services and accommodations specifically match the demonstrated needs of the child based on the results of individual assessment of learning strengths and weaknesses.

   Voigt RG, Macias MM, Myers SM, Tapia CD. *Developmental and Behavioral Pediatrics.* Itasca, IL: American Academy of Pediatrics; 2018.

6. Understand the range of prognoses for children with a reading disorder

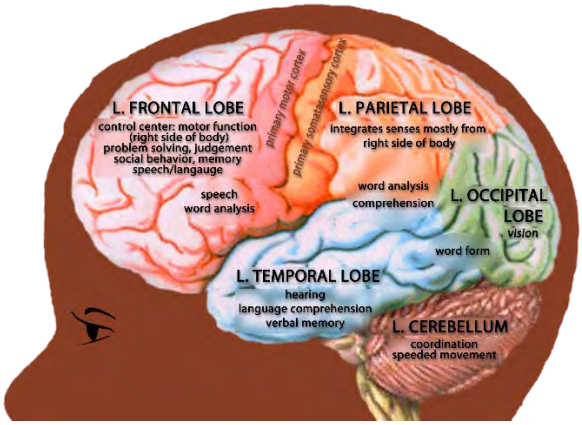
   The prognosis for children who have LD can range from academic and social success to ongoing vocational and personal-social problems in adulthood. Affected children completing high school, and exhibiting behaviors that, in some cases, may escalate and lead to expulsion. However, with proper parent, school, and community support, children who have LD can reach high levels of achievement.

   Rimrodt SL, Lipkin PH. Learning Disabilities and School Failure. *Pediatrics in Review*. 2011;32(8): 315-324

7. Know the specific CNS localization of deficits related to reading disorder

   Children with dyslexia demonstrated a disruption in neural systems for reading involving posterior brain regions, including parietotemporal sites and site in the occipitotemporal area. Reading skills was positivity correlated with the magnitude of activation in the left occipitotemporal region.

   Shaywitz BA, Shaywitz SE, Pugh KR, et al. Disruption of posterior brain systems for reading in children with developmental dyslexia. *Biol Psychiatry.* 2002; 52(2):101-10



8. Know the conditions commonly associated with reading disorder

   Genetic Syndromes:
   Neurofibromatosis type 1, 50% frequency. (LD and school failure)
   Klinefelter Syndrome

   Boada R, Janusz J, Hutaff-Lee C, Tartaglia N. The Cognitive Phenotype in Klinefelter Syndrome: A Review of the Literature Including Genetic and Hormonal Factors. *Dev Disabil Res Rev.* 2009; 15(4): 284-294

   Associated conditions:
   Dysgraphia
   ADHD
   Auditory processing disorder
   Developmental coordination disorder

9. Understand that reading disorders may result in academic failures in other subject areas

   In later grades, when children switch from learning to read to reading to learn, reading-impaired children are prevented from fully exploring science, history, literature, mathematics, and the wealth of information that is presented in print.

   Handler SM, Fierson WM, et al. Joint Technical Report-Learning Disabilities, Dyslexia, and Vision. *Pediatrics.* 2011: 2010-3670

10. Know the diagnostic criteria for reading disorder

    Specific Learning Disorder Diagnostic Criteria

    A. Difficulties learning and using academic skills, as indicated by the presence of at least one of the following symptoms that have persisted for at least 6 months, despite the provision of interventions that target those difficulties:
       1. Inaccurate or slow and effortful word reading (e.g., reads single words aloud incorrectly or slowly and hesitantly, frequently guesses words, has difficulty sounding out words).

2. Difficulty understanding the meaning of what is read (e.g., may read text accurately but not understand the sequence, relationships, inferences, or deeper meanings of what is read).
3. Difficulties with spelling (e.g., may add, omit, or substitute vowels or consonants).
4. Difficulties with written expression (e.g., makes multiple grammatical or punctuation errors within sentences; employs poor paragraph organization; written expression of ideas lacks clarity).
5. Difficulties mastering number sense, number facts, or calculation (e.g., has poor understanding of numbers, their magnitude, and relationships; counts on fingers to add single-digit numbers instead of recalling the math fact as peers do; gets lost in the midst of arithmetic computation and may switch procedures).
6. Difficulties with mathematical reasoning (e.g., has severe difficulty applying mathematical concepts, facts, or procedures to solve quantitative problems).

B. The affected academic skills are substantially and quantifiably below those expected for the individual's chronological age, and cause significant interference with academic or occupational performance, or with activities of daily living, as confirmed by individually administered standardized achievement measures and comprehensive clinical assessment. For individuals age 17 years and older, a documented history of impairing learning difficulties may be substituted for the standardized assessment.
C. The learning difficulties begin during school-age years but may not become fully manifest until the demands for those affected academic skills exceed the individual's limited capacities (e.g., as in timed tests, reading or writing lengthy complex reports for a tight deadline, excessively heavy academic loads).
D. The learning difficulties are not better accounted for by intellectual disabilities, uncorrected visual or auditory acuity, other mental or neurological disorders, psychosocial adversity, lack of proficiency in the language of academic instruction, or inadequate educational instruction.

Note: The four diagnostic criteria are to be met based on a clinical synthesis of the individual's history (developmental, medical, family, educational), school reports, and psychoeducational assessment.

Diagnostic and Statistical Manual of Mental Disorders: DSM-5. Washington, DC; American Psychiatric Publishing; 2013.

11. Recognize the symptoms of reading disorder

Children with a reading disorder may be able to use compensation strategies such as rote memorization of a large number of sight words, and their reading disorder may not be apparent until upper elementary school. When apparent, the signs and symptoms of a reading disorder are similar to those of all learning disabilities, which include increased learning effort, school distress, and school failure.

Increasing learning effort: “school is boring,” school anxiety, class clown behavior, spending a much longer time completing homework than classmates
School distress: frequent failing grades, frequent absences, social disengagement, frequent detention, suspensions, aggression and bullying behaviors.
School failure: retention, expulsion, dropping out

Rimrodt SL, Lipkin PH. Learning Disabilities and School Failure. *Pediatrics in Review*. 2011;32(8): 315-324

12. Know the differential diagnosis for low achievement in reading

Normal variations in academic achievement
Intellectual disability
Learning difficulties due to neurological or sensory disorders
Neurocognitive disorders
ADHD
Psychotic disorders

Diagnostic and Statistical Manual of Mental Disorders: DSM-5. Washington, DC; American Psychiatric Publishing; 2013.

13. Know subtypes of reading disorder (eg, word reading; reading comprehension)

- Phonological (word reading accuracy) – implicating a core problem in the phonological processing system of oral language
- Processing speech/orthographic processing deficit (reading rate or fluency) – affecting speech and accuracy of printed word recognition (also call naming speed problem or fluency problem)
- Comprehension deficit – often coinciding with the first two types of problems, but specifically found in children with social-linguistic disabilities (i.e. ASD), vocabulary weaknesses, generalized language learning disorders, and learning difficulties that affect abstract reasoning and logical thinking.

Moats L, Tolman C. Excerpted from Language Essentials for Teachers of Reading and Spelling (LETRS): The Challenge of Learning to Read (Module 1). Boston: Sopris West.

Fletcher JM. Dyslexia: The evolution of a scientific concept. *J Int Neuropsychol Soc.* 2009; 15(4): 501-508.

C. Specific learning disorder with impairment in mathematics (Puji)
    1. Understand the appropriate educational interventions and accommodations for a child with mathematics disorder
    2. Understand the range of prognoses for children with mathematics disorder
    3. Know the conditions commonly associated with mathematics disorder
    4. Know the diagnostic criteria for mathematics disorder
    5. Recognize the symptoms of mathematics disorder
    6. Understand the cognitive and adaptive skills that are necessary for the typical development of mathematics abilities
    7. Know the current concepts regarding the underlying neuropsychological deficits in mathematics disorder
    8. Know the differential diagnosis for low achievement in mathematics
    9. Understand the genetics of mathematics disorder
D. Specific learning disorder with impairment in written expression (Jillian)

1. Know the diagnostic criteria for disorder of written expression
2. Understand the cognitive and adaptive skills that are necessary for the typical development of writing and spelling abilities
3. Know the differential diagnosis of disorders of written expression and spelling
4. Know the components of the evaluation of children for disorders of written expression or spelling
5. Know the appropriate educational accommodations for children with spelling disorder or disorder of written expression
6. Recognize the symptoms of disorders of written expression
7. Know the underlying neuropsychological deficits in disorders of written expression

E. Nonverbal learning disorder

1. Recognize the signs and symptoms of a non-verbal learning disorder

   Children with non-verbal learning disorders have difficulties with visual and spatial awareness, higher-order comprehension, social communication, math concepts, and executive function. They have difficulty recognizing patterns or concepts and then applying them to new situations. They may have difficulty understanding visual imagery (unable to copy of cube) and evaluating visual-spatial information (making them appear physically awkward because they are unable to grasp the relationship between things they see and having a clear sense of where they are). They may have difficulty with reading comprehension, writing, or telling a story effectively. They may also have difficulty reading emotions in facial cues and body language leading to difficulties with social communication. They may have difficulty with more advanced mathematical problems based on recognizing concepts and patterns. They tend to have difficulty learning things that aren't rote or literal.

   https://childmind.org/article/what-is-non-verbal-learning-disorder/

2. Understand the natural history of non-verbal learning disorders

   Since a child's earliest mode of communication is non-verbal, parents may suspect that "something is amiss" early on, but they can't quite "put a finger on it." The child may have difficulty in motor coordination and balance, visual spatial organization, and difficulty with non-verbal communication. They may appear awkward and with poor fine and gross motor coordination despite high intelligence and high scores on receptive and expressive language measures. Due to their exceptional memory and rote learning skills, they do well in elementary school and usually go undiagnosed until late elementary school or middle school, when their difficulty with higher order reasoning causes them to fall behind. As less and less is "spelled out" for them, they will reach a point where functioning n school is impossible without specific compensations, accommodations, modifications, and strategies. If they are not supported appropriately, they may cease to try or "burn-out" attempting to succeed under these impossible demands, and develop other psychiatric disorders such as depression or anxiety.

   http://www.ldonline.org/article/6114/

   https://childmind.org/article/what-is-non-verbal-learning-disorder/

3. Know the conditions commonly associated with non-verbal learning disorders

   Autism spectrum disorder

Social communication disorder
ADHD

https://childmind.org/article/what-is-non-verbal-learning-disorder/

4. Know the appropriate educational interventions and accommodations for children with non-verbal learning disorder

There is no standard treatment approach for non-verbal learning disorders; each child requires individualized approaches to educational intervention in order to succeed in school. They may require direct verbal training in planning, organizing, studying, written expression, social cognition, and interpersonal communication. Physical therapy and social skills groups may also help. Although non-verbal learning disorders are not covered under IDEA specifically, it can be included in the DSM-5 category of Unspecified Neurodevelopmental Disorders, which may help the child receive additional services through an IEP.

Voigt RG, Macias MM, Myers SM, Tapia CD. *Developmental and Behavioral Pediatrics.* Itasca, IL: American Academy of Pediatrics; 2018.

http://www.ldonline.org/article/6114/

https://www.additudemag.com/what-is-nonverbal-learning-disorder-symptoms-and-diagnosis/

Content Category 8- Specific Learning Disabilities- Section C and F-5

ABP Content Specs for Developmental-Behavioral Pediatrics Fellow Collaboration Project

Prepared by Puji Jonnalagadda, MD, Colorado Children's DBP Fellow

Reviewed by Abigail Angulo, MD, Colorado Children's DBP Fellowship Director

8. Specific Learning Disorders

C. Specific learning disorder with impairment in mathematics

1. Understand the appropriate educational interventions and accommodations for a child with mathematics disorder
2. Understand the range of prognoses for children with mathematics disorder
3. Know the conditions commonly associated with mathematics disorder
4. Know the diagnostic criteria for mathematics disorder
5. Recognize the symptoms of mathematics disorder
6. Understand the cognitive and adaptive skills that are necessary for the typical development of mathematics abilities
7. Know the current concepts regarding the underlying neuropsychological deficits in mathematics disorder
8. Know the differential diagnosis for low achievement in mathematics
9. Understand the genetics of mathematics disorder

F. Communication disorders

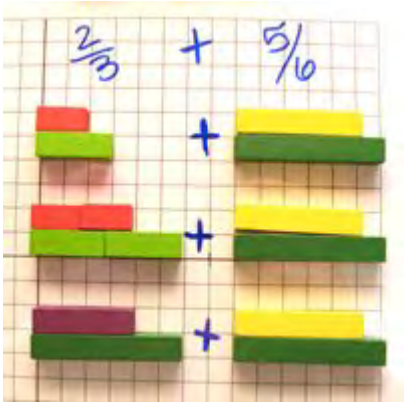
5. Social (pragmatic) communication disorder
   a. Know the criteria for social (pragmatic) communication disorder
   b. Know the differential diagnosis for social (pragmatic) communication disorder

C. Specific learning disorder with impairment in mathematics

1. Understand the appropriate educational interventions and accommodations for a child with mathematics disorder

Drilling on mathematical facts to increase automation of arithmetic skills DOES NOT help in children with mathematics learning disability (MLD) as it does when compared to typically developing peers. Following are things that are helpful for children with MLD:

- Visual cues and experiential materials are helpful with concepts of basic math such as numerical values and quantity.
- Cuisenaire rods (shown below) are color-segmented rods with different colors representing different numbers 1 to 10 that can be used help children understand some math concepts.



https://cmschoolsupply.com/math/1394-connecting-cuisenaire-rods-introductory-set.html
http://mathfour.com/arithmetic/adding-fractions-with-cuisenaire-rods

- Number lines are another visual aid that can be used to help children understand how numbers related to each other.
- Word problems might be difficult for some with MLD, especially those with also a reading disability. These children will need intervention to assist with understanding word problems and then developing strategies to work through word problems. For example, they need to practice understanding particular words or symbols.
- Children who are more severe MLD struggling self-monitoring and setting realistic goals for themselves based on their performance. For these children, allowing accommodations for assisting in executive functions may be helpful.

Table 2 Methods for accommodating students with MD

Methods	Accommodations
Improving reading skills	Break up the text into smaller sections
	Use a simple font
	Do not justify the text
	Use colored overlays to reduce glare
Improving mathematical problem solving skills	Photocopy math books with the relevant sections placed in order
	Separate complicated problems into small steps
	Use markers to highlight, and guide attention
	Use color to delineate columns and rows in spreadsheets
	Simplify tables
General instructional design	Supplement incomplete notes
	Use posters to remind students of various basic concepts
	Use flash cards
	Provide flow charts to clarify procedures
	Engage visual learners with manipulatives
	Encourage students to move at their own pace
	Teach organization, studying, and time management skills
	Focus on revision prior to an examination

Not an exhaustive list. MD, math disability.

2. Understand the range of prognoses for children with mathematics disorder

There is little prognostic data about the short-term and long-term outcomes of MLD. Some sources suggest that MLD appears to persist, at least for the short-term, in about half of affected preteen children. Individuals who struggle with procedural mathematical tasks (frequent careless errors, using their fingers to count, struggle to understand the basic mathematical process behind an arithmetic task) could improve with age and grade levels. There is a persistence in mathematical difficulties. Factors associated with persistence of MLD are severity of the disorder at the time of initial diagnosis and the presence of similar problems in siblings.

3. Know the conditions commonly associated with mathematics disorder

Isolated learning disability in mathematics is unusual but can be found in children with non-verbal learning disabilities. It is unclear what percentage, but a proportion of children with Math learning disability also have Reading disability and/or ADHD. On study found between 57 - 64% of those with MLD (depending on the diagnostic criteria used for MLD) also have a reading disability (RD).

MLD is frequently encountered in disorders of epilepsy, Turner syndrome, 22q11.2 deletion syndrome, Fragile X syndrome, myelomeningocele and Neurofibromatosis-1. Children with Williams syndrome have significant visuospatial deficits that may contribute to MLD features.

4. Know the diagnostic criteria for mathematics disorder

Specific learning disorder in DSM-V combines the DSM-IV diagnoses for reading disorder, mathematics disorder, disorder of written expression and learning disorder not otherwise

specified. Because learning deficits in all these areas can occur together, coded specifier for each deficit type are now included in DSM-V instead.

DSM-V criteria for Specific learning disorder:
A. Difficulties in learning and academic skills as indicated by at least one symptom in any area of difficulty (reading, comprehension, spelling, written, numbers, math reasoning) that have persisted for at least 6 months, despite the provision of interventions to target specific difficulties. When considering impairment in mathematics, these can include:
- Difficulty mastering number sense, number facts or calculation (example, has poor understanding of numbers, their magnitude, and relationships, counts on fingers to add single digit numbers instead of recalling the math facts when compared to peers; gets lost in the midst of arithmetic computation and may switch procedures).
- Difficulty in mathematical reasoning (example, has severe difficulty applying mathematical concepts, facts or procedures to solve quantitative problems.

B. Difficulties in academics are significantly and quantifiably below expectations for individual's age, and are interfering with academic or occupational performance.
C. Difficulties begin during school age years, but may not become fully manifested until academic demands increase and excess the individual's limited capacities in this area
D. Learning difficulties are not better explained by intellectual disability, uncorrected visual or auditory acuity, other mental or neurological disorders, psychosocial adversity, and lack of proficiency in the language in which academic instruction is being provided or inadequate educational instruction.

Specifier, 315.1 (F81.0) Specific Learning Disorder with impairment in mathematics:
Number sense
Memorization of arithmetic facts
Accurate or fluent calculation
Accurate math reasoning

Dyscalculia is the alternative term used to refer to these mathematical difficulties. "If dyscalculia is used to specify this particular pattern of mathematic difficulties, it is important also to specify any additional difficulties that are present, such as difficulties with math reasoning or word reasoning accuracy."

5. Recognize the symptoms of mathematics disorder

Symptoms are age and grade related. Three theoretical subtypes have been suggested:
1. Procedural: frequent careless errors, using their fingers to count, struggle to understand the basic mathematical process behind an arithmetic task.
    - Have deficits in retrieval of learned information and will use inefficient strategies in order to bypass their difficulties in solving arithmetic problems.
2. Semantic Memory: show problems with memory and retrieval of math facts.
3. Visuospatial and Number Sense: problems with special representation, aligning columns, understanding the relationship between number and quantity.

Symptoms of MLD:
- Difficulty learning to count or counting

- Difficulty sorting
- Difficulty corresponding numbers to objects
- Difficulty with auditory memory of numbers (such as a phone number)
- Difficulty subitizing (ability to instantly recognize how many object are contained within a small group without counting)
- Trouble with number recognition
- Difficulty comparing magnitudes
- Trouble learning match facts
- Difficulty with math problem solving skills
- Over reliance on finger counting for basic arithmetic
- Anxiety during math tasks
- Difficulties with precision during math work
- Difficulty remembering previously encountered patterns
- Difficulty sequencing multiple steps of math problem
- Difficulty understanding real-world representation of math formula
- Trouble applying math to money matters, estimating speech and distance while driving
- Trouble with measurements
- Difficulty with graphs or charts

6. Understand the cognitive and adaptive skills that are necessary for the typical development of mathematics abilities

Most school aged children learn to count by rote. This ability to count is not in and of itself a diagnostic marker for MLD. Pre-math skills such as one-to-one correspondent counting, correct order of numbers and understanding sorting and categorizing objects are some developmental important skills. Counting is a big one, as some findings indicate that children with mathematics disorder do not understand the counting process that underlies most mathematical skills such as addition. Arithmetic skills are initially learned and memorized. These then become more automated with practice. When younger, children use their fingers to count that they don't need to do once arithmetic skills become automated. Children with mathematic learning disorder (MLD) do not move from finger counting strategy to automatic processing until much later than their peers. Children with MLD also learn problem solving and memory retrieval of number facts much later that their peers.

7. Know the current concepts regarding the underlying neuropsychological deficits in mathematics disorder

Visuospatial, verbal or phonological and auditory-perceptual dysfunctions have been proposed as a neuropsychological mechanism underlying MLD. Working memory, speech of processing and overall intelligence (IQ) have been studied and identified as cognitive correlates that cause or modify expression of MLD. Working memory deficits, especially in the central executive function are seen in children with MLD. Tasks that utilize the central executive require a child to hold one or several pieces of information in mind, while simultaneously performing another mental tasks.

Individuals with MLD have difficulties most consistently in domains of number sense, semantic memory and procedural competence. These domains in turn affect developmental of number, counting and arithmetic development. There are implicit principles of counting that

mature during the preschool years that promote mathematical skills in a child with typical development. These are listed and defined below. Children with MLD do not detect a violation of these principles. For example, they may be confused when counting deviates from the standard of left to right or may not detect errors such as double counting a single object.

Implicit Counting Principles and Unessential Features of Counting

Implicit Principle	Description
One-one correspondence	One and only one word tag (e.g., *one*, *two*) is assigned to each counted object.
Stable order	Order of the word tags must be invariant across counted sets.
Cardinality	The value of the final word tag represents the quantity of items in the counted set.
Abstraction	Objects of any kind can be collected together and counted.
Order irrelevance	Items within a given set can be tagged in any sequence.
Unessential Feature	**Description**
Standard direction	Counting proceeds from left to right.
Adjacency	Consecutive count of contiguous objects.
Pointing	Counted objects are typically pointed at, but only once.
Start at an end	Counting starts at one of the end points of an array of objects.

There is evidence to indicate that children with MLD have difficulties with subitizing and the ability to represent approximate quantities. Subitizing means to judge the number of objects in a group rapidly, accurately and confidently without counting them. Children and adults can subitize for sets of 1 to 3, sometimes 4, objects, but not more than this.

8. Know the differential diagnosis for low achievement in mathematics

Differential for MLD should include:

- Intellectual disability
- Learning difficulties due to a neurological process or sensory disorder
- ADHD, especially of the inattentive type: Children with ADHD can make arithmetic mistakes as a results of impaired recall, careless errors and inattention to detail.
- Gerstmann syndrome: Disorder of cognitive impairment characterizing by four primary symptoms (deficits in writing, difficulty with mathematics, finger agnosia, left-right disorientation) that is often associated with a brain lesion
- Dyslexia or SLD in reading
- Neurological conditions such as Epilepsy, especially if the mathematical difficulties are newly developed or have regressed from prior abilities
- Environmental deprivation
- Psychiatric or psychotic disorder, especially when there is a rapid domain in one or more domains of learning

9. Understand the genetics of mathematics disorder

MLD is a brain-based disorder with familial/genetic predisposition. Similar to other forms of SLD, both genetic and environmental factors (such as poor teaching and environmental

deprivation) contribute to development of MLD. One study sounds that children family members of children with MLD are 10 times more likely to be diagnosed also with MLD compared to the general population. There are no specific identified genes but rather genetic influences on MLD. About 2/3rd of the genetic influences on MLD are the shared with learning in other academic areas such as reading, and 1/3rd of these are specific to mathematics learning. This could explain the co-morbidity of MLD with a reading disability or other difficulties that interfere with learning such as ADHD.

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F. Communication disorders

5. Social (pragmatic) communication disorder

a. Know the criteria for social (pragmatic) communication disorder

Characterized by primary difficulties in pragmatic or social use of language or communication, manifested by deficits in understanding and following social rules of communication, and modifying language according to the needs of the audience. These can results in functional limitations in effective communication, social participation and development of relationships, academic achievement or occupational performance. Usually associated with language impairments or language delays. Individuals with this might avoid social interactions.

DSM-IV Criteria for Social (Pragmatic) Communication Disorder – 315.39 (F80.89)
A. Persistent difficulties in the social use of verbal and nonverbal communication as manifested by all of the following:
1. Deficits in using communication for social purposes, such as greeting and sharing information, in a manner that is appropriate for the social context.
2. Impairment of the ability to change communication to match context or the needs of the listener, such as speaking differently in a classroom than on a playground, talking differently to a child than to an adult, and avoiding use of overly formal language.
3. Difficulties following rules for conversation and storytelling, such as taking turns in conversation, rephrasing when misunderstood, and knowing how to use verbal and nonverbal signals to regulate interaction.

b. Know the differential diagnosis for social (pragmatic) communication disorder

It is important to note that some of these in the differential are also co-morbidities. ADHD, behavioral problems and specific learning disorders are common among affected individuals. Differential diagnosis for social communication disorders include:
1. Autism Spectrum Disorder
2. Attention-Deficit/hyperactivity disorder
3. Social anxiety disorder (social phobia)
4. Intellectual disability (intellectual developmental disorder) and global developmental delay

References:

1. American Psychiatric Association. *Diagnostic and Statistical Manuel of Mental Disorders, Fifth Edition.* Arlington, Virginia: American Psychiatric Association; 2013.

Child Neuropsychology

Content Category 8- Communication Disorders- Section F

ABP Content Specs for Developmental-Behavioral Pediatrics Fellow Collaboration Project

Prepared by Sara Williams, MD, Colorado Children's DBP Fellow

Reviewed by Abigail Angulo, MD, Colorado Children's DBP Fellowship Director

F. Communication disorders

1. Language
    a. Understand the distinction between speech and language
    b. Distinguish the phonological, semantic, grammatical, prosodic and pragmatic aspects of language
    c. Know the differential diagnosis of language disorders
    d. Understand the role of hereditary factors in language disorders
    e. Understand theories about the causes of language disorder
    f. Recognize disorders in semantic skills
    g. Recognize disorders in phonological skills
    h. Recognize disorders in syntax
    i. Recognize impairments in prosodic skills
    j. Understand the distinction between receptive and expressive language skills
    k. Know the diagnostic criteria for language disorder
    l. Know how to plan the evaluation of a child with language delay
    m. Know how to plan the management of a child with a language disorder
    n. Know the range of prognoses for children with different types of language disorders
    o. Understand the neural basis of language functioning and language development
    p. Understand the impact of exposure to more than one language (bilingual household) on language development
    q. Understand the role of environmental factors in language disorders
    r. Understand the neural basis for language disorders
    s. Know the epidemiology of language disorders
    t. Recognize the signs and symptoms of language disorders
    u. Understand gender differences in language development
2. Speech sound disorders
    a. Understand the distinctions among articulation, voice/resonance, and fluency
    b. Know the pathophysiologic factors that affect articulation
    c. Recognize the developmental progression of articulation skills
    d. Know the definitions of dysarthria and oral-motor apraxia
    e. Know the difference between articulation disorders, phonological disorders, and childhood apraxia of speech
    f. Know how to plan the evaluation of a child with articulation problems
    g. Know the differential diagnosis of a child with articulation delays
    h. Know how to plan the management of a child with speech abnormalities
    i. Understand the range of prognoses for children with articulation and phonological disorders
3. Childhood onset fluency disorder (stuttering)
    a. Understand the normal development of speech fluency and normal dysfluency of childhood
    b. Understand the normal development of speech fluency
    c. Know the criteria for referral of a child with speech dysfluency
    d. Understand the management of a child with speech dysfluency
    e. Understand the prognosis for a child with speech dysfluency
4. Voice/resonance
    a. Know the pathophysiologic factors that affect voice and resonance
    b. Understand the evaluation of a child with abnormalities of voice or resonance
    c. Understand the management of a child with abnormalities of voice or resonance
    d. Know the prognosis for a child with abnormalities of voice or resonance

e. Know the role of velopharyngeal function in speech sound production

F. Communication disorders

1. Language
    a. Understand the distinction between speech and language
        i. Language: distinctly human form of communication that uses arbitrary but socially agreed upon signals and rules to convey meaning[1]; involves both expressive and receptive components
        ii. Speech: the output of the language system, created by coordinated movements of respiratory, laryngeal, velopharyngeal, and oral structures; sign language and written language can also be outputs
    b. Distinguish the phonological, semantic, grammatical, prosodic and pragmatic aspects of language
        i. Phonological (system of speech sounds): related to the individual sounds (*phonemes*), which are the simplest "units" of language, and the articulation of these sounds
        ii. Semantic (meaning): related to the individual word and sentence meanings
        iii. Grammatical (syntax): related to the order of words in phrases and sentences; the set of rules which govern the combinations of morphemes and words into sentences
        iv. Prosodic: related to the vocal intonation used when pronouncing words
        v. Pragmatic (social aspects that consider the speaker and context): related to the social use of language, including body language, volume of speech, and conversational skills
    c. Know the differential diagnosis of language disorder: If a language delay is suspected, the following diagnoses should also be considered.
        i. Hearing loss: With severe to profound hearing loss (threshold <55 dB), language and speech disorders are highly likely.[1] With mild (threshold of 26-40 dB) to moderate (threshold of 41-55 dB) sensorineural hearing loss, the child may be able to detect vowel sounds and low-frequency consonants (*m, b*), but may not detect high-frequency consonants (*f, th, sh*). The child should be referred for a formal audiologic examination, as even mild hearing loss can cause language delays and may not be picked up by newborn screening.[2]
        ii. Autism spectrum disorder: One of the two key criteria for autism is deficits in social communication. ASD may present with speech/ language delays or a regression in this domain. An autism-specific screening tool should be used along with general developmental screening, as early intervention improves the prognosis.
        iii. Intellectual disability/ global developmental delay: Children with global developmental delays may initially present with language and speech delays. If these delays persist and are accompanied by adaptive skill deficits, a child may meet criteria for intellectual disability. Generally, language skills are commensurate with other developmental abilities.[1]
        iv. Genetic disorders: Genetic conditions have distinctive language profiles. The following are several examples:

1. Klinefelter syndrome (47, XXY): Consider this, especially if gynecomastia or hypogonadism is also present in a male with language delays and language-related learning difficulties.
2. Rett syndrome: Consider this, especially in a female with language regression.
3. Trisomy 21/ Down syndrome: Speech and language deficits are often more severe than deficits in nonverbal skills.
4. Fragile X syndrome (trinucleotide repeats in the FMR1 gene): Individuals often have poor language skills accompanied by poor social skills, hyperactivity, and anxiety.

v. Social factors: Screen for possible trauma, psychosocial deprivation or other environmental factors (see part q below).

vi. Neurologic disorders:

1. Structural brain injuries may affect language, especially with injury to the frontal and temporal regions.

2. Landau-Kleffner syndrome is characterized by the sudden or gradual development of aphasia, usually occurring between 3 to 7 years of age. The first indication of the language problem is often auditory verbal agnosia, in which patients do not recognize familiar sounds and are impaired in their ability to localize sound. Clinical or electrographic seizures are also common.

d. Understand the role of hereditary factors in language disorders: Disorders of language cluster in families. In families with children with language impairments, the median incidence of language difficulties in first-degree relatives is more than 3 times higher than in unaffected families.[1] There is also a higher risk of speech/ language disorders in monozygotic vs. dizygotic twin pairs and a greater risk of a reading disorder in a child whose family members have reading disorders.[1] Specific genes associated with language disorders are an ongoing topic of research.
e. Understand theories about the causes of language disorder: Many congenital, environmental, and genetic conditions are known risk factors for the development of language disorders. Focal (e.g. intracranial hemorrhage) or diffuse brain injury (e.g. blunt head trauma, abuse) can cause language disorders ranging from mild to profound impairment. Additionally, there is a genetic component to the development of many speech/ language disorders, although this is an area of ongoing research. A child's language learning environment and exposure to neglect or abuse also affect language development.
f. Recognize disorders in semantic skills: Language disorders in semantic skills involve a lack of understanding of the meaning and appropriate use of single words, phrases, and sentences. Poor semantic skills may include an inability to recognize and name categorical labels, difficulty in using descriptive words, or trouble with recognition of words by their definitions. A child with semantic language problems may therefore have difficulty following verbal instructions, finding the words to express himself, categorizing words, understanding figurative language, or choosing specific words (e.g. using *stuff* or *things* often because specific names of objects are difficult to produce).

g. Recognize disorders in phonological skills: Phonological disorders may involve independent articulation errors or phonological rule-based error patterns. There are characterized by the substitution (one or more sounds are substituted, like *wabbit* for *rabbit*), omission (deletion of certain sounds, like *poon* for *spoon*), addition (insertion of an extra sound(s), like *buhlack* for *black*), or distortion of phonemes (alteration of sounds, like a lateral *s* causing a lisp).[2] Most speech therapy referrals are for articulation difficulties.
h. Recognize disorders in syntax: Language disorders in syntax involve difficulties in understanding the rules that govern word order in a sentence or phrase. A child struggling with syntax may leave out words, use incorrect word order, or only use a limited number of complex sentences. S/he may have difficulty understanding the use of different verb tenses (e.g. using past/ present/ future, the progressive - *ing*, or auxiliary verbs like *to be*) or grammatical markers (e.g. the possessive *'s* or indefinite articles).
i. Recognize impairments in prosodic skills: Language disorders in prosodic skills involve difficulties with modulating the rhythm of speech. This can result in robotic-like speech (in which each syllable is pronounced one at a time and with equal stress), dysprosodic speech (in which the prosody does not match the expected intonation pattern), aprosodic speech (monotone or decreased intonation patterns), or speech that is too fast or slow.
j. Understand the distinction between receptive and expressive language skills: Receptive skills involve an individual's ability to *understand* language while expressive skills involve the ability to *produce* language (i.e. trouble sharing thoughts, ideas, and feelings). Deficits in receptive language almost always occur in conjunction with expressive language deficits.[2]
k. Know the diagnostic criteria for language disorder: A communication disorder is defined as "an impairment in the ability to receive, send, process, and/or comprehend verbal, nonverbal or graphic symbol systems."[2] While *delays* in language development imply that the child will catch up, a *disability* is diagnosed when deficits are sufficiently severe to interfere with daily functioning (e.g. communication, learning, social interactions).[1]

   The DSM-5 criteria for language disorder includes the following[7]:

   a. Persistent difficulties in the acquisition and use of language due to deficits in the comprehension or production of vocabulary, sentence structure and discourse
   b. Language abilities below those expected for age that result in functional limitations
   c. Symptom onset in the early developmental period
   d. The difficulties are not better explained by a medical or neurologic diagnosis

l. Know how to plan the evaluation of a child with language delay: If a delay falls below 75% of the child's expected development, a comprehensive evaluation should occur, as outlined in the chart below.[1]

TABLE 3. Evaluation of a Child with Language or Speech Delay or Disorder

COMPONENT OF EVALUATION	CRITICAL INFORMATION
History	Age at onset, initial presentation, and subsequent course Affected subcomponents Effects of previous treatments
Medical history	Birth history Previous illnesses and chronic conditions
Review of systems	Associated signs and symptoms
Family history	Family members with language, reading, or intellectual disorders Other health conditions
Psychosocial factors	Primary caregivers, parent education Number in the household, financial resources, stress Nature of parent-child interactions Amount and quality of child-directed speech Out-of-home care, including child care, preschool, or school
Physical examination	Growth parameters Dysmorphic features Oral-motor structure and function
Neurologic examination	Structural abnormalities, such as asymmetry in tone or reflexes Functional disorders, such as seizures
Audiologic assessment	Ear-specific information or sound field testing
Cognitive skills	Developmental assessment or an intelligence test
Symptoms of autism	Social communication and restrictive or repetitive behaviors

m. Know how to plan the management of a child with a language disorder: All children suspected of language impairment should be promptly referred for hearing testing and to their local early intervention program or school system. If the child fails a language screening test, an evaluation by a speech-language pathologist is indicated to determine a treatment plan. Speech-language therapy may then be provided by the early intervention program or school system and, if additional supports are indicated, on an outpatient basis as well. Additionally, parent education is key to encourage language stimulation activities. Childcare centers or preschool programs can also be beneficial, especially if the delay is related to lack of a stimulating home environment or developmental variation.[2]

   Treatment of speech-language disorders includes 3 components[2]:
   1. Causal: Repair defects and eliminate factors that contribute to the language problem (e.g. cleft palate repair or hearing aids)
   2. Habilitative: Work to directly improve the child's language skills through speech-language therapy, parent training to help parents engage in the child's language development, and other supports
   3. Supportive: Boost language acquisition by supporting the development of other skills (e.g. increase social interaction or initiate a training program for speech-related skills)

n. Know the range of prognoses for children with different types of language disorders:

i. Of those children delayed in language at 2 years of age, approximately half catch up by age 3.[1] Approximately 60% of children with early language delays will catch up by age 4 years with no persistent problems.[2]
ii. In children with late language emergence, factors associated with decreased risk of continued problems include: younger age at diagnosis, continued progress with language development, age-appropriate receptive language abilities, good symbolic play skills, and a greater number of gestures to compensate for the decreased verbal language.[1,2] Factors associated with increased risk of early delays include family history of language delay, low socioeconomic status, and less "rich" language environments (i.e. exposure to less language).[3]
iii. Importantly, while therapy may improve the degree of language impairment and the prognosis, many children with early language delays continue to have difficulties as they grow, although the delays may manifest differently with time. They may struggle, for example, with reading, written expression, learning appropriate social skills, or learning a foreign language.[2]

o. Understand the neural basis of language functioning and language development: Language learning is a product of both a child's language environment and his/ her learning capacities. "Parallel distributed processing" or "connectionist frameworks" describe computer simulation models that imitate characteristics of the human brain structure and function and have provided insights into how young children learn language.[1] Per this model, "Units that become active simultaneously develop connections, and connections that occur in unison form networks. Knowledge is conceptualized as patterns of activity within the network; learning represents changes in the patterns of activity within and across networks."[1] Auditory input is also critical for organizing the neural pathways associated with speech.[3] The sound stream is divided into meaningful units (e.g. words) that serve as inputs for learning. Once a child's vocabulary reaches approximately 50 words, the pace of development increases as his/ her neural network self-organizes and white matter language pathway myelination matures.[1]
p. Understand the impact of exposure to more than one language (bilingual household) on language development:
    i. Bilingual children have similar risks of language disorders as monolingual children; bilingual exposure is *not* an adequate explanation of language delays in itself.[1] These children may have smaller vocabularies in each of their languages compared to monolingual children, but their combined vocabulary should be comparable in size. They may also show mixing of the two languages, but separation of the 2 languages will occur over time, usually around 3-4 years of age.[3] Evaluation of the bilingual child with language delays should use the same criteria as for monolingual children, as bilingualism is not an adequate explanation for language delay in a typically-developing child.
q. Understand the role of environmental factors in language disorders:
    i. Home environment: Poverty, crowding, limited breastfeeding, and parental smoking are all risk factors for poor language development.[1] Infants raised in orphanages often have profound delays in speech/ language

development, which can be mitigated by foster care placement before 2 years of age.[1]

ii. Relationships and engagement: Parental education level, parental health, socioeconomic status, and the level of engagement of parents with children are all associated with the rate of language development.[1] The amount of speech that young children hear predicts speed of language processing and lexical growth as well.[1]

iii. Other health conditions: Preterm birth is associated with persistent delays in language development, even in the absence of major disabilities.[1] These delays have been attributed to subtle neurologic deficits that may not be apparent on neuroimaging. Likewise, children with congenital heart disease also show language delays associated with white matter changes.[1]

iv. Factors that do NOT often explain delays in language:

1. Birth order: Despite frequent parental reports of "his/ her older sibling always talks for him/ her," language development is rarely delayed because of birth order. Overall, studies show that later-born children do not acquire language later than firstborn children. Slthough they are less likely to hear adult-generated language directed toward them, they are more likely to overhear conversations between adults and their older siblings.[1]
2. Chronic otitis media with effusion (OME): Chronic OME is not a clear cause of language delays. OME usually results in minimal-to-mild conductive hearing loss, which has been shown to not significantly impair long-term language development in otherwise healthy children due to the mild and fluctuating characteristics of the deficit.[3] Randomized clinical trials have shown that although tympanostomy tube placement may decrease the duration of an effusion, it does not change language outcomes in preschool or school-age children.[1] However, chronic otitis media is more prevalent in situations that also put language learning at risk (including limited breastfeeding, parental smoking, poverty, crowding), suggesting that it may be a marker for adverse conditions associated with poor language/ speech development, rather than a cause.[1] Also, because a small proportion of children with OME may develop moderate hearing loss, hearing testing should be performed if effusions persist for 3 months or longer or at any time if hearing impairment or delays are suspected.[3]
3. Bilingual households: See section p, above

r. Understand the neural basis for language disorders: The neural basis for language disorders is an ongoing topic of research. It was previously taught that Broca's and Wernicke's areas were *the* language centers, bridged by the arcuate fasciculus. Broca's area in the (usually) left frontal region was responsible for speech comprehension, while Wernicke's area in the (usually) left temporal region was responsible for speech production. However, these concepts are now considered too simplistic, especially in the developing brain, as more advanced brain imaging capabilities have shown that language processing depends on many neural sites working in concert. In one recent analysis of five different studies, children with

speech disorders had “structural and functional anomalies in the left supramarginal gyrus and functional anomalies in the posterior cerebellum bilaterally – regions critical for sensory-motor integration or feedback.”[5] Additionally, this analysis found that children with language disorders demonstrated “increased mean and radial diffusivity of the left arcuate fasciculus, although a widespread cortical and subcortical network of regions was implicated.”[5] Even so, there is yet only limited evidence of specific regional brain anomalies or MRI prognostic markers for children with language disorders.

s. Know the epidemiology of language disorders:
   i. Approximately 16% of children have difficulties in the early phases of language learning, which approximately half of those children demonstrating persistent difficulties.[1] Additionally, 13-18% of 1.5 to 3-year-old children present with expressive language delays.[3]
   ii. Of the 2.3% to 24.6% of school-aged children estimated to have speech delay or speech sound disorders, 11% to 40% of children also have concomitant language impairment.[4]
   iii. In the 3-5 year age range, impairment in speech and language is the most common reason for eligibility and enrollment in special education preschool services (45% were eligible for services based on speech-language impairment).[1]
   iv. Among school-age children, speech-language impairment is the second most common eligibility criterion for special education (following learning disabilities, which may be a late manifestation of language/ speech disorders).[1] A 2012 survey from the National Center for Health Statistics estimated that, of those children with a communication disorder, 48.1% of 3- to 10-year old children and 24.4% of 11- to 17-year old children had speech sound problems only.[4]

t. Recognize the signs and symptoms of language disorders
   i. Early detection of language delays depends on a thorough knowledge of patterns of normal development.
   ii. “Red Flags” that indicate a high likelihood of a persistent disorder and

TABLE 2. **Red Flags Indicating High Risk of Language or Speech Disorder and Prompting Evaluation**

AGE	RED FLAG OR INDICATION FOR REFERRAL FOR EVALUATION
Any age	Failure to participate freely and frequently in social interactions
6 mo	Lack of ability to laugh, vocalize, respond to sound, participate in reciprocal vocal interactions
9 mo	Failure to respond differentially to name or to produce babble (such as *baba, dada*)
12 mo	Inability to point to objects or actions Lack of use of gestures, such as shaking head "no" Inability to participate in verbal routines, such ability to wave to "wave bye-bye" No use of *mama* or *dada* specifically for a parent
18 mo	<5 words beyond *mama* and *dada* Failure to follow simple commands with gestures
24 mo	Vocabulary <50 words No 2-word combinations <50% of utterances intelligible to unfamiliar adults
36 mo	Inability to follow simple directions without gestures No ≥3-word combinations <75% of utterances intelligible to unfamiliar adults
>36 mo	Loss of language and speech skills, particularly in the presence of regression in social abilities and in the absence of regression in motor skills

should prompt an evaluation include the following[1]:

u. Understand gender differences in language development:
    i. While studies have shown that girls are more talkative than boys at all ages (measured in total words), with significant gender differences found between 1 to 2.5 years of age, boys' language development generally still falls within the accepted time frame despite a potential few months delay in expressive language.[2] However, boys do have higher rates of language and speech disorders and it should not therefore be assumed that a boy with language delays will "catch up" without intervention.[1]

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2. Speech sound disorders

a. Understand the distinctions among articulation, voice/resonance, and fluency:

- Articulation involves the production of specific sounds of a specified language. The most frequently misarticulated sounds in English are *l, r, s, z, th, ch, dzh,* and *zh*).[1]
- Fluency involves the flow of speaking. Dysfluent speech includes pauses, interjections, prolongations, and interruptions.[2]
- Voice includes variations in volume, pitch, strain and voice quality. Resonance is the result of the transfer of sound by the vocal cords through the vocal tract. A hyper- or hyponasal resonance quality may suggest anatomical differences.

b. Know the pathophysiologic factors that affect articulation: Articulation may be affected by the placement, timing, direction and speed of the tongue, lips, jaw and airflow. For most children with articulation difficulties, there is not an identifiable physical reason for their problem. However, the following may contribute to articulation difficulties:

i. Hearing impairment: If a child has a limited ability to hear the speech of others and to monitor their own speech production, this will result in articulation problems.

ii. Neurologic problems: If a child has neuromuscular impairment causing dysarthria (poor control of muscles required for speech production) or dysphagia (swallowing difficulties), his/ her articulation will likely also be affected.

iii. Structural defects: Cleft lip, cleft palate, ankyloglossia (tongue tie), and complete or partial glossectomy impair articulation by allowing air to escape into the nasal cavity or by preventing the tongue tip from freely moving.

iv. Apraxia: Apraxia involves an impaired ability to plan and initiate motor movements involving the speech musculature.

c. Recognize the developmental progression of articulation skills: Learning to produce speech sounds clearly and accurately and then learning to produce connected speech in a fluent manner are developmental skills. As typically developing children learn to talk, they commonly use developmental processes for shortening words and syllables. By the time they reach 3 years age, their speech should be about 75% intelligible. By 4 years of age, children should be 100% intelligible. By 7 years of age, most children are able to clearly produce the majority of speech sounds and accurately pronounce most words. The "late eight" sounds (*l, r, s, z, th, ch, dzh,* and *zh*) are most frequently misarticulated, which could be considered within normal limits at up to age 8 years.[1] Children with speech sound disorders continue to use immature phonological simplification processes past the age when most children are able to produce words clearly.[1] While mild articulation disorders are very common and rarely disrupt communication or social functioning, severe disorders may affect other domains of function.[3] Moderate to severe phonologic difficulties occur in approximately 2-3% of young school age children.[4]

d. Know the definitions of dysarthria and oral-motor apraxia: Dysarthria is a disorder involving motor control of muscles required for speech production. It often occurs after a neurologic insult and is seen in disorders like cerebral palsy, acquired brain injury, and neuromuscular disorders.[4] Dysarthria is characterized by abnormal tone, strength, or coordination of facial, oral-motor, and respiratory muscles, resulting in speech that is disordered and labored.[4] It is treated with speech therapy and with augmentative communication indicated in severe cases. Oral-motor apraxia, in contrast, involves difficulties with *planning* speech, rather than with *execution* of speech production. When participating in an oral-motor exam (which may include activities like pursing the lips, smiling, frowning, and speaking tasks), a child with dysarthria will exhibit impaired movement due to muscle weakness or low tone in the lips, jaw, and/ or tongue, while a child with apraxia will exhibit difficulties with planning and inconsistent error patterns without weakness or abnormal tone.

e. Know the difference between articulation disorders, phonological disorders, and childhood apraxia of speech: Articulation and phonological disorders are often difficult to clearly differentiate between. Articulation disorders focus on errors in the production of individual speech sounds (like substitutions and distortions), while phonological disorders focus on predictable, rule-based errors that affect more than one sound (like final consonant deletion).[5] Childhood apraxia of speech (CAS) is a disorder of planning speech sounds,[3] rather than a weakness of the muscles required to produce speech (dysarthria). Hallmarks include inconsistent errors in producing words, delayed onset of babbling or single-word production, errors in articulating vowel sounds, increasing difficulty with words of increasing length, and increased difficulty with production of spontaneous speech (as opposed to well-rehearsed speech).[3] Treatment emphasizes intensive, individual treatment with a focus on motor learning and motor programming.

Unlike the child with a phonological disorder, who makes consistent and predictable articulation errors, a child with CAS exhibits irregular and inconsistent speech production patterns.

f. Know how to plan the evaluation of a child with articulation problems: The first step in evaluating a child with articulation difficulties is to screen for a speech sound disorder by having a child pronounce individual speech sounds in single words and in connected speech using informal or formal screening measures. A screening of their oral motor functioning and an orofacial examination is also indicated to assess the strength and range of motion of their oral musculature and to identify possible anatomic causes for articulation problems (submucous cleft palate, ankyloglossia, malocclusion).[5]

If a child is then referred for a comprehensive speech sound assessment with a speech-language pathologist, a referral should also be made to an audiologist for a hearing evaluation and to other medical services, as appropriate.

g. Know the differential diagnosis of a child with articulation delays: It is important to consider potential physiologic causes (hearing impairment, neurologic deficits, structural defects, and apraxia, as described in part b) and possible accompanying language disorders. Disorders of speech may occur in conjunction with disorders of language or in isolation. The influence of accent and dialect should also be considered, as there are regional, social, and cultural variations of speech that should be considered as normal.

h. Know how to plan the management of a child with speech abnormalities: Once a child has been referred for speech therapy, either through an Early Intervention Program, a school-based program, a private outpatient program, or a combination of the aforementioned, treatment usually involves three steps[5]:

- Establishment: Selecting initial sounds to target with therapy and stabilizing the production of those sounds on a voluntary level
- Generalization: Teaching the child to carry over the learned sound productions at increasingly difficult levels (e.g. syllables, then words, then phrases/ sentences, then at a conversational level)
- Maintenance: Further stabilizing sound production to be more automatic; encouraging self-monitoring for errors and self-correction

The child's speech-language therapist will then select treatment modalities based on a variety of factors, including the child's age, type of speech sound errors produced, the severity of the disorder, and the degree to which the disorder affects the child's overall intelligibility.[5]

i. Understand the range of prognoses for children with articulation and phonological disorders: Of the 2.3% to 24.6% of school-aged children estimated to have speech delay or speech sound disorder, 11% to 40% of children also have concomitant language impairment.[5] Only an estimated 1% to 2% of older children and adults continue to make persistent or residual speech errors.[5] A child with persisting speech difficulties may be at risk for difficulty developing reading and writing skills, difficulty communicating effectively on an interpersonal level, and psychosocial problems (e.g. bullying, low self-esteem).[5] Continued support and educational accommodations into adulthood may be indicated. When a language disorder is also present, the prognosis is poorer and may be associated with specific learning disorders.

3. Childhood onset fluency disorder (stuttering)

a. Understand the normal development of speech fluency and normal dysfluency of childhood: Around 2 to 3 years old, children often exhibit developmental dysfluency as their speech production rapidly increases while their thinking outpaces their speech processes.[3] They repeat entire words or phrases as they speak (*I want, I want, I want some juice* or *I, I, I want some juice*) and use incomplete phrases, interjections, parenthetical remarks, and unfilled pauses. This usually resolves by 4 years of age.

b. Understand the normal development of speech fluency: The ability to speak fluently is a skill that develops gradually as children grow; it is normal for a child to occasionally hesitate or stumble as they begin to put sounds, words, and sentences together, especially between the ages of 2- to 6-years old. The most common dysfluency is whole word repetitions, usually at the beginnings of sentences. Pauses filled with filler words (*I want my, um, toy*), silent pauses (*Mommy, I want (pause) my toy*), and infrequent part-word repetitions (*Y-you said I could have it*) may also occur. Normal dysfluencies may increase when a child is excessively tired, excited, or in a more formal speaking environment (such as speaking with an adult). Dysfluencies decrease in frequency as the child grows.

c. Know the criteria for referral of a child with speech dysfluency: A child should be referred when she/ he exhibits symptoms of stuttering, rather than normal dysfluency of childhood. Stuttering involves the repetition of individual speech sounds (*p-p-p-pop),* prolongations (*mmmme*), excessive use of fillers (*uh, um,* etc), abnormal pauses between sounds, and the use of behavioral strategies to try to get the words out (for example, self-slapping).[2] Stuttering also causes anxiety in the affected child about speaking or limitations in effective communication, academic or occupational performance, or social participation.[1]

d. Understand the management of a child with speech dysfluency: Even when younger than 4 years of age, features of stuttering should prompt a referral for direct speech therapy. Treatment is often effective and involves targeting both the speech production problem itself and any associated emotional and attitudinal issues.[4]

e. Understand the prognosis for a child with speech dysfluency: Stuttering occurs in approximately 1% of school age children and is three times more common in boys, with a strong family influence.[4] Onset is almost always before 10 years of age and usually around age 4 to 5 years old.[4] For many children, dysfluency resolves, with longitudinal estimates of remission varying from 65% to 85%.[1]

4. Voice/resonance

a. Know the pathophysiologic factors that affect voice and resonance:

Voice disorders may be organic, functional, or psychogenic in nature. Organic voice disorders have a physiological cause that results in alterations in respiratory, laryngeal or vocal tract mechanisms.[7] The alterations may result from a structural change (e.g. edema of the vocal cords, vocal cord nodules, post-intubation laryngeal trauma, laryngitis) or a neurogenic change due to dysfunction of the central or peripheral nervous system (e.g. vocal tremor, spasmodic dysphonia, recurrent laryngeal nerve paralysis).[7] With functional voice disorders, improper or inefficient use of the voice mechanism affects the individual's voice in the absence of structural abnormalities (e.g. vocal fatigue

due to overuse, phonotrauma due to excessive yelling, diplophonia). Psychogenic voice disorders result in altered voice due to anxiety, depression, a conversion reaction, or chronic stress.

Resonance disorders result from either too much or too little nasal and/ or oral sound energy in the speech signal.[8] The cause may be structural or functional and can result in hypernasality (excessive nasal resonance during production of voiced, oral sounds), hyponasality (insufficient nasal resonance during production of nasal sounds due to obstruction), cul-de-sac resonance (sound "trapped" in a cavity due to obstruction), or mixed resonance.[8] Genetic syndromes are often associated with resonance disorders because of co-occuring velopharyngeal insufficiency (e.g. 22q11.2 deletion syndrome, CHARGE syndrome, Treacher Collins syndrome, Stickler syndrome). Abnormal resonance may also develop with hearing loss, dysarthria, or after adenoidectomy or uvulopalatopharyngoplasty.[8]

b. Understand the evaluation of a child with abnormalities of voice or resonance: If a voice/ resonance disorder is suspected, screening should first occur. This involves an evaluation of the child's respiration, phonation, resonance, vocal range, and vocal flexibility in addition to an oral exam to look for anatomical differences.[7] A formal screening tool or informal tasks may be used. If not already performed, hearing screening should also occur. If concerns persist, a comprehensive assessment should include a physician and a speech-language pathologist. The evaluation will assess for impairments in anatomical structure and function, potential co-morbid health conditions that may be contributing, limitations in the child's day-to-day functioning in communication and daily living skills, and any environmental or personal factors that may be contributing to the problem.[7] Imaging of the larynx using videoendoscopy may be indicated.

c. Understand the management of a child with abnormalities of voice or resonance: The goal of treatment of a voice disorder is to achieve improved voice production and improved coordination of respiration and laryngeal valving.[7] If a voice disorder has a physiological cause, SLPs often coordinate efforts with other medical professionals (e.g. otolaryngologists, pulmonologists, gastroenterologists, neurologists, allergists) to develop treatment plans. In the absence of structural pathology, a psychologist may also be helpful in identifying possible psychologic contributing factors. For voice disorders, SLPs often begin by identifying behaviors that contribute to the voice problems, like shouting, coughing, throat clearing, or poor hydration, and implementing healthy "vocal hygiene" practices.[7] For resonance disorders, surgical management, prosthetic management, pharmacologic management, and behavioral speech therapy may all be indicated, depending on the cause of the disorder.[8] Interventions for voice and resonance disorders will also seek to build new communication skills and provide appropriate accommodations and supports.

d. Know the prognosis for a child with abnormalities of voice or resonance: In the pediatric population, the estimated prevalence of voice disorders ranges from 1.4% to 6%.[7] Based on previous studies, if voice disorders are left untreated, approximately 1/3 of children will not improve, while there is growing evidence of successful treatment with appropriate therapy.[9] For resonance disorders, speech therapy may be an effective treatment option for mild disorders. For moderate to severe velopharyngeal dysfunction, corrective surgery with post-operative speech therapy is often indicated.

e. Know the role of velopharyngeal function in speech sound production: Velopharyngeal function is key to normal speech sound production. To produce most speech sounds, the air and sound produced from the lungs and vocal cords must be directed into the mouth and blocked from entering the nasal

cavity. This is accomplished by closure of the velopharyngeal valve. The valve consists of the velum, or soft palate, and the lateral and posterior pharyngeal walls. Velopharyngeal insufficiency (VPI) may result from structural differences (e.g. unrepaired cleft palate, whether overt or submucous, or postsurgical insufficiencies), neurogenic etiologies (e.g. dysarthria, apraxia, TBI, stroke, neuromuscular disease), or from velopharyngeal mislearning (secondary to hearing loss, compensatory misarticulations, or phoneme-specific nasal emissions).[8] Cleft palate is most commonly associated with velopharyngeal dysfunction and up to 30% of individuals continue to have VPI post-cleft palate repair.[8]

5. Social (pragmatic) communication disorder (*Puji)*

a. Know the criteria for social (pragmatic) communication disorder

b. Know the differential diagnosis for social (pragmatic) communication disorder

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Content Category 9 Motor and Disabilites and Multiple Handicaps

ABP Content Specs for Developmental-Behavioral Pediatrics Fellow Collaboration Project

Prepared by Rebecca Christi, MD, Madigan Army Medical Center DBP Fellow

Reviewed by Michael Msall, MD, UChicago Medicine

9. Motor Disabilities and Multiple Handicaps

A. Cerebral palsy
1. Recognize signs in early infancy that are associated with the later development of cerebral palsy
2. Recognize the signs and symptoms of spastic cerebral palsy
3. Know the prevalence and epidemiology of cerebral palsy
4. Distinguish the different subtypes of spastic cerebral palsy
5. Recognize the signs and symptoms of dyskinetic cerebral palsy
6. Know specific causes of the different types of cerebral palsy
7. Distinguish cerebral palsy from spinal cord injuries, peripheral motor disorders, and lower motor neuron lesions
8. Know how to plan the management of a toddler or preschooler with cerebral palsy
9. Know how to plan the management of a school-age child or adolescent with cerebral palsy
10. Evaluate early intervention and physical therapy in the management of cerebral palsy
11. Know the pharmacologic management of spasticity
12. Know the natural history of cerebral palsy
13. Know the management of drooling in children with cerebral palsy
14. Understand the neurological, orthopedic and/or ophthalmological complications associated with cerebral palsy
15. Know the developmental and behavioral characteristics of individuals with cerebral palsy
16. Know the range of prognoses for children with different types of cerebral palsy
17. Understand the classification of cerebral palsy
18. Characterize degree of impairment in cerebral palsy using the Gross Motor Function Classification System
19. Recognize the signs and symptoms of ataxic cerebral palsy
20. Understand the role of neuroimaging in the assessment of cerebral palsy

B. Myelodysplasia
1. Know the high prevalence of hydrocephalus and Chiari malformation in children with myelodysplasia
2. Understand the relationship between the level of myelodysplasia and motor, cognitive, and adaptive dysfunction
3. Know how to plan the management of children with different levels of myelodysplasia
4. Know the conditions commonly associated with myelodysplasia
5. Understand the relationship between genetic and environmental factors in the etiology and prevention of myelodysplasia
6. Understand the urological, neurological, and/or orthopedic complications associated with myelodysplasia
7. Know the developmental and behavioral characteristics of individuals with myelodysplasia

C. Muscular dystrophies
1. Recognize the signs and symptoms of Duchenne and other muscular dystrophies
2. Understand the long term prognoses for youth with Duchenne and other muscular

dystrophies

3. Know how to plan the laboratory evaluation for a child with progressive muscular weakness
4. Understand the genetics of Duchenne and other muscular dystrophies
5. Know how to plan the management of a boy with Duchenne and other muscular dystrophies
6. Understand the neurological and orthopedic complications associated with muscular dystrophy
7. Know the developmental and behavioral characteristics of individuals with muscular dystrophy

D. Other

1. Plan the evaluation of an infant with hypotonia
2. Recognize the typical presentation of developmental coordination disorder
3. Know appropriate management strategies for a child with developmental coordination disorder
4. Know the causes of congenital hypotonia
5. Know the signs and symptoms of spinal muscular atrophy (SMA)
6. Plan the evaluation of a child with developmental coordination disorder

9. **Motor Disabilities and Multiple Handicaps**

A. Cerebral palsy

1. Recognize signs in early infancy that are associated with the later development of cerebral palsy
    - CP is diagnosed clinically with physical findings, but it is supported by the prenatal, perinatal, and postnatal history
    - The diagnosis of CP is based on a combination of significant gross and fine motor developmental delay and focal abnormalities on neurologic evaluation
        - Significant motor developmental delay is defined as development occurring at 50% or less than the expected rate (e.g. a child walking for the first time at more than 24 months of age is considered > 50% delayed in gross motor skills)
    - Signs in early infancy:
        - Significant motor delay, as above
        - Deviancy in motor skills: e.g., attaining motor skills early like rolling at 2 months can be a sign of hypertonia
        - Abnormal neurological exam findings such as abnormal resting muscle tone (can be either hypertonia or hypotonia), increased DTRs, pathologic reflexes (e.g. abnormal Babinski), abnormal primitive reflexes, and delayed protective or postural responses
            - Examples:
                - Moro or ATNR persisting beyond 6 months of age
                - Postural responses not beginning to emerge by 6 months

2. Recognize the signs and symptoms of spastic cerebral palsy
    - Spasticity: refers to the usual hypertonia that may be elicited when muscles are rapidly and passively extended across a joint. It is a velocity-dependent increase in tone, and it is described as having a “catch-then-release” quality. Note that spasticity is not elicited when muscles are stretched slowly.
    - Signs and symptoms of spastic CP: spasticity, hyperreflexia, clonus, positive Babinski reflex (extensor plantar response) typically can all be elicited to various degrees in spastic CP. Also, voluntary muscle control and strength overall are diminished proportionally to the degree of CNS dysfunction.
    - Note: though spasticity is the key finding in spastic CP, global hypotonia is often found as an early phase finding during the natural course of spastic CP. Also, in spastic CP, hypotonia can persist in the neck and trunk, coexisting with spasticity in the extremities.

3. Know the prevalence and epidemiology of cerebral palsy
    - The prevalence of CP has remained fairly constant at a rate of approximately 2 per 1,000 live births.
        - However, there is some variance in how this is distributed; for example, recent studies have shown that for term infants the prevalence of CP is less than 2 per 1,000 live births, and that for preterm or low birth weight infants it can range as high as 50-80 per 1,000 live births.
        - About half of the total cases of CP are attributed to preterm infants.
    - Etiologies:
        - In most cases, the etiology remains unknown or unproven
        - Prenatal in origin:
            - Congenital brain malformations
            - Neuronal migration disorders

        - Vascular disturbances
        - Genetic syndromes
        - Maternal infections
        - Other maternal factors
    - Peri- and postnatal causes:
        - Trauma
        - Asphyxia
            - HIE (regardless of gestational age) due to a severe anoxic event may acutely manifest as seizures or specific neurologic deficits, but in can later present as CP (especially as the spastic quadriplegic or extrapyramidal (dyskinetic) subtypes)
        - Infections
        - Cerebral hemorrhage
            - Preemies are especially vulnerable to cerebral hemorrhage, particularly IVH with subsequent PVL – this often results in spastic CP
        - Kernicterus
            - Historically a causative factor for choreoathetoid or mixed CP

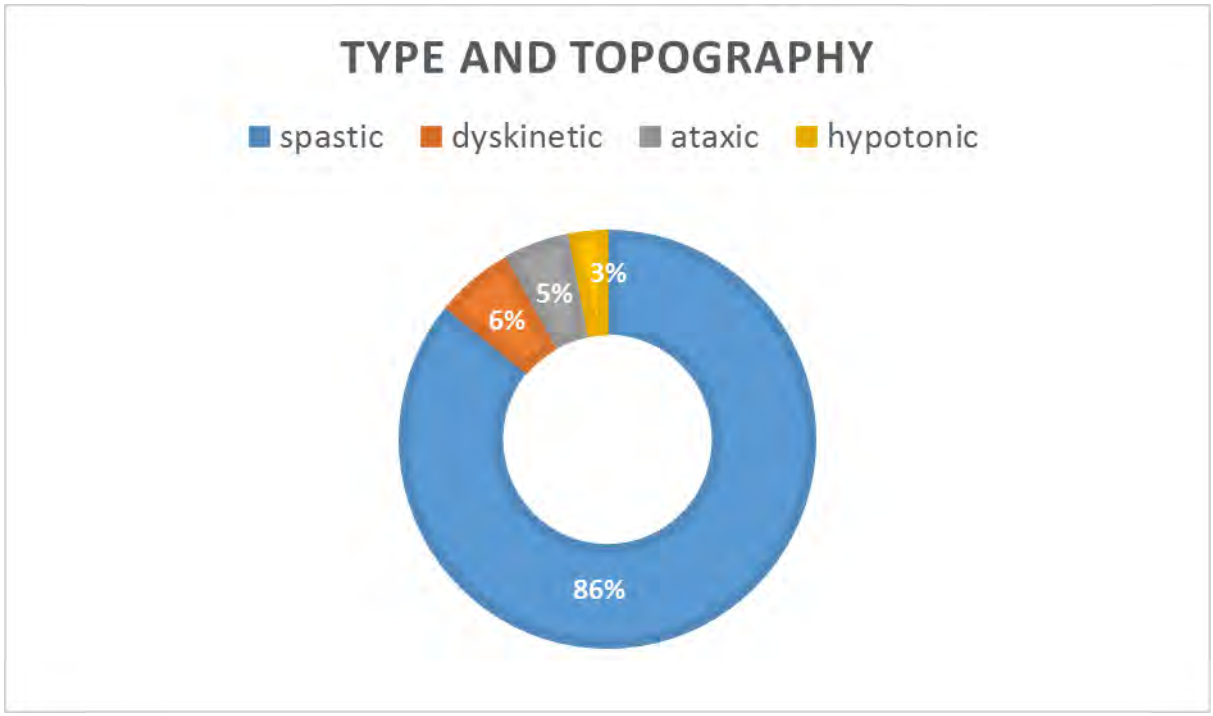
4. Distinguish the different subtypes of spastic cerebral palsy

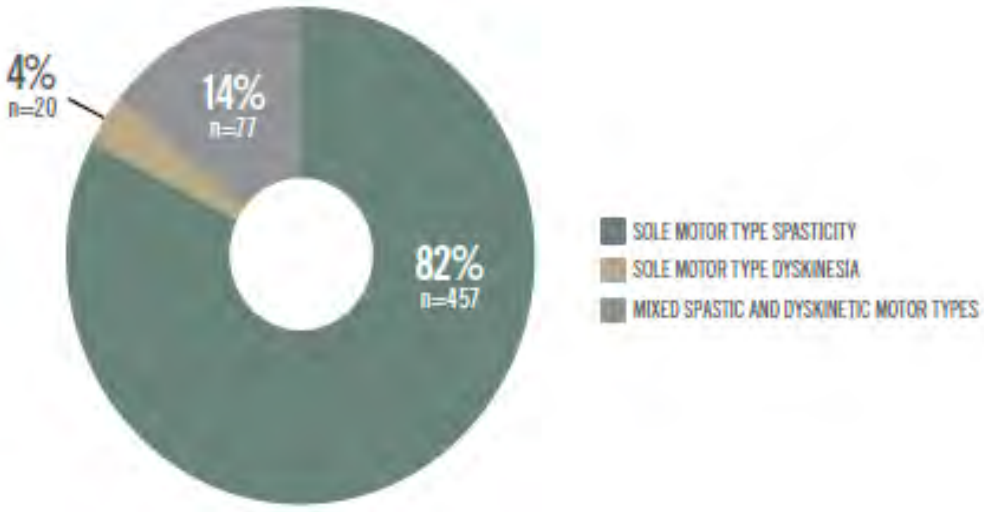
- Spastic (dyskinetic): classified based on topography
    - Quadriplegia
        - The most severe type of spastic CP
        - Lesions in the brain are large and often extend beyond just the periventricular areas
        - Spasticity is the predominant finding in the extremities, with hypotonia often being the predominant central finding
        - Often associated with associated medical and developmental conditions:
            - Seizures
            - Intellectual disability
            - Nonverbal
            - Difficulties feeding/swallowing
            - Breathing difficulties
            - Orthopedic concerns/contractures
    - Diplegia
        - Spastic findings are present in all four limbs, though the lower extremities exhibit substantially greater involvement than the upper extremities
        - Lesions in the brain are typically confined to the periventricular white matter, though the size of the lesion and the degree of the resultant impairment can vary
        - Associated with better function overall than spastic quadriplegia, and prognosis for independent ambulation (with or without assistive devices) is usually felt to be good if the child has the ability to sit by 2 years of age
        - Seizures and other accompanying medical or cognitive conditions are not nearly as common as in quadriplegia, and there is a generally good prognosis for intellectual development
    - Hemiplegia
        - Refers to the predominant findings of motor involvement on one side of the body, although minor abnormalities may be noted on the contralateral side as well

        - Represents about 1/3 cases of CP
        - Predominantly unilateral motor cortex involvement is responsible for the contralateral signs.
        - Usually the arm is somewhat more affected than the leg
            - Thus, we can see large delays in obtaining fine motor milestones than in obtaining gross motor milestones
            - Decorticate posturing of the affected arm is often seen
        - Most children can walk, but they can have gait asymmetries (internal rotation of the leg and toe walking on the affected side)
        - More commonly have seizures than in spastic diplegia
        - Cognition is usually not affected, but unilateral visual field deficits may be present, which can affect learning

5. Recognize the signs and symptoms of dyskinetic cerebral palsy

- Dyskinetic (extrapyramidal) CP: defined by involvement of areas outside of the motor pyramidal tracts
    - It may occur secondary to lesions in one of several central brain regions that serve to regulate muscle coordination and tone (e.g. the cerebellum, basal ganglia, thalamus, and other central structures)
    - Signs and symptoms:
        - Tone: variable due to fluctuating motor control. When increased tone is found, it tends to be more constant in quality (i.e., lead-pipe rigidity)
            - Hypertonia is markedly reduced in times of relaxation or sleep
            - Repeated passive movements may also decrease the degree of hypertonia for a brief period of time
        - Global problems with motor coordination: issues with moving all four extremities, the trunk, and the head are often all present
        - Speech and feeding are typically functionally limited
        - Involuntary movements: seen in the choreoathetoid form of dyskinetic CP
            - Chorea: quick and jerking movements
            - Athetosis: slow and writhing movements
            - Arms, trunk, and face are most commonly involved in these abnormal movements





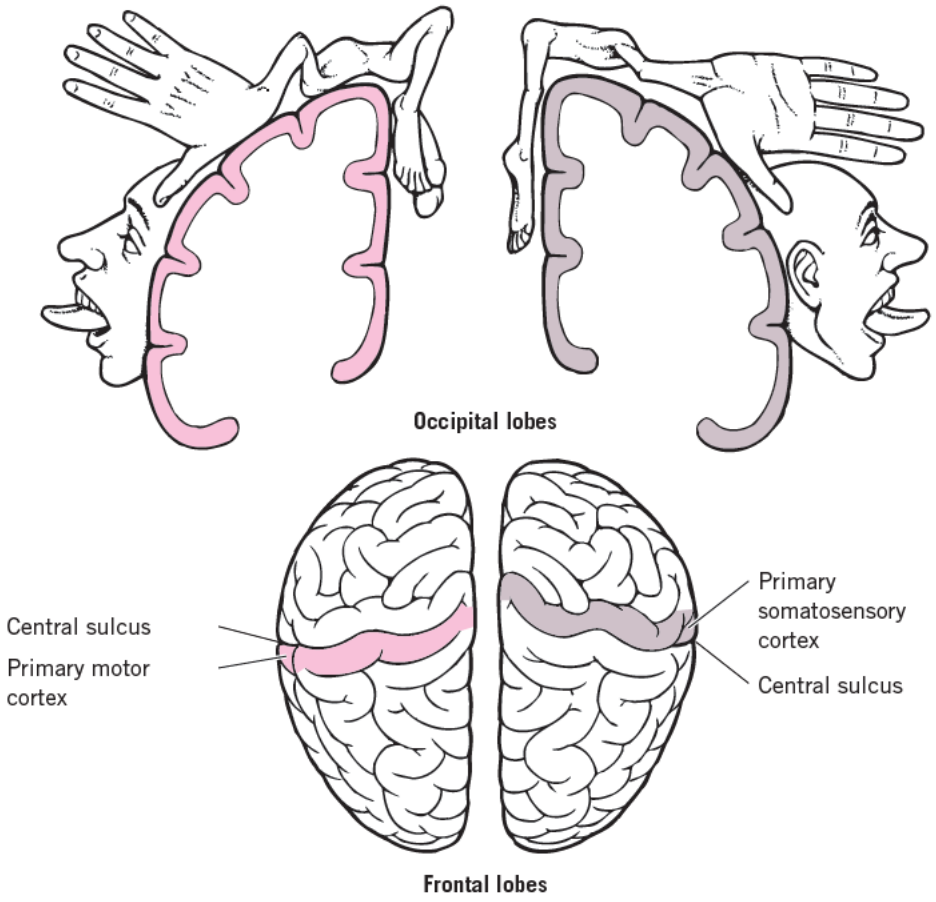
6. Know specific causes of the different types of cerebral palsy

- Spastic CP:
    - Quadriplegia:
        - Large lesion in the brain; can be due to a brain bleed (trauma, prematurity with hemorrhage, etc.), an anoxic event (asphyxia, etc.), or another cause
        - Think term infants with history of profound HIE and extreme premature infants with grade IV bilateral IVH with subsequent PVL
    - Diplegia:
        - Most often due to a more central brain bleed, also most often seen as intraventricular hemorrhage in the premature infant with resultant periventricular leukomalacia

        - Lower extremities usually most often affected than upper extremities
    - Hemiplegic:
        - Lesion in the brain (due to bleed, anoxia, etc.) as above, but typically unilateral in character
- Dyskinetic CP:
    - Lesion in the brain (due to bleed, anoxia, etc.) as above, but typically in one of several central brain regions that serve to regulate muscle coordination and tone (e.g. the cerebellum, basal ganglia, thalamus, and other central structures)
    - Choreoathetoid CP: historically caused by kernicterus, not seen as often anymore

- Remembering the motor homunculus and where damage to brain happens (and how) will help with remembering causes of each type of CP

Homunculus. The term *homunculus* means "little person." This drawing of a strange "little person" indicates where in the brain the different parts of the body are controlled and how much space is allocated to each part.



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7. Distinguish cerebral palsy from spinal cord injuries, peripheral motor disorders, and lower motor neuron lesions

- Cerebral Palsy:

    - See above. This is an upper motor neuron (UMN) disorder with various presentations, broadly either spastic in character (pyramidal symptoms) or dyskinetic in character (extrapyramidal symptoms). Etiologies as listed above, most commonly seen with prematurity and brain hemorrhage, but other causes (like anoxia or head trauma) exist as well.
- Spinal Cord Injuries:
    - Specific injury to part of the spinal cord can be caused by multiple etiologies such as trauma, a mass lesion like a neoplasm, a bleed in the spinal cord, or an ischemic event
    - Location of the injury in the spinal cord and the degree of the injury correspond with the nature of the neurologic deficit; tracts involved (motor or otherwise) are affected distal to the injury
        - Thus, lesions higher in the spinal cord affect the motor tracts higher, and thus will be seen with much more broad and significant motor involvement, whereas injuries lower in the spinal cord will not affect the motor tracts above the level of the lesion
- Peripheral Motor Disorders:
    - Quick Anatomy Review:
    - A motor unit consists of
        - An anterior horn cell
        - Its motor axon
        - The muscle fibers it innervates
        - The connection between them (neuromuscular junction)
- The anterior horn cells are located in the gray matter of the spinal cord and thus are technically part of the CNS. In contrast to the motor system, the cell bodies of the afferent sensory fibers lie outside the spinal cord, in dorsal root ganglia.
- Nerve fibers outside the spinal cord join to form anterior (ventral) motor nerve roots and posterior (dorsal) sensory nerve roots. The ventral and dorsal roots combine to form a spinal nerve. Thirty of the 31 pairs of spinal nerves have dorsal and ventral roots; C1 has no sensory root
- The term peripheral nerve refers to the part of a spinal nerve distal to the root and plexus.
    - Peripheral nerves are bundles of nerve fibers
    - Peripheral nerve disorders can result from damage to or dysfunction of the one of the following:
        - Cell body
        - Myelin sheath
        - Axons
        - Neuromuscular junction
    - Peripheral nerve damage is termed neuropathy
    - There are many different types of peripheral neuropathy, depending on the type(s) and amount of nerves involved
    - Etiology can vary; it can be due to nerve damage/injury, medications, systemic diseases like diabetes, vitamin deficiencies (namely vitamin B1, B12, copper; but also vitamin E deficiency and vitamin B6 deficiency in setting of TB treatment with isoniazid for instance), or other causes
    - There is often a sensory component (tingling or numbness are common)
    - Motor symptoms include weakness and impaired coordination

- Other specific examples are: diabetic neuropathy and certain genetic diseases like Friedreich's ataxia, Fabry disease, and Charcot-Marie-Tooth Disease

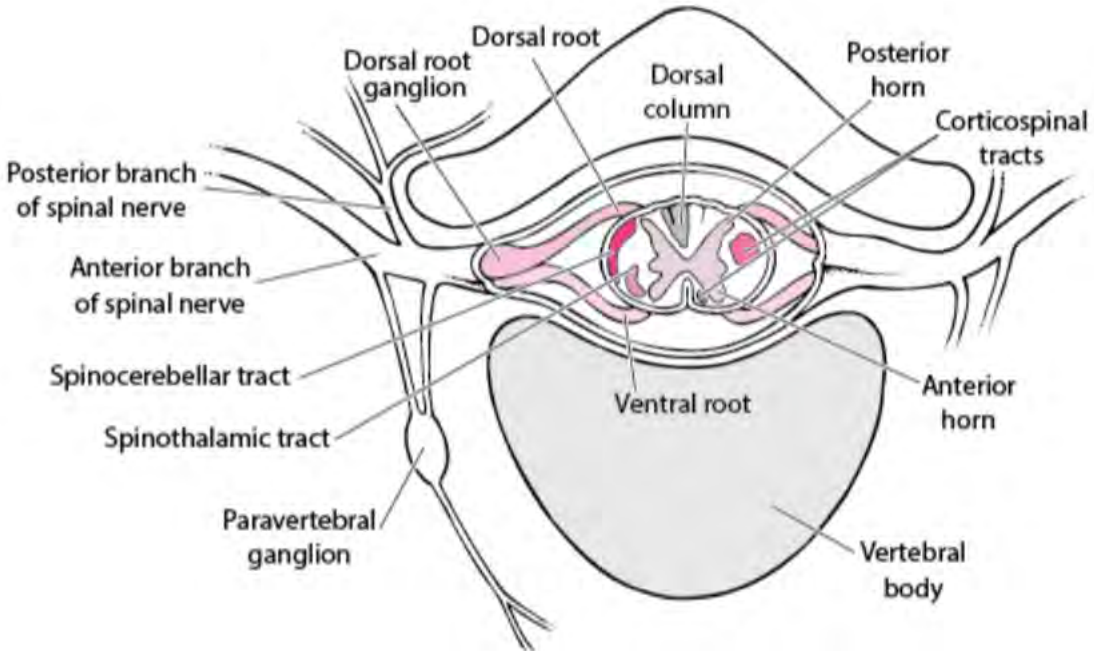


Figure outlining spinal nerves →peripheral nerve disorders involve diseases and disorders that involve anything distal to the root and plexus

Peripheral nerve	Entrapment	Carpal tunnel syndrome, cubital tunnel syndrome, radial nerve palsy, peroneal nerve palsy, tarsal tunnel syndrome
	Hereditary	Hereditary adult-onset neuropathies, hereditary sensory and motor neuropathies (eg, Charcot-Marie-Tooth disease), hereditary sensory and autonomic neuropathies
	Infectious	Hepatitis C, herpes zoster, HIV infection, Lyme disease, syphilis In developing nations: Diphtheria, leprosy, parasite infections
	Inflammatory	Chronic inflammatory demyelinating polyradiculoneuropathy, Guillain-Barré syndrome and variants
	Ischemic	Femoral nerve infarction (diabetic amyotrophy), vasculitis causing multiple mononeuropathy (mononeuritis multiplex)
	Toxic-metabolic	Amyloidosis, diabetes mellitus, dysproteinemic neuropathy, chronic excessive alcohol consumption with undernutrition (particularly deficiency of B vitamins), ICU neuropathy, leukodystrophies (rare), renal insufficiency, toxins (eg, arsenic, lead, mercury, thallium, chemotherapy drugs, pyridoxine toxicity)

- Lower Motor Neuron Lesions:
  - Lesions involving the lower motor neurons affect the nerves that travel from the anterior gray column of the spinal cord to the target peripheral muscles

- Differentiate from peripheral motor disorders in that lower motor neuron disorders (e.g., spinal muscular atrophies) technically involve the CNS because the cell body of the motor neuron (anterior horn cell) is located in the spinal cord. (see above figure)
- Identified by the key motor symptom of flaccid paralysis (paralysis accompanied by loss of muscle tone)
    - Other signs and symptoms: fibrillations, fasciculations, hyporeflexia, weakness/decreased muscle strength
- Examples of etiologies: trauma to peripheral nerves, viruses that lead to Guillain-Barre Syndrome, polio, botulism, ALS

8. Know how to plan the management of a toddler or preschooler with cerebral palsy.

- Care should involve a primary care provider who understands CP and who works with the family to understand the desired goals and outcomes for the child
    - Multi-disciplinary care is helpful given that multiple specialists can provide care for the many different medical problems that a child with CP may have
    - Management/care is dependent on multiple factors individual to a particular patient with CP:
        - Etiology – what caused it?
        - Pathophysiology – how can it affect global development?
        - Impairment – degree of loss of body function
        - Functional Limitation – is the ability to perform activities of daily living limited?
        - Disability – restriction to participate in desired and/or expected roles in society
        - Societal Limitations – what environmental or societal barriers are present?
    - Management should involve not only a multidisciplinary medical approach, but it should also involve the community (particularly the school)
        - School involvement is important because:
            - ID and learning disabilities are common
            - Dysarthria is common
            - Medical needs (such as needs related to ambulation, feeding, and toileting) are sometimes present, especially in young children
                - This includes involving personnel that are able to help with medical equipment
    - Other factors (such as specific medical problems like seizures, need for medication, drooling, and feeding issues) are unique to each child and must also be addressed

9. Know how to plan the management of a school-age child or adolescent with cerebral palsy

- This is similar to the management above for toddlers and preschoolers (please see #8)
- Efforts must be made to ensure that the child (especially an older child or adolescent) has his/her desires and concerns understood
- The school must continue to be involved to help children and adolescents with CP related to issues like learning difficulties, ambulation difficulties, toileting and feeding difficulties, etc.
- When children with CP are in a period of rapid growth, as in adolescence, providers must be cognizant of evolving equipment needs for these children
- Counseling families of older adolescents on the services available to them can help the adolescent with CP to transition effectively to college/adulthood, and achieve to the best of their potential

10. Evaluate early intervention and physical therapy in the management of cerebral palsy

- Early Intervention is crucial when children are diagnosed with CP prior to the age of 3

    - Parents should be encouraged to pursue an evaluation from Birth-to-Three services so that an IFSP can be developed and so that physical therapy (and other necessary therapies) can take place in the home.
- Physical Therapy:
    - PT (and OT) roles are broad in taking care of children with CP and their families
    - Provides direct treatment, participates in diagnostic evaluations, recommends braces and assistive devices, and provides training and support to children and caregivers
    - Goals during preschool years and subsequent years:
        - Improve strength, endurance, and speed
        - Gait training, particularly with new orthoses or devices
        - To assess when there is a change in motor skills or emerging skills such as independent walking
        - Postoperative services, when a child is removed from casts after surgery
        - Managing impending joint contractures
        - Services such as prescribing new braces or ambulatory aides
    - Evidence supporting PT/OT in CP is limited, however
        - Some studies have shown improvement in strength and functional capacity
        - Others have supported the use of constraint therapy in hemiplegia

11. Know the pharmacologic management of spasticity
- Children with marked spasticity and/or dystonia are likely to benefit from a combination of treatments (such as oral medications, IM injections of botox, intrathecal baclofen, nerve blocks, and selective dorsal rhizotomy)
- A major goal of early tone management is the prevention of orthopedic complications like contractures
- Oral medications for spasticity:
    - Baclofen and Benzodiazepines (GABA Agonists)
    - Dantrolene (Calcium Release Inhibitor)
    - Clonidine and Tizanidine (Alpha-2 Adrenergic Agonists)
    - Gabapentin (Increases GABA in the brain)
    - Side Effects of oral medications; sedation, drowsiness, and weakness
        - Side effects sometimes are severe enough to limit the modest benefits of oral medications
    - Evidence of oral medications is scarce and weak
    - Baclofen: abrupt withdrawal can result in serious side effects, including pruritus, increase in spasticity, confusion, hallucinations, and seizures
- Injected Phenol and Alcohol: limited use due to necessary technical expertise and risk for significant side effects
- Botox Injections: used for neuromuscular blockade
    - Easy to administer, low risk of side effects, rapid onset of action
    - Interferes with release of acetylcholine at the NMJ
    - Lasts up to 3 months after the initial injection
    - Limited by the 3 month duration, and by the fact that a limited number of muscles can receive injections at one time
    - Studies have shown significant reduction in spasticity and functional improvement in both the upper and lower extremities
- Intrathecal Baclofen: GABA agonist with site of action in the spinal cord

- Given by a continuous-infusion pump because the effects of a single dose last only a few hours
- Reviews have shown that IT baclofen decreases tone in the upper and lower extremities, improves function in the upper and lower extremities, improves ease of care and sleep, decreases pain, and decreases truncal tone
- Benefits seen in spastic and dystonic CP
- Despite prior concerns, studies have not shown any risk for IT baclofen causing an increased risk of seizures or hip subluxation
- Withdrawal as above for oral baclofen; can be life-threatening

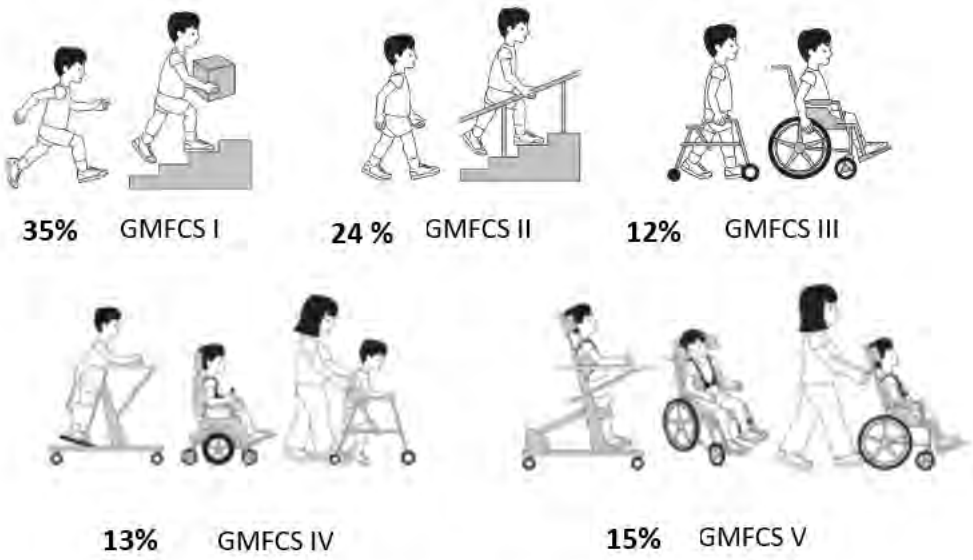
12. Know the natural history of cerebral palsy
    - As noted above, CP can be diagnosed at various ages and has multiple possible etiologies and presentations/classifications
    - Different classifications of CP are associated with different natural courses
        - Spastic quadriplegia:
            - Often diagnosed early with significant motor/movement differences and abnormal findings on neurological exam
            - Hypotonia may be the predominant initial finding
            - Spasticity progresses to be manifest as hypertonia in the extremities, with hypotonia being seen in the trunk
            - Spasticity in all 4 limbs can be severe
            - Children can develop significant complications as they continue to grow, and they may need pharmacologic, medical, or surgical management for these issues
                - Contractures, scoliosis, and other orthopedic problems
                - Feeding difficulties
                - Cognitive impairment and speech difficulties
                - Seizures more common than in other types of CP
            - As children grow, they likely will continue to require significant support and prospects for independence are limited
                - They will likely need lifelong assistance with ambulation, feeding, toileting, etc.
        - Spastic Diplegia:
            - Often diagnosed early in childhood, but this can vary depending on the severity
            - Can frequently be diagnosed after delay in motor milestones (for instance, significant delay in independent sitting or walking), and not necessarily with significant deficits or neurological findings in infancy (which can be seen in spastic quadriplegia)
            - More likely than quadriplegia to have normal cognition (or at least less cognitive impairment)
            - Less risk for seizures, orthopedic problems, problems with ambulation, feeding, drooling, etc.
            - Still: will likely need physical therapy and also will likely benefit from assistive devices for ambulation and other tasks (especially when the child is young)
        - Dyskinetic CP:
            - Since this appears with variable (and sometimes significant) brain lesions, it often occurs as a mixed CP with features also consistent with spastic quadriplegia
            - Tone is variable, making identifying it and managing sometimes difficult
            - Natural course (such as need for therapies and more significant interventions) is very similar to spastic quadriplegia, as above.

- As with spastic quadriplegia, capacity for independent function is very limited
- Involuntary movements are also present, however, and this can make management more difficult

o Functional classifications are very helpful for clinicians both for describing severity of impairment and for prognosticating about future needs. Two motor classifications for both gross motor function and manual ability are now widely used- Gross Motor Function Classification Scale (GMFCS) and Manual Ability Classification System (MACS)/Mini-MACS (<4 years old)

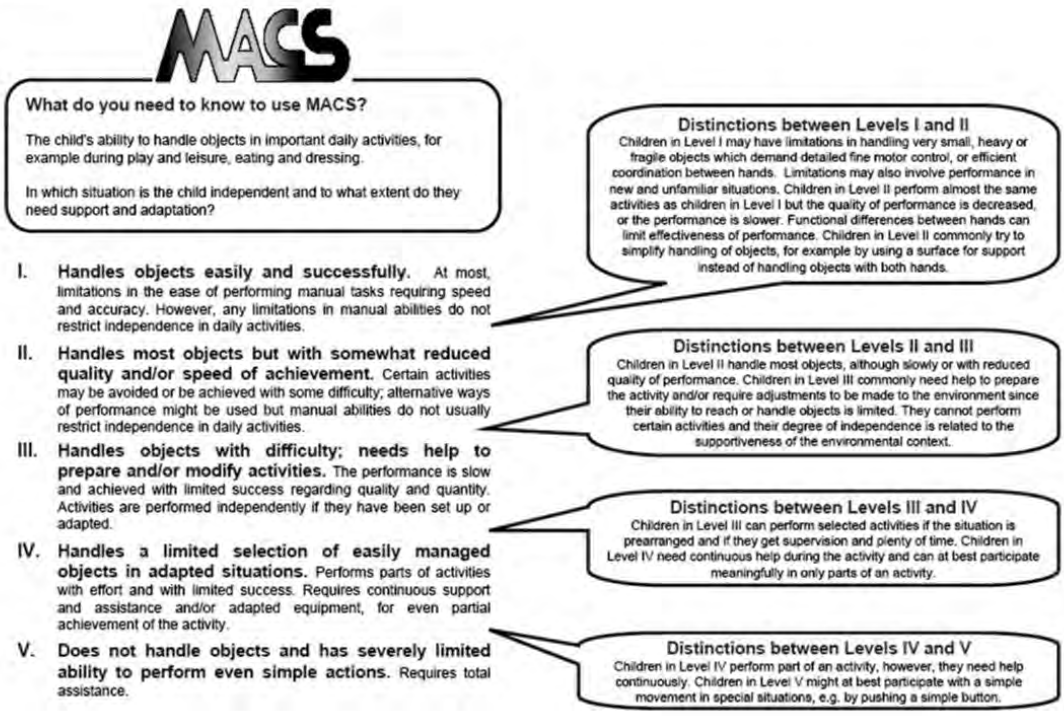
The Gross Motor Function Classification Scale (GMFCS), considered to be the "contemporary gold standard" of CP classification, is a 5 level classification describing a person's functional mobility[11,13]. Register data shows that the great majority of people with CP are ambulant.

Figure 3 demonstrates the 5 levels of GMFCS and percentage of people at each level according to ACPR.



GMFCS Illustrations 6-12: © Bill Reid, Kate Willoughby, Adrienne Harvey and Kerr Graham, The Royal Children's Hospital Melbourne.

Appendix I: The Manual Ability Classfication System (MACS) leaflet (which can be downloaded in several languages from www.macs.nu)



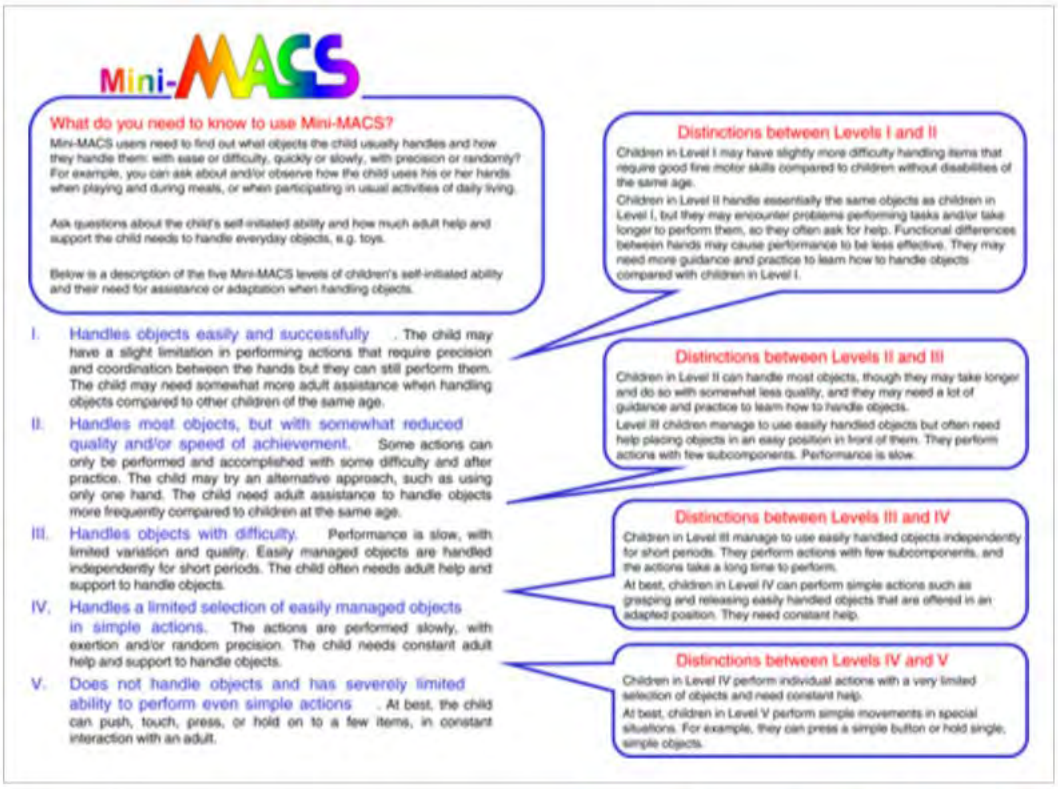


Figure 1 Open in figure viewer | PowerPoint

The Mini-Manual Ability Classification System (Mini-MACS) classification. [Colour figure can be viewed at wileyonlinelibrary.com].

13. Know the management of drooling in children with cerebral palsy
    - Drooling occurs in 10-20% of the patients with CP.
    - It's a consequence of poor oral-motor control and/or swallowing abnormalities
    - Sometimes can be a severe social problem
    - Management:
        - Speech therapy directed to improve swallowing
        - Anticholinergic Medications:
            - Glycopyrrolate (0.01 to 0.82 mg/kg/day)
        - Surgery to reduce saliva production can be an option for those who fail the above therapy approaches
            - Submandibular duct relocation
            - Sublingual gland excision
            - Bilateral parotid duct ligation
            - Surgeries can be isolated or in combination
            - Risk of these surgeries: produce excessive reduction of saliva, which can increase feeding problems

14. Understand the neurological, orthopedic and/or ophthalmological complications associated with cerebral palsy

- Neurological: Tone and movement are discussed above.
    - Seizures: frequent complication of CP, especially quadriplegia
        - Those with ID have a higher risk of seizures
        - Spastic Diplegia: 16-27% have seizures
        - Spastic Quadriplegia: 50-94% have seizures
        - Seizures in hemiplegia correlates with a poor cognitive outcome
        - In Spastic Diplegia, patients with seizures are more likely to respond to AEDs; in spastic quadriplegia, patients will likely need multiple AEDs
    - Hearing: 10% of people with CP will have hearing loss
    - Urinary Incontinence: not always neurologic in origin, but it is a common complications
        - CP patients with quadriplegia and/or ID: by 8 years old, only 40-55% have urinary continence
- Orthopedic:
    - Hip subluxation and dislocation: adults with CP and untreated hip dislocation can have chronic hip pain
        - Monitor hips with plain radiographs starting at about 18 months of age
            - Migration percentage (percentage of femoral head that is uncovered by the acetabulum) is the principal measure of hip stability
            - Migration percentage >/=40% at the time of surgery is a risk for progression of the migration
    - Scoliosis: spinal deformity is a common problem in children with quadriplegia
        - A spinal curve of 40% is likely to worsen and necessitate surgical stabilization
- Ophthalmologic:
    - Visual acuity deficits are very frequent in children with CP
        - Children with hemiplegia often have visual field restriction
        - Children with dyskinetic CP can have abnormal control of eye movements and may be wrongly assumed to be blind
        - Children with CP due to IVH can have visual problems including blindness
        - Children with CP should have early ophthalmologic evaluation and frequent follow-up

15. Know the developmental and behavioral characteristics of individuals with cerebral palsy

- As above, ID and learning disabilities present at varying rates in the different types of CP
- Also, speech and language delays present, especially when there is oral-motor involvement
- Other developmental issues:
    - Impaired written communication
    - Visual motor and visual perceptual dysfunction
    - ADHD
    - Autism
    - Low self-esteem, social isolation, depression
- Behavioral issues:
    - Behaviors can be hard to interpret, especially automatic motor responses like truncal hyperextension
    - Children with cognitive deficits can behave as if they were a younger chronological age

- Excessive or aggressive behaviors can be a sign of a medical problem, and in such cases it is important to rule out medical conditions that could produce pain (e.g. sinusitis, GE reflux, headaches, toothaches, joint pains, etc.)

16. Know the range of prognoses for children with different types of cerebral palsy
    - Prognosis can vary depending on the type of CP; see #12 above about natural history for details on prognosis
17. Understand the classification of cerebral palsy
    - Please see #4-5 above
18. Characterize degree of impairment in cerebral palsy using the Gross Motor Function Classification System
    - GMFCS: (see above chart under #12)
        - Level 1: Walks without restrictions; limitations in advanced skills only
        - Level 2: Walks without assistive devices; limited outdoors/community mobility
        - Level 3: Walks with assistive mobility devices: limited outdoors/community mobility
        - Level 4: Self-mobility with limitations: transported in wheelchair or use power mobility in outdoors/community
        - Level 5: Self-mobility is severely limited, even with the use of assistive technology
    - The original classification was developed for children 6-12 years, and an expanded and revised version was later developed and includes ages 2-18
    - Since the GMFCS is known to be unstable under 2 years, with at least 40% changing GMFCS level after 2 years, caution should be taken before applying a classification level at such an early age (Gorter, 2009).
    - GMFCS classification is based on the child's ability to sit, transfer, and mobilize
    - Motor development curves for CP that are useful for predicting GMFCS level are widely used as a tool to discuss prognosis with families and plan realistic rehabilitation goals
        - The study that produced these curves demonstrated that children with CP achieve 90% of their gross motor development potential by age 5 across all GMFCS levels, after which a plateau effect occurs
        - Furthermore, children at GMFCS level V reach this point before 3 years of age.
        - It is important to note that, based on GMFCS prevalence rates, mild CP is more common than severe CP.
    - When comparing the MACS and GMFCS for use in counseling parents/families on prognosis there can be discrepancies in Gross motor function and manual ability in children CP and the patterns seem to vary across the different subgroups based on the predominant neurological findings
        - For this reason, use of both measures is recommended to give a complete clinical picture for counseling families
19. Recognize the signs and symptoms of ataxic cerebral palsy
    - Ataxic CP: results from disturbances in regulation of motor coordination (like dyskinetic CP)
        - Typically manifested by tremors associated with intentional movements and/or gait abnormalities
            - Wide-based, sometimes staggering, gait often develops
20. Understand the role of neuroimaging in the assessment of cerebral palsy
    - Brain imaging is widely used to document the presence and extent of brain structural integrity in CP
    - A number of systematic reviews have demonstrated that certain defined patterns of injury in the grey and or white matter nearly always lead to CP, indicating the importance of appropriately timed neuroimaging in the diagnostic process
    - There is also considerable evidence that CP can be predicted with neuroimaging in the newborn period, provided the optimal sequences and timing are used (Massaro, 2015)

- Evidence based recommendations for timing and use of preferred imaging modalities are available for preterm infants and those with neonatal encephalopathy (BAPM, 2016); however, recommendations for infants not considered high risk are more complicated.
- The American Academy of Pediatrics suggests MRI be carried out in the case of increased tone,[4] and the American Academy of Neurology (AAN) and the Child Neurology Society (CNS) have produced an evaluation parameter also recommending imaging be conducted during the diagnostic process.

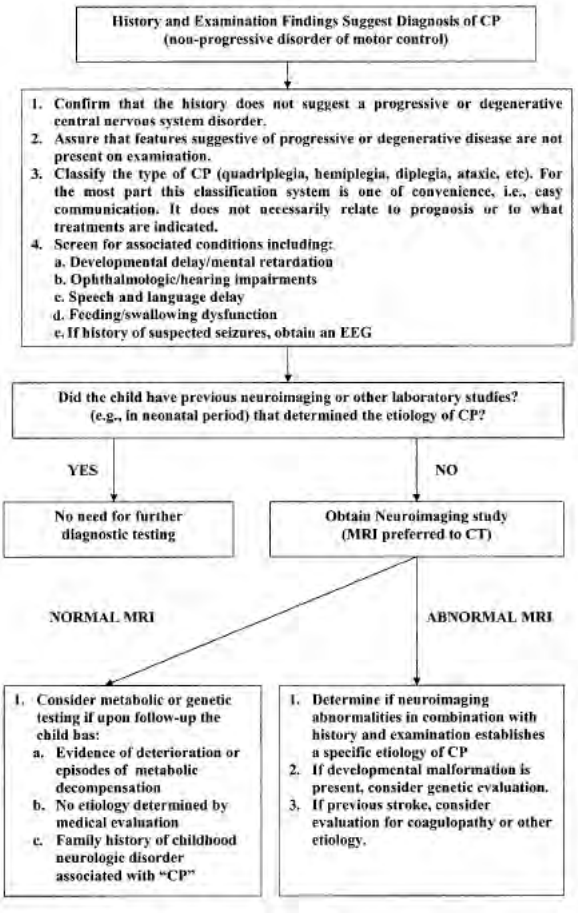


Figure. Algorithm for the evaluation of the child with cerebral palsy (CP). Screening for associated conditions (mental retardation, vision/hearing impairments, speech and language delays, oral motor dysfunction, and epilepsy) is recommended. Neuroimaging (MRI preferred to CT) is recommended for further evaluation if the etiology of the child's CP has not been previously determined. In some children, additional metabolic or genetic testing may be indicated.

- However, clinical research also documents outcomes that do not seem to "match" imaging findings. It is estimated up to 15% of children with CP have normal neuroimaging

- Several studies have shown that serial ultrasonography and cranial MRI are both sensitive and specific in the detection of CP

- MRI exams are particularly sensitive and specific in the examination of preterm infants with very low and extremely low birth weight
    - Also helpful in diagnostic evaluation for full term infants with history of HIE
    - Abnormalities of the thalamus and basal ganglia are highly predictive of subsequent neurodevelopmental problems, including CP
    - MRI can help evaluate more diffuse PVL and brain injury better than ultrasound and can also evaluate the white matter pathways better

- Utility of MRI in preterm infants is evolving: MRI at 36-40 weeks post conceptual age may be a valuable addition to cranial ultrasound in detecting PVL in the highest risk cohort of preterm infants→still important to remember that there will still be a small percentage of missed cases in this method of diagnosis

- Cranial ultrasonography:
  - Findings that are predictive of subsequent CP:
    - Persistent ventricular dilatation
    - Cystic PVL
    - Grades III and IV IVH
  - Child Neurological Society practice parameter recommends cranial ultrasonography at 7 to 14 days and again at 36-40 weeks post conceptual age
- Brain MRI:
- It should also be noted that although understanding of the etiology of CP is growing, methods of CP detection in high-risk populations still need much improvement, as there is currently no imaging strategy with 95% sensitivity and specificity as well as accurately predicting the severity of disability
- Best clinical practice, therefore, supports the combined use of neuroimaging, standard motor assessment, and neurological examination to provide the best information for early and accurate diagnosis of CP. [18](Novak Jama PEDES 2017)
- In infants with risks detectable in newborns, a diagnosis of CP can most often be made early using a combination of this sort

B. Myelodysplasia

1. Know the high prevalence of hydrocephalus and Chiari malformation in children with myelodysplasia.
   - Myelodysplasia is simply defined as any developmental anomaly of the spinal cord
   - Hydrocephalus and Chiari malformation (usually type II, but can see type IV) well connected specifically with myelomeningocele
   - **Hydrocephalus** is an excessive buildup of CSF in the brain—a Chiari malformation can block the normal flow of this fluid and subsequently lead to an obstructive or non-communicating hydrocephalus→can occur with any type of Chiari malformation, but is most commonly associated with Type II;
     - Estimated that hydrocephalus occurs in myelomeningoceles in up to 85% of cases
     - the risk of developing hydrocephalus requiring shunting is lower among infants with sacral lesions, as compared with those with a higher vertebral level of involvement
     - hydrocephalus usually develops secondary to impaction of the posterior fossa contents on the foramen magnum, leading to occlusion of the outlets of the fourth ventricle with cerebrospinal fluid (CSF) outflow blocked, or impaired, at the foramina of Luschka and Magendie and resulting in progressive ventriculomegaly
     - Moreover, vertical translocation of the brain stem causes increased resistance of CSF flow through the tentorial hiatus
     - The small volume of the posterior fossa, in conjunction with the very abnormal tilt of the tentorium, and cerebellar prolapse causes increased

tension inside the posterior fossa, which leads to increased resistance of the venous outflow through the sigmoid sinus and venous hypertension→This may create an element of communicating hydrocephalus along with obstructive component

- **Chiari malformations classified as follows, but most common ones are Chiari malformation Types I and II (table and figures below illustrating difference)**
  - **Chiari malformation Type I**
    Type 1 happens when the lower part of the cerebellum (called the cerebellar tonsils) extends into the foramen magnum. Normally, only the spinal cord passes through this opening. Type 1—which may not cause symptoms—is the most common form of CM. It is usually first noticed in adolescence or adulthood, often by accident during an examination for another condition. Adolescents and adults who have CM but no symptoms initially may develop signs of the disorder later in life.
  - **Chiari malformation Type II**
    Individuals with Type II have symptoms that are generally more severe than in Type 1 and usually appear during childhood. This disorder can cause life-threatening complications during infancy or early childhood, and treating it requires surgery.
    - In Type II, also called classic CM, both the cerebellum and brain stem tissue protrude into the foramen magnum. Also the nerve tissue that connects the two halves of the cerebellum may be missing or only partially formed
    - Type II is usually accompanied by a myelomeningocele—a form of spina bifida that occurs when the spinal canal and backbone do not close before birth→A myelomeningocele usually results in partial or complete paralysis of the area below the spinal opening.
    - The term Arnold-Chiari malformation specific to Type II malformations
    - Multiple ventricular anomalies are commonly found in CM II patients
    - The fourth ventricle, is typically small and poorly visualized, and is frequently displaced into the cervical canal along with its choroid plexus
    - The aqueduct is similarly small and rarely seen on routine imaging
    - Third ventricle is rarely enlarged but may take a narrow-angled appearance, giving rise to the term “shark tooth deformity”
    - The lateral ventricle varies from nearly normal to being severely deformed and hydrocephalic
    - Colpocephaly is common with the occipital horns disproportionately enlarged compared with the frontal horns
    - This finding is often present even in patients with MMC who do not have hydrocephalus and frequently persists in patients in whom a shunt has been placed

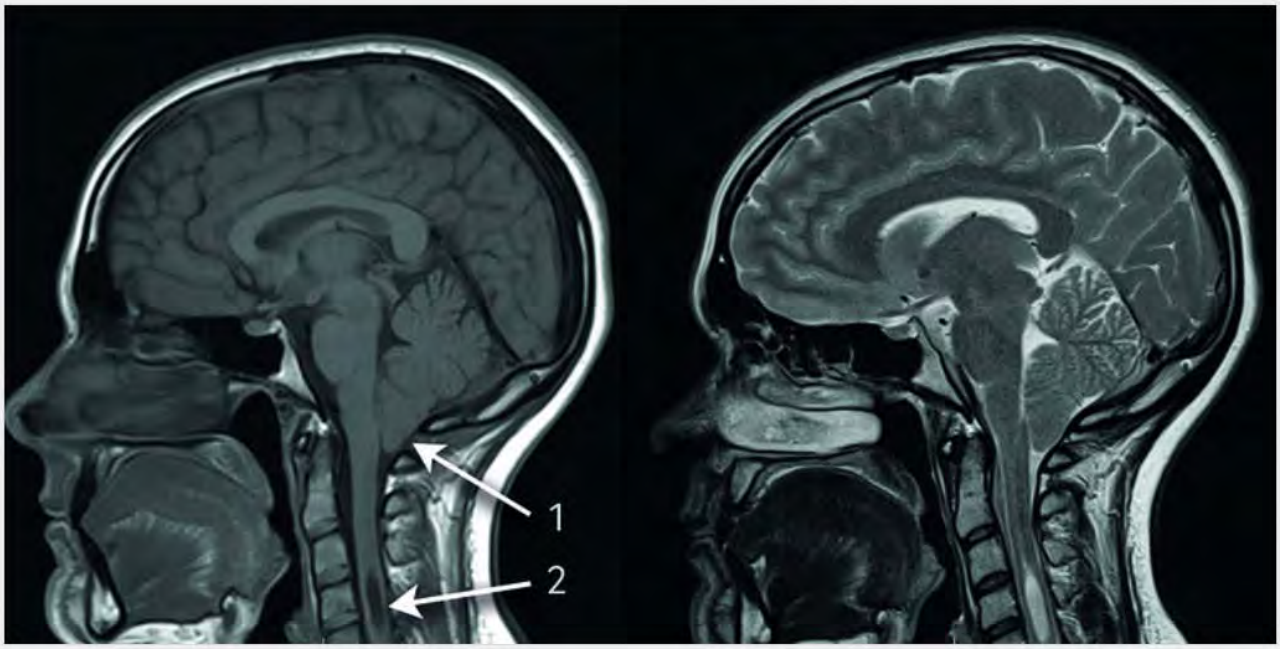


Fig 2

Sagittal MRI head images of a patient with Chiari 1 malformation, with T1-weighted (left) and T2-weighted (right) sequences. Arrows show (1) the cerebellar herniation and (2) the cervical cord syringomyelia

Table 1 Comparison between the Chiari 1 and Chiari 2 malformations

	Chiari 1 malformation	Chiari 2 malformation
Hindbrain structures herniating caudally	Cerebellar tonsils	Cerebellar tonsils and vermis, medulla and fourth ventricle
Hydrocephalus	Uncommon	Very common
Age of presentation	Childhood or early adulthood	Infancy or childhood
Association with spinal dysraphism (myelomeningocele)	Uncommon	Almost always

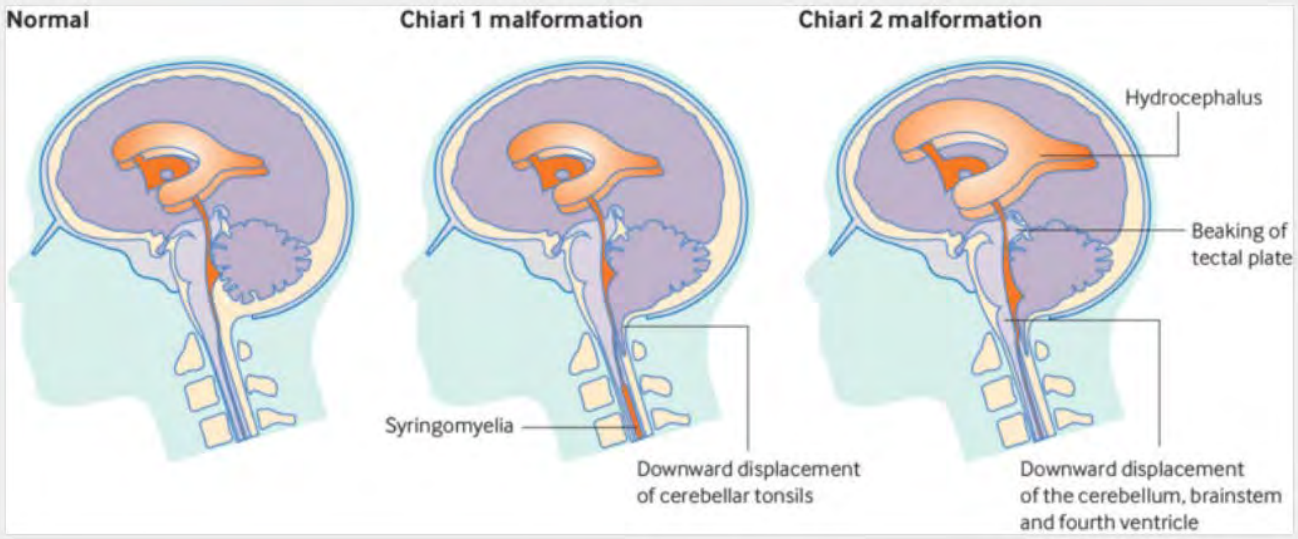


Fig 1

Key anatomical features of the Chiari 1 and Chiari 2 malformations

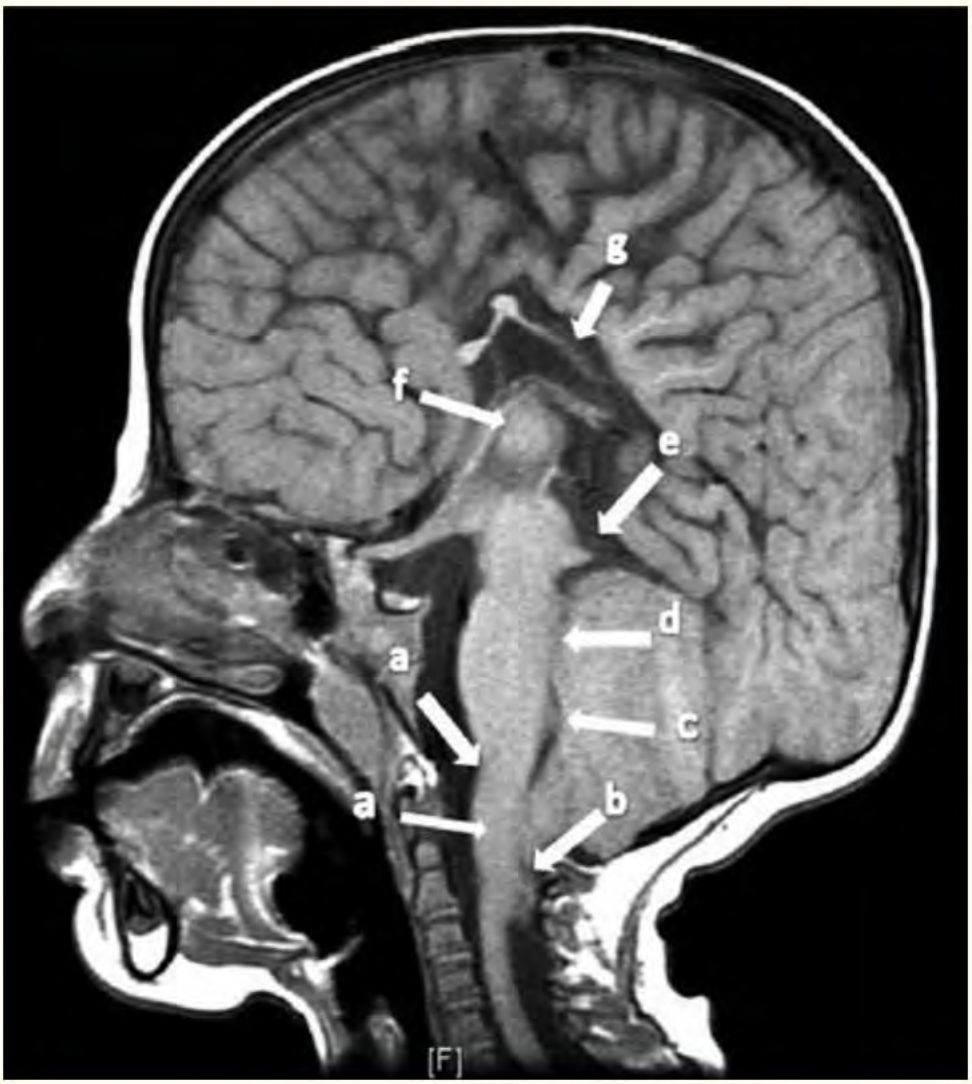


Figure 1

Sagittal T1-weighted image showing Chiari II malformation; the features include Elongated brainstem that extends into the cervical spinal canal; (a). Downward herniation of the cerebellar tonsils into cervical spinal canal; (b). Small fourth ventricle; (c). Aqueductal stenosios; (d). Tectal beaking; (e). Large massa intermedia; (f). There is also thin corpus callosum (g)

- **Chiari malformation Type III**
  - Type III is very rare and the most serious form of Chiari malformation. In Type III, some of the cerebellum and the brain stem stick out, or herniate, through an abnormal opening in the back of the skull. This can also include the membranes surrounding the brain or spinal cord.
  - The symptoms of Type III appear in infancy and can cause debilitating and life-threatening complications. Babies with Type III can have many of the same symptoms as those with Type II but can also have

additional severe neurological defects such as mental and physical delays, and seizures.

- **Chiari malformation Type IV**
  - Type IV involves an incomplete or underdeveloped cerebellum (a condition known as cerebellar hypoplasia). In this rare form of CM, the cerebellum is located in its normal position but parts of it are missing, and portions of the skull and spinal cord may be visible
- **Chiari malformation Type 0**
  - Researchers have determined that some individuals with a Chiari malformation have minimal or no herniation of the cerebellar tonsils through the foramen magnum
  - these individuals often have syringomyelia despite the lack of cerebellar tonsil herniation.
  - Occipital headaches may also occur. Symptoms in these cases are most likely due to abnormalities in the flow of cerebrospinal fluid at the level of the foramen magnum at the skull base, although there is often no identifiable cause. Individuals with this condition have improved after decompression surgery. The addition of Chiari malformation type 0 as a classification for Chiari malformations is controversial; some physicians believe that, for a diagnosis of a Chiari malformation, tonsillar herniation must be present

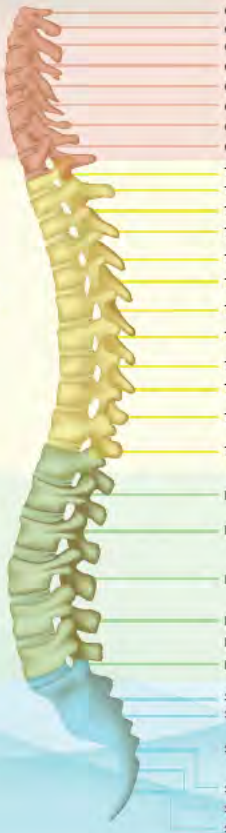
2. Understand the relationship between the level of myelodysplasia and motor, cognitive, and adaptive dysfunction.

- The effects of myelodysplasia relate to the location and size of the defect and the presence of hydrocephalus, brain abnormalities such as the Chiari II malformation, and other neurologic and orthopedic conditions
- Thoracic and higher lumbar myelomeningocele lesions are more likely to be associated with significant motor and sensory deficits and structural abnormalities in the lower extremities than are those in the lower lumber or sacral regions
- Functional defects of the urogenital and lower intestinal tract are likely to occur at all levels
- Likewise, the presence of Chiari II malformation, hydrocephalus, and other structural abnormalities of the brain (are also more commonly seen in thoracic and higher lumbar myelomeningocele lesions→ this would also predict greater concerns then for cognitive impairments and increase of seizure disorders (which would also impact cognitive functioning possibly)
- Adaptive functioning would parallel with cognitive abilities, but also be strongly impacted by ambulation and fine motor dexterity abilities with motor and sensory function (see below from Spina Bifida Association)

SBA National Resource Center:
800-621-3141

SPINA BIFIDA ASSOCIATION

Level of SB Function



CERVICAL REGION

- **C1**
- **C2**
- **C3**
- **C4**
- **C5** Elbow flexors: Partial upper extremity function
- **C6** Wrist extensors: Standing with stander/orthotics
- **C7** Elbow extensors
- **C8** Finger flexors

THORACIC REGION

- **T1**
- **T2** Complete upper extremity function
- **T3–T8** Standing with stander/orthotics
- **T4** Possible exercise ambulation
- **T5**
- **T6**
- **T7** Partial function of trunk muscles
- **T8**
- **T9–T12** Exercise ambulation
- **T10–L2** Bladder: Sympathetic input from hypogastric nerve
- **T11** Some function of trunk muscles
- **T12–S5** Sexual function varies

LUMBAR REGION

- **L1** Complete trunk function: exercise ambulation, sometimes household ambulation
- **L2** Hip flexor muscles present: exercise ambulation, household ambulation
- **L3** Knee extensors or Quadriceps muscles present: household ambulation, possible community ambulation
- **L4** Medial knee flexors present. Ankle dorsiflexors, 3/5 strength
- **L4–S5** Community ambulation
- **L5** May walk with or without crutches in home

SACRAL REGION

- **S1** Hip abductors, 3/5 strength
- **S2** Hip extensors, 4/5 strength. Ankle, plantarflexors, 3/5 strength. May walk with or without crutches
- **S2-S4** Bowel and bladder function varies. Bladder: parasympathetic input from the pelvic nerve. Somatic input from pudendal nerve to urethral sphincter
- **S3** All muscle activity may be within normal limits
- **S4**
- **S5 and above** Be aware of signs and symptoms of shunt malfunction and tethered spinal cord

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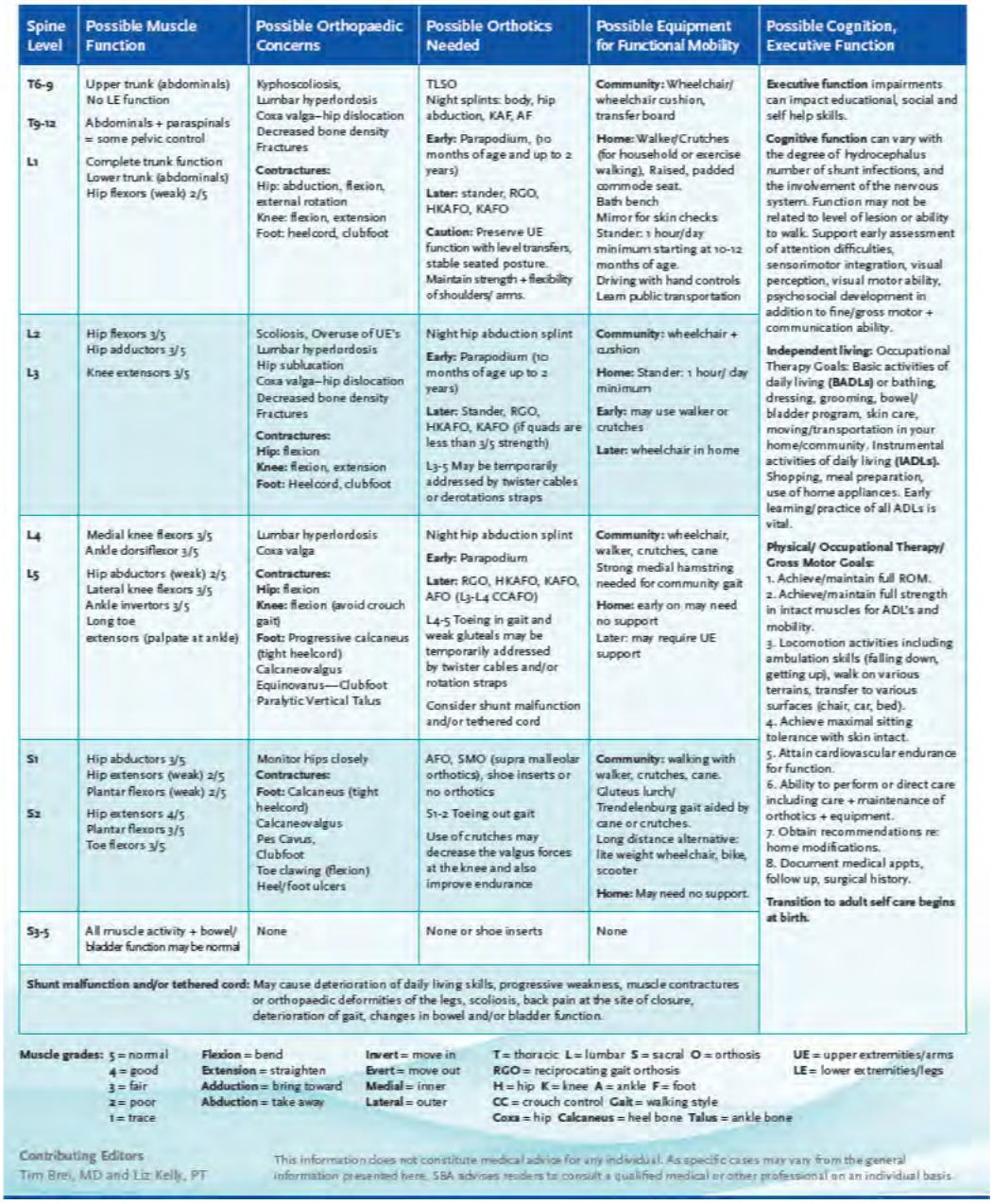


Spine Level	Possible Muscle Function	Possible Orthopaedic Concerns	Possible Orthotics Needed	Possible Equipment for Functional Mobility	Possible Cognition, Executive Function
T6-9 T9-12 L1	Upper trunk (abdominals) No LE function Abdominals + paraspinals = some pelvic control Complete trunk function Lower trunk (abdominals) Hip flexors (weak) 2/5	Kyphoscoliosis, Lumbar hyperlordosis Coxa valga–hip dislocation Decreased bone density Fractures **Contractures:** Hip: abduction, flexion, external rotation Knee: flexion, extension Foot: heelcord, clubfoot	TLSO Night splints: body, hip abduction, KAF, AF **Early:** Parapodium, (10 months of age and up to 2 years) **Later:** stander, RGO, HKAFO, KAFO **Caution:** Preserve UE function with level transfers, stable seated posture. Maintain strength + flexibility of shoulders/ arms.	**Community:** Wheelchair/ wheelchair cushion, transfer board **Home:** Walker/Crutches (for household or exercise walking), Raised, padded commode seat. Bath bench Mirror for skin checks Stander: 1 hour/day minimum starting at 10-12 months of age. Driving with hand controls Learn public transportation	**Executive function** impairments can impact educational, social and self help skills. **Cognitive function** can vary with the degree of hydrocephalus number of shunt infections, and the involvement of the nervous system. Function may not be related to level of lesion or ability to walk. Support early assessment of attention difficulties, sensorimotor integration, visual perception, visual motor ability, psychosocial development in addition to fine/gross motor + communication ability. **Independent living:** Occupational Therapy Goals: Basic activities of daily living **(BADLs)** or bathing, dressing, grooming, bowel/ bladder program, skin care, moving/transportation in your home/community. Instrumental activities of daily living **(IADLs).** Shopping, meal preparation, use of home appliances. Early learning/practice of all ADLs is vital. **Physical/ Occupational Therapy/ Gross Motor Goals:** 1. Achieve/maintain full ROM. 2. Achieve/maintain full strength in intact muscles for ADL's and mobility. 3. Locomotion activities including ambulation skills (falling down, getting up), walk on various terrains, transfer to various surfaces (chair, car, bed). 4. Achieve maximal sitting tolerance with skin intact. 5. Attain cardiovascular endurance for function. 6. Ability to perform or direct care including care + maintenance of orthotics + equipment. 7. Obtain recommendations re: home modifications. 8. Document medical appts, follow up, surgical history. **Transition to adult self care begins at birth.**
L2 L3	Hip flexors 3/5 Hip adductors 3/5 Knee extensors 3/5	Scoliosis, Overuse of UE's Lumbar hyperlordosis Hip subluxation Coxa valga–hip dislocation Decreased bone density Fractures **Contractures:** **Hip:** flexion **Knee:** flexion, extension **Foot:** Heelcord, clubfoot	Night hip abduction splint **Early:** Parapodium (10 months of age up to 2 years) **Later:** Stander, RGO, HKAFO, KAFO (if quads are less than 3/5 strength) L3-5 May be temporarily addressed by twister cables or derotations straps	**Community:** wheelchair + cushion **Home:** Stander: 1 hour/ day minimum **Early:** may use walker or crutches **Later:** wheelchair in home	
L4 L5	Medial knee flexors 3/5 Ankle dorsiflexor 3/5 Hip abductors (weak) 2/5 Lateral knee flexors 3/5 Ankle invertors 3/5 Long toe extensors (palpate at ankle)	Lumbar hyperlordosis Coxa valga **Contractures:** **Hip:** flexion **Knee:** flexion (avoid crouch gait) **Foot:** Progressive calcaneus (tight heelcord) Calcaneovalgus Equinovarus—Clubfoot Paralytic Vertical Talus	Night hip abduction splint **Early:** Parapodium **Later:** RGO, HKAFO, KAFO, AFO (L3-L4 CCAFO) L4-5 Toeing in gait and weak gluteals may be temporarily addressed by twister cables and/or rotation straps Consider shunt malfunction and/or tethered cord	**Community:** wheelchair, walker, crutches, cane Strong medial hamstring needed for community gait **Home:** early on may need no support Later: may require UE support	
S1 S2	Hip abductors 3/5 Hip extensors (weak) 2/5 Plantar flexors (weak) 2/5 Hip extensors 4/5 Plantar flexors 3/5 Toe flexors 3/5	Monitor hips closely **Contractures:** **Foot:** Calcaneus (tight heelcord) Calcaneovalgus Pes Cavus, Clubfoot Toe clawing (flexion) Heel/foot ulcers	AFO, SMO (supra malleolar orthotics), shoe inserts or no orthotics S1-2 Toeing out gait Use of crutches may decrease the valgus forces at the knee and also improve endurance	**Community:** walking with walker, crutches, cane. Gluteus lurch/ Trendelenburg gait aided by cane or crutches. Long distance alternative: lite weight wheelchair, bike, scooter **Home:** May need no support.	
S3-5	All muscle activity + bowel/ bladder function may be normal	None	None or shoe inserts	None	
Shunt malfunction and/or tethered cord: May cause deterioration of daily living skills, progressive weakness, muscle contractures or orthopaedic deformities of the legs, scoliosis, back pain at the site of closure, deterioration of gait, changes in bowel and/or bladder function.					

Muscle grades: 5 = normal, 4 = good, 3 = fair, 2 = poor, 1 = trace

Flexion = bend, **Extension** = straighten, **Adduction** = bring toward, **Abduction** = take away

Invert = move in, **Evert** = move out, **Medial** = inner, **Lateral** = outer

T = thoracic **L** = lumbar **S** = sacral **O** = orthosis
RGO = reciprocating gait orthosis
H = hip **K** = knee **A** = ankle **F** = foot
CC = crouch control **Gait** = walking style
Coxa = hip **Calcaneus** = heel bone **Talus** = ankle bone

UE = upper extremities/arms
LE = lower extremities/legs

Contributing Editors
Tim Brei, MD and Liz Kelly, PT

This information does not constitute medical advice for any individual. As specific cases may vary from the general information presented here, SBA advises readers to consult a qualified medical or other professional on an individual basis.

3. Know how to plan the management of children with different levels of myelodysplasia.

- The optimal care of the infant, child, or youth with spina bifida is best provided by a multispecialty team and a primary care provider who collaborate to provide comprehensive and coordinated care and support to the child and family
- The spina bifida team typically includes a clinical nurse specialist or nurse practitioner; pediatric specialists in neurosurgery, orthopedics, urology, developmental pediatrics, and physical medicine; physical therapists; orthotists; psychologists; social workers; and health education professionals.
- All these specialists might not be available to all teams and for all clinic settings. The care team's goal should be to meet the individual needs of each child and family by providing comprehensive and coordinated care, support, and education to the patients and families and support and assistance to the primary care provider, the child's school or early intervention program, and other service providers
- See tables below from AAP Clinical Report on providing a primary care medical home for patients with spina bifida- good summary

TABLE 4 Primary Care Interventions Typically Provided to Children and Teenagers With Spina Bifida

Fetuses (prenatal)
Counsel families in planning and decision-making
Provide information on spina bifida
Options and assistance with family choices
Consult with obstetric, neonatal, and neurosurgical teams
Prenatal (fetal)/postnatal surgery
Discuss postnatal planning and treatment
Family support
Newborns
Postnatal care and stabilization
Surgical repair of spinal lesion
Monitoring and surgery for hydrocephalus
Family support
Referral to multidisciplinary spina bifida term
Primary and specialty follow-up
Infants
Provide early and frequent follow-up
Monitor hydrocephalus
Provide routine and diagnostic-specific primary care
Give family and sibling teaching and support
Discuss recurrence risk and prevention
Refer to early-intervention program
Communicate and coordinate with the spina bifida team
Toddlers
Preventive and well-child care
Developmental monitoring
Mobility
Growth and weight management
Preschool-aged children
Transition from early-intervention program to preschool program
Ambulation and mobility
Bowel and bladder program
Social inclusion
School-aged children
Identify and characterize learning abilities
Ensure appropriate school-based services, Individualized Educational Plan or 504 plan
Monitor secondary conditions including latex allergy
Encourage physical activities, friends, and household responsibilities
Plan for educational transition
Encourage independent self-care and toileting
Preteens
Begin health care transition planning
Advocate for physical activities, recreation and community inclusion,
Monitor progress in school
Address bullying and safety
Monitor growth and puberty
Encourage independence and self-advocacy
Develop and maintain friendships
Teens
Continue transition and transfer process to adult care, activities and social participation
Educate regarding spina bifida and self-care
Provide private health care visits
Provide anticipatory guidance regarding sexuality and reproduction
Encourage independence in health care decision-making
Monitor growth and vital signs (blood pressure)
Encourage physical activities
Manage weight and nutrition
Encourage cardiopulmonary health and fitness
Young adults
Transfer care to a provider of routine adult health care
Provide resources for specialty care: neurosurgery, orthopedics, urology, and others
Monitor weight and physical fitness
Provide information regarding finances such as Social Security Disability Income and health insurance
Monitor education and employment
Help to build and maintain friendships and social support groups
Monitor for deterioration and late-onset complications

TABLE 3 Care of the Child and Youth With Spina Bifida: Potential Roles of the Pediatric Primary Care Provider

Time	Concerns	Actions
Fetuses (prenatal)	Diagnosis	Consult with diagnostic team
	Lesion: type and location	Review obstetric plan
	Other central nervous system findings (hydrocephalus, Chiari II malformation)	Review findings and implications with family
	Other physical findings (orthopedic, other)	Discuss options and plan
	Maternal and family stress and anxiety	Referral to spina bifida team
	Family choices and plan	Update primary care provider and other provider knowledge base
		Family education and support
		Communicate among family and all professionals
Newborns	Stabilization	Consult obstetric and neonatal teams, neurosurgery, others specialists
	Protection of lesion	Family support
	Examination	Monitor head growth
	Surgical closure	Neonatal screening and newborn hearing assessment
	Hydrocephalus	Discharge planning
	Motor function	Primary care and specialty follow-up
	Bowel and bladder function	
	Orthopedic conditions	
	Maternal/family anxiety and depression	
Infants	General health status	Health care per *Bright Futures* recommendations
	Growth	Referral to spina bifida team and support group
	Development	Referral to early-intervention program
	Immunizations	Monitor head growth and shunt function
	Spina bifida-specific concerns: hydrocephalus and shunt function, bowel and bladder function, UTI	Assess for feeding or swallow problems
		Discuss latex precautions
		Supplemental Security Income, Medicaid, and Medicaid Waiver
Preschool-aged children	General health status	Health care per *Bright Futures* recommendations
	Growth and development	Follow-up with spina bifida team and support group
	Spina bifida-specific concerns: vision and strabismus, motor function and mobility, hydrocephalus and shunt function, bowel and bladder function, UTI	Obtain early-intervention assessment, individualized family service plan
		Developmental team evaluation
		Discuss social inclusion and activities
		Bracing and ambulation
		Activity and weight management
		Dental referral
		Toileting
School-aged children	General health status	Health care per *Bright Futures* recommendations
	Growth and development	Follow-up with spina bifida team
	Weight control and healthy diet	School transition, service and support, Section 504/individualized education plan
	Spina bifida-specific concerns: motor function and mobility, hydrocephalus and shunt function, bowel and bladder function, tethered cord	Screen for attention-deficit/hyperactivity disorder, learning disabilities, and executive function disorders
		Bullying and safety
		Self-care and independence
		Physical activities and social group participation
		Neurology and physical therapy follow-up
Teens	General health status	Adolescent health care per American Academy of Pediatrics recommendations
	Weight control and healthy diet	Confidentiality and private visits
	Sexuality	Birth control and folic acid
	Psychosocial stressors	Health care transition
	Spina bifida specific concerns: motor function and mobility, tethered cord	Self-care and independence
	Developing independence	Driving and transportation
		Spina bifida teen support group
		Educational and vocational planning
Adults	General health status	Identify health care resources
	Weight control and healthy diet	Health insurance, Social Security Disability income
	Spina bifida specific concerns: motor function and mobility, bowel and bladder management, skin care and pressure sores	Education and employment
		Living situation
		Spina bifida adult support group
		Transfer to accessible adult health care

Concerns and actions are not intended to be limited to a single time period but should be addressed and readdressed as part of ongoing health care across the life span.

4. Know the conditions commonly associated with myelodysplasia.

- In addition to the primary deficits in motor and sensory function, children and youth with spina bifida experience a range of comorbid conditions including learning disabilities- math learning difficulties are particularly connected with SB patients- even without occurrence of reading learning disabilities, as well as problems with attention and executive function, dysfunction of upper extremities, strabismus, and seizures
- Also subject to other functional complications such as limitations of movement and ambulation, scoliosis, joint instability, fractures secondary to lower bone mineral density as well as secondary osteoporosis, bowel and bladder dysfunction, altered growth including precocious puberty, and obesity
- these children and teenagers will need ongoing prevention because of the increased risk of developing latex allergies
- the physical and psychological consequences of impaired mobility and independence, altered appearance, and the long-term needs of the condition also require identification and intervention
- the child's physical and developmental disabilities, limited mobility, and chronic health conditions can be barriers to social integration that can have lifelong consequences
- Below table offers good summary of systemic effects of myelomeningocele specifically

TABLE 2 Systemic Effects of Myelomeningocele

System	Effects
Neurologic	
Central	Hydrocephalus, Chiari II malformation, dysgenesis of the corpus callosum
Cranial nerves	Vision, strabismus, hearing, speech, stridor, swallow
Spinal	Tethered cord, progressive loss of motor and sensory function
Motor	Paraplegia, upper extremity hypotonia
Sensory	Loss of sensation and proprioception
Cognitive	Cognitive deficits including learning disabilities, executive function disorders, attention-deficit/hyperactivity disorder
	Lower cranium nerve dysfunction
Vision	
Acuity	Visual acuity problems, rarely severe
Alignment	Strabismus, oculomotor disorders
Oromotor	Oromotor dysfunction, swallowing disorder
Respiratory	
Central	Central ventilatory disorder, apnea, sleep apnea, hypoventilation
Pulmonary	Aspiration, restrictive and obstructive lung disease, pneumonia
	Stridor
Gastrointestinal	
Swallow	Swallowing dysfunction; oral, pharyngeal
Neurogenic bowel	Constipation, soiling, accidents
Urologic	
Neurogenic bladder	Bladder atonia/dystonia, increased bladder pressure, leakage/incontinence, reflux, renal failure
UTI	Bacterial colonization, acute/chronic UTIs
Genital	
Insensitivity	Lack of sexual sensation, erectile dysfunction, and retrograde ejaculation in males
Infertility	Early pubertal onset and increased risk of NTD in infants of women with NTD
Orthopedic	
Spinal	Scoliosis/kyphosis
Extremities	Decreased mobility, hip dislocation, clubbed feet, osteopenia, fracture, linear growth abnormalities
Growth	
Stature	Short stature resulting from limb-length disorders and precocious puberty in females
Weight	Overweight and obesity
Psychology	
Anxiety/depression	Increased anxiety and depression
Behavior	Social isolation, anxiety, depression, immaturity, risk-taking behavior, at increased risk for abuse
Social	
Inclusion	Social isolation, limited friendship, physical and transportation barriers
Employment	Limited work experience, physical/society barriers, loss of health coverage or benefits

Myelomeningocele is the most serious and complex NTD and affects multiple body systems; the severity of these effects is related to the type and location of the NTD and the extent of neuronal injury.

5. Understand the relationship between genetic and environmental factors in the etiology and prevention of myelodysplasia.

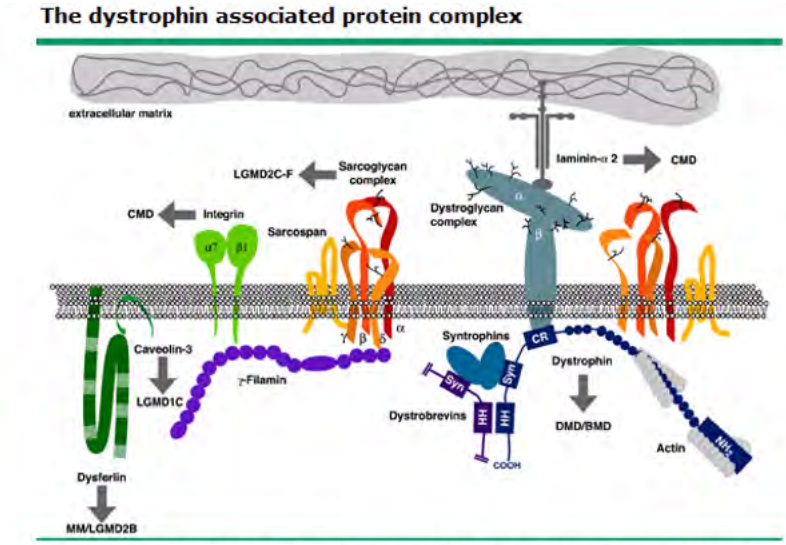
- Spina bifida, anencephaly, and other NTDs occur as a result of defective neurulation or closure during the third week after conception of the embryonic neural fold, which becomes the neural tube.
- Failure of closure in a cephalic direction leads to the development of anencephaly; failure of closure in the caudal direction leads to spina bifida. Myelomeningocele arises as a failure of neurulation by approximately day 28 after conception.
- The etiology of spina bifida includes genetic and environmental factors
    - spina bifida occurs worldwide and in all racial groups, although there are geographic and ethnic variations
    - genetic factors are likely to be related to the increased risk in some populations, notably the Irish, Scottish, and other northern Europeans
    - this risk may be related to altered folate metabolism—in the United States, a slightly higher rate occurs in families of Latin descent
    - Chromosome disorders including trisomy 13 or 18 and microdeletion of 22q11 are linked to NTDs
    - Exposure to some prenatal medications increases the risk of spina bifida→ including valproic acid, carbamazepine, isotretinoin, methotrexate and other folic acid antagonists, excess vitamin A or its analogs, and retinoic acid
    - Maternal nutrition (especially folate deficiency), alcohol consumption, obesity, and fever, either attributable to illness or hot tub/sauna use, can increase risk, as does maternal diabetes mellitus
    - Family history of previous NTDs is a significant risk factor, increasing the risk more than 20-fold (e.g., from 0.1%–2.5%)

6. Understand the urological, neurological, and/or orthopedic complications associated with myelodysplasia. **See above tables and figures**
7. Know the developmental and behavioral characteristics of individuals with myelodysplasia. **See above tables and figures**

C. Muscular dystrophies

1. Recognize the signs and symptoms of Duchenne and other muscular dystrophies

- The term muscular dystrophy refers to those inherited disorders of skeletal muscle that have no central or peripheral nervous system involvement
  - Muscular dystrophies are an inherited group of progressive myopathic disorders resulting from defects in a number of genes required for normal muscle function
  - Some of the genes responsible for these conditions have been identified
  - Muscle weakness is the primary symptom
  - Nine major forms of muscular dystrophy: Myotonic, Duchenne, Becker, Limb-Girdle, Facioscapulohumeral, Congenital (separate from Congenital Myotonic Dystrophy), Oculopharyngeal (not discussed below as typical age of onset is after age 40), Distal, and Emery-Dreifuss
  - Most, but not all, are associated with progressive muscle weakness, some of the mild dystrophies may be relatively static, and children who have congenital muscular dystrophy (CMD) may show periods of improvement



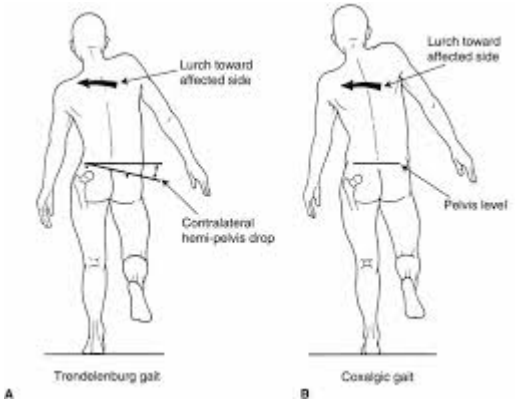
Dystrophin is located on the cytoplasmic face of the plasma membrane of muscle fibers. Arrows indicate the protein components mutated in various muscular dystrophies. Alterations in the dystrophin gene cause Becker and Duchenne muscular dystrophies. Mutations in the sarcoglycan proteins, caveolin-3 and dysferlin, lead to limb girdle muscular dystrophies. The laminin a2-chain is mutated in a subtype of congenital muscular dystrophy without structural brain anomalies, as is a7-integrin.
BMD: Becker muscular dystrophy; CMD: congenital muscular dystrophy; COOH: carboxy-terminal domain; CR: cysteine-rich domain; DMD: Duchenne muscular dystrophy; HH: two helices of the coiled-coil domain; LGMD: limb-girdle muscular dystrophy; MM: Miyoshi myopathy; NH2: amino-terminal domain; Syn: syntrophin-binding domain.

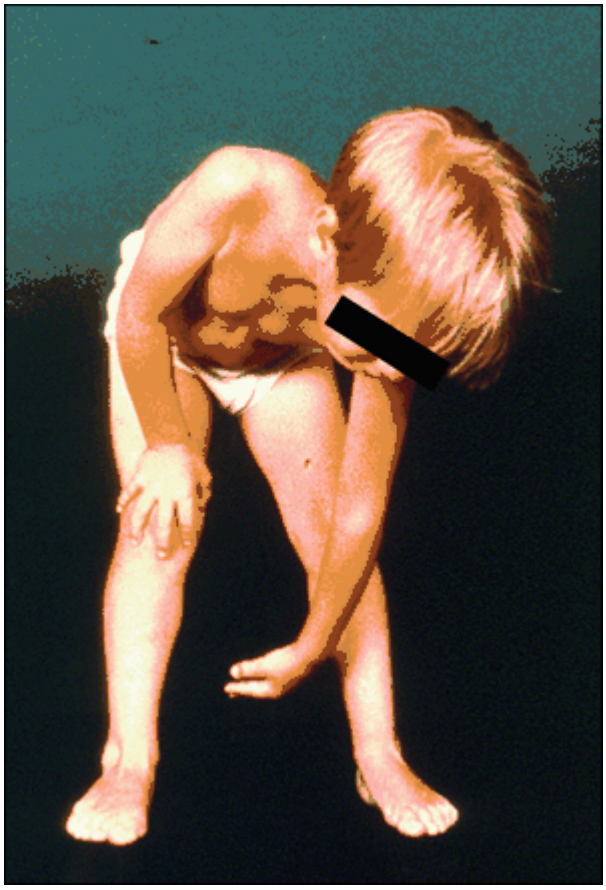
- DMD is an X-linked recessive disorder affecting primarily skeletal and cardiac muscle
  - The incidence is 1 in 3,300-5,000 liveborn males
  - The gene is located on the short arm of the X chromosome

- Most genetic mutations involve deletions; less often, point mutations and duplications are seen
- Without dystrophin, the glycoprotein structure of the muscle sarcolemma is less stable.
- Membrane instability leads to muscle damage, with the initiation of an inflammatory cascade contributing further to muscle damage, necrosis, and fibrosis

o Boys who have DMD exhibit a progressive and predictable loss of muscle function
- The muscles are affected at birth, but clinical symptoms of proximal muscle weakness usually manifest between 3 and 5 years of age
- Proximal muscles are involved first
- The boys may walk later than their siblings, but most are walking by 18 months of age.
- Toe-walking is common
- Vertical growth velocity is slowed in first few years of life→short stature; even before introduction of steroids
- Running, jumping, and hopping are awkward and difficult, if not impossible
- Muscle weakness often is apparent when the boys are observed playing with their siblings or other children

o As the pelvic muscles weaken further, affected boys develop a lumbar lordosis and a Trendelenburg gait (see picture below)
- They fall more often and have difficulty rising
- To raise themselves up, they get into a knee-elbow position, extend their elbows and knees, bring their hands and feet as close together as possible, and place one hand at a time on their knees→They then place their hands on their thighs and move proximally in alternating steps ("climbing up their legs") to become erect; known as the Gower maneuver (see picture below)





- Boys who have DMD are at increased risk for certain cognitive concerns
  - On average, individuals with DMD perform at 1 SD below the general population on full-scale IQ assessments
  - Dystrophin is present in the brain and plays an essential role in myelination during postnatal brain development
  - Intellectual dysfunction is associated with particular genetic mutations affecting specific dystrophin-protein isoforms, leading to intellectual disability (IQ<70) in about a quarter of the DMD population (20%–27%)
  - Their motor and language development may be delayed
  - More at risk for anxiety, ADHD, and OCD
  - Estimated that as many as 1 in 4 boys with DMD also carry a diagnosis of ASD
  - Individuals with DMD frequently have delayed diagnoses of cognitive and behavioral conditions, because focus is generally on medical treatment and intervention
  - Learning disabilities typically involve executive dysfunction (eg, inattention, disorganization) that is not disruptive in the classroom and compound the likelihood the learning disability will go undiagnosed
  - Almost half the individuals with DMD have a specific learning disability
  - Unlike their muscle weakness, which is progressive, their cognitive skills do not deteriorate over time→many boys require specifically designed instruction and implementation of IEPs for school support
- Muscle weakness continues, with the legs affected earlier than the arms

        - The boys begin to use wheelchairs full time between 8 and 12 years of age, most by 10 years
        - Approximately 3 to 4 years after losing ambulation, 90% of the boys develop a spinal curvature of greater than 20 degrees
        - Surgery is required to stabilize the spine
        - Joint contractures develop initially in the lower extremities, with the feet assuming the typical equinovarus position
        - Upper extremity function declines in the mid-teens, and the boys lose the ability to feed and care for themselves
        - This impairment frequently occurs after spinal surgery
    - Pulmonary function begins to deteriorate between 9 and 11 years of age
        - The forced vital capacity declines by 5% to 10% per year, their cough becomes weaker, and the ability to clear respiratory tract secretions is impaired
        - Recurrent pneumonia is common
        - Nocturnal assisted ventilation frequently is required in their mid-to-late teens.
        - Improved pulmonary care and aggressive treatment of pulmonary infections have improved life expectancy.
- Electrocardiographic and echocardiographic changes are present in more than 50% of boys who have DMD
    - They usually are free of cardiac symptoms (fatigue and reduced exercise tolerance) because they use a wheelchair full time and do not exercise vigorously
    - Some boys experience tachycardia and are aware of their hearts beating
    - They usually die in their late-teens to mid-twenties—75% from respiratory causes and approximately 25% from severe left ventricular failure.

- Becker's Muscular Dystrophy (BMD)
    - BMD is also an X-linked muscular dystrophy disorder caused by a mutation in the DMD gene
    - BMD has a milder clinical course and occurs less frequently than DMD, affecting 1:11,500–19,000 males; patients with BMD usually survive beyond the age of 30 years
    - typically, BMD manifests later in life with a mean onset of 12 years (though it varies widely from 5 to 60 years of age for age of onset of symptoms), loss of ambulation is delayed until the third or even fourth decade of life, onset of cardiac involvement is variable,→ course is overall more variable and unpredictable when compared to DMD
    - Intellectual disability and contractures are also not as common or severe and there is relative preservation of neck flexor muscle strength in BMD and intermediate types of muscular dystrophy
    - BMD results from an inherited or spontaneous/de novo mutation in the DMD gene
        - Approximately 1/3 of cases arise from spontaneous mutations
        - The correlation between mutation and phenotype →best explained by the reading frame theory (Monaco, 1988)
        - This theory argues that if a deletion leads to a shift in the open reading frame, then premature termination results in a truncated protein (or no protein), which is often associated with the DMD phenotype
        - By contrast, if a deletion does not result in a frameshift, then a partially functional, abnormally small dystrophin protein is often produced, resulting in the BMD phenotype
    - Individuals with BMD typically have average cognitive function as mentioned above

        - Both DMD and BMD have higher frequencies of learning disabilities and behavioral comorbidities than the general population (including ADHD, anxiety, autism symptoms)
        - About 1/3 of individuals with BMD have a learning disability
    - In patients with BMD, serum CK concentrations are usually elevated above the upper limit of normal by a factor of five or more
    - Although muscle involvement is less severe than in DMD, cardiac involvement in BMD is often a predominant feature of the presentation
        - Pathogenic changes in the cardiac muscle begin at an early age for both DMD and BMD, but clinically significant dysfunction is typically seen during the second decade
        - most individuals with DMD and BMD will eventually have dilated cardiomyopathy, but the age of onset and time to death are variable, and to date there have been no clear predictors of an individual's cardiac prognosis

        - Some studies have shown that the mean age of onset of cardiomyopathy is consistent across both BMD and DMD at ~14.5 years, but other reports indicate that ~85%–90% of DMD will have cardiomyopathy by age 18 years and individuals with BMD will present later with cardiomyopathy in either in the third or fourth decades
        - Echocardiography reveals early right ventricular involvement with the later development of left ventricular dysfunction in BMD
            - All four chambers are eventually involved with fibrosis, and a cardiomyopathy with heart failure can be rapidly progressive
            - In addition, abnormalities of the AV node and infranodal conduction system can result in fascicular and bundle branch block and can progress to complete heart block
            - Although not typically performed, endomyocardial biopsy shows a variable distribution of dystrophin in cardiomyocytes
            - Discontinuous immunostaining of cardiac dystrophin is characteristic of BMD and the absence of immunostaining may be associated with more severe cardiac disease.
        - individuals with BMD are much more likely to qualify for cardiac transplantation when compared to individuals with DMD are unable to qualify for transplant surgery, due to skeletal-muscle impairment, rejection, and low levels of pulmonary function
        - A recent review of the literature indicates individuals with BMD who are eligible for cardiac transplant will tolerate the procedure on par with individuals with other forms of heart failure
- Intermediate phenotype & symptomatic females
    - There is an known intermediate group of patients, also known as outliers, has a clinical phenotype best characterized as mild DMD or severe BMD; these individuals usually become confined to wheelchairs between the ages of 13 and 16 years
    - Clinically apparent symptoms occur in approximately 22 percent of female carriers of a mutated dystrophin gene
        - Symptomatic female DMD carriers have a high variability in age of onset and degree of muscle weakness or cardiac involvement
        - Weakness ranges from mild to, rarely, a classic DMD phenotype.
        - Serum CK is increased in approximately 70 and 50 percent of Duchenne and Becker female carriers, respectively, although the values decline with age

        - Hence, many carriers have CK values in the normal range, so genetic testing (ideally with the mutation first identified in the affected boy) should be used to determine carrier status wherever possible
        - The elevations are usually mild (up to three times the upper limit of normal)
        - In symptomatic carriers, however, the CK levels may be much higher
        - Female carriers are more at risk for having a DMD phenotype if one of the following genetic abnormalities is present:
            - A normal karyotype but nonrandom (skewed) X chromosome inactivation, resulting in diminished expression of the normal dystrophin allele
            - 45,X, 46,XY, or Turner mosaic karyotypes
            - Apparently balanced X/autosome translocations with breakpoints in Xp21, within the dystrophin gene, and preferential inactivation of the normal X
            - Uniparental disomy of the X chromosome
- Limb-Girdle Muscular Dystrophy (LGMD)
    - LGMD is defined as a muscular dystrophy with predominantly proximal distribution of weakness
        - It includes a number of heterogeneous genetic disorders that vary in severity, phenotype, pathology, and age of onset, which ranges from childhood through adulthood
        - LGMD is still used as a generic term to describe those patients with muscular dystrophy of girdle distribution
        - Together, the group of disorders that constitute LGMD is the fourth most common genetic cause of muscle weakness – behind the dystrophinopathies (Duchenne and Becker muscular dystrophy), myotonic dystrophy, and facioscapulohumeral muscular dystrophy – with an estimated minimum prevalence of 1 in 20,000
    - Most cases of LGMD are inherited in an autosomal recessive or an autosomal dominant pattern
        - In some families, the inheritance pattern cannot be determined
        - The discovery of genetically distinct subtypes has redefined the classification of LGMD and has led to nomenclature designating the autosomal dominant forms as LGMD1A, 1B, 1C, etc, and the autosomal recessive forms as LGMD2A, 2B, 2C, etc

    - The distinction between BMD and limb-girdle muscular dystrophy (LGMD) is often hard to make in patients with a negative family history for BMD

        - However, the calf muscle pseudohypertrophy is usually not as striking in LGMD
        - General features of the disorders that make up LGMD are progressive weakness and muscle atrophy mainly involving the shoulder girdle (scapulohumeral type), the pelvic girdle (pelvifemoral type), or both. Most childhood-onset cases have a pelvifemoral distribution of weakness. By comparison, adult-onset disease usually involves both shoulder and pelvic

girdles with gradually increasing proximal limb weakness, thereby leading to restriction of mobility and eventually to wheelchair confinement.

- Facial weakness is usually mild and, in some cases, totally absent
- Extraocular muscles are completely spared in the LGMDs.
- Distal muscle strength is usually preserved, even at the late stage of the disease, but distal muscle weakness can be an early or prominent feature in some LGMD subtypes
- Intellect is usually normal, though intellectual disability is a feature of LGMD2K and has been reported in LGMD2N
- Many of the subtypes of LGMD have characteristic though nonspecific clinical manifestations
- The most prevalent LGMD subtypes are the following:
    - LGMD2A (calpainopathy)
    - LGMD2B (dysferlinopathy)
    - LGMD2C through 2F (the sarcoglycanopathies)
    - LGMD2I (dystroglycanopathy)
    - LGMD2L (anoctamin 5)
    - LGMD1B (laminopathy)
- Autosomal dominant subtypes — The presentation of autosomal dominant LGMD (designated LGMD1) shows greater clinical heterogeneity, later age of onset, more gradual progression, and a creatine kinase elevation that may be minimal compared with autosomal recessive subtypes.
- LGMD1B (laminopathy) is the most common autosomal dominant type, and accounts for 5 to 10 percent of all cases of LGMD
    - Though autosomal dominant, there may be variable penetrance from generation to generation
    - The typical age of onset ranges from 2 to 25 years of age, but earlier or later onset can occur
    - The pattern of weakness is limb-girdle or humeroperoneal
    - Cardiac conduction defects, arrhythmias, cardiomyopathy, and mild elbow and neck contractures are common manifestations, thus overlapping with autosomal dominant Emery-Dreifuss muscular dystrophy
    - Creatine kinase levels range from 200 to 2000 units/L
    - Cardiac involvement is generally seen by the second or third decade in LGMD1B
    - Most affected individuals require pacemakers; worsening heart failure due to arrhythmogenic cardiomyopathy may necessitate heart transplantation.
    - findings with normal sarcoglycan, merosin, and dystrophin staining
- Autosomal recessive subtypes — Compared with autosomal dominant disease, autosomal recessive LGMD (designated LGMD2) is usually associated with earlier age of onset, more rapid progression, and relatively high creatine kinase values.
    - An affected child with autosomal recessive LGMD, particularly one with LGMD2I, may be indistinguishable on examination from a child with Duchenne or Becker muscular dystrophy. However, unlike Duchenne muscular dystrophy, cognitive function is typically normal in children with autosomal recessive LGMD. An exception is LGMD2K, which is associated with ID

- LGMD2A (calpainopathy) is caused by a mutation in the gene (CAPN3) encoding the proteolytic enzyme calpain-3
    - It is considered the most common type of LGMD worldwide, accounting for 15 to 40 percent of all cases of LGMD with variation depending in part on the geographic region
    - The onset of LGMD2A occurs between 6 and 18 years of age in 71 percent
    - There is significant involvement of the parascapular muscles, biceps, gluteus maximus, adductors, and hamstrings
    - Hip girdle muscles are weaker than shoulder girdle muscles, with severe weakness involves hip extension, adduction, and knee flexion
    - Scapular winging, abdominal laxity calf hypertrophy, hyperlordosis, and a waddling gait are common
    - Contractures are extensive and tend to develop early
    - Facial weakness may occur in cases with early-onset or severe disease
    - Creatine kinase levels range from 500 to 20,000 units/L. Muscle biopsy often shows lobulated fibers
    - Cardiac and pulmonary involvement can occur, but is not common
    - Requirement for a wheelchair occurs between the ages of 21 and 40 years in approximately 80 percent
    - The scapular winging, abdominal laxity, and variable facial weakness can cause diagnostic confusion with fascioscapulohumeral muscular dystrophy
    - Although the inheritance of calpainopathy was long considered to be exclusively recessive, autosomal dominant transmission over several generations has been observed among some families in patients who carry a single in-frame c.643_663del21 deletion of the CAPN3 gene →patients with this autosomal dominant calpainopathy have a phenotype that resembles but is generally milder than the recessive form.

- LGMD2B (dysferlinopathy) is caused by mutations in the DYSF gene, and is the second most common subtype of LGMD, accounting for 5 to 35 percent of all cases

    - prior to onset, development is normal and some individuals excel at athletics
    - the typical age of onset ranges from 15 to 35 years, and legs are usually affected first
    - Distal involvement is often present, with early weakness and atrophy of the gastrocnemius and inability to walk on the toes.
    - When the involvement is only distal, it is known as Miyoshi distal myopathy
    - In LGMD2B, arm weakness may occur with progression, but scapular winging is not seen
    - Likewise, there is no facial weakness or dysphagia
    - Cardiac or pulmonary involvement is uncommon; when it does occur, it is usually asymptomatic and late in the course of the disease

- Serum creatine kinase levels can be highly elevated, and range from 1000 to 40,000 units/L
- Muscle biopsy shows inflammatory features in 40 percent or more of cases
- LGMD2B is usually slowly progressive, with need of a wheelchair 10 to 20 years after onset
- However, occasional patients have rapid onset and progression with loss of ambulation over one to two years, in some cases apparently triggered by pregnancy

- LGMD2C, 2D, 2E, and 2F (the sarcoglycanopathies) together are a common cause of LGMD, accounting for 10 to 20 percent of cases

    - Onset usually occurs at 5 to 15 years of age
    - Typical features include proximal leg weakness, scapular winging, calf hypertrophy, macroglossia, and lumbar lordosis
    - Thus, the pattern of weakness is reminiscent of Duchenne muscular dystrophy, but cognitive function is preserved.
    - Cardiac and respiratory involvement is frequent with disease progression
    - serum creatine kinase levels range from 500 to 20,000 units/L. Muscle biopsy typically shows abnormal staining for all four sarcoglycans
    - There is progression to wheelchair use within 10 years of onset in many cases, but occasional patients have only mild disease characterized by exercise intolerance, myoglobinuria, minimal weakness, and slower progression.

- LGMD2I is a frequent form of LGMD, accounting for 20 to 40 percent of all cases worldwide

    - It is caused by mutations in the fukutin-related protein gene (FKRP)
    - While phenotypically heterogeneous, common clinical features of LGMD2I include proximal muscle weakness and atrophy, calf muscle hypertrophy, lumbar lordosis, scapular winging, macroglossia, and an increased incidence of cardiomyopathy, and elevated plasma levels of creatine kinase
    - These features are similar to those of Becker muscular dystrophy.
    - Cardiac and respiratory involvement is frequent with disease progression but can also arise early in the course of the disease and may not correlate with the severity of skeletal muscle weakness
    - Creatine kinase levels range from 500 to 20,000 units/L
    - Muscle biopsy shows reduced or absent immunostaining for alpha-dystroglycan

- The disease course can be sporadic, with periods where the strength remains stable for years followed by deterioration
- Some patients maintain ambulation into the fifth decade of life, but up to 30 percent require noninvasive ventilation
- In a study that screened 102 patients with a suspected diagnosis of sporadic Duchenne or Becker muscular dystrophy who were negative for dystrophin gene deletions or duplications, the FKRP gene mutation that causes LGMD2I was found in 13 percent of patients
    - This result suggests that a substantial number of patients with a phenotype of Duchenne or Becker muscular dystrophy who are negative for dystrophin gene mutations may have a form of LGMD and should be tested for the FKRP gene mutation

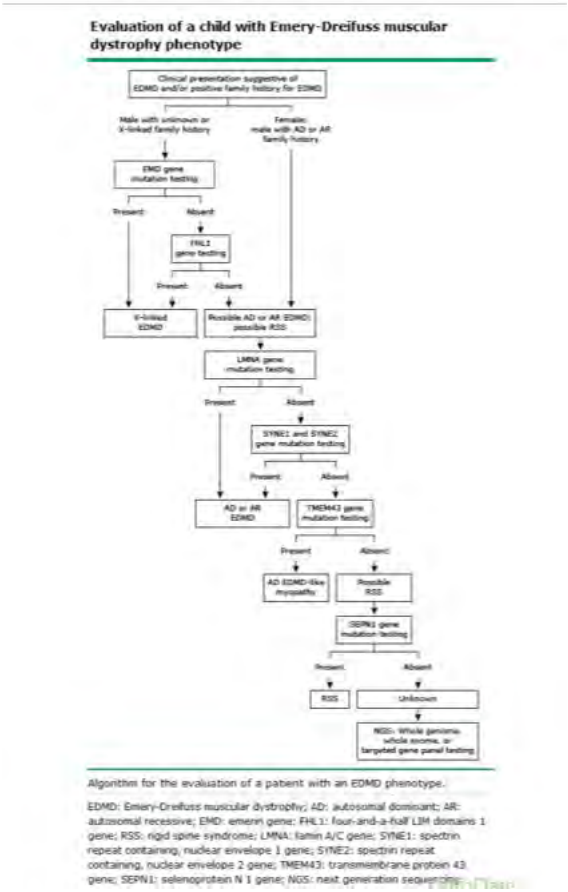
- LGMD2K is characterized by intellectual disability and abnormal alpha-dystroglycan, and is caused by a mutation in the POMT1 gene encoding the protein-O-mannosyltransferase
    - The same gene is associated with congenital muscular dystrophy-dystroglycanopathy with brain and eye anomalies (type A or MDDGA), which includes the phenotypes known as Walker-Warburg syndrome and muscle-eye-brain disease

- Cardiac involvement
    - Cardiac involvement is common in LGMD1B (laminopathy), LGMD1E, LGMD2E, and LGMD2I (MDDGC5)
    - It is unusual in LGMD1A (myotilinopathy), LGMD1D, LGMD2A, and LGMD2D
    - Dilated cardiomyopathy and cardiac conduction system abnormalities are the most common types of cardiac involvement

✓ Emery-Dreifuss muscular dystrophy (EDMD), also known as humeroperoneal muscular dystrophy, can be inherited as an X-linked recessive, autosomal dominant, or autosomal recessive disorder

- X-linked forms of the disorder are EDMD1 (*EMD* gene on Xq28)and EDMD6 (*FHL1* gene on Xq26.3)
- Both an autosomal dominant (EDMD2) and a rare autosomal recessive (EDMD3) form are linked to mutations of the *LMNA* gene on chromosome 1q21.2 that encodes for lamin A and lamin C
- In addition to EDMD2 and EDMD3, a number of other laminopathies (ie, disorders caused by *LMNA* gene mutations) have been identified
- major features of EDMD are early contractures, slowly progressive humeroperoneal muscle weakness/wasting, and cardiac disease with conduction defects, arrhythmias, and cardiomyopathy
- The different forms of EDMD have generally similar symptoms, which usually begin in the first or second decade of life. However, the disease exhibits significant inter- and intra-familial variability with regard to age at onset, severity, and progression of the major manifestations
- Cases of adult onset with slow progression have been observed

- Contractures at the elbows are noted early and are commonly the first manifestations of EDMD. Contractures of the posterior aspect of the neck, the entire spine, and the Achilles tendons also occur. Achilles tendon contractures are frequently associated with toe walking. Severe spine and leg contractures eventually may lead to loss of ambulation.
- Muscle weakness and wasting has a humeroperoneal distribution; it typically begins in the arms, involving both the biceps and triceps, with relative preservation of the deltoid muscles. Subsequently, distal leg weakness with atrophy of the peroneal muscles is noted. In some cases, mild facial weakness may also be observed. The myopathy tends to be slowly progressive during the first three decades of life, but more rapid thereafter.
- A dilated cardiomyopathy is seen in most patients with EDMD
    - It is typically associated with atrioventricular conduction abnormalities such as first-degree atrioventricular block, but also with sinus bradycardia or supraventricular tachycardia, which may be early signs of cardiac involvement
    - Other findings include atrial paralysis, atrial fibrillation, and atrial flutter; these are most common in EDMD1
    - Symptoms of hypoperfusion (syncope or near syncope) often result from infranodal or atrioventricular conduction block with the development of slow junctional rhythms, requiring pacemaker insertion
    - The onset of cardiac abnormalities is usually in the third decade of life but earlier onset has been observed
    - Sudden death may develop in patients not previously diagnosed because they have little or no skeletal myopathy
    - There is no correlation between the degree of neuromuscular involvement and the severity of cardiac abnormalities
    - Arrhythmogenic dilated cardiomyopathy, cardiac failure, and ventricular tachyarrhythmias are more common and more severe in autosomal dominant EDMD2 (due to *LMNA* mutations) but also occur in X-linked EDMD1 (caused by *EMD* mutations)
- EDMD is associated with abnormalities in serum creatine kinase (CK), electrocardiography (ECG), electromyography (EMG), muscle imaging, and muscle biopsy
- The clinical diagnosis of EDMD can be made for patients with the cardinal features:
    - Early contractures of elbow flexors, ankle plantar flexors, and spine
    - Childhood onset of humeroperoneal weakness and wasting
    - Cardiac disease with conduction defects, arrhythmias, and cardiomyopathy, which may present later in life
- The diagnosis of EDMD is established in a patient with a compatible clinical phenotype by genetic testing that identifies a hemizygous pathogenic variant in *EMD* or *FHL1*, a heterozygous pathogenic variant in *LMNA*, or rarely biallelic pathogenic variants in *LMNA (see algorithm below)*



Facioscapulohumeral muscular dystrophy (FSHD)

- third most common type of muscular dystrophy
- typical or classic form of FSHD is characterized by muscle weakness involving the facial, scapular, upper arm, lower leg, and abdominal muscles, usually with asymmetric involvement
    - The age of symptom onset varies from infancy to middle age, but is usually in the second decade
    - By age 20 years, findings are seen in approximately 90 percent of affected patients, although some or all of the signs may be subclinical
    - Progression is usually slow with a normal or near-normal life span
    - However, disease severity is also highly variable
    - Other manifestations of FSHD may include chronic pain, retinal vasculopathy, progressive hearing loss, cardiac arrhythmia, cognitive impairment, and epilepsy
- two main types: FSHD1 and FSHD2- both caused by different mutations of the DUX4 gene that lead to inappropriate expression of the double homeobox protein 4 gene (*DUX4*) → *DUX4* gene lies within each unit of a macrosatellite array known as D4Z4, located in the 4q35 region.
- there is an infantile variety of FSHD, which is often sporadic in inheritance, is observed in approximately 4 percent of patients with FSHD
    - The onset is within the first few years of life and the course is rapidly progressive in most cases, with wheelchair confinement by the age of 12 years or earlier
    - Children with this form of FSHD develop early facial weakness, with an inability to close the eyes in sleep, and inability to smile or show facial expression

- The weakness soon involves the shoulder and hip girdles resulting in severe lumbar lordosis, pronounced forward pelvic tilt, and hyperextension of the knees and the head upon walking
- Marked weakness of the wrist extensors may result in a wrist drop

- The clinical diagnosis of typical FSHD is suspected in patients who present with relatively selective weakness of the face and shoulder girdle muscles, typically including weakness of the scapular fixators with scapular winging
- clinical diagnostic criteria for FSHD have been largely supplanted by molecular genetic testing→some patients who now are given a confirmed genetic diagnosis of FSHD do not fulfill all the clinical criteria
- Electromyography (EMG) and muscle biopsy are not necessary when the diagnosis of FSHD is confirmed by genetic testing. However, EMG and muscle biopsy are suggested for patients with a clinical suspicion of FSHD who have negative standard genetic testing for FSHD1 and FSHD2

✓ Myotonic Dystrophy
- Myotonic dystrophy comprises the most common forms of adult-onset MD
- two major forms of myotonic dystrophy: DM1 and DM2
- there are congenital and childhood forms of DM1
- Myotonic dystrophy type 1 (DM1) and type 2 (DM2) are similar in that both are multisystem disorders→present with skeletal muscle weakness and myotonia (ie, abnormally slow or delayed muscle relaxation following normal muscle contraction with a characteristic neurophysiologic signature on electromyographic [EMG] testing), cardiac conduction abnormalities, cataracts, and other abnormalities
- Congenital DM1- DM1 results from an expansion of a cytosine-thymine-guanine (CTG) trinucleotide repeat in the 3'-untranslated region of the dystrophia myotonica protein kinase (*DMPK*) gene
  - characterized by profound hypotonia, facial diplegia, poor feeding, arthrogryposis (congenital joint contractures), especially of the legs, and respiratory failure
  - the majority of affected infants (at least 80 percent) have a characteristic "V" shape of the upper lip that results from facial diplegia
  - in addition to the profound hypotonia and facial weakness, physical examination also shows truncal and appendicular weakness as well as areflexia or marked hyporeflexia
  - Arthrogryposis usually involves at least the ankles, leading to clubfoot deformity
  - In some cases, DM1 may present before birth as polyhydramnios, talipes (clubfoot), and reduced fetal movement
  - In the most severely affected infants, polyhydramnios is common during pregnancy and is related to disturbance in swallowing; polyhydramnios in the mother usually indicates serious involvement of the fetus
  - Labor also tends to be either prolonged or abbreviated, presumably on the basis of maternal uterine muscle involvement
  - Myotonia is not usually present in the first year of life, and electrical myotonia is rare; therefore, the hallmark of congenital DM1 is hypotonia rather than myotonia
  - Respiratory involvement is common and is the leading cause of death in the neonatal period→it may be so severe that the newborn sustains an asphyxial episode leading to hypoxic brain injury; the encephalopathy is these cases may so dominate the clinical presentation that the underlying myopathy is overlooked
  - neonates with congenital DM1 may be misdiagnosed with hypoxic-ischemic encephalopathy when white matter abnormalities on brain magnetic resonance imaging (MRI), which are commonly seen in DM1, are misattributed to perinatal injury
  - Mechanical ventilation is required for 70 to 80 percent or more of patients

- Gastrointestinal (GI) and feeding difficulties are also common, with many children requiring a nasogastric or gastric feeding tube→the feeding difficulties involve both sucking and swallowing and are related to weakness of the facial, masticatory, and pharyngeal muscles
- A disturbance of gastric motility, probably related to the smooth muscle involvement in DM1, may play a major role in the feeding difficulties in some infants.
- With intensive support, most infants survive the neonatal period, but the overall mortality rate is approximately 15 to 20 percent, and approaches 40 percent in severely affected infants; in severely affected infants, cardiomyopathy may be apparent early on and contribute to neonatal death
- in early childhood, there is often a gradual improvement of motor function→Despite this improvement, some degree of hypotonia and facial weakness persists; at age three to five, foot deformities, learning, and behavioral abnormalities present as the main clinical problems
- Subsequently, pronounced delays are experienced in motor and mental development, with intellectual disability in 50 to 60 percent of children → Intelligence quotient (IQ) scores in the 50 to 65 range are common in congenital DM1 patients who survive the neonatal period; majority of children require assistive services
- There appears to be no correlation between the severity of congenital myotonic dystrophy at birth and the extent of complications during teenage years
    - As patients with congenital DM1 age, they develop many of the symptoms and signs of classic, adult-onset DM1, such as the distal predominance of muscle weakness, myotonia, and electrocardiogram (ECG) abnormalities
    - Those patients who survive early childhood typically experience significant cardiorespiratory morbidity and mortality. Serious cardiac rhythm disturbances may occur as early as the second decade of life in children with the congenital or infantile form of DM1
- The CTG repeat size associated with congenital DM1 is usually >1000
- Inheritance of congenital DM1 is maternal in approximately 90 percent of cases
    - stems from the much greater likelihood for anticipation (ie, expansions of CTG repeats) to occur in maternal compared with paternal transmissions
    - not uncommon for an adult (typically the mother) to be diagnosed with DM only after giving birth to an affected neonate, underscoring the potential for subclinical presentation of this disorder.
- Paternally inherited cases of congenital DM are less common- only 8-12% of cases
- Whereas mothers of infants with maternally transmitted congenital DM have larger mean trinucleotide (CTG) repeat sizes than mothers of patients with childhood or adult-onset disease, fathers of infants with paternally transmitted congenital DM1 have small repeat sizes and/or are asymptomatic at the time of the affected child's birth

- Childhood (infantile) form of DM1 typically presents before the age of 10 years with the involvement of systems and organs other than skeletal muscle
    - In most cases, the initial manifestations are cognitive and behavioral problems such as intellectual impairment with low IQ, attentional deficits, executive dysfunction, anxiety, and mood disorders
    - The CTG repeat size is usually >500 in childhood DM1, although some affected children have a repeat size ≤500
    - Over time, affected children develop muscle symptoms and physical disability that is similar to severe adult-onset classic DM1
    - Serious cardiac rhythm disturbances may occur in asymptomatic adolescents with no or only subtle signs of DM

- The rate of cardiac conduction abnormalities in childhood DM1 is 15 to 20 percent, most commonly atrioventricular block or incomplete bundle branch block
  - Sports and physical exercise precipitate arrhythmias in over one-half of these patients
  - Fewer than 10 percent of patients have clinical evidence of structural heart disease including cardiomyopathy and heart failure

✓ Distal myopathy (or distal muscular dystrophy)
- general term for a group of rare progressive genetic disorders characterized by wasting (atrophy) and weakness of the voluntary distal muscles
- Inheritance is autosomal dominant or recessive
- although age of onset can occur anytime from infancy to adulthood, most forms develop later in life and are slowly progressive →Laing Distal Myopathy (Laing Early-Onset Distal Myopathy; Distal Myopathy 1; MPD1), Inclusion Body Myopathy Type 2 (IBM2; Distal Myopathy with Rimmed Vacuoles (DMRV); Nonaka Myopathy), and Miyoshi Myopathy can present in childhood or adolescence initially

✓ Congenital Muscular Dystrophy
- include muscular dystrophies with onset in the first two years after birth
- Arthrogryposis (contracture of two or more joints at birth) is commonly observed in the newborn period
- The serum creatine kinase (CK) concentration is usually elevated and muscle biopsy is characteristically abnormal with extensive fibrosis, degeneration and regeneration of muscle fibers and proliferation of fatty and connective tissue
- In some cases, the clinical course is static but, in most patients, it progresses very slowly and actual improvement has been observed in a few cases
- original classification of the CMDs was based mainly upon the presence or absence of structural central nervous system abnormalities detected by neuroimaging or at autopsy
  - The absence of structural changes distinguished "occidental" or "classic" CMD from "syndromic" forms of CMD such as Fukuyama muscular dystrophy, Walker-Warburg syndrome, or muscle-eye-brain disease but structural lesions have been described in some cases of classic CMD
  - Likwise, cognitive impairment is a frequent manifestation of CMD, particularly in patients with structural brain lesions and it has also been detected in patients with CMD and normal brain MRI
- The syndromic CMDs are caused by defective post-translational modification of alpha-dystroglycan (dystroglycanopathies) and other proteins, and are caused by mutations in multiple genes
  - Dystroglycanopathies are characterized clinically by the involvement of multiple organ systems, severe brain malformations, and developmental delay
  - Cardiac involvement ranges from absent or mild to severe, and is most often associated with dystroglycanopathies such as Fukuyama type, Walker-Warburg syndrome, and muscle-eye-brain disease
  - Cardiac involvement is also seen in merosin-deficient CMD
  - In addition, cardiac involvement is a common manifestation of the congenital form of limb-girdle muscular dystrophy type 1B (see above discussion in LGMD section)

- Classic form — The identification of mutations within the laminin alpha-2 chain gene (LAMA2; merosin) led to the subclassification of classic CMD into merosin-negative and merosin-positive groups
    - Merosin-deficient CMD (MDC1A), described in Caucasian children, is characterized by a combination of severe CMD, demyelination of the cerebral hemispheres (typically without structural CNS anomalies) and high CK levels
        - The associated mutated gene (LAMA2) was mapped to chromosome 6q22-23 and identified as encoding the alpha-2 chain of laminin, also known as merosin
    - Merosin-positive CMD without structural brain abnormalities usually has a milder phenotype
        - This group is clinically and genetically heterogeneous, and includes classic CMD without distinguishing features, rigid spine syndrome associated with mutations in the selenoprotein N1 gene (SEPN1), CMD with hyperextensible distal joints (Ullrich type), and CMD with intellectual disability or sensory abnormalities
- Ullrich congenital muscular dystrophy and Bethlem myopathy
    - The presence of multiple proximal joint contractures and hyperextensible distal joints in a child with congenital generalized weakness is suggestive of Ullrich congenital muscular dystrophy
        - The course is characterized by a progressive decline in motor and respiratory function in the first decade of life, with a majority confined to wheelchair by 11 years of age
        - The phenotype was originally associated with recessive mutations in type VI collagen genes (COL6A1, COL6A2, and COL6A3), although dominant mutations were subsequently reported
    - Mutations in the same genes also cause Bethlem myopathy, a relatively less severe disorder typically presenting with proximal weakness and flexion contractures involving primarily distal joints (e.g., ankles and interphalangeal joints of the fingers) but also involving the knees, hips, elbows, shoulders, and neck
        - Bethlem myopathy was originally associated with autosomal dominant mutations in COL6A1, COL6A2, and COL6A3 genes, but was later reported in two patients with compound heterozygous COL6A2 mutations and recessive inheritance
    - While Ullrich congenital muscular dystrophy and Bethlem myopathy were once believed to be separate entities, they are now considered to represent opposite ends of a phenotypic spectrum
- Dystroglycanopathies
    - The dystroglycanopathies are associated with mutations in different genes that cause defective post-translational modification of alpha-dystroglycan
    - They are both genetically and phenotypically heterogeneous

        - in current nomenclature, these phenotypes are referred to as the "MDDG" series and include mild to severe forms of congenital muscular dystrophy and mild forms of limb-girdle muscular dystrophy
        - The dystroglycanopathies are characterized by a variety of developmental brain abnormalities, best identified on MRI, including lissencephaly, cerebellar cysts, pontine hypoplasia, and posterior concavity of the brainstem (bowing)
    - Congenital muscular dystrophy-dystroglycanopathy with brain and eye anomalies (type A or MDDGA) includes the more severe phenotypes historically known as Walker-Warburg syndrome and Fukuyama congenital muscular dystrophy, and a milder phenotype called muscle-eye-brain disease
        - Fukuyama type — The Fukuyama type of CMD (MIM 253800) is among the most common autosomal recessive disorders in Japan (0.7 to 1.2 per 10,000 births), and is characterized by hypotonia, generalized weakness, severe developmental delay, seizures, microcephaly, and elevated serum CK levels
            - The electroencephalogram is abnormal in this disorder and shows epileptiform activity
            - Cortical dysgenesis is detected by cerebral CT or MRI
            - The specific lesions are pachygyria and polymicrogyria in the temporal and occipital regions
            - transient T2 hyperintensities appear in the white matter, and hypoplasia of the pons and cerebellar cysts may occur
            - Ocular involvement is limited to simple myopia without structural changes. The locus for the mutated Fukuyama-type (MDDGA4) congenital muscular dystrophy gene (FKTN gene) is located on chromosome 9q31-33
            - The respective protein, fukutin, is secreted outside the cell and may be a component of the extracellular matrix reinforcing muscle membranes
            - Pathologic studies of the brain have suggested that fukutin is a constituent of the basement membrane
            - FKTN mutations have also been associated with severe dilated cardiomyopathy accompanied by a mild form of limb-girdle muscular dystrophy
            - In addition, FKTN gene mutations have been identified in children with an LGMD phenotype and normal intelligence and brain structure designated muscular dystrophy-dystroglycanopathy (limb-girdle) type C4 (MDDGC4)

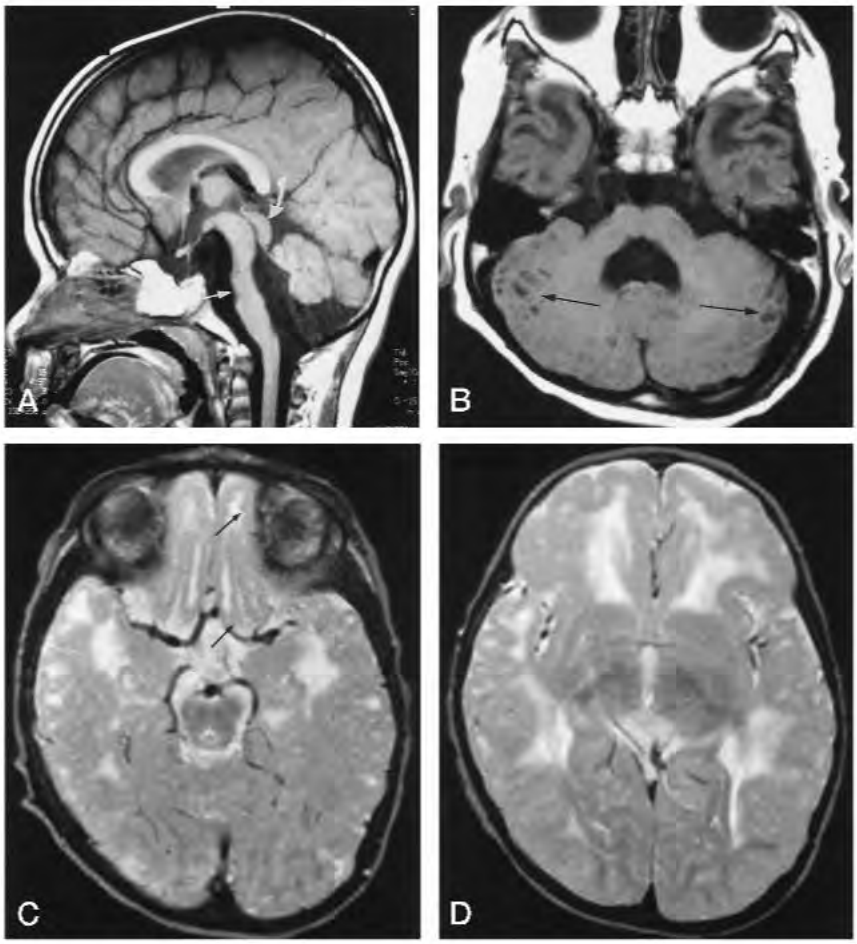


FIG 2. 20-month-old girl with Fukuyama CMD.

A, Sagittal SE (500/15) image shows marked pontine hypoplasia (*straight arrow*), fusion of the midbrain colliculi (*curved arrow*), and hypoplasia of the inferior cerebellar vermian lobules.

B, Axial SE (600/15) image shows low intensity in the temporal white matter, enlarged fourth ventricle associated with vermian hypoplasia, and the cystic type of cerebellar polymicrogyria, with the cysts (*arrows*) primarily in the lateral cerebellar cortex.

C and *D*, Axial (2500/90) images show T2-weighted prolongation of the central cerebral white matter and normal-appearing occipital and temporal gyral patterns. The subcortical white matter is better myelinated. Note that the inferomedial frontal cortex (*arrows, C*) has an irregular cortical-white matter junction, suggesting polymicrogyria.

- Walker-Warburg syndrome — Cerebro-ocular dysplasia or Walker-Warburg syndrome (WWS) is a type of CMD associated with ocular dysplasia, hydrocephalus, and cerebral malformations
    - Ocular abnormalities include cataracts, optic nerve hypoplasia, corneal clouding, and retinal dysplasia or detachment
    - Serum CK concentration is mildly to moderately elevated in this disorder and the electrodiagnostic findings are myopathic
    - Brain MRI shows hypodense white matter, hypoplastic cerebellum and pons, ventricular dilatation (with or without hydrocephalus), and abnormal cortical development known as cobblestone type brain malformation (also called Type II lissencephaly)
    - Other malformations include Dandy-Walker cyst, sometimes associated with posterior encephaloceles
    - The median survival is only four months
    - The Walker-Warburg phenotype is associated with mutations in the POMT1, POMT2, FKTN, FKRP, POMGNT1, LARGE, ISPD, GTDC2, and DAG1 genes
    - WWS associated with POMT1 mutations (MIM 236670) is designated as muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A1 (MDDGA1)

- Other mutations in the POMT1 gene are linked to a milder phenotype of congenital muscular dystrophy with microcephaly and ID, but without the ocular manifestations or structural brain malformations of WWS
- In addition, POMT1 and POMT2 gene mutations have been identified in children with subtypes of autosomal recessive limb-girdle muscular dystrophy (MDDGC1 and MDDGC2)

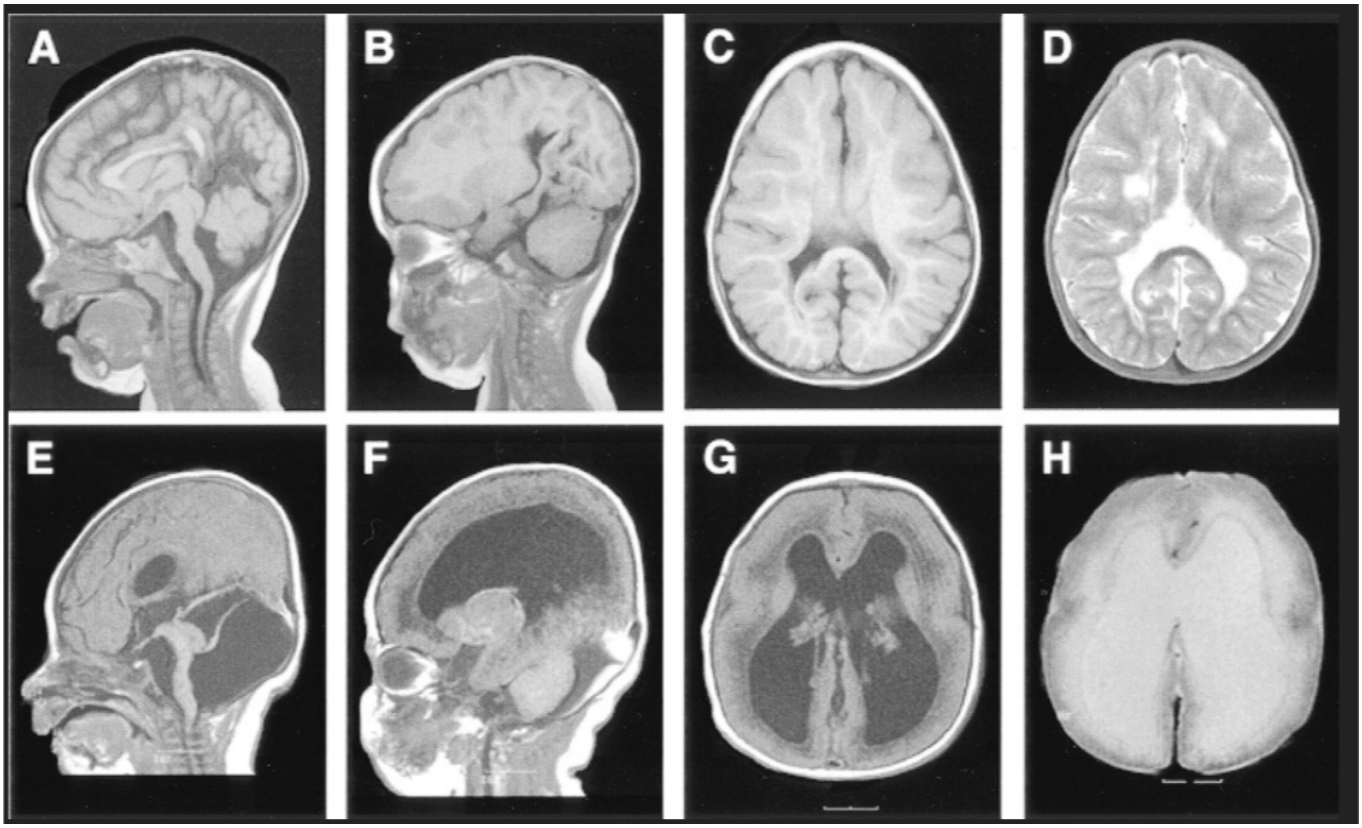
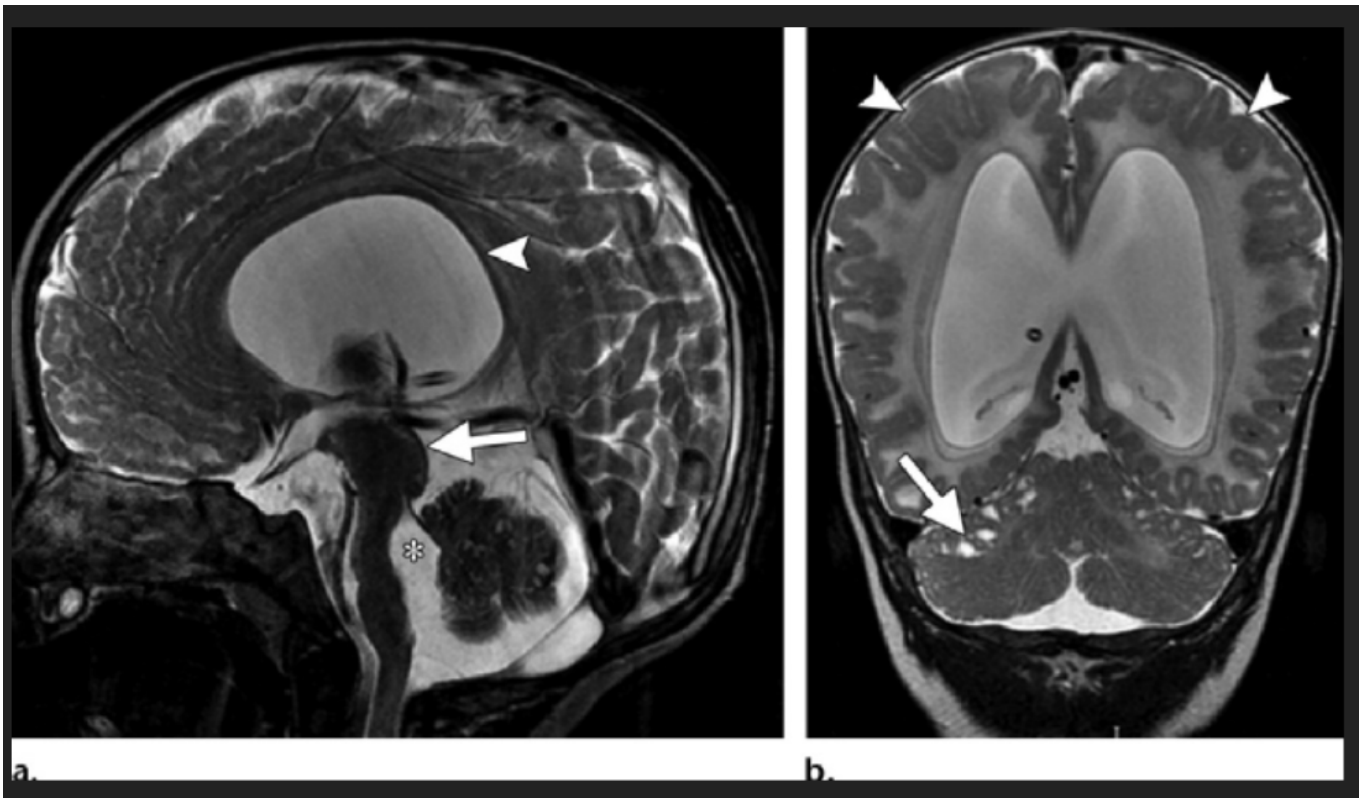


Figure 2. Comparison of MR abnormalities in muscle–eye–brain disease (MEB) (A to D) and Walker–Warburg syndrome (WWS) (E to H). Cranial MR images of patient II-1 (family 18) at age 3 years and patient II-1 (family 12) at age 4 days demonstrate typical MEB and WWS patterns of cobblestone complex. Midline sagittal images (A, E) show an intact although deformed corpus callosum, flat brainstem, and mild cerebellar vermis hypoplasia in MEB (A) compared with a severely hypoplastic corpus callosum, flat brainstem, and severe pancerebellar hypoplasia in WWS (E). Parasagittal images (B, F) show an irregular surface and atypical pachygyria in MEB (B) compared with a pebbled surface resembling lissencephaly in WWS (F). Axial T1-weighted images (C, G) show the same anteroposterior gradient with intermediate (5 to 8 mm) increased thickness of the cortex in MEB (G) compared with an almost smooth surface with no definite gradient, slightly thicker (7 to 10 mm) cortex, and absent or disrupted septum pellucidum in WWS (G). Finally, axial T2-weighted images show patchy increased signal of the white matter in MEB (D) compared with diffuse abnormal signal in WWS (H), although the child with WWS is much younger. The irregular cortical surface and cortical–white matter interface are well seen in both figures, although better in WWS (H).

- Muscle-eye-brain disease — Muscle-eye-brain (MEB) disease has a milder phenotype than WWS → especially prevalent in Finland.
  - Patients with MEB typically present with hypotonia, severe progressive myopia from infancy, and developmental delay

- Pale retina, low or flat electroretinogram, and visual failure related to retinal degeneration develop with advancing age
- Seizures are common and cognitive impairment is often severe
- At approximately five years of age, most patients decline motorically and develop contractures and spasticity
- Laboratory findings in MEB disease include an elevated serum CK level
- Electromyography shows myopathic findings and the electroencephalogram is always abnormal
- Brain MRI shows cobblestone lissencephaly, although it is less severe than in WWS; the brainstem in MEB disease is characteristically flat
- Ventriculomegaly and white matter hypodensities may also be seen
- Visual evoked potentials are delayed and giant (>50 μv) in most patients
- The clinical phenotype of MEB can be caused by mutations in multiple different genes including POMGNT1, FKRP, POMT2, POMT1, FKTN
- POMGNT1-related muscle-eye-brain disease is designated as muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A3 (MDDGA3)



Muscle-eye-brain disease and POMGnT1 mutation in a 5-month-old infant who presented with muscular hypotonia, weakness, intractable seizures, and poor visual function. (a) Sagittal T2- weighted MR image shows hypoplasia of the vermis, flattening of the ventral pons, a dysmorphic tectum and midbrain (arrow), an abnormal concave posterior border of the brainstem (*), an enlarged fourth ventricle, and supratentorial ventriculomegaly (arrowhead). (b) Coronal T2-weighted MR image shows cerebellar hypoplasia, multiple bilateral subcortical cysts in the cerebellar hemispheres (arrow), cerebellar dysplasia, generalized polymicrogyria (arrowheads), abnormal signal intensity of the supratentorial white matter, absence of the septum pellucidum, and marked ventriculomegaly.

- Congenital muscular dystrophy-dystroglycanopathy with or without ID (type B or MDDGB) is also genetically heterogeneous
    - The type B phenotypes of congenital muscular dystrophy-dystroglycanopathy are less severe than the type A but more severe than the type C (limb-girdle muscular dystrophy-dystroglycanopathy)
- Diagnosis — For patients with a clinical presentation that is suspicious for CMD, we suggest an evaluation that includes cranial MRI, eye examination, and molecular genetic testing with next generation sequencing techniques
    - A muscle biopsy for histology and immunostaining is indicated if the molecular genetic testing is negative, but muscle biopsy is not needed if a genetic diagnosis is made
    - The infant with any type of CMD typically presents in the newborn period as a floppy baby, often with arthrogryposis
    - The clinical features are similar to those of an infant with a severe congenital myopathy, a disorder that is more frequent than the rare congenital muscular dystrophies
    - MRI of the brain is useful to look for structural lesions or white matter abnormalities that accompany some CMDs
    - Examination of the eyes is important to exclude an ocular abnormality
    - the infant with a CMD has variably elevated serum CK levels.
    - Even with availability of next generation sequencing techniques, no genetic diagnosis can be made in many cases, suggesting that additional genetic causes of CMD remain to be identified
    - Patients with LMNA-related congenital muscular dystrophy may have prominent inflammatory changes on muscle biopsy and thus be misdiagnosed as having an inflammatory myopathy

2. Understand the long term prognoses for youth with Duchenne and other muscular dystrophies.
- See #1-3 above
3. Know how to plan the laboratory evaluation for a child with progressive muscular weakness.
See #1-3 above in addition to below

- A dystrophinopathy should be suspected in the following situations:
    - Any evidence of delayed motor milestones in a young child with a positive family history of DMD
    - When there is no family history of DMD, a child not walking by 16 to 18 months, or the presence of Gower's sign, toe walking, or calf hypertrophy
    - Unexplained increases in transaminases (e.g., aspartate transaminase and alanine transaminase)
    - Additional symptoms and signs that may be associated with DMD include the following:
        - Poor head control
        - Not running by age 3 years
        - Struggling to hop, climb stairs, or get up from the floor in school-aged children
        - Frequent trips or falls
        - Abnormal gait

        - Muscle pain or cramps
        - Episodes of myoglobinuria
        - Learning difficulties and behavioral issues
        - Speech and language delay
        - Autistic spectrum disorder
- When dystrophinopathy is suspected based on clinical findings, a creatine kinase (CK) level should be obtained and the child should be referred to a neuromuscular specialist
    - An increased CK level is compatible with the diagnosis and should lead to genetic analysis, but a normal CK level makes DMD and BMD unlikely and should prompt an evaluation for alternative diagnoses
    - Serum creatine kinase (CK) concentrations are elevated in children with DMD prior to the appearance of any clinical signs of disease; increased levels are even observed among newborns
        - Serum CK peaks by age 2 years; it is usually 10 to 20 times the upper limit of normal and may be higher
        - these levels then progressively fall at a rate of about 25 percent per year, eventually reaching the normal range in many cases, as more and more muscle is replaced by fat and fibrosis
        - Aldolase levels and other muscle enzymes, such as aspartate transaminase (AST) and alanine transaminase (ALT), are also elevated
    - In most cases, molecular genetic testing can confirm a definitive diagnosis of a dystrophinopathy without recourse to muscle biopsy
    - Given the high frequency of deletions and duplications, it is advisable to pursue "large" deletion/duplication genetic testing first and, if negative, proceed to sequence analysis, which includes not only "small" mutation scanning but also micro deletion/duplication analysis→ diagnosis is established if a disease-causing mutation of the *DMD* gene is identified
    - However, a small percentage of patients with a dystrophinopathy do not have coding region pathogenic variants and, thus, a mutation may be hard to detect
    - When no disease-causing mutation of the *DMD* gene is found and other conditions like spinal muscular atrophy (SMA), inflammatory myopathy, or myopathy of systemic disease have been excluded, genetic testing targeted at certain gene mutations that cause limb-girdle muscular dystrophy (LGMD) or a next generation sequencing-based muscular dystrophy gene panel is the next step because of the phenotypic resemblance between DMD/BMD and certain LGMDs
    - When thorough molecular genetic testing fails to identify a disease-causing mutation, muscle biopsy should be obtained for dystrophin analysis
    - For other less common muscular dystrophies in children, focus on the clinical phenotype and let that direct you in further evaluation→ focus continues to shift to use of molecular genetic testing as gold standard for diagnosis in all muscular dystrophies

Molecular genetic testing used in Duchenne and Becker muscular dystrophy

Gene symbol	Test method	Mutations detected	Mutation detection rate by test method*			
			DMD		BMD	
			Affected males	Heterozygous females ¶	Affected males	Heterozygous females ¶
DMD	Deletion/duplication analysis Δ	Deletion of one or more exons or the whole gene	~50-65%	~50-65%	~65-70%	Unknown
		Duplication of one or more exons	~5-10%	~5-10%	~5-10%	Unknown
	Sequence analysis/mutation scanning ◇	Point mutations (sequence variants) §	~20-35% ¥	~25-35% ‡	~20-30% ¥	Unknown

DMD: Duchenne muscular dystrophy; BMD: Becker muscular dystrophy; CGH: comparative genomic hybridization; PCR: polymerase chain reaction.

* The ability of the test method used to detect a mutation that is present in the indicated gene. The mutation detection rates reflect approximate values.

¶ The mutation spectrum for females heterozygous for a *DMD* mutation resulting in BMD has not been well documented, but is presumed to be similar to the spectrum for males with BMD. Similarly, there are less data about females heterozygous for a *DMD* mutation resulting in DMD, but it is also presumed to be similar to that seen in males with DMD.

Δ Testing that identifies deletions/duplications not readily detectable by sequence analysis of genomic DNA; a variety of methods including quantitative PCR, long-range PCR, multiplex ligation-dependen probe amplification, or targeted array CGH (gene/segment-specific) may be used. A full array CGH analysis that detects deletions/duplications across the genome may also include this gene/segment.

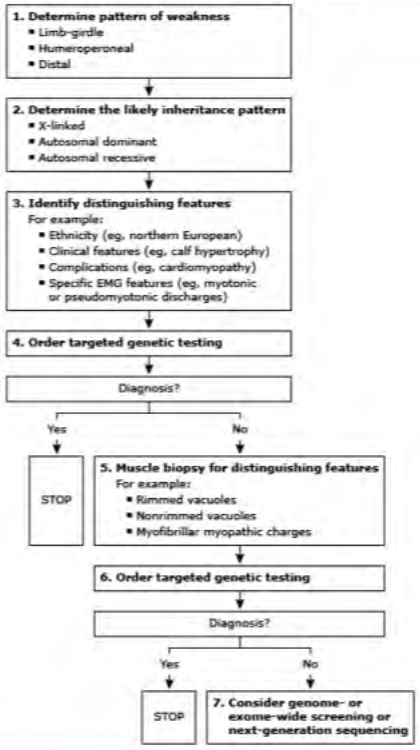
◇ Sequence analysis and mutation scanning of the entire gene can have similar detection frequencies; however, detection rates for mutation scanning may vary considerably between laboratories based on specific protocol used.

§ Examples of mutations detected by sequence analysis may include small intragenic deletions/insertions and missense, nonsense, and splice site mutations.

¥ Lack of amplification by PCR prior to sequence analysis can suggest a putative deletion of one or more exons or the entire X-linked gene in a male; confirmation may require additional testing by deletion/duplication analysis.

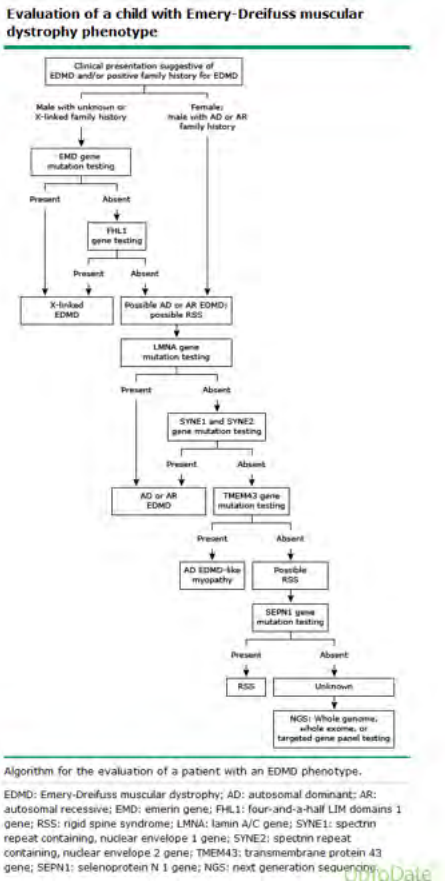
‡ Sequence analysis of genomic DNA cannot detect deletion of one or more exons or the entire X-linked gene in a heterozygous female.

Conceptual approach to a patient with a suspected limb-girdle muscular dystrophy



EMG: electromyography.

From: Narayanaswami P, Weiss M, Selcen D, et al. Evidence-based guideline summary: Diagnosis and treatment of limb-girdle and distal dystrophies: Report of the Guideline Development Subcommittee of the American Academy of Neurology and the Practice Issues Review Panel of the American Association of Neuromuscular & Electrodiagnostic Medicine. Neurology 2014; 83:1453. DOI: 10.1212/WNL.0000000000000892.

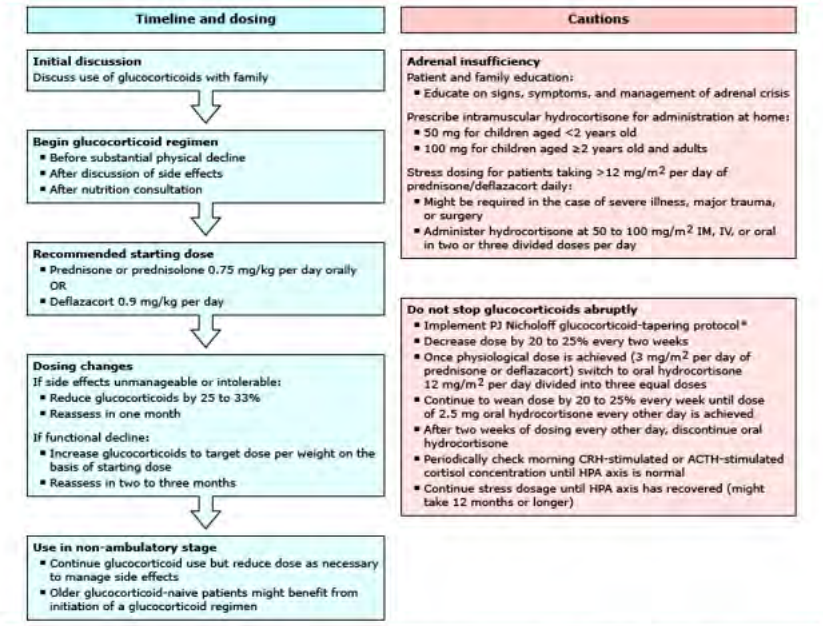


4. Understand the genetics of Duchenne and other muscular dystrophies. – see responses in #1-3 above.

5. Know how to plan the management of a boy with Duchenne and other muscular dystrophies.

- Up until recently there were really no treatments available that could stop or reverse the course of muscular dystrophies
- Broad categories of treatment focus on support of symptoms and maintaining motor and adaptive functioning→ may include physical therapy, occupational therapy, respiratory therapy and respiratory support, speech therapy, feeding therapy, orthopedic appliances used for support, pacemaker for cardiac support, assistive technology for help with communication and other adaptive functions, and corrective orthopedic surgery
- Medications used to treat muscular dystrophy are also usually focused on treating secondary medical conditions (i.e. anti-epileptic drugs in patients who also have seizures, immunosuppressants to delay some damage to dying muscle cells, and antibiotics to fight respiratory infections) and for improving overall motor function and adaptive functioning- as in use of glucocorticoids for Duchenne's and Becker's to slow muscular deterioration→
- glucocorticoids have long been a mainstay in treatment of DMD and BMD in their ability to improve motor function and pulmonary function, reducing the risk of scoliosis, delaying the loss of ambulation, and possibly for delaying progression of cardiomyopathy and improving survival→started early before signs of damage apparent (see below)

Care considerations for glucocorticoid (steroid) initiation and use for patients with Duchenne muscular dystrophy

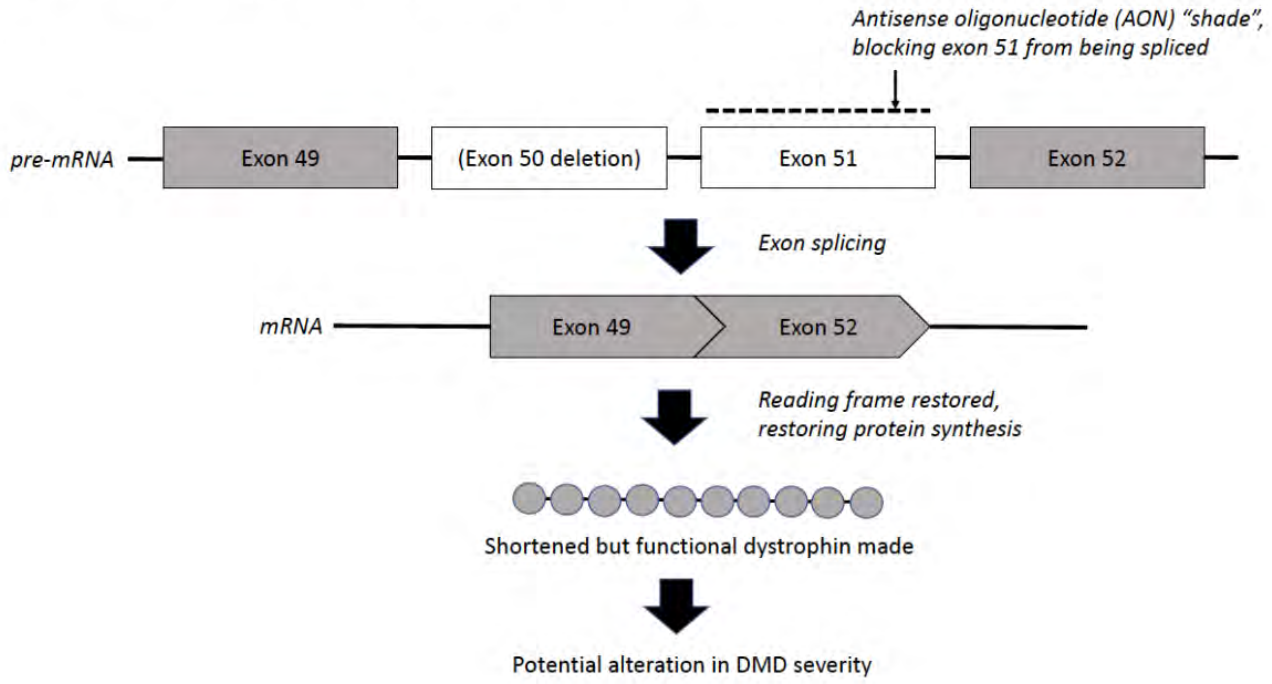


CRH: corticotropin-releasing hormone; ACTH: adrenocorticotropic hormone; HPA: hypothalamic-pituitary-adrenal; IM: intramuscular; IV: intravenous.
* The PJ Nicholoff tapering protocol is available online.

Original figure modified for this publication. From: Birnkrant DJ, Bushby K, Bann CM, et al. Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and neuromuscular, rehabilitation, endocrine, and gastrointestinal and nutritional management. Lancet Neurol 2018; 17:251. Illustration used with the permission of Elsevier Inc. All

- Novel disease modifying therapies available now and under active investigation for DMD primarily
    - Golodirsen - FDA approval in December 2019 to treat DMD for patients who have a confirmed mutation of the dystrophin gene that is amenable to exon 53 skipping
        - Estimated that approximately 8% of patients with DMD have this mutation
        - antisense oligonucleotide that is designed to modify the splicing of exon 53 of dystrophin pre-messenger RNA, resulting in exon 53 skipping in patients with amenable pathogenic variants of the dystrophin gene→increased dystrophin production
        - end note→clinical benefit not clearly proven yet over risk of harms →ongoing clinical trial still being conducted
    - Eteplirsen — granted accelerated approval in September 2016 by FDA for treatment of patients with DMD who have a confirmed mutation of the dystrophin gene amenable to exon 51 skipping- this mutation is present in 13% of patients with DMD; accelerated approval with some controversy→FDA can still remove approval if further clinical study does not indicate clear benefits over harms

Figure 1b. Exon-Skipping Therapy Leading to Shortened but Functional Dystrophin Production



- Ataluren — Ataluren (PTC124) is an orally administered drug being developed for the treatment of genetic defects caused by nonsense (stop) mutations

    - promotes ribosomal read-through of nonsense (stop) mutations, allowing bypass of the nonsense mutation and continuation of the translation process to production of a functioning protein
    - This approach could benefit the estimated 10 to 15 percent of patients with DMD/BMD who harbor nonsense (stop) mutations
    - Only licensed in certain countries (EU, UK); not currently in the U.S.→in these countries where approved, it is an option to treat patients age 2 years and older with DMD caused by nonsense mutations

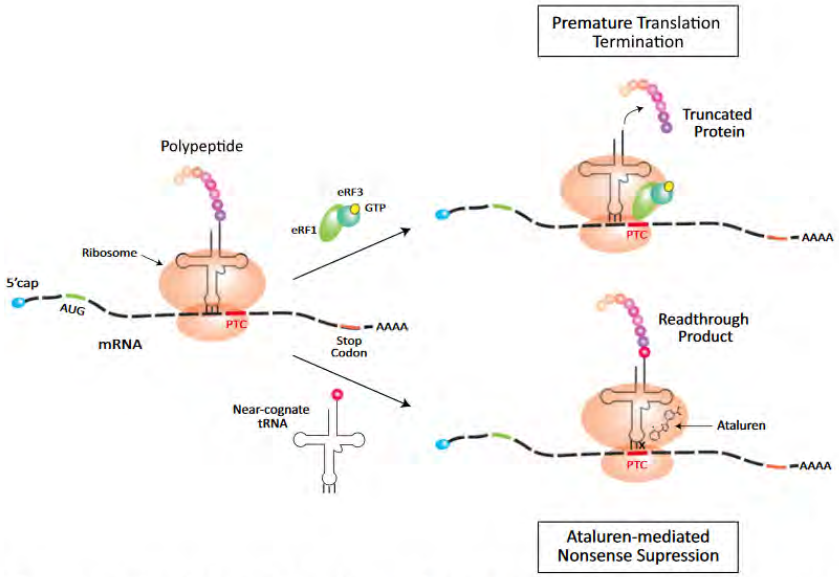


Fig. 1. Schematic diagram illustrating ataluren's proposed mechanism of action. In normal circumstances, ribosomes move along the mRNA linking amino acids into protein until arriving at the stop codon. When a ribosome encounters a premature stop codon (PTC) due to a nonsense mutation, eukaryotic release factors [eRF1 (green) and eRF3 (turquoise) in complex with GTP (yellow)] are recruited and translation is terminated prematurely, generating a truncated protein. Ataluren is proposed to interact with the ribosome and facilitate the recruitment of near-cognate tRNAs, which supresses the nonsense mutation and allows for the readthrough of a PTC and synthesis of a full-length protein. The amino acid colored in red on the near-cognate tRNA is incorporated into the readthrough protein product. The X on the tRNA in the ataluren-mediated nonsense suppression model indicates mispairing at codon position three. The 5'cap is shown in blue, ribosomes are shown in brown, and amino acids in multi-colors on the polypeptide.

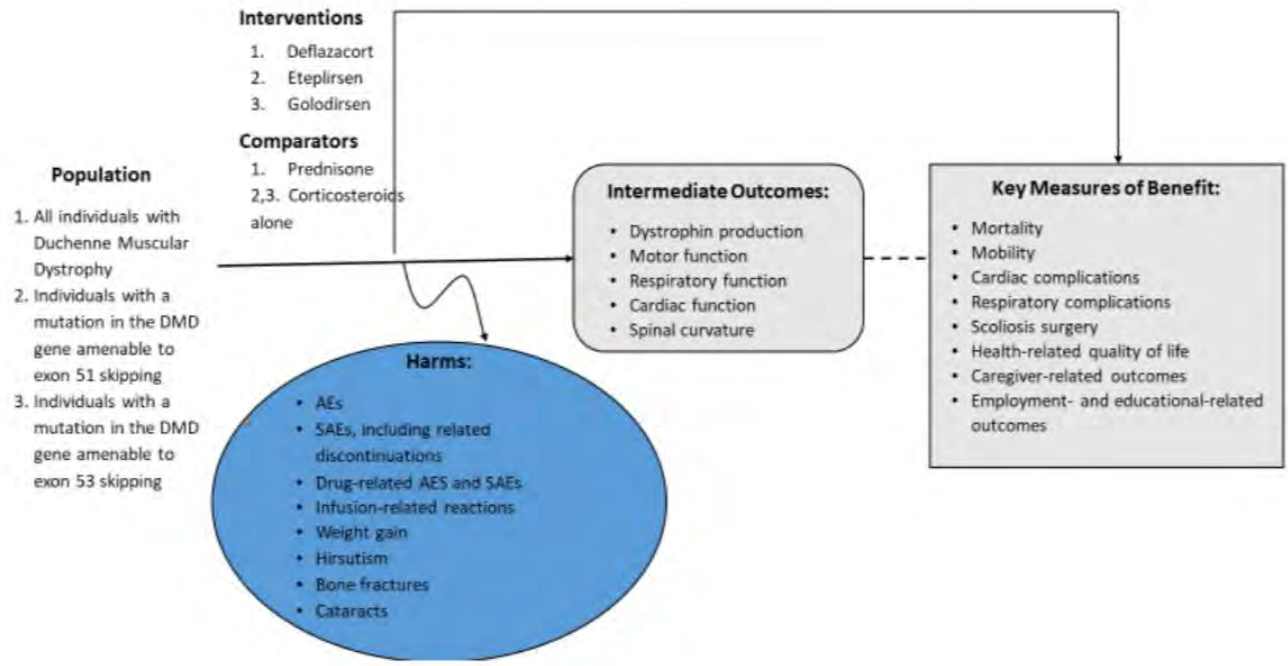
- Other investigational therapies — Investigational therapies for DMD and BMD include gene therapy, creatine, myostatin inactivation, skeletal muscle progenitors, and idebenone
  - Gene therapy
    - Preliminary clinical studies are evaluating systemic gene transfer by intravascular administration of recombinant adeno-associated viral (rAAV) vectors that carry microdystrophin or minidystrophin genes→major hindrance has been that cellular immunity may be an obstacle to successful dystrophin transgene therapy
    - Discovery of pre-existing T-cell immunity to dystrophin may be caused by priming of the cellular immune system to revertant dystrophin myofibers that express truncated dystrophin protein and are present in low levels in most patients with DMD

  - **Creatine** — Creatine monohydrate has been studied for its potential to increase muscle strength in neuromuscular disorders and muscular dystrophies
    - **There has been some evidence based medicine to suggest that creatine treatment was associated with improved grip strength of the dominant hand and increased fat free mass**

    - **Beneficial effects of creatine were independent of corticosteroid use→that being said, creatine treatment was not associated with actual significant improvement on functional measures or ADL's**
    - **All things considered, when weighing benefits versus risks in terms of CAM→ would still consider creatine still possible benefit and low risk when counseling parents (as long as it is used in dosages recommended in current medical evidence) in terms of CAM**
- **Myostatin inactivation** — Myostatin is a protein that has an inhibitory effect on muscle growth; animal models thus far have demonstrated that when expressing the DMD phenotype but lacking myostatin they have an increased muscle mass compared with those with a wild-type myostatin gene
    - Antibodies to myostatin also have a beneficial effect; treated animals have increased muscle mass, strength, lower serum creatine kinase, and less histologic evidence of muscle damage
    - Clinical trials of myostatin inhibitors are underway

- **Cell therapy** — The use of skeletal muscle progenitors in the treatment of DMD and BMD continues to be under investigation but remains experimental
- **Idebenone** — analogue of coenzyme Q_{10} (CoQ10), the lipophilic electron carrier and endogenous antioxidant found in all cellular mitochondrial membranes→ antioxidant drug used in experimental treatment of a multitude of other medical diagnoses (most notably, Alzheimer's)
    - Its application and use in DMD treatment has been more in reference with treatment of cardiac and respiratory complications

Figure 2. Analytic Framework: Therapies for DMD



AE: adverse event, SAE: serious adverse event

6. Understand the neurological and orthopedic complications associated with muscular dystrophy- see #1-3 above

7. Know the developmental and behavioral characteristics of individuals with muscular dystrophy – see #1-3 above

D. Other

1. Plan the evaluation of an infant with hypotonia.

- Initial first step is to be able to clinically differentiate between hypotonia and weakness
    - **Hypotonia** is described as reduced resistance to passive range of motion in joints→ability to sustain postural control and movement against gravity whereas **weakness** is reduction in the maximum power that can be generated.
    - Thus, floppy infants exhibit poor control of movement, delayed motor skills, and hypotonic motor movement patterns.
        - The abnormal motor patterns include alterations in postural control, increased range of motion of joints, and abnormal stability and movement mechanics.
        - A weak infant will always have hypotonia, but not all infants with hypotonia will have weakness
- Generating a differential diagnosis can be overwhelming, but most important delineation to help break the differential down is determining whether it is a central cause (UMN) or peripheral cause (LMN)
    - Need to utilize neurological examination skills to be able to help you pinpoint to what level of the nervous system is being affected and causing the hypotonia

- Central causes, both acute and chronic, are more common than are peripheral disorders→60-80% central; 15-30% peripheral
    - Central conditions include hypoxic-ischemic encephalopathy, other encephalopathies, brain insult, intracranial hemorrhage, chromosomal disorders, congenital syndromes, inborn errors of metabolism, and neurometabolic diseases.
    - Peripheral disorders include abnormalities in the motor unit, specifically in the anterior horn cell (ie, spinal muscular atrophy), peripheral nerve (ie, myasthenia), neuromuscular junction (ie, botulism), and muscle (ie, myopathy).
    - The most common central cause of hypotonia is cerebral palsy or hypoxic encephalopathy in the young infant.
        - However, this dysfunction may progress in later infancy to hypertonia.
    - The most common neuromuscular causes, although still rare, are congenital myopathies, congenital myotonic dystrophy, and spinal muscular atrophy.
    - Some disorders cause both central and peripheral manifestations, such as acid maltase deficiency/ alpha glucosidase deficiency (Pompe disease), congenital myopathy, congenital myotonic dystrophy, some mitochondrial and peroxisomal disorders

Central (most common)	▪ Hypoxic ischaemic encephalopathy ▪ Intracranial haemorrhage ▪ Cerebral malformations ▪ Chromosomal abnormalities (e.g.Trisomy 21, Prader-Willi syndrome) ▪ Congenital infections (TORCH) ▪ Acquired infections ▪ Peroxisomal disorders ▪ Drug effects (e.g. benzodiazepines)
Spinal cord	▪ Birth trauma (especially Breech delivery) ▪ Syringomyelia
Anterior Horn Cell	▪ Spinal Muscular Atrophy ▪ Pompe's disease (acid maltase deficiency)
Neuromuscular junction	▪ Myasthenia gravis (transient/ congenital) ▪ Infantile botulism
Muscle	▪ Muscular dystrophies (inc. congenital myotonic dystrophy) ▪ Congenital myopathies (e.g. central core disease)
Peripheral nerves	▪ Hereditary motor and sensory neuropathies
Metabolic myopathies	▪ Acid maltase deficiency ▪ Carnitine deficiency ▪ Cytochrome-*c*-oxidase deficiency

Table 1. **Differential Diagnosis of Neuromuscular Disorders Presenting in Newborns**

Anterior Horn Cell Disorders
• Acute infantile spinal muscular atrophy
• Traumatic myelopathy
• Hypoxic-ischemic myelopathy
• Neurogenic arthrogryposis
• Infantile neuronal degeneration
Congenital Motor or Sensory Neuropathies
• Hypomyelinating neuropathy
• Congenital hypomyelinating neuropathy
• Charcot-Marie-Tooth disease
• Dejerine-Sottas disease
• Hereditary sensory and autonomic neuropathy
Neuromuscular Junction Disorders
• Transient acquired neonatal myasthenia
• Congenital myasthenia
• Magnesium toxicity
• Aminoglycoside toxicity
• Infantile botulism
Congenital Myopathies
• Nemaline myopathy
• Central core disease
• Myotubular myopathy
• Congenital fiber type disproportion myopathy
• Multicore myopathy
Muscular Dystrophies
• Dystrophinopathies
• Congenital muscular dystrophy with merosin deficiency
• Congenital muscular dystrophy without merosin deficiency
• Congenital muscular dystrophy with brain malformations or intellectual disability
• Walker-Warburg disease
• Muscle-eye-brain disease
• Fukuyama disease
• Congenital muscular dystrophy with cerebellar atrophy/ hypoplasia
• Congenital muscular dystrophy with occipital argyria
• Early infantile facioscapulohumeral dystrophy
• Congenital myotonic dystrophy
Metabolic and Multisystem Diseases
• Disorders of glycogen metabolism
• Acid maltase deficiency
• Severe neonatal phosphofructokinase deficiency
• Severe neonatal phosphorylase deficiency
• Debrancher deficiency
• Primary carnitine deficiency
• Peroxisomal disorders
• Neonatal adrenoleukodystrophy
• Cerebrohepatorenal syndrome (Zellweger)
• Disorders of creatine metabolism
• Mitochondrial myopathies
• Cytochrome-c oxidase deficiency

- **Central hypotonia** – Central hypotonia is generally characterized by the following:
    - Tone is reduced relatively more than is muscle strength
    - Hypotonia is typically mild to moderate
    - Limbs typically retain antigravity power
    - There may be some head lag
    - Deep tendon reflexes are present (may be decreased or increased)
    - Positive Babinski sign
    - Persistent neonatal reflexes

- If the pattern of hypotonia is chiefly central, a central nervous system disorder is most likely (eg, cerebral palsy or Leigh syndrome)
    - The latter is predominantly a central nervous system disorder, though some patients may have associated myopathy and peripheral neuropathy
- **Peripheral hypotonia** – Peripheral hypotonia is generally characterized by the following:
    - Weakness is the most significant finding

    - Absent or extremely reduced antigravity movements
    - Significant head lag
    - Absent deep tendon reflexes
    - Negative Babinski sign
    - Absence of neonatal reflexes
- If the pattern of weakness and hypotonia is predominantly peripheral, a neuromuscular disorder is most likely, but many exceptions can occur
    - i.e. Prader-Willi syndrome (primarily central, but presents with both profound hypotonia and weakness)
    - infants with a mild congenital myopathy can retain antigravity power in the limbs

Table 2. **Localization of Disorders Producing Hypotonia**

Variable	Central Injury	Central Developmental	Anterior Horn Cell	Peripheral Nerve	Neuromuscular Junction	Muscle
Strength	Normal or slight weakness	Normal or slight weakness	Weakness	Weakness	Weakness	Weakness
Deep tendon reflexes	Normal to increased	Normal	Decreased	Decreased	Normal to decreased	Decreased to absent
Babinski sign	+/−	+/−	Absent	Absent		
Infantile reflexes	Persistent	Persistent/Absent	Absent	Absent	Absent	Absent
Muscle fasciculations	Absent	Absent	Prominent	Absent	Absent	Absent
Muscle mass	Normal or disuse atrophy	Normal or disuse atrophy	Prominent atrophy (proximal)	Distal atrophy	Normal or decreased	Proximal atrophy; increased or decreased distal pseudohypertrophy
Sensation	Normal	Normal	Normal	Increased or decreased	Normal	Normal
Tone	Decreased evolving to increased	Decreased	Decreased	Decreased	Decreased or normal	Decreased

- First step in evaluation, therefore, must be collecting a thorough history and doing a comprehensive physical examination
    - Family and PMH points to focus on:
        - prenatal, perinatal, and neonatal assessment
            - The prenatal history should include information on fetal movement in utero, fetal presentation, and the amount of amniotic fluid present.
            - The obstetric history occasionally may identify both a cause and the timing of onset.
            - Maternal exposures to toxins or infections suggest a central cause.
            - Low Apgar scores may suggest floppiness from birth, and a hypotonic newborn should be considered septic until proven otherwise.
            - A term infant who is born healthy but develops floppiness after 12 to 24 hours may have an inborn error of metabolism.
            - Cervical spinal cord trauma is a complication of a breech delivery or cervical presentation and can present with hypotonia, with other neurologic signs appearing days to weeks later.
            - The diagnosis of myotonic dystrophy in a floppy newborn is suggested by a history of uterine dystonia and a difficult delivery and even an examination of the mother can be important in suspected cases of congenital myotonic dystrophy (inability to relax the hand)

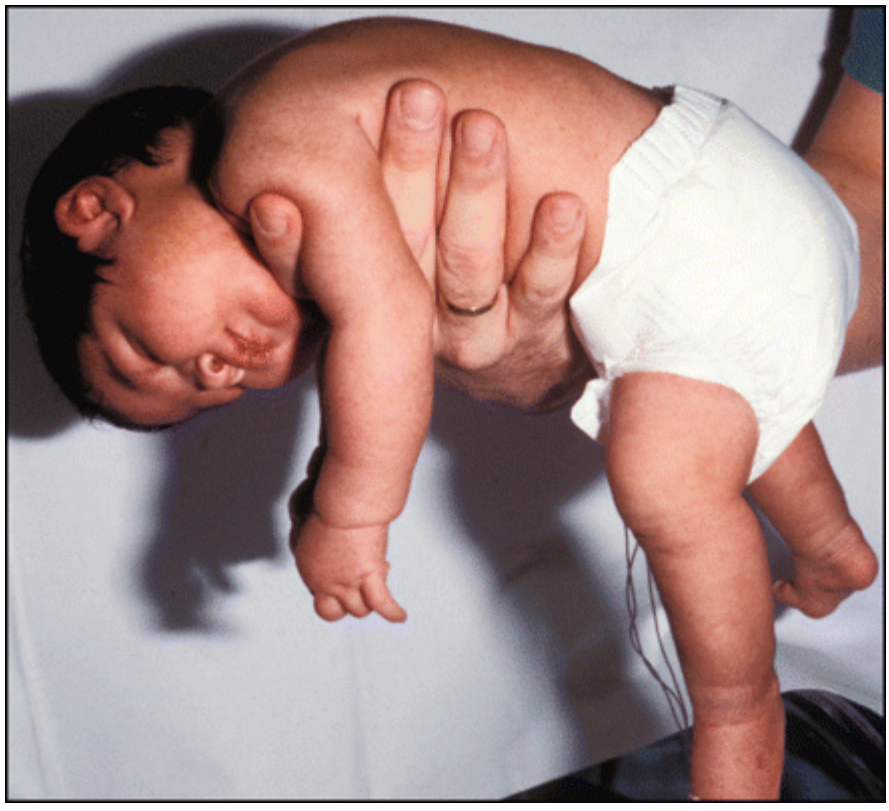
- After the newborn period, the course of floppiness can be revealing. Central congenital hypotonia does not worsen with time but may become more readily apparent, whereas infants suffering central injury usually develop increased tone and deep tendon reflexes

- A family history of neuromuscular abnormalities may be informative because many disorders are inherited. Examples of familial neuromuscular diseases include:

  - Congenital myotonic dystrophy
  - Spinal muscular atrophy
  - Familial dysautonomia
  - Many metabolic disorders (eg, disorders of mitochondrial metabolism, acid alpha-glucosidase deficiency [Pompe disease]

- The physical examination should include:

  - the assessment of pertinent clinical features (e.g. the presence of fixed deformities as in myotonic dystrophy or presence of arthrogryposis in myasthenia syndromes– see below)
  - careful evaluation for any dysmorphic features to further direct any genetic testing

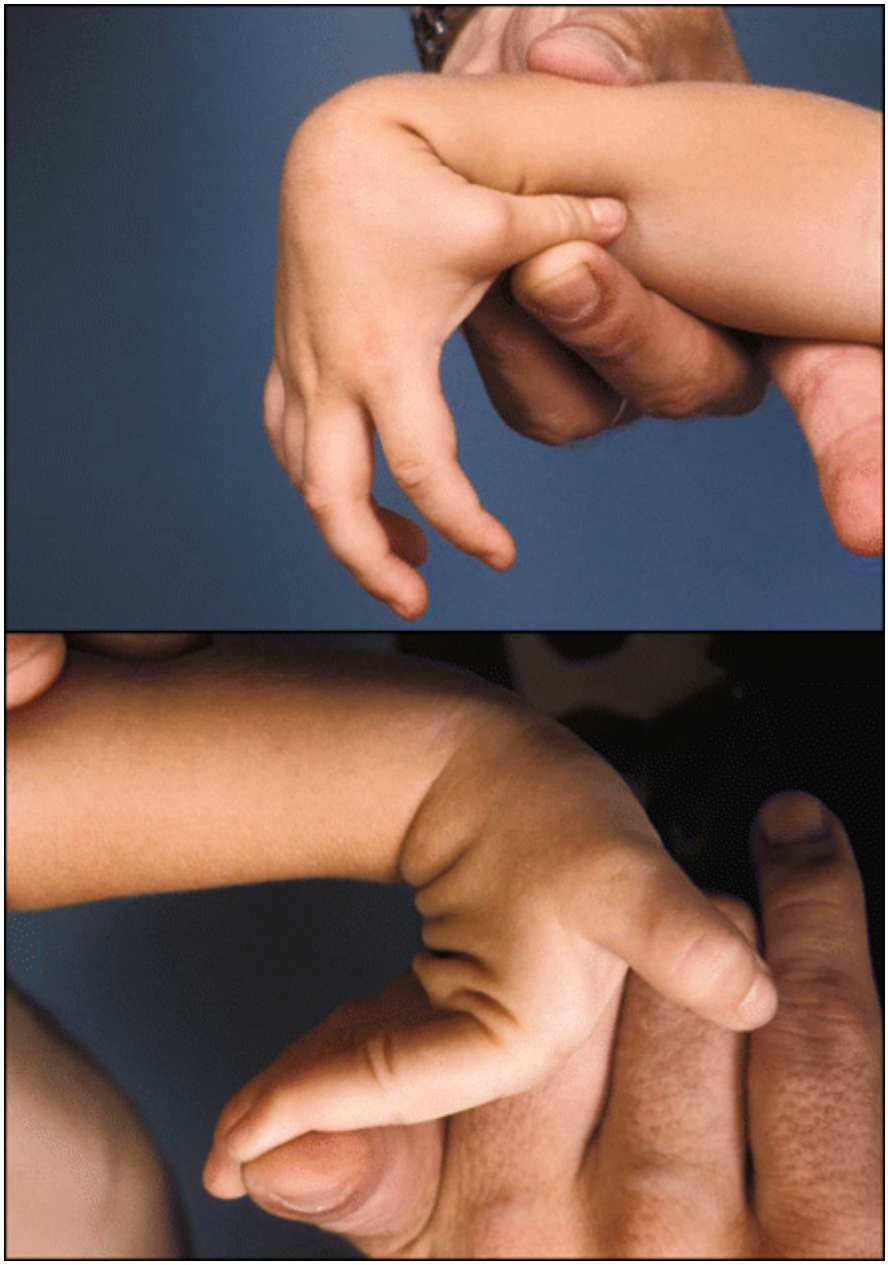


  - a comprehensive neurologic evaluation examining tone, strength, and reflexes

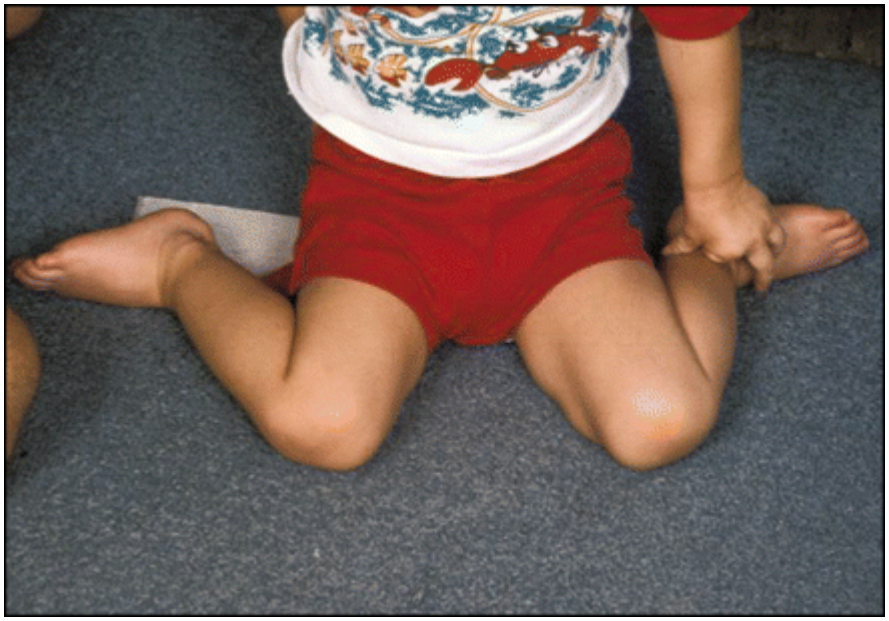
- to begin assessing tone, a clinician should examine an infant's head size and shape, posture, and movement
- floppy infant often lies with limbs abducted and extended.
- plagiocephaly frequently is present in older infants
- additional techniques for positioning and examining tone include horizontal and vertical suspension and traction
    - to demonstrate decreased tone, an infant is suspended in the prone position with the examiner's palm underneath the chest (horizontal suspension)→the head and legs hang limply, forming an inverted “U” posture (see picture above)
    - an infant who has hypotonia “slips through” at the shoulders when the examiner grasps him or her under the arms in an upright position (vertical suspension) – see below

    - Head lag or hyperextension is evident when the infant is pulled by the arms from a supine to sitting position (traction)

- Other pertinent findings may include poor trunk extension, astasias (inability to stand due to muscular incoordination) in supported standing, decreased resistance to flexion and extension of the extremities (see pictures below of hyperlaxity of finger and wrist joints), exaggerated hip abduction, and exaggerated ankle dorsiflexion



- Abnormalities in stability and movement may manifest in an older infant as a combat crawl, W-sitting (see picture below), a wide-based gait, genu recurvatum, and hyperpronation of the feet

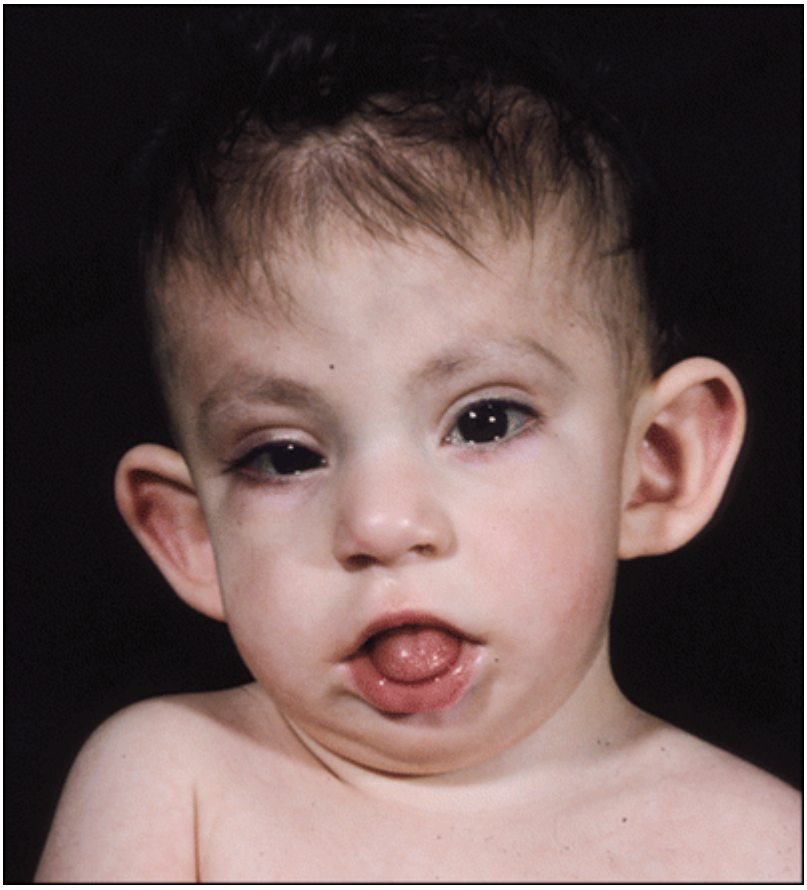


- In addition, the child who has hypotonia may exhibit oral-motor dysfunction, poor respiratory support, and gastroesophageal reflux.
- Deep tendon reflexes (DTRs) often are hyperactive in central conditions, and clonus and primitive reflexes persist while DTRs are normal, decreased, or absent in peripheral disorders
- Hypotonia also may manifest in the face (hypotonic facies- see pictures)

    - Weakness also can manifest as decreased strength and frequency of spontaneous movements
        - A complete assessment for weakness includes evaluating for cry, suck, facial expressions, antigravity movements, resistance to strength testing, and respiratory effort
        - Infants who can generate a full motor response when aroused are more likely to be hypotonic than weak
        - The distribution and course of weakness is crucial to note, that is, if the face is spared versus the trunk and extremities.
    - Important to note if hypotonia is fluctuating, static, or progressive
        - This differentiation discriminates whether you have a static encephalopathy (as is seen in intellectual disability) versus a degenerative neurologic condition (e.g., spinal muscular atrophy)
- Important to consider the heritable disorders associated with hypotonia- especially the more common ones- when launching an initial evaluation (see chart below)
    - Trisomy 21- most common chromosomal abnormality causing developmental disability
        - characterized by hypotonia, intellectual disability, and congenital heart defects (in 50%)
        - particular physical features in the neonate include flattened posterior occiput with brachycephaly, flat facial profile and nasal bridge, upslanting palpebral

fissures, small or anomalous ears, short neck with excess nuchal folds, single transverse palmar creases, hypoplasia of the midphalanx of the fifth digit with clinodactyly, joint hyperextensibility, dysplasia of the pelvis, and a poor Moro reflex
        - high-resolution chromosomal study is diagnostic for most patients
        - if chromosomes are normal on high resolution and concern remains, a trisomy screen or FISH testing should be requested for rare cases of partial mosaic trisomy (only 2% of cases)
- Fragile X syndrome – leading cause of inherited intellectual disability
    - characterized by mild-to-profound intellectual disability, autistic features, macrocephaly, large ears, epicanthal folds, a thickened nasal bridge, and increased testicular size in puberty
    - An expansion of a trinucleotide repeat (CGG) in the promoter region of the *FMR1* gene at Xq27.3 is responsible for the phenotype – diagnosis is based on molecular genetic testing of FMR1 gene
    - Affected individuals have more than 200 repeats
    - Premutation carriers (55-200 repeats) also can be detected with molecular genetic testing
    - Although hypotonia generally is a feature during infancy, it is mild, and most children who have fragile X syndrome are not diagnosed early in life until a delay in developmental milestones is detected or emergence of autistic features (or both)
- Prader-Willi syndrome- most common genetic cause of potentially life-threatening childhood obesity
    - characterized by hypotonia, hypogonadism, intellectual disability, and short stature as well
    - affected patients present at birth with profound hypotonia and feeding problems until 8 to 11 months of age, when they develop low-normal muscle tone and insatiable appetites
    - prominent physical features during childhood include a narrow bifrontal diameter, strabismus, almond-shaped eyes, enamel hypoplasia, and small hands and feet
    - the genetic abnormality in 75% of patients is a deletion of the long arm of chromosome 15 at q11-q13→ in these cases, the paternally derived chromosome has been deleted
    - Maternal uniparental disomy accounts for an additional 20% of cases
    - The remaining 5% are due to a mutation of the imprinting center or to a chromosomal translocation. Methylation analysis can detect all three molecular defects. If the methylation study result is abnormal, a FISH study can be used to define the diagnosis further.
- Kabuki syndrome is a rare multiple congenital anomaly syndrome
    - Associated with hypotonia and feeding problems and is characterized by specific facial features (see picture below)
        - distinctive facial features including arched eyebrows; long eyelashes; blue sclerae; long openings of the eyelids (long palpebral fissures) with the lower lids turned out (everted) at the outside edges; a flat, broadened tip of the nose; and large protruding earlobes

    - also commonly have dental abnormalities and cleft palate
- Typically with mild-to-moderate intellectual disability, postnatal growth delay, skeletal abnormalities (scoliosis and short fifth fingers, or problems with the hip and knee joints)
- possibility of accompanying seizure disorders, nystagmus, and esotropia
- Also have unusual dermatoglyphic patterns with prominent finger pads →called fetal finger pads because they normally occur in human fetuses; in most people they disappear before birth
- cardiac and renal defects, frequent ear infections (otitis media), hearing loss, and early puberty can also occur
- In the absence of major birth defects, this syndrome is difficult to recognize in neonates. No definitive genetic test or mechanism of inheritance has been determined, but research is ongoing.
- Kabuki syndrome is caused by mutations in the KMT2D gene (also known as *MLL2*) or the KDM6A gene.
    - Between 55 and 80 percent of cases of Kabuki syndrome are caused by mutations in the KMT2D gene. This gene provides instructions for making an enzyme called lysine-specific methyltransferase 2D that is found in many organs and tissues of the body. Lysine-specific methyltransferase 2D functions as a histone methyltransferase. Histone methyltransferases are enzymes that modify proteins called histones. Histones are structural proteins that attach (bind) to DNA and give chromosomes their shape. By adding a molecule called a methyl group to histones (a process called methylation), histone methyltransferases control (regulate) the activity of certain genes. Lysine-specific methyltransferase 2D appears to activate certain genes that are important for development.
    - Between 2 and 6 percent of cases of Kabuki syndrome are caused by mutations in the KDM6A gene. This gene provides instructions for making an enzyme called lysine-specific demethylase 6A. This enzyme is a histone demethylase, which means that it helps to remove methyl groups from certain histones. Like lysine-specific methyltransferase 2D, lysine-specific demethylase 6A regulates the activity of certain genes, and research suggests that the two enzymes work together to control certain developmental processes.
    - The KMT2D and KDM6A gene mutations associated with Kabuki syndrome lead to the absence of the corresponding functional enzyme. A lack of the enzymes produced from these genes disrupts normal histone methylation and impairs proper activation of certain genes in many of the body's organs and tissues, resulting in the abnormalities of development and function characteristic of Kabuki syndrome.
    - Some people with Kabuki syndrome have no identified KMT2D or KDM6A gene mutation. The cause of the disorder in these individuals is unknown.



- X-linked intellectual disability (formally known as X-linked intellectual disability (XLMR))
  - Hypotonia in infancy and developmental delays are common manifestations in X-linked ID
  - Affected children typically present with decreased muscle tone early in life, and striking progression to spasticity may occur
  - Thirty syndromes exist in which infantile hypotonia is associated with X-linked ID

  - Some of the X-liked genetic syndromes connected with ID:
    - Coffin–Lowry syndrome
    - MASA syndrome
    - MECP2 duplication syndrome
    - X-linked alpha thalassemia intellectual disability syndrome (see below)
    - intellectual disability and microcephaly with pontine and cerebellar hypoplasia
  - estimated there are ~200 genes involved in this syndrome; of these ~100 have been identified

  - Many of these genes are found on the short 'p' arm of the chromosome, and duplications at Xp11.2 are associated with the syndromic form of the condition.

- X-linked intellectual disability accounts for ~16% of all cases of intellectual disability in males
- Early diagnosis often is difficult because distinctive syndromic findings may be absent in the early years and only hypotonia and developmental delays may exist. One such disorder is the ATRX syndrome (X-linked alpha thalassemia-intellectual disability), which is associated with hypotonic facies and developmental delays where *XNP* is the causative gene.

- Myotonic dystrophy is a multisystem disorder transmitted by autosomal dominant inheritance and caused by an unstable DNA trinucleotide repeat on chromosome 19 that can expand in successive generations
    - Symptoms usually begin in young adult life and include weakness of the face and distal limb muscles, cataracts, multiple endocrinopathies, frontal baldness in males, and myotonia (see figure below)
    - Congenital myotonic dystrophy (Steinert disease) can afflict infants born to affected mothers.
    - Polyhydramnios is common, labor is prolonged, and delivery usually requires mechanical assistance.
    - Severely affected infants have inadequate diaphragm and intercostal muscle function and require assisted mechanical ventilation.
    - Perinatal asphyxia can be a consequence of a prolonged and difficult delivery and resuscitation.
    - Facial diplegia, generalized muscular hypotonia, joint deformities, gastrointestinal dysfunction, and oral motor dysfunction can occur.
    - Affected infants have a characteristic facial appearance, with tenting of the upper lip, thin cheeks, and wasting of the temporalis muscles.
    - They also tend to have dislocated hips, arthrogryposis, and club feet.
    - Limb weakness is proximal, tendon reflexes usually are absent, and myotonia may not be elicited on electromyography (EMG)
    - Intellectual disability is common
    - Cardiomyopathy contributes to early death, and the long-term prognosis is poor
    - Respiratory failure and an increased risk of aspiration also lead to early death. If the infant survives the first 3 postnatal weeks, motor function may improve, although facial diplegia usually persists.

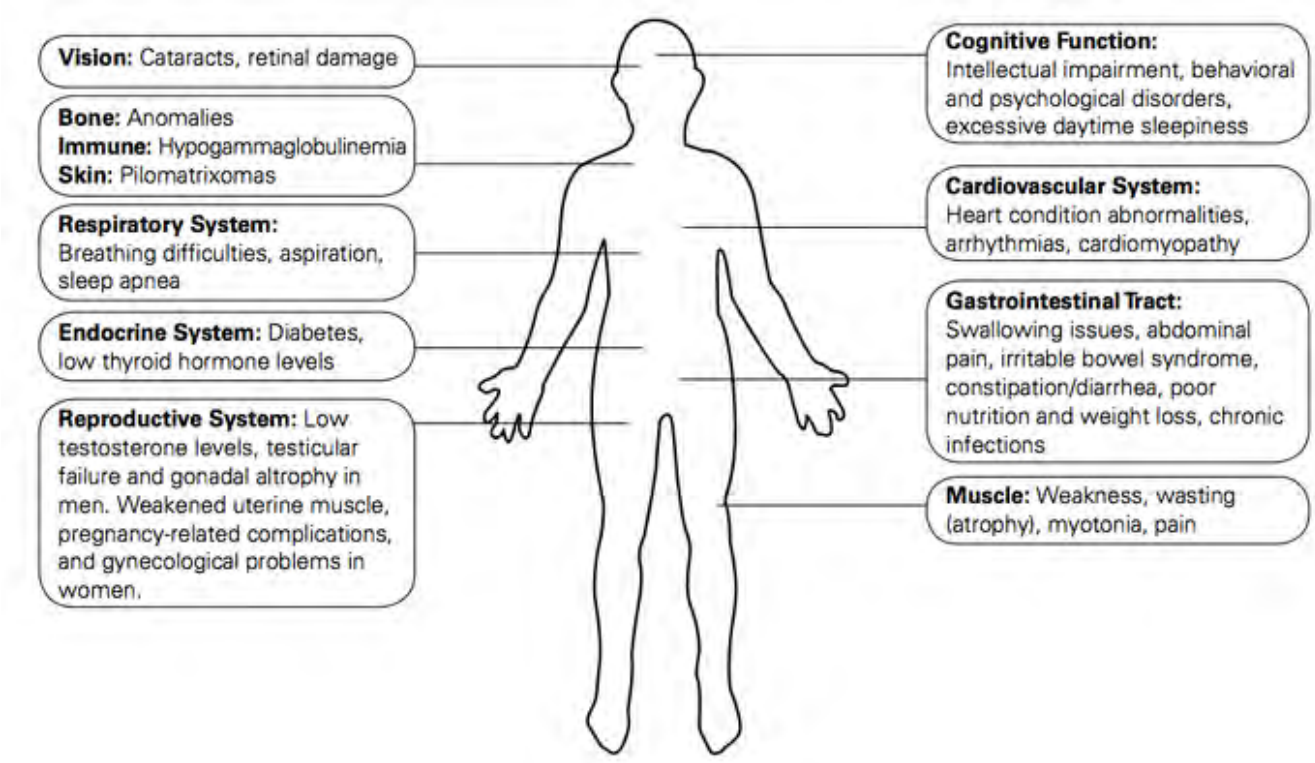


Figure above indicating multiple systems impacted with myotonic dystrophy

- Spinal muscular atrophies are a heterogeneous group of disorders, usually genetic in origin, characterized by the degeneration of anterior horn cells in the spinal cord and motor nuclei of the brainstem
    - Symptoms can present from birth to adult life
    - Disorders that begin in infancy are marked by a generalized distribution of weakness
    - Several clinical syndromes of infantile spinal muscular atrophy exist, one form manifesting between birth and 6 months of age (Werdnig-Hoffmann disease)
    - Inheritance is autosomal recessive
    - Newborns often have arthrogryposis at birth, and weakness progresses rapidly to respiratory insufficiency and death.
- Myasthenia syndromes can be caused by genetic defects or can occur as a transitory phenomenon in 10% to 15% of infants born to women who have myasthenia gravis
    - Most myasthenia syndromes are transmitted via autosomal recessive inheritance and are extremely rare
    - Many infants require assisted ventilation at birth.
    - Arthrogryposis may be present, as well as ptosis and generalized weakness.
    - The infants are able to be weaned from mechanical ventilation in weeks, but persistent episodes of weakness and apnea may occur.
    - The diagnosis is established by the patient's response (temporary reversal of weakness) to an intravenous or subcutaneous injection of edrophonium chloride (0.15 mg/kg) as well as presence of antibodies in the infant's blood
        - Ptosis and oculomotor paresis are the only functions that can be tested reliably
        - The transitory myasthenic syndrome is due to the passive placental transfer of antibodies against the acetylcholine receptor protein from the mother who has myasthenia to her unaffected fetus.
        - The severity of symptoms correlates with the newborn's antibody concentration.
        - Difficulty feeding and generalized hypotonia are the major clinical features.
        - Symptoms usually occur within hours of birth or up to 3 days later
        - Respiratory insufficiency is uncommon
        - Although weakness initially worsens, dramatic resolution subsequently occurs
        - The duration of symptoms averages 18 days, and recovery is complete

Table 3. **Prevalence of Causes of Hypotonia**

Cause of Hypotonia	Percentage in Three Hypotonic Series (n=277)	Prevalence	Distinguishing Features	Test Available?
Hypoxic-ischemic Encephalopathy	19%			
Genetic/Chromosomal Syndromes	31%			
Down syndrome	13%	1:800 to 1:1,000		Karyotype
Prader–Willi syndrome	5%	1:10,000 to 1:30,000		Methylation
Other dysmorphic syndromes	9%			
Other chromosomal anomalies	4%			Karyotype
Fragile X syndrome		1:4,000 males 1:8,000 females		*FMR1* test
Trisomy 18 (Edwards syndrome)		1:5,000 to 1:6,000		Karyotype
1p36 deletion syndrome		1:5,000 to 1:10,000		Array CGH
22q13 deletion syndrome		*		Array CGH
22q11.2 deletion syndrome (velocardiofacial/DiGeorge syndrome)		1:4,000 to 1:6,400		Array CGH
Williams syndrome		1:7,500		Array CGH
Trisomy 13 (Patau syndrome)		1:10,000		Karyotype
Smith–Magenis syndrome		1:15,000 to 1:25,000		Array CGH
Sotos syndrome		1:14,000		Array CGH
Wolf–Hirschhorn syndrome		1:50,000		Array CGH
Kabuki syndrome		1:30,000		None
Cri du chat syndrome		1:20,000 to 1:50,000		Karyotype
Brain anomalies	13%			
Myopathies	5%			
Central core disease		**		
Nemaline myopathy		1:50,000		
X-linked myotubular myopathy		1:50,000		
Congenital myotonic dystrophy	4%	1:100,000		
Metabolic disorders	3%			
Peroxisome biogenesis disorders, Zellweger syndrome spectrum		1:50,000		
Smith–Lemli–Opitz syndrome		1:20,000 to 1:60,000		
Glycogen storage disease Pompe (Type II)		1:14,000 to 1:100,000 1:40,000 (in United States)	Cardiomegaly	GAA gene; Alpha glucosidase
Benign neonatal hypotonia	3%			
Spinal muscular atrophy	2%	1:10,000		
Muscular dystrophies	2%	1:20,000 to 1:40,000		
Joint laxity	1.4%			
Neuropathy	1.4%			
Teratogens	1%			
Brain tumor	0.4%			
Myoclonic encephalopathy	0.4%			
Neuromuscular junction disorder	0.4%			
Familial infantile myasthenia (not transient)		1 to 4.4:1,000,000		Decremental EMG, negative antibodies, multiple gene tests for AcHR
Unknown	13%			

AcHR=anticholine receptor, EMG=electromyography, CGH=comparative genomic hybridization
*Second most common subtelomeric deletion after 1p36 deletion syndrome (*GeneReviews*)
**Most common congenital myopathy (*GeneReviews*)

- Next steps in evaluation for hypotonia in an infant is selection of appropriate laboratory and radiological testing that would be indicated given the clinical scenario and history
  - The initial laboratory evaluation of the hypotonic newborn is directed at ruling out systemic disorders
    - Hypotonia in a neonate should always be presumed to be sepsis until otherwise proven!...therefore, routine studies should include an evaluation for sepsis (blood culture, urine culture, cerebrospinal fluid culture and analysis)
    - Would also include measurement of serum electrolytes, including liver functions and ammonia, glucose, calcium, magnesium, and creatinine; a complete blood count; and a urine drug screen are also appropriate If hepatosplenomegaly is present and calcifications are noted on head ultrasonography, TORCH titers (toxoplasmosis, rubella, cytomegalovirus infection, herpesvirus infections) and a urine culture for cytomegalovirus should be undertaken.

- Sepsis evaluations in older infants should be driven by mental status and immunization status for the infant of course per standard of care measures
- If the hypotonia is considered to be central, the practitioner should evaluate for genetic and metabolic causes
    - Initial first tier genetic test of choice (when a specific chromosomal or genetic etiology is not known) is currently a chromosomal microarray (either CGH or SNP- SNP offers advantages over CGH when available – including being able to detect long contiguous stretches of heterozygosity = LCSH)
    - A high resolution karyotype can still be the best (and more cost effective choice) when clinical index of suspicion is high for instances of aneuploidy (i.e. Turner's, Trisomy 21)
    - Methylation study for 15q11.2 (Prader-Willi/Angelman) imprinting defects and testing for known disorders with specific mutational analysis may still be clinically indicated
    - Molectular genetic testing may also be faster and more cost efficient when index of suspicion is high→Molecular genetic testing provides the advantage of speed and diagnostic specificity without invasive procedures and these tests should be chosen according to the clinical presentation of the infant.
- If the clinical evaluation suggests complex multisystem involvement, screening for inborn errors of metabolism is indicated
    - If acidosis is present, plasma amino acids and urine organic acids (aminoacidopathies and organic acidemias), serum lactate (disorders of carbohydrate metabolism, mitochondrial disease), pyruvate, ammonia (urea cycle defects), and acylcarnitine profile (organic acidemia, fatty acid oxidation disorder) should be measured
    - Very long-chain fatty acids and plasmalogens are specific for the evaluation of a peroxisomal disorder
    - A creatine kinase and acylcarnitine/carnitine concentration should be determined if the child is weak and exhibits hypotonia to aid in diagnosis of a muscular dystrophy or carnitine deficiency
- To evaluate causes of peripheral hypotonia, creatine kinase concentrations should be measured.
    - This value is elevated in muscular dystrophy but not in spinal muscular atrophy or in many myopathies
    - Specific DNA testing can be performed for myotonic dystrophy and for spinal muscular atrophy
    - Other potentially useful screening tools include electrophysiologic studies, which show abnormalities in nerves, myopathies, and disorders of the neuromuscular junction
    - With the exception of a few myopathies, normal EMG findings suggest that the hypotonia is central in origin
    - Muscle biopsy with immunohistochemical staining and electron microscopy is the method of choice for differentiating myopathies and muscular dystrophies, although it is more invasive

        - If biopsy shows specific abnormalities, it can be an essential part of the diagnostic evaluation in the newborn to guide subsequent DNA molecular diagnostic studies.
    - Neuroimaging is a valuable tool for detecting central nervous system abnormalities. Magnetic resonance imaging can delineate structural malformations, neuronal migrational defects, abnormal signals in the basal ganglia (mitochondrial abnormalities), or brain stem defects (Joubert syndrome)
        - Deep white matter changes can be seen in Lowe (oculocerebrorenal) syndrome- a peroxisomal defect
        - Abnormalities in the corpus callosum may occur in Smith-Lemli-Opitz syndrome (hypoplasia, agenesis)
        - Heterotopias may be seen in congenital muscular dystrophy
        - Magnetic resonance spectroscopy also can be revealing for metabolic disease
- Other diagnostic testing would have to be driven by clinical exam findings and index of suspicion based on other evaluation findings

    - The findings of an enlarged heart in a floppy, weak infant who has delayed milestones would hugely support the possibility of a glycogen storage disease (type II Pompe disease or acid maltase deficiency)
        - this condition is caused by a deficiency in the lysosomal enzyme acid alpha glucosidase, present in all tissues, which hydrolyzes maltose to yield glucose but has no function in maintaining blood glucose
        - Absent enzyme activity in the infantile form of Pompe disease results in abnormal glycogen deposition in the skeletal, cardiac, and smooth muscles, leading to hypertrophic cardiomyopathy, feeding abnormalities, hypotonia, weakness, respiratory insufficiency, and ultimately, death
        - Inherited in an autosomal recessive pattern, the infantile form may present perinatally, but onset of symptoms usually occurs in the second postnatal month.
        - Profound generalized hypotonia without atrophy and congestive heart failure are the initial signs
        - Hypotonia is the result of glycogen storage in the brain, spinal cord, and muscle, producing a mixed central and peripheral clinical picture
        - Cardiomegaly almost always is diagnostic→Most patients die of cardiac failure by 12 months of age
        - The diagnosis of Pompe disease can be made by blood draw measuring the function/activity of the GAA enzyme can be measured in white blood cells (leukocytes),
        - When a diagnosis of Pompe disease is based on a leukocyte or blood spot assay, it must be confirmed through molecular genetic testing (DNA analysis) or by another enzyme assay, preferably using cultured skin fibroblasts obtained by a skin biopsy
        - Muscle biopsies are not usually indicated anymore in diagnosis of Pompe disease
        - Early diagnosis of Pompe disease results in early institution of enzyme replacement therapy, which minimizes morbidity and prolongs survival
            - However, improving the function of skeletal muscle has proven to be a more challenging prospect for enzyme replacement therapy, which has not been shown to affect outcomes in severe cases presenting in the first few postnatal months with associated congenital anomalies or ventilator dependence.
            - Recent assays using tandem mass spectrometry and included in some states on newborn metabolic screening are likely to prove useful for early diagnosis and institution of therapy.

- Other important diagnostic considerations for hypotonia in the infant is when the presentation is acute or subacute in terms of onset

    - Human botulism one important diagnosis to consider on differential in this case→ ordinarily results from eating foods contaminated by preformed exotoxin of the organism *Clostridium botulinum*
        - the exotoxin blocks the release of acetylcholine at the neuromuscular junction, which results in cholinergic blockade of skeletal muscle and end organs innervated by autonomic nerves
        - infantile botulism is an age-limited disorder in which *C botulinum* is ingested, colonizes the intestinal tract, and produces toxin in situ. In only 20% of cases, contamination with honey or corn syrup is identified
        - infants afflicted with botulism are usually between 2 and 26 weeks of age, usually live in a dusty environment adjacent to construction or agricultural soil disruption, and become symptomatic between March and October
        - A prodrome of constipation, lethargy, and poor feeding is followed in 4 to 5 days by progressive bulbar and skeletal muscle weakness and loss of DTRs
        - Progressive muscle paralysis can lead to respiratory failure. Symmetric bulbar nerve palsies manifested as ptosis, sluggish pupillary response to light, ophthalmoplegia, poor suck, difficulty swallowing, decreased gag reflex, and an expressionless face are primary features of infantile botulism
        - Treatment for infantile botulism needs to be instituted promptly with IV human botulium IG along with respiratory and nutrition support →self-limited course that lasts 2-6 weeks- relapse is possible in 5%

        - The differential diagnosis includes sepsis, intoxication, dehydration, electrolyte imbalance, encephalitis, myasthenia gravis, and polyneuropathies such as Guillain-Barré syndrome
        - Spinal muscular atrophy type I and metabolic disorders can mimic infantile botulism
            - Patients who have spinal muscular atrophy type I generally have a longer history of generalized weakness, do not typically have ophthalmoplegia, and have normal anal sphincter tone
- The most common clinical condition, although a diagnosis of exclusion, is benign congenital hypotonia

    - nonprogressive neuromuscular disorder that presents at birth with delays in achieving developmental milestones
    - improves with the maturity of the central nervous system
    - characteristics include generalized symmetric flaccidity of muscles and hypermobile joints
    - as a diagnosis of exclusion, the history should not be suggestive of a neurologic or metabolic disorder
    - Muscle stretch reflexes are normal or only slightly exaggerated and routine laboratory test results are within normal limits
    - patients must be counseled about the possibility of joint dislocations in the future
    - an increased incidence of intellectual disability, learning disability, or other sequelae of cerebral abnormality often is evident later in life, despite the recovery of normal muscle tone
    - a high familial incidence also is reported
    - this condition must be differentiated from congenital muscular dystrophy, which has a high risk of life-threatening malignant hyperthermia from anesthesia.

- The cause of hypotonia in most affected patients is central→bottom line is the greatest diagnostic yield starts with a detailed medical history and examination, including a neurologic evaluation and the search for dysmorphic features

2. Recognize the typical presentation of developmental coordination disorder

- The terms developmental dyspraxia, minimal brain dysfunction, minimal neurological dysfunction and clumsy child syndrome have all been used to describe the same clinical presentation, however, the accepted terminology in most places is now developmental coordination disorder, or DCD
- Developmental coordination disorder is a common neurodevelopmental condition, affecting as many as about 5%–6% of school-aged children according to some reports (European Academy of Childhood Disabilities, EACD)
- Children born preterm are 6-8 times more likely to be diagnosed with the condition.
- DCD is often not diagnosed until at least 5 years of age although screening and the start of treatment can occur earlier
- See below chart for current diagnostic criteria under the DSM-5
    - Ideally, the diagnosing team should involve a multidisciplinary approach and include a physician (e.g., child psychiatrist, developmental pediatrician, child neurologist) and an occupational therapist or physical therapist trained in the standardized motor tools used to assess children suspected of having the disorder.
    - Other possible causes of the child's motor difficulties (e.g., cerebral palsy, muscular dystrophy, systemic illness) should be ruled out through a comprehensive clinical examination that includes the neuromotor status, medical status, sensory status and behavioral status.
    - The child's cognitive status will also need to be assessed if learning difficulties have been reported by the teacher. Body mass index should be part of the general physical exam, because children with developmental coordination disorder have a propensity to be overweight or obese.
    - The American Academy of Pediatrics has outlined components of the neuromotor examination appropriate for young children, including:

        - **Cranial nerve examination**

            - Observe eye movements
            - Check response to visual fields by confrontation
            - Observe pupillary responses to light
            - Observe eye opening and closing
            - Observe facial expression
            - Have child drink through a straw or blow kisses

        - **Strength and flexibility**

            - Palpate muscle bulk and texture
            - Assess joint flexibility
            - Assess quality and intensity of grasp

        - **Motor planning**

- Observe functional gross motor skills (e.g., running, hopping, skipping, balancing on one foot)
- Observe functional fine motor skills (e.g., buttoning, zipping, cutting with scissors, tying shoelaces)
- Determine hand dominance (or lack thereof)

Box 4:

Diagnostic criteria for developmental coordination disorder[4]

A. The acquisition and execution of coordinated motor skills is substantially below that expected given the individual's chronological age and opportunity for skill learning and use. Difficulties are manifested as clumsiness (e.g., dropping or bumping into objects) as well as slowness and inaccuracy of performance of motor skills (e.g., catching an object, using scissors or cutlery, handwriting, riding a bike or participating in sports).

   How to assess this criterion: According to recent review articles,[13,14] the most widely used motor test to assist in the diagnosis of this disorder is the Movement Assessment Battery for Children, second edition (Movement ABC-2).[15] Another test often used is the Bruininks–Oseretsky Test of Motor Proficiency, second edition.[16] Both tests are norm-referenced, reliable and valid in diagnosing developmental coordination disorder.

B. The motor skills deficit in criterion A significantly and persistently interferes with activities of daily living appropriate to chronological age (e.g., self-care and self-maintenance) and affects academic/school productivity, prevocational and vocational activities, leisure and play.

   How to assess this criterion: Parents should be asked about their child's performance and length of time to learn motor tasks, such as dressing, tying shoelaces, brushing teeth, using a knife and fork, and printing and handwriting. In addition, checklists or questionnaires for parents and teachers can be helpful, such as the Developmental Coordination Disorder Questionnaire (DCDQ)[11] and the Movement ABC-2 checklist.[17] The Little DCDQ is currently undergoing validation to identify preschool-aged children with the disorder.[18]

C. Onset of symptoms is in the early developmental period.

   How to assess this criterion: The pattern and trajectory of the child's motor development should be ascertained to determine whether motor delay was present in early life. Parents can be questioned about their child's early developmental history and milestones (Box 2). Although some children with developmental coordination disorder may have delayed motor milestones, many do not.

D. The motor skills deficits are not better explained by intellectual disability (intellectual developmental disorder) or visual impairment and are not attributable to a neurologic condition affecting movement (e.g., cerebral palsy, muscular dystrophy, degenerative disorder).

   How to assess this criterion: To rule out intellectual disability, a standardized IQ test administered by a school psychologist is useful; however, a formal IQ assessment may not be necessary if there has been typical achievement at school.[1] As with screening, a neurologic examination is important to rule out other medical conditions that could explain impaired motor development.[1] A family physician or optometrist could rule out impairments in visual acuity.[1]

- Children with this condition experience significant difficulties with activities of daily living and academic achievement due to poor performance of gross and fine motor skills
- Common comorbidities include ADHD, learning disabilities and speech and language impairments. (see chart below for other comorbidities, resultant conditions, and risk factors)
    - Whereas the DSM-IV-TR did not allow for concurrent diagnoses, the DSM-5 does
    - However, as coordination difficulties are a common marker for higher risk for comorbid communicative, cognitive, and autism spectrum disorders, a broader perspective is justified
    - The DSM-V and the European Academy of Child Neurology and Neurodevelopmental Pediatrics have continued to emphasize the critical importance of a qualitative and quantitative approach to diagnosis of this disorder
    - Bottom Line: Developmental coordination disorder (DCD) construct has continued evolving as a comprehensive developmental condition in which impairments in a child's capacity to develop and execute coordinated motor skills cannot be explained by cognitive or sensory dysfunction or other neurological conditions

Box 5:

Summary of risk factors, concurrent diagnoses, secondary consequences and therapies relevant to developmental coordination disorder

Risk factors

- Male sex (odds ratio [OR] 1.7–2.8)[7,8]
- Very preterm birth (< 32 wk) or low birth weight (< 1500 g) or both (OR 6.29–8.66)[6]
- Small-for-gestational age (OR 1.74)[7]
- Independent walking at age 15 months or later (OR 3.05)[7]
- Difficulties in attention (OR 1.94), social communication (OR 1.87), non-word repetition (OR 1.83), spelling (OR 2.81) and reading (OR 3.35)[10]

Concurrent diagnoses

- Anxiety (16.7%–33.8%)[19] and depression (9.1%–11.8%)[19]
- Attention-deficit/hyperactivity disorder (50%)[1]
- Autism spectrum disorder (4.1%–8.2%)[1]
- Specific learning disabilities (17.8%–27.5%)[33]
- Specific language impairment (up to 70%)[1]

Secondary consequences

- Compromised physical fitness (OR unknown)
- Decreased participation in daily-living, physical and social activities (OR unknown)
- Hypermobility of joints (OR 1.92)[23]
- Mental health difficulties (OR 2.08–4.23)[20]
- Overweight or obesity (OR 1.79–2.28)[22]

Therapies

- Occupational therapy[29]
- Physical therapy[29]
- Task-oriented interventions[29]
- Methylphenidate[29,30]
- Dietary supplementation with fatty acids[32]

3. Know appropriate management strategies for a child with developmental coordination disorder.
- See above chart regarding therapies and other treatment options recommended with DCD.
- Children with developmental coordination disorder typically receive different types of therapies (e.g., occupational therapy or physical therapy) aimed at improving their motor performance.
- For children who also have ADHD, drug therapies (e.g., methylphenidate) have been used. Recent evidence is summarized below.
    - Recent studies have been done that have looked at motor performance measured on the Movement Assessment Battery for Children, second edition→ scores substantially improved when children with developmental coordination disorder and concurrent ADHD were taking methylphenidate.
    - Lack of studies, however, that look at the effects of chemical supplements in children who had developmental coordination disorder without a diagnosis of ADHD.
    - In a 2014 review of prospective clinical trials examining the safety of medications for children with ADHD, long-term (≥ 1 yr) adverse effects associated with methylphenidate included headache, decreased appetite, insomnia, abdominal pain, tics and weight loss (with incidence ranging from about 5% to 20%).[31]
        - As with other research on this topic, the authors reported that most of the adverse effects were mild or moderate.
- In a comprehensive systematic review and meta-analysis (Smits-Engelsman BC et al, 2013), the efficacy of various interventions on motor performance in children with developmental coordination disorder was examined
    - Twenty-six studies (systematic reviews, randomized controlled trials and crossover studies) were included and divided into four categories: task-oriented intervention; traditional occupational therapy or physical therapy; process-oriented therapies; and chemical supplements.
    - The task-oriented interventions were aimed at learning specific motor skills that are particularly difficult for the child, and the process-oriented therapies focused on more global functions (e.g., sensory integration, visual-motor perception and muscle strength).
        - The largest effect sizes were shown for task-oriented interventions and for occupational or physical therapy with only a weak effect for process-oriented approaches
        - Thus, referral to an occupational therapist or physical therapist, or both, who can focus treatment on helping the child perform everyday tasks is recommended.
    - In this same systematic review, the effects of methylphenidate on fine motor performance were reported from three studies involving children who had developmental coordination disorder and ADHD; the effect size was medium
    - The systematic review only found one randomized controlled trial that examined the effects of dietary supplementation with fatty acids
        - Although there were no changes in motor performance, improvements in reading, spelling and behavior were reported

4. Know the causes of congenital hypotonia.

5. Know the signs and symptoms of spinal muscular atrophy (SMA)
6. Plan the evaluation of a child with developmental coordination disorder

References for Content Category 9- Motor Disabilities and Multiple Handicaps

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Content Category 10- Autism Spectrum Disorder

ABP Content Specs for Developmental-Behavioral Pediatrics Fellow Collaboration Project

Sections 1-5 Prepared by Tanaporn Jasmine Wilaisakditipakorn, UH Rainbow Babies and Children's-Cleveland DBP Fellow

Sections 10-14 Prepared by Carrie Cuffman, UH Rainbow Babies and Children's-Cleveland DBP Fellow

Reviewed by Shanna Kralovic, MD, UH Rainbow Babies and Children's-Cleveland -DBP Fellowship Director

Sections 6-9 Prepared by Preeya Desai, MD, UCLA DBP Fellow

Reviewed by Irene Koolwijk, MD, UCLA DBP Fellowship Director

10. Autism Spectrum Disorder

A. Autism spectrum disorder

1. Know the comorbid conditions commonly associated with autism spectrum disorder
2. Know the etiologies of autism spectrum disorders
3. Know the diagnostic criteria and severity levels for autism spectrum disorder
4. Understand the deficits of children with autism spectrum disorder in joint attention, social referencing, and theory of mind
5. Know the differential diagnosis for autism spectrum disorder
6. Know how to plan the psycho-educational evaluation of a child with autism spectrum disorder
7. Develop management plans for children with autism spectrum disorders taking into account age, severity of autistic behaviors, and language and cognitive abilities
8. Understand the role of psychopharmacologic agents in the management of children with autism spectrum disorder
9. Understand the role of behavior therapies in the treatment of children with autism spectrum disorder
10. Know the range of prognoses for a child with autism spectrum disorder
11. Know the neurologic, developmental, and behavioral characteristics of Landau-Kleffner syndrome
12. Know how to plan the medical evaluation of a child with autism spectrum disorder
13. Know appropriate educational interventions and accommodations for a child with autism spectrum disorder
14. Understand the genetics of autism spectrum disorders

A. Autism spectrum disorder

1. Know the comorbid conditions commonly associated with autism spectrum disorder

Motor impairment

- Excessive reliance on proprioceptive feedback and weak integration of visual feedback
- Impairment in basic aspects of motor coordination, dyspraxia and impairment in motor learning activities
- Tic disorders including Tourette disorder is also common
- Clinically: delayed motor milestones, clumsiness, toe-walking and difficulty with handwriting
- Neuro exam: hypotonia, decreased postural stability, poor motor imitation abilities, dysrhythmia

Epilepsy

- Onset has bimodal peaks in early childhood and adolescent
- Complex partial seizures, with or without secondary generalization are more common than primary generalized seizures
- Estimated cumulative prevalence is 26%
- The most robust risk factors are low IQ and older age
- There are very little information to support the idea that epilepsy cause ASD

Neurobehavioral Symptoms/Coexisting Psychiatric Disorders

- Common neurobehavioral features: irritability, anger outbursts, aggression, self-injurious behavior, property destruction, elopement or wandering, sleep disturbance, mood instability, anxiety, hyperactivity, impulsivity, inattention and other disruptive behaviors or emotional dysregulation
- Common psychiatric diagnoses: ADHD (28-44%), anxiety (42-56%), depression (12-70%), psychotic disorders (12-17%), tic disorders (14-38%), and oppositional defiant disorder (16-28%)
- Sleep problems
    - reported 40-80%, common at all ages and levels or intellectual ability
    - associated with exacerbation of core ASD symptoms
    - common problems: bedtime resistance, prolonged sleep-onset latency, decreased sleep efficiency, increased waking after sleep onset, reduced total sleep time and daytime sleepiness
    - Behavioral factors: inadequate sleep hygiene, maladaptive sleep-onset associations, and problems with limit-setting
    - Other factors: ASD-associated melatonin or GABA abnormalities, coexisting medical/psychiatric problems and adverse effects of medications
    - Other occasionally reported sleep problems: obstructive sleep apnea, parasomnias, and periodic limb movements of sleep

Gastrointestinal and Feeding Problems

- Comparing to children without ASD, children with ASD experience more general GI symptoms, diarrhea, constipation, abdominal pain
- No evidence suggesting a unique GI pathophysiology in ASD
- Common feeding problems: food selectivity, rituals around food presentation, compulsive eating of certain foods, food refusal , rumination and pica

[Reference: Voigt, R. G. (2018), Developmental and Behavioral Pediatrics 2nd edition, the American Academy of Pediatrics.]

2.Know the etiologies of autism spectrum disorders

The etiology of ASD is heterogenous and the pathophysiology has not been identified.

Genetics and Genomics

- ASD is heritable. Studies showed increased risk of ASD in siblings of children with ASD and higher concordance rates for monozygotic twins than dizygotic twins.
- No specific mutation has been identified that is unique to ASD.
- There is substantial genetic overlap between ASD and other NDD which is associated with increased copy number variants (CNV).
- Increased maternal and paternal age are independently associated with ASD-risk. ASD-risk also increased with a combined parental age effect and when the difference between the parental ages is 10 years or more.

Environment

- Prenatal environment: maternal medications (eg, valproate), short interpregnancy interval, multiple gestation, maternal obesity, gestational diabetes, gestational bleeding, and infections (eg, rubella, CMV, influenza)
- Perinaltal factors: prematurity, low birth weight, fetal growth restriction, intrapartum hypoxia, and neonatal encephalopathy
- Toxic environmental chemicals have not been proven to cause ASD but associations between ASD risk and some compounds such as organophosphates and certain other pesticides, metals,and volatile organic compounds.

Epigenetics

- Epigenetic modifications such as DNA cytosine methylation and posttranslational histone modification, produce heritable change in gene expression without change in the DNA sequesce
- Examples: Rett syndrome, CHARGE syndrome, Angelman, Prader-Willi , and Fragile X syndrome

Neuropathology

- No uniform neuropathology has been identified.
- Focal disruption of the normal laminar architecture, minicolumnar abnormalities and variations in neuronal density are among the most common cerebral cortical abnormalities described. However it is not yet clear whether this represents primary pathology or a secondary process.

Biomarkers

- No biomarker has yet been shown to be clinically valuable.
- Proposed biomarkers of ASD include head circumference growth trajectory; structural and functional magnetic resonance imaging (MRI) markers; electroencephalographic characteristics; eye tracking markers; genetic and biochemical markers in blood, uring, or brain tissue; placental pathology and maternal autoantibody profiles.
- Early brain overgrowth and neuroimaging markers
    - Retrospectively, children with ASD have average or below-average head circumference (HC) at birth, with abnormal acceleration in brain growth during the first 1-2 years leading to above-average HC and MRI brain volumes in toddlers, followed by a plateau resulting in average brain volume in adolescence and adulthood.
    - However in a large prospective study, this atypical pattern did not predict the diagnosis of ASD in the first 3 years of life.
- Electrophysiological markers and eye tracking

    - No evidence for the use of any marker in clinical practice.
    - However studies show electrophysiological correlates of abnormalities in low-level and higher cognitive auditory and visual procession (including language processing and face procession), somatosensory response, multisensory integration, recognition memory, selective attention, attentional shifting and neural connectivity in association with ASD.
    - Children with ASD may have decreased fixation on the eyes' region of the face and increased fixation on the mouth and background elements.
    - Retrospectively, children with ASD exhibit a decline in eye fixation between the ages of 2 and 6 months
- Biochemical, gene expression, and other tissue markers
    - The most consistently reported biochemical markers associated with children with ASD include increased platelet serotonin level, decreased plasma melatonin and urine melatonin sulphate, indicators of increased oxidative stress or altered redox status and markers of altered immune response such as irregular cytokine profiles and central nervous system microglial activation.
    - Studies using gene expression to classify ASD risk is still preliminary.

[Reference: Voigt, R. G. (2018), Developmental and Behavioral Pediatrics 2nd edition, the American Academy of Pediatrics.]

3.Know the diagnostic criteria and severity levels for autism spectrum disorder

DSM-5 Criteria for Autism Spectrum Disorder

Domains	Criteria: Deficits	Examples
A. Persistent deficits in social communication and social interaction across multiple contexts, as manifested by the following, currently or by history; must have all 3 symptoms in this domain	1. Social-emotional reciprocity	Abnormal social approach and failure of normal back and-forth conversation; reduced sharing of interests, emotions, or affect; failure to initiate or respond to social interactions
	2. Nonverbal communicative behaviors used for social interaction	Poorly integrated verbal and nonverbal communication; abnormalities in eye contact and body language or deficits in understanding and use of gestures; total lack of facial expressions and nonverbal communication
	3. Developing, maintaining, and understanding relations	Difficulties adjusting behavior to suit various social contexts; difficulties in sharing imaginative play or in making friends; absence of interest in peers
B. Restricted, repetitive patterns of behavior, interests, or activities, as manifested by at least 2 of the following, currently or by history; must have 2 of the 4 symptoms	1. Stereotyped or repetitive motor movements, use of objects, or speech	Simple motor stereotypies, lining up toys or flipping objects, echolalia, idiosyncratic phrases
	2. Insistence on sameness, inflexible adherence to routines, or ritualized patterns or verbal nonverbal behavior	Extreme distress at small changes, difficulties with transitions, rigid thinking patterns, greeting rituals, need to take same route or eat food every day
	3. Highly restricted, fixated interests that are abnormal in intensity or focus	Strong attachment to or preoccupation with unusual objects, excessively circumscribed or perseverative interest

	4. Hyper- or hyporeactivity to sensory input or unusual interests in sensory aspects of the environment	Apparent indifference to pain/temperature, adverse response to specific sounds or textures, excessive smelling or touching of objects, visual fascination with lights or movement

- Symptoms must be present in the early developmental period (but may not become fully manifest until social demands exceed limited capacities or may be masked by learned strategies in later life).
- Symptoms cause clinically significant impairment in social, occupational, or other important areas of current functioning.
- These disturbances are not better explained by intellectual disability (intellectual developmental disorder) or global developmental delay.
- Intellectual disability and ASD frequently co-occur; to make comorbid diagnoses of ASD and intellectual disability, social communication should be below that expected for the general developmental level. Specify whether: with or without accompanying intellectual impairment, language impairment or associated with a known medical or genetic condition or environmental factor.

Severity levels for autism spectrum disorder
(Ref: Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition)

Severity Level	Social Affective	Restricted and Repetitive Behaviors
Level 1. "Requiring support"	Without supports in place, deficits in social communication cause noticeable impairments. Difficulty initiating social interactions, and clear examples of atypical or unsuccessful response to social overtures of others. May appear to have decreased interest in social interactions.	Inflexibility of behavior causes significant interference with functioning in one or more contexts. Difficulty switching between activities. Problems of organization and planning hamper independence.
Level 2. "Requiring substantial support"	Marked deficits in verbal and nonverbal social communication skills. Social impairments apparent even with supports in place. Limited initiation of social interactions and reduced or abnormal responses to social overtures from others.	Inflexibility of behavior, difficulty coping with change, or other restricted and repetitive behaviors appear frequently enough to be obvious to the casual observer and interfere with functioning in a variety of contexts Distress and/or difficulty changing focus or action.

Level 3. "Requiring very substantial support"	Severe deficits in verbal and nonverbal social communication skills cause severe impairments in functioning, very limited initiation of social interactions, and minimal response to social overtures from others.	Inflexibility of behavior, extreme difficulty coping with change, or other restricted and repetitive behaviors markedly interfere with functioning in all spheres. Great distress at or difficulty with changing focus or action.

4.Understand the deficits of children with autism spectrum disorder in joint attention, social referencing, and theory of mind

Deficits in joint attention

- Deficits in joint attention for both receptive (responding to other's initiated joint attention) and expressive (initiating joint attention)
- Joint attention ability is a precursor and predictor of language and theory of mind
- Infants, toddlers: no responsive smile, lack of posture showing desire to be picked up.
- Young children: no coordinating attention with others, no social engagement or awareness of other's mutual interest.
- Older children; difficulty in reciprocal social behaviors such as seeking to share enjoyment with others, recognizing and reacting to the mental states of others and forming reciprocal friendships

Deficits in social referencing

- Social referencing is the process that infants look at parents or adults to decide how to response to an unfamiliar situation
- This is limited and lacking in children with ASD

Theory of mind deficit

- Social cognition impairment is an impairment in the capacity to represent and reason about the thoughts, beliefs, and emotional states of others
- Theory of mind deficit is impaired ability to take the perspective of others and understand that other people have intentions, knowledge, and beliefs that may differ from their own.

References for #1-4:

1. Voigt, R. G. (2018), Developmental and Behavioral Pediatrics 2nd edition, the American Academy of Pediatrics.

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5.Know the differential diagnosis for autism spectrum disorder

Mental health disorders	This is a broad category of differential diagnoses with variable symptomatology depending on the specific diagnosis. a. Obsessive compulsive disorder (OCD) – The obsessive thoughts and repetitive actions seen in OCD can appear very similar to the ritualistic behaviors and motor stereotypes seen in ASD. b. Anxiety – Children who have problems with anxiety may be hesitant to interact with others. They may have difficulties with transitions. Children with anxiety are still socially related and have appropriate social insight. c. Depression – Depression in children can present in a variety of ways. Children may be withdrawn and isolate themselves. They may have a blunted affect and avoid eye contact. d. Attention deficit-hyperactivity disorder (ADHD) – Children with ADHD may have impairments in their social skills due to their hyperactivity and impulsivity. They may have difficulty sustaining a conversation because of inattention. Children with ASD often have problems with hyperactivity, impulsivity, and inattention. e. Oppositional defiant disorder (ODD)/behavior problems – The behavior problems seen in children with ODD are usually intentional. Most children will have temper tantrums at some point. Children with ASD are more likely to have tantrums associated with transitions or "for no apparent reason." f. Tourette syndrome – Tics seen in Tourette syndrome may appear similar to motor stereotypes associated with ASD. Children with Tourette syndrome will usually not have the social or communication impairments seen with ASD. However, there may be some social isolation due to embarrassment or peer avoidance.
Psychosocial (e.g., neglect)	Children who have a history of significant abuse or neglect may be withdrawn and hesitant to interact with others. They may also have regression of skills, such as loss of language, and behavior problems.
Sensory problems	Children with ASD often have sensory issues such as being hypersensitive to loud noises or avoiding certain food textures. A child that has sensory impairments but is not on the autism spectrum will not have the core features of ASD (impairments in social communication, etc.).
Speech/language disorder	Children with speech/language disorders will compensate with nonverbal forms of communication such as pointing and gestures. They lack severe social deficits, although there may be some social impairment due to the communication difficulties.

Suggested Citation: Macferran K, Major N, Fussel J, High P. Differential and Etiologic Diagnosis of Autism Spectrum Disorder. Developed for the Autism Case Training: A Developmental-Behavioral Pediatrics Curriculum. 2011.

Condition	Features that may help distinguish the condition from ASD
Global developmental delay/ intellectual disability	Social responsiveness and communication appropriate for developmental level
Intellectual giftedness	Normal pragmatic language skills Intense interests are functional, varied, and can be explained by the child Social interaction is generally enjoyed
Social (pragmatic) communication disorder	Absence of restricted, repetitive patterns of behavior, interests, or activities
Developmental language disorder	Normal reciprocal social interactions Normal desire and intent to communicate Appropriate imaginative play
Language-based learning disorder	Normal reciprocal social interactions Normal desire and intent to communicate Appropriate imaginative play Pragmatic language more typical than in ASD Desire to communicate (even if competency is lacking)
Nonverbal learning disorder	Impairment in social skills and pragmatic language milder than in ASD Lack of restricted, repetitive patterns of behavior, interests, or activities
Hearing impairment	Normal reciprocal social interactions Normal eye-to-eye gaze Facial expressions indicate intention to communicate
Landau-Kleffner syndrome	Usually have typical development until approximately 3 to 6 years of age Typically presents with auditory verbal agnosia (behaving as if deaf)
Rett syndrome	Female predominance Head growth deceleration Stereotypic hand movements Gait abnormalities

	Abnormal respiratory pattern
Fetal alcohol spectrum disorder	Characteristic facial features (not always present): Short palpebral fissures Thin vermillion border Smooth philtrum
Attachment disorder	History of severe neglect or mental health issues in caregiver Social deficits tend to improve in appropriate caregiving environment
Attention deficit hyperactivity disorder	Normal pragmatic language skills Normal nonverbal social behavior Normal imaginative play Lack of restricted, repetitive patterns of behavior, interests, and activities
Anxiety disorder (includes social anxiety and selective mutism)	Normal nonverbal social behavior and imaginary play Lack of circumscribed interests Absence of restricted, repetitive patterns of behavior, interests, or activities
Obsessive compulsive disorder	Normal social skills Normal pragmatic language Symptoms are a source of anxiety rather than a pleasure
Stereotypic movement disorder	Normal social skills Normal pragmatic language
Tic disorder/Tourette syndrome	Normal social skills Normal pragmatic language

[Reference: Uptodate. Differential diagnosis of autism spectrum disorder. Available at: https://www.uptodate.com/contents/image?imageKey=PEDS%2F119225&topicKey=PEDS%2F628&search=autism&rank=1~150&source=see_link&sp=0. Accessed May 15, 2020.]

6. Know how to plan the psycho-educational evaluation of a child with autism spectrum disorder

- The following are validated observation tools used to provide structured data to confirm the diagnosis of ASD:
    - Autism Diagnostic Observation Schedule, 2nd Edition (ADOS-2): developed to elicit atypical social language and behaviors

        - Used in conjunction with the history of peer interactions, social relationships and functional impairment from symptoms to determine if the DSM-5 criteria are met
        - Semi-structured assessment by trained clinician of social interaction, play, imaginative use of materials, communication and atypical behaviors
        - Can be used in individuals age 12 months - adulthood
        - 5 Modules (Toddler, Module 1-4) based on child's expressive language abilities
    - Childhood Autism Rating Scale, 2nd Edition (CARS-2)
        - 15-point scale completed by the trained clinician based on history and direct-observation
        - Items scored on a 4-point scale ranging from 1 (normal) to 4 (severely abnormal)
        - The sum of the ratings is used to determine a total score and the severity of autistic behaviors
            - Non-autistic: 15-29
            - Mildly-moderately autistic: 30-37
            - Severely autistic: >37
        - Can be used in children >= 2 years of age
- The following can be used to help with history taking when considering a diagnosis of Autism Spectrum Disorder. Not sufficient to make a diagnosis alone, but help elicit symptoms.
    - Social Communication Questionnaire (SCQ)
    - Social Responsiveness Scale (SRS)
    - Autism Diagnostic Interview – Revised (ADI-R)
    - Gilliam Autism Rating Scale – Third Edition (GARS)
- The following are used to assess for behavioral health conditions, but may also identify profiles consistent with ASD
    - Behavior Assessment System for Children (BASC)
    - Diagnostic Interview for Social and Communication Disorders (DISCO)
    - Child Behavior Checklist (CBCL)
- Formal evaluation of language, cognitive, adaptive abilities, and sensory status is an important part of the diagnostic process in order to assess functional impairment, define the child's strengths and weaknesses for education planning, identify associated conditions, and evaluate conditions with symptoms that mimic ASD. The following could be included when evaluating a child for ASD:
    - Cognitive/Developmental Testing
    - Academic Testing
    - Adaptive Skills Functioning
    - Speech, language, and communication assessment
    - Sensorimotor and/or occupational therapy evaluation
    - Hearing and vision evaluation

7. Develop management plans for children with autism spectrum disorders taking into account age, severity of autistic behaviors, and language and cognitive abilities

- Goals of treatment
    - minimize the core deficits of ASD and co-occurring associated impairments

    - maximize functional independence by enabling learning and attainment of adaptive skills
    - eliminate, minimize, or prevent problem behaviors that may interfere with functional skills
- Treatments should be
    - Individualized based on child's age, specific needs, and strengths and weaknesses
    - Developmentally appropriate
    - Intensive
    - Include performance data relevant to treatment goals to assess and adjust intervention
- Interventions for children with ASD are provided through educational practices, developmental therapies, and behavioral interventions
- Consider the following when developing an intervention plan
    - Theoretical approach and scope: focused and targeted vs comprehensive
        - Comprehensive Treatment Model
            - Should be replicable, intense, and designed to address, multiple therapeutic goals over a period of time
        - Focused interventions:
            - address a single or limited range of skills (developmental, behavioral or educational) and may be delivered over a short period of time
    - Setting and/or modality of delivery
        - Individual vs group or classroom
        - School vs home setting
    - Targets of intervention
- Management Plans may consist of the following:
    - Behavioral intervention to address the core symptoms of ASD
    - Medication to address interfering behaviors refractory to behavioral intervention
    - Educational interventions
    - Speech-Language Therapy
    - Occupational Therapy
    - Social Skills Training

8. Understand the role of psychopharmacologic agents in the management of children with autism spectrum disorder

- There are currently no medications that treat the core social and communication symptoms of ASD
- Medications may be used for:
    - Associated behavioral health disorders, such as ADHD, mood disorders, anxiety disorders, etc
    - Associated problem behaviors or symptoms causing significant impairment and distress, such as aggression, self-injurious behavior, sleep disturbance, mood lability, anxiety, hyperactivity, impulsivity, inattention
- Medications should only be considered after:
    - History and physical exam have been completed to evaluate for medical factors that can exacerbate challenging behaviors
    - Review of when behavior started and exacerbating factors
    - A functional behavioral assessment has been completed to support development of a treatment plan

    - Referral to a behavior therapist to assess the reasons for the behavior, provide the family with strategies, and collaborate in care
- Consider medication in conjunction with the patient and his/her family after treatable medical conditions and behavioral factors have been assessed and intervention does not address the symptoms of concern

The following are medications that may be used to address co-occurring behavioral health condition, associated problem behaviors, or symptoms causing impairment and distress

Medication Class	Target Behavior	Comment
Psychostimulants	Hyperactivity, impulsivity, inattention, distractibility	
SNRI (atomoxetine)	Hyperactivity, impulsivity, inattention, distractibility	Consider in those with social anxiety
Alpha-adrenergic agonists (guanfacine and clonidine)	Hyperactivity, impulsivity, inattention, distractibility	- May have beneficial effects on irritability based on small studies though larger studies need to be completed - Can be considered in treatment of anxiety to address physiologic symptoms and behavioral dysregulation
Atypical antipsychotics (risperidone, aripiprazole)	Irritability, severe disruptive behavior, stereotyped or repetitive movements, self-injury, mood dysregulation disorder	- Risperidone and aripiprazole are the only FDA approved medications for treatment of irritability in ASD - Medication is most effective if combined with behavioral strategies addressing causes for the behavior and appropriate responses - Comprehensive behavioral approaches should be used in treatment of repetitive behaviors - More effective in treatment of tantrums, aggression, and self-injurious behavior vs repetitive behavior - Adverse effects: weight gain, increased appetite, dyslipidemia,

		fatigue, drowsiness, dizziness, drooling - Monitoring: for extrapyramidal symptoms; weight, height, BMI; glucose and lipid levels
SSRI's	Anxiety, depression, OCD	- Generalized anxiety disorder, separation anxiety disorder, and social phobias are most responsive - Adverse effects: Hyperactivation - Medication should be used in conjunction with a comprehensive behavioral approach
Anticonvulsant mood stabilizers (valproic acid and divalproex sodium)	Repetitive behaviors	- Modest improvements reported with divalproex sodium - May have improvement with topiramate as a second agent with risperidone - Small studies suggestive of improvement in irritability though larger studies are needed
Short-acting benzodiazepine	Could be considered for event-related anxiety	

9. Understand the role of behavior therapies in the treatment of children with autism spectrum disorder

- Behavioral interventions target the core symptoms of ASD
- Children should participate in behavioral therapies as early as possible
- Intensive Behavioral Intervention Programs
    - ABA: set of principles that form the basis for many behavioral treatments.
        - Is based on the science of learning and behavior
        - Used to increase useful or desired behaviors; reduce behaviors that may interfere with learning or behaviors that may be harmful; increase language and communications skills; improve attention, focus, social skills memory, and academics; decrease problem behaviors
        - Considered an evidence based "best" practice treatment by the US Surgeon General and American Psychological Association
        - Focuses on antecedents and consequences
        - ABA therapies:
            - Are Structured

            - Collect data for target skills or behaviors – provides monitoring of effectiveness of individual treatment programs and promotes alteration in program and goals when needed
        - Focuses on positive reinforcement strategies
        - Addresses problem behaviors that interfere with functioning through a functional behavioral assessment
        - Types of ABA therapies include:
            - Pivotal Response Training: Relies on naturally occurring teaching opportunities and consequences, and works to increase a child's motivation to learn, monitor their own behavior, and initiate communication with others
            - Incidental Teaching aka Natural Environment Training: focuses on teaching skills in settings where the child will naturally use them to facilitate generalization and maximize reinforcement. The child chooses an activity or situation and the therapist or caregiver follows the child's lead
            - Natural language paradigms: based on the idea that learning can be assisted by intentional arrangement of the environment in order to increase opportunities to use language. Emphasizes the child's initiative. Uses natural reinforcers and it promotes skill generalization
            - Discrete Trial Learning (Training): uses a series of trials to teach each step of a desired behavior or response. Specific behaviors are broken down into their simplest parts and positive reinforcement is used to reward correct answers and behaviors.
            - Verbal behavior: Focuses on teaching verbal skills in a structured, intensive one-to-one therapy by developing a connection between a word and its meaning
            - Early Intensive Behavioral Intervention (EIBI): Used in children younger than 5. Structured teaching approach to promote positive behaviors and decrease unwanted behaviors.
- Developmental Relationship-Focused Interventions
    - Designed to promote social development in children with ASD by focusing on the relationship between the child with ASD and his/her caregiver. Utilizes coaching to help increase responsiveness to the adult through imitating, expanding on, or joining into child-initiated play activities
    - Addresses core symptoms of ASD including joint attention, imitation, and affective social engagement
    - Examples
        - DIRFloortime
- Naturalistic Developmental Behavioral Intervention
    - Integrates elements of ABA and developmental principals by utilizing child-initiated opportunities for learning within play routines and instituting ABA-based approaches to address measurable goals
    - Ex: Early Start Denver Model
- Programs may use a combination of behavioral and developmental approaches
- Focused interventions delivered by trained parents or other caregivers can be an important part of the therapeutic program

References:
Hyman SL, Levy SE, Myers SM. “Identification, Evaluation, and Management of Children with Autism Spectrum Disorder.” *Pediatrics*. 2020; 145(1): e20193447
“Treatment and Intervention Services for Autism Spectrum Disorder.” Ceners for Disease Control and Prevention. cdc.gov/ncbddd/autism/treatment.html
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Weissman, L. “Autism spectrum disorder in children and adolescents: Overview of management.” *UpToDate.* https://www.uptodate.com/contents/autism-spectrum-disorder-in-children-and-adolescents-overview-of-management?topicRef=628&source=see_link

10. Know the range of prognoses for a child with autism spectrum disorder

- Prognosis and trajectory of development for a child with ASD cannot be predicted at the time of diagnosis
- >80% of children diagnosed with ASD by comprehensive evaluation at less than 3 years of age have retained their diagnosis. Approximately 9% of children who are diagnosed with ASD in early childhood may not meet criteria by young adulthood
    - Youth who no longer meet criteria more likely to have: history of higher cognitive skills at 2 yo, participated in earlier intervention services and to have demonstrated a decrease in repetitive behaviors over time
- Change in clinical diagnosis (i.e. to ADHD, OCD) more likely in children diagnosed before 30 months of age or diagnosis of PDD-NOS per DSM-IV
- Severity scores most likely to improve in youth who have had the greatest increase in tested verbal IQ
- Executive function difficulties are associated with poorer adaptive measures, independent of IQ
- Young children with ASD with language impairment tend to have more social difficulty than children w/ ASD without language impairment
- Children with ASD and ID have the most difficulty developing social competence
- IQ and language ability in childhood tend to predict outcome in adulthood
- Reported quality of life in high-functioning adults with ASD was associated more with the presence of family and community supports than ASD-related symptoms

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11.Know the neurologic, developmental, and behavioral characteristics of Landau-Kleffner syndrome

Landau Kleffner Syndrome is a rare age-related epileptic encephalopathy, with no more than a few hundred cases reported in the literature. Males > females. Typically between ages of 3 and 8.

Neurologic characteristics:

- Seizures occur in 2/3 of patients. Seizure types most commonly described are partial motor (most common), generalized clonic, and atypical absence seizures (eye blinking, chewing gestures, as well as lip-smacking, or slight jerking movements of the lips).
- **Seizures occur more often during sleep, making sleep EEG critical for diagnosis**
- EEG shows unilateral or bilateral activity most pronounced over posterior temporal regions around the Sylvian fissure. EEG activity becomes more widespread and intense during sleep (non-REM sleep) when it becomes characterized by an almost continuous spike-wave pattern with a frequency of 1.5 to 2.5 spikes/sec.

Developmental characteristics:

- New onset aphasia: initially receptive aphasia followed by expressive aphasia. Children may babble, use neologisms, become mute or have verbal perseveration.
- Social cognitive deficits
- Typically have normal development prior to onset

Behavioral characteristics:

- Attention deficit, hyperactivity, impulsiveness, and the tendency to get distracted
- Emotional lability
- Anxiety and depression
- Sleep disorders
- Working memory impairment (but not long-term memory)
- Hypersensitivity to sound

Muzio MR, Cascella M, Al Khalili Y. Landau Kleffner Syndrome. [Updated 2020 Jan 2]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2020 Jan-. Available from: https://www.ncbi.nlm.nih.gov/books/NBK547745/

12.Know how to plan the medical evaluation of a child with autism spectrum disorder

Begin work-up with careful medical, developmental-behavioral and family history and a thorough physical and neurologic examination. History should include potential prenatal exposure to teratogens. Physical exam: growth curves (including head circumference), dysmorphic features, organomegaly, skin manifestations of neurocutaneous disorders (e.g. tuberous sclerosis, neurofibromatosis), neurologic abnormalities.

Genetic Testing: Recommended and offered to all families. Dysmorphic features or ID increases likelihood of finding genetic abnormality.

- CMA analysis
- Fragile X-analysis
- Consider MECP2 testing & evaluation for Rett syndrome if female patient
- Other genetic/metabolic testing guided by family history and physical exam
- If no etiology from above testing, can consider whole exome testing
- Referral to genetics/neurology is appropriate if specific syndrome or metabolic disorder suspected (i.e. male w/ marked macrocephaly, pigmented macules on penis – evaluate for PTEN)

Neuroimaging: Need for MRI directed by history and physical exam

- Atypical regression
- Micro or macrocephaly
- Seizures
- Intracranial manifestations of genetic disorders
- Abnormal neurologic exam

Metabolic testing: Not recommended for regular use. Should be informed by history, family history, symptoms, and exam.

- Atypical regressions (later than 2 years of age, motor regression, multiple regressions)
- Family history of early childhood death or diagnosed metabolic disorders
- Physical features: significant hypotonia or weakness, visual and hearing impairment, dysmorphic features
- Motor delay: creatine kinase and TSH

EEG: Not recommended as routine although children with ASD do have increased risk of seizures.

- Clinical concern for seizures
- Atypical regression
- Late or atypical loss of language evaluate with overnight EEG

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13.Know appropriate educational interventions and accommodations for a child with autism spectrum disorder

- School-aged children should be educated in classroom setting with supports
- Typically require an IEP, although some may not qualify based on educational criteria and may be supported with accommodations through a 504 plan or classroom-level accommodations
    - Most with ASD + normal intelligence require academic intervention due to coexisting learning disabilities, executive function challenges, ADHD, motor processing challenges, effects of weaknesses of pragmatic language differences on reading/writing/comprehension
- Many students are served in inclusive classrooms
- IDEA laws apply: right to free and appropriate public education, least restrictive setting
- Education programs should promote language, academic, adaptive and social skills development and prepare the student for postsecondary education or employment
- Some children benefit from disorder-specific approaches (i.e. Learning Experiences, Alternative Programs for Preschoolers and their Parents (LEAP), and TEACCH)
    - LEAP: blends ABA with special and general education techniques for elementary aged students (RCT found improvement in socialization, cognition, language and challenging behavior and superior to treatment-as-usual)

- TEACCH: assessment-based curriculum development and an emphasis on structure, using class settings that are visually organized to promote learning (minimize distractions and sensory dysregulation). Visual schedules and predictable organization of instruction is used to promote independence in the activities planned. Small benefit in perceptual, motor, verbal, and cognitive skills. Associated with more reported improvement in ASD severity for students with greater cognitive delays.
- For social skills:
  - School-aged children and adolescents with ASD should have social skills support in school and other therapeutic settings if needed. Can be adult-mediated, peer-mediated and mixed.
  - Families should be encouraged to advocate for including social goals into IEP

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14. Understand the genetics of autism spectrum disorders

- Highly heritable. Twin studies demonstrate a high concordance rate (98% in monozygotic twins, 53-67% in dizygotic twins). Siblings also at risk for ASD (risk as high as 18-20% in some studies) and symptoms related to ASD that don't meet threshold for diagnosis
- Risk of ASD is increased in children of both older mothers and older fathers. Unclear mechanism for older mothers. For older fathers, may be related to germline mutations.
- Many aspects of genetics of ASD still poorly understand, including role of common variants, epistasis, environmental modification of genotype effects, epigenetics
- Researchers of microanalysis and whole exome studies have established that although de novo and inherited rare variants of large effect size are collectively common, no individual pathogenic variant accounts for more than 1% of cases of ASD
- Genes that contribute to ASD are involved in a variety of biological functions, with convergence on aspects of brain development and function
- No mutation has been identified that is unique to ASD: substantial overlap between ASD and other neurodevelopmental disorders, including intellectual disability, epilepsy and schizophrenia
- Environmental factors may present at independent risk or may affect gene function in individuals with genetic predisposition (i.e. medications, prenatal influences – short interpregnancy interval, multiple gestation, maternal obesity, gestation bleeding, infections; preterm birth, low birth weight, fetal growth restriction, neonatal encephalopathy)
- Also proposed that children with ASD-associated copy number variants may be susceptible to environmental insult in form of maternal immune activation – needs further research

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Content Category 11- Attention Deficit Hyperactivity Disorder

ABP Content Specs for Developmental-Behavioral Pediatrics Fellow Collaboration Project

Prepared by Anishia Srinivasan, MD, Seattle Children's/UW

Reviewed by Emily Myers, MD, Samuel Zinner, MD, Seattle Children's

11. Attention Deficit Hyperactivity Disorder (ADHD)

A. Etiology

1. Know the epidemiology of ADHD
2. Understand current concepts regarding the underlying neuropsychologic deficits in ADHD
3. Know current research on neuroimaging in ADHD
4. Understand the current research on the neurobiologic etiology of ADHD
5. Know the prenatal and environmental exposures that can cause ADHD symptoms
6. Know the environmental situations that exacerbate ADHD symptoms
7. Know the familial pattern of ADHD
8. Understand the evidence relating specific neurotransmitters to ADHD

B. Evaluation

1. Distinguish between ADHD, problems with and normal variations in activity, impulsivity, and attention at different ages
2. Know the diagnostic criteria for ADHD and differences in criteria between childhood and older adolescents
3. Understand the limitations of the diagnostic criteria for ADHD
4. Know the characteristics and diagnostic criteria for the presentations of ADHD
5. Understand the severity levels of ADHD
6. Know the differential diagnosis of ADHD
7. Recognize the common co-morbid externalizing conditions in ADHD
8. Recognize the common co-morbid internalizing conditions in ADHD
9. Know the motor deficits commonly associated with ADHD
10. Know the common speech and language deficits associated with ADHD
11. Know how to plan the evaluation for children with ADHD
12. Know the issues associated with the assessment of preschool-age children for ADHD
13. Know the issues associated with the assessment of adolescents for ADHD
14. Know the learning disorders commonly associated with ADHD
15. Recognize the frequent co-occurrence of ADHD and tic disorders

C. Treatment

1. Know how to plan the pharmacological management of children with ADHD
2. Know the appropriate dosing of short acting and extended release stimulant medications in treating ADHD
3. Know the limitations and benefits of tricyclic antidepressants in treating ADHD
4. Know the benefits, limitations, and side effects of centrally acting alpha-adrenergic agonists in treating ADHD
5. Understand the rationale and risks associated with combining medications in treating ADHD
6. Know the behavioral management of ADHD in the home setting
7. Know the behavioral management of ADHD that can be utilized in the classroom

8. Know how to plan a comprehensive intervention program for children with ADHD
9. Know the mechanism of action of the stimulants in the treatment of ADHD
10. Know the benefits, limitations, and side effects of short acting and extended release stimulant medications in treating ADHD
11. Know the benefits, limitations, dosing, and side effects of norepinephrine reuptake inhibitors in treatment of ADHD
12. Know the mechanism of action of centrally acting alpha-adrenergic agonists in treating ADHD
13. Know the appropriate dosing of short acting and extended release centrally acting alpha-adrenergic agonists in treating ADHD

D. Outcome

1. Know the relationship between ADHD and motor vehicle accidents
2. Know the range of prognoses for children with ADHD
3. Understand the natural history of ADHD
4. Understand the relationship between ADHD and problems with peer relationships
5. Understand the relationship between diet and ADHD symptoms

DBP Boards Study Guide
Attention Deficit Hyperactivity Disorder (ADHD)
Content Category 11

Author: Anisha srinivasan, MD
Reviewers: Emily Myers, MD & Samuel Zinner, MD

A. Etiology

1. Know the epidemiology of ADHD

- The best estimate of worldwide ADHD prevalence based on a meta-analysis is 7.2%. The prevalence rate is ~10% when looking at parent survey data from the National Health Interview Survey (NHIS).
- In the last 20 years, ADHD prevalence has risen for a combination of reasons, including increased awareness and more widely available diagnostic tools.
- There are sex disparities in diagnosis [males (14%) versus females (6%)]. There are many theories as to why; one often mentioned is that girls present less often with hyperactive/impulsive symptoms, but with more inattentive symptoms, which tend to go underrecognized. Girls with ADHD also experience substantial functional impairments and are at risk of the same coexisting conditions as boys.
- In large community adult studies, the prevalence is similar between sexes so it is likely that ADHD is underrecognized in females in childhood.
- There are also racial disparities in diagnosis, with a higher prevalence seen in non-Hispanic White children (11.5%) versus non-Hispanic Black children (8.9%), and Hispanic children (6.3%).

2. Understand current concepts regarding the underlying neuropsychologic deficits in ADHD

- Executive function impairments are primarily responsible for hyperactive and inattentive behaviors.
- There are 3 core executive functions- 1) cognitive flexibility, 2) inhibitory control, and 3) working memory.
- The 3 core executive functions are necessary to perform higher-order executive functions such as problem solving, reasoning, and planning.
- Based on a recent meta-analysis, neurocognitive deficits in individuals with ADHD have been found in the domains of reaction time variability, intelligence/achievement, vigilance, working memory, and response inhibition.

3. Know current research on neuroimaging in ADHD

- Neuroimaging research has shown that brains of children with ADHD are 3-4% smaller in volume than that of children without ADHD.
- Neuroimaging in children with ADHD is associated with a delay in cortical maturation, most prominently in the prefrontal regions.
- Pathophysiological features are not just regional but likely involve large-scale neuronal networks such as the frontostriatal circuits and frontal-to-parietal cortical connections.
- Neuroimaging is not currently used for diagnosis due to substantial overlap of findings with the general population.

4. Understand the current research on the neurobiologic etiology of ADHD

- Genetics plays the largest role in the expression of ADHD, with heritability estimated at 76%
- ADHD is associated with dysfunctions in the prefrontal cortex (PFC) association circuits, which are responsible for regulating attention, behavior, and emotions. The right hemispheric prefrontal cortex specializes in behavioral inhibition.

5. Know the prenatal and environmental exposures that can cause ADHD symptoms

- There are no known causes of ADHD but there is a strong link between genetics and ADHD symptomatology.
- ADHD symptoms are likely a result of multiple interacting genes, and gene-environmental interactions.
- Non-genetic factors such as premature birth or low birth weight, exposure to environmental lead, pesticides, and polychlorinated biphenyls, and brain injury may interact with genetic predisposition to cause ADHD symptoms.
- Maternal smoking and stress were thought to be linked with ADHD symptoms, but emerging research is shedding doubt on this link.

6. Know the environmental situations that exacerbate ADHD symptoms

- Watching too much television, eating sugar, family stress, and traumatic experiences do not cause ADHD symptoms but these factors may exacerbate ADHD symptoms for some children.
- Parenting difficulties and certain parenting styles may make ADHD symptoms worse but do not cause the disorder.

7. Know the familial pattern of ADHD

- ADHD has a very strong genetic component, with multiple twin studies estimating heritability at ~76% .
- Parents and siblings of a child with ADHD are much more likely to have ADHD themselves.
- Adoption studies suggest that genetics plays a larger role than shared environmental factors in ADHD.

8. Understand the evidence relating specific neurotransmitters to ADHD

- There have been strong associations between ADHD and genes regulating dopamine, norepinephrine, and serotonin pathways in the brain.
- The prefrontal cortex (PFC) requires an optimal balance of stimulation from catecholamines like noradrenaline and dopamine. Genetic differences in ADHD are linked to weakened signaling of the catecholamine signaling systems in the PFC.
- Stimulants improve ADHD symptoms by improving the catecholamine signaling in the PFC.

B. Evaluation

1. Distinguish between ADHD, problems with and normal variations in activity, impulsivity, and attention at different ages

- It is normal, especially for children younger than 6 years of age, to be easily distractible, highly active, and impulsive.
- For children and youth with ADHD, these symptoms tend to be more severe, are long-lasting and significantly impair their ability to function at home, school, and/or in the community.

2. Know the diagnostic criteria for ADHD and differences in criteria between childhood and older adolescents

- Diagnostic criteria are based on the DSM-5 (see table below).
- For children and youth <16 years of age, diagnosis requires meeting 6/9 symptoms in either or both the inattentive or the hyperactive-impulsive domain.
- For adolescents and adults $\geq$ 17 or older, diagnosis requires meeting 5/9 symptoms in either or both domains.
- There are 3 different subtypes: Impulsive/hyperactive type, inattentive type, and combined type. Combined type is the most common, but more common in males than in females.
- Complex ADHD is defined by:
  - age of presentation < 4 years or > 12 years *or*
  - presence of coexisting condition (neurodevelopmental, mental health, medical or psychosocial factors adversely impacting health) *or*
  - moderate to severe functional impairment *or*
  - diagnostic uncertainty *or*
  - inadequate response to treatment.

Inattentive symptoms	Hyperactive-Impulsive symptoms
Often fails to give close attention to details or makes careless mistakes in schoolwork, at work, or with other activities.	Often fidgets with or taps hands or feet, or squirms in seat.
Often has trouble holding attention on tasks or play activities.	Often leaves seat in situations when remaining seated is expected.
Often does not seem to listen when spoken to directly.	Often runs about or climbs in situations where it is not appropriate (adolescents or adults may be limited to feeling restless).
Often does not follow through on instructions and fails to finish schoolwork, chores, or duties in the workplace (e.g., loses focus, side-tracked).	Often unable to play or take part in leisure activities quietly.
Often has trouble organizing tasks and activities.	Is often "on the go" acting as if "driven by a motor".
Often avoids, dislikes, or is reluctant to do tasks that require mental effort over a long period of time (such as schoolwork or homework).	Often talks excessively.
Often loses things necessary for tasks and activities (e.g. school materials, pencils, books, tools, wallets, keys, paperwork, eyeglasses, mobile telephones).	Often blurts out an answer before a question has been completed.
Is often easily distracted	Often has trouble waiting their turn.
Is often forgetful in daily activities.	Often interrupts or intrudes on others (e.g., butts into conversations or games)

3. Understand the limitations of the diagnostic criteria for ADHD

- Core symptoms are inattention, hyperactivity, and impulsivity. Core symptoms are not always observable in the clinical setting, which is why rating scales are used.
- Rating scales have some good evidence in population studies but at an individual level can be highly subjective based on the rater (For example, it is not uncommon for 2 co-habitant parents to rate their child very differently from each other).
- It is essential that rating scales are filled out by >1 adult, each of whom spends time with the child in different settings (home, school, work, community).
- In adolescents, self-reports should also be used.

- It is essential that all rating scales and/or symptoms checklist used to make the diagnosis include functional impairment criteria. In other words, symptoms must not only be present, but also cause performance (academic, social, and/or work) impairment. Studies have shown that prevalence rate of ADHD diagnosis drops significantly when functional impairment is required for a diagnosis.
- The sensitivity and specificity of the rating scales vary and are not 100%. See chart below for comparison of the most commonly used rating scales.

Scale	Sensitivity (pooled)	Specificity (pooled)	Diagnostic Odds Ratio (accuracy)
Child Behavior Checklist- Attention Problem (CBCL-AP)	0.77 (0.69 – 0.84)	0.73 (0.64 – 0.81)	9.37 (5.71 – 15.38)
Conners Parent Rating Scale – Revised (CPRS-R)	0.75 (0.64 – 0.84)	0.75 (0.64 – 0.84)	8.95 (3.39 – 23.61)
Conners Teacher Rating Scale – Revised (CTRS-R)	0.72 (0.63 – 0.79)	0.84 (0.69 – 0.93)	13.68 (4.22 – 44.29)
Conners Abbreviated Symptom Questionnaire (ASQ)	0.83 (0.59 – 0.95)	0.84 (0.68 – 0.93)	26.72 (4.15 – 171.87)
Vanderbilt Parent Rating Scale	0.80	0.75	

- Studies have shown racial differences in ADHD symptom rating scales. Use caution when interpreting symptom ratings in non-White patients, particularly in African-American patients. In children from racially diverse backgrounds, gathering information about functional impairment and direct classroom observations are even more critical to making the diagnosis.
- ADHD is a “diagnosis of exclusion”. Most children with ADHD also have non-ADHD co-existing features that may partially overlap ADHD symptoms and/or exacerbate ADHD symptoms. Similarly, environmental circumstances (toxic stress, poverty, abuse, etc.), health conditions (e.g., sleep disorders, visual or hearing impairment), and the presence of other mental health disorders (such as a mood or anxiety disorder) must be taken into account. Table 1, “Differential Diagnosis”, below, explores others. These potential contributions must be excluded as the full explanation for the ADHD symptoms in order to diagnose ADHD.
- Forgetfulness, disorganization, inability to follow directions, and inability to complete tasks are also symptoms that maybe seen in children living in chronic or toxic stress and/or children with a mood disorder.

4. Know the characteristics and diagnostic criteria for the presentations of ADHD

- Symptom onset must be < 12 years of age.
- Symptoms should be present for ≥6 months and occur in ≥2 settings.
- It is important to ensure that these symptoms are not better explained by another mental health disorder particularly depression or anxiety.

5. Understand the severity levels of ADHD

- 3 levels of severity exist: mild, moderate, and severe
- Level of severity is slightly arbitrary and is left up to the clinician's judgement:
  - "*Mild*" indicates that few to no symptoms in addition to those required to make the diagnosis are present.
  - "*Severe*" indicates that several symptoms in addition to those required to make the diagnosis are present.
  - "*Moderate*" is anywhere in between mild and severe.

6. Know the differential diagnosis of ADHD

- ADHD can co-occur with other mental health disorders, but determine which disorder is the primary source of impairment.
- Differential diagnoses include seizure disorders, central nervous system trauma or infection, hearing impairment, visual impairment, sleep disorders, hyperthyroidism, physical or sexual abuse, and substance abuse.
- Certain conditions that coexist with ADHD such as learning disorders, mood disorders, sleep disorders, autism spectrum disorder, disruptive behavior disorder, cognitive disorders, and substance abuse are also on the differential for ADHD.

TABLE 1 DIFFERENTIAL DIAGNOSIS OF ADHD.
TABLE FROM "DEVELOPMENTAL BEHAVIORAL PEDIATRICS" TEXTBOOK CHAPTER ON "ATTENTION AND DEFICITS OF ATTENTION"

ADHD symptom caused by or overlapping with another concern	Alternative diagnoses to consider	Red Flags suggestive of a non-ADHD Diagnosis	Other considerations

Inattention	Hearing impairment Vision impairment Seizure disorder Sleep disorder Anxiety disorder	Excessive worries Fearfulness	Cognitive level
Talks excessively	Anxiety disorder Bipolar disorder	Excessive worries Fearfulness Grandiosity	
School failure or underachievement	Learning disorders Adjustment disorder Psychosocial stressors	ADHD symptoms only in settings requiring academics (school; homework)	Consider behaviors during summer versus school year
Fidgety	Anxiety disorder Tic disorder Stereotypies	Repetitive vocal or motor movements	High level of activity but not affecting performance

7. Recognize the common co-morbid externalizing conditions in ADHD

- Disruptive behavior disorders (which include oppositional defiant disorder and conduct disorder) co-occur in 40% of children with ADHD.
- Common symptoms of externalizing conditions include anger outbursts, defiance of adult/authority figures, aggression towards other people or animals, destruction of property, lying, stealing, and delinquency.

8. Recognize the common co-morbid internalizing conditions in ADHD

- Internalizing conditions such as anxiety and depression often coexist with ADHD.
  - 30% of children with ADHD have an anxiety disorder.
  - 14% of children with ADHD have depression.

9. Know the motor deficits commonly associated with ADHD

- Motor coordination deficits are usually present in children with ADHD.
- Motor coordination deficits are seen more often in children with inattentive ADHD or combined ADHD compared to those with hyperactive-impulsive ADHD.
- Motor impairments may exist in the domains of strength, visual-motor coordination, modulating speed, and dexterity. Such children may appear clumsy, or “dyspraxic.”
- Manual dexterity, including impact on handwriting, is often the most impaired domain of motor coordination.

- Motor coordination difficulties likely stem from abnormalities in the nigrostriatal pathway.

10. Know the common speech and language deficits associated with ADHD

- Speech and language deficits impairments can exist in receptive, expressive, pragmatic speech and language domains.
- These deficits may cause difficulty understanding instructions, making social inferences, and having conversations.
- Speech and language deficits may impair social functioning.

11. Know how to plan the evaluation for children with ADHD

- In DBP, we are often assessing children with complex ADHD and therefore a comprehensive medical and psychosocial assessment is essential. With a comprehensive assessment, coexisting conditions are identified in 70% of children with ADHD.
- There is no single evaluation protocol to assess for ADHD. The evaluation is based on the child's severity level, functional impairment, and developmental level.
- The goal of the evaluation is not just to rule in/out ADHD but also to evaluate for coexisting conditions and differentiate between ADHD and other psychiatric disorders. A comprehensive assessment helps guide appropriate behavioral, social and environmental, academic, and pharmacological interventions.
- A comprehensive evaluation should include:
    - verification of previous diagnosis and assess for coexisting conditions,
    - comprehensive medical history and physical examination,
    - psychological assessment that is developmentally appropriate, culturally sensitive, and includes data from multiple individuals and settings (home, school, community). Psychological assessments can include rating scales, structured interviews, and/or performance based tests.
    - Assessment of how symptoms are functionally impairing at home, school, and/or community settings.
- For children < 4 years of age or those with suspected coexisting conditions, evaluate developmental and cognitive status using formal, standardized assessments.
- For children with ADHD and coexisting learning disorders, psychological evaluation in addition to the school psychoeducational evaluation may be important especially if current school interventions are insufficient. Psychological evaluation here refers to formal measures of cognitive ability and academic achievement, which usually involve assessments involving diagnostic interviews, standardized questionnaires, and/or direct assessment using psychometric instruments.
- For children with ADHD and behavioral disorders, a functional behavioral assessment (FBA) is helpful.
- For children with other neurological conditions (traumatic brain injury, brain tumor, stroke, spina bifida), neuropsychological testing is usually necessary. Neuropsychological testing refers to measurement of cognitive functions (such as memory, executive function) in addition to those used in formal psychological testing. This is typically measured directly using psychometric instruments.

12. Know the issues associated with the assessment of preschool-age children for ADHD

- Hyperactive and impulsive behavior is part of typical development in preschoolers and it generally declines with age. This complicates ADHD diagnosis in preschool age children. There is little research on diagnostic thresholds of ADHD in preschoolers.
- Symptom duration must be present for at least 9 months in this age group, compared to the 6-month criterion used for older children and adolescents. This is because in many preschool children with parental or teacher concern about ADHD symptoms, symptoms remit at 12-month follow up.
- Preschoolers have similar patterns of coexisting conditions and functional impairment as older children but the presentation of symptoms differs from older children.
- Preschoolers diagnosed with hyperactive-impulsive ADHD may later meet the criteria for either predominantly inattentive or combined ADHD when older. The hyperactive-impulsive subtype of ADHD is most common in preschool whereas the inattentive subtype of ADHD is common in school-age children.
- Rating scales are the most practical way to assess for ADHD in this group such as:
  - ADHD Rating Scale-IV-Preschool Version,
  - ADHD Symptoms Checklist-4,
  - Attention Deficit Disorders Evaluation Scale-Third Edition (ADDES-3),
  - Behavior Assessment Scale for Children-2 (BASC-2),
  - Behavioral Rating Inventory of Children,
  - Child Behavior Checklist for Ages 1.5 to 5 (CBCL),
  - Children's Problems Checklist,
  - Conners Early Childhood,
  - Early Child Inventory-4,
  - Preschool and Kindergarten Behavior Scales- Second Edition (PKBS-2).
- Structured interviews useful in this age group include the Diagnostic Interview School, Young Children (DISC-YC) and the Preschool Age Psychiatric Assessment (PAPA).
- There is very little research on performance-based tests in this age group.
- The largest study ever done on preschool ADHD was the multisite, clinical trial, Preschool ADHD Treatment Study (PATS). PATS has informed many clinical guidelines on preschool ADHD treatment.

13. Know the issues associated with the assessment of adolescents for ADHD

- DSM-5 criteria require presence of symptoms prior to 12 years of age, but recalling childhood symptoms can be difficult and fraught with recall bias.
- Symptom-onset after 12 years of age is one of several forms of "complex ADHD."
- Many of the DSM-5 criteria ("runs about or climbs" "often unable to play") are not applicable to adolescents.
- Middle and high school teachers are less useful informants of an individual student's behavior compared to elementary school teachers because teachers of these older students spend much less time with each student. Inter-teacher rating scale agreement is poor at the secondary school level.
- Adolescent self-report rating scales often underestimate ADHD symptoms and level of impairment compared to the parent and teacher ratings.

- By adolescence, signs and symptoms of many of the co-existing conditions have arisen, so a comprehensive assessment is even more essential.

14. Know the learning disorders commonly associated with ADHD

- 30-50% of children with ADHD also have a learning disorder.
- Dyslexia and dyscalculia are *specific* learning disorders and are the most common learning disabilities seen with ADHD.
- Many children with ADHD who do not meet criteria for a learning disorder nevertheless do have some degree of learning difficulties.

15. Recognize the frequent co-occurrence of ADHD and tic disorders

- 7% of those with ADHD also have tics or Tourette Syndrome but 50-70% of those with Tourette syndrome have ADHD.

C. Treatment

1. Know how to plan the pharmacological management of children with ADHD

- Ideally, psychosocial treatment should already be in place before starting pharmacological treatment of ADHD.
- Prior to starting pharmacological treatment:
  - Patients and families should receive psychoeducation about risks, benefits, side effects of medications.
  - Assess for any symptoms identical to side effects that already exist (such as headache, abdominal pain, poor appetite) so they are not later confused with medication side effects.
  - In children with tic disorders, it is safe to provide stimulant medication. Tics wax and wane in severity naturally, and families should be alerted that stimulant medications are unlikely to influence tic behaviors.
  - Baseline ADHD-specific rating scales should be obtained to understand the degree of functional impairment.
  - Identify target symptoms and treatment goals to inform treatment progress.
- Initial pharmacological treatment should be with either of 2 classes of stimulants-methylphenidates or amphetamines. Selection of one vs. the other class is mostly arbitrary. About 40% of children respond better to one or the other, and about 40% responding equally well to either. If there is a personal or family history of cardiovascular symptoms, a more detailed cardiologic workup maybe necessary prior to starting stimulants. In general, treatment should begin at the lowest reasonable dose with slow upward titration at weekly intervals until maximal functional improvement is seen with tolerable side effects.

- Treatment effects should be assessed using ADHD-specific rating scales such as the Vanderbilt Assessment Scale.
- If one stimulant class appears insufficiently effective, consider switching to the other stimulant class.
- If there are significant side effects of treatment, consider adjusting the dose, switching to an alternative medication, or adding an adjunct medication.
- During the initial titration period, patients should be seen in clinic every 3-4 months to monitor side effects and response to treatment. Once stable, patients may be seen on an annual basis to monitor for coexisting conditions.

2. Know the appropriate dosing of short acting and extended release stimulant medications in treating ADHD

- When starting stimulant medication treatment for ADHD, first decide which stimulant class to use (methylphenidate versus amphetamine) and then decide the desired duration of action of the preparation (long-acting versus short-acting).
- Once-daily preparations are preferred for pediatric patients.
- The starting dose of any stimulant medications for most children is 2.5 to 5 mg, with a suggested daily dose ranging from 0.3 to 1mg/kg for amphetamines and 0.6 to 2mg/kg for methylphenidate.
- After initial dosing, the dose for each child or adolescent can be titrated based on therapeutic response and side effects.
- Extended-release formulations are available in osmotic release oral system (OROS), beaded, transdermal and liquid forms.
    - OROS formulations (such as Concerta) are made of an osmotic pump system, which allow for controlled release of the drug. OROS capsules have triple-release. One the outer layer is drug that is released within 10-15 minutes of taking the medication. Under this outer layer is a push system that is activated by water and allows for release of drug from 2 inner compartments. This type of release allows for an extended effect. OROS capsules must be swallowed whole.
    - Beaded formulations (such as Metadate CD, Ritalin-LA, Adderrall XR) consist of capsules that contain beads that can be sprinkled into food. There are generally 2 types of beads within a capsule—immediate release and slow release. This allows for extended coverage.
    - Other capsule formulations (such as Aptensio XR or Focalin XR) can be taken whole as a capsule or opened and mixed in with food.
    - Liquid formulations (such as Quillivant XR or Dynavel XR) are useful for those who cannot swallow pills or have difficulty with beads. Liquid formulations are also easier to titrate than the other forms.
    - Transdermal formulations (ie, Daytrana) come in the form of a patch. These formulations can be useful for those who cannot swallow pills. They also allow for bypassing the GI tract as they are absorbed through the skin immediately into the

bloodstream. Immediate release in not possible with this formulation and typically takes about 2 hours to get to therapeutic level.

3. Know the limitations and benefits of tricyclic antidepressants in treating ADHD

- Tricyclic antidepressants (TCA) act as SNRIs (serotonin norepinephrine reuptake inhibitors) and thereby increase the concentration of these neurotransmitters at the synaptic junction.
- There are 3 TCAs used in treatment of ADHD: desipramine, imipramine, and nortriptyline. Of these, desipramine is the best studied.
- Desipramine has been found to be effective in children with ADHD who failed to respond to stimulants.
- An important benefit of TCAs is the positive effect they have on co-occurring conditions such as anxiety, depression, and tic disorders.
- However, TCA use is limited due to adverse cardiovascular side effects such as elevated blood pressure, heart rate, and conduction defects.

4. Know the benefits, limitations, and side effects of centrally acting alpha-adrenergic agonists in treating ADHD

- These agents tend to be more effective for ADHD symptoms of hyperactivity/impulsive behavior than for inattention.
- Drowsiness, headache, fatigue, nausea and abdominal pain are common side effects seen with guanfacine.
- Sedation, irritability, low blood pressure, and rebound hypertension can be seen with clonidine.
- The cardiovascular side effects (bradycardia, syncope, low blood pressure) are less common with guanfacine than with clonidine.
- Alpha-2-adrenergic agonists are useful as adjunctive or alternative agents to stimulant medications.

5. Understand the rationale and risks associated with combining medications in treating ADHD

- Rationales for combining medication in treating ADHD include:
  - Greater therapeutic effect from a combination of medications while minimizing side effects of each individual medication.
  - Treatment of coexisting conditions such as anxiety, depression, tic disorders, or sleep disorders.
  - Extending coverage beyond stimulant duration.
- Risks of combining medications include adverse drug interactions, decreased adherence to medication regimen, and increased side effects.

- Guidelines recommend treating the most impairing condition and avoiding polypharmacy as this has limited evidence.

6. Know the behavioral management of ADHD in the home setting

- Behavior parent training is an essential component of the multimodal treatment of ADHD.
- Treatment begins with behavior parent training alone, without use of prescription medication, for children under 6 years of age. Medication may be added in this age group as needed.
- A home environment that provides routine, structures, clear expectations and limits is necessary to optimal for ADHD management.
- An effective discipline system is one that is consistent across caregivers and includes both rewards for appropriate behavior and loss of privileges for misbehavior.
- Daily special time (a daily time for positive interaction without negative feedback), assisting children with social skills, and helping children identify their strengths are also helpful.

7. Know the behavioral management of ADHD that can be utilized in the classroom

- Classrooms that are structured with consistent rules and routines, smooth transitions between activities, minimal distracting classroom displays are helpful in behavior management of ADHD. Establishing and practicing rules for routines and classroom tasks with students also help.
- Providing positive feedback that is specific is powerful in changing behavior.
- Information delivery in the classroom should be done in ways that provide multisensory instruction and gives one instruction at a time. Lesson plans should minimize interruptions. Large projects should be broken up into a series of small tasks with deadlines.
- Some other classroom accommodations may include preferential seating (single desks, away from distraction, near teachers or responsible peers), shortened assignments, and/or allowing fidget objects.

8. Know how to plan a comprehensive intervention program for children with ADHD

- The goal of ADHD treatment is improvement in function, especially long-term functional outcomes.
- A comprehensive intervention program (multimodal treatment) for children with ADHD includes both pharmacological and psychosocial treatment. Psychosocial treatment includes behavior parent training and behavioral classroom management. Both should be part of the intervention plan from the outset of treatment.
- Shared decision-making and family engagement are essential in planning multimodal treatment.
- ADHD medication use is lower in children of color and children who live in poverty. This maybe partially due to decreased awareness and different perceptions of ADHD medication, therefore family engagement is essential.

9. Know the mechanism of action of the stimulants in the treatment of ADHD

- Stimulants block the reuptake and increase release of dopamine and norepinephrine in the extraneuronal space, thereby increasing the concentration of these neurotransmitters at the synaptic junction.

10. Know the benefits, limitations, and side effects of short acting and extended release stimulant medications in treating ADHD

- Stimulant treatment for ADHD has shown a large effect size and a response rate of 70%.
- Data on preschoolers aged 3-5 years is less robust and suggests that this group may have a lower response rate to stimulants. The Preschool ADHD treatment study (PATS) is the largest preschool stimulant treatment study done in 165 children. Of these about 85% were thought to respond to methylphenidate. Data on stimulant efficacy for ADHD is also limited in adolescents, though current research suggests moderate response.
- Among adolescents with ADHD with prescriptions, there is a higher rate of diversion and misuse of stimulant medications. Prevention strategies such as medication contracts, pill counts, prescribing small number of pills, and prescribing long-acting formulations are some ways to reduce risk of diversion and misuse.
- It should be noted that most studies on stimulant efficacy are conducted on white males, with limited available information on females and children of color.
- Common side effects of stimulants include delayed sleep onset, headache, appetite suppression, and abdominal pain. Loss of appetite is the most common sustained side effect.
- There is a small but clinically insignificant reduction in weight with stimulant use. Children treated with stimulants grow 1.0-1.5 cm lower than their expected height during the first 2 years of treatment, though there maybe catch-up growth with chronic treatment.
- Stimulant therapy for ADHD does not cause emergence of or exacerbation of tic disorders. This hypothesis has been largely disproven by several well-controlled studies.
- Sudden cardiac death is an extremely rare event in children with ADHD and there is not sufficient data to suggest a causal link between stimulant use and sudden death. However, blood pressure and pulse should be monitored closely during stimulant treatment. A family history of premature sudden death and a personal history of cardiovascular symptoms should be obtained as well.
- Psychosis or mania is a rare adverse side effect seen with stimulant therapy, and in most cases, resolves with stimulant discontinuation. An alternative agent should be considered in the case of psychotic or manic symptoms.
- Over the first 6 months of treatment, methylphenidates are associated with a mild tolerance and may require a 20-30% escalation of dose to maintain effectiveness.

11. Know the benefits, limitations, dosing, and side effects of norepinephrine reuptake inhibitors in treatment of ADHD

- Atomoxetine is the norepinephrine reuptake inhibitor used in treatment for ADHD.

- Unlike stimulant medications, full therapeutic effect of atomoxetine may not be seen for up to 8 weeks.
- Somnolence, decreased appetite, nausea, and vomiting are common side effects seen with atomoxetine. Insomnia, irritability, and mood swings may also occur but are less common. Small reductions in weight and height can be seen, though less than with stimulants.
- Benefits of atomoxetine include that it has some therapeutic effect for co-occurring conditions such as oppositional defiant disorder, conduct disorder, and anxiety disorders. For patients with significant anxiety symptoms and ADHD, atomoxetine may be considered as first line.
- Atomoxetine is less effective than stimulants in ADHD treatment. Atomoxetine also has a Black Box warning of suicidal behavior and liver toxicity. All of these features limit its use.
- The maximum daily recommended dose is 1.4 mg/kg/day. Dosing should begin at 0.5mg/kg/day, slowly increasing after a minimum of 3 days to the target dose. BID dosing improves tolerability compared to once daily dosing.

12. Know the mechanism of action of centrally acting alpha-adrenergic agonists in treating ADHD

- There are 2 primary centrally acting alpha-adrenergic agonists used in treating ADHD: clonidine and guanfacine.
- Moderate stimulation of the alpha-2 adrenergic receptors improves the prefrontal cortex's ability to regulate attention, behavior and emotions.
- Clonidine and guanfacine both stimulate the alpha-2 adrenergic receptors on the prefrontal cortex (PFC) neurons, mimicking norepinephrine's actions in the PFC.
- Guanfacine is more selective for alpha-2 adrenergic receptors than is clonidine, and therefore has less sedative and hypotensive side effects.

13. Know the appropriate dosing of short acting and extended release centrally acting alpha-adrenergic agonists in treating ADHD

- Clonidine is available in both short acting and extended release formulations. Behavioral effect of the short-acting clonidine last for 3 to 6 hours, whereas the extended release formulation can have effect up to 12 hours.
- Short-acting formulations require TID or QID dosing, whereas extended release formulations should be dosed BID.
- For ADHD treatment, clonidine is generally dosed at 3-5mcg/kg/day. It is recommended to start treatment at a low dose and increase slowly to minimize cardiovascular and sedation side effects. A general rule is not to increase the dose more than 0.05 mg every 3 days.
- Extended release clonidine dosing should be initiated at 0.1mg at bedtime, followed by 0.1mg BID.
- Short-acting clonidine is available in oral and transdermal forms. It is recommended that if using the short acting formulations, begin with the oral form and switch to transdermal once the optimal dose has been determined.

- Guanfacine is 10 times less potent than clonidine and therefore 1 mg of guanfacine is equivalent to 0.1mg of clonidine
- Short-acting guanfacine dose can range from 0.5 mg to 4mg/day given on a BID or TID schedule
- Extended release guanfacine dose can range from 0.05 – 0.08 mg/kg and is administered once daily.
- Guanfacine has a longer half-life than clonidine and therefore dose should be increased at a slower rate, with a suggested titration of 0.5mg dose increase every 3 to 4 days.

D. Outcome

1. Know the relationship between ADHD and motor vehicle accidents

- Adolescents with ADHD are at greater risk for motor vehicle accidents, speeding tickets, and reckless driving.
- Adolescents with ADHD are also more likely to drive with a suspended license or without a license.
- Adolescents and adults with ADHD are particularly at risk of distractions during low stimulus driving settings like long-distance or highway driving. Distractions include texting, talking, changing radio stations, checking make up, eating, drinking, etc.
- Pharmacological treatment of ADHD is associated with improved driving performance and decreased motor vehicle accident rates in adolescents with ADHD.

2. Know the range of prognoses for children with ADHD

- It should be noted that most of studies on ADHD prognosis evaluated small groups of mostly white males, so the generalizability is limited
- Most children with ADHD will also develop 1 or more coexisting condition during their lifetime. It is rare for children to have “pure” ADHD without any coexisting conditions.
- Approximately 60% of those with ADHD have at least 1 coexisting psychiatric condition, and 35% have two or more coexisting psychiatric conditions.
- There are few studies looking at long-term outcomes of children diagnosed with ADHD. From what we know currently, those with ADHD are at greater risk of:
    - Sustaining unintentional injuries
    - Getting into motor vehicle accidents
    - Impaired academic functioning (less schooling, lower achievement scores, failing classes)
    - Engaging in substance abuse
- Important childhood predictors of positive adult outcomes include high IQ, higher household income, and decreased ADHD symptom severity. These children tend to have better

educational, emotional, and occupational outcomes. They are also less likely to engage in risky sexual behaviors or have police involvement in adulthood. Therefore, children who do not have these positive predictors, require additional attention and assistance.

3. Understand the natural history of ADHD

- At least 2/3 of children with ADHD have persistent symptoms into adulthood.
- Hyperactive symptoms generally decrease over time, but inattentive symptoms and executive function deficits persist.
- Significant functional impairment maybe worse in adulthood due to increased demands on individuals to perform day-to-day activities.
- Preschoolers with ADHD tend to have a predominantly hyperactive-impulsive presentation, school age children tend to have a combined presentation, and adolescents tend to have a predominantly inattentive presentation.

4. Understand the relationship between ADHD and problems with peer relationships

- Children with ADHD tend to have difficulty making and maintaining friendships, which is thought to be due to impaired executive function.
- It is thought that children with ADHD have acquired appropriate social skills but do not always use them efficiently, which causes problems with peer relationships.
- Social skills training is most helpful for children with ADHD when it is embedded within an intensive behavioral intervention, such as a specialized summer program.

5. Understand the relationship between diet and ADHD symptoms

- Children who eat a substantial breakfast are able to focus longer on tasks in late morning than those who do not. Better cognitive effects are seen throughout the morning with a protein-rich or balanced carbohydrate-protein diet.
- Several nutrient deficiencies have been associated with ADHD including iron, zinc, magnesium, and polyunsaturated fatty acids. These deficiencies are not associated with worsening of ADHD symptoms.
- Several studies have looked at the impact of diet and certain food groups on worsening or improving ADHD symptoms and none thus far have found a relationship between diet and ADHD.
- The following foods/diets have *not* been found to change ADHD symptomatology:
    - Sugar intake
    - Gluten intake
    - Vitamin or trace mineral deficiency
    - Elimination of artificial colors or preservatives
    - Organic foods compared to non-organic foods
    - Food allergies
- There is some emerging evidence that supplementing the diet of expectant mothers with omega-3 fatty acids may reduce the likelihood of a child developing ADHD.

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Content Category 12- Externalizing Conditions

ABP Content Specs for Developmental-Behavioral Pediatrics Fellow Collaboration Project

Prepared by Cassandra Conrad, MD[a], Liesl Windsor, MD[a], Lianna Lipton, MD, MS[a], Melissa Harada, MD[b], Audrey Christiansen, MD[a], Helene Pinches, MD[a]

Reviewed by Demetra Pappas, MD, MPH[a], Irene Koolwijk, MD, MPH[b], Leonard Rappaport, MD, MS[a]

[a]Boston Children's Hospital, [b]UCLA Mattel Children's Hospital

12. Externalizing Conditions

A. Aggressive behavior

1. Differentiate aggressive behavior problems from normal variants at different developmental stages
2. Know the factors associated with aggressive and violent behavior at different ages
3. Plan the behavioral management for aggressive behavior at different ages
4. Know the pharmacological interventions for aggressive behavior
5. Characterize effective preventive programs for aggressive behavior
6. Understand the natural history of aggressive behavior
7. Know how to evaluate a child with aggressive behavior
8. Know the epidemiology of aggressive behavior at various stages of development
9. Understand the association between child maltreatment and later aggressive behavior
10. Understand the etiologies of bullying behavior
11. Know how to advise families on the management of bullying behavior
12. Know how to advise a school or day care center on the management of aggressive or bullying behaviors
13. Understand the components of school or community programs to teach social skills and non-violent conflict resolution
14. Understand the epidemiology and management of biting behavior at various stages of development
15. Understand the influences of exposure to violence in media on aggressive behavior in children and adolescents
16. Know how to plan the management of a child with aggressive behavior
17. Understand the association between corporal punishment and aggression in children, adolescents, and adults

B. Oppositional defiant disorder (ODD)/Conduct disorder/Intermittent explosive disorder

1. Recognize the natural history of ODD/conduct disorder
2. Distinguish between ODD, oppositional behavioral problems, and temperamental variations
3. Recognize the environmental contributors to oppositional and defiant behaviors
4. Recognize the behavioral and functional characteristics of ODD and the variations in presentation based on developmental stage
5. Recognize the common co-morbid conditions of ODD
6. Plan the management of a child with ODD
7. Understand the range of prognoses for children with ODD
8. Recognize the environmental situations that contribute to the development and maintenance of antisocial, aggressive, and delinquent behaviors
9. Recognize the signs and symptoms of conduct disorder at different developmental stages
10. Recognize the common co-morbid conditions of conduct disorder
11. Understand the range of prognoses for children with conduct disorder
12. Know the pharmacologic treatments for conduct disorder
13. Know the psychologic treatments for conduct disorder
14. Describe the characteristics of effective training programs for parents of children with

ODD/conduct disorder

15. Understand the stages of development typically associated with oppositional behaviors
16. Know the diagnostic criteria for ODD
17. Know how to evaluate a child for ODD/conduct disorder
18. Understand the benefits and limitations of different venues (eg, family, community, residential) for the treatment of conduct disorder
19. Know the diagnostic criteria for conduct disorder
20. Recognize family systems factors that contribute to the development and maintenance of ODD, intermittent explosvie disorder, and conduct disorder
21. Know the diagnostic criteria for intermittent explosive disorder
22. Recognize the signs and symptoms of intermittent explosive disorder at different developmental stages
23. Recognize the common co-morbid conditions of intermittent explosive disorder
24. Be able to differentiate intermittent explosive disorder from ODD and conduct disorders
25. Know the psychological and pharmacologic treatments for intermittent explosive

ABP DBP Board Specifications for Externalizing Conditions

Fellow Contributors: Cassandra Conrad, MD[a], Liesl Windsor, MD[a], Lianna Lipton, MD, MS[a], Melissa Harada, MD[b], Audrey Christiansen, MD[a], Helene Pinches, MD[a]

Faculty Mentors: Demetra Pappas, MD, MPH[a], Irene Koolwijk, MD, MPH[b], Leonard Rappaport, MD, MS[a]

[a]Boston Children's Hospital, [b]UCLA Mattel Children's Hospital

A. Aggression

1. Differentiate aggressive behavior problems from normal variants at different developmental stages

	Normal Variant	Behavioral Problem
Toddler	Time-limited aggressive behaviors (crying, hitting, biting, kicking, throwing, temper tantrums) which typically present around 17 months to communicate frustration prior to adequate language development. Behaviors peak at 18-24 months and decrease with language development.	Persistent pattern of tantrums, irritability, anger, and/or blaming others regardless of language development
School-Age	Mild/rare aggressive behaviors towards peers may appear early in school age (teasing, irritability, isolating peers from groups). Aggressive behaviors tend to decrease overtime as social relationships and empathy continue to develop.	**Oppositional Defiant Disorder:** Usually presents before age 8. Symptoms last longer than 6 months. Trouble with authority, breaking rules, persistent anger, noncompliant. **Child Conduct disorder**: Onset before age 10. Three incidences within year, one within past 6 months: Animal cruelty, theft, property destruction, setting fires, law breaking, substance abuse. **Intermittent Explosive Disorder:** Diagnosed after age 6. Marked by recurrent behavioral outburst manifested by either 1) Verbal aggression (e.g., temper tantrums, tirades, verbal arguments or fights) or physical aggression toward property, animals, or other individuals, occurring twice weekly on average, for a period of three months. 2) Three behavioral outburst involving damage of property and or physical assault involving injury against animals or another individual occurring with 12-month period.
Adolescent	Increased limit testing, rebellion, peer pressure, mild resistance to authority	**Adolescent Conduct Disorder**: Onset between 10-16 years. Criteria same as above. May also include weapon use, gang activities, sexual assault. **Antisocial Personality Disorder**: Conduct disorder that persists after age 18. Pervasive disregard for others.

2. Know the factors associated with aggressive and violent behavior at different ages

	Social	Psychiatric	Medical
Toddler	Neglect, psychological maltreatment, physical discipline, exposure to violent media, exposure to domestic violence	Developmental Delay	Lead exposure, congenital adrenal hyperplasia (in males may not be identified until age 5-6 years)
School-Age Adolescent	Psychological maltreatment, media exposure, exposure to domestic violence, peer victimization, Low/inconsistent parental involvement, unstable community	Intellectual disability, language deficits, ADHD, academic underachievement, mood disorders, anxiety, ASD, trauma disorders	Substance abuse
Genetic Syndromes	Cri du Chat, Smith-Magenis, Fragile X, Angelman, Cornelia de Lange, and Prader-Willi		

3. Plan the behavioral management for aggressive behavior at different ages

	Behavioral Management
Overview	Management involves targeting biological, psychological, and environmental risk factors from the individual to community level. Developing positive home environments and minimizing family psychosocial stressors is key.
Toddler	Parent training to develop child's behavioral skills, which focuses on consequences, reinforcement of positive behavior, and emotional regulation of both parent and child.
School-Age and Adolescent	Interventions at individual, parent/family, school, and community level (see board specification 16 for details)

4. Know the pharmacological interventions for aggressive behavior

- Medication should only be considered if psychosocial and behavioral treatment interventions have failed
- Initial medication treatment should target the underlying disorder(s) (ADHD, Anxiety, Depression, other medical etiology)
- Consider adding an antipsychotic medication if severe aggression persists after an adequate trial of treatments for the underlying disorder
- Medication management is effective only in the context of impulsive aggression (vs. covert/premeditated aggression)

	Possible Indication	Side Effects	Monitoring
Alpha 2 Agonists (clonidine, guanfacine)	ADHD	Drowsiness and dizziness, dry mouth, irritability, abdominal pain, headache	
Stimulants	ADHD	**Appetite suppression**, insomnia, abdominal pain, nausea, headache, palpitations, tachycardia, weight loss	
SSRI	Anxiety or Depression	Nausea, vomiting, fatigue, decreased appetite, abdominal pain, somnolence, insomnia, constipation, dry mouth, dizziness, sexual dysfunction, urinary hesitancy, **black box warning for suicidal thinking in**	

		individuals	
Atypical Antipsychotics	ASD, other treatments for underlying etiology failed	**Extrapyramidal symptoms**, **dyslipidemia**, somnolence, **weight gain**, **metabolic syndrome**, sexual dysfunction, hyperprolactinemia, gynecomastia, menstrual irregularity, prolonged QTc	Total Cholesterol AST, ALT Fasting Glucose CBC with differential Prolactin TSH, T4 EKG BMI Abnormal Involuntary Movement Scale

5. Characterize effective preventive programs for aggressive behavior

- Target risk factors including academic failure, problematic family dynamics (i.e., parental mental health challenges, trauma, incarceration, violence, drugs), easy access to firearms and drugs, and community disorganization.
- Successful prevention programs often incorporate mental health systems, the legal system, and community-based recreational and faith systems

6. Understand the natural history of aggressive behavior

- Early risk factors and/or behavioral reinforcement → persistent aggressive behaviors → academic failure and social rejection during school age →deviant peer clustering→ persistent violence and delinquency into adulthood

7. Know how to evaluate a child with aggressive behavior

- Interview (Parent, Child/Adolescent, School)
- Assess for Psychological, Biological, and Environmental Risk Factors
- Validated Scale:
    - Childhood Aggression Scale (age 5-18, parent and teacher)
    - Buss-Perry Aggression Scale (age >9, self)
    - Vanderbilt Assessment Scales (age 6-12, parent and teacher
    - Behavior Assessment System for Children (age 2-25, parent, teacher, and self)

8. Know the epidemiology of aggressive behavior at various stages of development

- Prevalence:
    - Toddler: up to 72%
    - School-age: generally decreases with age
    - Adolescence: up to 25%
- For clinical characteristics and risk factors, see board specifications 1 and 2

9. Understand the association between child maltreatment and later aggressive behavior

- Neglect (especially 0-2years old) – lack of social modeling and bonding/attachment
- Psychological maltreatment – being ridiculed/rejected, witnessing intimate partner violence
- Physical abuse/trauma – highly associated with chronic aggression

10. Understand the etiologies of bullying behavior

- Bullying:
    - Three characteristics: intention to harm, repetitive, and imbalance of power

    - Three types: physical, verbal (taunting, hate speech), social (exclusion, rumor-starting)
- Cyberbullying:
    - Use of some form of electronic communication with the intent to harm another person
    - Victims more likely to experience subsequent mental and physical problems (e.g. suicidal ideation/attempt) than victims of traditional bullying
- Causes: Multifactorial, vary by person/situation including:
    - Popularity – feeling of power/control reinforced by having "followers"
    - "Bully-victim" – child with personal history of being bullied then victimizes others to regain a sense of control.
    - Personality/temperament – children who are more impulsive, aggressive, view violence as being positive are more likely to bully others.
    - Family functioning – lower SES, less parental involvement, exposure to maltreatment, witness to domestic violence more likely to bully others.

11. Know how to advise families on the management of bullying behavior

- Set clear expectations that bullying is never OK
- Model appropriate behavior and show how to resolve conflicts positively.
- Help the child understand how bullying hurts other children. Give real examples of the good and bad results of the child's actions.
- Work with the child's school team (teacher, principal, psychologist/counseling) and parents of victims to find effective ways to address bullying.

12. Know how to advise a school or day care center on the management of aggressive or bullying behaviors

- Stop negative interactions immediately and separate children involved.
- Remain calm and model appropriate problem-solving strategies.
- Ensure that everyone is safe and manage any medical or mental health needs.
- Set clear expectations with consistent consequences for aggressive or bullying behavior.

13. Understand the components of school or community programs to teach social skills and non-violent conflict resolution

- Create unified, school-wide systems engaging all members of the school community, which are more effective than one-time prevention programs.
- Establish clear policies focused on prevention of victimization.
- Enact close monitoring and frequent reassessment of strategies to ensure effectiveness.
- Ensure prompt and consistent interventions using positive discipline.
- Offer social, emotional, and mental health supports for those involved in incidents (bullies, victims, and bystanders).

14. Understand the epidemiology and management of biting behavior at various stages of development

- Epidemiology:
    - Biting may initially occur in infants and toddlers accidently during teething. May also be learned when used during games or play.
    - Older children often bite when frustrated or trying to communicate. After 2-3 years old, children may bite deliberately to scare other children or get what they want.
- Management:
    - Set simple and clear expectations ("No biting").

- Stop biting immediately.
- Consistently praise for not biting and institute consequences for biting behavior (time-out, removal of preferred item).
- Do not bite child to show them how it feels or punish them for biting.
- Teach appropriate ways of making requests, and do not give in when biting is used to get preferred items.

15. Understand the influences of exposure to violence in media on aggressive behavior in children and adolescents

- Social learning theories suggest that children exhibit aggressive behaviors because they observe others acting aggressively and see reinforcement of these behaviors (e.g. watching peers or family members behave aggressively)
- Similarly, exposure to violence in the media is considered one of many risk factors that predispose to aggressive behavior
- Both past and present violent media preferences are correlated to reports of violent/aggressive behavior in children

16. Know how to plan the management of a child with aggressive behavior

- Treatment plan should target psychological, social, and biological risk factors
- Programs to manage a child with aggressive behavior typically focus on the following areas:
  - Development of social skills
  - Improving frustration tolerance
  - Enhancing problem-solving skills
- Intervention ideally focused on individual, family, school/peers, and community
- Individual: May include mental health management, such as cognitive behavior therapy, and/or medication
- Family: Focus on developing positive home environment and reducing family stressors. Parent training programs focus on parent education and skills building, consistency, stress coping strategies, and positive parenting styles
- School: School-based interventions most helpful when focused on promoting positive peer interactions, reducing ostracizing and bullying behaviors, and enhancing academic achievement. If individual has mental health or medical diagnosis, he/she may qualify for services under the IDEA if academic functioning has been impacted.
- Community: Programs are offered for families and children and may include parent training, linkage to other services, and a variety of family supports across areas/risk factors.

17. Understand the association between corporal punishment and aggression in children, adolescents, and adults

- Children exposed to family violence may be more likely to exhibit aggressive behaviors themselves
- Parent verbal aggression and corporal punishment are associated with children's externalizing behavioral issues
  - Causation unclear in part due to bidirectional relationship (for example, children with aggressive behaviors may be more likely to be physically disciplined)
  - In addition, children exposed to corporal punishment are more likely to be exposed to other known risk factors of aggressive behavior

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B. Oppositional defiant disorder (ODD)/Conduct disorder/Intermittent explosive disorder

1. Recognize the natural history of ODD/conduct disorder

- Oppositional behavior can occur in all children from time to time particularly during toddler and early adolescence
- More severe disruptive behavior disorders (e.g. conduct disorder) almost always preceded by oppositional behavior disorders
- Earlier age of symptom onset associated with poorer prognosis
- If behaviors persist, children are at increased risk of psychiatric disorders in adulthood
- Higher number of disruptive behaviors also associated with worse long-term outcomes

2. Distinguish between ODD, oppositional behavioral problems, and temperamental Variations

- ODD:

- DSM-5 criteria: "A pattern of angry/irritable mood, argumentative/defiant behavior, or vindictiveness lasting at least 6 months"
- Need at least 4 symptoms in those categories, which are exhibited during interaction with at least one individual who is not a sibling
- Behavior disturbance associated with distress in individual or others or impacts negatively on function and is not part of another mental health condition

- Oppositionality:
  - Mild opposition with mild negative impact (no one hurt, no damage to property, no significant alteration in parents' plans)
  - Considered normal developmental variation
  - May be observed as a complicating behavior problem in children with other developmental and behavioral disorders

- Temperamental variations:
  - "Difficult" temperament – irregularity, negative response to withdrawal from novel stimuli, slow or poor adaptability to change, intense/negative mood
  - "Goodness of fit" between parenting style and child's temperament matters in how oppositional behaviors evolve

3. Recognize the environmental contributors to oppositional and defiant behaviors

- Socioeconomic disadvantage, including parental unemployment, residing in public housing, and high neighborhood crime
- Physical or sexual abuse
- Adopted children
- Exposure to parental separation

4. Recognize the behavioral and functional characteristics of ODD and the variations in presentation based on developmental stage

- Developmental stage leads to different presentations
- Behavioral symptoms:
  - Frequent temper tantrums
  - Excessive arguments with adults
  - Actively refusing to comply with requests/rules
  - Often questioning rules
  - Deliberately annoying and upsetting others
  - Often touchy/annoyed by others
  - Blaming others for their mistakes
  - Frequent outbursts of anger and resentment
  - Spiteful attitude and revenge seeking
  - Children do NOT typically engage in delinquent behavior

5. Recognize the common co-morbid conditions of ODD

- Rates of ODD are more common in children with ADHD; ADHD is the most common comorbidity.
- Some research indicates that children develop ODD to cope with anxiety or uncertainty

6. Plan the management of a child with ODD

- Treatment is tailored to the needs of each patient. There is no one size fits all treatment.

- Stepped Management:
    - There is a high rate of response to brief interventions including books and printed material with or without psychotherapy. Start with a trial of brief intervention and then move to more intensive behavior therapy and/or parent training.
- Behaviors may occur at home, school or multiple locations. Behavioral interventions should address each context where the behaviors occur.
- Treating comorbid mental health and learning difficulties has been shown to improve behavioral symptoms.

- Parent Training: Help parent and others manage the child's behavior
    - Structured program that empowers caregivers to provide the child with positive feedback, logical consequences, and (rare) brief and specific punishment
    - Examples:
        - Incredible Years
        - PCIT
        - Helping the Noncompliant Child
        - Parent Management Training Oregon
        - Triple P
    - Intensive parent training typically 12 sessions over 2 to 3 months, followed by additional reinforcement over a year
    - Includes the following components:
        - Understanding developmentally appropriate mood and behavior
        - Managing difficult temperaments
        - Social learning principles
        - Developing a warm supportive relationship with the child
        - Child-directed interaction and play
        - Predictable and structured home environment
        - Clear and simple house rules
        - Consistent praise and reward of positive behavior
        - Ignoring annoying behavior
        - Consistent consequences
    - *Parent Child Interaction Therapy*
        - 2 phases of training:
            - child-directed interaction = parents trained in nondirective play skills to alter quality of parent child interactions
            - parent-directed interaction = focuses on improving parenting skills by teaching parents to give clear instructions, praise for compliance, and time-out for non-compliance
    - Cochrane review found group parenting programs to be effective and cost effective
    - Adherence and completion of the program limits effectiveness, premature termination rates as high as 50-60%
    - There is evidence from RCTs that PMT strategies are effective, but may not bring children out of the clinically impaired range of functioning
    - Demonstrated to result in clinically significant improvement in ODD

- Anger Management Training:
    - Examples:
        - Coping Power

        - Problem solving skills training
    - Found to be effective for misbehaving youth
    - Typical 16-20 weekly sessions
    - Focus on emotional awareness, perspective taking, anger management, social problem solving, and goal setting
- Medication:
    - Stimulants and atypical antipsychotics have strong evidence for management of impulsive and anger driven aggression

7. Understand the range of prognoses for children with ODD

- Approximately two-thirds of children diagnosed with ODD no longer meet criteria at 3-year follow up
- One-third of patients with ODD will progress to CD
    - Approximately 30% higher likelihood of progression if comorbid ODD and ADHD
    - Defiant and vindictive symptoms of ODD carry highest risk of later development of CD
- 10% of children and adolescents diagnosed with ODD will eventually develop a more lasting personality disorder, such as antisocial personality disorder
- Irritable and anger symptoms of ODD carry highest risk for development of depression or anxiety

8. Recognize the environmental situations that contribute to the development and maintenance of antisocial, aggressive, and delinquent behaviors

- Temperamentally difficult children are likely to elicit negative parenting responses.
- Ineffective parenting:
    - Inconsistent rules
    - Unclear commands
    - Parent is more likely to respond to their child on the basis of their mood as opposed to the child's behavior
    - Parent is less likely to monitor the child's whereabouts
    - Parent is relatively unresponsive to the child's prosocial behavior
- Other family level influences:
    - Parent-child attachment
    - Child maltreatment
    - Exposure to marital conflict or domestic violence
    - Family poverty
    - Crime
    - Family history of behavior problems, bipolar disorder, substance use, schizophrenia, somatization and personality disorders.
- Peer level influences:
    - peer rejection
    - antisocial peer groups

9. Recognize the signs and symptoms of conduct disorder at different developmental stages

- Symptoms can begin as early as preschool
    - Typical begin as less severe symptoms (lying)
- Severe symptoms that emerge earlier convey a poorer prognosis
- Early onset: (before 8 years)
    - Lower IQ
    - More attentional challenges and impulsivity
    - Greater peer difficulties
    - More likely to have adverse family circumstances

- Late Onset: (adolescence)
    - Delinquent Behavior is more related to social influences
- Preschool years: irritable temperament; inattentiveness; poor maternal-child attachment; and aberrant behaviors
- Elementary school: quick, angry temperaments; poor social skills; tendency to "blame the victim" in cases of physical aggression
- Middle/high school: commonly break rules; overreact emotionally; fail to take responsibility for actions

10. Recognize the common co-morbid conditions of conduct disorder

- ADHD and ODD are common in children with CD
- CD may also cooccur with depression, anxiety, bipolar disorder, learning, language and substance related disorders.

11. Understand the range of prognoses for children with conduct disorder

- At risk for later development of mood, anxiety, post-traumatic stress, impulse control, psychotic, somatic and substance-related disorders.
- At risk for suicidal behavior, physical injury, delinquency, early pregnancy, social instability, marital failure and academic and occupational underachievement.
- 40% of children and adolescent with CD will go on to develop a personality disorder.
- 50% of children with early onset (before 8 years) persist into adulthood.
- 85% of children with late onset persist into adulthood.
- Factors predicting poor outcome:
    - Early onset
    - Hyperactivity and attention problems
    - Lower IQ
    - Family History of parental criminality and alcoholism
    - Parenting style: harsh, inconsistent parenting, low warmth, low involvement and low supervision
    - Socioeconomic: low income with poor schools
- Adult Outcomes:
    - Antisocial behavior: violence and criminal behavior
    - Psychiatric problems: increased rate of antisocial personality, substance abuse, anxiety, depression, episodes of self harm, attempted suicide, psychiatric hospitalization
    - Education: Increased truancy and higher rate of dropping out of school
    - Increased unemployment
    - Increased rate of short relationships, often partners with antisocial behaviors
    - Increased rates of child abuse
    - Earlier death

12. Know the pharmacologic treatments for conduct disorder

- There is no empiric support for psychopharmacologic treatment of oppositional behaviors in general.
- There has been suggestion that mood stabilizers, typical and atypical antipsychotics, clonidine, and stimulants may be useful; but few RCTs have been performed.
- Careful assessment for comorbid disorders (mood, ADHD) is indicated because behavioral problems may be accounted for or aggravated by the comorbid conditions.
    - ADHD? Stimulant
    - Depression? SSRI
    - Learning disability? IEP

- Substance use disorder? Treat accordingly.

13. Know the psychologic treatments for conduct disorder

- Best = multimodal Treatment
- Need long-term coordination among PCP, mental health practitioner, and family
- May include:
  - Parent Training (please see objective 7 for details)
  - Individual Psychotherapy to develop more effective anger management
  - May be most effective as part of a broader treatment program
  - Family Psychotherapy to improve communication and mutual understanding
  - Cognitive Problem-Solving Skills Training and Therapies to decrease negativity
  - Social Skills Training to increase flexibility and improve social skills and frustration tolerance with peers
  - Mentoring program
  - Academic skills training
  - Proactive classroom management and teacher training
  - Group therapy
  - Playground behavior program
- Multisystemic therapy = effective in reducing antisocial behavior and cost-effective
  - Proactive and flexible
  - Address risks at individual, family, per, school and neighborhood level
- Treatment is intensive and designed to address therapeutic barriers including parental substance abuse, psychopathology, and marital conflict; associations with delinquent peers; poor school performance; and deficient problem-solving or perspective taking skills
- Treatment providers participate in supervision and team case-review sessions to ensure treatment fidelity and overcome obstacles
- Role of Prevention:
  - Successful prevention programs typically used at least two modes of intervention, included a parent-directed component, and included social-cognitive skills training, academic skills training, proactive classroom management and teacher training and group therapy
  - Addressing risk factors:
    - Early parental rejection
    - Harsh, erratic discipline
    - Poor parental supervision
    - Separation from parents without alternative caregiver
    - Family neglect
    - Abuse
    - Domestic violence
    - Parental mental illness
    - Parental substance use
    - Poverty

14. Describe the characteristics of effective training programs for parents of children with ODD/conduct disorder

- Interventions addressing multiple needs from multiple domains tend to be more successful.
- Effective interventions are based on an understanding of the need to reverse the negative parent-child interactions that perpetuate oppositional behaviors.

- General approach:
    - Re-establish positive parent-child interactions through guided instruction and practice in positive, play-based interactions.
        - Goal = replace negative parental attention and reinforcement of undesirable behaviors with positive parental attention and reward for desirable behaviors
    - Teach limit setting and giving directions.
        - Goal = increase child's compliance with more appropriate, well-define set of rules
    - Teach effective discipline (implementing time out, avoiding power struggles, maintaining consistency in implementation).
        - Goal = replace coercive, ineffective discipline with a more effective non-coercive approach
    - Generalizing to other settings.

15. Understand the stages of development typically associated with oppositional behaviors

- Oppositional behaviors (defiance, passive noncompliance, self-assertion, avoidance) are often developmentally appropriate, and can be seen in all age groups.
- Oppositional behaviors typically emerge in toddlerhood when children begin to assert their independence and individuality.
- Manifestations change with the child's developmental stage:
    - Infant: push away, gesture refusal, dawdle; run away from or ignore parent
    - Toddler: says no, tantrums, does opposite of what is asked
    - School-age: procrastination, makes excuses, engages in arguments, negative attitudes
    - Adolescent: argues, demands reasons, breaks rules

16. Know the diagnostic criteria for ODD

- DSM-5: Disorder Class: Disruptive, Impulse-Control, and Conduct Disorders
    - A pattern of angry/irritable mood, argumentative/defiant behavior, or vindictiveness lasting at least 6 months as evidenced by at least four symptoms of the following categories, and exhibited during interaction with at least one individual who is not a sibling:
    - Angry/Irritable Mood
        - Often loses temper
        - Is often touchy or easily annoyed
        - Is often angry and resentful
    - Argumentative/Defiant Behavior
        - Often argues with authority figures or, for children and adolescents, with adults
        - Often actively defies or refuses to comply with requests from authority figures or with rules
        - Often deliberately annoys others
        - Often blames others for his or her mistakes or misbehavior
    - Vindictiveness
        - Has been spiteful or vindictive at least twice within the past 6 months.

Note: The persistence and frequency of these behaviors should be used to distinguish a behavior that is within normal limits from a behavior that is symptomatic. For children younger than 5 years, the behavior should occur on most days for a period of at least 6 months unless otherwise noted (Criterion AB). For individuals 5 years or older, the behavior should occur at least once per week for at least 6 months. Unless otherwise noted (Criterion AB). While these frequency criteria provide guidance on a minimal level of frequency to define symptoms, other factors should also be considered, such as whether the frequency and

intensity of the behaviors are outside a range that is normative for the individual's developmental level, gender, and culture.

17. Know how to evaluate a child for ODD/conduct disorder

- Careful history
    - Parents
    - Teachers/others sources
- Diagnostic interview
- Evaluating for deliberate, rule-breaking behaviors:
    - 4 general categories:
        - Aggression toward people and animals
        - Destruction of property
        - Lying and theft
        - Other serious violation of rules that can include truancy or substance use

18. Understand the benefits and limitations of different venues (eg, family, community, residential) for the treatment of conduct disorder

- Interventions should be implemented in the setting in which the behaviors occur.
- Family:
    - Parent psychopathology, expectations regarding treatment, and family stressors are predictive of retention in and success of treatment [11]
- Community:
    - Group interventions with peers: potentially problematic, as can reinforce deviant behavior and cause worsened problem behaviors [12]
- School-based intervention programs: mild positive outcomes at best, with little behavioral change; Data inconsistent, lacking in rigor
- Residential:
    - Treatment Foster care: only a modest positive change in general behavior problems
- Policy can effect change in reduction to exposures to toxins, reduction in community crime, and programs to reduce parental risk factors; can also organize services and management among agencies (mental heath, education, health, child welfare, juvenile justice) [13]

19. Know the diagnostic criteria for conduct disorder

- "A repetitive and persistent pattern of behavior in which the basic rights of others or major age-appropriate societal norms or rules are violated"
- Must have presence of **≥3** of the following 15 criteria in past 12 months, with at least one criterion in the past 6 months:
    - **Aggression to People and Animals:**
        1. Often bullies, threatens or intimidates others
        2. Often initiates physical fights
        3. Has used a weapon that can cause serious physical harm to others
        4. Has been physical cruel to people
        5. Has been physical cruel to animals
        6. Has stolen while confronting a victim
        7. Has forced someone into sexual activity
    - **Destruction of Property**
        8. Has deliberately engaged in fire setting with the intention of causing serious harm

9. Has deliberately destroyed others' property

- **Deceitfulness or Theft**
  10. Has broken into someone's house, building or car
  11. Often lies to obtain goods or favors or to avoid obligations
  12. Has stolen items of nontrivial value without confronting a victim (ie: shoplifting)
- **Serious Violations of Rules**
  13. Often stays out at night despite parental prohibitions, beginning before age 13 years
  14. Has run away from home overnight at least twice while living in parental or parental surrogate home, on once without returning for a lengthy period
  15. Is often truant from school, beginning before age 13 years

- Clinically significant impairment in academic, social or occupational functioning
- If ≥18 years of age, criteria are not met for antisocial personality disorder
- Based on age of onset:
  - CD, childhood-onset type = at least one CD criterion before 10 years of age
  - CD, adolescent-onset type = absence of any CD criterion before 10 years of age
  - CD, unspecified onset = unknown age of onset

20. Recognize family systems factors that contribute to the development and maintenance of ODD, intermittent explosive disorder, and conduct disorder

- Single parent or divorce
- Domestic violence
- Lack of permanent family
- Parental substance abuse or antisocial behavior
- Child maltreatment or neglect
- Parent-child conflict
- Excessive parental control
- Lack of parental supervision
- Maternal depression or anxiety

21. Know the diagnostic criteria for intermittent explosive disorder

- "Recurrent behavioral outburst representing a failure to control aggressive impulses as manifested by either of the following:"
  - **Verbal Aggression** (ie: temper tantrums, tirades, verbal arguments) **OR physical aggression** toward property, animals or other individuals, occurring **twice weekly**, on average, for a period of **3 months**. The physical aggression does not result in damage or destruction of property and does not result in physical injury to animals or other individuals.
  - **Three behavioral outbursts** involving damage or destruction of property and/or physical assault involving physical injury against animals or other individuals in a 12 month period.
- Magnitude of aggressiveness expressed during the recurrent outbursts is **grossly out of proportion** to the provocation or to any precipitating psychosocial stressors.
- Recurrent aggressive outbursts are **not premeditated** (ie: they are impulsive and/or anger based) and are not committed to achieve some tangible objective (ie: money, power)
- The recurrent aggressive outbursts cause either marked distress in the individual or impairment in occupational or interpersonal functioning, or as associated with financial or legal consequences.

- Chronological age is at least **6 years** (or equivalent developmental level)
- Recurrent aggressive outbursts are not better explained by another mental disorder and are not attributable to another medical condition or physiological effects of a substance. For children 6-18 years, aggressive behavior that occurs as part of an adjustment disorder should not be considered for this diagnosis.

22. Recognize the signs and symptoms of intermittent explosive disorder at different developmental stages

- Stability Over Time: Onset Age and Duration of Condition
  - Appears as early as prepubertal childhood
  - Rare to see before 6 years of age
  - Mean age of onset: 12 years of age
  - Peaks in mid-adolescence
  - Average Duration: 12 years to lifetime
- Explosive symptoms tend to improve slightly with age

23. Recognize the common co-morbid conditions of intermittent explosive disorder (IED)

- Depression
- Anxiety Disorders (Social Anxiety Disorder, Specific Phobia, Generalized Anxiety Disorder)
- Disruptive Behaviors (Oppositional Defiant Disorder, Conduct Disorder, ADHD)
- Alcohol/substance abuse
- Self-harm: suicide attempts and non-suicidal self-injury
- Bipolar Disorder
- Posttraumatic Stress Disorder
- Cluster B Personality Disorders
- Note: Comorbid diagnoses often present later than IED

24. Be able to differentiate intermittent explosive disorder from ODD and conduct disorders

- All three involve problems in behavioral and emotional regulation
- Conduct disorder criteria:
  - Focuses on poorly controlled behaviors
  - Predatory or premeditated aggression (ie: robbery, extortion, sexual abuse, physical cruelty
- Intermittent explosive disorder criteria:
  - Focuses on poorly controlled emotions
- ODD criteria:
  - More evenly distributed between behavioral and emotional symptoms
  - Impulsive aggression (ie: arguing, temper tantrums)

Disorder	Behavior	Emotions	Age of Onset
Conduct Disorder	• Predatory or premeditated aggression • Violate rights of others • Violate major societal norms		• Can emerge in preschool • Significant symptoms middle childhood to adolescence • Onset rare after 16

			years of age
Intermittent Explosive Disorder	• Impulsive aggression • Can include physical assault (not seen in ODD)	• Poorly controlled emotion --> anger outbursts that are disproportionate to the provocation or stressor	• At least 6 years of age • Late childhood or adolescence • Onset rare after 40 years of age
Oppositional Defiant Disorder	• Temper tantrums • Argumentative/Defiance with authority	• Emotional dysregulation • Angry or irritable mood	• Onset in preschool • Onset rare later than early adolescence

25. Know the psychological and pharmacologic treatments for intermittent explosive disorder

- General Treatment Principles:
  - Psychotherapy and/or pharmacotherapy
  - Goals:
    - Remission = resolution of symptoms or improvement to 1-2 mild symptoms
    - Or if remissions is not possible: Response = stabilize safety of patient and others decreased frequency and intensity of symptoms
- Psychotherapy: Cognitive Behavior Therapy (CBT)
  - Teaches patients how to manage aversive stimuli in their day to day environment to prevent aggressive impulses
  - CBT Techniques: Cognitive restructuring, relaxation training, coping skills training, relapse prevention
  - CBT best for highly motivated patients
  - Group or individual CBT
- Pharmacologic:
  - First Line: SSRI's
    - Fluoxetine (preferred; most studied)
    - Other SSRI's are reasonable
  - Resistant Patients (if no response to SSRI's):
    - Phenytoin
    - Oxcarbazepine/carbamazepine
  - Refractory Patients (if no response to above)
    - Lamotrigine
    - Topiramate
    - Valproate

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Content Category 13- Internalizing Behaviors & Conditions

ABP Content Specs for Developmental-Behavioral Pediatrics Fellow Collaboration Project

Prepared by Matthew Scott- Madigan Army Medical Center, DBP Fellow

Reviewed by Eric Flake- Madigan Army Medical Center, Staff DBP

13. Internalizing Behaviors and Conditions

A. Anxiety disorders

1. Understand the natural history of anxiety disorders
2. Understand the relationship between temperamental characteristics and anxiety
3. Recognize the environmental situations that contribute to or exacerbate anxious and fearful behaviors
4. Recognize the signs and symptoms of phobias and anxiety disorders and the range of common presentations
5. Know the initial management of phobias and anxiety disorders
6. Know the pharmacologic management of phobias and anxiety disorders
7. Know the psychological management of phobias and anxiety disorders
8. Recognize the common co-morbid conditions of anxiety disorders
9. Understand the typical prognoses and the range of prognoses for children with anxiety disorders
10. Know the diagnostic criteria for anxiety disorders
11. Differentiate worries from anxiety disorders
12. Recognize the signs and symptoms of selective mutism
13. Differentiate between selective mutism and other conditions affecting speech and language
14. Know how to plan the evaluation of a child with selective mutism
15. Know how to plan the management of a child with selective mutism
16. Differentiate selective mutism from normal variations in a child's comfort speaking in social settings
17. Understand the etiology of selective mutism

B. Trauma and stress-related disorders

1. Recognize the signs and symptoms of post-traumatic stress disorder in children and adolescents and the range of common presentations
2. Plan the treatment of a child or adolescent with post-traumatic stress disorder
3. Understand the etiology and environmental contributors to PTSD
4. Understand the role of medication in the management of acute stress disorder
5. Understand the role of medication in the management of PTSD
6. Understand the natural history of PTSD
7. Recognize the importance of active outreach and screening for PTSD after a traumatic event
8. Recognize that PTSD may have a delayed onset after a traumatic event
9. Plan the management of a child with acute stress disorder
10. Differentiate the prognosis and treatment for PTSD from that for an acute stress disorder
11. Know the diagnostic criteria, treatment and prognosis for a reactive attachment disorder
12. Know the diagnostic criteria, treatment and prognosis for an adjustment disorder

C. Depressive disorders

1. Understand the natural history of depressive disorders including disruptive mood dysregulation disorder, persistent depressive disorder (dysthymia), premenstrual dysphoria, and major depressive disorder
2. Understand the relationship between temperamental characteristics and depressive

disorders

3. Recognize the social and environmental contributors to the development of depressive symptoms
4. Recognize the signs and symptoms of dysthymia and the variations in presentation based on developmental stage
5. Recognize the signs and symptoms of major depression and the variations in presentation based on developmental stage
6. Recognize the common co-morbid conditions of depressive disorders
7. Understand the pharmacologic treatment of depressive disorders
8. Understand the psychological interventions for the treatment of depressive disorders
9. Understand the range of prognoses for children and adolescents with dysthymia or depressive disorders
10. Differentiate between normal grieving associated with a significant loss and major depressive disorder
11. Know the familial risk of major depressive disorder in children
12. Know the diagnostic criteria for depressive disorders
13. Recognize the family systems factors that contribute to depressive disorders
14. Understand the epidemiology of depression in children and adolescents (eg, gender-based differences, age-based differences, etc)

D. Bipolar and related disorders
1. Recognize the signs and symptoms of bipolar disorders
2. Understand the range of prognoses for children and adolescents with bipolar disorders
3. Know potential side effects of SSRI treatment for teens with bipolar disorders

E. Ritualistic/obsessive compulsive behavior
1. Know the natural history of OCD
2. Understand the impact of OCD symptoms on the child's functioning in his family, at school and with peers
3. Differentiate OCD from normal variations in obsessive or compulsive personality traits
4. Understand the range of prognoses for children with obsessive-compulsive disorder
5. Understand the etiologies of obsessive-compulsive disorder
6. Know the pharmacological treatments for obsessive-compulsive disorder
7. Know the diagnostic criteria for OCD
8. Recognize the signs and symptoms of OCD
9. Understand the genetics of OCD
10. Know the psychological treatments for OCD
11. Recognize the common co-morbid conditions that may be associated with OCD

F. Suicidal behavior
1. Know the risk factors associated with suicidal behavior in children and adolescents
2. Know the risk factors associated with a poor prognosis for children and adolescents who have attempted suicide
3. Know the steps in the prevention of suicidal behavior and management of an adolescent at risk of suicidal behavior
4. Know how to assess a child or adolescent with suicidal ideation
5. Know the indications for hospitalization of a child or adolescent at risk of suicide
6. Recognize the impact of suicide on peers and members of the family
7. Plan the management of a child with suicidal ideation or behavior

13. Internalizing Behaviors and Conditions

A. Anxiety disorders

1. Understand the natural history of anxiety disorders

Usually, anxiety disorders develop during childhood and persist in adulthood unless they are treated. Most are more common in females (2:1).

Separation anxiety disorder: children experience typical separation anxiety starting around 12 months of age (related to stranger danger). Separation anxiety disorder usually starts in preschool but can start at any time during childhood. The majority of children do not have impairing anxiety as an adult.

Specific Phobia: Sometimes develops after a traumatic event or observation of a traumatic event, but not always (almost everyone develops a phobia of choking after a near choking episode). Most cases start before age 10, will wax and wane, and often persist to adulthood.

Social Anxiety Disorder: "75% of individuals have an age at onset between 8 and 15 years." "The disorder sometimes emerges out of a childhood history of social inhibition or shyness... Onset can also occur in early childhood. Onset of social anxiety disorder may follow a stressful or humiliating experience (e.g., being bullied, vomiting during a public speech), or it may be insidious, developing slowly." (DSM 5)

Panic Disorder: The "median age at onset for panic disorder in the United States is 20–24 years. A small number of cases begin in childhood." (DSM 5)

Generalized Anxiety Disorder: A diagnosis of generalized anxiety disorder before adolescence is rare, but symptoms of excessive worry and an anxious temperament begin at a young age. In younger children, consider separation anxiety disorder, social anxiety disorder (social phobia), and obsessive-compulsive disorder before making a diagnosis of generalized anxiety disorder.

2. Understand the relationship between temperamental characteristics and anxiety

Symptoms of excessive worry and anxiety may occur early in life but are then manifested as an anxious temperament. Negative affectivity/neuroticism (i.e., proneness to experiencing negative emotions), anxiety sensitivity (i.e., the disposition to believe that symptoms of anxiety are harmful), behavioral inhibition, and fear of negative evaluation are temperaments associated with the multiple anxiety disorders. (DSM 5)

Behavioral inhibition is a temperament that is described as a "biologically based characteristic that presents as wariness, hesitance, and avoidance in the presence of novel situations, objects, places, and people." (Ryan et al) Children with higher behavioral inhibition levels are at increased risk for internalizing disorders (anxiety, depression, etc).

3. Recognize the environmental situations that contribute to or exacerbate anxious and fearful behaviors

From the DSM 5: Childhood maltreatment and adversity are risk factors for social anxiety disorder, generalized anxiety disorder, and phobias. Separation anxiety disorder often develops after a significant life stress (death or relative or pet, illness, change of school, divorce, move). Negative or traumatic encounters with the feared object or situation sometimes (but not always) precede the

development of specific phobia. Authoritarian and permissive parenting also increase the risk for internalizing disorders.

4. Recognize the signs and symptoms of phobias and anxiety disorders and the range of common presentations

Separation anxiety disorder: Excessive fear or anxiety concerning separation from home or attachment figures. Younger children may be reluctant to go to school or avoid school. At a young age they cannot express their fears and the anxiety is only manifested when the separation occurs.

Specific phobia: "First, young children may express their fear and anxiety by crying, tantrums, freezing, or clinging. Second, young children typically are not able to understand the concept of avoidance. Therefore, the clinician should assemble additional information from parents, teachers, or others who know the child well. Excessive fears are quite common in young children but are usually transitory and only mildly impairing and thus considered developmentally appropriate. In such cases a diagnosis of specific phobia would not be made." (DSM 5)

Social Anxiety Disorder: "The essential feature of social anxiety disorder is a marked, or intense, fear or anxiety of social situations in which the individual may be scrutinized by others. In children the fear or anxiety must occur in peer settings and not just during interactions with adults." "When exposed to such social situations, the individual fears that he or she will be negatively evaluated. The individual is concerned that he or she will be judged as anxious, weak, crazy, stupid, boring, intimidating, dirty, or unlikable. The individual fears that he or she will act or appear in a certain way or show anxiety symptoms, such as blushing, trembling, sweating, stumbling over one's words, or staring, that will be negatively evaluated by others (Criterion B). Some individuals fear offending others or being rejected as a result." "The social situations almost always provoke fear or anxiety (Criterion C). Thus, an individual who becomes anxious only occasionally in the social situation(s) would not be diagnosed with social anxiety disorder. However, the degree and type of fear and anxiety may vary (e.g., anticipatory anxiety, a panic attack) across different occasions." "In children, the fear or anxiety may be expressed by crying, tantrums, freezing, clinging, or shrinking in social situations." "Adolescents endorse a broader pattern of fear and avoidance, including of dating, compared with younger children." (DSM 5)

Generalized Anxiety Disorder: "Children and adolescents tend to worry more about school and sporting performance." "In children and adolescents with generalized anxiety disorder, the anxieties and worries often concern the quality of their performance or competence at school or in sporting events... There may be excessive concerns about punctuality. They may also worry about catastrophic events, such as earthquakes or nuclear war. Children with the disorder may be overly conforming, perfectionist, and unsure of themselves and tend to redo tasks because of excessive dissatisfaction with less-than-perfect performance. They are typically overzealous in seeking reassurance and approval and require excessive reassurance about their performance and other things they are worried about… Generalized anxiety disorder may be overdiagnosed in children. When this diagnosis is being considered in children, a thorough evaluation for the presence of other childhood anxiety disorders and other mental disorders should be done to determine whether the worries may be better explained by one of these disorders. Separation anxiety disorder, social anxiety disorder (social phobia), and obsessive-compulsive disorder are often accompanied by worries that may mimic those described in generalized anxiety disorder." (DSM 5)

5. Know the initial management of phobias and anxiety disorders

Advise parents about fears or anxieties in their child at different ages

- Anticipatory guidance and developmental surveillance targeted to life transitions (e.g. beginning school, entering high school, travel, divorce.)

Initial management

- Evaluate symptoms.
- Evaluate child's stresses and strengths.
- Consider strategies to reduce stress or provide more support.

Ongoing management

- Cognitive Behavior Therapy (CBT)
- Augment with psychopharmacological treatment

6. Know the pharmacologic management of phobias and anxiety disorders

SSRIs: supported by open label and controlled studies. Demonstrate efficacy and short-term safety for children and adolescents with anxiety disorders.

Tricyclic antidepressants: Multiple controlled studies. Poor side-effect profile

Benzodiazepines: Limited data on efficacy. Theoretical potential for abuse in adolescents

Starting at a very low dose of SSRI for the first week or two with anxiety disorders is especially essential to reduce the child's experience of side effects (augmented by associated somatic anxieties).

Name	Dosage Form	Usual starting dose for adolescents	Increase increment (after ~4 weeks)	RCT anxiety treatment benefit in kids	FDA anxiety approved for children?	Editorial Comments
Fluoxetine (Prozac)	10, 20, 40mg 20mg/5ml	5-10 mg/day (60mg max)*	10-20mg**	Yes	Yes (For OCD ≥7yr) (For MDD ≥8yr)	Long 1/2 life, no SE from a missed dose
Sertraline (Zoloft)	25, 50, 100mg 20mg/ml	25 mg/day (200mg max)*	25-50mg**	Yes	Yes (For OCD ≥6yr)	May be prone to SE from weaning off
Sertraline and Fluoxetine are both first line medications for child anxiety disorders, per the evidence base						
Fluvoxamine (Luvox)	25, 50, 100mg	25 mg/day (300mg max)*	50 mg**	Yes	Yes (For OCD ≥8yr)	Often more side effect than other SSRI's, has many drug interactions
Paroxetine (Paxil)	10, 20, 30, and 40 mg 10mg/5ml 12.5, 25, 37.5mg CR forms	5-10 mg/day (60mg max)*	10-20mg**	Yes	No	Not preferred if child also has depression. Can have short 1/2 life
Citalopram (Celexa)	10, 20, 40 mg 10mg/5ml	5-10 mg/day (40mg max)*	10-20mg**	Yes	No	Very few drug interactions
Escitalopram (Lexapro)	5, 10, 20mg 5mg/5ml	2.5 to 5 mg/day (20mg max)*	5-10mg**	No	No	Active isomer of citalopram
Duloxetine (Cymbalta)	20, 30, 40, 60mg	30 mg/day (120mg max)	30mg	Yes	Yes (For generalized anxiety ≥7yr)	

* Recommend decrease maximum dosage by at least 1/3 for pre-pubertal children

** Recommend using the lower dose increase increments for younger children.

Successful medication trials should continue for 6-12 months.

https://www.seattlechildrens.org/healthcare-professionals/access-services/partnership-access-line/resources/

7. Know the psychological management of phobias and anxiety disorders

Cognitive Behavioral Therapy (CBT): First line treatment for all anxiety disorders, including social anxiety disorder, separation anxiety disorder, and phobias

- efficacy supported by randomized clinical trials. (Kendall (1994).
- Treatment based on The Coping Cat workbook (Kendall 1990).
- Treatment is 18 weeks of 50- 60-minute sessions (individual) or 90 minutes (group treatment).
- Involves gradual exposure to fears
- Results: 73% of children with individual treatment and 50% of children with group CBT versus 8% of wait-list controls, no longer met criteria.
- Replicated with 55 children with OAD, 22 children with SAD, 17 children with avoidant disorder; 50% were free of disorder after Rx compared with 6% waitlist controls. Those who continued to have anxiety disorder had a significant decrease in severity. Treatment gains maintained at 1-year follow-up.

8. Recognize the common co-morbid conditions of anxiety disorders

Separation Anxiety Disorder: highly comorbid with generalized anxiety disorder and specific phobia. In adults, common comorbidities include specific phobia, PTSD, panic disorder, generalized anxiety disorder, social anxiety disorder, agoraphobia, obsessive-compulsive disorder, and personality disorders.

Selective Mutism: other anxiety disorders (usually social anxiety disorder), oppositional behaviors (often in situations requiring speech), communication delays.

Specific Phobia: Often, specific phobia have an earlier onset and patients develop anxiety disorders, depression, bioplar, substance use, dependent personality disorder.

Social Anxiety Disorder: "other anxiety disorders, major depressive disorder, and substance use disorders, and the onset of social anxiety disorder generally precedes that of the other disorders, except for specific phobia and separation anxiety disorder. Chronic social isolation in the course of a social anxiety disorder may result in major depressive disorder. Substances may be used as self-medication for social fears, but the symptoms of substance intoxication or withdrawal, such as trembling, may also be a source of (further) social fear. Social anxiety disorder is frequently comorbid with bipolar disorder or body dysmorphic disorder; for example, an individual has body dysmorphic disorder concerning a preoccupation with a slight irregularity of her nose, as well as social anxiety disorder because of a severe fear of sounding unintelligent. The more generalized form of social anxiety disorder, but not social anxiety disorder, performance only, is often comorbid with avoidant personality disorder. **In children, comorbidities with high-functioning autism and selective mutism are common.**" (DSM 5)

Panic Disorder: Patients almost always present with other psychopathology, mainly other anxiety disorders, depression, bipolar. Patients often have other general medical symptoms: "dizziness, cardiac arrhythmias, hyperthyroidism, asthma, COPD, and irritable bowel syndrome." (DSM 5) Thyroid disease and mitral valve prolapse have been associated with panic disorder. Caffeine can make symptoms worse.

Generalized Anxiety Disorder: Most common comorbidities include other anxiety disorders and depression. Less common comorbidities include "substance use, conduct, psychotic, neurodevelopmental, and neurocognitive disorders." (DSM 5)

9. Understand the typical prognoses and the range of prognoses for children with anxiety disorders

Last et al (1996) evaluated children with anxiety disorders every 12 months for four years with favorable prognosis:

- 82 % remitted
- Rates of recovery were: 96% SAD, 80% GAD
- 1/3 of participants developed new psychiatric disorders.
- Prospective study (Pine et al 1998) Children with SAD were not more likely to develop panic disorder.

10. Know the diagnostic criteria for anxiety disorders

Separation Anxiety Disorder (F93.0)

A. Developmentally inappropriate and excessive fear or anxiety concerning separation from those to whom the individual is attached, as evidenced by **at least three of the following**:
    1. Recurrent excessive distress when anticipating or experiencing separation from home or from major attachment figures.
    2. Persistent and excessive worry about losing major attachment figures or about possible harm to them, such as illness, injury, disasters, or death.
    3. Persistent and excessive worry about experiencing an untoward event (e.g., getting lost, being kidnapped, having an accident, becoming ill) that causes separation from a major attachment figure.
    4. Persistent reluctance or refusal to go out, away from home, to school, to work, or elsewhere because of fear of separation.
    5. Persistent and excessive fear of or reluctance about being alone or without major attachment figures at home or in other settings.
    6. Persistent reluctance or refusal to sleep away from home or to go to sleep without being near a major attachment figure.
    7. Repeated nightmares involving the theme of separation.
    8. Repeated complaints of physical symptoms (e.g., headaches, stomachaches, nausea, vomiting) when separation from major attachment figures occurs or is anticipated.
B. The fear, anxiety, or avoidance is persistent, lasting at least 4 weeks in children and adolescents and typically 6 months or more in adults.
C. The disturbance causes clinically significant distress or impairment in social, academic, occupational, or other important areas of functioning.
D. The disturbance is not better explained by another mental disorder, such as refusing to leave home because of excessive resistance to change in autism spectrum disorder; delusions or hallucinations concerning separation in psychotic disorders; refusal to go outside without a trusted companion in agoraphobia; worries about ill health or other harm befalling significant others in generalized anxiety disorder; or concerns about having an illness in illness anxiety disorder.

Specific Phobia

A. Marked fear or anxiety about a specific object or situation (e.g., flying, heights, animals, receiving an injection, seeing blood).
    - **Note:** In children, the fear or anxiety may be expressed by crying, tantrums, freezing, or clinging.
B. The phobic object or situation almost always provokes immediate fear or anxiety.
C. The phobic object or situation is actively avoided or endured with intense fear or anxiety.
D. The fear or anxiety is out of proportion to the actual danger posed by the specific object or situation and to the sociocultural context.
E. The fear, anxiety, or avoidance is persistent, typically lasting for 6 months or more.
F. The fear, anxiety, or avoidance causes clinically significant distress or impairment in social, occupational, or other important areas of functioning.
G. The disturbance is not better explained by the symptoms of another mental disorder, including fear, anxiety, and avoidance of situations associated with panic-like symptoms or other incapacitating symptoms (as in agoraphobia); objects or situations related to obsessions (as in obsessive-compulsive disorder); reminders of traumatic events (as in posttraumatic stress disorder); separation from home or attachment figures (as in separation anxiety disorder); or social situations (as in social anxiety disorder).

Specify if:

- Code based on the phobic stimulus:
- **300.29 (F40.218) Animal** (e.g., spiders, insects, dogs).
- **300.29 (F40.228) Natural environment** (e.g., heights, storms, water).
- **300.29 (F40.23x) Blood-injection-injury** (e.g., needles, invasive medical procedures).
    - **Coding note:** Select specific ICD-10-CM code as follows: **F40.230** fear of blood; **F40.231** fear of injections and transfusions; **F40.232** fear of other medical care; or **F40.233** fear of injury.
- **300.29 (F40.248) Situational** (e.g., airplanes, elevators, enclosed places).
- **300.29 (F40.298) Other** (e.g., situations that may lead to choking or vomiting; in children, e.g., loud sounds or costumed characters).

Social Anxiety Disorder (F40.10)

A. Marked fear or anxiety about one or more social situations in which the individual is exposed to possible scrutiny by others. Examples include social interactions (e.g., having a conversation, meeting unfamiliar people), being observed (e.g., eating or drinking), and performing in front of others (e.g., giving a speech).

Note: In children, the anxiety must occur in peer settings and not just during interactions with adults.

B. The individual fears that he or she will act in a way or show anxiety symptoms that will be negatively evaluated (i.e., will be humiliating or embarrassing; will lead to rejection or offend others).
C. The social situations almost always provoke fear or anxiety.
    - **Note:** In children, the fear or anxiety may be expressed by crying, tantrums, freezing, clinging, shrinking, or failing to speak in social situations.
D. The social situations are avoided or endured with intense fear or anxiety.
E. The fear or anxiety is out of proportion to the actual threat posed by the social situation and to the sociocultural context.
F. The fear, anxiety, or avoidance is persistent, typically lasting for 6 months or more.
G. The fear, anxiety, or avoidance causes clinically significant distress or impairment in social, occupational, or other important areas of functioning.
H. The fear, anxiety, or avoidance is not attributable to the physiological effects of a substance (e.g., a drug of abuse, a medication) or another medical condition.
I. The fear, anxiety, or avoidance is not better explained by the symptoms of another mental disorder, such as panic disorder, body dysmorphic disorder, or autism spectrum disorder.
J. If another medical condition (e.g., Parkinson's disease, obesity, disfigurement from burns or injury) is present, the fear, anxiety, or avoidance is clearly unrelated or is excessive.

Specify if:

- **Performance only:** If the fear is restricted to speaking or performing in public.

Panic Disorder (F41.0)

A. Recurrent unexpected panic attacks. A panic attack is an abrupt surge of intense fear or intense discomfort that reaches a peak within minutes, and during which time four (or more) of the following symptoms occur:

Note: The abrupt surge can occur from a calm state or an anxious state.

1. Palpitations, pounding heart, or accelerated heart rate.
2. Sweating.
3. Trembling or shaking.
4. Sensations of shortness of breath or smothering.
5. Feelings of choking.
6. Chest pain or discomfort.
7. Nausea or abdominal distress.
8. Feeling dizzy, unsteady, light-headed, or faint.
9. Chills or heat sensations.
10. Paresthesias (numbness or tingling sensations).
11. Derealization (feelings of unreality) or depersonalization (being detached from oneself).
12. Fear of losing control or "going crazy."
13. Fear of dying.

- **Note:** Culture-specific symptoms (e.g., tinnitus, neck soreness, headache, uncontrollable screaming or crying) may be seen. Such symptoms should not count as one of the four required symptoms.

B. At least one of the attacks has been followed by 1 month (or more) of one or both of the following:

1. Persistent concern or worry about additional panic attacks or their consequences (e.g., losing control, having a heart attack, "going crazy").
2. A significant maladaptive change in behavior related to the attacks (e.g., behaviors designed to avoid having panic attacks, such as avoidance of exercise or unfamiliar situations).

B. The disturbance is not attributable to the physiological effects of a substance (e.g., a drug of abuse, a medication) or another medical condition (e.g., hyperthyroidism, cardiopulmonary disorders).

C. The disturbance is not better explained by another mental disorder (e.g., the panic attacks do not occur only in response to feared social situations, as in social anxiety disorder; in response to circumscribed phobic objects or situations, as in specific phobia; in response to obsessions, as in obsessive-compulsive disorder; in response to reminders of traumatic events, as in posttraumatic stress disorder; or in response to separation from attachment figures, as in separation anxiety disorder).

Panic Attack Definition

Note: Symptoms are presented for the purpose of identifying a panic attack; however, panic attack is not a mental disorder and cannot be coded. Panic attacks can occur in the context of any anxiety disorder as well as other mental disorders (e.g., depressive disorders, posttraumatic stress disorder, substance use disorders) and some medical conditions (e.g., cardiac, respiratory, vestibular, gastrointestinal). When the presence of a panic attack is identified, it should be noted as a specifier (e.g., "posttraumatic stress disorder with panic

attacks"). For panic disorder, the presence of panic attack is contained within the criteria for the disorder and panic attack is not used as a specifier.

An abrupt surge of intense fear or intense discomfort that reaches a peak within minutes, and during which time four (or more) of the following symptoms occur:

Note: The abrupt surge can occur from a calm state or an anxious state.

1. Palpitations, pounding heart, or accelerated heart rate.
2. Sweating.
3. Trembling or shaking.
4. Sensations of shortness of breath or smothering.
5. Feelings of choking.
6. Chest pain or discomfort.
7. Nausea or abdominal distress.
8. Feeling dizzy, unsteady, light-headed, or faint.
9. Chills or heat sensations.
10. Paresthesias (numbness or tingling sensations).
11. Derealization (feelings of unreality) or depersonalization (being detached from oneself).
12. Fear of losing control or "going crazy."
13. Fear of dying.

Note: Culture-specific symptoms (e.g., tinnitus, neck soreness, headache, uncontrollable screaming or crying) may be seen. Such symptoms should not count as one of the four required symptoms.

Agoraphobia (F40.00)

A. Marked fear or anxiety about two (or more) of the following five situations:
    1. Using public transportation (e.g., automobiles, buses, trains, ships, planes).
    2. Being in open spaces (e.g., parking lots, marketplaces, bridges).
    3. Being in enclosed places (e.g., shops, theaters, cinemas).
    4. Standing in line or being in a crowd.
    5. Being outside of the home alone.
B. The individual fears or avoids these situations because of thoughts that escape might be difficult or help might not be available in the event of developing panic-like symptoms or other incapacitating or embarrassing symptoms (e.g., fear of falling in the elderly; fear of incontinence).
C. The agoraphobic situations almost always provoke fear or anxiety.
D. The agoraphobic situations are actively avoided, require the presence of a companion, or are endured with intense fear or anxiety.
E. The fear or anxiety is out of proportion to the actual danger posed by the agoraphobic situations and to the sociocultural context.
F. The fear, anxiety, or avoidance is persistent, typically lasting for 6 months or more.
G. The fear, anxiety, or avoidance causes clinically significant distress or impairment in social, occupational, or other important areas of functioning.
H. If another medical condition (e.g., inflammatory bowel disease, Parkinson's disease) is present, the fear, anxiety, or avoidance is clearly excessive.
I. The fear, anxiety, or avoidance is not better explained by the symptoms of another mental disorder—for example, the symptoms are not confined to specific phobia, situational type; do not involve only social situations (as in social anxiety disorder); and are not related exclusively to obsessions (as in obsessive-compulsive disorder), perceived defects or flaws in physical appearance (as in body dysmorphic disorder), reminders of traumatic events (as in posttraumatic stress disorder), or fear of separation (as in separation anxiety disorder).

Note: Agoraphobia is diagnosed irrespective of the presence of panic disorder. If an individual's presentation meets criteria for panic disorder and agoraphobia, both diagnoses should be assigned.

Generalized Anxiety Disorder (F41.1)

A. Excessive anxiety and worry (apprehensive expectation), occurring more days than not for at least 6 months, about a number of events or activities (such as work or school performance).
B. The individual finds it difficult to control the worry.
C. The anxiety and worry are associated with three (or more) of the following six symptoms (with at least some symptoms having been present for more days than not for the past 6 months):
    - **Note:** Only one item is required in children.
    1. Restlessness or feeling keyed up or on edge.
    2. Being easily fatigued.
    3. Difficulty concentrating or mind going blank.
    4. Irritability.
    5. Muscle tension.
    6. Sleep disturbance (difficulty falling or staying asleep, or restless, unsatisfying sleep).
B. The anxiety, worry, or physical symptoms cause clinically significant distress or impairment in social, occupational, or other important areas of functioning.
C. The disturbance is not attributable to the physiological effects of a substance (e.g., a drug of abuse, a medication) or another medical condition (e.g., hyperthyroidism).
D. The disturbance is not better explained by another mental disorder (e.g., anxiety or worry about having panic attacks in panic disorder, negative evaluation in social anxiety disorder [social phobia], contamination or other obsessions in obsessive-compulsive disorder, separation from attachment figures in separation anxiety disorder, reminders of traumatic events in posttraumatic stress disorder, gaining weight in anorexia nervosa, physical complaints in somatic symptom disorder, perceived appearance flaws in body dysmorphic disorder, having a serious illness in illness anxiety disorder, or the content of delusional beliefs in schizophrenia or delusional disorder).

American Psychiatric Association. Diagnostic and Statistical Manual of Mental Disorders. 5th ed. Washington, DC, USA: APA; 2013

11. Differentiate worries from anxiety disorders

"The essential feature of generalized anxiety disorder is **excessive** anxiety and worry (apprehensive expectation) about a number of events or activities. The intensity, duration, or frequency of the anxiety and worry is out of proportion to the actual likelihood or impact of the anticipated event. The individual finds it difficult to control the worry and to keep worrisome thoughts from interfering with attention to tasks at hand. Adults with generalized anxiety disorder often worry about everyday, routine life circumstances, such as possible job responsibilities, health and finances, the health of family members, misfortune to their children, or minor matters (e.g., doing household chores or being late for appointments). Children with generalized anxiety disorder tend to worry excessively about their competence or the quality of their performance. During the course of the disorder, the focus of worry may shift from one concern to another.

Several features distinguish generalized anxiety disorder from nonpathological anxiety. First, the worries associated with generalized anxiety disorder are **excessive and typically interfere significantly with psychosocial functioning**, whereas the worries of everyday life are not excessive and are perceived as more manageable and may be put off when more pressing matters arise. Second, the worries associated with generalized anxiety disorder are **more pervasive, pronounced, and distressing; have longer duration; and frequently occur without precipitants**. The greater the range of life circumstances about which a person worries (e.g., finances, children's safety, job performance), the more likely his or her symptoms are to meet criteria for generalized anxiety disorder. Third, everyday worries are much less likely to be **accompanied by physical symptoms (e.g., restlessness or feeling keyed up or on edge)**. Individuals with generalized anxiety disorder report subjective distress due to constant worry and related impairment in social, occupational, or other important areas of functioning."

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12. Recognize the signs and symptoms of selective mutism

Selective mutism is defined as "consistent failure to speak in social situations in which there is an expectation to speak (e.g., school) even though the individual speaks in other situations." (DSM 5) Selective mutism significantly impacts the ability of a child to succeed in school and work. They are often able to communicate easily and typically with their families or close friends. However, they become unable to communicate in certain situations. It is caused by extreme anxiety and not usually by opposition or a refusal to speak. The most common comorbid conditions are social anxiety disorder, separation anxiety disorder, and specific phobia. Other communication delays or disorders may also be diagnosed in children with selective mutism.

DSM Diagnostic Criteria for Selective Mutism (F94.0):

A. Consistent failure to speak in specific social situations in which there is an expectation for speaking (e.g., at school) despite speaking in other situations.
B. The disturbance interferes with educational or occupational achievement or with social communication.
C. The duration of the disturbance is at least 1 month (not limited to the first month of school).
D. The failure to speak is not attributable to a lack of knowledge of, or comfort with, the spoken language required in the social situation.
E. The disturbance is not better explained by a communication disorder (e.g., childhood-onset fluency disorder) and does not occur exclusively during the course of autism spectrum disorder, schizophrenia, or another psychotic disorder.

13. Differentiate between selective mutism and other conditions affecting speech and language

Unlike selective mutism, the speech disturbance in other conditions is not restricted to a specific social situation. The differential diagnosis includes language disorders, speech sound disorder, stuttering, and social communication disorder. However, these speech disturbances do not disappear in certain settings, like when talking to parents at home. Selective mutism should be diagnosed only when a child has an established capacity to speak in some social situations (e.g., typically at home). For example, children with autism or intellectual disability also have difficulty speaking in social situations, but the difficulties don't disappear in other social situations.

14. Know how to plan the evaluation of a child with selective mutism

The general development of the child should be evaluated. The key to diagnosis is the child speaks in some situation, usually at home with family. The evaluation should include either direct observation of the child alone with the family or a recording of the child in a comfortable setting where he or she speaks normally. Other communication disorders should be evaluated for because they are often associated. The child should be screened for other anxiety disorders.

15. Know how to plan the management of a child with selective mutism

Treatment usually involves an SSRI to target associated anxiety symptoms (many are also diagnosed with anxiety or social anxiety disorder/social phobia) and behavioral therapy. Cognitive Behavioral Therapy has shown the most benefit. Other therapies have been developed specifically for selective mutism. Most use a gradual increase in a requirement for verbal communication, such as speaking to the therapist from a distance with a parent present, and gradually having the parent move farther away and then leaving.

16. Differentiate selective mutism from normal variations in a child's comfort speaking in social settings

Shyness is often mistaken for selective mutism. One of the big differences is that selective mutism "interferes with educational or occupational achievement or with social communication." Children who are shy will typically warm up over time (which is why symptoms should last more than 1 month at school to be diagnosed with selective mutism). Children who are shy usually do not like speaking in class or other situations, but they are able to when required. Children with selective mutism do not just outgrow their symptoms. The earlier intervention is started, the better the outcomes.

17. Understand the etiology of selective mutism

- Genetic: There is thought to be a genetic component, so look for a family history of anxiety, social phobia, or selective mutism.
- Temperament: Behavioral Inhibition is when children show fear and avoidance in unfamiliar situations. This temperament style is often linked with selective mutism.
- There is a higher rate of diagnosis of selective mutism in immigrant children who are learning a second language. That is why the D criteria is so important. The lack of speech should not attributable to a lack of knowledge of, or comfort with, the spoken language required in the social situation.

References for Selective Mutism Sections:
American Psychiatric Association. Diagnostic and Statistical Manual of Mental Disorders. 5th ed. Washington, DC, USA: APA; 2013
Black, D., & Andreasen, N. (2014). Introductory Textbook of Psychiatry (6th ed.)
Hua A, Major N. Selective mutism. Curr Opin Pediatr. 2016;28(1):114–120.

B. Trauma and stress-related disorders

1. Recognize the signs and symptoms of post-traumatic stress disorder in children and adolescents and the range of common presentations

The essential feature of posttraumatic stress disorder (PTSD) is the development of characteristic symptoms following exposure to one or more traumatic events. The clinical presentation of PTSD varies. In some individuals, fear-based re-experiencing, emotional, and behavioral symptoms may predominate. In others, anhedonic or dysphoric mood states and negative cognitions may be most distressing. In some other individuals, arousal and reactive-externalizing symptoms are prominent, while in others, dissociative symptoms predominate. Finally, some individuals exhibit combinations of these symptom patterns.

Presentations vary by development: Young children may report new onset of frightening dreams without content specific to the traumatic event. Before age 6 years, young children are more likely to express reexperiencing symptoms through play that refers directly or symbolically to the trauma. They may not manifest fearful reactions at the time of the exposure or during reexperiencing. Parents may report a wide range of emotional or behavioral changes in young children. Children may focus on imagined interventions in their play or storytelling. In addition to avoidance, children may become preoccupied with reminders. Because of young children's limitations in expressing thoughts or labeling emotions, negative alterations in mood or cognition tend to involve primarily mood changes. Children may experience co-occurring traumas (e.g., physical abuse, witnessing domestic violence) and in chronic circumstances may not be able to identify onset of symptomatology. Avoidant behavior may be associated with restricted play or exploratory behavior in young children; reduced participation in new activities in school-age children; or reluctance to pursue developmental opportunities in adolescents (e.g., dating, driving). Older children and adolescents may judge themselves as cowardly. Adolescents may harbor beliefs of being changed in ways that make them socially undesirable and estrange them from peers (e.g., "Now I'll never fit in") and lose aspirations for the future. Irritable or aggressive behavior in children and adolescents can interfere with peer relationships and school behavior. Reckless behavior may lead to accidental injury to self or others, thrill-seeking, or high-risk behaviors. Individuals who continue to experience PTSD into older adulthood may express fewer symptoms of hyperarousal, avoidance, and negative cognitions and mood compared with younger adults with PTSD, although adults exposed to traumatic events during later life may display more avoidance, hyperarousal, sleep problems, and crying spells than do younger adults exposed to the same traumatic events. In older individuals, the disorder is associated with negative health perceptions, primary care utilization, and suicidal ideation.

Posttraumatic Stress Disorder DSM 5 Diagnostic Criteria (F43.10)

Note: The following criteria apply to adults, adolescents, and **children older than 6 years**.

A. Exposure to actual or threatened death, serious injury, or sexual violence in one (or more) of the following ways:
    1. Directly experiencing the traumatic event(s).
    2. Witnessing, in person, the event(s) as it occurred to others.
    3. Learning that the traumatic event(s) occurred to a close family member or close friend. In cases of actual or threatened death of a family member or friend, the event(s) must have been violent or accidental.

4. Experiencing repeated or extreme exposure to aversive details of the traumatic event(s) (e.g., first responders collecting human remains; police officers repeatedly exposed to details of child abuse).
    - **Note:** Criterion A4 does not apply to exposure through electronic media, television, movies, or pictures, unless this exposure is work related.

B. Presence of one (or more) of the following intrusion symptoms associated with the traumatic event(s), beginning after the traumatic event(s) occurred:
    1. Recurrent, involuntary, and intrusive distressing memories of the traumatic event(s).
        - **Note:** In children older than 6 years, repetitive play may occur in which themes or aspects of the traumatic event(s) are expressed.
    2. Recurrent distressing dreams in which the content and/or affect of the dream are related to the traumatic event(s).
        - **Note:** In children, there may be frightening dreams without recognizable content.
    3. Dissociative reactions (e.g., flashbacks) in which the individual feels or acts as if the traumatic event(s) were recurring. (Such reactions may occur on a continuum, with the most extreme expression being a complete loss of awareness of present surroundings.)
        - **Note:** In children, trauma-specific reenactment may occur in play.
    4. Intense or prolonged psychological distress at exposure to internal or external cues that symbolize or resemble an aspect of the traumatic event(s).
    5. Marked physiological reactions to internal or external cues that symbolize or resemble an aspect of the traumatic event(s).

B. Persistent avoidance of stimuli associated with the traumatic event(s), beginning after the traumatic event(s) occurred, as evidenced by one or both of the following:
    1. Avoidance of or efforts to avoid distressing memories, thoughts, or feelings about or closely associated with the traumatic event(s).
    2. Avoidance of or efforts to avoid external reminders (people, places, conversations, activities, objects, situations) that arouse distressing memories, thoughts, or feelings about or closely associated with the traumatic event(s).

B. Negative alterations in cognitions and mood associated with the traumatic event(s), beginning or worsening after the traumatic event(s) occurred, as evidenced by two (or more) of the following:
    1. Inability to remember an important aspect of the traumatic event(s) (typically due to dissociative amnesia and not to other factors such as head injury, alcohol, or drugs).
    2. Persistent and exaggerated negative beliefs or expectations about oneself, others, or the world (e.g., “I am bad,” “No one can be trusted,” “The world is completely dangerous,” “My whole nervous system is permanently ruined”).
    3. Persistent, distorted cognitions about the cause or consequences of the traumatic event(s) that lead the individual to blame himself/herself or others.
    4. Persistent negative emotional state (e.g., fear, horror, anger, guilt, or shame).
    5. Markedly diminished interest or participation in significant activities.
    6. Feelings of detachment or estrangement from others.
    7. Persistent inability to experience positive emotions (e.g., inability to experience happiness, satisfaction, or loving feelings).

B. Marked alterations in arousal and reactivity associated with the traumatic event(s), beginning or worsening after the traumatic event(s) occurred, as evidenced by two (or more) of the following:
    1. Irritable behavior and angry outbursts (with little or no provocation) typically expressed as verbal or physical aggression toward people or objects.
    2. Reckless or self-destructive behavior.
    3. Hypervigilance.
    4. Exaggerated startle response.
    5. Problems with concentration.
    6. Sleep disturbance (e.g., difficulty falling or staying asleep or restless sleep).

B. Duration of the disturbance (Criteria B, C, D, and E) is more than 1 month.

C. The disturbance causes clinically significant distress or impairment in social, occupational, or other important areas of functioning.

D. The disturbance is not attributable to the physiological effects of a substance (e.g., medication, alcohol) or another medical condition.

Specify whether:

- **With dissociative symptoms:** The individual's symptoms meet the criteria for posttraumatic stress disorder, and in addition, in response to the stressor, the individual experiences persistent or recurrent symptoms of either of the following:
    1. **Depersonalization:** Persistent or recurrent experiences of feeling detached from, and as if one were an outside observer of, one's mental processes or body (e.g., feeling as though one were in a dream; feeling a sense of unreality of self or body or of time moving slowly).
    2. **Derealization:** Persistent or recurrent experiences of unreality of surroundings (e.g., the world around the individual is experienced as unreal, dreamlike, distant, or distorted).
    - **Note:** To use this subtype, the dissociative symptoms must not be attributable to the physiological effects of a substance (e.g., blackouts, behavior during alcohol intoxication) or another medical condition (e.g., complex partial seizures).

Specify if:

- **With delayed expression:** If the full diagnostic criteria are not met until at least 6 months after the event (although the onset and expression of some symptoms may be immediate).

Posttraumatic Stress Disorder for Children 6 Years and Younger Diagnostic Criteria

A. In children 6 years and younger, exposure to actual or threatened death, serious injury, or sexual violence in one (or more) of the following ways:
    1. Directly experiencing the traumatic event(s).
    2. Witnessing, in person, the event(s) as it occurred to others, especially caregivers.
        - **Note:** Witnessing does not include events that are witnessed only in electronic media, television, movies, or pictures.
    3. Learning that the traumatic event(s) occurred to a parent or caregiving figure.

B. Presence of one (or more) of the following intrusion symptoms associated with the traumatic event(s), beginning after the traumatic event(s) occurred:
    1. Recurrent, involuntary, and intrusive distressing memories of the traumatic event(s).

        - **Note:** Spontaneous and intrusive memories may not necessarily appear distressing and may be expressed as play reenactment.
    2. Recurrent distressing dreams in which the content and/or affect of the dream are related to the traumatic event(s).
        - **Note:** It may not be possible to ascertain that the frightening content is related to the traumatic event.
    3. Dissociative reactions (e.g., flashbacks) in which the child feels or acts as if the traumatic event(s) were recurring. (Such reactions may occur on a continuum, with the most extreme expression being a complete loss of awareness of present surroundings.) Such trauma-specific reenactment may occur in play.
    4. Intense or prolonged psychological distress at exposure to internal or external cues that symbolize or resemble an aspect of the traumatic event(s).
    5. Marked physiological reactions to reminders of the traumatic event(s).

C. One (or more) of the following symptoms, representing either persistent avoidance of stimuli associated with the traumatic event(s) or negative alterations in cognitions and mood associated with the traumatic event(s), must be present, beginning after the event(s) or worsening after the event(s):

- Persistent Avoidance of Stimuli
    1. Avoidance of or efforts to avoid activities, places, or physical reminders that arouse recollections of the traumatic event(s).
    2. Avoidance of or efforts to avoid people, conversations, or interpersonal situations that arouse recollections of the traumatic event(s).
- Negative Alterations in Cognitions
    3. Substantially increased frequency of negative emotional states (e.g., fear, guilt, sadness, shame, confusion).
    4. Markedly diminished interest or participation in significant activities, including constriction of play.
    5. Socially withdrawn behavior.
    6. Persistent reduction in expression of positive emotions.

D. Alterations in arousal and reactivity associated with the traumatic event(s), beginning or worsening after the traumatic event(s) occurred, as evidenced by two (or more) of the following:
    1. Irritable behavior and angry outbursts (with little or no provocation) typically expressed as verbal or physical aggression toward people or objects (including extreme temper tantrums).
    2. Hypervigilance.
    3. Exaggerated startle response.
    4. Problems with concentration.
    5. Sleep disturbance (e.g., difficulty falling or staying asleep or restless sleep).

E. The duration of the disturbance is more than 1 month.

F. The disturbance causes clinically significant distress or impairment in relationships with parents, siblings, peers, or other caregivers or with school behavior.

G. The disturbance is not attributable to the physiological effects of a substance (e.g., medication or alcohol) or another medical condition.

Specify whether:

- **With dissociative symptoms:** The individual's symptoms meet the criteria for posttraumatic stress disorder, and the individual experiences persistent or recurrent symptoms of either of the following:
    1. **Depersonalization:** Persistent or recurrent experiences of feeling detached from, and as if one were an outside observer of, one's mental processes or body (e.g., feeling as though one were in a dream; feeling a sense of unreality of self or body or of time moving slowly).
    2. **Derealization:** Persistent or recurrent experiences of unreality of surroundings (e.g., the world around the individual is experienced as unreal, dreamlike, distant, or distorted).
    - **Note:** To use this subtype, the dissociative symptoms must not be attributable to the physiological effects of a substance (e.g., blackouts) or another medical condition (e.g., complex partial seizures).

Specify if:

- **With delayed expression:** If the full diagnostic criteria are not met until at least 6 months after the event (although the onset and expression of some symptoms may be immediate).

Acute stress disorder Diagnostic Criteria

A. Exposure to actual or threatened death, serious injury, or sexual violence in one (or more) of the following ways:
    1. Directly experiencing the traumatic event(s).
    2. Witnessing, in person, the event(s) as it occurred to others.
    3. Learning that the event(s) occurred to a close family member or close friend. **Note:** In cases of actual or threatened death of a family member or friend, the event(s) must have been violent or accidental.
    4. Experiencing repeated or extreme exposure to aversive details of the traumatic event(s) (e.g., first responders collecting human remains, police officers repeatedly exposed to details of child abuse).
        - **Note:** This does not apply to exposure through electronic media, television, movies, or pictures, unless this exposure is work related.

B. Presence of nine (or more) of the following symptoms from any of the five categories of intrusion, negative mood, dissociation, avoidance, and arousal, beginning or worsening after the traumatic event(s) occurred:

Intrusion Symptoms

1. Recurrent, involuntary, and intrusive distressing memories of the traumatic event(s). **Note:** In children, repetitive play may occur in which themes or aspects of the traumatic event(s) are expressed.
2. Recurrent distressing dreams in which the content and/or affect of the dream are related to the event(s). **Note:** In children, there may be frightening dreams without recognizable content.
3. Dissociative reactions (e.g., flashbacks) in which the individual feels or acts as if the traumatic event(s) were recurring. (Such reactions may occur on a continuum, with the most extreme expression being a complete loss of awareness of present surroundings.) **Note:** In children, trauma-specific reenactment may occur in play.

4. Intense or prolonged psychological distress or marked physiological reactions in response to internal or external cues that symbolize or resemble an aspect of the traumatic event(s).
   - Negative Mood
5. Persistent inability to experience positive emotions (e.g., inability to experience happiness, satisfaction, or loving feelings).

Dissociative Symptoms

6. An altered sense of the reality of one's surroundings or oneself (e.g., seeing oneself from another's perspective, being in a daze, time slowing).
7. Inability to remember an important aspect of the traumatic event(s) (typically due to dissociative amnesia and not to other factors such as head injury, alcohol, or drugs).

Avoidance Symptoms

8. Efforts to avoid distressing memories, thoughts, or feelings about or closely associated with the traumatic event(s).
9. Efforts to avoid external reminders (people, places, conversations, activities, objects, situations) that arouse distressing memories, thoughts, or feelings about or closely associated with the traumatic event(s).

Arousal Symptoms

10. Sleep disturbance (e.g., difficulty falling or staying asleep, restless sleep).
11. Irritable behavior and angry outbursts (with little or no provocation), typically expressed as verbal or physical aggression toward people or objects.
12. Hypervigilance.
13. Problems with concentration.
14. Exaggerated startle response.

C. Duration of the disturbance (symptoms in Criterion B) is 3 days to 1 month after trauma exposure.

   Note: Symptoms typically begin immediately after the trauma, but persistence for at least 3 days and up to a month is needed to meet disorder criteria.

D. The disturbance causes clinically significant distress or impairment in social, occupational, or other important areas of functioning.

E. The disturbance is not attributable to the physiological effects of a substance (e.g., medication or alcohol) or another medical condition (e.g., mild traumatic brain injury) and is not better explained by brief psychotic disorder.

2. Plan the treatment of a child or adolescent with post-traumatic stress disorder

First-line treatment for PTSD is trauma-focused psychotherapy/trauma-focused cognitive behavioral therapy. There are different types of trauma-based psychotherapy, but there is limited evidence comparing the different psychotherapies. "There is growing support for the use of trauma-focused psychotherapies that (1) directly address children's traumatic experiences, (2) include parents in treatment in some manner as important agents of change, and (3) focus not only on symptom improvement but also on enhancing functioning, resiliency, and/or developmental trajectory." (Cohen et al) If patients do not respond to trauma-focused therapy, there is new evidence for Eye Movement Desensitization and Reprocessing (EMDR).

Medications are not recommended for treatment of PTSD in children or adolescents. Sometimes they are used to treat specific symptoms of PTSD (see below).

3. Understand the etiology and environmental contributors to PTSD
Risk factors: lower socioeconomic status, lower education, exposure to prior trauma, economic deprivation, family dysfunction, lower intelligence, minority race/ethnicity, family history of mental illness, female gender, increased severity of trauma, perceived life threat, personal injury, interpersonal violence, dissociation that occurs during the trauma and persists afterwards, and repeated upsetting reminders. Girls are 2-3 times more likely than boys to develop PTSD.
Protective: Social support (including family stability)

4. Understand the role of medication in the management of acute stress disorder
There is no proven effective medication in the management of acute stress disorder.

5. Understand the role of medication in the management of PTSD
While SSRIs are often used to treat adults with PTSD, there is not enough evidence to recommend using SSRIs for the treatment of PTSD in children and adolescents. The FDA has not approved any medications for the treatment of PTSD in children and adolescents. Some suggest that medications should be trialed in patients who do not respond to or are unable to participate in psychotherapy. There is no consensus on the ideal medications to try. Most recommend using medications to treat the specific symptoms the patient is experiencing (for example, giving a medication to help with sleep for a patient who is experiencing sleep disturbance). In addition, patients should be treated for any comorbid conditions (ADHD, depression, anxiety, etc.).

6. Understand the natural history of PTSD
The natural history of PTSD can vary significantly. Some patients can recover on their own without intervention, while some continue to have symptoms years after the trauma. Additional traumatic events while having PTSD makes recovery less likely.

7. Recognize the importance of active outreach and screening for PTSD after a traumatic event
Recognizing PTSD in children is important because earlier treatment can help prevent long term symptoms. Often, practitioners worry about asking sensitive questions to patients. However, it is important to learn more about the trauma and symptoms the patient is experiencing in order to provide him or her with helpful therapy. There are structured interviews and patient reported questionnaires to help with the diagnosis. Some examples of PTSD interviewing tools include the PTSD Reaction Index for Children and Adolescents, the Clinician-administered PTSD Scale for Children and Adolescents, and Child PTSD Symptom Scale – Interview (CPSS-5-I). Some self-report questionnaires include the Children's Revised Impact of Event Scale (CRIES-8), Child Trauma Screening Questionnaire (CTSQ), Child PTSD Symptom Scale (CPSS), and Child and Adolescent Trauma Screen (CATS). Screening large groups after a public traumatic event is also important for early identification and intervention. Screening typically occurs about 4 weeks after the event in order to accurately detect patients with PTSD. However, schools and local resources should be used early on to provide mental health first aid and support.

8. Recognize that PTSD may have a delayed onset after a traumatic event
Usually symptoms start within 3 months after the trauma. But, sometimes there can be a delay of months or even years. This is now called "delayed expression", not delayed onset.

9. Plan the management of a child with acute stress disorder

The most helpful treatment is trauma-focused cognitive behavioral therapy. Medications have not proven to be beneficial and there are no FDA approved medications for treatment of acute stress disorder. Like PTSD, some will treat the symptoms of acute stress disorder. There have been some studies looking at imipramine along with SSRIs to help with the symptoms of acute stress disorder. However, there is not strong enough evidence to suggest the routine use of such medications.

10. Differentiate the prognosis and treatment for PTSD from that for an acute stress disorder
In the past, many believed that acute stress disorder was the precursor to PTSD and that most patients would progress along the path from acute stress disorder to PTSD if not treated. However, the relationship between these two disorders is not always linear. Patients often do not meet diagnostic criteria for acute stress disorder shortly after the traumatic event, but will later develop PTSD. Conversely, patients with acute stress disorder do not always progress to PTSD. In other words, the "stress responses change over time" (Bryant, 2018) and are hard to predict. The first-line treatment for both PTSD and acute stress disorder is trauma-focused cognitive behavioral therapy. Please see above for more information about management for PTSD and acute stress disorder.

References for PTSD and acute stress disorder:
American Psychiatric Association. Diagnostic and Statistical Manual of Mental Disorders. 5th ed. Washington, DC, USA: APA; 2013
Bryant R. The Current Evidence for Acute Stress Disorder. Curr Psychiatry Rep. 2018;20(12). doi:10.1007/s11920-018-0976-x
Cohen JA, Bukstein O, Walter H, et al. Practice parameter for the assessment and treatment of children and adolescents with posttraumatic stress disorder. J Am Acad Child Adolesc Psychiatry. 2010;49(4):414-430.
Smith P, Dalgleish T, Meiser-Stedman R. Practitioner Review: Posttraumatic stress disorder and its treatment in children and adolescents. J Child Psychol Psychiatry. 2019;60(5):500-515. doi:10.1111/jcpp.12983
Smith P, Perrin S, Dalgleish T, Meiser-Stedman R, Clark DM, Yule W. Treatment of posttraumatic stress disorder in children and adolescents. Curr Opin Psychiatry. 2013;26(1):66-72. doi:10.1097/YCO.0b013e32835b2c01

11. Know the diagnostic criteria, treatment and prognosis for a reactive attachment disorder

Diagnostic Criteria for Reactive Attachment Disorder (F94.1)

A. A consistent pattern of inhibited, emotionally withdrawn behavior toward adult caregivers, manifested by both of the following:
    1. The child rarely or minimally seeks comfort when distressed.
    2. The child rarely or minimally responds to comfort when distressed.

B. A persistent social and emotional disturbance characterized by at least two of the following:
    1. Minimal social and emotional responsiveness to others.
    2. Limited positive affect.
    3. Episodes of unexplained irritability, sadness, or fearfulness that are evident even during nonthreatening interactions with adult caregivers.

B. The child has experienced a pattern of extremes of insufficient care as evidenced by at least one of the following:
    1. Social neglect or deprivation in the form of persistent lack of having basic emotional needs for comfort, stimulation, and affection met by caregiving adults.
    2. Repeated changes of primary caregivers that limit opportunities to form stable attachments (e.g., frequent changes in foster care).
    3. Rearing in unusual settings that severely limit opportunities to form selective attachments (e.g., institutions with high child-to-caregiver ratios).

B. The care in Criterion C is presumed to be responsible for the disturbed behavior in Criterion A (e.g., the disturbances in Criterion A began following the lack of adequate care in Criterion C).

C. The criteria are not met for autism spectrum disorder.

D. The disturbance is evident before age 5 years.

E. The child has a developmental age of at least 9 months.

Specify if:

- **Persistent:** The disorder has been present for more than 12 months.

Specify current severity:

- Reactive attachment disorder is specified as **severe** when a child exhibits all symptoms of the disorder, with each symptom manifesting at relatively high levels.

Clinical observation of attachment:

1	5 minutes	Clinician observes parent-child "free play."	Note especially familiarity, comfort, and warmth in child as he/she interacts with attachment figure.
2	3 minutes	Clinician talks with, then approaches, then attempts to engage child in play.	Most young children exhibit some reticence, especially initially, about engaging with an unfamiliar adult.
3	3 minutes	Clinician picks up child and shows him/her a picture on the wall or looks out window with child.	This increases the stress for the child. Again, note the child's comfort and familiarity with this stranger.
4	3 minutes	Caregiver picks up child and shows him/her a picture on the wall or looks out window with child.	In contrast to stranger pick-up, child should feel obviously more comfortable during this activity.

4a	1 minute	Child is placed between caregiver and stranger, and remote control novel (e.g., scary/exciting) toy is introduced.	Child should seek comfort preferentially from parent. If interested rather than frightened, child should share positive affect with parent.
5	3 minutes	Clinician leaves the room.	This separation should not elicit much of a reaction in the child, as the clinician is a stranger.
6	1 minute	Clinician returns.	Similarly, the child should not be much affected by the stranger's return.
7	3 minutes	Caregiver leaves the room.	Child should definitely take notice of caregiver's departure, although not necessarily exhibit obvious distress. If the child is distressed, the clinician should be of little comfort to the child.
8	1 minute	Caregiver returns.	Child's reunion behavior with caregiver should be congruent with separation behavior. That is, distressed children should seek comfort, and nondistressed children should re-engage positively with caregiver, by introducing him or her to the toy or activity or talking with him or her about what occurred during the separation.

Treatment of reactive attachment disorder: First, ensure the child has an emotionally available caregiver (most important). This is accomplished through therapy by building the caregiver's sensitivity to the child's need to help the child build an internal sense of security. Then, help build positive interactions between the caregiver and child. Then the caregiver can be coached on "sensitive responsiveness." Of note, pharmacological interventions are not recommended

Prognosis of reactive attachment disorder: Prognosis appears to depend on the quality of the caregiving environment following serious neglect. In one study, 22 months after placement from institutions into foster care showed levels of RAD similar to controls. In groups that remained in institutions, symptoms of RAD lasted past age 8. In studies of adopted children, they did not have any symptoms of RAD after a few months to a few years. There are no studies that show an age maximum for which children can no longer form attachments. Long term effects on other areas (interpersonal relationships, mental health) have not been studied.

American Psychiatric Association. Diagnostic and Statistical Manual of Mental Disorders. 5th ed. Washington, DC, USA: APA; 2013

Zeanah CH, Chesher T, Boris NW. Practice parameter for the assessment and treatment of children and adolescents with reactive attachment disorder and disinhibited social engagement disorder. J Am Acad Child Adolesc Psychiatry. 2016;55:990–1003. doi: 10.1016/j.jaac.2016.08.004.

12. Know the diagnostic criteria, treatment and prognosis for an adjustment disorder

Adjustment Disorder DSM 5 Diagnostic Criteria

A. The development of emotional or behavioral symptoms in response to an identifiable stressor(s) occurring within 3 months of the onset of the stressor(s).
B. These symptoms or behaviors are clinically significant, as evidenced by one or both of the following:
    1. Marked distress that is out of proportion to the severity or intensity of the stressor, taking into account the external context and the cultural factors that might influence symptom severity and presentation.
    2. Significant impairment in social, occupational, or other important areas of functioning.
C. The stress-related disturbance does not meet the criteria for another mental disorder and is not merely an exacerbation of a preexisting mental disorder.
D. The symptoms do not represent normal bereavement.
E. Once the stressor or its consequences have terminated, the symptoms do not persist for more than an additional 6 months.

Specify whether:

- **309.0 (F43.21) With depressed mood:** Low mood, tearfulness, or feelings of hopelessness are predominant.
- **309.24 (F43.22) With anxiety:** Nervousness, worry, jitteriness, or separation anxiety is predominant.
- **309.28 (F43.23) With mixed anxiety and depressed mood:** A combination of depression and anxiety is predominant.
- **309.3 (F43.24) With disturbance of conduct:** Disturbance of conduct is predominant.
- **309.4 (F43.25) With mixed disturbance of emotions and conduct:** Both emotional symptoms (e.g., depression, anxiety) and a disturbance of conduct are predominant.
- **309.9 (F43.20) Unspecified:** For maladaptive reactions that are not classifiable as one of the specific subtypes of adjustment disorder.

Specify if:

- **Acute:** If the disturbance lasts less than 6 months.
- **Persistent (chronic):** If the disturbance lasts for 6 months or longer.

Adjustment Disorder treatment: First line treatment involves CBT. Other therapies, including family therapy and group therapy can also help. Medications are not first line, but should be considered to treat anxiety or depression if present (see other sections for treatments of anxiety and depression).

Adjustment Disorder Prognosis: One study found that after 5 years, 50% of adolescents had resolution of their symptoms. The others in the study developed other psychiatric diagnoses, including depression, anxiety, bipolar, and schizophrenia. Not treating adjustment disorder increases the risk of future mental health problems.

C. Depressive disorders

1. Understand the natural history of depressive disorders including disruptive mood dysregulation disorder, persistent depressive disorder (dysthymia), premenstrual dysphoria, and major depressive disorder

See Below

2. Understand the relationship between temperamental characteristics and depressive disorders

See Below

3. Recognize the social and environmental contributors to the development of depressive symptoms

Disruptive Mood Dysregulation Disorder (DMDD) (content specs C1-C3): The diagnosis of DMDD is for patients with chronic, severe, persistent irritability and frequent, extreme temper outbursts that are far beyond temper tantrums (see below for diagnostic criteria). The onset must be before age 10 years, and the diagnosis should not be made for children less than 6 years developmentally. Most are male (unlike bipolar disorder which has an equal gender prevalence). Children with a chronic irritability temperament (but don't meet DMDD criteria) often have complicated psychiatric histories. They are sometimes diagnosed with oppositional defiant disorder (ODD) before criteria for DMDD are met. Half of children with significant symptoms will continue to have symptoms 1 year later. Children with chronic irritability (rather than episodic) are at risk to develop unipolar depression/anxiety disorders as an adult (and not bipolar disorder). Comorbid conditions include ADHD, anxiety disorders, and major depressive disorder.

Persistent Depressive Disorder (Dysthymia) (content specs C1-C3): A negative affectivity temperament leads to greater symptom severity. Childhood risk factors include parent loss or separation. Early onset (in childhood) is associated with a higher likelihood of comorbid personality disorders and substance use disorders.

Premenstrual dysphoria (content specs C1-C3): "Onset of premenstrual dysphoric disorder can occur at any point after menarche." (DSM 5) "Environmental factors associated with the expression of premenstrual dysphoric disorder include stress, history of interpersonal trauma, seasonal changes, and sociocultural aspects of female sexual behavior in general, and female gender role in particular." (DSM 5) "Women who use oral contraceptives may have fewer premenstrual complaints than do women who do not use oral contraceptives." (DSM 5)

Major Depressive Disorder (content specs C1-C3): "Major depressive disorder may first appear at any age, but the likelihood of onset increases markedly with puberty. In the United States, incidence appears to peak in the 20s … The course of major depressive disorder is quite variable, such that some individuals rarely, if ever, experience remission (a period of 2 or more months with no symptoms, or only one or two symptoms to no more than a mild degree), while others experience many years with few or no symptoms between discrete episodes … Recovery typically begins within 3 months of onset for 40% with major depression and within 1 year for 80% of patients. Recency of onset is a strong determinant of the likelihood of near-term recovery, and many individuals who have been depressed only for several months can be expected to recover spontaneously. Features associated with lower recovery rates, other than current episode duration, include psychotic features, prominent anxiety, personality disorders, and symptom severity. The

risk of recurrence becomes progressively lower over time as the duration of remission increases. The risk is higher in individuals whose preceding episode was severe, in younger individuals, and in individuals who have already experienced multiple episodes. The persistence of even mild depressive symptoms during remission is a powerful predictor of recurrence." (DSM 5) "Neuroticism (negative affectivity) is a well-established risk factor for the onset of major depressive disorder, and high levels appear to render individuals more likely to develop depressive episodes in response to stressful life events." (DSM 5) "Adverse childhood experiences, particularly when there are multiple experiences of diverse types, constitute a set of potent risk factors for major depressive disorder. Stressful life events are well recognized as precipitants of major depressive episodes, but the presence or absence of adverse life events near the onset of episodes does not appear to provide a useful guide to prognosis or treatment selection." (DSM 5) "First-degree family members of individuals with major depressive disorder have a risk for major depressive disorder two- to four-fold higher than that of the general population. Relative risks appear to be higher for early-onset and recurrent forms. Heritability is approximately 40%, and the personality trait neuroticism accounts for a substantial portion of this genetic liability." (DSM 5)

4. Recognize the signs and symptoms of dysthymia and the variations in presentation based on developmental stage

Dysthymia (now known as Persistent Depressive Disorder in DSM 5) is defined as a depressed or irritable mood for most of the day, for more days than not, as indicated by either subjective account or observation by others, for at least 1 year (2 years in adults). While depressed, patients have 2 or more of: poor appetite or overeating, insomnia or hypersomnia, low energy or fatigue, low self-esteem, poor concentration or difficulty making decisions, and feelings of hopelessness. Often, younger children will present with more physical complaints. See Table below for similar symptoms related to major depressive disorder.

5. Recognize the signs and symptoms of major depression and the variations in presentation based on developmental stage

"The Centers for Disease Control and Prevention estimate the incidence at 0.5% in children 3-5 years old, 2% for 6- to 11-year-olds, and up to 12% for 12- to 17-year-olds." "Depressed children 3-8 years of age often present with more somatic complaints, are more irritable, display fewer signs of depression, present with symptoms of anxiety, and have other problem behavior…"

TABLE 1: Clinical presentation variation compared to adult symptom onset for major depressive disorder[10]

Age, y	Clinical Presentation Variation
3-5	Trouble verbalizing feelings, marked decreased interest in play, self-destructive themes in play, thoughts of worthlessness or suicide, symptoms do not need to be present for 2 wk
6-8	Trouble verbalizing feelings, increased somatic complaints, crying or shouting outbursts, unexplained irritability, observed anhedonia
9-12	Low self-esteem, guilt, hopelessness, increased boredom, feelings of wanting to run away, and fear of death
13-18	Increased irritability, impulsivity, and behavior changes; decreased grades and poor school performance; increased disturbances in sleep and appetite; suicidality similar to adults; increased likelihood of chronic course of depression; stronger genetic association
≥19	Symptoms similar to adult presentation

Mullen S. Major depressive disorder in children and adolescents. Ment Health Clin. 2018;8(6):275-283. Published 2018 Nov 1. doi:10.9740/mhc.2018.11.275

6. Recognize the common co-morbid conditions of depressive disorders

Substance related disorders, panic disorders, OCD, anorexia nervosa, bulimia nervosa, borderline personality disorder. Persistent depressive disorder: higher risk for anxiety and substance abuse.

7. Understand the pharmacologic treatment of depressive disorders

If symptoms are mild (noticeable but functioning well), the patient does not need to start medication right away. Start with education, improve sleep hygiene, reduce stress, improve peer interactions, and remove guns from the home. Follow up within 2 weeks. If not better, consider counseling. For moderate to severe depression, patients should be referred for individual psychotherapy (CBT or IPT, see below) and considered for medication.

First line: Fluoxetine, second line: escitalopram or sertraline, third line: other SSRIs, bupropion, mirtazepine. Wait four weeks between dose increases to see changes. Check for side effects every 1-2 weeks after first starting.

"Only fluoxetine has been approved by the FDA for use in children and adolescents with depression, and only escitalopram has been approved for use in adolescents aged 12 years and older." (Cheung et al)

Drug Name	Dosage Form	Usual starting dose for adolescent	Increase increment (after ~4 weeks)	RCT evidence in kids	FDA depression approved for children?	Editorial Comments
Fluoxetine (Prozac)	10, 20, 40mg 20mg/5ml	10 mg/day (60mg max)*	10-20mg**	Yes	Yes (Age ≥8)	Long 1/2 life, no side effect from a missed dose
Fluoxetine considered first line per the evidence base in children						
Sertraline (Zoloft)	25, 50, 100mg 20mg/ml	25 mg/day (200mg max)*	25-50mg**	Yes	No	May be prone to side effects when stopping
Escitalopram (Lexapro)	5, 10, 20mg 5mg/5ml	5 mg/day (20mg max)*	5-10mg**	Yes	Yes (Age ≥12)	The active isomer of citalopram.
Escitalopram and Sertraline considered second line per the evidence base in children						
Citalopram (Celexa)	10, 20, 40mg 10mg/5ml	10 mg/day (40mg max)*	10-20mg**	Yes	No	Few drug interactions, dose maximum 40mg/day due to risk of QT prolongation

Bupropion (Wellbutrin)	75, 100mg 100, 150, 200mg SR forms 150, 300mg XL forms	75 mg/day (later dose this BID) (400mg max)*	75-100mg**	No	No	Can have more agitation risk. Avoid if eat d/o. Also has use for ADHD treatment. Seizure risk limits dose.
Mirtazapine (Remeron)	15, 30, 45mg	15 mg/day (45mg max)*	15mg**	No	No	Sedating, increases appetite
Venlafaxine (Effexor)	25, 37.5, 50, 75, 100mg 37.5,75, 150 mg ER forms	37.5 mg/day (225mg max)*	37.5 to 75mg**	No (May have higher SI risk than others for children)	No	Only recommended for older adolescents. Withdrawal symptoms can be severe.
Duloxetine (Cymbalta)	20, 30, 40, 60mg	30 mg/day (120mg max)*	30mg	No	No	May cause nausea. May help with somatic symptoms.
Citalopram, bupropion, mirtazapine, venlafaxine, and duloxeline considered third line treatments per the evidence base in children						

Starting doses in children less than 13 may need to be lowered using liquid forms

Successful medication trials should continue for 6 to 12 months

* Recommend decrease maximum dosage by around 1/3 for pre-pubertal children

** Recommend using the lower dose increase increments for younger children.

There is an FDA black box warning about using antidepressants in children and adolescents because they found an increased risk of suicidality in patients taking antidepressants when compared to placebo (4% and 2%). The FDA recommends that "Pediatric patients being treated with antidepressants for any indication should be closely observed for clinical worsening, as well as agitation, irritability, suicidality, and unusual changes in behavior, especially during the initial few months of a course of drug therapy, or at times of dose changes, either increases or decreases." Later studies have found that the benefits of using antidepressants outweigh the risks, but patients and families should be educated about and closely monitored for potential side effects.

Side effects: Worsening depressive symptoms (eg, suicidal ideation and behavior, anxiety and panic attacks, or insomnia), Agitation, Irritability or hostility, Impulsivity, Akathisia (ie, restlessness and inability to sit down), Hypomania or mania (UpToDate)

Side Effect	Symptoms	Incidence	When most commonly occurs	Response
Suicidality	Thoughts or acts of self-harm	2 percent	1-4 weeks	Discontinue, monitor resolution of suicidality, consider alternative antidepressant

Akathisia	Inner restlessness; sense of being "driven": pacing or movements usually bilateral, symmetrical, often specific muscle groups	5-25 percent	2-6 weeks	Consider switch to alternative agent; if very positive response for depression, consider propanolol, or <4 weeks augmentation with clonazapam
Manic switching	Silliness, giggling, angry outbursts, lack of sleep	2-70 percent in bipolar depression, but 1-10 percent in unipolar depression (TCA >SSRI)	2-4 weeks, or within weeks of dose increases	Discontinue antidepressant; consider mood stablizers if impairing mania (vs milder hypomanic symptoms); after manic symptoms resolve, if prominent depression, consider alternative antidepressants, but titrate slowly and attempt low doses
Discontinuation syndrome	Fear, dizziness, lethargy, parasthesias, nausea, vivid dreams, insomnia, increased irritability or depression	4-18 percent; shorter half-live antidepressants >longer half-life agents	Within 1-7 days of stopping, decreasing antidepressant	Resume antidepressant and titrate down slowly; consider switching/adding long half-life antidepressant (fluoxetine) to allow more gradual taper
Serotonergic syndrome	Confusion, restlessness, agitation, fever, hyperthermia, diaphoresis, hypertonia/clonus (usually symmetrical), tremor, shivering, hyper-reflexia	Rare (<1 percent)	When multiple serotonergic medications are added or combined	Hospital management of hyperthermia, benzodiazepines for seizures or muscle hyperactivity; serotonin antagonists such as cyproheptadine 4-8 mg up to four times a day

Apathy	Disinterest, confusion, lack of enjoyment in previously enjoyed activities; NOT depressed		24-78 weeks	Consider augmentation with additional antidepressant, but at low dose (eg, bupropion SR 100 mg every morning)

Rappaport, N, Bostic, JQ, Prince JB, Jellinek, M. Treating pediatric depression in primary care: Coping with the patients' blue mood and the FDA's black box. J Pediatr 2006; 148:567. Copyright ©2006 Elsevier.

Cheung AH, Zuckerbrot RA, Jensen PS, Laraque D, Stein REK; GLAD-PC STEERING GROUP. Guidelines for Adolescent Depression in Primary Care (GLAD-PC): Part II. Treatment and Ongoing Management. Pediatrics. 2018;141(3):e20174082. doi:10.1542/peds.2017-4082
Hilt R, Barclay R; Primary Care Principles for Child Mental Health. Seattle, Seattle Children's Hospital, Partnership Access Line, 2019. https://www.seattlechildrens.org/healthcare-professionals/access-services/partnership-access-line/resources/

8. Understand the psychological interventions for the treatment of depressive disorders

There are 2 main psychological interventions for depression: Cognitive Behavioral Therapy (CBT) and
Interpersonal Psychotherapy for Depressed Adolescents (IPT-A). CBT is more common and focuses on changing thoughts and behaviors. IPT-A focuses on adolescents' relationship problems as an underlying contributor to depression.

Therapy	Key Components
CBT	Thoughts influence behaviors and feelings and vice versa. Treatment targets patient's thoughts and behaviors to improve his or her mood. Essential elements of CBT include increasing pleasurable activities (behavioral activation), reducing negative thoughts (cognitive restructuring), and improving assertiveness and problem-solving skills to reduce feelings of hopelessness. CBT for adolescents may include sessions with parents and/or caregivers to review progress and to increase compliance with CBT-related tasks.
IPT-A	Interpersonal problems may cause or exacerbate depression, and that depression, in turn, may exacerbate interpersonal problems. Treatment targets patient's interpersonal problems to improve both interpersonal functioning and his or her mood. Essential elements of interpersonal therapy include identifying an interpersonal problem area, improving interpersonal problem-solving skills, and modifying communication patterns. Parents and/or caregivers are involved in sessions during specific phases of the therapy.

Cheung AH, Zuckerbrot RA, Jensen PS, Laraque D, Stein REK; GLAD-PC STEERING GROUP. Guidelines for Adolescent Depression in Primary Care (GLAD-PC): Part II. Treatment and Ongoing Management. Pediatrics. 2018;141(3):e20174082. doi:10.1542/peds.2017-4082

9. Understand the range of prognoses for children and adolescents with dysthymia or depressive disorders

For children, the duration of depression is about 1 year. 30-70 percent with remission had a relapse (UpToDate). Children with depressive and anxiety symptoms that are not sufficient for diagnosis of a true disorder are at a significantly increased risk for major depression or anxiety within the next 10 years (Uchida et al). For adolescents, the average depression episode lasted 6-9months.

90% had remission of symptoms by 2 years. Recurrence occurs in 20-70% of adolescents (UpToDate).

Birmaher B, Arbelaez C, Brent D. Course and outcome of child and adolescent major depressive disorder. Child Adolesc Psychiatr Clin N Am. 2002 Jul;11(3):619-37, x. doi: 10.1016/s1056-4993(02)00011-1. PMID: 12222086.
Uchida M, Fitzgerald M, Woodworth H, Carrellas N, Kelberman C, Biederman J. Subsyndromal Manifestations of Depression in Children Predict the Development of Major Depression. J Pediatr. 2018 Oct;201:252-258.e1. doi: 10.1016/j.jpeds.2018.05.049. Epub 2018 Jul 13. PMID: 30007773; PMCID: PMC6153024.

10. Differentiate between normal grieving associated with a significant loss and major depressive disorder

Normal grieving associated with a significant loss often resembles major depressive disorder and many symptoms overlap. However, just because someone is grieving does not mean they should not be diagnosed with major depressive disorder. The patient's symptoms should be taken in context with the patient's history, cultural norms, and severity of symptoms.

"In distinguishing grief from a major depressive episode (MDE), it is useful to consider that in grief the predominant affect is feelings of emptiness and loss, while in an MDE it is persistent depressed mood and the inability to anticipate happiness or pleasure. The dysphoria in grief is likely to decrease in intensity over days to weeks and occurs in waves, the so-called pangs of grief. These waves tend to be associated with thoughts or reminders of the deceased. The depressed mood of an MDE is more persistent and not tied to specific thoughts or preoccupations. The pain of grief may be accompanied by positive emotions and humor that are uncharacteristic of the pervasive unhappiness and misery characteristic of an MDE. The thought content associated with grief generally features a preoccupation with thoughts and memories of the deceased, rather than the self-critical or pessimistic ruminations seen in an MDE. In grief, self-esteem is generally preserved, whereas in an MDE feelings of worthlessness and self-loathing are common. If self-derogatory ideation is present in grief, it typically involves perceived failings vis-à-vis the deceased (e.g., not visiting frequently enough, not telling the deceased how much he or she was loved). If a bereaved individual thinks about death and dying, such thoughts are generally focused on the deceased and possibly about "joining" the deceased, whereas in an MDE such thoughts are focused on ending one's own life because of feeling worthless, undeserving of life, or unable to cope with the pain of depression." (DSM 5)

11. Know the familial risk of major depressive disorder in children

"First-degree family members of individuals with major depressive disorder have a risk for major depressive disorder two- to fourfold higher than that of the general population. Relative risks appear to be higher for early-onset and recurrent forms. Heritability is approximately 40%, and the personality trait neuroticism accounts for a substantial portion of this genetic liability." (DSM 5)

12. Know the diagnostic criteria for depressive disorders

Major Depressive Disorder DSM 5 Diagnostic Criteria

A. Five (or more) of the following symptoms have been present during the same 2-week period and represent a change from previous functioning; at least one of the symptoms is either (1) depressed mood or (2) loss of interest or pleasure.
   - **Note:** Do not include symptoms that are clearly attributable to another medical condition.

   1. Depressed mood most of the day, nearly every day, as indicated by either subjective report (e.g., feels sad, empty, hopeless) or observation made by others (e.g., appears tearful). (**Note:** In children and adolescents, can be irritable mood.)
   2. Markedly diminished interest or pleasure in all, or almost all, activities most of the day, nearly every day (as indicated by either subjective account or observation).
   3. Significant weight loss when not dieting or weight gain (e.g., a change of more than 5% of body weight in a month), or decrease or increase in appetite nearly every day. (**Note:** In children, consider failure to make expected weight gain.)
   4. Insomnia or hypersomnia nearly every day.
   5. Psychomotor agitation or retardation nearly every day (observable by others, not merely subjective feelings of restlessness or being slowed down).
   6. Fatigue or loss of energy nearly every day.
   7. Feelings of worthlessness or excessive or inappropriate guilt (which may be delusional) nearly every day (not merely self-reproach or guilt about being sick).
   8. Diminished ability to think or concentrate, or indecisiveness, nearly every day (either by subjective account or as observed by others).
   9. Recurrent thoughts of death (not just fear of dying), recurrent suicidal ideation without a specific plan, or a suicide attempt or a specific plan for committing suicide.

B. The symptoms cause clinically significant distress or impairment in social, occupational, or other important areas of functioning.

C. The episode is not attributable to the physiological effects of a substance or another medical condition.

Note: Criteria A–C represent a major depressive episode.

D. The occurrence of the major depressive episode is not better explained by schizoaffective disorder, schizophrenia, schizophreniform disorder, delusional disorder, or other specified and unspecified schizophrenia spectrum and other psychotic disorders.

E. There has never been a manic episode or a hypomanic episode.

Persistent Depressive Disorder (Dysthymia) DSM 5 Diagnostic Criteria

A. Depressed mood for most of the day, for more days than not, as indicated by either subjective account or observation by others, for at least 2 years.

Note: In children and adolescents, mood can be irritable and duration must be at least 1 year.

B. Presence, while depressed, of two (or more) of the following:
    1. Poor appetite or overeating.
    2. Insomnia or hypersomnia.
    3. Low energy or fatigue.
    4. Low self-esteem.
    5. Poor concentration or difficulty making decisions.
    6. Feelings of hopelessness.

C. During the 2-year period (1 year for children or adolescents) of the disturbance, the individual has never been without the symptoms in Criteria A and B for more than 2 months at a time.

D. Criteria for a major depressive disorder may be continuously present for 2 years.

E. There has never been a manic episode or a hypomanic episode, and criteria have never been met for cyclothymic disorder.

F. The disturbance is not better explained by a persistent schizoaffective disorder, schizophrenia, delusional disorder, or other specified or unspecified schizophrenia spectrum and other psychotic disorder.

G. The symptoms are not attributable to the physiological effects of a substance (e.g., a drug of abuse, a medication) or another medical condition (e.g. , hypothyroidism).

H. The symptoms cause clinically significant distress or impairment in social, occupational, or other important areas of functioning.

Note: Because the criteria for a major depressive episode include four symptoms that are absent from the symptom list for persistent depressive disorder (dysthymia), a very limited number of individuals will have depressive symptoms that have persisted longer than 2 years but will not meet criteria for persistent depressive disorder. If full criteria for a major depressive episode have been met at some point during the current episode of illness, they should be given a diagnosis of major depressive disorder. Otherwise, a diagnosis of other specified depressive disorder or unspecified depressive disorder is warranted.

Disruptive Mood Dysregulation Disorder (DMDD) DSM 5 Diagnostic Criteria

A. Severe recurrent temper outbursts manifested verbally (e.g., verbal rages) and/or behaviorally (e.g., physical aggression toward people or property) that are grossly out of proportion in intensity or duration to the situation or provocation.
B. The temper outbursts are inconsistent with developmental level.
C. The temper outbursts occur, on average, three or more times per week.
D. The mood between temper outbursts is persistently irritable or angry most of the day, nearly every day, and is observable by others (e.g., parents, teachers, peers).
E. Criteria A–D have been present for 12 or more months. Throughout that time, the individual has not had a period lasting 3 or more consecutive months without all of the symptoms in Criteria A–D.
F. Criteria A and D are present in at least two of three settings (i.e., at home, at school, with peers) and are severe in at least one of these.
G. The diagnosis should not be made for the first time before age 6 years or after age 18 years.
H. By history or observation, the age at onset of Criteria A–E is before 10 years.
I. There has never been a distinct period lasting more than 1 day during which the full symptom criteria, except duration, for a manic or hypomanic episode have been met.
   Note: Developmentally appropriate mood elevation, such as occurs in the context of a highly positive event or its anticipation, should not be considered as a symptom of mania or hypomania.
J. The behaviors do not occur exclusively during an episode of major depressive disorder and are not better explained by another mental disorder (e.g., autism spectrum disorder, posttraumatic stress disorder, separation anxiety disorder, persistent depressive disorder [dysthymia]).
   Note: This diagnosis cannot coexist with oppositional defiant disorder, intermittent explosive disorder, or bipolar disorder, though it can coexist with others, including major depressive disorder, attention-deficit/hyperactivity disorder, conduct disorder, and substance use disorders. Individuals whose symptoms meet criteria for both disruptive mood dysregulation disorder and oppositional defiant disorder should only be given the diagnosis of disruptive mood dysregulation disorder. If an individual has ever experienced a manic or hypomanic episode, the diagnosis of disruptive mood dysregulation disorder should not be assigned.
K. The symptoms are not attributable to the physiological effects of a substance or another medical or neurological condition.

Premenstrual Dysphoric Disorder DSM 5 Diagnostic Criteria

A. In the majority of menstrual cycles, at least five symptoms must be present in the final week before the onset of menses, start to *improve* within a few days after the onset of menses, and become *minimal* or absent in the week postmenses.

B. One (or more) of the following symptoms must be present:
   1. Marked affective lability (e.g., mood swings; feeling suddenly sad or tearful, or increased sensitivity to rejection).
   2. Marked irritability or anger or increased interpersonal conflicts.
   3. Marked depressed mood, feelings of hopelessness, or self-deprecating thoughts.
   4. Marked anxiety, tension, and/or feelings of being keyed up or on edge.

C. One (or more) of the following symptoms must additionally be present, to reach a total of *five* symptoms when combined with symptoms from Criterion B above.
   1. Decreased interest in usual activities (e.g., work, school, friends, hobbies).
   2. Subjective difficulty in concentration.
   3. Lethargy, easy fatigability, or marked lack of energy.
   4. Marked change in appetite; overeating; or specific food cravings.
   5. Hypersomnia or insomnia.
   6. A sense of being overwhelmed or out of control.
   7. Physical symptoms such as breast tenderness or swelling, joint or muscle pain, a sensation of "bloating," or weight gain.

Note: The symptoms in Criteria A–C must have been met for most menstrual cycles that occurred in the preceding year.

B. The symptoms are associated with clinically significant distress or interference with work, school, usual social activities, or relationships with others (e.g., avoidance of social activities; decreased productivity and efficiency at work, school, or home).

C. The disturbance is not merely an exacerbation of the symptoms of another disorder, such as major depressive disorder, panic disorder, persistent depressive disorder (dysthymia), or a personality disorder (although it may co-occur with any of these disorders).

D. Criterion A should be confirmed by prospective daily ratings during at least two symptomatic cycles. (**Note:** The diagnosis may be made provisionally prior to this confirmation.)

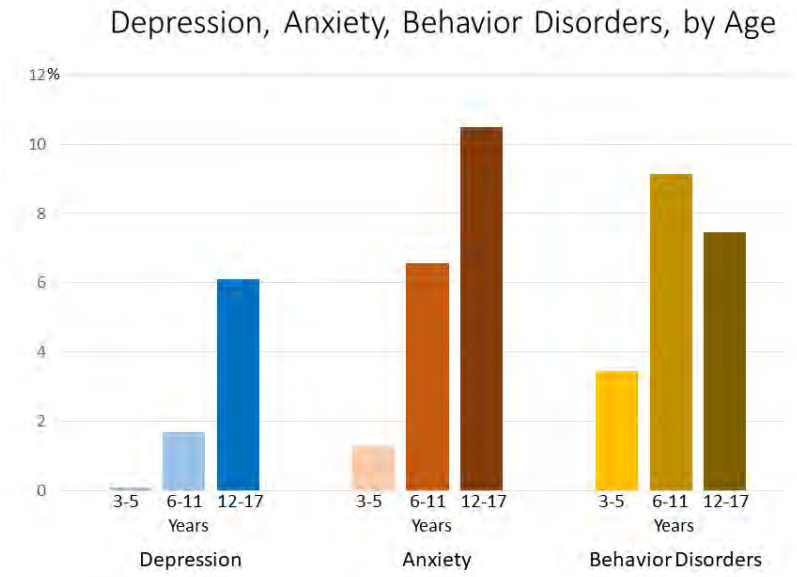
E. The symptoms are not attributable to the physiological effects of a substance (e.g., a drug of abuse, a medication, other treatment) or another medical condition (e.g., hyperthyroidism).

13. Recognize the family systems factors that contribute to depressive disorders
Authoritarian and distant/neglectful parenting are associated with higher levels of depression; authoritative parenting is associated with less depression. Increased parental warmth decreases the risk of internalizing disorders. Psychological control and harsh control increase the risk of anxiety and depression. More autonomy granting is negatively associated with internalizing symptoms.

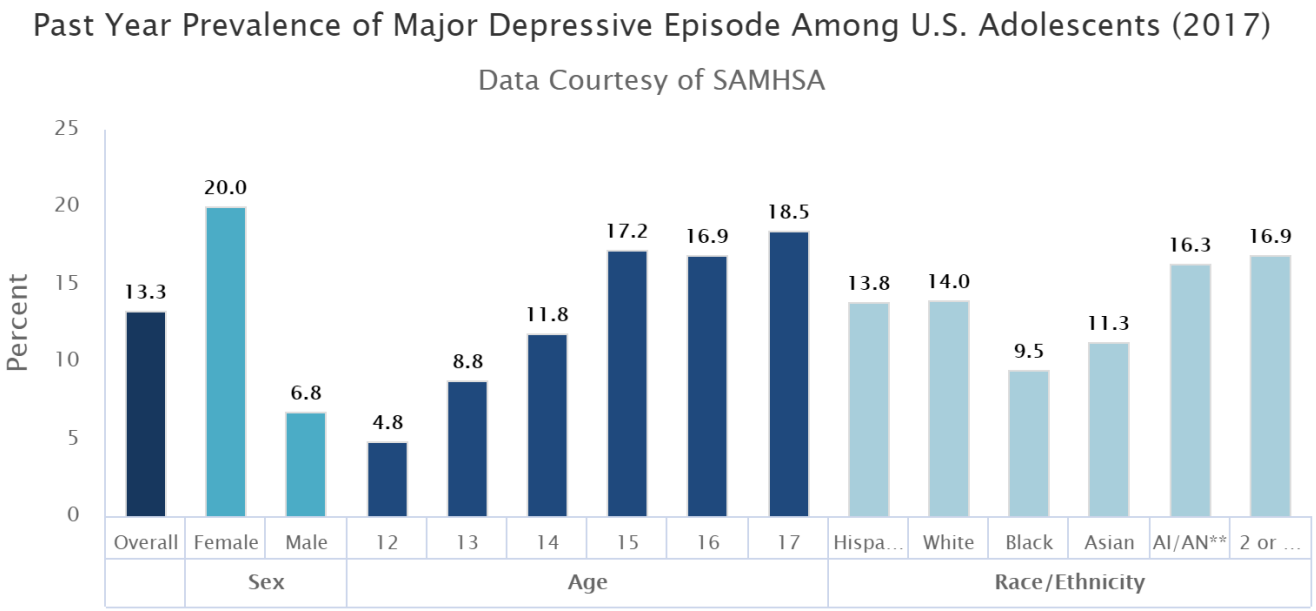
In addition, Adverse Childhood Experiences (ACEs) are associated with depression. All categories of ACEs increased the risk for depression, but children with exposure to violence and household dysfunction had the highest rates of depression. Exposure to only 2 or more ACEs increased the risk for depression and anxiety. There was a slight increase in risk with each additional ACE, but it was relatively stable between 2 or more and 6 or more ACEs.

Elmore AL, Crouch E. The Association of Adverse Childhood Experiences With Anxiety and Depression for Children and Youth, 8 to 17 Years of Age. Acad Pediatr. 2020;20(5):600-608. doi:10.1016/j.acap.2020.02.012
Gorostiaga A, Aliri J, Balluerka N, Lameirinhas J. Parenting Styles and Internalizing Symptoms in Adolescence: A Systematic Literature Review. Int J Environ Res Public Health. 2019;16(17):3192. Published 2019 Sep 1. doi:10.3390/ijerph16173192
King KA, Vidourek RA, Merianos AL. Authoritarian parenting and youth depression: Results from a national study. J Prev Interv Community. 2016;44(2):130-139. doi:10.1080/10852352.2016.1132870
Lindblom J, Vänskä M, Flykt M, et al. From early family systems to internalizing symptoms: The role of emotion regulation and peer relations. J Fam Psychol. 2017;31(3):316-326. doi:10.1037/fam0000260

14. Understand the epidemiology of depression in children and adolescents (eg, gender-based differences, age-based differences, etc)
The rates of depression increase with age. In a 2016 study, the prevalence of depression in children 3-5 years was 0.08%, children 6-11 years 1.7%, and 12-17 years 6.1%. In that study, the rates of depression between males and females were the same. But, the rates of depression were not broken down by gender and age. In adolescents, the rate of depression is higher in females than males (about 2-3:1).



https://www.cdc.gov/childrensmentalhealth/data.html#ref



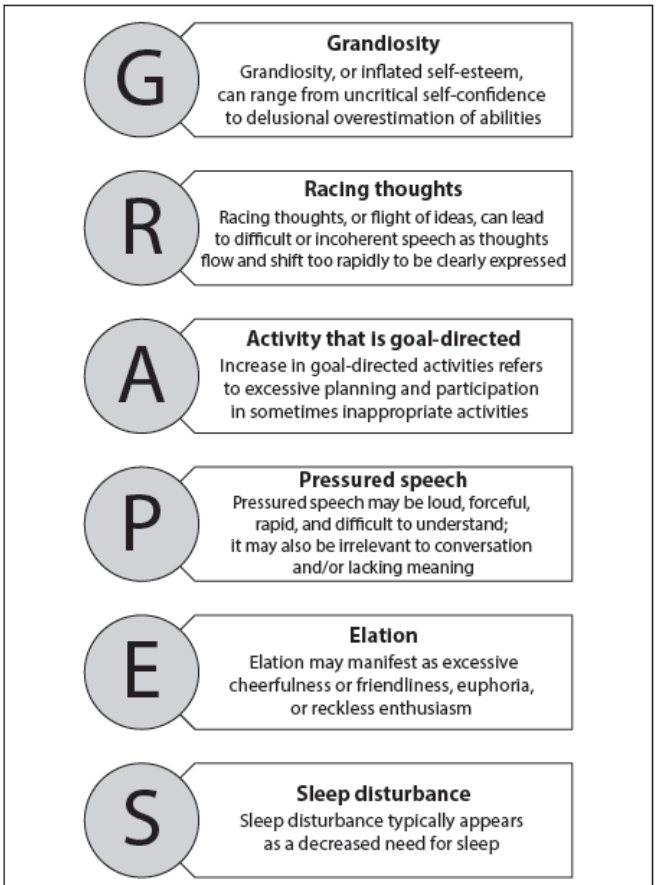
https://www.nimh.nih.gov/health/statistics/major-depression.shtml

D. Bipolar and related disorders

1. Recognize the signs and symptoms of bipolar disorders

The diagnosis of bipolar disorder in children is challenging. The DSM 5 diagnostic criteria for children and adults are the same. When children show signs of a manic episode, the diagnosis of bipolar should be considered. The DSM 5 criteria below define manic and hypomanic episodes, but see the figure below for common signs of mania in children and adolescents. Diagnosing manic episodes in children and adolescents can be challenging because they typically have more mood fluctuations than adults and their mood can change quickly. For children and adolescents, the elevated/irritable mood must last for most of each day over at least 1 week for mania and 4 days for hypomania.

Figure 2. Common Symptoms of Mania in Children and Adolescents[a]



[a]Based on Freeman et al[2] and the American Psychiatric Association.[25]

There was concern in the past that children were being over diagnosed with bipolar disorders, so DSM 5 included a new diagnosis: Disruptive Mood Dysregulation Disorder (DMDD). The main difference between bipolar disorders and DMDD is that the symptoms and irritability of DMDD are persistent. The irritability may wax and wane, but is always present. Bipolar symptoms are episodic changes from the child's usual baseline. Parents should be able to identify a discrete time period when the child's behavior was different from his or her baseline. The irritable mood of DMDD persists over several months.

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Symptom	Bipolar mania hypomania	Attention deficit hyperactivity disorder	Oppositional defiant disorder
Elation	Episodic, prolonged, pathological (inappropriate to context or uncharacteristic), associated with change in functioning, "travels" with ≥3 other manic symptoms	If present, not clearly episodic or pathological	If present, not clearly episodic or pathological
Irritability	Episodic, prolonged, pathological, associated with change in functioning, "travels" with ≥4 other manic symptoms	Can be an associated feature, related to stimulant rebound, or due to a comorbid illness (eg, ODD)	Diagnostic criterion, lacks distinct prolonged episodes, does not "travel" with other manic symptoms
Sleep	Reduced need for sleep (ie, significantly less sleep than usual without increased daytime fatigue or somnolence); change must be mood-related	Insomnia (ie, difficulty falling asleep); can be an associated feature or associated with stimulants, but need for sleep is unchanged	Not a symptom or common characteristic; may defy bedtime rules or routine
Grandiosity	Distinct uncharacteristic increase in confidence or self-importance; change must be mood-related	Not a symptom or common characteristic	Defiance toward authority figures is common but not necessarily mood-related
Hyperactivity and distractibility	Episodic; if comorbid ADHD is diagnosed, then distinctly "worse than usual" change must be mood-related	Diagnostic criteria, nonepisodic	Not prominent or episodic

Differentiating manic symptoms from ADHD can also be challenging. "Notably, ADHD is an ongoing condition whereas BD is episodic, and decreased sleep, hypersexuality, hallucinations or delusions, and homicidal or suicidal thoughts and actions occur with childhood mania, but are rare or absent in uncomplicated ADHD." (Natham et al)

DSM 5 Diagnostic Criteria
Manic Episode, Hypomanic Episode, Major Depressive Episode

Manic Episode

A. A distinct period of abnormally and persistently elevated, expansive, or irritable mood and abnormally and persistently increased activity or energy, lasting at least 1 week and present most of the day, nearly every day (or any duration if hospitalization is necessary).
B. During the period of mood disturbance and increased energy or activity, three (or more) of the following symptoms (four if the mood is only irritable) are present to a significant degree and represent a noticeable change from usual behavior:
    - Inflated self-esteem or grandiosity.
    - Decreased need for sleep (e.g., feels rested after only 3 hours of sleep).
    - More talkative than usual or pressure to keep talking.
    - Flight of ideas or subjective experience that thoughts are racing.
    - Distractibility (i.e., attention too easily drawn to unimportant or irrelevant external stimuli), as reported or observed.
    - Increase in goal-directed activity (either socially, at work or school, or sexually) or psychomotor agitation (i.e., purposeless non-goal-directed activity).
    - Excessive involvement in activities that have a high potential for painful consequences (e.g., engaging in unrestrained buying sprees, sexual indiscretions, or foolish business investments).

C. The mood disturbance is sufficiently severe to cause marked impairment in social or occupational functioning or to necessitate hospitalization to prevent harm to self or others, or there are psychotic features.
D. The episode is not attributable to the physiological effects of a substance (e.g., a drug of abuse, a medication, other treatment) or another medical condition.

Hypomanic Episode

A. A distinct period of abnormally and persistently elevated, expansive, or irritable mood and abnormally and persistently increased activity or energy, lasting at least 4 consecutive days and present most of the day, nearly every day.
B. During the period of mood disturbance and increased energy and activity, three (or more) of the following symptoms (four if the mood is only irritable) have persisted, represent a noticeable change from usual behavior, and have been present to a significant degree:Inflated self-esteem or grandiosity.
    a. Decreased need for sleep (e.g., feels rested after only 3 hours of sleep).
    b. More talkative than usual or pressure to keep talking.
    c. Flight of ideas or subjective experience that thoughts are racing.
    d. Distractibility (i.e., attention too easily drawn to unimportant or irrelevant external stimuli), as reported or observed.
    e. Increase in goal-directed activity (either socially, at work or school, or sexually) or psychomotor agitation.
    f. Excessive involvement in activities that have a high potential for painful consequences (e.g., engaging in unrestrained buying sprees, sexual indiscretions, or foolish business investments).
C. The episode is associated with an unequivocal change in functioning that is uncharacteristic of the individual when not symptomatic.
D. The disturbance in mood and the change in functioning are observable by others.
E. The episode is not severe enough to cause marked impairment in social or occupational functioning or to necessitate hospitalization. If there are psychotic features, the episode is, by definition, manic.
F. The episode is not attributable to the physiological effects of a substance (e.g., a drug of abuse, a medication, other treatment) or another medical condition .

Major Depressive Episode

A. Five (or more) of the following symptoms have been present during the same 2-week period and represent a change from previous functioning; at least one of the symptoms is either (1) depressed mood or (2) loss of interest or pleasure.
    a. Depressed mood most of the day, nearly every day, as indicated by either subjective report (e.g., feels sad, empty, or hopeless) or observation made by others (e.g., appears tearful). (Note: In children and adolescents, can be irritable mood.)
    b. Markedly diminished interest or pleasure in all, or almost all, activities most of the day, nearly every day (as indicated by either subjective account or observation).
    c. Significant weight loss when not dieting or weight gain (e.g., a change of more than 5% of body weight in a month), or decrease or increase in appetite nearly every day. (Note: In children, consider failure to make expected weight gain.)

d. Insomnia or hypersomnia nearly every day.
e. Psychomotor agitation or retardation nearly every day (observable by others; not merely subjective feelings of restlessness or being slowed down).
f. Fatigue or loss of energy nearly every day.
g. Feelings of worthlessness or excessive or inappropriate guilt (which may be delusional) nearly every day (not merely self-reproach or guilt about being sick).
h. Diminished ability to think or concentrate, or indecisiveness, nearly every day (either by subjective account or as observed by others).
i. Recurrent thoughts of death (not just fear of dying), recurrent suicidal ideation without a specific plan, or a suicide attempt or a specific plan for committing suicide.

B. The symptoms cause clinically significant distress or impairment in social, occupational, or other important areas of functioning.
C. The episode is not attributable to the physiological effects of a substance or another medical condition.

Bipolar I Disorder Diagnostic Criteria
A. Criteria have been met for at least one manic episode (Criteria A–D under "Manic Episode" above).

B. The occurrence of the manic and major depressive episode(s) is not better explained by schizoaffective disorder, schizophrenia, schizophreniform disorder, delusional disorder, or other specified or unspecified schizophrenia spectrum and other psychotic disorder.

Bipolar II Disorder Diagnostic Criteria
A. Criteria have been met for at least one hypomanic episode (Criteria A–F under "Hypomanic Episode" above) and at least one major depressive episode (Criteria A–C under "Major Depressive Episode" above).
B. There has never been a manic episode.
C. The occurrence of the hypomanic episode(s) and major depressive episode(s) is not better explained by schizoaffective disorder, schizophrenia, schizophreniform disorder, delusional disorder, or other specified or unspecified schizophrenia spectrum and other psychotic disorder.
D. The symptoms of depression or the unpredictability caused by frequent alternation between periods of depression and hypomania causes clinically significant distress or impairment in social, occupational, or other important areas of functioning.

2. Understand the range of prognoses for children and adolescents with bipolar disorders

"More than 90% of individuals who have a single manic episode go on to have recurrent mood episodes." (DSM 5) In a 2009 study based on children with a DSM IV diagnosis of bipolar, 80% of youth recovered after 2.5 years. However, 60% of those children had a recurrence of symptoms within 1.5 years. Thus, pediatric patients often have recovery of symptoms, but recurrence of mood episodes at some point over the next few years. Worse outcomes are associated with "early age of onset (eg, <13 years), long duration of illness, low socioeconomic status, mixed or rapid cycling episodes, psychosis, subsyndromal symptoms, comorbid disorders (eg, anxiety

disorders or attention deficit hyperactivity disorder), negative life events (eg, abuse), family psychopathology, poor adherence to pharmacotherapy, and lack of psychotherapy" (Uptodate).

3. Know potential side effects of SSRI treatment for teens with bipolar disorders

While there is limited data, SSRI's should be used very cautiously in patients with bipolar disorders as SSRI's may destabilize children and lead to a manic episode. Nevertheless, they are sometimes used in combination with a mood stabilizing medication to treat children with bipolar depression.

For children with a diagnosis of depression, physicians should remember that the child may actually have bipolar disorder, but there was no reported history of manic episodes. If a child develops manic symptoms, the SSRI should be stopped. In addition, physicians should be aware when treating depression or ADHD in children of parents with bipolar disorder, as they are at an increased risk of bipolar disorder. However, this does not mean these children should be denied these medications and there are not different guidelines in treating children with depression or ADHD if they have a parent with bipolar disorder.

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E. Ritualistic/obsessive compulsive behavior

1. Know the natural history of OCD

"In the United States, the mean age at onset of OCD is 19.5 years, and 25% of cases start by age 14 years." (DSM 5) One study showed that about half of adults with OCD started to have symptoms during childhood, but many were not diagnosed with OCD. Many go untreated or undiagnosed for more than 7 years after onset of symptoms. Symptoms usually worsen gradually over time. "If OCD is untreated, the course is usually chronic, often with waxing and waning symptoms (Ravizza et al. 1997; Skoog and Skoog 1999). Some individuals have an episodic course, and a minority have a deteriorating course. Without treatment, remission rates in adults are low (e.g., 20% for those reevaluated 40 years later) (Skoog and Skoog 1999). Onset in childhood or adolescence can lead to a lifetime of OCD. However, 40% of individuals with onset of OCD in childhood or adolescence may experience remission by early adulthood (Stewart et al. 2004). The course of OCD is often complicated by the co-occurrence of other disorders." (DSM 5)

2. Understand the impact of OCD symptoms on the child's functioning in his family, at school and with peers

Children with OCD are often bullied because of their symptoms. Often, they are aware of their behavior and that their peers are watching and judging their OCD behaviors. In schools, teachers can educate the class about OCD and allow the child with OCD to explain his or her disorder to the other children. OCD symptoms can also impact the child's ability to learn and participate in school. OCD is not included specifically in the IDEA, but services can fall under the Other Health Impairment (OHI)" or "Emotional Disturbance (ED)" exception. If children do not qualify for an IEP, they may be eligible for a 504 plan

Families can also be disrupted by OCD symptoms. Plans often have to be changed, the OCD behaviors can be disruptive in many different environments, and parents and siblings can develop frustration and resentment. Families should be involved closely with the child's therapy.

3. Differentiate OCD from normal variations in obsessive or compulsive personality traits

"Obsessive-compulsive personality disorder is not characterized by intrusive thoughts, images, or urges or by repetitive behaviors that are performed in response to these intrusions; instead, it involves an enduring and pervasive maladaptive pattern of excessive perfectionism and rigid control."

4. Understand the range of prognoses for children with obsessive-compulsive disorder

OCD is a challenging disorder to treat and many patients have a persistent diagnosis despite treatment. New treatments may produce improvement, but some patients continue with chronic and debilitating course. About 60% of children with OCD will have persistent full or subthreshold OCD. "Earlier age of OCD onset, increased illness duration, inpatient status, the presence of comorbidities and a positive family his-tory seem to predict greater rates of persistence." (Fineberg et al) In another study, 68% of children still met diagnostic criteria for OCD 2-7 years after presentation and 48% had an additional diagnosis (most often depression or anxiety). (Flament 1990)

5. Understand the etiologies of obsessive-compulsive disorder

OCD is highly genetic (see below). Physical and sexual abuse in childhood and other stressful or traumatic events increase the risk for OCD.

There is a lot of debate surrounding pediatric autoimmune neuropsychiatric disorder associated with group A streptococci (PANDAS). PANDAS is a disorder in which children have a worsening of their OCD or tics after an infection with group A beta-hemolytic streptococcus. It could be considered when the OCD or tics have an acute onset associated with symptoms of acute pharyngitis. It is likely that a small subset of patients with OCD and tics have onset and exacerbations linked to PANDAS.

6. Know the pharmacological treatments for obsessive-compulsive disorder
SSRIs (fluoxetine, fluvoxamine, and sertraline): Short term efficacy established in multiple publications. Generally a 12-week trial with adequate dosage is necessary (including 4-6 weeks at a maximum tolerable dose). Many patients do not have symptom relief until 6-12 weeks. The SSRI can be continued beyond the 12 week trial if symptoms improve. If symptoms do not improve after the 12 week trial, a different SSRI can be started. If that fails, the patient can then trial Clomipramine.
Clomipramine (tricyclic antidepressant and SRI): Approved for use in children with OCD. Some trials have shown clomipramine to be more effective than SSRIs in children. However, clomipramine has a higher side effect profile, so SSRIs are still first line. Children need an EKG prior to starting treatment as clomipramine can cause delayed cardiac conduction, arrhythmia, prolonged QTc, and tachycardia. Other side effects include metabolic issues (weight gain, increased appetite), nausea, sedation, dry mouth, and others.
Augmentation with CBT or combined treatment may be necessary.

TABLE 1 Dosing Guidelines

Drug	Starting Dose (mg)		Typical Dose Range (mg) (Mean Dose)[a]
	Preadolescent	Adolescent	
Clomipramine[b,c]	6.25–25	25	50–200
Fluoxetine[b,d]	2.5–10	10–20	10–80 (25)
Sertraline[b,d]	12.5–25	25–50	50–200 (178)
Fluvoxamine[b,c]	12.5–25	25–50	50–300 (165)
Paroxetine[e]	2.5–10	10	10–60 (32)
Citalopram[d]	2.5–10	10–20	10–60

Note: [a]Mean daily doses used in randomized controlled trials.
[b]Approved by the Food and Drug Administration for obsessive-compulsive disorder in children and adolescents.
[c]Doses lower than 25 mg/day may be administered by compounding 25 mg into a 5-mL suspension.
[d]Oral concentrate commercially available.
[e]Oral suspension commercially available.

7. Know the diagnostic criteria for OCD

Obsessive-Compulsive Disorder (OCD) (F42.2)

A. Presence of obsessions, compulsions, or both:
- Obsessions are defined by (1) and (2):

1. Recurrent and persistent thoughts, urges, or images that are experienced, at some time during the disturbance, as intrusive and unwanted, and that in most individuals cause marked anxiety or distress.

2. The individual attempts to ignore or suppress such thoughts, urges, or images, or to neutralize them with some other thought or action (i.e., by performing a compulsion).

- Compulsions are defined by (1) and (2):

1. Repetitive behaviors (e.g., hand washing, ordering, checking) or mental acts (e.g., praying, counting, repeating words silently) that the individual feels driven to perform in response to an obsession or according to rules that must be applied rigidly.
2. The behaviors or mental acts are aimed at preventing or reducing anxiety or distress, or preventing some dreaded event or situation; however, these behaviors or mental acts are not connected in a realistic way with what they are designed to neutralize or prevent, or are clearly excessive.
    - **Note:** Young children may not be able to articulate the aims of these behaviors or mental acts.

B. The obsessions or compulsions are time-consuming (e.g., take more than 1 hour per day) or cause clinically significant distress or impairment in social, occupational, or other important areas of functioning.

C. The obsessive-compulsive symptoms are not attributable to the physiological effects of a substance (e.g., a drug of abuse, a medication) or another medical condition.

D. The disturbance is not better explained by the symptoms of another mental disorder (e.g., excessive worries, as in generalized anxiety disorder; preoccupation with appearance, as in body dysmorphic disorder; difficulty discarding or parting with possessions, as in hoarding disorder; hair pulling, as in trichotillomania [hair-pulling disorder]; skin picking, as in excoriation [skin-picking] disorder; stereotypies, as in stereotypic movement disorder; ritualized eating behavior, as in eating disorders; preoccupation with substances or gambling, as in substance-related and addictive disorders; preoccupation with having an illness, as in illness anxiety disorder; sexual urges or fantasies, as in paraphilic disorders; impulses, as in disruptive, impulse-control, and conduct disorders; guilty ruminations, as in major depressive disorder; thought insertion or delusional preoccupations, as in schizophrenia spectrum and other psychotic disorders; or repetitive patterns of behavior, as in autism spectrum disorder).

Specify if:

- **With good or fair insight:** The individual recognizes that OCD beliefs are definitely or probably not true or that they may or may not be true.
- **With poor insight:** The individual thinks OCD beliefs are probably true.
- **With absent insight/delusional beliefs:** The individual is completely convinced that OCD beliefs are true.

Specify if:

- **Tic-related:** The individual has a current or past history of a tic disorder.

8. Recognize the signs and symptoms of OCD

"The characteristic symptoms of OCD are the presence of obsessions and compulsions (Criterion A). Obsessions are repetitive and persistent thoughts (e.g., of contamination), images (e.g., of violent or horrific scenes), or urges (e.g., to stab someone). Importantly, obsessions are not pleasurable or experienced as voluntary: they are intrusive and unwanted and cause marked distress or anxiety in most individuals. The individual attempts to ignore or suppress these obsessions (e.g., avoiding triggers or using thought suppression) or to neutralize them with another thought or action (e.g., performing a compulsion)." (DSM 5) Obsessions can be very challenging to recognize in children because they may be unable to describe their obsessions well. However, children often have obsessions even when only their compulsions are obvious. "Compulsions (or rituals) are

repetitive behaviors (e.g., washing, checking) or mental acts (e.g., counting, repeating words silently) that the individual feels driven to perform in response to an obsession or according to rules that must be applied rigidly. Most individuals with OCD have both obsessions and compulsions (Foa et al. 1995). Compulsions are typically performed in response to an obsession (e.g., thoughts of contamination leading to washing rituals or that something is incorrect leading to repeating rituals until it feels "just right"). The aim is to reduce the distress triggered by obsessions or to prevent a feared event (e.g., becoming ill). However, these compulsions either are not connected in a realistic way to the feared event (e.g., arranging items symmetrically to prevent harm to a loved one) or are clearly excessive (e.g., showering for hours each day). Compulsions are not done for pleasure, although some individuals experience relief from anxiety or distress." (DSM 5) "Certain symptom dimensions are common in OCD, including those of cleaning (contamination obsessions and cleaning compulsions); symmetry (symmetry obsessions and repeating, ordering, and counting compulsions); forbidden or taboo thoughts (e.g., aggressive, sexual, and religious obsessions and related compulsions); and harm (e.g., fears of harm to oneself or others and related checking compulsions)." (DSM 5) The obsessions and compulsions will often wax and wane as well as vary with developmental stage. "Higher rates of sexual and religious obsessions in adolescents than in children; higher rates of harm obsessions [e.g., fears of catastrophic events, such as death or illness to self or loved ones] in children and adolescents than in adults." (DSM 5)

Some good screening questions are: "Do you ever have unwanted thoughts or worries that won't go away? Are there things you have to do over and over again, even though you don't want to or that don't make sense?" One of the best diagnostic screening tools is the Children's Yale-Brown-Obsessive-Compulsive Scale (CY-BOCS). This is an involved tool and requires training.

9. Understand the genetics of OCD
The rate of OCD among first-degree relatives of *adults* with OCD is approximately two times that among first-degree relatives of those without the disorder; however, among first-degree relatives of individuals with onset of OCD in *childhood or adolescence*, the rate is increased 10-fold. Familial transmission is due in part to genetic factors (e.g., a concordance rate of 0.57 for monozygotic vs. 0.22 for dizygotic twins). Genetic linkage studies of OCD have found evidence for susceptibility loci on chromosomes 1q, 3q, 6q, 7p, 9p, 10p, and 15q. There is increasing evidence that glutamate receptor/modulating genes may be associated with OCD.

10. Know the psychological treatments for OCD
First line treatment for OCD is Cognitive Behavioral Therapy (CBT). When symptoms are more severe, CBT failed, or the patient has comorbid condition (such as depression), an SSRI should be trialed (see above). CBT is the only psychological treatment that has been shown to help OCD. An important piece of CBT is Exposure and Response Prevention (ERP). Patients choose to expose themselves to the anxiety trigger/what causes the obsession. Then, they avoid doing the compulsive behavior (or response).

Some other psychological treatments include: a structured treatment protocol for children entitled: "How I ran OCD off my Land": A cognitive-behavioral program (March & Mulle 1998); psychodynamic psychotherapy may have indirect benefit e.g. understanding how OCD affects self-esteem and encouraging compliance with behavior of medication focusing directly on OCD symptoms; and family therapy is an important treatment to assist with family discord and other factors that may complicate the child's suffering.

11. Recognize the common co-morbid conditions that may be associated with OCD
Lifetime diagnosis of an anxiety disorder in 76% of patients, or a depressive or bipolar disorder in 64% of patients. Up to 30% of individuals with OCD also have a lifetime tic disorder. A comorbid tic disorder is most common in males with onset of OCD in childhood. Patients with schizophrenia or schizoaffective disorder, bipolar disorder, anorexia, bulimia nervosa, and Tourette's disorder should be assessed for OCD.

Sources for the OCD section:

Fineberg NA, Hollander E, Pallanti S, et al. Clinical advances in obsessive-compulsive disorder: a position statement by the International College of Obsessive-Compulsive Spectrum Disorders. Int Clin Psychopharmacol. 2020;35(4):173-193. doi:10.1097/YIC.0000000000000314
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F. Suicidal behavior

The AAP's 2016 Policy Statement on Suicide and Suicide Attempts in Adolescents is a good resource for this topic: Shain B; COMMITTEE ON ADOLESCENCE. Suicide and Suicide Attempts in Adolescents. Pediatrics. 2016 Jul;138(1):e20161420. doi: 10.1542/peds.2016-1420. PMID: 27354459.

1. Know the risk factors associated with suicidal behavior in children and adolescents
"Previous suicide attempts, impulsivity, a mood or disruptive behavior disorder, recent psychiatric hospitalization, substance abuse, a family history of suicide, or a history of sexual or physical abuse; are homeless/runaway; or who identify as lesbian, gay, bisexual, or transgender."

Top risk factors for suicide:

- Conflict in a romantic relationship
- Bullying
- Childhood affective and disruptive disorders
- Abuse
- Mental illness
- Substance and alcohol abuse
- Social difficulties

Subjects who attempted suicide older than 12 years were predominantly females, whereas children attempting suicide under 12 years of age were predominantly males.

Risk factors for suicide among adolescents

Demographic	Clinical	Family and environmental	Mental State
- Male sex	- Psychiatric illness (major depression, bipolar disorder, conduct disorder)	- Particularly stressful events	- Thoughts on suicide
- Older age	- Recent discharge from a psychiatric hospital	- Availability of lethal means	- Abuse of alcohol or illegal substances
- Non heterosexual	- Previous attempted suicide	- Lack of social support	- State of anxiety, agitation, being without hope
	- Family history of depression or suicide	- Contact with subjects with suicidal behaviors	- Impulsiveness
	- Physical/sexual abuse	- Loss of a parent (death/divorce)	- Difficulty in troubleshooting
	- History of trauma in childhood	- Difficult relationship with parents	
	- Sleep disorders	- Perception of excessive control/poor care by parents	
	- Organic pathologies that determine functional limitations	- Bullying	
	- Personality disorders		
	- Low self-esteem		
	- Low compliance to therapy		

2. Know the risk factors associated with a poor prognosis for children and adolescents who have attempted suicide

Risk Factors for recurrence of attempted suicide

Case history:	Demographic data:
- Constant thoughts of suicide	- Living alone
- Previous attempted suicide	- Male gender
Mental state:	
- Depression, mania, hypomania, anxiety, or all of these mental states	
- Substance abuse	
- Irritability, agitation, hallucinations, violent attitude	

Adapated from: Shaffer D, Pfeffer CR et al. Practice parameter for assessment and treatment of children and adolescents with suicidal behavior. J Am Acad Child Adolesc Psychiatry. 2001; 40(7 Supplement): 24S-51S)

3. Know the steps in the prevention of suicidal behavior and management of an adolescent at risk of suicidal behavior

Even if there are no specific tests to identify subjects susceptible to suicide, risk factors must be investigated by the pediatrician and taken into consideration suicidal ideation is suspected, the following specific questions should be investigated:

- Have you ever thought to kill yourself or would you rather be dead?
- Have you ever done something with the purpose to do yourself harm or kill yourself?
- Have you ever considered methods to put an end to your existence?

Family factors. Familiarity for suicidal behaviors is a risk factor for suicide regardless of the presence of psychiatric illnesses. Strife in the family, loss of a family member (divorce/death), possible abuse or relationship difficulties between parents and children. In particular, a recent review showed a close association between suicidal behaviors and a parent/child relationship characterized by poor care or overprotection.

Substance abuse. Abuse of drugs and alcohol should always be investigated, since it increases the possibility of suicide, especially among male adolescents.

Correlation between attempted suicide and "Heavy Episodic Drinking" (HED), a phenomenon increasingly observed among young people: HED increases the risk of suicide by 2.6 times in children under 13 years and by 1.2 times in subjects aged more than 18 years. Drinking alcohol when one feels "low" increases the risk of attempted suicide by 3 times.

Sexual and physical abuse. Sexual and physical abuse are important risk factors and higher in subjects aged between 16 and 25 years [21]. 50 % of women and 33 % of men who attempted suicide experienced physical or sexual abuse or domestic violence.
Sexual orientation. The pediatrician should also assess the sexual orientation of adolescents who are at risk of suicide. Literature shows that young homosexual men are 2-3 times more likely to commit suicide compared to their heterosexual peers. In particular, those living in families that refuse their sexual orientation have a risk of suicide 8 times greater than those whose sexual orientation is accepted by the family.

Bullying. Both "bullies" and "victims" of bullying are at an increased risk of suicidal behaviors. Victim of bullying affects mental growth in terms of self-esteem and personal well-being, and may lead to the development of depressive syndromes.

Safety of the home environment

- environmental precautions are aimed at restricting access to means of suicide (e.g. guns, ropes, medications
- Restriction of access to means for suicide is a fundamental suicide prevention strategy in adolescents.
- Limiting availability of firearms in countries with high prevalence of suicide by shooting is mandatory: presence of firearms in households increases the risk of adolescent suicide and restriction of their availability reduces this risk.
- Limiting availability of storage of pesticides in rural areas of developing countries, where suicide by pesticide ingestion is common, has also been suggested
- Limiting the availability of pack sizes of drugs that are commonly used for self-poisoning in young people.

4. Know how to assess a child or adolescent with suicidal ideation

Consensus warning signs of suicide

A person at risk for suicidal behavior most often will exhibit warning signs such as:		
I	Ideation	Expressed or communicated ideation
		Threatening to hurt or kill him/herself or talking of wanting to hurt or kill him/herself
		Looking for ways to kill him/herself by seeking access to firearms, available pills or other means
		Talking or writing about death, dying or suicide when these actions are out of the ordinary
S	Substance abuse	Increased substance (alcohol or drug) use
P	Purposelessness	No reasons for living; no sense of purpose in life
A	Anxiety	Anxiety, agitation, unable to sleep or sleeping all the time
T	Trapped	Feeling trapped – like there's no way out
H	Hopelessness	Hopelessness
W	Withdrawal	Withdrawing from friends, family and society
A	Anger	Rage, uncontrolled anger, seeking revenge
R	Recklessness	Acting reckless or engaging in risky activities, seemingly without thinking
M	Mood changes	Dramatic mood changes

5. Know the indications for hospitalization of a child or adolescent at risk of suicide

"Adolescents who have made previous attempts, exhibit a high degree of intent to commit suicide, show evidence of serious depression or other psychiatric illness, engage in substance use or have an active substance use disorder, have low impulse control, or have families who are unwilling to commit to counseling are at high risk and may require psychiatric hospitalization." (Shain B et al)

Consensus warning signs of suicide

Seek help immediately when you see one of these behaviors:

- Someone who threatens to kill or hurt themselves
- Someone means to kill themselves: medicines, weapons and other means
- Someone who speaks or writes about death or suicide

Seek help, without the need of immediate assistance, when you see someone that manifests itself:

- Complete loss of hope
- Anger, rage, a desire for revenge
- Imprudence and desire to carry out dangerous activities
- Feeling of being trapped, no way out
- Abuse of alcohol/drugs
- Isolation from friends, from family or from society
- Anxiety, agitation, insomnia or hypersomnia
- Excessive change in tone of the mood
- Absence of a reason to live, absence of purposes in life

6. Recognize the impact of suicide on peers and members of the family

Suicide-bereaved (SB) families report higher levels of rejection, shame, stigma, the need to conceal the loved one's cause of death, and blaming. Stigma may derive from a "societal perception that the act of suicide is a failure by the victim and the family to deal with some emotional issue." Stigma and shame are barriers to seeking help and receiving support.

Guilt and Blame

• Feelings of guilt often overlap with shame, compounding the sense of stigma. SB individuals often experience "intense guilt or feelings of responsibility for the death."
• Self-blame is particularly strong when the deceased is an individual's child.

Rumination and Anger

• Rumination is common in SB individuals and is unique compared with the responses of bereaved individuals to other losses.
• Suicide sometimes comes as a "total shock" to the survivors and coupled with rumination are feelings of rejection and abandonment. These feelings can lead to anger at the deceased, which can compound the guilt.

Complicated Grief and Depression

• Rumination contributes to complicated grief (CG), a "painful and debilitating condition…characterized by prolonged, acute grief and complicating psychological features such as self-blaming thoughts and excessive avoidance of reminders of the loss."
  - Conversely, instead of avoiding reminders of the deceased, some SB people may "spend long periods of time trying to feel closer to the deceased through pictures, keepsakes,

clothing, or other items associated with the loved one." Left untreated, CG can last for years, if not indefinitely.

• "In contrast to mourning, CG is a "chronic impairing form of grief brought about by interference with the healing process" that "derails" the mourning process and "prevents the natural healing process from progressing."

Mental and Physical Health Sequelae

• SB individuals are vulnerable to physical, psychological, and psychosomatic difficulties.
  - 1/4 of people bereaved by suicide experience elevated levels of depression and stress and close to one-fifth have elevated levels of anxiety, as well as posttraumatic stress disorder (PTSD) and impairment in social and employment settings.

• Survivors are themselves at high risk for suicidal thoughts or completed suicide.
  - A study of 3432 young adults who had lost close friends or family members to suicide found they had a higher probability of attempting suicide than individuals bereaved by deaths due to sudden, natural causes.

Impact on the Family Unit

• The suicide of a family member leaves an indelible mark on the survivors, affecting each individual, the family as a whole, and also larger social networks.
  - The suicide may affect family communication and the developmental processes of children.
  - Marital breakup is also more common in parents of children who died by suicide.

Support for the suicide bereaved (SB) families

• Complicated grief therapy (CGT), a manualized, structured, 16-session protocol, has been shown to be effective in treating CG in SB adults.
  -It includes self-regulation, focusing on aspirational goals, rebuilding connections, revisiting the story of the death, revisiting the world, and creating memories/continuing bonds.

• Education and information about support groups can be found at the American Foundation for Suicide Prevention (https://afsp.org/) and the American Association of Suicidology (https://www.suicidology.org/).

• Bereavement family counseling can facilitate the grieving process.

• Helpful activities might include rituals, ceremonies, lighting candles, reviewing pictures and mementoes, finding new information about the deceased person or even his/her death, and engaging in artistic expression.2 Religious and spiritual activities can be helpful to some people, as can engaging in regular physical activity, good nutrition, sleep hygiene, and "taking time out" from grief.

7. Plan the management of a child with suicidal ideation or behavior

- Patients should be medically stabilized first after suicidal behavior
- Patients with suicidal ideation or after medical stabilization should be admitted to the hospital if they meet the above criteria (see section 5)
- If the patient does not meet hospitalization criteria, does not have a specific plan or intent for suicide, outpatient management is indicated. A safety plan needs to be completed. Here is one example: https://suicidepreventionlifeline.org/wp-content/uploads/2016/08/Brown_StanleySafetyPlanTemplate.pdf
  Firearms and other weapons should be removed from the home. Prescription and over the counter medications should be locked up.

- Patients should be connected with mental health professionals and have same day or next day evaluation
- Consider starting an SSRI, but patients should have close follow up when starting an SSRI.

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Content Category 14- Substance Use Disorder

ABP Content Specs for Developmental-Behavioral Pediatrics Fellow Collaboration Project

Prepared by: Beatrice Egboh, MD, University of Nebraska, DBP Fellow

Reviewed by Howard Needelman, MD, University of Nebraska, DBP Fellowship Director

Final Version/Faculty Reviewed

Substance Use Disorder

A. General

1. Know the factors that are associated with substance use/abuse
2. Know the factors that are protective against substance use/abuse
3. Understand that early academic failure predisposes to adolescent social-emotional dysfunction, including substance use/abuse
4. Know surveillance and screening techniques for detection of substance use disorder in children and adolescents
5. Know the criteria for substance use disorder
6. Know the range of substance-induced disorders, including intoxication, withdrawal and other substance/medication-induced mental disorders
7. Know the ranges of severity of substance use, from mild to severe

B. Tobacco use

1. Know the factors that predict high risk for the onset of tobacco smoking in adolescents
2. Understand the major strategies for prevention of tobacco use in adolescents
3. Know how to plan a smoking cessation intervention directed at adolescents
4. Understand the natural history of tobacco use during adolescence

C. Alcohol use

1. Understand that familial tendencies toward violence or alcoholism may involve both genetic mechanisms and social learning
2. Know how to screen for excessive alcohol use in adolescents
3. Recognize the behavioral consequences of alcohol use in adolescents
4. Know the epidemiology of alcohol use among adolescents
5. Know the health and behavioral effects of alcohol use
6. Recognize the co-incidence of chronic alcohol use disorder with other health risks during adolescence (eg, tobacco, depression, sexual activity, school failure)
7. Recognize the signs and symptoms of increasing alcohol tolerance
8. Know how to plan an intervention for a child whose parent chronically abuses alcohol
9. Know the familial pattern of alcohol use disorder
10. Know how to counsel parents regarding prevention and early intervention of alcohol use disorder
11. Know the epidemiology of alcohol use disorder in adolescents
12. Know the natural history of alcohol use disorder in children and adolescents
13. Know the criteria for referral of adolescents for alcohol use disorder
14. Know the appropriate treatment for alcohol use disorder
15. Know office-based interventions for adolescents who are abusing alcohol (eg, rescue plans) 16. Identify the stages in the continuum of alcohol use disorder (eg, alcohol intoxication, alcohol tolerance, alcohol withdrawal)

D. Illicit drug abuse

1. Recognize the signs and symptoms of illicit drug use, including cannabis, cocaine, and prescription drugs
2. Recognize the common signs and symptoms of abuse of stimulant drugs
3. Recognize the common signs and symptoms of abuse of depressant drugs

4. Understand that there are problems associated both with the particular substance used and with the vehicle of use (eg, needles, sniffing, etc)
5. Differentiate between patterns of experimentation and evidence of addiction to illicit drugs
6. Understand the natural history of drug use disorders
7. Know the elements of treatment for an adolescent who is abusing illicit chemicals (eg, withdrawal, family therapy, group treatment, such as AA/NA)
8. Understand the use and limitations of urine and serum drug screens
9. Know surveillance and screening techniques for detection of substance use in adolescents

DSM 5 Features of Substance Use Disorder.

Addiction is defined as a chronic, relapsing disorder characterized by compulsive drug seeking and use despite adverse consequences.

A cluster of cognitive, behavioral and physiological symptoms indicating that the individual continues using the substance despite significant substance related problems.

It is considered a brain disorder, because it involves functional changes to brain circuits involved in reward, stress, and self-control, and those changes may last a long time after a person has stopped taking drugs[1]

DSM 5 Diagnostic Criteria

1. **Impaired Control**
    a. Taking the substance in larger amounts or over a longer period than intended.
    b. Unsuccessful attempts to cut down or decrease the substance use.
    c. Spending a lot of time getting, using, or recovering from its effects. (In severe use, virtually all of the individual's daily activity revolves around the substance).
    d. Craving (intense desire or urge for the drug).

2. **Social Impairment**
    a. Failure to fulfill major role obligations in school, home, work.
    b. Continued substance use despite having recurrent or persistent social or interpersonal problems.
    c. Reduced or giving up on important social, occupational, or recreational activities.

3. **Risky Use**
    a. Take the form of recurrent substance use in situations in which it is physically hazardous.
    b. Continued use despite having a persistent or recurrent physical or psychological problem known to be caused or exacerbated by the substance.

4. **Pharmacological Criteria**
    a. Tolerance
    b. Withdrawal

Severity – is based on the number of symptoms endorsed

no diagnosis- zero to one symptom
mild - two to three symptoms,
moderate - four to five symptoms
severe - six or more symptoms

Risk Factors Associated with Substance Use Disorder.

- Genetic,
- familial,
- social,
- environmental
- Parental use of alcohol or drugs or permissive attitudes about substance

- Patterns of use in the community
- Results are mixed
- urbanicity has been associated with higher rates of smoking, cannabis use
- and alcohol use
- substance use is associated with a low average socioeconomic status
- Peer influences do not predict the development of substance use problems or disorders, adolescents who use for social reasons are more likely to stop than are those who use for psychological reasons

Personality characteristics

- low assertiveness
- low self-esteem
- low social confidence
- external locus of control
- lower religiosity
- higher identification with counterculture
- higher impulsivity
- higher anxiety
- stronger need for peer approval

Age- the most powerful predictor of substance use disorders is younger age at first use.
The majority of adolescent substance abuse is initiated between 15 and 17 years of age but can begin as early as age 10 years[5].

Early life stress exposure has been associated with anxiety, depression, drug abuse, and schizophrenia[5]. The effects of early stress exposure emerge later in adolescence. Toxic stress includes physical/sexual abuse, neglect, loss of a caregiver, and exposure to a dysfunctional household.

Surveillance and Screening Techniques for Detection of Substance Use Disorder in Children and Adolescents.

According to the American academy of Pediatrics, screening for substance use should be part of routine adolescent health care visits especially those with behavioral, emotional and academic challenges.
The Substance Abuse and Mental Health Services Administration (SAMHSA) recommends universal screening with Substance use, Brief intervention, and/or Referral to Treatment (SBIRT) as part of routine health care[7]. Although the United States Preventive Services Task Force (USPSTF) state that there is insufficient evidence to recommend screening for unhealthy substance use in youth, new research lends support for effectiveness of Screening and Brief Intervention (SBI) in adolescents, the AAP recommends that pediatricians: increase their capacity in substance use detection, assessment, and intervention; and become familiar with adolescent SBIRT practices and their potential to be incorporated into universal screening and comprehensive care of adolescents in the medical home.

Discussion about and treatment can be kept confidential in general as preserving confidentiality may help to strengthen trust.[2]

The clinician should consider breaking confidentiality and engage the parents to ensure safety, the NIAAA also suggests engaging parents even against the minors wishes for any alcohol use by elementary school or alcohol related mild problems in middle school or significant problems in high school[2]

The assessment should also include a screening for co-occurring mental disorders, parent or sibling alcohol and drug use, and other risk behaviors such as illegal activities and sexual promiscuity

Screening in Adolescents.

Substance use, Brief intervention, and/or Referral to Treatment (SBIRT): Evidence based brief intervention to reduce and prevent problematic use, abuse and dependence on alcohol and illicit drug. It consists of 3 major components:

Screening — a healthcare professional assesses a patient for risky substance use behaviors using standardized screening tools. Screening can occur in any healthcare setting
Brief Intervention — a healthcare professional engages a patient showing risky substance use behaviors in a short conversation, providing feedback and advice
Referral to Treatment — a healthcare professional provides a referral to brief therapy or additional treatment to patients who screen in need of additional services

SBIRIT for Adolescents- usually assess patient self-reported information about substance use, and any health care professional can easily score the results involves the use of a brief one-three question prescreen

CRAFFT Screen.
A validated, reliable and easy to use survey.

Part A
During the PAST 12 MONTHS, did you:

1. Drink any alcohol (more than a few sips, do not count sips of alcohol taken during family or religious events.)
2. Smoke any marijuana or hashish?
3. Use anything else to get high? (“anything else” includes illegal drugs, over the counter and prescription drugs, and things that you sniff or “huff”)

If you get a Yes for any one of the above,
Continue to Part B

1. Have you ever ridden in a CAR driven by someone (including yourself) who was “high” or had been using alcohol or drugs?
2. Do you ever use alcohol or drugs to RELAX, feel better about yourself, or fit in?
3. Do you ever use alcohol or drugs while you are by yourself, or ALONE?
4. Do you ever FORGET things you did while using alcohol or drugs?
5. Do your FAMILY or FRIENDS ever tell you that you should cut down on your drinking or drug use?
6. Have you ever gotten into TROUBLE while you were using alcohol or drugs?

Each YES scores 1 point. A total score of 2 or higher is a positive screen, indicating a need for additional assessment. Also, children who screen negative should be praised and counselled on the continuation of their healthy behavior.
Probable substance addiction is indicated by red-flag findings, including a CRAFFT score of 2 or more in an adolescent aged 14 years or younger, daily or near-daily use of any substance, a CRAFFT score of 5 or higher, and alcohol-related blackouts (memory lapses)

Problem Oriented Screening Instrument for Teenagers (POSIT)- 139-item yes or no questionnaire developed by the National Institute on Drug Abuse, good reliability, easy to use in general outpatient setting.

- The National Institute on Alcohol Abuse and Alcoholism, in collaboration American Academy of Pediatrics, has encouraged using the following 2 questions, adapted for age, for screening for risk of alcohol-related harm:
  1. “Do you have any friends who drank beer, wine, or any drink containing alcohol in the past year?”
  2. “Have you ever had more than a few sips of beer, wine, or any drink containing alcohol?”

Know the criteria for substance use disorder-

Stages of Substance Use.
According to the *Diagnostic and Statistical Manual for Primary Care, Child and Adolescent Version* (DSM-PC), substance use occurs on a continuum from the “developmental variation” of experimentation, through “substance use problems,” to the disorders of abuse and dependence.
Abstinence: not yet using any substance.
Experimental use: tried substances usually from friends, may have experienced mild euphoria with return to normal baseline/ no serious adverse consequences. Voluntary and infrequent use. People are able to stop on their own.

Non-problematic use: intermittent continuing use of substance without negative consequences. Use in a regular/ predicted pattern.

Problematic/Risky Use: adverse consequence occurs as a result of use. May be associated with anxiety, guilt feeling. There is increase in frequency and quantity used.

Substance Abuse: maladaptive pattern of use, causing impairment in social functioning during a 12-month period without a diagnosis of dependence.

Substance Dependence: maladaptive pattern of use characterized by compulsive use, negative consequences, preoccupation and loss of control over use, tolerance and withdrawal.

Range of substance-induced disorders.
Intoxication: - is the result of being under the influence of, and responding to, the acute effects of alcohol or another drug[2].
These symptoms are due to the CNS effects of the drugs and cannot be attributable to another medical or mental health disorder.
Intoxication can mimic a variety of medical and mental health disorders.

It is characterized by altered mood states, psychomotor behavior, sensorium and perception, there may also be impaired judgement and function. Severity can range from mild euphoria to severe life -threatening emergencies depending on the substance.
Management depends on diagnosis and treatment.

Withdrawal- according to the WHO is defined as group of symptoms of variable clustering and degree of severity which occur on cessation or reduction of use of a psychoactive substance that has been taken repeatedly, usually for a prolonged period and/ or in high doses. They are usually the opposite of a substance pharmacological effects.
These cause significant clinical distress, impairment in social, educational or other important areas of functioning. Most individual have an urge to re-administer the substance to reduce symptoms.
Management aims to reduce the medical consequences; pain and craving from withdrawal.

Detoxification is the process of clearing toxins; detoxification programs are relatively short-term inpatient programs whose goal is the medical management of physiologic withdrawal symptoms.
The American Society of Addiction Medicine immediate goals for detoxification of alcohol and other toxic materials is

1. To provide safe withdrawal from the drugs of dependence and enable the individual become drug – free.
2. To provide withdrawal that is humane and thus protect the patient's dignity
3. To prepare the patient for his or her ongoing treatment of his or her dependence.

Substance/medication-induced mental disorders- these are CNS syndromes that develop due to the effects of substances of abuse, medication or toxins. They are usually temporal but sometimes persist despite discontinuation of the substance.
DSM 5 Criteria:

A. The disorder represents a clinically significant symptomatic presentation of a relevant mental disorder.

B. There is evidence from the history, physical examination, or laboratory findings of both of the following:
    1. The disorder developed during or within 1 month of a substance intoxication or withdrawal or taking a medication; and
    2. The involved substance/medication is capable of producing the mental disorder.
C. The disorder is not better explained by an independent mental disorder (i.e., one that is not substance- or medication-induced). Such evidence of an independent mental disorder could include the following:
    1. The disorder preceded the onset of severe intoxication or withdrawal or exposure to the medication; or
    2. The full mental disorder persisted for a substantial period of time (e.g., at least 1 month) after the cessation of acute withdrawal or severe intoxication or taking the medication. This criterion does not apply to substance-induced neurocognitive disorders or hallucinogen persisting perception disorder, which persist beyond the cessation of acute intoxication or withdrawal.
D. The disorder does not occur exclusively during the course of a delirium.
E. The disorder causes clinically significant distress or impairment in social, occupational, or other important areas of functioning.

Most substance/medication-induced mental disorders tend to improve relatively quickly with abstinence and unlikely to remain clinically relevant for more than 1 month after complete cessation of use.

There are 11 DSM 5 subclassifications of Substance Use Disorders:

1. Substance-induced depressive disorder
2. Substance-induced anxiety
3. Substance-induced sleep disorders
4. Substance-induced delirium
5. Substance-induced amnestic disorder
6. Substance-induced neurocognitive disorders
7. Substance-induced obsessive-compulsive and related disorders
8. Substance-induced psychotic disorder
9. Substance-induced bipolar and related disorders
10. Hallucinogen persisting perception disorder
11. Substance-induced sexual dysfunctions

Tobacco Use.

Tobacco product use is started and established primarily during adolescence[8]. Nearly 9 out of 10 cigarette smokers first try cigarette smoking by age 18.
Teenagers who smoke are 3 times more likely than nonsmokers to use alcohol, 8 times more likely to use marijuana, and 22 times more likely to use cocaine.
Nicotine addiction is not just a bad habit but a chronic disease.

Flavoring in tobacco makes it more appealing to youths. The number of middle and high school students using e-cigarettes rose from 3.6 million in 2018 to 5.4 million in 2019[9].

According to the CDC, in 2019, tobacco use among high school students

- Any tobacco product: 31.2%
- E-cigarettes: 27.5%
- Cigars: 7.6%
- Cigarettes: 5.8%
- Smokeless Tobacco: 4.8%
- Hookah: 3.4%
- Pipe tobacco: 1.1%

Natural History of Tobacco Use During Adolescence.
Nicotine is the addictive substance in tobacco, it activates the brain rewards system involved in pleasurable activities. Tobacco addiction usually begins in adolescence, with experimenting. Use may also be a means of self-treating mental health disorder. Children with at least one smoking parent are twice as likely to experiment compared with others whose parents do not smoke. This may be due to the priming or the nicotine receptors in utero or due to secondhand smoke. Polymorphic genes have been hypothesized to be involved in the risk of dependence including genes affecting dopamine receptors and nicotine metabolism. Symptoms of dependence may be evident after a short-term use, and adolescents who smoke only a a few cigarettes per month can suffer physical and psychological withdrawal. The addictive nature and unpleasant withdrawal symptoms (anxiety, irritability, restlessness, dizziness, sweating) make quitting difficult.
Tobacco is seen as the gateway to use of other substances, teenagers who smoke are more likely than a non-smoker to use other substances.

- Adolescent smokers are more likely to become nicotine dependent through smoking fewer cigarettes a day than are adult smokers.
- Adolescents see the positive aspects of smoking as helping with boredom, dealing with stress, staying thin, appearing more mature
- The negative aspects adolescents see include yellow teeth, interfering with playing sports, being harder to quit, bad breath.
- Nicotine's effect on the brain takes < 20 seconds (nicotinic acetylcholine receptors).

Factors That Predict High Risk for Onset of Tobacco Smoking in Adolescents.

Population.

- Populations found to be at higher risk for tobacco use include American Indian and Alaskan native ethnic groups, sexual minorities.

Biological and genetic factors.

- Maternal smoking in pregnancy (hypothesized role of nicotine receptor priming as a result of being exposed to nicotine in utero and secondhand smoke after birth).
- It is more difficult for teens to quit smoking compared to adults
- Adolescents are more likely to feel dependent on nicotine sooner than adults

Social and physical environments.

- Adolescents are more likely to smoke when they see friends smoking.
- Parents smoking: Teenagers with at least 1 smoking parent are twice as likely to become smokers compared with teenagers whose parents do not smoke.
- mass media/ entertainment presenting tobacco product use as a normal activity
- Lower socioeconomic groups,
- Lower educational attainment.
- Adolescents using e-cigarettes are more likely to use cigarette.

Mental health:

- Those with mental health disorders There is a strong relationship between youth smoking and depression, anxiety, and stress.[2]

Personal:

- Low self-esteem.
- Poor academic performance
- Poor will power to say no.
- When young people expect positive things from smoking, such as coping with stress better or losing weight, among adolescent girls a relatively consistent association between having higher body weight or concerns about weight and a greater likelihood of smoking
- Lesbian, gay, bisexual, and transgender youth are significantly more likely than heterosexual youth to engage in high-risk behaviors including tobacco use.

Protective Factors.

- Community programs and school and college policies that encourage tobacco-free places and lifestyles
- Community programs that lower tobacco advertising, promotions, and help make tobacco products less easily available
- Being part of a religious group
- Racial/ethnic pride and strong racial identity

- Higher academic achievement

Major Strategies for Prevention of Tobacco Use in Adolescents.
AAP Public Policy Statement.

1. **The FDA should regulate all tobacco products to protect the public health**
    a. **Tobacco control should be adequately funded including** treatment and research programs.
2. TV and radio commercials, posters, and other media messages aimed at kids and teens in order to counter tobacco product ads[2].
3. **Tobacco product advertising and promotion in forms that are accessible to children and youth should be prohibited.**
4. **Point-of-sale tobacco product advertising and product placement that can be viewed by children should be prohibited.**
5. **Depictions of tobacco products in movies and other media that can be viewed by youth should be restricted**
6. **The promotion and sale of electronic nicotine delivery systems to youth should be prohibited.**
7. **Tobacco control programs should change the image of tobacco by telling the truth about tobacco**
    a. Tobacco industry should be excluded from development and implementation of control measures as their programs has been shown to be ineffective and counterproductive.
8. **Tobacco product prices should be increased to reduce youth tobacco use initiation.**
    a. **Mandating minimum package size**
    b. **Taxes should escalate with inflation**
    c. **Ban free products, coupon or samples**
9. **The minimum age to purchase tobacco should be increased to 21 years.**
10. **Flavoring agents, including menthol, should be prohibited in all tobacco products.**
11. **Comprehensive smoking bans should be enacted.**
    a. Prohibiting smoking in indoor areas of workplaces and public places

    b. Incorporate e-cigarettes into current tobacco-free laws and ordinances where children and adolescents live, learn, play, work, and visit.
12. **Smoking in multi-unit housing should be prohibited.**
    a. This involuntarily exposes those in nearby units.
13. **Prohibitions on smoking and use of tobacco products should include prohibitions on use of electronic nicotine delivery systems.**
14. **Children younger than 18 years should be legally prohibited from working on tobacco farms and in tobacco production**
15. **Concentrated nicotine solution for electronic nicotine delivery systems should be sold in child-resistant containers with amounts limited to that which would not be lethal to a young child if ingested.**

AAP Policy Statement (2015) Recommended Actions for Pediatricians.

1. Inquire about tobacco use and exposure during all health supervision visits and visits for diseases caused or exacerbated by tobacco.
2. **Include tobacco use prevention as part of anticipatory guidance beginning at age 5. Message should be clear, personally relevant and age appropriate.**
3. **Address parent/caregiver tobacco dependence as part of pediatric health care. Recommend tobacco dependence treatment of tobacco-dependent parents and caregivers and implement systems to identify and offer referral, counselling and treatment.**
4. **Offer tobacco dependence treatment and/or referral to adolescents who want to stop smoking, pharmacotherapy can be considered for moderate to severe tobacco dependent adolescents.**
5. **Offer tobacco-dependent individuals Quitline referral.**

Pharmacotherapy.

Research is Limited in adolescents due to high rate of non- adherence and relapse after discontinuation. Medications are not FDA approved for children younger than 18.

1. Bupropion.
2. Varenicline: It decreases cravings and pleasurable effects of nicotine.
3. Nicotine Replacement Therapies- Nicotine gum, patch, Nasal sprays inhalers, Lozenges.

Smoking Cessation Intervention Directed at Adolescents.

The 6 A's for Brief Intervention to Treat Tobacco Dependence

1. Anticipate risk of tobacco use
2. Ask about tobacco use
3. Advise to quit
4. Assess willingness to make a cessation attempt
5. Assist in cessation attempt
6. Arrange follow-up

Alcohol Use in Adolescents.

Adolescence “a critical risk period for initiation of alcohol” as areas of the brain that control impulsivity have not fully developed[13]

An individual’s tendency toward alcohol-induced aggression depends not just on neurobiological factors, but also on personal expectations of the effects of alcohol, on prior experience of violent conflicts, and on the environmental conditions of early childhood, especially social exclusion and discrimination. Gene–environment interactions affecting the serotonergic and other neurotransmitter systems play an important role.

According to the National Institute on Alcohol Abuse and Alcoholism (NIAA), 1 in 3 children starts drinking by the end of 8th grade and of them half report having been drunk.

Alcohol is usually the first risky behavior tried, it is most strongly linked to violence among adolescents, it is also a marker for other unhealthy behaviors (drinking, smoking, illicit drugs, unprotected sex). It contributes to more deaths than all other substances combined.
Children of alcoholic parents – threefold to nine-fold increased risk for alcoholism.
Genetic influences for the predisposition: family, twin, adoption studies.
Rates of drinking and alcohol-related problems are highest among White and American Indian or Alaska Native youth, followed by Hispanic youth, African Americans, and Asians

Risks.

- Young people who are disruptive, hyperactive, aggressive, have conduct problems
- Depressed, withdrawn, or anxious.
- Youths who smoke cigarettes.
- Child of an alcoholic or having several alcoholic family members places a person at greater risk for alcohol problems.
- An adolescent girl with an older or adult boyfriend is more likely to use alcohol and other drugs and to engage in delinquent behaviors.
- Many youths want to assert their independence and try alcohol, many have access through family members/ find at home.
- Peer pressure.
- Stress.
- Adolescents appear to be particularly sensitive to the positive effects of drinking, such as feeling more at ease in social situations, and young people may drink more than adults because of these positive social experiences.

Binge Drinking.
People ages 12 through 20 drink 11 percent of all alcohol consumed in the United States, although they drink less often, they consume more than 90% of their alcohol by binge drinking.
Children have a higher blood alcohol concentration after drinking smaller amounts of alcohol.
Number of drinks for binge drinking

- Girls (9-17 years)3 drinks
- Boys (9-13 years) 3 drinks
- Boys (14-15 years) 4 drinks
- Boys (16+) 5 drinks

Danger of Underage Drinking.
Death (homicide, vehicle crashes, alcohol poisoning, falls, burns, drowning, suicides)
Injury / MVA
Impaired Judgement

In the short term, adolescent drinking:

- Primarily acts as a CNS depressant.
- Cause Euphoria, grogginess, talkativeness; impairs short term memory; increases pain threshold
- Lowers inhibition, impairs judgement resulting in unintentional injuries and death; suicidality;
- aggression and
- victimization;

- infections
- pregnancies from unplanned, unprotected sex
- academic (missing school, lower grades) and social problems
- vasodilation and hypothermia.
- Inhibitory effect on pituitary antidiuretic hormone release (diuresis).
- Erosive gastritis, pancreatitis

Long term, drinking in adolescence is associated with
- increased risk for alcohol dependence later in life.
- Lasting functional and structural changes in the brain.
- Coordination problems.

The DSM-5 can be used to specifically query for 11 alcohol abuse criteria, and the presence of ≥2 of these criteria indicates an Alcohol use disorder. The DSM 5 is considered the gold standard for diagnosis.

1. Had times when you ended up drinking more, or longer than you intended?
2. More than once wanted to cut down or stop drinking, or tried to, but couldn't?
3. Spent a lot of time drinking? Or being sick or getting over the aftereffects?
4. Experienced craving — a strong need, or urge, to drink?
5. Found that drinking — or being sick from drinking — often interfered with taking care of your home or family? Or caused job troubles? Or school problems?
6. Continued to drink even though it was causing trouble with your family or friends?
7. Given up or cut back on activities that were important or interesting to you, or gave you pleasure, in order to drink?
8. More than once gotten into situations while or after drinking that increased your chances of getting hurt (such as driving, swimming, using machinery, walking in a dangerous area, or having unsafe sex)?
9. Continued to drink even though it was making you feel depressed or anxious or adding to another health problem? Or after having had a memory blackout?
10. Had to drink much more than you once did to get the effect you want? Or found that your usual number of drinks had much less effect than before?
11. Found that when the effects of alcohol were wearing off, you had withdrawal symptoms, such as trouble sleeping, shakiness, irritability, anxiety, depression, restlessness, nausea, or sweating? Or sensed things that were not there?

National Institute on Alcohol Abuse and Alcoholism (NIAAA) 2-question screen - is a brief and valid adolescent alcohol screening tool used to ask about a patient's drinking frequency and their friends' drinking.

Friends' drinking question is an early warning signal that strongly predicts the patient's future drinking levels and a nonthreatening "side-door" entrance to begin talking about alcohol with younger patients in particular, for high school students, having friends who binge drink heightens concerns.
Personal drinking question zeroes in on frequency, the best predictor of current risk for alcohol-related harm in adolescents who are already drinking.

It that can accurately characterize an adolescent for future Alcohol use disorder.

The questions are age specific (for elementary, middle and high school)

Elementary and Middle School:

1. "Do you have any friends who drank beer, wine, or any drink containing alcohol in the past year?"
2. "Do you have any friends who drank beer, wine, or any drink containing alcohol in the past year?"

High School:

1. "In the past year, on how many days have you had more than a few sips of beer, wine, or any drink containing alcohol?"
2. "If your friends drink, how many drinks do they usually drink on an occasion?"

Signs of Alcohol Use

1. Have health problems that might be alcohol related, such as: -
    a. Accidents or injury –
    b. Changes in eating or sleeping patterns
    c. sexually transmitted
    d. Gastrointestinal disturbances infections
    e. Unintended - chronic pain
    f. Pregnancy
2. Show substantial behavioral changes, such as: -
    a. Increased oppositional behaviors
    b. Change of friends' behavior
    c. A drop-in grade point average
    d. Significant mood changes
    e. Large number of unexcused / loss of interest in activities school absences

Stages of Alcohol Use.

Abstinence – child does not use any alcohol.

- -Be aware of children and adolescents at risk
- Encourage and support (positive reinforcement)
- Discuss what child would do if there is peer pressure (patient & parent education)

Experimental Use (Substance use without a disorder) – infrequent use. Infrequent use in social situations, without related problems, typically, use occurs at predictable times (e.g., weekends)

- Educate about potential consequences.
- Develop a "**rescue plan**" with the child or adolescent and parents. A rescue plan should specify that the child or adolescent will receive a ride home if he finds himself in an unsafe situation, including being intoxicated or high, and the commitment that discussion about the behavior will take place at a time when it can be rational.
- Advise the patient to stop.

Regular Use- uses alcohol on a regular basis.

- Counsel adolescent on consequences of alcohol use.
- Rescue plan.
-

Mild to moderate substance use disorder – used in high risk situations, such as when driving or with strangers; use associated with a problem (fight, arrest, or school suspension, use for emotional regulation (relieve stress or depression) – defined as meeting 2-5 of the 11 criteria for a substance use disorder in the DSM5

-Brief assessment to explore patient-perceived problems associated with use
-Give clear, brief advice to quit
-Provide counseling regarding the medical harms of substance use
-negotiate a behavior change to quit or cut down
-Provide close patient follow-up
-consider referral to substance use disorder treatment

Severe substance use disorder – Loss of control or compulsive drug use associated with neurological changes in the reward system of the brain; defined as meeting 6 or more of the 11 criteria for a substance use disorder in DSM-5

-As above
-Involve parents in treatment planning whenever possible
-Refer to the appropriate level of care
-Follow-up to ensure compliance with treatment and to offer continued support

Problem Use- experienced adverse consequences associated with use

- Discuss concerns and options for change.
- Consider '**abstinence challenge'.**
- Develop rescue plan.

Substance Abuse- Child or adolescent engages in ongoing use of alcohol, despite harm.

- Loss of control over use.
- Continue to work with the child or adolescent and family until the child or adolescent
- is ready to engage in substance abuse treatment
- Refer to formal treatment program

Substance Dependency- Child or adolescent is preoccupied with use.

- Development of tolerance or withdrawal symptoms.
- Refer to formal treatment program

Secondary Abstinence is the Goal of treatment

- Continue to monitor closely
- Relapse is part of early recovery
- Avoid Stigmatization.

Parents in prevention and early intervention of alcohol use disorder.

- Parents' have the ability to influence whether their children drink across all racial and ethnic groups.

- Talk to children about dangers of drinking.
- Setting and enforcing clear rules.
- Drink responsibly
- Role model
- No alcohol available
- Regular conversations about life
- Know children's friends
- Supervising Parties
- Encourage kids to participate in healthy and fun activities.

Treatment.

1. Medication (The safety and efficacy not established in pediatrics)
    a. Naltrexone - blocks the euphoric effects and feelings of intoxication
    b. Acamprosate- reduces the desire to drink alcohol.
    c. Disulphiram - producing an acute sensitivity to ethanol
2. Cognitive-behavioral therapy
3. Support Groups- AA

- In the treatment of adolescents with alcohol use disorders, the greatest number of studies have supported a family therapy component to be the most efficacious, although cognitive behavioral therapy, motivational enhancement therapy, and multisystemic therapy have demonstrated some efficacy.
- The vast majority of treatments for alcohol use disorders in adolescents are psychosocial based treatments. There are limited numbers of studies of pharmacologic treatments.
- The common goals of family therapy consist of psychoeducation of the disorder, assisting initiation and effort into obtaining treatment, establishing structure, limit-setting, monitoring, improving communication amongst family members, and ensuring that family members seek their own treatment and support.
- The primary goal of treatment of adolescents with alcohol use disorder should be achieving and maintaining abstinence from the use of alcohol; however, harm reduction may be the initial goal.

Illicit drug Use.

Cannabis.

- Marijuana is the most commonly used substance among adolescents after alcohol.
- Use among adolescents has increased in the past 10 years.
- Fewer adolescents believe that marijuana use is a threat than in the past.
- **Vaping** is a popular and relatively new way to use marijuana and other substances.
- Smoking, vaping, eating, or dabbing (smoking in form of harsh oil/wax).
- Tetrahydrocannabinol (THC) is the active ingredient in marijuana.

Effects:

- Addictive
- Withdrawal symptoms: restlessness, decreased appetite, mood disorder, disrupted sleep.
- Driving under the influence of marijuana can also lead to injury or death
- Mood swings
- Distorted sensory perception
- Increased appetite
- Dilated pupils.
- Declining academic performance
- Careless with grooming/
- Changes with sleeping habits
- Health effects: testicular cancer, heart attacks, respiratory disease, a weakened immune system, pregnancy complications, and low birthweight
- cognitive problems; low academic achievement and other educational outcomes; impaired social functioning; and mental health disorders, including depression and anxiety
- **Cannabis hyperemesis syndrome:** recurring episodes of intractable nausea, vomiting and abdominal pain that occurs after long term use of cannabis.
  Episodes typically last 24-48 hours.
  Resolves with discontinuation of cannabis/ marijuana and recurs when intake resumes.

Cocaine.

- Highly addictive
- Crack, Snow, Coke, Blow, flake,
- Fine white Powder/ Solid rock.
- Snorted or injected
- Dilated pupils
- Restlessness and/or high energy
- Insomnia
- A runny nose and nosebleeds
- A hoarse voice
- Weight loss
- An increase in anxiety, depression, panic attacks,
- Paranoia
- violent behavior
- Seizures, cardiac arrest, strokes.

Inhalants.

- Rapid action, easy availability, low cost.
- Includes toluene (gasoline, paint thinners), freon (refrigerant), amyl nitrite, benzene
- Sniffing, huffing, snorting, bagged.
- Effects
    - Produces a high
    - Euphoria
    - Lightheadedness
    - Hallucination and delusions
- Complications

- Asphyxiation: fumes displacing O2, suffocation (air blocked when inhaling from plastic bag)
- Headaches,
- syncope, hypotension,
- Vasoconstriction, cutaneous flushing
- Arrhythmia, Tachycardia, EKG changes.
- Methemoglobinemia
- Increased bronchial irritation
- Increased intraocular pressure
- Dermatitis
- Pulmonary edema/ pulmonary hypertension,
- Seizures, cerebral or cerebellar atrophy.
- lack of coordination, disruptive behavior, hallucinations
- muscle weakness
- Bone marrow suppression

- Treatment – supportive

Prescription drug abuse.

Most common: opioid, anti-anxiety medications, sedatives and stimulants.
Symptoms depend on the medication used.

Stimulant drugs.

- Methamphetamines/ amphetamines.
- Binge and Crash (rapid high and fades quickly)
- Increased wakefulness and physical activity
- Decreased appetite
- Increased temp, BP, HR
- Tremors
- Dry mouth
- Euphoria: Stimulant misuse can create feelings of euphoria, because stimulants increase the levels of available dopamine in the brain.
- Cognitive problems
- Weight loss
- Anxiety
- Violent behavior
- Paranoia
- Meth mouth
- Hallucinations
- Individuals who misuse stimulant medications can develop a stimulant use disorder in as little as a week. Tolerance may follow. The use of higher doses can result in aggressive behaviors, anxiety similar to a panic disorder, generalized anxiety, and paranoia.
- Short-acting stimulants are more commonly misused, especially for their euphoric properties, when compared to extended-release stimulants and prodrugs. The euphoric properties of short-acting

stimulants are caused by their earlier peak drug concentration, which results in more immediate dopamine blockade. However, extended-release medications may also be misused for performance enhancement.

* Treatment of attention-deficit/hyperactivity disorder with stimulants does not increase risk for substance use disorders; rather, the literature suggests a protective effect.

Anti-anxiety / CNS Depressants.

Benzodiazepines (alprazolam/ clonazepam/ diazepam)
Non – Benzodiazepine Sedatives (Zolpidem/ Zaleplon)
Barbiturates (Phenobarbital, mephobarbital)

- Drowsiness
- Confusion
- Dry Mouth
- Slurred speech
- Poor concentration
- Dizziness
- Problems with memory
- Slowed breathing
- Long term use – Dependence, withdrawal, tolerance.

Opioids.

Prescription Opioids – Oxycodone, Hydrocodone, Codeine, Morphine, Fentanyl
Non-prescription – Heroin

- 'Euphoria'
- Short term effect
- Nausea/ Vomiting
- Dry mouth
- Slowed breathing rate
- Drowsiness
- Long term
- Constipation
- Depression
- Antisocial personality Disorder
- Worsening or increased sensitivity to pain with higher doses (hyperalgesia)

Opioid Medication/Treatment.

- Lofexidine α2a adrenergic receptor agonist for treatment of Opioid withdrawal symptoms (safety and efficacy have not been established in children or adolescents 17 years of **age** and younger).
- Buprenorphine / Naloxone
- Methadone
- Naltrexone

New Psychoactive Substances (NPS)

Unregulated analogues of drugs designed to mimic its pharmacological effect of illegal drugs and not detected easily by routine screen.
Targets the younger population.

Rohypnol/ Flunitrazepam (Forget Me Pill, La Rocha, Lunch Money Drug)

- Date rape drug
- Amnesia/ sleep
- Muscle relaxant
- Anxiolytic
- Impaired mental function
- Addictive.

K-2/ Spice/ Synthetic THC.

- Synthetic cannabinoids /may be more powerful than marijuana and unpredictable
- Falsely marketed as safe legal alternative to marijuana.
- Elevated mood,
- Altered perception
- Anxiety
- Paranoia
- Elevated troponins/ EEG changes

MDMA (Ecstasy/Molly)

- Psychoactive (stimulant +hallucinogen)
- Increased energy
- Distortion of perception
- Increased energy
- Hyperthermia
- Impulsivity and aggression
- Decreased appetite
- Loss of appetite
- Depression

Hallucinogens.

- 'Acid', 'angel dust', and 'vitamin k'
- Potent hallucinogen/ altered perception (time, colors, sounds, motion)
- Bizarre behaviors- unreal and frightening sensations.
- Extreme mood changes

Problems associated both with the particular substance used and with the vehicle of use.

Intravenous:

- Infections: cutaneous, abscesses, phlebitis, bacteremia, endocarditis, hiv, hepatitis b/c.
- Scarring / needle tracks
- Overdose risk.

Inhalation/ smoking

- Nasal septum perforation (cocaine vasoconstriction)
- Pulmonary edema (heroin)
- Hemoptysis
- e-cigarette or vaping product use-associated lung injury (evali)
- Diffuse alveolar hemorrhages (crack lung)
- Alveolar damage (cocaine vasoconstriction)
- Opioid induced anaphylactoid reaction
- Septic emboli of pulmonary origin

Ingestion.

- Parotid Gland enlargement.
- Gastritis/Peptic ulcer.
- Mallory Weiss Syndrome.

Referral to Treatment.

Substance Abuse and Mental Health Services Administration (SAMHSA) has estimated that fewer than 10% of adolescents in need of specialty substance use treatment receive it and majority of referrals are from the justice system. Adolescents should be treated in the least restrictive environment that supports their clinical needs.

Prochaska Stages of Change.

This model describes the stages people go through on their way to new and healthier behavior.

- **Precontemplation:** Not yet beginning to consider change.
- **Contemplation:** Beginning to understand that there is a problem.
- **Determination:** Making a clear decision to change. A quit day may be set. Individual may struggle with urges.
- **Action:** evidence of actual change
- **Maintenance:** able to sustain behavior; may lead to termination or relapse.

Treatment Options for Substance Use Disorder.

Individual counselling: Specific individualized therapy including motivational interviewing, cognitive behavioral therapy, contingency management

1. Group Therapy: cost-effective and takes advantage of the developmental preference for congregating with peers

2. Family Therapy: best-validated approach for treating adolescent SUDs. It targets domains that figure prominently in the etiology of SUDs in adolescents: family conflict, communication, parental monitoring, discipline, child abuse/neglect, and parental SUD.

3. Intensive Outpatient Treatment: Intermediate level of care. It comprises of a combination of support group therapy, educational, family therapies, case mgt and after – care. 2-9 hours/ day; 2-5 days per week lasting for 1- 3 months.

4. Partial Hospital Program: Short term comprehensive outpatient program in affiliation with an inpatient program, 7-8 hours / day, at least 5 days per week and lasting for 1-3 weeks.

5. Inpatient/ Residential Programs:
    a. Detoxification: Medical management of withdrawal.
    b. Acute Residential Treatment: short term, for patient in crisis.
    c. Residential Treatment: Structured 24-hour care severe SUD, mental illness, or behavioral problems that require 24-hour care. short-term programs of ≤30 days
    d. long-term programs >30 days.
    e. Therapeutic boarding schools: Educational institutions that provide constant supervision for their students by professional staff. These schools offer a highly structured environment with set times for all activities, smaller, more specialized classes, and social and emotional support.

Drug Screening.

Immunoassays are common qualitative tests for drug exposure. They are inexpensive, noninvasive, and rapid. However, determination of exact timeline, route of exposure, and level of intoxication is difficult with urine immunoassays. Each category of drug has variable durations of detection after exposure, and has potential false-positive and -negative results[6]

There is a potential for cross reactivity with similar antigens.

A screening method may be more reactive with one drug than another in the same group.

Basic screening tests includes amphetamines, cocaine, marijuana, opioids, phencyclidine, benzodiazepines.

Many synthetic substances used are not routinely screened for and may thus have limited use.

Common Drug Classes Found in Urine Drug Screening Immunoassays and Testing Characteristics

Urine Drug Screening Categories	Duration of Detection[a]	False Positives and Negatives[b]
Amphetamines	1-4 days	Can cross-react with decongestants, ADHD medications, Methamphetamines, MDMA, ephedrine, bupropion
Barbiturates	1-4 days	Phenobarbital can be detected for 3-4 weeks
Benzodiazepines	1-4 days	Can have false negatives for lorazepam, alprazolam, midazolam, and clonazepam. Diazepam can be detected for 3-4 weeks
Cocaine (benzoylecgonine)	2-4 days	Do not commonly occur
Opiates	1-4 days	False negatives can occur with synthetic and semi-synthetic opioids (oxycodone, hydromorphone, fentanyl, etc.)
Phencyclidine	1-8 days	Can cross-react with ketamine, dextromethorphan, diphenhydramine.
Marijuana (THC metabolites)	7-30 days	Reported false positives include dronabinol, efavirenz, proton pump inhibitors, nonsteroidal anti-inflammatory drugs

ADHD=attention-deficit/hyperactivity disorder; MDMA=3,4-methylenedioxymethamphetamine; THC=tetrahydrocannabinol.

a Detection period varies and depends on chronicity of exposure.

b This is not an all-inclusive list of potential false positives/cross reactants and false negatives.

Wang, G. S., & Hoyte, C. (2018, August 1). Common Substances of Abuse. https://pedsinreview.aappublications.org/content/39/8/403

Mass Spectrometry + Chromatography.
Confirmatory
Has greater specificity

Screening tests should never be considered as proof a drug is present, at same time a negative test should not rule out presence of a drug.

Choice of Matrix.
Should depend on drug of interest, desired window, subversion of patient and available resources.

- Urine: simple; noninvasive; wider window of detection. Limitation (unobserved collection).
- Blood and breath: the earliest and shortest windows of detection for substances
- Oral fluid
- Sweat
- Hair: longest window
- Meconium (wide window)

Indications for Drug Screen.

- Drug treatment Programs
- Seizure/ Status epilepticus
- Cardiovascular event
- Acute psychosis

Screening Techniques for Detection of Substance Use In Adolescents

- S2B1 Single frequency of use question per substance
- CAGE (Cut down, Annoyed, Guilty, Eyeopener) >16 years
- CRAFFT (Car, Relax, Alone, Forget, Family or Friends, Trouble) for adolescents, quickly assesses for problems with Substance use
- NIAAA 2-Question Screen for adolescents.
- The Alcohol, Smoking and Substance Involvement Screening Test (ASSIST): not validated for adolescents <18.
- Alcohol Use Disorders Identification Test (AUDIT) can be used for adolescents ages 14 to 18
- Drug Abuse Screening Test- Adolescents (DAST-A) 28 item.
- GAIN (Global Appraisal of Individual Needs) screens both SUD and Mental health disorders.

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Content Category 15- Child Abuse and Neglect

ABP Content Specs for Developmental-Behavioral Pediatrics Fellow Collaboration Project

Prepared by: Devina Savant, MD, Brown/Hasbro Children's- DBP Fellow, Dalal Elsori- DBP Fellow, Starrina Gianelloni- DBP Fellow, & Stephanie Klees- DBP Fellow, Rene Bartos- DBP Fellow

Prepared by: Matthew Scott, MD, Madigan Army Medical Center- DBP Fellow

Reviewed by: Carrie Kelly, MD, MPH, Brown/Hasbro Children's – Staff/Faculty DBP

15. Child Abuse and Neglect

A. Physical abuse

1. Know the parental risk factors associated with physical abuse of young children (eg, stress, isolation, parental abuse, substance abuse, poverty)
2. Know the child risk factors that predispose to physical abuse (eg, prematurity, disability, irritability, male gender)
3. Describe common screening techniques to identify children at risk of physical abuse
4. Recognize signs and symptoms of physical abuse
5. Understand the legal and clinical implications of reporting physical abuse
6. Recognize characteristics of fractures caused by physical abuse
7. Know the signs and symptoms of abusive head trauma
8. Know the appropriate management for a child suspected of having been abused
9. Know the advantages and disadvantages of "family preservation" vs removal of the child from the home in the face of repeated serious physical abuse
10. Know the components of effective programs for the prevention of child abuse
11. Know the long-term outcome of physical trauma

B. Sexual abuse

1. Know the risk factors for sexual abuse
2. Recognize physical signs and symptoms of sexual abuse
3. Recognize psychological symptoms of sexual abuse
4. Know appropriate interviewing techniques for assessing possible victims of sexual abuse
5. Know how to plan the management of a child who has been sexually abused
6. Know the long-term outcomes of childhood sexual abuse
7. Know the epidemiology, including the most common perpetrators, of sexual abuse
8. Know the components of effective school-based programs to prevent sexual abuse
9. Recognize the heightened risk of individuals with developmental disabilities to become victims of sexual abuse
10. Know how to evaluate a child for possible sexual abuse

C. Psychological abuse

1. Know the risk factors for psychological abuse
2. Recognize the signs and symptoms of psychological abuse
3. Know how to plan the management of a child who has been psychologically abused
4. Know the long term outcomes including impact on somatic health, mental health, and future parenting of own children

D. Factitious disorder imposed on another (Munchausen syndrome by proxy)

1. Recognize signs and symptoms suggestive of factitious disorder imposed on another
2. Know family risk factors often seen in cases of factitious disorder imposed on another
3. Know how to plan the management of cases of factitious disorder imposed on another
4. Know how to diagnose factitious disorder imposed on another

E. Child neglect

1. Know common developmental and behavioral sequelae of chronic neglect
2. Understand how the developmental and behavioral symptoms of neglected children vary with stages of development

3. Know the legal definition of child neglect
4. Know how to plan the management of chronic neglect
5. Know the parental risk factors associated with child neglect
6. Recognize child neglect as the most common form of child maltreatment
7. Know the child risk factors that predispose to child neglect
8. Describe interventions that can lower the risk of child neglect (eg, home nurse visits)

CHILD ABUSE AND NEGLECT

A. Physical abuse

1. Know the parental risk factors associated with physical abuse of young children (eg, stress, isolation, parental abuse, substance abuse, poverty)

Individual Risk Factors

- Parents' lack of understanding of children's needs, child development and parenting skills
- Parental history of child abuse and or neglect
- Substance abuse and/or mental health issues including depression in the family
- Parental characteristics such as young age, low education, single parenthood, large number of dependent children, and low income
- Nonbiological, transient caregivers in the home (e.g., mother's male partner)
- Parental thoughts and emotions that tend to support or justify maltreatment behaviors

Family Risk Factors

- Social isolation
- Family stress, separation or divorce, and violence, including intimate partner violence
- Parenting stress, poor parent-child relationships, and negative interactions

Community Risk Factors

- Community violence
- Concentrated neighborhood disadvantage (e.g., high poverty, high unemployment rates, and high density of alcohol outlets), and poor social connections.

Other relevant information:

Protective Factors for Child Abuse and Neglect

Protective factors may lessen the likelihood of children being abused or neglected. Identifying and understanding protective factors are equally as important as researching risk factors.

Family Protective Factors

- Supportive family environment and social networks
- Concrete support for basic needs
- Nurturing parenting skills
- Stable family relationships
- Household rules and child monitoring
- Parental employment
- Parental education
- Adequate housing
- Access to health care and social services
- Caring adults outside the family who can serve as role models or mentors

Community Protective Factors

- Communities that support parents and take responsibility for preventing abuse

2. Know the child risk factors that predispose to physical abuse (eg, prematurity, disability, irritability, male gender)

Child factors

· Emotional/behavioral difficulties

- Chronic illness
- Physical disabilities
- Developmental disabilities
- Preterm birth
- Unwanted
- Unplanned
- Some normal developmental phases that may cause difficulty for some parents, specifically colic, awakening at night, separation anxiety, normal exploratory behavior, normal negativism, normal poor appetite, and toilet-training resistance.
- Frequent crying

3. Describe common screening techniques to identify children at risk of physical abuse

It is imperative that pediatricians play an active role in screening children for abuse and neglect at every clinical encounter. Universal screening means assessing everyone; selective screening indicates only those who are 'high risk'.

Some examples of screening techniques:

1. Self-administered Questionnaires, such as

-The *Kempe Family Stress Inventory (KFI)* - A retrospective cohort study found that a high score on the KFI was the only statistically significant predictor of maltreatment at 1 and 2 years and, when compared with a low score, was associated with more clinic visits during the first year and hospital admissions during the first 6 months.

-*Hawaii Risk Indicators Screening Tool.*

-*Child Abuse Potential (CAP)* inventory

2. Clinical Staff-administered Questionnaires, such as

-The *Maternal History Interview (MHI-2)* utilizes open-ended questions and subscales to evaluate parenting skills, personality, discipline philosophy, life stress, and others to determine risk for child abuse. Mothers determined to be high-risk by the MHI-2 had a higher incidence of reported child abuse than low-risk mothers in a study of young pregnant women.

-The *Parenting Profile Assessment (PPA)* is a 21-item nurse interview designed for the primary care setting. Responses on the PPA were compared with self-reports about past episodes and indicated 75% sensitivity and 86% specificity.

3. Other Techniques: Clinician Observation

It must be noted however that false-negative results may hinder identification of those who are truly at risk. False-positive results could lead to inappropriate labeling and punitive attitudes. Additional possible harms include psychological distress, escalation of abuse and family tension, loss of personal residence and financial resources, erosion of family structure, loss of autonomy for the victim, and lost time from work. Children could lose contact with established support systems including neighbors, siblings, school contacts, and peer groups.

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4. Recognize signs and symptoms of physical abuse

Red Flag History to evaluate a child for possible physical abuse:

● History does not explain degree or type of injury

●Delay in seeking care

●Injury attributed to in-home resuscitation efforts

●Caregiver histories that change with retelling or conflict with versions from other observers

●Severe injury explained as self-inflicted or blamed on other young children or pets

It is important to interview a verbal child about a suspicious injury without caregivers present whenever possible.

Red Flag Findings on Physical Exam:

Inflicted Bruises

●Any bruising in infants < 6 months old

● >1 bruise in a pre-mobile infant and > 2 bruises in a crawling child

●Bruises located on the torso, ear, neck, or buttocks

●Bruises with a pattern of the striking object (eg, slap, belt, or loop marks; spoons; spatulas; or other objects)

●Human bite marks

Of note: estimating the age of a bruise using its color is inaccurate and misleading and should not be used.

Oral injuries (highly suspicious especially in non-ambulatory infants)

●Lip lacerations or bruising

●Lingual or labial frenulum tears

●Tongue lacerations

●Bruising or wounds of the buccal mucosa, gums, or palate

●Missing or fractured teeth with an absent or inconsistent history

●Maxillary or mandibular fractures with an absent or implausible history

●Bruising, lichenification, or scarring at the corners of the mouth from being gagged

Intentional burns

●Scalds in children < 5 years of age that do not fit the pattern of an unintentional spill

●Scalds from hot tap water due to immersion, demonstrating a sharp upper line of demarcation, affecting both sides of the body symmetrically, or involving the lower extremities and/or perineum

●Burns that have a sharply demarcated edge in the shape of the burning object (eg, metal tops of butane cigarette lighters)

●Cigarette burns that appear as discrete circular burns 8 to 12 mm in diameter and are deep

●Stun gun burns usually are multiple and appear as paired lesions (0.5 cm in diameter and 5 cm apart)

Visceral injuries: severe esophageal, pulmonary, cardiac, or abdominal injury in a child with a questionable or absent mechanism of injury is an important red flag for abuse, especially when it

occurs in children <4 years of age. Associated head injury, fractures, bruises or burns should increase suspicion for physical abuse.

5. Understand the legal and clinical implications of reporting physical abuse

- Mandatory reporting of a suspicion of abuse is required for physicians and other medical providers in many regions
- Documentation is key. Each step of the evaluation and management of suspected child abuse is essential. Statements made by the child or parents should be recorded as direct quotations to decrease the chance of misinterpretation. Injuries should be described in as much detail as possible. Sketches and/or high-quality photographs (if permissible) are helpful in documenting extensive injuries.
- Interviewing a verbal child without the presence of the caregivers is of utmost importance
- Children with serious injuries (eg, intracranial/intra abdominal injury, femur fracture, or extensive burns) should be admitted to a pediatric trauma center and managed appropriately
- Hospitalization may be indicated when there is ongoing concern for a child's safety. This permits continued contact between the child and a possibly non-abusive caretaker while the assessment is being completed rather than removal from the family.
- Children with injuries that do not require hospitalization may be discharged when the abuse evaluation is complete and a safe outpatient setting has been identified in conjunction with Child Protective Services

6. Recognize characteristics of fractures caused by physical abuse

Fractures

Highly suspicious fractures:

●Metaphyseal corner (bucket handle) fractures

●Rib fractures

●Fractures of the sternum/scapula/spinous processes

●Long bone fracture in a non-ambulatory infant

●Multiple fractures in various stages of healing

●Bilateral acute long bone fractures

●Vertebral body fractures and subluxations in the absence of a history of high force trauma

●Digital fractures in children <36 months of age or without a corresponding history

●Epiphyseal separations

●Severe skull fractures (eg, multiple, stellate, or depressed) in children <18 months of age, particularly without a corresponding history

Less specific fractures:

●Isolated long bone fractures in ambulatory children

●Linear skull fractures

●Clavicle fractures

●Sub-periosteal new bone formation

7. Know the signs and symptoms of abusive head trauma

Abusive head trauma —Features that are most predictive of child abuse include intracranial hemorrhage (especially subdural hemorrhage) associated with one or more of the following:

●Inadequate/inconsistent history with regards to mechanism of injury

●Apnea or seizures

●Associated fractures of the ribs, metaphyseal region, or long bones (i.e high risk fractures)

●Retinal hemorrhages

●Any skull fracture other than an isolated, unilateral, non-diastatic, linear, parietal skull fracture

●Any associated bruising of the child's ears, neck, or torso

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8. Know the appropriate management for a child suspected of having been abused

Workup:

Parent/caregiver history:Each involved should be interviewed separately and away from the child

- Open-ended questions in non-judgemental manner

- Detailed notes should include:
    - Sequence of events leading to injury
    - When child was last well/healthy
    - Identify all adults/caregivers with the child during time of injury
    - Identify potential witnesses
    - Full history including developmental and behavioral
    - Risk factors-interpersonal violence, substance use, mental health issues, history with CPS, prior law enforcement involvement
    - Protective factors-community and family supports, child care assistance, child's consistent participation in school and other activities, family's strong meaningful connections to community agencies and parental willingness to seek assistance

Child history: Only if the child is verbal and developmentally able to answer questions

- Questions appropriate to developmental age
- Consider ability to answer questions based on behavioral and developmental diagnoses
- Open-ended questions, do not lead. Ask-"What happened here?" and follow up "tell me more about that"
- Immediately document quotes as well as the question that elicited the answer
- Obtain only information necessary for medical diagnosis and treatment-A detailed interview should be completed by specially trained professionals in response to report of suspicious injuries to local child welfare agency
- Reassure child-what happened not his/her fault, ok to tell you

Physical exam

- Complete, in gown, inspect entire skin
- Attention to privacy
- Document location, size, pattern, use photographs and/or drawings when possible
- Occult injury workup:
    - Head imaging-CT or MRI if younger than 4 months or older but with possible neurological sx
    - Skeletal survey-if younger than 24 months or older and nonmobile, nonverbal
    - AST/ALT-abdominal trauma, Abd CT for bruising or AST/ALT over 80
    - Urine tox screen
    - Additional labs per history and physical findings

Consider differential diagnosis/alternative explanations such as bleeding disorder, connective tissue disorder, accidental injuries, birthmarks, skin conditions, infection, leukemia, rickets, syndromes, etc.

<u>Management</u>

- Report suspected abuse
- Consult child abuse specialist about any positive findings
- Alert family to possible reactions of child
- Educate parent/caregiver to provide sense of physical security, normal routines, affection, limit-setting
- Educate parent/caregiver not to pry or try to make patient forget experience-be open, listen, support
- Parent/caregiver self-care and care for other family members
- Refer to experienced mental health clinician with training in trauma-informed therapy

- Know and utilize local resources-mental health resources, parent groups, advocates, children's groups, offender treatment
- Educate families that new questions and feelings will arise with successive developmental change and be prepared to normalize these feelings
- Continue to monitor for any signs of recurrence of abuse
- Important to schedule follow-up visits in the medical home more frequently after suspected maltreatment and particularly if out of home placement is made. Guideline for youth entering foster care: 3 visits in 3 months after CPS involvement or leaving foster care and every 6 months after that. For children returning to his or her family, child should be seen in the first week, at 1 month and then 3 months after the transition.
- Be alert/monitor carefully for sequelae of abuse, including behavioral and developmental/academic progress as well as specific attention to problems could develop as the result of acute head trauma

9. Know the advantages and disadvantages of "family preservation" vs removal of the child from the home in the face of repeated serious physical abuse

- Two-thirds of children who have been determined by CPS to have been maltreated will remain in the care of their families while receiving supportive and therapeutic services.
- Even when children are placed in out-of-home care, approximately ½ will be returned to their families within days to months. The median length of stay in foster care for children who are later re-unifed with their family of origin is 8 months.
- Research on ACEs demonstrates that child abuse and neglect can have a profound impact on the development of a child. When children are removed from their homes due to abuse or

neglect, it is important to realize that this placement can potentially be another negative stressor for the child.

- Children are often separated from family, friends, neighborhoods and schools
- Half of foster children experience more than one foster care placement
- Plans for reunification or adoption are often uncertain with no clear timeline in place.
- Children in foster care have higher rates of mental health issues, developmental delays, and learning problems compared to peers. Up to 25% of young children in foster care have a developmental delay, up to 25% of teenagers in foster care have PTSD, and 80% of adolescents aging out of the foster care system have a mental health diagnosis. Over 40% of foster children receive special education services, and their rates of acute and chronic illness are also increased.
- Central to a child's social and emotional development is his or her ability to form a secure attachment to a stable, caring, responsive adult.
- The age and developmental level of a child at foster placement, as well as the quality of care he or she receives, will contribute to the emotional consequences of abuse and placement.
- Multiple foster care placements can further disrupt a child's ability to form secure attachments.
- By educating foster and adoptive parents about the effects of toxic stress on a child's learning, behavior, and health, the professional can encourage understanding of maladaptive and frustrating behaviors.
- A child may benefit from referral to trauma-informed counseling services, such as Parent-Child Interaction Therapy or Trauma Focused Cognitive Behavioral Therapy

10. Know the components of effective programs for the prevention of child abuse

- Address both primary and secondary prevention. Children who have already been victims of abuse may be abused again. Recognizing and intervening when abuse is suspected is a form of prevention. Need to be vigilant in follow up and also provide programs and resources to parents/guardians to prevent future abuse. Prevention can include programs and resources known to be general protective factors
- Efforts need to be broad and sustained
- Pediatrician, as trusted advisor, can play an important role in assessing parent/caregiver strengths and deficits, providing education and connecting families to resources to address family needs
- Pediatrician actions for prevention/protective effects:
    - Promote positive parenting through individual counseling, integrating parenting programs, and referring families to evidence-based programs and evidence-informed resources (Some evidence that therapeutic school-based centers (eg, Head Start) and parenting programs (eg, Triple P) have protective effects when there is a parent inclusion component that includes support and education)
    - Screen for developmental/behavioral problems and maternal depression
    - Monitor families for signs of parental stress and maternal depression and look for early warning signs of child abuse and neglect
    - Connect families to high-quality child care and preschool and maternal-child-home-visiting programs (note that there is evidence that home-visitation programs reduce the risk of child maltreatment)
    - Advocate for funding of programs that ameliorate negative environmental influences on families

- Help parents understand the importance of child's social-emotional development and work with families to provide a stable, loving and nurturing environment
- Although most people in the United States would agree that not all forms of corporal punishment are child abuse, most child abuse starts as corporal punishment. For these reasons, practitioners should discourage parents from using corporal punishment and should emphasize proactive and positive approaches

Selected tables from AAP Clinical Report:The Pediatrician's role in child maltreatment prevention (reference 8 below):

TABLE 2 Protective Factors

Dispositional/Temperamental Attributes of the Child	Warm and Secure Family Relationships	Availability of Extrafamilial Support
Above-average cognitive ability	Presence of a caring and supportive adult	Structured school environment
High ego control (high degree of impulse control and modulation)	Positive family changes (eg, family interventions, father no longer allowed on visitations)	Involvement with a religious community
Internal locus of control (belief in one's ability to control own destiny)		Involvement in extracurricular activities or hobbies
External attribution of blame (attribute cause to something outside oneself [eg, some external pressure])		Access to good health, educational, and social welfare services
Presence of spirituality		
Ego control and ego resilience (able to modify impulses and insulate themselves from environmental distracters)		
High self-esteem or sense of self-worth		

TABLE 3 Incorporating Primary Child Maltreatment Prevention Into the Health Supervision Visit

	Parent Coping Skills and Support System
Prenatal or first visit	Who lives in the home? History of mental health problems, substance abuse/alcohol abuse, or intimate partner violence?
	How were the parents parented and disciplined?
	What were the parents' experience(s) with trauma?
	Are there financial problems and/or poverty?
	Was the pregnancy planned?
	Who will care for the infant?
Newborn	Infant crying
	Expectations
	Identify 3 friends or family members who can help (safety line)
First months	Infant crying
	Normal development and expectations
	Maternal depression
	Identify 3 friends or family members who can help (safety line)
	Loving is not "spoiling"
Cruiser/toddler	Discipline = teaching
	Toilet-training
	Normal development and age-appropriate expectations
Preschool	Teach child names for genitalia
	Safe touch/unsafe touch
	Normal sexual behavior
	Normal development and age-appropriate expectations
	Discipline = teaching
	Model nonviolent anger management and conflict resolution
School	Discipline = teaching
	Model nonviolent anger management and conflict resolution
	Appropriate supervision
	Respect private parts of others and others to do the same
	Personal safety; peer pressure; Internet use
Adolescence	Discipline = teaching
	Dating violence
	Model nonviolent anger management and conflict resolution

Note that topics may be reintroduced at successive visits

Table (Reference 9 below) Preventing Child Abuse and Neglect (CDC):

Contextual and Cross-Cutting Themes

The strategies and approaches that have been included in this technical package represent different levels of the social ecology, with efforts intended to impact the community and societal levels, as well as individual and relationship levels. The strategies and approaches are intended to work in combination and reinforce each other to prevent child abuse and neglect (see box below). The strategies are arranged in order such that those strategies hypothesized to have the greatest potential for broad public health impact on child abuse and neglect are included first, followed by those that might impact more select populations (e.g., first-time parents or those for whom child abuse and neglect is already present).

Preventing Child Abuse and Neglect

Strategy	Approach
Strengthen economic supports to families	• Strengthening household financial security • Family-friendly work policies
Change social norms to support parents and positive parenting	• Public engagement and education campaigns • Legislative approaches to reduce corporal punishment
Provide quality care and education early in life	• Preschool enrichment with family engagement • Improved quality of child care through licensing and accreditation
Enhance parenting skills to promote healthy child development	• Early childhood home visitation • Parenting skill and family relationship approaches
Intervene to lessen harms and prevent future risk	• Enhanced primary care • Behavioral parent training programs • Treatment to lessen harms of abuse and neglect exposure • Treatment to prevent problem behavior and later involvement in violence

11. Know the long-term outcome of physical trauma

Child maltreatment often has significant short- and long-term consequences

- Risk for behavior problems
- Risk for poor cognitive function and learning problems
- Risk for PTSD, anxiety, depression, SI and suicide attempt
- Important to note child may seem to be functioning well at the time of evaluation-problems can emerge months or years later-need long term follow up care in the medical home
- Risk for maltreatment of the child's future children

Protective factors and intervention early associated with better outcomes

B. Sexual abuse

Sexual abuse is defined as the engagement of a child in sexual activities that the child cannot comprehend, for which he or she is developmentally unprepared and cannot give informed consent, and/or that violate laws and/or societal taboos. A spectrum of activities constitutes sexual abuse, from exposure to pornographic materials or sexual situations, to use of a child to produce pornography, to inappropriate touching and penetration. Coercion and threats by the perpetrator are common

1. Know the risk factors for sexual abuse

Risk factors for sexual abuse-overlap with risk factors for child abuse generally

Note: Female gender-higher risk for sexual abuse based on reporting

3 general categories: Child risk factors, parental/family risk factors, environmental risk factors

TABLE 1 Factors and Characteristics That Place a Child at Risk for Maltreatment

Child	Parent	Environment (Community and Society)
Emotional/behavioral difficulties	Low self-esteem	Social isolation
Chronic illness	Poor impulse control	Poverty
Physical disabilities	Substance abuse/alcohol abuse	Unemployment
Developmental disabilities	Young maternal or paternal age	Low educational achievement
Preterm birth	Parent abused as a child	Single parent
Unwanted child	Depression or other mental illness	Nonbiologically related male living in the home
Unplanned pregnancy	Poor knowledge of child development or unrealistic expectations for child	Family or intimate partner violence
	Negative perception of normal child behavior	

Reproduced with permission from Flaherty et al.[34]

(Table from Reference 4-AAP Clinical Report referenced below)

2. Recognize physical signs and symptoms of sexual abuse

Note most cases of sexual abuse have normal or nonspecific exams-in suspected sexual abuse, rely on the history to make the diagnosis. A normal exam neither rules out nor confirms possibility of sexual abuse

- Genital or rectal pain, discharge, bleeding, injury, infection
- Evidence of STI

- NO symptoms
- Important to note that of premenarchal girls with vaginal discharge-less than 5-10% occur in the context of sexual abuse and that only a small percentage of sexually abused children will have an abnormal genital or anal finding.

3. Recognize psychological symptoms of sexual abuse

Note that these do not specifically indicate abuse

- Fears/phobias, can be triggered by reminders
- Sleep problems/nightmares
- Appetite change/eating disorders
- Enuresis/encopresis
- Change in behavior, attitude or school performance
- Depression, anxiety, withdrawal from social situations, suicidality
- Excessive anger, aggression, or running away
- Concerning sexualized behavior (note that this can also demonstrate exposure to age-inappropriate media and information)
- Multiple sexual partners at a young age
- Substance abuse
- Sexualized behaviors or knowledge of words that would be unanticipated for age

4. Know appropriate interviewing techniques for assessing possible victims of sexual abuse

History from parent/caregiver

- Determine if child made disclosure

- Determine last exposure to potential abusing person
- Assess current safety
- Identify developmentally normal vs concerning sexualized behavior

History from child

- Apart from caregiver
- Eye-level
- Establish rapport
- Speak at his/her developmental level
- Can use line drawings with young children. Do NOT use anatomic drawings or dolls
- Introduce topic of possible sexual abuse in general way
- Use non-leading questions
- Document exact language/quotes to specific questions asked
- Reassure the child it is not his/her fault. Do not make promises to child that they will not need to talk to someone else
- Take the child's statements and behavior seriously
- Child (particularly older child) may be embarrassed, feel partially responsible, may have been threatened if they disclose, may sacrifice themselves for the family (responsible party would go to jail, house will be sold, others will be angry at the child)
- Child may retract statement when they see the family/person they disclosed to become upset

5. Know how to plan the management of a child who has been sexually abused

- 3 primary domains:
    1. Providing appropriate medical care

2. Reporting to protective services
3. Ensuring mental health service for child and family

- 5 important steps by pediatrician:

1. Decision about how much history to obtain directly from the child

2. Decision about extent of physical exam and any lab or forensic evidence will be collected

3. Careful documentation in the medical record

4. Meet with non-offending parents and the child, if old enough to explained that a report will be made to protective services and an investigation conducted

5. Arrange follow-up with the family and provide ongoing support

- Use children's advocacy centers (CACs) and multidisciplinary teams for coordinated and rational approach if available
- Immediate referral if within 72 hours of sexual assault to local child protection team, advocacy center or ED

6. Know the long-term outcomes of childhood sexual abuse

- Sexual abuse can have a long-lasting and devastating effect on the development of children, adolescents and adults
- Domains of functioning affected include survivor's emotional state, sense of self (victim, powerless, worthless), relationships with others

- Targets for treatment including feelings of self-blame, sexual awareness, poor self esteem and feelings of powerlessness and mistrust of adults.
- Boys may be concerned about their masculinity and whether they are gay. Men who were sexually abused as children are at increased risk of mental health or substance abuse problems and are more likely to become a perpetrator of sexual abuse
- Overall sense of loss created by disclosure and potential anger/upset family members
- Girls/teens are at increased risk for mental health problems such as eating disorder, multiple personality disorders and PTSD as well as pregnancy at a younger age

7. Know the epidemiology, including the most common perpetrators, of sexual abuse

- In the US in 2016, CPS evaluated 3.5million children and identified 676,000 victims. The majority of children (75%) were found to have been the victims of child neglect. Approximately 18% were victims of physical abuse and 8.5% were victims of sexual abuse. Some were victims of more than one type of maltreatment.
- Approx. 75% of children evaluated for sexual abuse are girls (based on CPB reporting), majority of reports are unsubstantiated-important to note this does not mean abuse did not occur but that CPS was unable to substantiate that it did occur
- All social classes and educational backgrounds affected, although racial and socioeconomic factors influence reports to CPS.
- Most victims know the perpetrator
- Perpetrator-sexual arousal to children and willingness to act on this. Factors more likely to decrease control of behavior-drugs/alcohol, lack of empathy for the child, thought that the behavior is acceptable and not harmful to the child, history of being abused as a child

- Children often selected based on perceived vulnerability (ex. Intellectual disability), in situations where they have increased contact with the child)
- Initial attention, gifts commonly used by perpetrators
- Often occurs in home or setting where there are no witnesses
- Can occur in conjunction with other types of abuse
- Comes to clinical attention usually by:
    - Report of child to clinician or another adult
    - Note that children may lack the vocabulary to describe what happened to them (e.g., "We played the hugging game")
- Older children may be more likely to tell a peer, often at first with a vague disclosure

8. Know the components of effective school-based programs to prevent sexual abuse

- Attempts have been directed toward developing programs to teach children about good and bad touches. Children 4-6 years are able to learn and retain this over a short period in studies. No conclusive evidence yet that these programs have resulted in prevention or earlier recognition of sexual abuse
- Conclusion of one review/meta analysis (Reference 10 below): Children's self-protective skills and knowledge can be increased by participation in school-based sexual abuse prevention programs. However, it is unknown whether gains in skills and knowledge actually decrease the likelihood of child sexual abuse

  www.childwelfare.gov -has resources for teachers include webinars, resources and programs such as Circles of Safety. For example:

https://www.stopitnow.org/circles-of-safety-training/circles-of-safety-for-youth-serving-organizations

Participants who complete Circles of Safety:

- Use the knowledge gained about warning signs and situations to define their own rules about personal space, privacy, and appropriate interactions with children;
- Proactively communicate their boundaries to adults or older youth interacting with their children or the children in their care;
- Speak up or intervene when they are concerned about an adult or older youth's behavior around children; and
- Gather allies to create a circle of safety around children.

9. Recognize the heightened risk of individuals with developmental disabilities to become victims of sexual abuse

- Young infants are highest risk for abuse overall due to fewer developmental skills and nonverbal status
- Children/youth with disabilities are at high risk for physical, sexual and emotional abuse

10. Know how to evaluate a child for possible sexual abuse

History from parent/caregiver

- Determine if child made disclosure
- Determine last exposure to potential abusing person
- Assess current safety

- Identify developmentally normal vs concerning sexualized behaviors

History from child

- Apart from caregiver
- Eye-level with the child
- Establish rapport
- Speak at his/her developmental level
- Can use line drawings with young children. Do NOT use anatomic drawings or dolls
- Introduce topic of possible sexual abuse in general way
- Use non-leading questions
- Document exact language/quotes to specific questions asked
- Reassure the child it is not his/her fault. Do not make promises to child that they will not need to talk to someone else

Physical Examination

- Important to do full examination (not just genital exam), explain to child in developmentally appropriate way and with privacy and good light source, stop exam if child is unable to cooperate and discuss plan for further exam attempt with PCP vs. child abuse specialist
- Female genital exam positions-supine frog-leg, prone knee-chest or lithotomy
- Male exam should include entire genital area and anal exam
- Know genital anatomy, developmental variation with age and puberty and anatomic variants for both male and female
- Consider possible abnormal findings not due to abuse such as lichen sclerosis, urethral prolapse, hemangiomas, straddle injuries

Lab testing for STIs, pregnancy when indicated based on history and physical, pubertal stage

- GC, chlamydia
- Serologic testing- syphilis, HIV, Hep B, Hep C
- If symptomatic, can be cultured for other genital infections-trichomonas, gardnerella, Strep pyogenes Candida. HSV may be considered if it would change management although differentiating between HSV1 and 2 does not assist in determining whether an HSV infection was transmitted through sexual contact
- Condyloma acuminata is diagnosed clinically
- Pregnancy test on peripubertal girls

Reporting guidelines for lab tests: Note that positive GC, syphilis, chlamydia have a certain likelihood of sexual abuse in prepubertal children and should be reported. Trichomonas-possible likelihood and should be reported. Condyloma and HSV 1, 2-possible likelihood-should be reported if sexual abuse is suspected on history and physical. BV-uncertain likelihood of sexual abuse and is not recommended to be reported

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HHS/ACF: www.acf.hhs.gov/programs/cb

HHS/Child Welfare gateway: www.childwelfare.gov

C. Psychological abuse

1. Know the risk factors for psychological abuse

Social and environmental risk factors include

- social isolation

- poverty
- severe housing difficulty
- problematic immigration status

Caregiver risk factors include:

- mental health difficulties
- alcohol and drug misuse
- domestic violence/significant parental conflict
- caregiver childhood maltreatment

Demographics:

- Older children (>6 years of age)
- Children from lower income families
- Boys slightly more likely than girls

2. Recognize the signs and symptoms of psychological abuse

- A key part of identifying psychological abuse is being attuned to interactions between children and their caregivers.
- Suspicion of psychological abuse relies heavily on history or direct observation of caregiver-child interactions and child/adolescent emotional and behavioral well-being
- Important to note that psychological abuse can co-occur with other forms of maltreatment
- signs to look for during observation/history taking of the caregiver-child interactions: negativity/rejection, threatening the child, modeling and encouraging anti-social behavior and failure to show affection
- Psychologically abusive behavior by caregivers include:
  - spurning (eg, ridiculing or humiliating),

- terrorizing (eg, threatening violence against a child or a child's loved ones)
- isolating (eg, restricting social interactions)
- corrupting/exploiting (eg, involving in illegal activities)
- denying emotional responsiveness (eg, providing no praise).

- important to ask nonjudgmental questions about relationships in the family from as many members as possible. This should be done individually to protect anyone disclosing abuse from retaliation. Probing around caregiver discipline practices and how conflict is handled in the home and between family members can yield important information
- crucial to interview a child privately to avoid any undue influence by a caretaker on the child's responses

3. Know how to plan the management of a child who has been psychologically abused

- prevention is the best intervention, and from a family's first visit, pediatricians have the opportunity to discuss approaches to parenting and to work with parents about being sensitive to their child's needs
- If there are clear social and environmental, or parental risk factors, for instance homelessness or clear mental health problems, it is likely to be necessary to intervene first with these contextual problems
- clear evidence of harm is not a prerequisite for reporting psychological abuse: if a clinician suspects that psychological abuse has or may be occurring, a report to the authorities and child protection personnel should be made in accordance with state laws
- documenting statements made by the child, child's family, teachers, and child care personnel are critical to detailing a repeated pattern of caregiver behavior that is or can be detrimental to the child's well-being.

- Refer to appropriate services: child and adolescent mental health services, adult mental health services, domestic violence and substance misuse services
- Hostility toward the child is often based on negative beliefs which the caregiver has about the child's temperament or personality and which are difficult to shift. This requires skillful work and counseling with the caregivers to improve their understanding of their child's needs.
- Incorporating a psycho-educational parenting approach. Several parent training programs have been developed.
- The principles of support for the child within the family in coping with continuing emotional abuse include:

  · Acknowledging explicitly the reality of the child's experiences, while avoiding denigration of the caregivers.

  · Explaining the parents' difficulties.

  · A problem solving approach to enable the child to cope with the forms of emotional abuse and neglect.

  · Working with child's feelings of self blame and low self esteem

  · Enabling the child to maintain or develop an enduring and meaningful relationship with at least one positive adult.

  · Ensuring that the child is able to fulfill their educational potential, which will enhance the child's self esteem

4. Know the long term outcomes including impact on somatic health, mental health, and future parenting of own children

- Impact on somatic and mental health: depression, anxiety, and posttraumatic stress disorder, antisocial behavior, eating and weight-related disorders, lower educational attainment, and impairment in peer relationships that continue into adulthood.
- Caregiver childhood maltreatment is a risk factor for inflicting similar psychological maltreatment to their own children in the future

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D. Factitious disorder imposed on another (Munchausen syndrome by proxy) (Matt)

Factitious disorder imposed on another is the diagnosis from DSM-5 that applies to the perpetrator, not the victim. It has been known by many other names, including factitious disorder by proxy and Munchausen syndrome by proxy. Recently, many have started to use the term Medical Child Abuse in order to focus the attention on the harm or potential for significant harm caused to the child. This diagnosis can be given to the child.

1. Recognize signs and symptoms suggestive of factitious disorder imposed on another

Here are some red flags for possible factitious disorder imposed on another (from PIR article):

1) A diagnosis that does not match the objective findings

2) Inconsistent histories, signs, and symptoms that are present only in the presence of 1 caregiver

3) Failure of the child's illness to respond to normal treatments

4) Caregiver insists on invasive procedures

5) Caregiver does not express relief or pleasure when told that his or her child does not have a particular illness or improves

6) Caregiver or siblings have a history of unusual or unexplained illness

7) Doctor shopping

2. Know family risk factors often seen in cases of factitious disorder imposed on another

Demographics of the perpetrator: >90% are the patient's mother. The average age of the perpetrator is in the high 20's. About half of the time they work in healthcare. They often have a history of mistreatment as a child (30% in Yates, et al), an obstetric complication (24%), or factitious disorder themselves (31%).

3. Know how to plan the management of cases of factitious disorder imposed on another

Treatment of medical child abuse is complex and involves many different disciplines to treat both the patient and caregiver.

First, the family must be informed, including the perpetrating caregiver. The goals of this meeting are:

1) Detail the current medical condition of the child

2) Dispel any false claims of alternative medical conditions

3) Express concerns about the caregiver's distorted perception of the child's health

4) Focus on a path forward toward normalization of the child's medical condition

If the caregiver refuses a protective plan, separation of the child from the caregiver should be pursued with CPS. There is a possibility that the caregiver will escalate the malicious behavior in order to prove the child's illness.

The medical treatment plan of the patient must be changed. Unnecessary and harmful interventions and medications should be peeled away. Depending on the medications and interventions, they may be relatively quickly. Sometimes admission to the hospital is necessary for this step.

4. Know how to diagnose factitious disorder imposed on another

Medical child abuse is very difficult to diagnose, and the average time to diagnosis from onset of symptoms is 15-22 months. Diagnosis requires a thorough review of the medical records in order to document the reported symptoms and illnesses in comparison with the objective findings (labs, clinic visits, therapies, etc). "The diagnosis requires demonstrating that the individual is taking surreptitious actions to misrepresent, simulate, or cause signs or symptoms of illness or injury in

the absence of obvious external rewards. Methods of illness falsification can include exaggeration, fabrication, simulation, and induction." (DSM 5)

The AAP Committee on Child Abuse and Neglect recommended using these 3 questions when making a diagnosis:

1) Are the history, signs, and symptoms of disease credible?

2) Is the child receiving unnecessary and harmful or potentially harmful medical care?

3) If so, who is instigating the evaluation and treatment?

When gathering medical records, conversations should be conducted with the various medical providers to discuss any previous concerns. This includes reaching out to other institutions.

Sometimes children are admitted to the hospital to help make a diagnosis. The goals of the admission could include observing the child's symptoms, observing the caregiver interactions with the patient, meticulous documentation of the actual signs and symptoms. Caregivers should no administer medications or feeds. Rarely, video monitoring of the admission can help confirming malicious actions.

DSM 5 Diagnostic Criteria for Factitious Disorder Imposed on Another (F68.A)

A. Falsification of physical or psychological signs or symptoms, or induction of injury or disease, in another, associated with identified deception.

B. The individual presents another individual (victim) to others as ill, impaired, or injured.

C. The deceptive behavior is evident even in the absence of obvious external rewards.

D. The behavior is not better explained by another mental disorder, such as delusional disorder or another psychotic disorder.

Note: The perpetrator, not the victim, receives this diagnosis.

Specify:

- Single episode
- Recurrent episodes (two or more events of falsification of illness and/or induction of injury)

References for section D:

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E. Child neglect

1. Know common developmental and behavioral sequelae of chronic neglect :

There are various developmental and behavioral sequelae of chronic neglect that present from childhood to adulthood.

In young children these are some of the effects seen

I. Cognitive Outcomes

a. Executive Function and Attention

Child abuse and neglect have profound effects on the prefrontal cortex, a brain structure centrally involved in executive functioning. Executive functioning refers to higher-order cognitive processes that aid in the monitoring and control of emotions and behavior. Children who experience abuse and neglect appear to be especially at risk for deficits in executive functioning and attention, which have implications for behavioral regulation.

b. Academic Underachievement

There is a strong body of evidence that suggests that both abuse and neglect are predictive of academic problems. They have a significant impact on reading ability, IQ scores, and academic achievement.

II. Psychosocial Outcomes

a. Attachment

Children develop *secure* attachments to parents who are responsive to them when they are distressed. Children typically develop *insecure (avoidant or resistant)* attachments when parents are unresponsive or inconsistent in responsiveness, but not frightening or bizarre. Secure, avoidant, and resistant attachments are referred to as *organized* attachment strategies because they are organized around the caregiver's availability and provide a child a template for dealing with distress. On the other hand, *disorganized* attachment represents a breakdown in or a lack of strategy for dealing with distress when in the parent's presence. Disorganized attachments are the most problematic in terms of outcomes for children. Relative to organized

attachment, disorganized attachment is most predictive of long-term problems, especially externalizing symptoms. Child abuse and neglect are predictive of disorganized attachment, as well as insecure attachment.

In early childhood, abused or neglected children may develop *attachment disorders.* Childhood attachment disorders are phenomena distinct from insecure, disorganized, or nonexistent attachment types; they have been redefined in the DSM-V to include two distinct disorders: (i) reactive attachment disorder and (ii) disinhibited social engagement disorder Reactive attachment disorder involves inhibited or emotionally withdrawn behavior, including rarely seeking and responding to comforting; it results from a lack of or incompletely formed selective attachments to adult caregivers. Disinhibited social engagement disorder is marked by a pattern of overly familiar behavior with strangers; it may occur even in children with established or secure attachments. Previously, each attachment disorder was considered the inhibited or disinhibited type of reactive attachment disorder, respectively.

b. Emotion Regulation:

The scaffolding important for the development of emotion regulation is challenged in abusing or neglecting families. It is likely that abused and neglected children experience not only a lack of modeling and support and an absence of positive affect but also harsh, inconsistent, and insensitive parenting. In the case of abuse, parents often respond in threatening or unpredictable ways to children's distress. In the case of neglect, parents may be unresponsive or non empathic. As a result of either response, children are at risk of failing to develop effective strategies for regulating emotions.

Compared to their non maltreated and abused counterparts, physically and emotionally neglected preschoolers often demonstrate notable problems in coping, personality development, and emotion regulation. Neglected preschoolers are generally confused by the emotional displays of others and are less able to discriminate emotions than non maltreated and abused children

c. Peer Relations

Abused and neglected children have problematic peer relations at disproportionately high rates as do children with a history of institutional care. Problematic emotion regulation and higher levels of aggression and withdrawal found in abused and neglected children can become apparent to peers when frustrations and challenges arise in school and playground environments.

d. Representations of self and others

Abused and neglected preschoolers often have negative mental representations (i.e., internal working models) of the self and others. The unresponsive, insensitive, or traumatizing care that they have experienced often leaves them with models of themselves as unworthy of love and others as unavailable or rejecting.

e. Externalizing Problems

Findings from several studies indicate that children who have experienced abuse and neglect are at greater risk for a number of externalizing behaviors, including oppositional defiant disorder,conduct disorders, aggression, and delinquency.

f. Internalizing Problems

Internalizing symptoms refers to symptoms of anxiety and depression. Child abuse and neglect have been found to put children at increased risk of internalizing symptoms from early childhood through adolescence and adulthood. An article published by Gilbert et al in 2009 cite a body of studies reporting adjusted odds ratios ranging from 1.3 to 2.4 for depression after childhood among those subjected to abuse and neglect as children. (1)

g. Dissociation

Dissociation is defined as a "disruption of and/or discontinuity in the normal, subjective integration of one or more aspects of psychological functioning, including—but not limited to—memory, identity, consciousness, perception, and motor control". Child abuse and neglect have been associated with dissociation among both preschool-aged and elementary-aged children as well as among adults.

h. Post Traumatic Stress Disorder

DSM-V defines PTSD as a trauma- and stressor-related disorder. PTSD develops following "exposure to actual or threatened death, serious injury, or sexual violation," including directly experiencing the traumatic event, witnessing the event firsthand, learning that an actual or threatened violent or accidental death occurred to a family member or close friend, and experiencing repeated or extreme firsthand exposure to the details of the traumatic event. A number of prospective and retrospective studies have found elevated rates of PTSD among individuals with a history of abuse and neglect Numerous studies have found that PTSD was preceded by abuse and neglect; links with sexual abuse were especially strong.

i. Personality Disorders

Evidence links child abuse and neglect with personality disorders. A 1999 study by Johnson et al found that adults with a history of abuse and neglect had a fourfold increase in personality disorders relative to those without a history of abuse or neglect. Physical abuse was associated with elevated antisocial and depressive personality disorder symptoms; sexual abuse was associated with elevated borderline personality disorder symptoms; and neglect was associated with elevated symptoms of antisocial, avoidant, borderline, narcissistic, and passive-aggressive personality disorders, as well as with attachment difficulties and other interpersonal and psychological problems (2).

III. Health Outcomes

a. Growth

Extreme neglect and abuse can lead to stunted growth with effect on weight, height and head circumference.

b. Illness

Child abuse and neglect have been linked to various forms of long term chronic debilitating illness as well as various indicators of physical health problems including GastroIntenstinal Problems, Lung Disease, liver disease and ischemic heart disease. Other physical problems include injuries, ingestions, dental problems, malnutrition, neurologic deficits and death.

c. Obesity

In various studies, different forms of child abuse and neglect have been linked with increased body mass index and higher rates of obesity in childhood, adolescence, and adulthood.

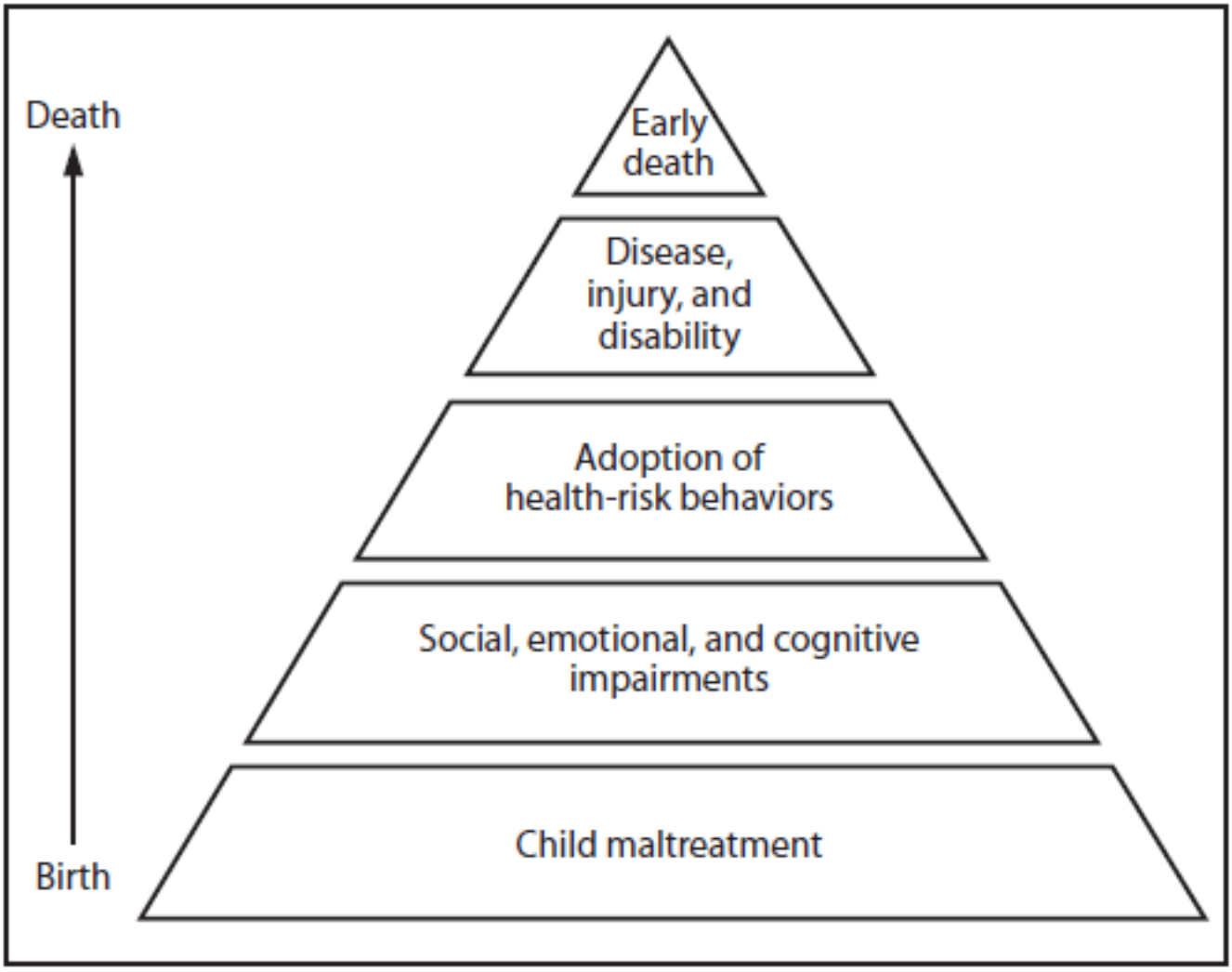
In older children/adolescence these are some of the effects seen

a. Delinquency and Violence

b. Increased risk of Alcohol and Substance use

c. Increased risk of Suicide Attempts

d. Early sex initiation and high risk sexual behaviors that may result in STD such as HIV, Teen pregnancy and in some extreme cases prostitution



2. Understand how the developmental and behavioral symptoms of neglected children vary with stages of development

Symptoms of neglected children vary depending on their stage of development as well as cognition. For e.g. excessive inconsolable crying in infant stage, attachment difficulties in preschool age, academic underachievement and anxiety in young children and externalizing and/or internalizing behaviors in adolescence.

3. Know the legal definition of child neglect

Child neglect is a form of child abuse. It occurs when a person who is responsible for the child fails to care for the minor’s emotional or physical needs. Neglect involves not meeting children's basic needs: physical, medical, educational, and emotional.

Federal legislation provides guidance to States by identifying a minimum set of acts or behaviors that define child abuse and neglect. The Federal Child Abuse Prevention and Treatment Act (CAPTA) (42 U.S.C.A. § 5106g), as amended by the CAPTA Reauthorization Act of 2010, defines child abuse and neglect as, at minimum:

- "Any recent act or failure to act on the part of a parent or caretaker which results in death, serious physical or emotional harm, sexual abuse or exploitation"; or

- "An act or failure to act which presents an imminent risk of serious harm."

This definition of child abuse and neglect refers specifically to parents and other caregivers. A "child" under this definition generally means a person who is younger than age 18 or who is not an emancipated minor.

"Neglect" or "neglected" means:

(a) The inability or unwillingness of a parent, guardian or custodian of a child to provide that child with supervision, food, clothing, shelter or medical care if that inability or unwillingness causes unreasonable risk of harm to the child's health or welfare, except if the inability of a parent, guardian or custodian to provide services to meet the needs of a child with a disability or chronic illness is solely the result of the unavailability of reasonable services.

(b) Permitting a child to enter or remain in any structure or vehicle in which volatile, toxic or flammable chemicals are found or equipment is possessed by any person for the purposes of manufacturing a dangerous drug as defined in section 13-3401.
(c) A determination by a health professional that a newborn infant was exposed prenatally to a drug or substance

4. Know how to plan the management of chronic neglect

Pediatricians may encounter a variety of forms of neglect.

● Noncompliance (nonadherence) with health-care recommendations

● Delay or failure in getting health care

● Hunger, failure to thrive, and unmanaged morbid obesity

● Drug-exposed newborns, older children

● Ingestions; injuries; exposure to second-hand smoke, guns, domestic violence; failure to use car seats/ belts (may reflect inadequate protection from environmental hazards)

● Emotional (eg, excessive quietness or apathy in a toddler), behavior (eg, repetitive movements), and learning problems, especially if not being addressed; extreme risk-taking behavior (may reflect inadequate nurturance, affection, or supervision)

● Inadequate hygiene, perhaps contributing to medical problems ● Inadequate clothing, perhaps contributing to medical problems ● Unmet educational needs ● Abandoned children

● Homelessness

Core Issues for Assessing Child Neglect

1. Do the circumstances indicate that the child's need(s) is (are) not being adequately met? To what extent? Is there evidence of actual harm? Is there evidence of potential harm, based on knowledge of the child (eg, severe asthma) or epidemiologic data (eg, risk from not wearing a bike helmet)?

2. Is there a pattern of neglect? For how long has there been a problem? Are there indications of other forms of neglect? Has CPS been involved?

3. What is contributing to the neglect? Consider factors at different levels:

–Child (eg, child does not tell parent about the problem)

–Parent (eg, parent is ignorant about the condition and the need for help; depressed)

–Family (eg, domestic violence/ other stresses, social isolation)

–Community (eg, poor access to health care, few parent supports)

4. What strengths/resources are available?

–Child (eg, child wants to play sports, requiring better health; child likes school) –Parent (eg, parent interested in knowing more about child's condition)

–Family (eg, other family members willing to help)

–Community (eg, programs for parents, families)

5. What interventions have been tried and with what results? Knowing the details of the interventions can be useful (eg, agency name, nature of the intervention, frequency of contacts, duration). What has the pediatrician done to address the problem?

6. What is the prognosis? Is the family motivated to improve the circumstances and accept help or is the family resistant? Are suitable resources, formal or informal, available?

By employing a systematic approach, the clinician usually can provide the most thorough assessment and management of child neglect.

1. Convey concerns to the family kindly but forthrightly about why one is worried. 2. State an interest in helping or suggest another pediatrician.

3. Address contributory factors, prioritizing those most important and amenable to being remedied (eg, recommending treatment for the mother's depression compared with diminishing neighborhood violence).

4. Begin with the least intrusive approach, usually not CPS.

5. Recognize that neglect often requires long-term intervention, support, follow-up. 6. Try to ensure continuity of care as primary clinician.

7. Establish specific objectives (eg, family will always use a car seat) with measurable outcomes (eg, family reports routine use of car seat at next visit).

8. Engage the family in developing the plan, soliciting their input and agreement. 9. Build on family strengths (eg, a parent's wish to see his or her child do well).

10. Encourage informal supports (ie, family, friends).

11. Consider support through religious affiliation.

12. Consider need for concrete services (eg, Medical Assistance, Temporary Assistance to Needy Families [TANF], Food Stamps).

13. Be knowledgeable about community resources and facilitate referrals.

14. Consider need to involve CPS, particularly when there is serious harm or risk or when less intrusive interventions have failed. Even when a CPS report is substantiated, the vast majority of children remain with their parents and are not placed in foster care. Therefore, constructive efforts to work with families are needed.

15. Provide support, follow-up, review of progress, and adjustment of plan if needed. (5)

Science Helps to Differentiate Four Types of Unresponsive Care

	OCCASIONAL INATTENTION	CHRONIC UNDER-STIMULATION	SEVERE NEGLECT IN A FAMILY CONTEXT	SEVERE NEGLECT IN AN INSTITUTIONAL SETTING
Features	Intermittent, diminished attention in an otherwise responsive environment	Ongoing, diminished level of child-focused responsiveness and developmental enrichment	Significant, ongoing absence of serve and return interaction, often associated with failure to provide for basic needs	"Warehouse-like" conditions with many children, few caregivers, and no individualized adult-child relationships that are reliably responsive
Effects	Can be growth-promoting under caring conditions	Often leads to developmental delays and may be caused by a variety of factors	Wide range of adverse impacts, from significant developmental impairments to immediate threat to health or survival	Basic survival needs may be met, but lack of individualized adult responsiveness can lead to severe impairments in cognitive, physical, and psychosocial development
Action	No intervention needed	Interventions that address the needs of caregivers combined with access to high-quality early care and education for children can be effective	Intervention to assure caregiver responsiveness and address the developmental needs of the child required as soon as possible	Intervention and removal to a stable, caring, and socially responsive environment required as soon as possible

5. Know the parental risk factors associated with child neglect

    1. Parent factors:

        - Low self-esteem
        - Poor impulse control
        - Substance abuse/alcohol abuse
        - Young maternal or paternal age
        - Abused as a child
        - Depression or other mental illness
        - Poor knowledge of child development or unrealistic expectations for child
        - Negative perception of normal child behavior

2. Environment factors (Community/Society)

   - Social isolation
   - Poverty
   - Unemployment
   - Low educational achievement
   - Single-parent home
   - Non–biologically related male living in the home
   - Family or intimate partner violence

6. Recognize child neglect as the most common form of child maltreatment

It is estimated that 1 in 4 children experience some form of child abuse or neglect in their lifetimes and 1 in 7 children have experienced abuse or neglect in the last year (2018-2019). The US Dept of Health and Human services, Child Maltreatment 2018 data shows more than four-fifths (84.5%) of victims suffer a single type of maltreatment. Sixty percent (60.8) are neglected only, 10.7 percent are physi cally abused only, and 7.0 percent are sexually abused only. More than 15 percent (15.5%) are victims of two or more maltreatment types. For 2018, an estimated 1,770 children died of abuse and neglect at a rate of 2.39 per 100,000 children in the national population.(6)

7. Know the child risk factors that predispose to child neglect

Child factors

- Emotional/behavioral difficulties
- Chronic illness
- Physical disabilities
- Developmental disabilities
- Preterm birth
- Unwanted
- Unplanned
- Some normal developmental phases that may cause difficulty for some parents, specifically colic, awakening at night, separation anxiety, normal exploratory behavior, normal negativism, normal poor appetite, and toilet-training resistance.
- Frequent crying

8. Describe interventions that can lower the risk of child neglect (eg, home nurse visits)

1. Connect families to supports that can help them. Build relationships with *local programs that can support families under stress*. In most communities there are a host of programs and resources available to support families. These may include family resource centers, home visiting programs, parent education programs and support groups.
2. Trauma-informed counseling services for children and referrals, as appropriate, to evidence-based treatment options (connecting family with home based therapeutic services, counselors)

3. Resources within the health system of which the practice may be a part (such as home nurse visits, regular clinic follow ups, connecting families with a social worker)

4. Providing access to contact numbers for domestic violence shelters, supportive services, peer support groups and other programs that can be easily accessed by both staff and families

5. Having materials (brochures, Web-based information, etc) that can explain to families the effects that traumatic experiences can have on their child in both the immediate and long-term future – and constructive ways for them to respond and build resilience. Provide educational materials for parents and children on appropriate topics

6. Ensuring that all staff members in a clinical practice know and understand the protocols for making referrals to child protective services

7. Ensuring that the child's environment at home or school is safe – and contact child protective services if it is not

8. Providing supports for parent or caregiver as they cope with a potentially stressful situation (Respite, Home based services, Subsidized daycare, home nurse visits, mental health care for the caregivers)

9. In a number of states resources such as Help Me Grow and 211 can provide an important and easy way to connect families with needed resources and support.

Other relevant information

Protective Factors against Child Neglect

1. Dispositional/Temperamental Attributes of the Child
    - Above-average cognitive ability
    - High ego control (high degree of impulse control and modulation)
    - Internal locus of control (belief in one's ability to control own destiny)
    - External attribution of blame (attribute cause to something outside oneself [eg, some external pressure])
    - Presence of spirituality
    - Ego control and ego resilience (able to modify impulses and insulate themselves from environmental distracters)
    - High self-esteem or sense of self-worth

2. Warm and Secure Family Relationships
    - Presence of a caring and supportive adult
    - Positive family changes (eg, family interventions, father no longer allowed on visitations)

3. Availability of Extrafamilial Support
    - Structured school environment
    - Involvement with a religious community
    - Involvement in extracurricular activities or hobbies
    - Access to good health, educational, and social welfare services

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Content Category 16- Somatic Symptom and Related Disorders

ABP Content Specs for Developmental-Behavioral Pediatrics Fellow Collaboration Project

Prepared by Maja Katusic, MD, Veronica Villarreal, MD, & Ann Kennelly, MD, Baylor/Texas Children's DBP Fellows

Reviewed by Noel Mensah-Bonsu, MD, Baylor/Texas Children's DBP Fellowship Director

16. Somatic Symptom and Related Disorders

A. Chronic pain syndromes and somatic symptom disorder

1. Recognize patterns of chronic or recurrent pain that are commonly seen without evidence of associated tissue damage
2. Know the psychological symptoms and disorders commonly associated with chronic pain
3. Know how to plan initial treatment strategies for a child/adolescent with a recurrent pain syndrome
4. Know the appropriate evaluation of a child with recurrent pains
5. Know the diagnostic criteria for a somatic symptom disorder
6. Know the differential diagnosis for somatic symptom disorder
7. Know the signs and symptoms of complex regional pain syndrome
8. Know the signs and symptoms of fibromyalgia
9. Understand the interaction between psychologic and physiologic factors that produce pain
10. Understand the principles underlying effective medication regimens for the treatment of chronic pain

B. Conversion disorders

1. Know how to plan the management of children/families with somatic symptoms not fully explained by a medical condition
2. Know how to plan the evaluation for a child/adolescent suspected of having a conversion disorder
3. Recognize the common complications of conversion disorders
4. Understand the distinction between primary and secondary gain in conversion disorder
5. Understand the etiology of conversion disorder
6. Understand the importance of symptom modeling in the development of conversion symptoms (eg, pseudoseizures in a patient with seizures)
7. Understand the natural history of conversion symptoms
8. Understand the role of psychosocial stressors in conversion disorder
9. Understand the role of social and cultural factors in the development of somatic symptoms
10. Understand that the particular manifestations of a conversion disorder result from an unconscious process
11. Understand that conversion disorder rarely presents prior to 5 years of age
12. Understand the characteristics and management of group conversion disorder (ie, mass hysteria)

C. Factitious disorder

1. Differentiate between a conversion disorder and a factitious disorder

D. Dissociative disorders

1. Recognize the symptoms of a dissociative disorder

16. Somatic Symptom and Related Disorders

A. Chronic pain syndromes and somatic symptom disorder

1. Recognize patterns of chronic or recurrent pain that are commonly seen without evidence of associated tissue damage
- Allodynia = pain with a nonpainful stimulus
- Hyperalgesia = disproportionate pain with a painful stimulus
- Pain in multiple areas of the body that cannot be explained by a medical condition (i.e. labs and/or imaging are negative)
- Ongoing pain usually for longer than 3 months

2. Know the psychological symptoms and disorders commonly associated with chronic pain
- Sleep problems/fatigue
- Anxiety/depression (mood disorders), potential associated suicidal thoughts
- Childhood neglect or abuse, history of other trauma (or PTSD)
- Alcohol or drug abuse

3. Know how to plan initial treatment strategies for a child/adolescent with a recurrent pain syndrome
- Stress coping skills rather than a cure
- Spend time listening and acknowledge that what they are feeling is real
- Emphasize the mind-body connection; avoid comments such as “there is nothing medically wrong with you”
- Ensure participation in school, exercise, regular sleep, engaging in self-care (i.e. hygiene, drinking enough water) (Mayo Clinic)
- Limit diagnostic testing and referrals to subspecialists and provide reassurance that there are no concerns for serious medical diseases
- Behavioral interventions: Cognitive Behavior Therapy with biofeedback, Mindfulness-based therapy

https://www.aafp.org/afp/2016/0101/p49.pdf (Somatic Symptom Disorder article from American Academy of Family Physicians)

4. Know the appropriate evaluation of a child with recurrent pains
- Limit lab testing especially if the person has had a thorough prior workup
- Differential diagnosis (other than behavioral): rheumatologic, orthopedic, or neurologic
- Blood/urine tests: chemistry panel, thyroid testing, CBC, markers of inflammation/autoimmune function, muscle damage (CK), urine drug screen
- Imaging studies: x-ray, CT, MRI, bone scan, EMG
- Usually these blood tests are normal in children with somatic symptom disorder, fibromyalgia, or complex regional pain syndrome (these are diagnoses of exclusion)

5. Know the diagnostic criteria for a somatic symptom disorder
- Characterized by somatic symptoms (one or more physical symptoms) that are either very distressing or result in significant disruption of functioning
- Excessive thoughts, disproportionate feelings, and behaviors regarding those symptoms
- Symptoms must be present for 6 months or longer and can be associated with a known medical condition (DSM 5/American Psychiatry Association)

6. Know the differential diagnosis for somatic symptom disorder

- Excessive thoughts: adjustment disorder, body dysmorphic disorder, obsessive-compulsive disorder, and illness anxiety disorder
- Functional disorders of unclear etiology: fibromyalgia and irritable bowel syndrome (Mayo Clinic)

7. Know the signs and symptoms of complex regional pain syndrome

- Chronic peripheral pain in affected extremity, both at rest and worsened by movement
- Allodynia (pain with a nonpainful stimulus) and hyperalgesia (disproportionate pain with a painful stimulus)
- Autonomic changes (changes in skin color, temperature sensitivity, swelling, edema)
- Decrease in hair and nail growth of the affected extremity
- Atrophy, weakness, dystonia, tremors, and spasms of the muscle (possibly due to disuse)
- Anxiety, frequent school absences

8. Know the signs and symptoms of fibromyalgia

- In pediatric cases, number of tender points may be less than 11 that cause some level of impairment in daily functioning for 3 months or longer. Some researchers have proposed a cut-off of ≥5 tender points for pediatric fibromyalgia.
- Fatigue, stiffness, headaches, and anxiety
- Non-restorative or disrupted sleep is common

9. Understand the interaction between psychologic and physiologic factors that produce pain

- Fibromyalgia potential pathogenesis: Neuroendocrinologic dysfunction affecting the central nervous system is a popular theory.
- Complex regional pain syndrome possible pathogenesis: Disturbances in the central nervous system after initial injury could result in increased connectivity and overactivation in the sensory cortex, emotional processing centers, and pain sensory regions. Peripheral nerve fibers could be damaged by an initial insult, resulting in small-fiber polyneuropathy.
- Understanding and managing the thoughts, emotions and behaviors that accompany the discomfort can help with coping with pain (American Psychology Association)

10. Understand the principles underlying effective medication regimens for the treatment of chronic pain (Table taken from Chronic Pain: Medication Decisions on Mayo Clinic website)

- Antidepressants used: TCAs: amitriptyline and nortriptyline or SNRIs: duloxetine (Cymbalta), venlafaxine (Effexor XR) and milnacipran (Savella)
- Antiepileptics used: gabapentin (Gralise, Neurontin) and pregabalin (Lyrica)

Medication type	How they work	First line option for	Benefits	Risks
NSAIDs	Block COX-1 and COX-2 enzymes involved in pain and inflammation	Mild to moderate pain accompanied by swelling and inflammation Arthritis pain and pain resulting from muscle sprains and strains, back and neck injuries, overuse injuries, and menstrual cramps	When taken as directed, generally safe for short- and long-term use	May cause nausea, stomach pain, stomach bleeding or ulcers When taken in high doses, can lead to kidney problems, fluid retention and high blood pressure Increased risk of side effects for older adults
Acetaminophen	Unknown, but possibly blocks a COX-3 enzyme	Mild to moderate pain	Acetaminophen	Unknown, but possibly blocks a COX-3 enzyme

COX-2 inhibitors	Block COX-2 enzymes	Rheumatoid arthritis, osteoarthritis, menstrual cramps and injury-related pain	As effective as NSAIDs without damaging stomach lining at regular doses	Respiratory infection, headache and dizziness When taken in high doses, can lead to stomach bleeding, kidney problems, fluid retention and high blood pressure Increased risk of side effects for older adults
Antidepressants	Interfere with certain chemical processes that cause you to feel pain	Neuropathic pain, chronic daily headaches, fibromyalgia May be considered for chronic low back pain	Can be in doses much lower than what is currently used to treat depression Side effects generally mild	Drowsiness possible with tricyclic antidepressants Can take several weeks to produce desired effects May worsen depression and cause suicidal thoughts in a small number or people

Anti-seizure medications	Quiet pain signals from damaged nerves	Postherpetic neuralgia, diabetic neuropathy, fibromyalgia	Side effects generally mild	May cause dizziness, drowsiness, nausea, reduced coordination and weight changes May worsen depression and cause suicidal thoughts in a small number of people
Opioids	Activate feel-good neurotransmitters, called endorphins, that suppress pain and boost a sense of well-being	Acute pain, such as pain that follows surgery or a bone fracture Typically prescribed for maximum of three days	Powerful relief during short periods of severe pain	Tolerance, dependence, misuse, addiction and overdose, which may begin to develop within one week of use Responsible for the majority of accidental overdose deaths in the U.S.

B. Conversion disorders

1. Know how to plan the management of children/families with somatic symptoms not fully explained by a medical condition

- Physical symptoms of emotional distress are called **somatic symptoms**. A **Somatic Symptom and Related Disorder (SSRD)** is diagnosed when physical symptoms (eg. pain or fatigue) are not explained by a medical illness or when symptoms of a known illness affect a child much more than expected and these symptoms interfere with daily life such as missing school, not wanting to play with friends, or avoiding fun activities.
- **Management/ Treatment** consists of:
    - validating the patient's symptoms as real to the patient
    - educating the patient about the symptoms

    - remaining available to the patient and family for follow-up and re-assessment
    - and rehabilitating to prior function
    - treatments such as cognitive behavioral therapy and pharmacotherapy for co-existing conditions (eg, anxiety) may be considered
- If the patient is suspected of feigning the symptoms for secondary gain, the diagnosis of factitious disorder or malingering should be considered.

2. Know how to plan the evaluation for a child/adolescent suspected of having a conversion disorder
- Conversion disorder can present differently depending on the age.
    - **Younger prepubertal children** are more likely to have negative symptoms, such as loss of function, weakness, imbalance, vision loss.
    - **Adolescents** are more likely to have positive symptoms, such as nonepileptic movements.
    - Conversion occurs more readily as the patient gains experience with examples of illness and their potential for reward. Hence, it becomes more frequent in adolescence. (Conversion disorder only rarely presents prior to 5 years of age.)
- **Evaluation**:
    - Nondirective interviewing can be productive; ask the patient to explain his or her symptoms, noting the language used and the affect displayed. The diagnosis may lie in the patient's explanation.
    - It is best not to review the systems in the context of the presenting complaint lest new symptoms be encouraged inadvertently to emerge. The review can be undertaken best during the physical examination.
    - A physical examination, with particular attention to the symptomatic site, should be completed no matter how evident the diagnosis of conversion. It generally enables the pediatrician to eliminate other procedures as unnecessary.
    - There are some important diagnostic caveats:
        - Always keep the possibility of conversion in mind. Know the family; conversion occurs more commonly in certain families, both because they model the phenomenon and because they honor it. (There may be underlying genetic predilections as well.)
        - Do not insist on a complete picture. To diagnose conversion, it is not necessary to uncover the unconscious symbolism involved, although this is desirable for confirmation.
        - Limit diagnostic tests to screening procedures as much as possible.
        - Classic conversion patients describe the symptoms in highly colorful body language, and its use should alert the observer.
        - A casual, off-hand response even to apparently devastating symptomatology is helpful diagnostically when it occurs. It is uncommon in children, however, who are anything but indifferent if their symptoms arouse their parents' or the physician's anxiety. Because the symptoms are "contagious," it is understandable that such patients 'catch' the anxiety aroused in an observer by the symptoms.
        - The illness model in families prone to conversion can lead them to be curiously unconcerned about one of their member's symptoms, as if they know at some level whence the symptoms have arisen. By contrast, in other families, concern may be extreme; these families tend to be overprotective and enmeshed.

3. Recognize the common complications of conversion disorders.
- Physical complications/ symptoms of conversion disorder can include the following:

- General: weakness or paralysis, abnormal movements (eg, tremor), a feeling of a lump in the throat, speech alterations (dysarthria, dysphonia), altered skin sensation, and altered vision and hearing.
- Gastrointestinal symptoms: weight loss, dietary deficiency symptoms
- Cardiovascular symptoms: loss of muscle tone, weakness
- Dermatologic symptoms: edema, flushing, vesicles/ rashes
- Respiratory symptoms: hyperventilation causing respiratory alkalosis (with numbness, light headedness, pins-and-needles, tetany, contractures, muscle atrophy, osteoporosis)
- Neurologic Symptoms:
  - Psychogenic nonepileptic seizures (PNES) are a conversion disorder that mimics epileptic seizures and involves alterations in behavior, motor activity, consciousness, and sensation. Psychogenic nonepileptic seizures are not associated with epileptiform activity in the brain.
  - Syncope is the result of altered oxygen delivery to the brain rather than a disturbance in brain electrical function.

4. Understand the distinction between primary and secondary gain in conversion disorder.

- **Primary gain** is the lessening of anxiety by the presence of the symptom
- **Secondary gain** is reward to the patient as an outcome of illness

5. Understand the etiology of conversion disorder

- **Conversion disorder (functional neurological symptom disorder)** is characterized in Diagnostic and Statistical Manual of Mental Disorders (Fifth Edition) (DSM-5) by one or more symptoms of altered voluntary sensory or motor function, not better explained by another medical or mental disorder, and causing clinically significant distress or impairment. In any potential case of conversion, both somatic and psychological factors should be sought from the start. Conversion is a positive diagnosis, not a diagnosis of last resort following exhaustive negative tests.

6. Understand the importance of symptom modeling in the development of conversion symptoms (eg, pseudoseizures in a patient with seizures)

- **Symptom modeling** is a theory that in conversion disorders such as psychogenic nonepileptic seizures (PNES), organic signs and symptoms in the individual or others are mimicked to satisfy primary and secondary gains.
  - The primary gain may be a reduction of anxiety, and the secondary gain may be the attention of others.
  - There is a greater-than-chance occurrence of PNES in individuals with epilepsy.
  - In addition, in a 2008 study by Bautista and colleagues, a significantly larger number of patients with PNES (66 percent) admitted to having witnessed a seizure compared with patients who had epileptic seizures (11 percent). There was no significant difference in the prevalence of seizures between the families in the study.
  - Family modeling and learned behavior theories suggest children can develop conversion symptoms from seeing maladaptive response to illness in family members or being reinforced in their own illness behaviors.
  - Parents reported greater medical vulnerability in children with PNES than in those without, which could be inadvertent reinforcement. Parents of children with PNES have also reported more symptoms of anxiety, depression, and somatization than have parents of control children.

7. Understand the natural history of conversion symptoms

- On an individual level, conversion disorder, also known as functional neurological symptom disorder, is a condition where symptoms are suggestive of a medical illness but cannot be explained with a specific pathophysiology. Symptoms may include seizures, paresthesias, abnormal gait, paresis, and/or other abnormal disorders of movement.
- These symptoms are not intentionally produced, but cause the patient clinically significant distress or impairment in functioning.
- *La belle indifference*, or lack of concern about the nature/implication of the symptom(s), has been associated with conversion disorder; however, it is not definitive of a conversion disorder and is not required to make the diagnosis.
- It may be useful to view conversion as a form of suggestibility, that is, learned symptomatic behavior that rewards the patient sufficiently to be sustained. Such behavior is more likely if the family and medical professionals accept it. Rather than striving to uncover the presumed dynamic underpinnings of the system or becoming overly concerned with formal psychiatric nomenclature, the pediatrician can accept the patient's physical complaint most usefully as a metaphorical expression of emotional distress.

8. Understand the role of psychosocial stressors in conversion disorder
- In any potential cases of conversion: always evaluate somatic, psychological, and psychosocial stressors from the start of the work up.
    - Risk factors: maladaptive personality traits, history of childhood abuse or neglect, and the presence of neurological disease.
    - Comorbidity with psychological conditions is much more common than generally realized.
        - In particular, conversion and **depression** another condition underdiagnosed in youth are frequent partners, although neither usually is recognized or treated. The coexistence of the two conditions is significant because each can exacerbate the other.
        - Unresolved **grief** also promotes conversion.
        - Conversion frequently coexists with **anxiety**, particularly with panic. Anxiety levels can be heightened if conversion symptoms are regarded with gravity; each condition feeds off the other. That conversion and panic coexist is demonstrated readily in hyperventilation syndrome.
            - School refusal-separation anxiety- often incorporates conversion symptoms for its continuance.
        - **Posttraumatic stress disorder** may induce conversion symptoms, often in organs or systems involved in the original stressful incident (eg, "hysterical amaurosis," - conversion blindness-after.
    - Conversion in the parent reinforces conversion in the child. Children of depressed mothers also have an increased likelihood of conversion symptoms. Recent family stress is common.
    - Patients with PNES have reported more lifetime adversities, bullying, and learning struggles.
        - Parents of children with PNES have also reported more symptoms of anxiety, depression, and somatization than have parents of control children.
        - Risk factors for PNES in children include comorbid general medical, neurological, and psychiatric diagnoses.

9. Understand the role of social and cultural factors in the development of somatic symptoms
- Conversion in response to anxiety or conflict probably is more common in those ethnic and cultural populations that support or accept somatic symptoms more readily than psychological symptoms. (The converse also may be true.)
- There is much evidence that social tension, migration, and lower socioeconomic status all are associated with increased incidence of conversion.

- As patients and families become more sophisticated, obvious conversion symptoms are less common, although suggestibility and emotional needs for the rewards that conversion brings may lead to conversion in more subtle forms in patients from intelligent and knowledgeable families.
- The incidence of conversion is also increased after physical and sexual abuse and in children whose parents are seriously ill or have chronic pain.

10. Understand that the particular manifestations of a conversion disorder result from an unconscious process
    - Conversion disorders result from an unconscious process and there must always be clinical findings to show clear evidence that the symptoms are not consistent with neurological disease. A conceptual model proposes that unresolved psychological stress leads to the physical symptoms.

11. Understand that conversion disorder rarely presents prior to 5 years of age
    - Conversion occurs more readily as the patient gains experience with examples of illness and their potential for reward. Hence, it becomes more frequent in adolescence, and would be rare to present prior to 5 years of age.
    - Prior to 5 years of age the traditional diagnostic elements of conversion are difficulty to demonstrate for reasons of developmental immaturity.

12. Understand the characteristics and management of group conversion disorder (ie, mass hysteria)
    - **Mass psychogenic illness is also known as epidemic hysteria, sociogenic illness, or transient situational disturbance. It has been defined as symptoms of an organic illness without an identified organic cause in a cohesive group of people who share the same beliefs about the etiology of the symptoms**.
    - Mass psychogenic illness occurs more often in females than males; it is also commonly seen in groups that are under physical or psychological distress.
    - Mass psychogenic illness occurs as a social phenomenon, where healthy individuals jointly believe an illness is caused by some external factor, commonly an odor. There is an initial primary individual's illness with a response by emergency personnel to tend to the illness. Symptoms can spread rapidly amongst a group of people, often driven by sight, sound, or direct communication (including social media). There may be inconsistency amongst reporters about the offensive external factor with no plausible explanation for the stimuli. A skin rash may be seen, although usually on exposed skin in a distribution consistent with scratching.
    - The differential diagnosis may include bioterrorism, a spreading infectious disease, or acute exposure to toxic agents.
    - Management:
        - Early recognition with proper response can have a significant impact on outcomes.
        - **Once a reasonable work-up has been done to exclude any medical causes, and there is confidence that the outbreak is mass psychogenic illness, further work-up should cease**.
        - The treatment is self-limited and requires a sophisticated level of communication between the physician and patient with gentle but confident reassurance. **When providers and patients do not agree on the reality of the illness, the prognosis is worse; therefore, skillful communication is important**.
        - Skillfully communicating to the patient and family that stress, anxiety, and other factors can lead to physical symptoms is important; providers should avoid phrases like "there's nothing wrong with you."
    - Treatment:
        - **The treatment focus should be on restoring the individual (and group) back to typical functioning as soon as possible**. Negative media attention can add to public health, social, and economic consequences, so involved agencies must coordinate to send a consistent message to the community at large.

- There may be some hesitancy to label the outbreak as psychogenic, as this may create anger and shame, and possibly mistrust in the community.

C. Factitious disorder

1. Differentiate between a conversion disorder and a factitious disorder

- Conversion disorder = one or more symptoms of altered voluntary sensory or motor function, not better explained by another medical or mental disorder, and causing clinically significant distress or impairment. Phenomenon of la belle indifference = the lack of concern about the symptoms (seen in conversion)
- Factitious disorder = considered if the patient is suspected of feigning the symptoms for secondary gain

D. Dissociative disorders

1. Recognize the symptoms of a dissociative disorder

- Experience of detachment or feeling as if one it outside one's body, loss of memory or amnesia
- Frequently associated with previous experience of trauma (American Psychiatry Association)

Content Category 17- Sleep Problems and Sleep-Wake Disorders

ABP Content Specs for Developmental-Behavioral Pediatrics Fellow Collaboration Project

Prepared by: University of Michigan DBP Fellows

Reviewed by: Barbara Felt, MD, University of Michigan-Staff/Faculty DBP

17. Sleep Problems and Sleep-Wake Disorders

A. General
   1. Recognize the cultural variations in acceptance of co-sleeping and sleep expectations
   2. Know the association of sleep disorders with other psychiatric, developmental, and behavioral disorders
   3. Know the different stages of sleep
   4. Understand the changes in sleep cycles with development
   5. Understand the physiology of sleep cycles
   6. Know the behavioral effects of sleep deprivation
   7. Understand the changes in the need for sleep with age
   8. Know the effects of commonly used medications on sleep

B. Night-time awakening
   1. Know the concept of "sleep associations" and its relevance to night-time awakening
   2. Know the management of frequent night awakenings
   3. Know the epidemiology of night-time awakening
   4. Know how to evaluate a child with frequent night-time awakenings

C. Bedtime resistance
   1. Relate the ages of peak bedtime resistance to developmental theory
   2. Know how to manage bedtime resistance in a developmentally appropriate way
   3. Know the classifications of sleep disorders
   4. Know how to evaluate a child with bedtime resistance
   5. Understand factors that contribute to bedtime resistance
   6. Know the epidemiology of bedtime resistance

D. Narcolepsy
   1. Recognize the signs and symptoms of narcolepsy
   2. Know the familial pattern of narcolepsy
   3. Know how to plan the laboratory evaluation of narcolepsy
   4. Know the treatment of narcolepsy

E. Breathing-related sleep-wake disorders
   1. Know the signs and symptoms of obstructive sleep apnea
   2. Understand the impact of obstructive sleep apnea on development and behavior
   3. Know the treatment of obstructive sleep apnea
   4. Differentiate among obstructive sleep apnea, central sleep apnea, and sleep-related hypoventilation

F. Insomnia disorder
   1. Know the criteria for insomnia disorder in children and adolescents
   2. Know the differential diagnosis of insomnia
   3. Know how to plan the management of a child or adolescent with insomnia
   4. Know common pharmacologic management of insomnia

G. Parasomnias
   1. Know the primary parasomnias
   2. Understand the physiology of sleepwalking and night terrors
   3. Know the management of common parasomnias
   4. Know the sleep stages associated with common parasomnias

5. Know the epidemiology of common parasomnias
6. Differentiate between night terrors and nightmares

H. Circadian rhythm sleep-wake disorders
1. Know the criteria for delayed and advanced sleep phase type
2. Know the criteria for irregular sleep-wake type
3. Know the differential diagnosis for circadian rhythm sleep-wake disorders
4. Know the treatment for circadian rhythm sleep-wake disorders

I. Restless legs syndrome
1. Know the criteria for restless legs syndrome
2. Know the differential diagnosis for restless legs syndrome
3. Know the treatment for restless legs syndrome

J. Other sleep problems
1. Know the crtieria for rapid eye movement sleep behavior disorder
2. Know the criteria for substance/medication-induced sleep disorder
3. Know the differential diagnosis for substance/medication-induced sleep disorder

Sleep Problems and Sleep-Wake Disorders (per ABP content specifications):

A. General

1.Recognize the cultural variations in acceptance of co-sleeping and sleep expectations

While bedsharing rates are rising overall in the U.S., it is more common among Black, Hispanic, and Asian infants than for Non-Hispanic White infants. It is more common in families in which the mother is single and younger than 20, and for families with younger infants. In addition, bedsharing is more likely for mothers with low education, in low-income households, and in the U.S. West and South (Salm Ward & Ngui, 2015). Also, breastfeeding mothers and highly educated mothers who engage in "attachment parenting" are likely to bedshare. National Council on Family Relations

2. Know the association of sleep disorders with other psychiatric, developmental, and behavioral disorders

- Children with underlying neurologic disorders including cerebral palsy, intellectual disability, autism spectrum disorder and blindness are more likely to have sleep disorders due to the association of these conditions with neurobehavioral and circadian sleep disruption.
- ADHD-associated behavioral problems and stimulant medications tend to cause sleep disruption.
- Some types of sleep disorders like OSA and restless legs syndrome may cause behavioral symptoms that meet criteria for ADHD (inattention, hyperactivity, impulsivity, and irritability). Often when the sleep disorder is treated the behavioral symptoms resolve.

3. Know the different stages of sleep

	WAKE	NREM SLEEP			REM SLEEP
Stage of Sleep:	Stage 0	LIGHT SLEEP: Stage 1, Stage 2		DEEP SLEEP: Stage 3	Stage R
Description:	Eyes open, responsive to external stimuli, can hold intelligible conversation	Transition between waking and sleep. If awakened, person will claim was never asleep.	Main body of light sleep. Memory consolidation. Synaptic pruning.	Slow waves on EEG readings.	Brain waves similar to waking. Most vivid dreams happen in this stage. Body does not move.
Time Spent In:	16 to 18 hours per day	4 to 7 hours per night			90 to 120 min/night

4. Understand the changes in sleep cycles with development

Infants *— Sleep in the healthy, full-term newborn is distinguished from that of older individuals by:*

- *Longer sleep duration (16 to 18 hours per 24 hours)*
- *Rapid eye movement (REM) sleep occurring at sleep onset*
- *Increased proportion of REM sleep*

- *REM-nonrapid eye movement (NREM) cycle much shorter in duration as compared with older individuals*

With maturation of the child's central nervous system, predictable changes occur, including gradual decrease in total sleep time and the proportion of REM sleep, progressive lengthening of the REM-NREM cycle, and shift to the adult pattern of sleep onset via NREM sleep.

***Children and adolescents** — In normal older children and adolescents, sleep is characterized by:*

- *Onset via NREM sleep*
- *NREM sleep occupying approximately 75 percent of total sleep time*
- *REM and NREM sleep alternating throughout the night with a period of 90 to 100 minutes, and a progressive lengthening of the duration of REM sleep periods in the final one-third of the night*

Sleep in adolescents is further characterized by:

- *Decrease in slow-wave sleep beginning in puberty and continuing into adulthood*
- *Physiologic shift in sleep onset to a later time*
- *Increasing irregularity of sleep-wake patterns (primarily discrepancy between weeknights and weekend sleep patterns)*
- *Decrease in average sleep **duration** despite relatively stable sleep **requirement** of approximately nine hours*

Source: https://www.uptodate.com/contents/sleep-physiology-in-children?search=sleep%20cycle&source=search_result&selectedTitle=8~150&usage_type=default&display_rank=8

5. Understand the physiology of sleep cycles

Stage of Sleep	EEG (brain wave patterns	EMG (muscle tension)	EOG (eye movement s)	Other Physiological signs
Alert	Beta waves High frequency (13-24 cps) High amplitude	active	active	Normal
Waking (relaxed)	Brain activity begins to slow to Alpha waves Slower frequency 8-12 cps	High amplitude	Active, may be spiky with eye blinks	Normal, relaxed
Stage 1 (drowsy)	Only a few minutes Low amplitude, mixed frequency Theta waves (4-7 cps)	High amplitude, but not distorted Hypnic jerks	May be rolling	People will deny having been asleep Breathing, heart rate, blood pressure start to drop
Stage 2 (light sleep)	Theta waves continue Sleep spindles (low amplitude, high frequency) K-complexes (slow, high amplitude waves)	Medium amplitude	No eye movement	Breathing, heart rate, blood pressure, body temperature continue to drop
Stage 3 (SWS – deep sleep)	Delta waves High amplitude, low frequency rhythmic waves Delta waves make up 20% -50% of brain waves	Medium or low amplitude, muscles relax	No eye movement	Breathing, heart rate, blood pressure, body temperature continue to drop
Stage 4 (SWS – very deep sleep)	Delta waves are more than 50% Deepest stage of sleep	Relaxed	No eye movement	Lower body temp, respiration is decreased Harder to wake someone If awoken, confused, disoriented
REM sleep - -	Similar to stage 1. Beta and some alpha waves	Low Muscles are relaxed	Sharp intermittent eye movements L to R and up and down	Pulse rate, blood pressure quicken, respiration faster People report dreaming when woken

6. Know the behavioral effects of sleep deprivation

Motor hyperactivity is one of the most consistently reported symptoms in a number of sleep disorders, particularly in younger children, and may be construed as a behavioral compensation for an internal sense of sleepiness (in an attempt to remain awake), compounded by behavioral dysregulation. Accordingly, there is an overlap between sleep disorders including OSA and symptoms of attention deficit hyperactivity disorder (ADHD).

Sleep loss promotes risk-taking and sensation-seeking behavior. A number of studies, including some in adolescents, have suggested that insufficient sleep is linked to changes in reward-related decision-making, so that sleep-deprived individuals tend to take greater risks and are less concerned about the potential negative consequences of their behavior. These effects are mediated through the striatum (caudate, putamen, and nucleus accumbens), which is part of the basal ganglia and is particularly important for motivation and reward-related function. Risk behaviors during adolescence, such as using drugs, driving while impaired, or self-harm behaviors, depend on a complex relationship between risk perception, assessment of the cost/benefit ratio of risky behaviors, and reward salience.

Daytime behavioral disinhibition, including oppositional, defiant and aggressive behavior, and noncompliance as reported by teachers and parents, has been associated with disordered sleep in children. Sleepy school children more often show bullying behavior and receive discipline referrals compared with their peers. However, it is important to recognize that while children with poor

quality and/or insufficient sleep demonstrate more problematic behavior, the converse may also be true. For example, an average of 30 minutes of sleep extension in school-aged children was reported to be associated with improvements in students' emotional lability and restless/impulsive behavior as rated by their teachers, and with decreased aggressive and oppositional behavior in school.

The behavioral effects of sleep loss may be further exacerbated by "weekend oversleep." This phenomenon is common among teens who are exposed to chronic sleep loss during the week and who attempt to compensate by shifting bed and wake times later and extending sleep on weekends, causing progressive disruption of circadian rhythms.

SOURCE: https://www.uptodate.com/contents/cognitive-and-behavioral-consequences-of-sleep-disorders-in-children?search=behavioral%20effects%20of%20sleep%20deprivation&sectionRank=1&usage_type=default&anchor=H16053978&source=machineLearning&selectedTitle=1~150&display_rank=1#H16053978

7. Understand the changes in the need for sleep with age

Recommended sleep times for children

Age group	Recommended sleep time
Infants 4 to 12 months	12 to 16 hours (including naps)
Toddlers 1 to 2 years	11 to 14 hours (including naps)
Children 3 to 5 years	10 to 13 hours (including naps)
Children 6 to 12 years	9 to 12 hours
Teens 13 to 18 years	8 to 10 hours

For optimal health, daytime functioning, and development, the above sleep times are recommended on a regular basis. These consensus recommendations were made by the American Academy of Sleep Medicine[1] and endorsed by the American Academy of Pediatrics[2].

References:

1. *Paruthi S, Brooks LJ, D'Ambrosio C, et al. Recommended Amount of Sleep for Pediatric Populations: A Consensus Statement of the American Academy of Sleep Medicine. J Clin Sleep Med 2016; 12:785.*
2. *Recommended Amount of Sleep for Pediatric Populations. Pediatrics 2016; 138.*

8. Know the effects of commonly used medications on sleep
- Stimulants can delay sleep onset
- SSRIs may increase awakenings and abrupt withdrawal may worsen insomnia
- Alcohol leads to shortened sleep latency but can increase insomnia later in the night

B. Night-time awakenings

1. Know the concept of "sleep associations" and its relevance to night-time awakening.

   A sleep association is any action that helps a child or baby fall asleep. Children of all ages, and even adults, have sleep associations. Sleep associations can be positive or negative. For an infant or child, a negative sleep association requires someone else to do something for them to fall asleep. These actions are considered “negative” because when a child wakes up in the middle of the night, they have a hard time going back to sleep without it. A positive sleep association is something that a baby can do to fall asleep on his or her own.

2. Know the management of frequent night awakenings

Management of frequent night awakenings depends on evaluation including a comprehensive history and physical exam. Things to consider include:

The parent response to nighttime awakenings *– When children experience nighttime awakenings, the parents' response can promote or extinguish the behavior. As an example, excessive parental attention in response to nighttime awakenings (including routinely allowing the child to switch beds) may perpetuate the problem. The clinician should assess whether the parents' response to nighttime awakenings is likely to reinforce the behavior, and should offer advice on how to reverse this pattern. For healthy toddlers and older infants, habitual nighttime feedings may disrupt sleep and are unnecessary. Clinicians should inquire about nighttime feedings and offer guidance on eliminating these when appropriate.*

Other contributors *— The following problems contribute to insomnia in some children. In such cases, the insomnia is unlikely to respond to behavioral therapy alone until these issues are addressed.*

- ***Psychosocial dysfunction*** *– Psychosocial dysfunction may cause or present as a sleeping problem. Conversely, children's sleeping problems can contribute to household and marital stress. The clinician should probe the psychosocial history, including the presence of marital discord, use of alcohol or drugs by household members, and any indications of child abuse. In some cases, it may be necessary to interview the child alone to get a reliable history.*

- ***Anxiety or depression*** *– Anxiety (including separation anxiety) and depression are common causes of insomnia in children. All children have fears at some point in their lives, and these may interfere with sleep. If the fears are persistent and consistently interfere with functioning (ie, sleep), evaluation for a specific phobia or generalized anxiety disorder may be warranted. The prevalence of depression is approximately 2 percent among school-aged children and rises sharply during adolescence. In addition to insomnia, symptoms include depressed or irritable mood, diminished interest or pleasure (anhedonia), change in appetite or weight status, psychomotor agitation or retardation (eg, talking or moving more slowly than is usual for them), fatigue or loss of energy, feelings of worthlessness or guilt, impaired concentration, or recurrent thoughts of death or suicide.*

●***Concomitant medical problems*** *– The clinician should identify chronic medical problems that may influence sleep, including chronic or recurrent pain, symptoms suggestive of gastroesophageal reflux, breathing problems during wakefulness or sleep, and the medication history. Such medical problems may disrupt sleep because of discomfort or because of medical interventions (eg, medications, breathing treatments, or feedings) that are given during the night. Some medications, including stimulants used for attention deficit hyperactivity disorder (ADHD), may affect sleep latency and continuity.*

SOURCE: https://www.uptodate.com/contents/assessment-of-sleep-disorders-in-children?search=frequent%20night%20awakenings&topicRef=6353&source=related_link

3. Know the epidemiology of night-time awakening

Behavioral sleep problems are found in all age groups:

●*Infants and toddlers – Night awakenings are one of the most common sleep problems in infants and toddlers; 25 to 50 percent of children over the age of six months continue to awaken during the night. Bedtime resistance is found in 10 to 15 percent of toddlers.*
●*Preschool-aged children – Up to 20 percent of preschool- and early school-aged children have insomnia symptoms, with an increased risk for those with comorbid medical conditions.*
●*Middle childhood – Although previously thought to be less common in middle childhood, more recent surveys suggest that insomnia symptoms are present in 20 to 40 percent of school-aged children [5], are persistent in a substantial percentage, and appear more common in girls than boys (at age 11 to 12 years) [6]. Consistent with previous studies, difficulty falling asleep was the most common insomnia complaint.*
●*Adolescents – Studies have estimated that 11 percent of adolescents (13 to 16 years of age) have a history of significant insomnia. Children with neurodevelopmental (ie, autism, mental retardation) and psychiatric disorders (ie, depression, anxiety, attention deficit hyperactivity disorder [ADHD]) are at particularly high risk for sleep disturbances.*

SOURCE: https://www.uptodate.com/contents/behavioral-sleep-problems-in-children?search=frequent%20night%20awakenings&source=search_result&selectedTitle=8~150&usage_type=default&display_rank=8

4. Know how to evaluate a child with frequent night-time awakenings

If the primary problem is either bedtime resistance or difficulty initiating or maintaining sleep (sleeplessness or insomnia), behavioral origins are likely. Further evaluation is needed to identify potential contributors and solutions. The sleep history may be facilitated by the use of a screening tool, such as the BEARS survey, which a clinician uses to inquire about five sleep areas (**B**edtime issues, **E**xcessive daytime sleepiness, night **A**wakenings, **R**egularity and duration of sleep, and **S**noring). This type of screen can help identify patients who should be evaluated with a more detailed sleep history.

The sleep history is most contributory when it is structured, detailed, and systematic. The clinician should evaluate the sleep-wake schedule, difficulties initiating or maintaining sleep, abnormal movements or behavior during sleep, presence of snoring, and daytime accompaniments (eg, sleepiness, hyperactivity, inattentiveness, or irritability). The history should include details about the

duration and frequency of the problem, temporal profile of onset (abrupt, gradual, intermittent), and degree of variability from night to night. The clinician should note what interventions or strategies have been tried and whether medications have been used.

Because parents are generally asleep during the night, they may struggle to provide a full history, as they may witness only portions of nighttime events. Regardless of the accuracy of their account, for diagnosis and determining the character of certain behaviors (eg, seizures versus nonepileptic events), evaluation with polysomnography with expanded encephalography montage may be needed.

Completion of a sleep log during the two weeks prior to evaluation may provide important information regarding the sleep-wake pattern and nocturnal events. The log should include bed time, time of sleep onset, awakenings, rise time, nocturnal events, feeding pattern, naps, perceived quality of sleep, degree of alertness or sleepiness during the day, and observations regarding nocturnal events and medical or psychological stressors. The child's sleep patterns can then be compared with typical sleep patterns for his or her age group, although it should be recognized that the average sleep time of children in a given age group varies by as much as two hours.

SOURCE: https://www.uptodate.com/contents/assessment-of-sleep-disorders-in-children?search=frequent%20night%20awakenings&topicRef=6353&source=related_link#H4

C. Bedtime resistance

1. Relate the ages of peak bedtime resistance to developmental theory

Active bedtime resistance (i.e., calling out from or leaving one's room after bedtime) is one of the most common difficulties in young children, seen in approximately 20% to 25% of children 1 to 5 years of age (see Metzler & Mindell, 2004). According to the *Diagnostic and Statistical Manual of Mental Disorders* (*DSM-IV-TR*; American Psychiatric Association, 2000), bedtime resistance may be classified as dyssomnia not otherwise specified, and bedtime problems can be the fundamental component of behavior clusters that are classifiable with other diagnoses (e.g., oppositional defiant disorder, attention deficit hyperactivity disorder). Left untreated, bedtime problems can persist for years (Kataria, Swanson, & Trevathon, 1987). Further, pediatric sleep disturbances are related to various adverse outcomes (e.g., disturbed cognitive functioning, Steenari et al., 2003; increased behavior problems, Paavonen et al., 2002; poor parental satisfaction, Gelman & King, 2001). Thus, effective intervention is important.

SOURCE: Treating Bedtime Resistance with the Bedtime Pass: A Systematic Replication and Component Analysis with 3-Year-Olds
Kurt A Freeman
J Appl Behav Anal. 2006 Winter; 39(4): 423–428. doi: 10.1901/jaba.2006.34-05

2. Know how to manage bedtime resistance in a developmentally appropriate way

Interventions for pediatric bedtime resistance typically are behavioral in nature, the most common being extinction (i.e., planned parental ignoring). Although it is effective, parental acceptability of extinction is often low (France, 1994; Friman et al., 1999), partly due to temporary increases in bedtime resistance often seen early in intervention (i.e., the extinction burst; see Blum & Friman,

2000). Alternatively, the bedtime pass (Friman et al.) involves (a) a small notecard exchangeable for one trip out of the bedroom after being put to bed and (b) extinction.
SOURCE: Treating Bedtime Resistance with the Bedtime Pass: A Systematic Replication and Component Analysis with 3-Year-Olds
Kurt A Freeman
J Appl Behav Anal. 2006 Winter; 39(4): 423–428. doi: 10.1901/jaba.2006.34-05

Specific techniques

Bedtime routines — Establishment of a consistent bedtime routine is helpful for all manifestations of behavioral insomnia (bedtime resistance, prolonged sleep onset, and night wakings) [13]. The routine should last approximately 20 to 45 minutes and include three to four soothing activities, such as taking a bath, changing into pajamas, and reading stories; it should not include television or other electronic devices [25-29]. The introduction at bedtime of more appropriate sleep associations should be readily available to the child during the night and can include transitional objects such as a blanket or toy. The child should be put to bed drowsy but awake to minimize dependence upon parental presence at sleep onset.

An integral part of the bedtime routine is the institution of a bedtime and sleep schedule that ensures a developmentally appropriate amount of sleep. The bedtime should coincide with the child's natural sleep onset time. A consistent nightly bedtime will help to reinforce the circadian clock and enable the child to fall asleep more easily.

Systematic ignoring — Systematic ignoring addresses problems at sleep onset or night waking in which the child needs or demands a parent's assistance. Typically, this occurs when the child demands that the parent stay in the room while he or she falls asleep or when the child wakes the parent for reassurance during the night. The technique typically involves a program of abrupt or gradual withdrawal of parental assistance at sleep onset and during the night. When consistently applied, systematic ignoring usually achieves "extinction" of the need for parental assistance.

- Unmodified extinction ("crying it out") involves putting the child to bed at a designated bedtime and then ignoring the child until a set time the next morning. Although this approach has been documented to be a highly successful treatment, it is often not acceptable to families; parents are often unable to tolerate the child's crying and protest behavior and are less likely to be compliant.

- Graduated extinction is an alternative approach that weans the child from dependence upon parental presence; it involves putting the child to bed drowsy but awake and waiting progressively longer periods of time before checking on the child. On each subsequent night, the initial waiting period before checking is increased by a specified number of minutes. When parents check on their child, they should reassure the child but keep contact brief (one to two minutes) and neutral (eg, pat on shoulder rather than pick up and cuddle). Since the goal of this treatment is to allow the child to fall asleep independently, there is no recommended "optimal" period of time between checks, and the amount of time should be determined by the parents' tolerance for crying and the child's temperament.

A variation on this approach, especially with somewhat older children (preschool-aged and up), is to use positive reinforcement. For example, the caregiver should try to return to the child's bedroom only when he/she is engaged in more appropriate behavior such as remaining in bed instead of climbing out. Another option is to close the child's bedroom door until more appropriate behavior occurs. As an example, a child who becomes more agitated with brief parental checks may do better with infrequent checks.

Graduated extinction is effective even if instituted only at bedtime. Within one to two weeks after the child has learned to fall asleep easily and quickly at bedtime, the self-soothing skills usually generalize to nighttime arousals.

In order to develop a strategy that gradually eliminates adult intervention, the clinician and parents should collaborate to develop a specific plan. They should identify an end goal, such as falling asleep independently at bedtime, and outline successive steps to achieve that goal. For example, a plan might include three days of establishing a bedtime routine and target bedtime, three nights of the parent sitting with the child at the bedside until the child falls asleep, three nights of sitting in the child's bedroom doorway, followed by three nights of sitting outside of the doorway.

More gradual fading of adult intervention may be more appropriate for families that either are unable to tolerate the above extinction approaches or consider them unacceptable.

Bedtime fading — Bedtime fading addresses problems with insomnia at sleep onset, which may be related to a natural "evening" circadian preference with a resulting "mismatch" between the set bedtime and the child's fall asleep time. The technique involves temporarily setting the bedtime to the current sleep onset time and then gradually advancing the time of lights out [30]. The initial bedtime is set to coincide with the natural sleep onset time when the child is more physiologically ready for sleep, and the circadian preference is then gradually modified by setting the bedtime earlier over a period of several weeks.

Bedtime fading may also be used to address a common situation in which the child's "time in bed" exceeds his/her sleep needs (eg, when a parent establishes a set sleep schedule of 12 hours, conflicting with a sleep need of only 10 hours). This results in difficulty falling asleep, prolonged night wakings, or early morning waking. Setting a later bedtime, which provides a "sleep window" that approximates sleep needs, often eliminates the problem.

Strategic napping — Napping schedules should take into consideration normal developmental daytime sleep patterns, 24-hour sleep needs (nocturnal plus daytime sleep), and sleep drive. Children typically need at least four hours between sleep periods in order to build up enough of a sleep drive to allow them to fall asleep again. Thus, naps that are too close together, too long in duration in relation to nighttime sleep, or too late in the day can result in insomnia complaints.

Positive reinforcement — Reinforcement strategies, such as sticker charts, can be beneficial with preschoolers and older children. Such systems are most effective if rewards can be earned immediately. For example, the sticker reward should be given first thing in the morning if the child has met the goal. In addition, the goals must be obtainable to reinforce success. For example, a child may initially earn a sticker just for sleeping in his/her own bed all night, even in the face of frequent calls to parents. With time, more challenging goals can be implemented.

For school-aged children, the rewards can be modified as appropriate to the child's interests, but they should still be concrete and immediate. Multiple small rewards are generally more effective than fewer larger rewards.

Healthy sleep practices for children

1. Have a set bedtime and bedtime routine for your child.
2. Bedtime and wake-up time should be approximately the same time on school nights and non-school nights. There should not be more than about an hour difference from 1 day to another.
3. Make the hour before bed shared quiet time. Avoid high-energy activities, such as rough play, and stimulating activities, such as watching television or playing computer games, just before bed.
4. Don't send your child to bed hungry. A light snack (such as milk and cookies) before bed is a good idea. Heavy meals within an hour or 2 of bedtime, however, may interfere with sleep.
5. Avoid products containing caffeine. These include caffeinated sodas, coffee, tea, and chocolate.
6. Make sure your child spends time outside every day whenever possible and is involved in regular exercise.
7. Keep your child's bedroom quiet and dark. A low-level night light is acceptable for children who find completely dark rooms frightening.
8. Keep your child's bedroom at a comfortable temperature during the night (approximately 65°F).
9. Don't use your child's bedroom for time-out or punishment.
10. Avoid use of electronic media devices (televisions, laptop computers, smartphones) for at least 1 hour before bedtime, and keep these devices out of the bedroom. Children can easily develop the bad habit of using social media after bedtime or "needing" the television to fall asleep. It's much more difficult to control your child's electronic media if the devices are in the bedroom.

Source: Owens J, Mindell J. Sleep Hygiene: Healthy Sleep Habits for Children and Adolescents. In: A Clinical Guide to Pediatric Sleep, 2nd ed, Lippincott, Williams & Wilkins, Philadelphia 2010.

3. Know the classifications of sleep disorders

The International Classification of Sleep Disorders (ICSD) is the most widely used classification system for sleep disorders. The third edition of the ICSD (ICSD-3) includes seven major categories of sleep disorders:

- Insomnia
  - chronic versus short term
- Sleep-related breathing disorders
  - central sleep apnea
  - OSA
  - sleep-related hypoventilation disorders
- Central disorders of hypersomnolence
  - narcolepsy
  - idiopathic hypersomnia
  - Klein-Levin syndrome
  - other

- Circadian rhythm sleep-wake disorders
- Parasomnias
  - NREM-related – disorders of arousal (e.g. confusional arousals, sleepwalking, sleep terrors, sleep-related eating disorder)

- REM-related – intrusion of the features of REM sleep into wakefulness (e.g. sleep paralysis), exaggeration of features of REM sleep (e.g. nightmare disorder) or aberrations of REM sleep physiology (e.g. lack of atonia)

●Sleep-related movement disorders
- Restless legs syndrome
- Periodic limb movement disorder

●Other sleep disorders

4. Know how to evaluate a child with bedtime resistance

Clinical evaluation for children with suspected behavioral sleep problem

Detailed description of the sleep problem(s)
▪ Type of problem (eg, bedtime resistance, difficulty initiating sleep, and/or nighttime awakenings)
▪ Onset, frequency, and duration
▪ Child's usual sleep schedule (weekdays and weekends)
▪ Sleeping environment, including presence of siblings, television, or other noise or distraction
▪ Pre-sleep activities, including exercise, video games, or other stimulating activities
▪ Bedtime routine
▪ Parents' response to the problem and previous treatment attempts
Potential psychosocial contributors
▪ Potential triggers at time of onset (eg, change in schedule, stressful family event, or birth of a sibling)
▪ Other psychosocial triggers (eg, marital discord, mental health problems, or medical illness [in parents, child, or other family members])
▪ Child's developmental history, temperament, and any behavioral problems
▪ Screen time, including time of day
Potential biologic contributors (may also have psychosocial effects)
▪ Other primary sleep disorders, especially: • Obstructive sleep apnea – Symptoms may include frequent snoring; loud snoring; observed pauses in breathing; mouth breathing; or daytime hyperactive, inattentive, or somnolent behavior • Restless legs syndrome – Symptoms include the urge to move the legs (especially in the evening and at rest), leg discomfort, restless sleep, and kicking movements during sleep
▪ Child's medical history, especially disorders that may interfere with sleep, such as allergies and atopic dermatitis, seizure disorders, or rheumatologic conditions
▪ Medications or caffeine, especially stimulants or psychoactive medications
▪ Family history for insomnia or psychopathology
Additional focused history for adolescents with insomnia
▪ Bedtime, sleep onset, and waking time, especially on weekends versus school days
▪ Access to and use of electronics in bedroom (computer, television, e-readers)
▪ Medications or caffeine, specially stimulants or psychoactive medications; drug and/or alcohol use
▪ Preferred sleep schedule and if insomnia resolves when sleeping on preferred schedule
▪ Anxiety about falling or staying asleep and anticipated consequences
▪ Activities in bed other than sleeping (doing homework; use of social media, phone, texting, social games)
▪ Daytime napping (when and for how long)
▪ Daytime sleepiness (dozing off at school, while doing homework, on car rides)

Courtesy of Judith Owens, MD, PhD.

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5. Understand factors that contribute to bedtime resistance

Insomnia related to inadequate parental limit-setting — The limit-setting type of behavioral insomnia is most common in children preschool-aged and older. It is characterized by active resistance, verbal protests, and repeated demands at bedtime ("curtain calls") rather than night wakings. If sufficiently prolonged, the sleep onset delay may result in inadequate sleep. Some children present with nighttime fears characterized by fearful behaviors (eg, crying, clinging, or leaving the bedroom to seek parental reassurance), but these are a manifestation of bedtime stalling rather than anxiety.

This disorder most commonly develops from a caregiver's inability or unwillingness to set consistent bedtime rules and enforce a regular bedtime. The problem is often exacerbated by the child's oppositional behavior. In some cases, however, the child's resistance at bedtime reflects an underlying problem in falling asleep caused by other factors such as asthma, medication use, or other medical conditions; a sleep disorder, such as restless legs syndrome or anxiety; or a mismatch between the child's intrinsic circadian preferences ("night owl") and parental expectations.

SOURCE: https://www.uptodate.com/contents/behavioral-sleep-problems-in-children/print#:~:text=In%20some%20cases%2C%20however%2C%20the,the%20child's%20intrinsic%20circadian%20preferences%20(

6. Know the epidemiology of bedtime resistance

Behavioral sleep problems are found in all age groups:

●*Infants and toddlers – Night awakenings are one of the most common sleep problems in infants and toddlers; 25 to 50 percent of children over the age of six months continue to awaken during the night. Bedtime resistance is found in 10 to 15 percent of toddlers.*

●*Preschool-aged children – Up to 20 percent of preschool- and early school-aged children have insomnia symptoms, with an increased risk for those with comorbid medical conditions.*

●*Middle childhood – Although previously thought to be less common in middle childhood, more recent surveys suggest that insomnia symptoms are present in 20 to 40 percent of school-aged children [5], are persistent in a substantial percentage, and appear more common in girls than boys (at age 11 to 12 years) [6]. Consistent with previous studies, difficulty falling asleep was the most common insomnia complaint.*

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D. **Narcolepsy**

1. Recognize the signs and symptoms of narcolepsy
    - Core features: excessive daytime sleepiness (≥3 months), cataplexy, hypnagogic (upon falling asleep) hallucinations, sleep paralysis
    - Additional signs/symptoms: sleep disturbance (night waking, PLMs, REM sleep behavior disorder), depression, anxiety, inattention, poor academic performance, aggression
    - May be associated with obesity and precocious puberty

2. Know the familial pattern of narcolepsy
    - Sporadic/no specific inheritance pattern but increased risk (40x) in first degree relatives of individuals who have narcolepsy with cataplexy (type I)
3. Know how to plan the laboratory evaluation of narcolepsy
    - Polysomnogram (PSG; sleep study)
    - Multiple sleep latency test (MSLT)
        - ≤ 8-minute mean sleep latency
        - ≥ 2 sleep-onset REM periods
    - CSF orexin (hypocretin-1) – maintains alertness
        - narcolepsy type 1 = cataplexy + CNS orexin deficiency
    - Genetic testing – not clinically specific; most individuals with narcolepsy type I positive for HLADQB*0602 haplotype
4. Know the treatment of narcolepsy
    - Sleep hygiene (regular sleep/wake cycle)
    - Safety precautions (swimming, driving, etc)
    - Scheduled naps (associated school supports – 504 plan/IEP)
    - Daytime physical activity
    - Medications
        - Stimulants (methylphenidate/amphetamine salts)
        - Modafinil
        - Sodium oxybate (gamma-hydroxybutyrate (GHB)) – treatment for daytime sleepiness and cataplexy
        - Atomoxetine
        - SSRI/SNRI/Tricyclic antidepressants also used for cataplexy

E. **Breathing-related sleep-wake disorders**

- Know the signs and symptoms of obstructive sleep apnea
    - Snoring
    - Pauses in breathing/gasping
    - Mouth breathing/dry mouth
    - Positional sleep, with neck arched
    - Sweaty
    - Restless
    - Nighttime awakenings
    - Morning headaches
    - Daytime tiredness
- Understand the impact of obstructive sleep apnea on development and behavior
    - Attentional difficulties and increased activity
    - Behavioral problems (impulsivity, aggression, oppositionality)
    - Mood concerns (depression)
    - Academic challenges (executive functioning and memory difficulties)
    - Cardiovascular consequences (hypertension, cor pulmonale)
    - Failure to thrive
- Know the treatment of obstructive sleep apnea
    - Adenotonsillectomy (first line)

- CPAP
- Orthodontic procedures/devices (maxillary expansion, mandibular advancement)
- Additional therapies: allergy reduction (intranasal corticosteroids/leukotriene modifier), weight loss

- Differentiate among obstructive sleep apnea, central sleep apnea, and sleep-related hypoventilation
  - Obstructive sleep apnea: sleep related upper airway obstruction; apnea-hypoxnea index (AHI) ≥1 on PSG for children
  - Central sleep apnea: sleep related absent respiratory effort; impaired autonomic function; cessation of airflow without evidence of respiratory effort on PSG; example – central hypoventilation syndrome
  - Sleep-related hypoventilation: hypoventilation (increased CO2) during sleep; transcutaneous or end-tidal CO2 > 50mmHg for > ¼ time asleep on PSG

F. **Insomnia disorder**

1. Know the criteria for insomnia disorder in children and adolescents
   - Insomnia – difficulty with sleep onset, maintenance or duration
   - Behavioral insomnia of childhood – based on parental report
     - Sleep-onset association type
       - Delayed sleep onset and/or disrupted sleep
       - Specific process/conditions required to fall asleep
       - Negative/problematic associations
       - Nighttime awakenings require caregiver intervention
     - Limit-setting type
       - Delayed sleep onset and/or disrupted sleep
       - Bedtime resistance (“curtain calls”)
       - Lack of appropriate caregiver limit setting
     - Combined type
2. Know the differential diagnosis of insomnia
   - Behavioral insomnia of childhood
   - Circadian rhythm sleep disorder, delayed sleep phase type
   - Medication associated insomnia (such as in the setting of ADHD stimulant use)
   - Transient sleep disturbances (associated with life stressors – illness, travel, etc)
3. Know how to plan the management of a child or adolescent with insomnia
   - Behavioral interventions first line
   - Sleep hygiene
     - Maintain regular sleep/wake cycle
     - Avoid caffeine after 2pm
     - Limit electronics before bedtime and in room
     - Bedroom just for sleeping
     - Calming bedtime routine
     - Dark and cool bedroom
   - For behavioral insomnia, sleep-onset association type:
     - Extinction method (aka “cry it out” method)
     - Graduated extinction (aka Ferber method)
     - Fading

- For behavioral insomnia, limit-setting type:
    - Authoritative parenting supports
    - Bedtime pass system (paired with incentives)

4. Know common pharmacologic management of insomnia
    - Non-prescription medications:
        - Melatonin – not FDA regulated; useful for sleep-onset insomnia and circadian rhythm delay
        - Antihistamines (ie. Diphenhydramine, Hydroxyzine) – short term/occasional use
    - Common off-label medications:
        - Alpha-adrenergic agonists (ie. Clonidine, Guanfacine) – limited empirical evidence
        - Trazodone – useful for insomnia with comorbid mood issues; lacks support for use in children

G. **Parasomnias**

1. Know the primary parasomnias
    - Nightmares – peak 6-10yo; disturbing vivid dreams; alter and conscious; consolable; event recall
    - Night terrors – peak 4-12yo; genetic predisposition; awakening from sleep, screaming, sweating, flushed, tachycardia, tachypnea; unable to console; amnesia of event
    - Confusional arousal – peak 2-5yo; typically lasting 5-30 minutes; sitting up, distressed, crying out; no sweating, flushing, or stereotyped movements; not consolable; amnesia of event
    - Sleepwalking – peak 8-12yo; walking while asleep and altered consciousness (difficult to arouse, confused, event amnesia, inappropriate behaviors)
    - Sleep paralysis – atonia and paralysis of skeletal muscle; alert and conscious
    - Nocturnal enuresis – >5yo; nighttime incontinence ≥2x/week
2. Understand the physiology of sleepwalking and night terrors
    - Sleepwalking and night terrors occur during partial arousal from non-REM sleep (typically at transition from deep to lighter non-REM); associated autonomic dysfunction; no motor inhibition/atonia such as in REM
3. Know the management of common parasomnias
    - Nightmares
        - Sleep hygiene
        - Reassurance
        - Limiting exposure to violent/scary events or content
    - Night terrors
        - Sleep hygiene (adequate sleep duration)
        - Scheduled awakenings 15-30 minutes before events
        - Safety precautions (door locks, alarms, etc)
    - Sleep walking
        - Safety precautions (door locks, alarms, etc)
        - No need to awaken or restrain
    - Nocturnal enuresis
        - Bedwetting alarm

    - Desmopressin
4. Know the sleep stages associated with common parasomnias
    - Non-REM Parasomnias: more common first third of night
        - Night terrors
        - Sleepwalking
        - Confusional arousals
    - REM Parasomnias: more common last third of night
        - Nightmares
        - Sleep paralysis
        - REM sleep behavior disorder
    - Nocturnal enuresis may occur in non-REM or REM
5. Know the epidemiology of common parasomnias
    - Night terrors – 40%
    - Sleepwalking – 15%
    - Nocturnal enuresis – 25%
6. Differentiate between night terrors and nightmares

Night Terrors	Nightmares
Non-REM sleep	REM-sleep
First third of the night (earlier)	Last third of the night (later)
Confused, disoriented	Alert, scared/upset
Unable to be comforted	Able to be comforted
Typically unable to recall	Remembers dream
Returns to sleep quickly	Difficulty falling back asleep

H. **Circadian rhythm sleep-wake disorders**

1. Know the criteria for delayed and advanced sleep phase type
    - Delayed sleep phase
        - Chronic (≥ 3 months)
        - Inability to fall asleep at conventional/desired time, as well as difficulty awakening at acceptable/desired time
        - If allowed preferred sleep schedule (such as weekends/summer/school breaks), appropriate sleep duration and quality
        - Stable delayed sleep schedule for ≥1 week per actigraphy/sleep diary
    - Advanced sleep phase
        - Chronic (≥ 3 months)
        - Inability to stay awake until conventional/desired time, as well as early awakening
        - If allowed preferred sleep schedule (such as weekends/summer/school breaks), appropriate sleep duration and quality
        - Stable advanced sleep schedule for ≥1 week per actigraphy/sleep diary
2. Know the criteria for irregular sleep-wake type
    - Irregular sleep-wake type
        - Chronic (≥ 3 months)
        - Irregular sleep-wake pattern within 24 hours, with insomnia and/or daytime tiredness

        - Stable irregular bouts (≥ 3) of sleep within a 24 hours without major sleep period for ≥1 week per actigraphy/sleep diary
3. Know the differential diagnosis for circadian rhythm sleep-wake disorders
    - Insomnia
    - Depression/anxiety
4. Know the treatment for circadian rhythm sleep-wake disorders
    - Sleep hygiene (regular sleep/wake schedule)
    - Light therapy
    - Melatonin - ~1.5-3 hours before bedtime
    - Phase advancement (if < 3 hours): move up bedtime by 15 minutes every 3 nights
    - Phase delay "chronotherapy" (if > 3 hours): delay bedtime by 2 hours every day

I. **Restless legs syndrome**

1. Know the criteria for restless legs syndrome
    - Urge to move lower extremities, often with/because of discomfort/unpleasant sensation (described in patient's own words)
        - Worse at rest and at night
        - Relieved by movement
    - Periodic limb movements (PLMs) > 5 per hour on PSG (or family history of such)
    - First degree relative with RLS or PLMD
2. Know the differential diagnosis for restless legs syndrome
    - Periodic limb movement disorder (PLMD); RLS supersedes PLMD
    - Sleep-related rhythmic movement disorder
    - Seizure-disorder (ESES, Benign Rolandic Epilepsy)
3. Know the treatment for restless legs syndrome
    - Sleep hygiene
    - Iron supplementation with ferrous sulfate
        - Iron studies (CBC, serum iron, TIBC and ferritin) obtained initially and repeated every 3 months
            - Iron supplementation suggested for ferritin < 50mcg/L
    - No FDA approved medications for pediatric RLS
        - Off label use of gabapentin, dopamine agonists, clonidine, etc

J. Other sleep problems

1. Know the criteria for rapid eye movement sleep behavior disorder

The specific *DSM-5* criteria for rapid eye movement sleep behavior disorder are as follows: [5]

- Recurrent episodes of arousal during sleep associated with vocalization and/or complex motor behaviors that arise during rapid eye movement (REM) sleep
- On waking from these episodes, the individual is not confused or disoriented and is completely alert
- Either of the following is present: REM sleep without atonia on polysomnographic recordings; or a history suggestive of REM sleep behavior disorder and an established synucleinopathy diagnosis (e.g., Parkinson's disease, multiple system atrophy)

- The episodes cause significant distress or impairment in social, occupational or other areas of functioning which may include serious injury to self or the bed partner
- The disturbance cannot be explained by the effects of a drug of abuse or medication
- The episodes cannot be attributed to another mental disorder or medical condition

2. Know the criteria for substance/medication-induced sleep disorder

Substance/medication-induced sleep disorder *is a sleep disorder characterized by a severe change to sleeping patterns enough to warrant independent clinical attention and judged to be primarily caused by the pharmacological effects of a substance (i.e., a drug of abuse, a medication, toxin exposure). Depending on the substance involved, one of four types of sleep disturbances is reported. Insomnia type and day time sleepiness type are most common, while parasomnia-type is seen less often. The mixed type is noted when more than one type of sleep disturbance-related symptom is present and none predominates. As discontinuation/withdrawal states for some substances can be protracted, onset of the sleep disturbance can occur 4 weeks after cessation of substance use, and the disturbance may have features atypical of other sleep disorders (e.g., atypical age at onset or course).*

Prominent and severe sleep disturbances can occur in association with intoxication with the following classes of substances: alcohol; caffeine; cannabis; opioids; sedatives, hypnotics, or anxiolytics; stimulants (including cocaine); and other (or unknown) sub stances. Prominent and severe sleep disturbances can occur in association with withdrawal from the following classes of substances: alcohol; caffeine; cannabis; opioids; sedatives, hypnotics, or anxiolytics; stimulant (including cocaine); tobacco; and other (or unknown) substances. Some medications that invoke sleep disturbances include adrenergic agonists and antagonists, dopamine agonists and antagonists, cholinergic agonists and antagonists, serotonergic agonists and antagonists, antihistamines, and corticosteroids.

Criteria:

A. A prominent and severe disturbance in sleep.

B. There is evidence from time history, physical examination, or laboratory findings of both (1) and (2):

    1. The symptoms in Criterion A developed during or soon after substance intoxication or after withdrawal from or exposure to a medication.

    2. The involved substance/medication is capable of producing the symptoms in Criterion A.

C. The disturbance is not better explained by a sleep disorder that is not substance/medication-induced. Such evidence of an independent sleep disorder could include the following: the symptoms precede the onset of the substance/medication use; the symptoms persist for a substantial period of time (e.g., about 1 month) after the cessation of acute withdrawal or severe intoxication; or there is other evidence suggesting the existence of an independent non-substance/medication-induced sleep disorder (e.g., a history of recurrent non-substance/medication-related episodes).

D. The disturbance does not occur exclusively during the course of a delirium.

E. The disturbance causes clinically significant distress or impairment in social, occupational, or other important areas of functioning.

SOURCE: https://www.psychdb.com/sleep/parasomnias/substance-sleep#:~:text=Such%20evidence%20of%20an%20independent,there%20is%20other%20evidence%20suggesting

3. Know the differential diagnosis for substance/medication-induced sleep disorder

- Substance intoxication or substance withdrawal
- Delirium
- Other sleep disorders
- Sleep disorder due to another medical condition

SOURCE: https://www.psychdb.com/sleep/parasomnias/substance-sleep#:~:text=Such%20evidence%20of%20an%20independent,there%20is%20other%20evidence%20suggesting

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Content Category 18- Feeding & Eating Problems

ABP Content Specs for Developmental-Behavioral Pediatrics Fellow Collaboration Project

Prepared by Maja Katusic, MD, Veronica Villarreal, MD, & Ann Kennelly, MD, Baylor/Texas Children's DBP Fellows

Reviewed by Noel Mensah-Bonsu, MD, Baylor/Texas Children's DBP Fellowship Director

18. Feeding and Eating Problems

A. Obesity
1. Know the diagnostic criteria for obesity
2. Know the epidemiology of obesity
3. Describe the environmental factors that contribute to obesity
4. Know the psychosocial problems commonly associated with obesity
5. Know how to plan the management of an infant, child, or adolescent with obesity
6. Recognize the importance of behavioral interventions in the prevention and treatment of obesity
7. Know how to plan an obesity prevention program
8. Know how to evaluate a child with obesity for an underlying endocrinopathy or genetic syndrome
9. Understand the risk for obesity in children receiving atypical antipsychotics

B. Failure to thrive (FTT)
1. Know how to identify failure to thrive (FTT)
2. Understand the etiology of FTT
3. Understand the importance of the social circumstances for a child with FTT
4. Recognize the developmental and behavioral conditions associated with FTT
5. Understand the prognosis for children with FTT
6. Know how to plan the management of behavioral and developmental conditions associated with FTT

C. Anorexia nervosa, bulimia nervosa, and binge eating disorder
1. Know the diagnostic criteria for anorexia nervosa
2. Know the diagnostic criteria for bulimia nervosa
3. Know the diagnostic criteria for binge-eating disorder
4. Understand the association of body-image disorders with anorexia and bulimia
5. Know the pre-morbid behaviors that may be associated with anorexia or bulimia
6. Recognize the signs and symptoms of anorexia, bulimia, and binge eating disorder
7. Know the differential diagnosis of a patient with anorexia, bulimia, and binge eating disorder
8. Recognize the medical complications of anorexia, bulimia, and binge eating disorder
9. Know the criteria for hospitalization of a patient with anorexia or bulimia
10. Know the psychiatric co-morbidity of anorexia, bulimia, and binge eating disorder
11. Understand the prognoses of anorexia and bulimia

D. Disorders of feeding in infants and toddlers
1. Know the developmental progression of food selectivity in children
2. Know the diagnostic criteria for avoidant/restrictive food intake disorder
3. Know the medical, developmental, and psychosocial factors that affect appetite
4. Understand the benefits of a mealtime routine
5. Understand the development of feeding skills in healthy children
6. Know the medical factors that increase risk for feeding disorders in infants and toddlers
7. Know the developmental and behavioral conditions associated with feeding disorders
8. Understand that impairments in feeding skills may manifest as food selectivity

9. Know the physiologic phases of swallowing
10. Recognize the signs and symptoms of gastroesophageal reflux
11. Understand the importance of feeding observation for patients with feeding disorders
12. Know how to evaluate a child who is a picky/selective eater
13. Know how to evaluate a child with rumination
14. Know how to evaluate an infant or toddler with a feeding skill disorder
15. Be able to evaluate whether a child with oral-motor dysfunction is safe to feed orally
16. Know how to plan the management for infants or toddlers with feeding disorders
17. Know the management of a child with picky eating
18. Know the management of avoidant/restrictive food intake disorder
19. Know the differential diagnosis of avoidant/restrictive food intake disorder

18. Feeding and Eating Problems

A. Obesity

1. Know the diagnostic criteria for obesity
    - Overweight = BMI 85th to 94th percentile
    - Obesity = BMI greater than or equal to 95th percentile

2. Know the epidemiology of obesity
    - The CDC reports prevalence of childhood obesity in the US is 1 in 5 children

3. Describe the environmental factors that contribute to obesity
    - Lower socio-economic status has been related to increased risk of adult obesity
    - High availability of fast food restaurants and limited access to fresh fruits and vegetables
    - Low availability and safety of parks, bicycle paths, and sidewalks can discourage physical activity
    - Parents are the main role models for eating and physical activity, so parents who are obese are more likely to have children who are obese
    - Since the 1980s, studies have shown association between exposure to screen media and obesity. More TV viewing in childhood is associated with obesity also later into adulthood. Screen time factors thought to contribute to obesity: increased eating while viewing, exposure to marketing for high calorie low nutrient food and beverage; decreased exercise.
    - Adverse life events, such as social isolation, bullying, child neglect/abuse, domestic violence may also be factors that increase obesity. Food may be used in response to stress along with decreased physical activity.

4. Know the psychosocial problems commonly associated with obesity
    - Children who are overweight/obese more likely to be bullied and ostracized. Fear of bullying may lead to emotional eating behaviors or lead to children staying inside and exercising less.
    - Overweight/obese children may experience depression, anxiety, decreased self-esteem, behavioral problems. Risk for mental health conditions increase the longer a child is overweight.

5. Know how to plan the management of an infant, child, or adolescent with obesity
    - AAP Institute for Healthy Childhood Weight in 2016 released algorithm for assessment/management of obesity in patients 2+ years of age: assess healthy eating and living behaviors, prevention counseling, and workup for those in the overweight/obese range.
    - Recommended labs to order for children with obesity: fasting glucose, hemoglobin A1C, alanine aminotransferase, and aspartate aminotransferase. Screening with these labs for children with obesity may start as young as two years of age.
    - Bright Futures/AAP recommends using motivational interviewing and accommodating stages of change/readiness to change

6. Recognize the importance of behavioral interventions in the prevention and treatment of obesity
    - Family-based behavioral treatment: effective intervention for pediatric obesity in preschoolers to pre-adolescents. Goal is to create environment to promote healthy eating and

activity by building parenting skills (i.e. limit setting and problem solving). Studies show 26 to 75 hours contact over a six-month period is most effective (not just referring to dietitican).
- Cognitive behavioral therapy is effective in management of obesity in older children and adolescents, mostly by breaking negative behavior cycles.
- Changing food and drink available at home
- Repackaging snacks into age appropriate portions and decrease caloric intake and teach children about healthy eating habits

7. Know how to plan an obesity prevention program
- Providing incentives for supermarkets or farmers markets to establish their businesses in underserved areas, placing nutrition and calorie content on restaurant and fast food menus, and implementing and supporting nutrition standards for childcare, schools, hospitals, and worksites. Community leaders/city planners can increase places where people can be active, encourage use of public trails/paths. (CDC.gov)
- Schools can: encourage children to drink water rather than sweetened beverages, ensure that the available food and beverage options are healthy and help youth eat food that meets dietary recommendations. (CDC.gov)
- Families can: Encourage young children to engage in at least 60 minutes of unstructured physical activity per day. Encourage children age 6 to 17 to engage in greater than or equal to 60 minutes of exercise per day (at least 60 minutes should "generate sweat"). Avoid screen time for < 18 months of age and children 18 months to 4 years should have no more than 1 hour off screen time per day. Establish family media plan for usage of screen time. Ensure adequate sleep. (Bright Futures/AAP)
- Bright Futures has a document that gives additional recommendations for actions to prevent obesity at:
https://brightfutures.aap.org/Bright%20Futures%20Documents/BF4_HealthyWeight.pdf

8. Know how to evaluate a child with obesity for an underlying endocrinopathy or genetic syndrome
- AAP algorithm for Assessment/Management of Childhood Obesity in Patients 2+ Years from 2016 found at:
https://ihcw.aap.org/Documents/Assessment%20%20and%20Management%20of%20Childhood%20Obesity%20Algorithm_FINAL.pdf
- Potential endocrinopathies: Precocious puberty, polycystic ovarian syndrome (PCOS), prediabetes, premature adrenarche, Type 2 diabetes (found in above AAP algorithm)
- Potential genetic syndromes associated with childhood obesity, following table taken from the following article by Stephen O'Rahilly and I. Sadaf Farooqi Published online 2006 Jun 15. doi: 10.1098/rstb.2006.1850

Table 2

Human pleiotropic obesity syndromes.

syndrome	additional clinical features	locus	gene
autosomal dominant			
Prader–Willi syndrome	hypotonia, mental retardation, short stature, hypogonadotropic hypogonadism	lack of the paternal segment 15q11.2–q12	unknown
Albright hereditary osteodystrophy	short stature, skeletal defects, and impaired olfaction	20q13.2	GNAS1
ulnar-mammary syndrome	ulnar defects, delayed puberty, hypoplastic nipples	12q24.1	TBX3
autosomal recessive			
Bardet–Biedl syndrome	mental retardation, dysphormic extremities, retinal dystrophy or pigmentary retinopathy, hypogonadism and structural abnormalities of the kidney or functional renal impairment	1q13 (BBS1), 16q21 (BBS2), 3p13 (BBS3), 15q22 (BBS4), 2q31 (BBS5) 20p12 (BBS6), 4q27 (BBS7), 14q32 (BBS8)	
Alstrom syndrome	retinal dystrophy, neurosensory deafness, diabetes	2p13	ALMS1
Cohen syndrome	prominent central incisors, opthalmopathy, microcephaly	8q22	unknown
X-linked			
fragile X syndrome	mental retardation, hyperkinetic behaviour, macroorchidism, large ears, prominent jaw, and high-pitched jocular speech	Xq27.3	FMR1
Borjeson–Forssman–Lehmann syndrome	mental retardation, hypogonadism, large ears	Xq26	PHF6
Mehmo syndrome	mental retardation, epilepsy, hypogonadism, microcephaly	Xp22.13	unknown
Simpson–Golabi–Behmel—type 2	craniofacial defects, skeletal and visceral abnormalities	Xp22	unknown
Wilson–Turner syndrome	mental retardation, tapering fingers, gynaecomastia	Xp21.2	unknown

9. Understand the risk for obesity in children receiving atypical antipsychotics
    - Atypical antipsychotics are associated with diabetes, hyperlipidemia, and obesity
    - American Academy of Child and Adolescent Psychiatry endorse consensus statement of the American Diabetes Association and American Psychiatry Association to obtain fasting glucose and fasting lipid profile upon initiation of antipsychotic medication, as well as BMI and waist circumference measurements + then fasting glucose and lipid (and blood pressure) 3 months after initiation
    - Adapted from ADA guidelines and an article in Academic Pediatrics from 2014 https://doi.org/10.1016/j.acap.2014.05.009

B. Failure to thrive (FTT)

1. Know how to identify failure to thrive (FTT)

- Weight <3rd percentile for age (after period of stable weight gain)

-Weight for height <5th percentile

-Growth that has fallen, Crossing 2 percentile curves in a short time

-Weight 20% or more below ideal weight for height

-Weight gain <20 g/day from 0-3 months old or <15 g/day from 3-6 months old

2. Understand the etiology of FTT

-Inadequate nutrition due to either inadequate intake or altered metabolism

-Organic causes (prematurity, congenital malformations, neuromuscular disorders, abnormal suck/swallow coordination, intrauterine infections, or exposure to drug and/or other toxins) and Nonorganic psychosocial causes (familial dysfunction, maternal depression, poverty, low food security)

-If caloric intake appears to be adequate evaluate for organic cause of FTT due to Excessive caloric loss or Increased caloric requirements

-Neurologic conditions such as cerebral palsy may increase caloric expenditure due to increased tone or may cause decreased caloric intake due to feeding difficulties associated with abnormal pharyngeal tone/coordination.

-Behavioral Disorders such as Autism Spectrum Disorder or Attention Deficit Hyperactivity Disorder may be associated with symptoms, such as, “Picky” eating and when severe they may contribute to FTT.

3. Understand the importance of the social circumstances for a child with FTT

-In nonorganic causes of FTT to important to evaluate and address improper formula mixing, family dysfunction, child-care arrangements, maternal depression and risk for abuse/neglect.

-Risk for abuse/neglect include: Abnormal child-parent interaction, Poor parental functioning, Stressful home environment, Parental substance abuse/mental health issues, Domestic violence and Parental characteristics (i.e. young age, low education, single parenthood, large number of dependent children and poverty.)

4. Recognize the developmental and behavioral conditions associated with FTT

- Children with FTT may appear to have gaze disturbances or total avoidance of eye contact and apathetic withdrawal. Infants may resist being held or prefer interactions with inanimate objects. Infants may have co-occurring difficulties with attachment, communication, mood and affect.

-Most children likely have developmental delays in language and social behavior.

-Behavioral Disorders such as Autism Spectrum Disorder or Attention Deficit Hyperactivity Disorder may be associated with symptoms such as “Picky” eating and when these symptoms are severe, they may contribute to FTT. Behavioral issues are more common in children with FTT.

-FTT may be due to pain or discomfort associated with eating due to other medical issues (i.e. pain or discomfort due to dysphagia, aspiration, esophagitis, gastroesophageal reflux, constipation, or celiac disease).

5. Understand the prognosis for children with FTT

-Prognosis with organic FTT depends on the cause.

-Nonorganic FTT, the majority of children age > 1 year achieve a stable weight.

-Children who develop FTT before age 1 year are at high risk of cognitive delay, especially verbal and math skills. Behavioral problems occur in about 50% of children. Problems related to eating or elimination occur in about 50% of children (usually those with other behavioral problems).

-Severe FTT is associated with poor cognitive and academic outcomes. These poor outcomes may be caused by the insufficient intake of essential nutrients for neural development.

6. Know how to plan the management of behavioral and developmental conditions associated with FTT

-Education and emotional support to correct problems interfering with the parent-child relationship.

-Evaluation team (in hospital) should define the family's needs, provide initial instruction and support, and institute appropriate referrals to community agencies.

-A pre-discharge planning conference involving hospital-based personnel, representatives from the community agencies that will provide follow-up services, and the child's primary physician is ideal.

-The parents should be invited to a summary session after the conference so that they can meet the community workers, ask questions, and arrange follow-up appointments.

-Parents should have regular follow up appointments with PCP and community agencies to ensure families have necessary resources and that child is gaining weight appropriately.

-In some cases, the child must be placed in foster care.

-Parenting skill training and psychologic counseling may be necessary to ensure ability to adequately care for child.

FTT Resources:

-Pediatrics Core: Growth and Development

-Merck Manuals: FTT https://www.merckmanuals.com/professional/pediatrics/miscellaneous-disorders-in-infants-and-children/failure-to-thrive-ftt

-PREP Questions

C. Anorexia nervosa, bulimia nervosa, and binge eating disorder

1. Know the diagnostic criteria for anorexia nervosa

TABLE 1. DSM-5 Diagnostic Criteria for Anorexia Nervosa (4)

A. Restriction of energy intake relative to requirements, leading to a significantly low body weight in the context of age, sex, developmental trajectory, and physical health. *Significantly low weight* is defined as a weight that is less than minimally normal or, for children and adolescents, less than that minimally expected (e.g., falling off a previously followed growth curve).
B. Intense fear of gaining weight or of becoming fat or persistent behavior that interferes with weight gain, even though at a significantly low weight.
C. Disturbance in the way in which one's body weight or shape is experienced, undue influence of body weight or shape on self-evaluation, or persistent lack of recognition of the seriousness of the current low body weight.
Types:
Restricting type: During the last 3 months, the individual has not engaged in recurrent episodes of binge eating or purging behavior (i.e., self-induced vomiting or misuse of laxatives, diuretics, or enemas). This subtype describes presentations in which weight loss is accomplished primarily through dieting, fasting, and/or excessive exercise.
Binge-eating/purging type: During the last 3 months, the individual has engaged in recurrent episodes of binge eating or purging behavior (i.e., self-induced vomiting or the misuse of laxatives, diuretics, or enemas).

- Anorexia nervosa (AN) is characterized by severe caloric restriction due to a fear of weight gain and a disordered body image, resulting in low body weight (for children, BMI-for-age <5 percentile is considered underweight by the Centers for Disease Control) due to either weight loss or failure to gain expected weight given the previous growth trajectory. Caloric restriction is accompanied by either fear of weight gain or persistent behavior that interferes with weight gain (ie, intentional vomiting or misuse of laxatives/diuretics) in addition to disordered body image.
- Transition from DSM-4 to DSM-5:
    - DSM-5 retains the core characteristics but expands the diagnostic criteria to accommodate a wider range of individuals.
    - Unlike DSM-IV criteria, DSM-5 does not include a requirement for amenorrhea, increasing the applicability of the diagnosis in males, premenarchal females, and postmenopausal females.
    - The replacement of a specific weight criterion (<85% expected body weight) with a broader statement that allows clinicians to assess weight based on individual growth trajectory as well as numerical guidelines. In addition, the phrase "refusal to maintain weight" was removed and an alternative behavioral criterion ("persistent behavior that interferes with weight gain") was added to avoid making assumptions about the patient's intentions.

2. Know the diagnostic criteria for bulimia nervosa

TABLE 6. DSM-5 Diagnostic Criteria for Bulimia Nervosa (4)

A. Recurrent episodes of binge eating. An episode of binge eating is characterized by both of the following:
1. Eating, in a discrete period of time (e.g., within any 2-hour period), an amount of food that is definitely larger than what most individuals would eat in a similar period of time under similar circumstances.
2. A sense of lack of control over eating during the episode (e.g., a feeling that one cannot stop eating or control what or how much one is eating).
B. Recurrent inappropriate compensatory behaviors in order to prevent weight gain, such as self-induced vomiting; misuse of laxatives, diuretics, or other medications; fasting; or excessive exercise.
C. The binge eating and inappropriate compensatory behavior both occur, on average, at least once a week for 3 months.
D. Self-evaluation is unduly influenced by body shape and weight.
E. The disturbance does not occur exclusively during episodes of anorexia nervosa.

- The primary features of Bulimia Nervosa (BN), according to the Diagnostic and Statistical Manual of Mental Disorders (Fifth Edition), include:
  - Recurrent binge-eating episodes
    - Binge-eating episodes are characterized by eating large amounts of food, beyond what most other people would eat, within a certain period and in similar circumstances. Also, there is an associated lack of control in overeating, in which there is an inability to refrain from eating or cease eating once started. Guilt often follows these binge-eating episodes, which drives the compensatory purging. Celebratory meals, such as those on birthdays or holidays, are not included because they are special circumstances, nor is eating small portions of food throughout the day.
  - Recurrent inappropriate behaviors that are compensatory in nature, to prevent weight gain
    - Vomiting is the most frequent compensatory behavior for binge eating in BN. The vomiting allows for alleviating physical discomfort and helps assuage the fear of weight gain. Fingers and other instruments may be used to stimulate the gag reflex and induce vomiting. Physical symptoms of dental caries, enamel erosion, parotid swelling, and calluses on knuckles from self-induced vomiting (Russell sign) may be seen, as well as hematemesis from esophageal tears. There may be other compensatory methods such as the misuse of laxatives, which may lead to fluid and electrolyte loss, and use of diuretics, which would decrease fluid weight and cause electrolyte imbalances.
  - Self-evaluation excessively influenced by body shape and weight.
  - Occurrence not limited exclusively to episodes of anorexia nervosa.

3. Know the diagnostic criteria for binge-eating disorder

TABLE 7. DSM-5 Diagnostic Criteria for Binge-Eating Disorder (4)

A. Recurrent episodes of binge eating. An episode of binge eating is characterized by both of the following:
1. Eating, in a discrete period of time (e.g., within any 2-hour period), an amount of food that is definitely larger than what most individuals would eat in a similar period of time under similar circumstances.
2. A sense of lack of control over eating during the episode (e.g., a feeling that one cannot stop eating or control what or how much one is eating).
B. The binge-eating episodes are associated with three (or more) of the following:
1. Eating much more rapidly than normal.
2. Eating until feeling uncomfortably full.
3. Eating large amounts of food when not feeling physically hungry.
4. Eating alone because of feeling embarrassed by how much one is eating.
5. Feeling disgusted with oneself, depressed, or very guilty afterward.
C. Marked distress regarding binge eating is present.
D. The binge eating occurs, on average, at least once a week for 3 months.
E. The binge eating is not associated with the recurrent use of inappropriate compensatory behavior as in bulimia nervosa and does not occur exclusively during the course of bulimia nervosa or anorexia nervosa.

- Binge-eating disorder was introduced in the DSM-5. Like bulimia nervosa, individuals with binge-eating disorder experience recurrent episodes of binge eating (average of once weekly for 3 months) and feel as if they do not have control over the eating behavior during the episodes. For this diagnosis, episodes of binge-eating are also associated with a minimum of 3 of the following: **rapid consumption of food, consumption to the point of discomfort due to fullness, consumption of large amounts despite lack of hunger, consumption of food in private due to embarrassment by quantity of consumed food, and feeling of self-disgust after the binge-episode**. Individuals with this disorder are markedly distressed about the binge-episodes. This condition is significantly different from BN in that there are not compensatory behaviors to prevent weight gain.
    - **Medical complications** associated with binge-eating disorder are often **related to obesity** and include hypertension, coronary artery disease, dyslipidemia, diabetes mellitus type 2, osteoarthritis, and obstructive sleep apnea.
    - **Treatment approaches** for this condition include **cognitive behavioral therapy** and **dialectical behavioral therapy**. Weight loss treatment is often necessary for individuals with this condition who are also obese. While lisdexamfetamine has received FDA approval for treatment of binge-eating disorder in adults, approval has not been received for treatment of this disorder in pediatric patients. With therapy, 30%-80% of individuals with this disorder recover within 1-6 years.
    - Differential diagnosis includes: **Purging disorder**: a sub-classification of Other Specified Feeding or Eating Disorder characterized by recurrent purging behavior (ie, self-induced vomiting or misuse of laxatives/diuretics) without accompanied binge-eating episodes. Disorders in this category describe patient presentations that do not fully meet criteria for another eating or feeding disorder.

4. Understand the association of body-image disorders with anorexia and bulimia
    - Body image concerns in **anorexia** may include a distorted/negative body image; the patient's perception of body size differs from reality. They may perceive their bodies as large and

unattractive, despite being underweight. Therefore, a healthy weight may feel uncomfortable and unacceptable to someone with anorexia. Individuals with anorexia may be unable to recognize the seriousness or potential impact of their low weight on their general health.

- Although patients with **bulimia** are at or above a healthy weight, they still have concerns about their weight; however, their body image concerns may be to a lesser degree than in individuals with AN. The self-image of individuals with bulimia is what drives them to restrictive and compensatory behaviors. They may have maladaptive behaviors, including self-induced vomiting or laxative or diuretic use. Self-evaluation by individuals with bulimia places great importance on weight, body shape, or both, something that is related to the individual's self-esteem. Individuals with bulimia often feel a sense of shame and secrecy. They may often attempt to hide their symptoms as well as their binge-eating behaviors.

5. Know the pre-morbid behaviors that may be associated with anorexia or bulimia

- Common premorbid behaviors that should heighten concern for eating disorders include weight loss, growth stunting, pubertal delay, restrictive eating, frequent vomiting, extreme exercise habits, poor weight gain, or body image concerns.
    - Despite these behaviors, individuals with anorexia will often continue with academic and athletic success, and they may be even more driven to attain success by their need for control.
- Individuals with bulimia may often experience secrecy, shame, and guilt that lead to feelings of loss of control. They may be impulsive and exhibit risk-taking behaviors including substance abuse or self-injurious behaviors.
- Parents may provide additional information that an adolescent may not disclose.
    - They may talk about their child eating less, avoiding high-calorie foods, and increasing exercise.
- Caloric reduction may not be sudden, but occur over time, with the limitation of certain foods, such as fats, sweets, and protein.
- There may be external pressures in individuals involved in activities that highlight weight and/or appearance (eg, dance, wrestling, modeling, cheerleading, gymnastics).

6. Recognize the signs and symptoms of anorexia, bulimia, and binge eating disorder

Table - Features in Diagnosing Anorexia Nervosa and Bulimia Nervosa

Anorexia Nervosa	Bulimia Nervosa
• Restriction of calories leading to lower than expected body weight • Intense fear of weight gain or being fat • Body image distortion	• Binge eating • Repeated use of unhealthy behaviors after a binge to prevent weight gain (vomiting, laxative, or diuretic abuse, food restriction, excessive exercise) • Behaviors occur more than once weekly for 3 months • Self-worth overly based on body shape and weight • Behaviors distinctly apart from anorexia nervosa

Reprinted with permission from Golden NH, Schneider M, Wood C; AAP Committee on Nutrition, Committee on Adolescence, Section on Obesity. Preventing obesity and eating disorders in adolescents. *Pediatrics.* 2016;138(3):e20161649.

- Weight is not the only marker of clinically significant disease. As many as 66% of individuals with eating disorders are at normal weight and 33% are obese at the onset of disease.

- Both anorexia nervosa and bulimia nervosa frequently begin with dietary changes, such as adopting a vegetarian, low-fat, low-carbohydrate, or other "healthy" diet, along with changes in mealtime rituals, such as taking longer to complete meals, cutting food into small pieces, or shifting food around on the plate.
- Individuals who purge, often make frequent trips to the bathroom, particularly during or after meals, and may adjust their schedules to accommodate both binge eating and purging.
- Many adolescents, including those with binge-eating disorder, begin to avoid social eating. Increases in physical activity may precede changes in diet and often become progressively obsessive throughout the course of illness.
- In **anorexia**, many patients have weight concerns and behavioral changes related to weight loss that precede their clinical diagnosis by 6 to 12 months. Individuals with anorexia maintain academic functioning and may become even be more compulsive and driven, as cognitive skills typically stay intact until there is severe malnutrition.
- In **bulimia**, many individuals have exhibited symptoms for nearly 5 years prior to seeking treatment. Additionally, self-evaluation is unreasonably influenced by personal body shape and/or weight. Individuals with this condition may have body weight within the normal or overweight ranges.

7. Know the differential diagnosis of a patient with anorexia, bulimia, and binge eating disorder

TABLE 4. Differential Diagnosis for Weight Loss, Vomiting, and Binge Eating

Weight loss
• Celiac disease
• Inflammatory bowel disease
• Malabsorption
• Hyperthyroidism
• Addison disease
• Acquired immunodeficiency syndrome
• Occult malignancies
Vomiting
• Migraine
• Pseudotumor cerebri
• Hydrocephalus
• Central nervous system malignancy
• Gastrointestinal disease
• Cyclic vomiting
Binge eating
• Obesity
• Major depressive disorder
• Borderline personality disorder
• Prader Willi syndrome

- **Purging disorder** is a sub-classification of Other Specified Feeding or Eating Disorder characterized by recurrent purging behavior (ie, self-induced vomiting or misuse of laxatives/diuretics) without accompanied binge-eating episodes. Disorders in this category describe patient presentations that do not fully meet criteria for another eating or feeding disorder.

- **Anorexia nervosa** is characterized by lower than minimally normal or expected body weight due to intense caloric restriction accompanied by either fear of weight gain or persistent behavior that interferes with weight gain in addition to disordered body image.
- **Bulimia nervosa** is characterized by episodes of recurrent binge-eating with inappropriate compensatory behaviors to prevent weight gain occurring an average of once weekly for 3 months with accompanied self-evaluation that is unreasonably influenced by personal body shape and/or weight.
- **Binge-eating disorder** is significantly different from bulimia nervosa in that there are not compensatory behaviors to prevent weight gain. Binge-eating disorder was introduced in the DSM-5. Like bulimia, individuals with binge-eating disorder experience recurrent episodes of binge eating (average of once weekly for 3 months) and feel as if they do not have control over the eating behavior during the episodes.
- **Female athlete triad**:
    - The inadequate intake of calories may not be volitional. Although, female athletes in sports that have an aesthetic component, such as gymnastics, may have poor body image because of an inability to achieve their idea of the appropriate body weight or shape. Body image disorders can result in disordered eating or eating disorders such as anorexia and bulimia; although, further evaluation and questioning are needed to determine this.
    - The female athlete triad consists of 3 components: **disturbances in menstrual function**, **decreased bone mineral density**, and **deficient energy availability**. Female athletes may exhibit only 1 component of the female athlete triad. The presence of any 1 of the 3 components is sufficient to justify further evaluation. Deficient energy availability (EA) is the driving factor in the female athlete triad. The triad occurs when daily energy expenditure exceeds necessary energy intake and may or may not coincide with disordered eating. In fact, deficient energy availability often occurs unintentionally. Females with the triad often have normal physical findings.
    - Oral contraceptives are not indicated for the female athlete triad and do not result in increased bone density. Intake of calcium and vitamin D should be optimized. Treatment of the female athlete triad involves ensuring appropriate energy availability.
- Differential for **Delayed Menarche**: Menarche should occur within 2 years of thelarche; however, other causes of primary amenorrhea should be ruled out, including eating disorders, pregnancy, polycystic ovarian syndrome, prolactinoma, hyperthyroidism, or use of medications such as oral contraceptive pills or corticosteroids.

8. Recognize the medical complications of anorexia, bulimia, and binge eating disorder
    - **Binge-eating disorder**:
        - Medical complications are often related to obesity and include: hypertension, coronary artery disease, dyslipidemia, diabetes mellitus type 2, osteoarthritis, and obstructive sleep apnea.
    - **Anorexia**:

    - Caloric restriction and purging behaviors associated with AN can affect many organ systems and result in serious and life threatening medical conditions (see further complications below).
        - Osteopenia and euthyroid sick syndrome can result from starvation.
    - Cardiovascular complications: severe bradycardia, prolonged QTc interval, cardiac arrhythmia, hypotension, orthostatic hypotension.
        - Bradycardia, QTc prolongation, and electrolyte abnormalities are the most common reason for hospitalization.

- **Bulimia nervosa**:
    - Chronic dehydration can develop that may affect the renin-angiotensin-aldosterone axis; compensatory mechanisms such as vomiting may cause metabolic alkalosis with hypokalemia and hypochloremia. In bulimia nervosa, laxative abuse may result in metabolic acidosis with hypokalemia and hypochloremia.
    - Laboratory findings reflect the electrolyte disturbances associated with purging.
    - Chronic dehydration may affect the renin-angiotensin-aldosterone axis.
    - Vomiting may cause metabolic alkalosis, hypokalemia, and hypochloremia.
    - Cardiac arrhythmias may result from hypokalemia; cardiomyopathy may result from chronic purging, particularly in those who abuse ipecac.

Further Complications:

- **Gastrointestinal** complaints occur commonly when the body tries to compensate for insufficient nutritional intake by slowing gastric motility, leading to increased constipation, bloating, and abdominal discomfort.
- Complications in the **endocrine** system may include euthyroid sick syndrome (low total and unbound triiodothyronine values with normal thyroxine and thyrotropin values), an adaptive response to starvation.
- **Girls** may have **amenorrhea** with decreased FSH, LH, and estradiol concentrations
- **Boys** may have **low testosterone concentrations**.
- **Prepubertal patients** may experience delayed puberty and growth retardation, and **older patients** with suppressed hypothalamic pituitary gonadal axes may fail to deposit bone, leading to osteopenia and increased risk of stress fractures.
- Patients with **restrictive eating disorders** may develop pancytopenia, with white blood cell loss first, anemia second, and platelet suppression last.
- Closer examination of the **bone marrow** can reveal an increase in fat deposition in the hypocellular matrix despite loss of fat elsewhere in the body during prolonged starvation.
- In the **brain**, weight loss is associated with decreased brain tissue (both white and gray matter), reduced brain activity, and mood changes. Psychomotor retardation, or slowing of speech, thought, and movement, may occur with prolonged starvation. Patients may have deficits in concentration, ability to focus, and memory. Although some patients may continue to perform exceptionally well in school, others may show declining academic performance with ongoing undernutrition.
- Complications associated with **self-induced vomiting**: Metabolic alkalosis, hypochloremia, hypokalemia, orthostasis, Mallory-weiss esophageal tears and resultant anemia. Fluid and electrolyte abnormalities may cause or contribute to cardiac complications.
- Many of the medical complications of eating disorders improve and ultimately reverse with nutritional rehabilitation. Some complications, however, such as low bone mineral density, growth retardation, and structural brain changes, may not fully normalize after prolonged disease.

9. Know the criteria for hospitalization of a patient with anorexia or bulimia

TABLE 5. Criteria for Hospital Admission for Patients with Eating Disorders (5)

- Heart rate <50 beats per minute while awake
- Heart rate <45 beats per minute while asleep
- Systolic pressure <90 mm Hg
- Temperature <35.6°C (96°F)
- Prolonged QTc or other arrhythmia
- Orthostatic changes in blood pressure (>10 mm Hg)
- Orthostatic changes in pulse (>20 beats per minute)
- Syncope
- Electrolyte abnormalities
- Esophageal tears or hematemesis
- Intractable vomiting
- Suicide risk
- Weight <75% of expected body weight or body fat <10%
- Ongoing weight loss despite intensive management
- Acute weight loss and food refusal
- Failure to respond to outpatient treatment

Table. American Academy of Pediatrics Criteria for Inpatient Hospitalization in Bulimia Nervosa

Bulimia Nervosa
Syncope
Serum potassium <3.2 mmol/L
Serum chloride <88 mmol/L
Esophageal tears
Cardiac arrhythmias including prolonged QTc
Hypothermia
Suicide risk
Intractable vomiting
Hematemesis
Failure to respond to outpatient treatment

Data collected and reprinted with permission from Campbell K, Peebles R. Eating Disorders in Children and Adolescents: State of the Art Review. *Pediatrics*. 2014;134:582–592.

10. Know the psychiatric co-morbidity of anorexia, bulimia, and binge eating disorder
    - Eating disorders commonly occur in patients with other psychiatric conditions, particularly anxiety, depression, bipolar disorder, and/or substance abuse.
    - In general, when considering pharmacologic therapy, clinicians should weigh the adverse effects against the potential benefits of the medication on both eating disorder pathology and comorbid psychiatric symptoms.

11. Understand the prognoses of anorexia and bulimia
    - **Anorexia Nervosa**:
        - Cognitive behavioral therapy, dialectical behavioral therapy, and family-based treatment are the most common therapy strategies.
        - Recovery is achieved within 5 years by individuals who receive traditional treatment.
        - Of note, 50% of individuals who receive family-based treatment have been found to have recovery by 1 year.
        - While improved nutritional status may reverse many of the medical conditions which may present in settings of semi-starvation, structural brain changes, osteopenia, and growth retardation may not fully recover.
    - **Bulimia**:
        - Cognitive behavioral therapy, dialectical behavioral therapy, and family-based treatment are common treatment approaches for bulimia nervosa.
        - With treatment, 33%-66% of individuals recover within 5 years.
    - **Binge eating disorder**:

- Treatment approaches for this condition include cognitive behavioral therapy and dialectical behavioral therapy.
- Weight loss treatment is often necessary for individuals with this condition who are also obese.
- While lisdexamfetamine has received FDA approval for treatment of binge-eating disorder in adults, approval has not been received for treatment of this disorder in pediatric patients.
- With therapy, 30%-80% of individuals with this disorder recover within 1-6 years.

D. Disorders of feeding in infants and toddlers

1. Know the developmental progression of food selectivity in children

Inborn preferences: infants have a preference for energy-rich foods. They are born with preference for sweet tastes; all other tastes learned through experience.

Learned Preferences before birth: Taste preferences can be acquired by an infant from pregnant mother via amniotic fluid (flavored by foods eaten).

Breast feeding: The food eaten by a breast-feeding mother can influence the infant's taste preferences at the introduction of complementary foods.

Inherited Factors: Some infants and toddlers are particularly sensitive to bitter foods and drinks.

Learned taste acceptance: Infant taste preferences are learned through experience. The earlier infants are offered food with a specific strong taste (such as vegetables) the more likely they are to accept the food. More likely to accept other new foods and tastes if they are given earlier in period of food introduction

Disgust and Rejection: After 12-14 months infants have learned to recognize which foods they do not like and reject either by sight or taste

Neophobic stage (the fear of new foods) – after 12 months it is difficult to introduce new foods and will often refuse to eat foods they accepted before. The rejection of new foods is a normal response (peaks about 18-24 months and usually diminishes by 8 years old) Most children accept new foods only after repeated exposures

2. Know the diagnostic criteria for avoidant/restrictive food intake disorder

- An eating or feeding disturbance (e.g., apparent lack of interest in eating or food; avoidance based on the sensory characteristics of food; concern about aversive consequences of eating) as manifested by persistent failure to meet appropriate nutritional and/or energy needs associated with one (or more) of the following:
    - Significant weight loss (or failure to achieve expected weight gain or faltering growth in children).
    - Significant nutritional deficiency.
    - Dependence on enteral feeding or oral nutritional supplements.
    - Marked interference with psychosocial functioning.
- The disturbance is not better explained by lack of available food or associated cultural practice

- The eating disturbance does not occur exclusively during the course of anorexia nervosa or bulimia nervosa, and there is no evidence of a disturbance in the way in which one's body weight or shape is experienced.
- The eating disturbance is not attributable to a concurrent medical condition or not better explained by another mental disorder. When the eating disturbance occurs in the context of another condition or disorder, the severity of the eating disturbance exceeds that routinely associated with the condition or disorder and warrants additional clinical attention.

3. Know the medical, developmental, and psychosocial factors that affect appetite

Medical Factors: Blood Glucose Levels, Leptin Production, Medications, illness, neurologic abnormalities (does not signal hunger)

Developmental Factors: Energy Intake/Expenditure, infant/maternal bonding, breast feeding vs. bottle feeding, impaired feeding skills, schedule/routine meal times

Psychosocial Factors: Stress, Social environments/surroundings, food insecurity, parental knowledge of hunger cues

4. Understand the benefits of a mealtime routine

Routine helps child know what to expect at meal time, helps child get on a schedule and establish expectations, it can help teach children how to eat and important meal time skills like hand washing as well as healthy eating habits. It can also help children develop social skills.

5. Understand the development of feeding skills in healthy children

-Before Birth: sucking and swallowing observed in the womb

Birth: Opens mouth to suck fist (especially if hungry), the gag response to food and objects in the mouth is observed from birth, moves tongue in and out and up and down

2 weeks – 9 months: Can show open mouth for spoon at an early age so this response is present before onset of complementary feeding at 4-6 months.

2 months: Can move food from a spoon to the back of the mouth and has developed better head control

6 months: Gag response declines as mouth becomes more used to the feel of food but still observed, can move food from side to side of the mouth.

6-12 months: Eruption of teeth

6-14 months: Can chew softer lumps and keep most food in the mouth.

7-12 months: can close lips to clear the spoon

8 months: can hold bottle independently

8-12 months: can bite into harder foods (once teeth have erupted).

12 months – 48 months: can cope with most textures offered but chewing not fully matured.

14 months: can feed self independently with a spoon

24 months: can cope with most foods as offered as part of a family meal.

21-24 months: can feed self independently with a fork

6. Know the medical factors that increase risk for feeding disorders in infants and toddlers

-Disorders that affect oral, nasal, or pharyngeal function (macroglossia, extensive dental disease, cleft palate, velopharyngeal insufficiency, choanal atresia, tonsillar hypertrophy)

-Aerodigestive disease: Airway (i.e subglottic stenosis), Pulmonary (i.e. bronchopulmonary dysplasia) Gastrointestinal (i.e. Eosinophilic esophagitis), Congenital and Cardiac (Congenital Heart Disease i.e. hypoplastic left heart syndrome)

-Iatrogenic: conditions requiring prolonged hospitalization with critical care support, invasive operative procedures affective vital systems, aversive feeding

***Perinatal adversities, female gender, maternal smoking in pregnancy, prematurity, small for gestational age, congenital malformations, being the first born, and immigrant status all associated with increased risk of feeding and eating disorders (FED)

7. Know the developmental and behavioral conditions associated with feeding disorders

-Children with neurologic impairments such that nutritional needs exceed their feeding skills

-Children with motor delays as well as hyper/hypotonia (i.e. Cerebral Palsy and Muscular Dystrophies)

-Children with Autism Spectrum Disorder and ADHD may have associated symptoms that make them more likely to develop avoidant/restrictive food intake disorder as well as other feeding disorders

-Children in whom picky eating is severe are more likely to develop avoidant/restrictive feeding disorder

Examples of psychosocial conditions associated with Pediatric Feeding Disorder:

Psychosocial Restriction (Health Conditions and Problems*)	**Impact on Feeding Behaviors**

Developmental (child and/or caregiver)
• Delay
• Disorder
Mental/Behavioral Health (child and/or caregiver)
• Diagnosed disorder
• Undiagnosed signs/symptoms of disorder
• Deregulated temperament/personality characteristics
Social
• Caregiver-child interaction problems
• Cultural expectations are not commensurate with AAP nutrition guidelines
Environmental
• Disorganized/distracting feeding environment
• Disorganized or poorly timed schedule of feedings
• Access to food or other necessary resources
• Inadvertent reinforcement of food refusal behavior
	• **Learned aversion (child and/or caregiver)**
	• **Stress/distress (child and/or caregiver)**
	  • Caregiver disengagement
	  • Caregiver over-engagement
	• **Disruptive behavior**
	  • Food refusal (passive & active resistance)
	  • Gagging/vomiting
	  • Elopement/attempts to disengage or flee from meal
	• **Food over-selectivity**
	• **Failure to advance to age-appropriate diet or feeding habit despite adequate skill**
	  • Reliance on formula beyond expected chronological age
	  • Failure to consume age-typical texture
	  • Not feeding self at age-typical level
	• **Grazing behavior**
	• **Caregiver use of compensatory strategies to feed child**

Legend: * International Classification of Functioning, Disability, and Health (ICF) terminology

8. Understand that impairments in feeding skills may manifest as food selectivity

-A child that experiences neophobia at a delayed age

-A child that is unable to consume age-appropriate liquids and solids or that is unable to age-appropriately self-feed or use age-appropriate utensils may only be able to eat a limited number of foods and in comparison to peers appear to have food-selectivity.

9. Know the physiologic phases of swallowing

Oral Stage

The oral stage is often split into two steps:

1.) Oral preparatory – liquids sealed in oral cavity, solids undergo mastication and manipulation

2.) Oral Propulsion - The tongue elevates to move the bolus posteriorly into the oropharynx.

Pharyngeal Phase

This stage serves two primary purposes: 1) directs food into the esophagus and 2) protects the airway from aspiration. It is characterized by a wave of coordinated stimuli lasting about a second that ends as the food bolus reaches the upper esophageal sphincter.

1.) Nasopharynx closure

2.) Airway protection

3.) Elevation of the hyoid-laryngeal complex

4.) Bolus transport

5.) Transit through upper esophageal sphincter (UES)

Esophageal Phase

The bolus is propagated inferiorly by a wave of peristalsis once it reaches the esophagus. This is an autonomous process not under voluntary control, much like the pharyngeal phase. It occurs much slower than the pharyngeal phase. This phase ends once the bolus passes through the lower esophageal sphincter (LES) and into the stomach. At rest, the LES is tonically contracted to prevent reflux from the stomach and it undergoes relaxation during the swallowing phase.

10. Recognize the signs and symptoms of gastroesophageal reflux

Return of gastric contents into the esophagus (normal process) referred to as spitting up or vomiting if refluxed material passes out of the mouth. Occurs at least once daily in half of infants up to 3 months old and 2/3 of infants 4-6 months old. Usually this decreases rapidly by 8 months of age and most infants usually outgrow daily vomiting by 2 years old.

11. Understand the importance of feeding observation for patients with feeding disorders

-Displays impairments in oral sensory and/or oral motor functioning.

-Displays Skill Based Dysfunction (unsafe oral feeding, delayed feeding skills and/or inefficient oral feeding)

-Displays psychosocial factors that may contribute to feeding disorder.

-Psychosocial impairments include developmental delays, mental and behavioral health problems in the child and/or caregiver, social influences that may impact child's behavior at meal time and environmental factors that may contribute to feeding disorders.

12. Know how to evaluate a child who is a picky/selective eater

-Obtain a detailed history of parental concern as well as review of systems, dietary assessment, anthropometric measures and physical examination. Observe a live or videotaped feeding interaction.

-Assess for behavioral and/or organic "red flags"

-If necessary, evaluate for possible parental misperception of food selectivity in the setting of neophobia (the rejection of foods that are novel or unknown to the child) as this is normal (peaks 18-24 months) and usually resolves with repeated exposures.

-Picky eaters usually try many foods but only like a few, although this may not resolve with repeated exposure (neophobia) these children typically grow and develop normally but there is concern for family discord and behavioral consequences.

-Evaluate for psychosocial factors affecting meal times and eating

13. Know how to evaluate a child with rumination

-Does vomiting occur within seconds or minutes after initiating a meal?

-If adolescent: do they leave the table multiple times during a meal to "throw up" mouthfuls of food in the bathroom?

-What does the regurgitated food look and taste like? Is it Undigested food?

-How long after eating does regurgitation happen?

-Does regurgitation wake your child from sleep?

-Can you hear retching or see that your child is about to regurgitate or vomit?

-Has your child taken medications to treat reflux? Do medications for reflux help?

-Ask about symptoms of comorbid anxiety or depression

-Some patients may also have associated stomach pain, indigestion, halitosis, nausea, weight loss

14. Know how to evaluate an infant or toddler with a feeding skill disorder

-Unsafe oral feeding (chocking, aspiration etc), delayed feeding skills (unable to consume age-appropriate liquid and food textures), inefficient oral feeding (prolonged meal time duration or inadequate intake).

-Impairment in oral sensory functioning and oral motor functioning can be evaluated visually

-Impairment in pharyngeal sensation or pharyngeal motor functioning requires assessment using a modified barium swallow study or fiberoptic endoscopic evaluation of swallowing.

Impairment (Body functions and impairments *)	**Dysfunction** (Activities and participation/limitations and restrictions*)
Oral sensory functioning • Under- or over-response to sensory aspects of liquids and food textures inhibiting acceptance and/or tolerance **Oral motor function** • Reduced strength, coordination, range of motion, timing inhibiting oral movements required for acceptance, control, manipulation and/or oral transit of liquids and food textures **Pharyngeal sensory processing and/or motor function** • Under- or over-response to bolus during pharyngeal transit or residue remaining post-swallow • Reduced strength, coordination, range of motion, timing impacting pharyngeal transit of liquids and food textures • Inhibiting efficient swallowing and/or airway protection	**Limitation in oral feeding skills** • Unable to consume age-appropriate liquid and food textures • Unable to use age-appropriate feeding utensils and devices • Unable to self-feed at age-appropriate expectations • Unable to use age-appropriate mealtime seating • Requires more assistance or requires special strategies relative to other children of same age • Prolonged mealtime duration • Insufficient oral intake **Restrictions in mealtime participation due to safety concerns:** • Adverse mealtime events (e.g. coughing, choking, gagging, vomiting, discomfort, stress, fatigue, refusal) • Adverse cardio-respiratory events (e.g. apnea, bradycardia, increased work of breathing) • Aspiration

Legend: * International Classification of Functioning, Disability, and Health (ICF) terminology

15. Be able to evaluate whether a child with oral-motor dysfunction is safe to feed orally

-Evaluate for history of safety concerns during oral feeding episodes including adverse mealtime events (e.g. coughing, choking, gagging, vomiting, discomfort, stress, fatigue, refusal) and/or adverse cardio-respiratory events (e.g. apnea, bradycardia, increased work of breathing)

-Obtain and Evaluate a swallow study

-If it is safe to feed orally but significantly delayed feeding skills or inefficient oral feeding present will need to consider g-tube supplementation to ensure adequate nutrition and weight gain.

16. Know how to plan the management for infants or toddlers with feeding disorders

-Offer a desired food contingency on the progressive acceptance of less desired foods

-"Food Chaining," the replacement of one food with a similar one

-"Fading" and "Shaping' – gradually altering the taste, color, texture and exposure to the food coupled with positive reinforcement

-Oral motor therapist (feeding specialist)

-May require behavioral psychologist and/or other specialty physicians to provide ancillary recommendations to address specific impairments related to medical conditions

17. Know the management of a child with picky eating

-Functional Analysis, Mealtime atmosphere, Social cues, Positive reinforcement, Repeated exposure, Forced exposure, providing information, combining foods

-Educating parents to have reasonable expectations and counseling them to consistently and repeatedly expose children to new foods (foods must be offered 8-15 times without pressure to achieve acceptance)

-"Hiding" pureed vegetables in sauces, using "dips" to enhance flavor, modeling eating, giving foods appealing names, involving children in food preparation and presenting it in attractive designs

18. Know the management of avoidant/restrictive food intake disorder

Cognitive-behavioral therapy

Feeding Therapy with a specialized speech/language therapist

19. Know the differential diagnosis of avoidant/restrictive food intake disorder

Other medical conditions (Food Allergies, Malabsorption problems etc.), specific neurological/neuromuscular, structural, or congenital disorders and conditions associated with feeding difficulties, reactive attachment disorder, Autism Spectrum Disorder, Specific phobia or other anxiety disorder, Anorexia Nervosa, OCD, Major depressive disorder, Schizophrenia spectrum disorders, Factitious disorder.

Resources:

Physiologic phases of swallowing

Developmental Stages in Infant and Toddler Feeding:
https://infantandtoddlerforum.org/media/upload/pdf-downloads/3.5_Developmental_Stages_in_Infant_and_Toddler_Feeding_NEW.pdf

Risk of feeding and eating disorders in infants and toddlers 0-3:
https://pediatrics.aappublications.org/content/137/2/e20152575

Evaluation of feeding skill disorder: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6314510/

Managing feeding difficulties:
https://pediatrics.aappublications.org/content/pediatrics/135/2/344.full.pdf

Content Category 19- Elimination Disorders

ABP Content Specs for Developmental-Behavioral Pediatrics Fellow Collaboration Project

Prepared by: University of Michigan DBP Fellows

Reviewed by: Dr. Barbara Felt, MD, University of Michigan – Staff/Faculty DBP

19. Elimination Disorders

A. Normal development of continence

1. Know the sex related variability in the development of urinary daytime and night time continence
2. Know the typical developmental course for acquiring stool continence in American children
3. Know the cultural variability in the development of urinary continence

B. Encopresis

1. Know the epidemiology of encopresis
2. Know the medical and psychosocial factors that may predispose to encopresis
3. Know the psychological and behavioral complications of encopresis
4. Understand the pathophysiology of encopresis
5. Understand the role of initial catharsis in the treatment of encopresis
6. Know how to evaluate a child with encopresis
7. Know the treatment of a child with encopresis
8. Understand the time course of recovery of normal bowel function during treatment for encopresis
9. Formulate the differential diagnosis for encopresis
10. Know the diagnostic criteria for encopresis

C. Enuresis

1. Recognize the medical disorders that can present with urinary incontinence
2. Know the pathophysiologic theories of primary enuresis
3. Know that genetic factors, including sex, may predispose a child to nocturnal enuresis
4. Understand the natural history, including spontaneous remission, of nocturnal enuresis
5. Differentiate between primary and secondary enuresis and their etiologies
6. Know how to evaluate a child with nocturnal enuresis
7. Know how to treat a child with nocturnal enuresis
8. Understand the theoretical psychological explanation for the efficacy of urinary alarms
9. Know the indications for pharmacologic management of nocturnal enuresis
10. Know how to evaluate a child with diurnal enuresis
11. Know how to treat a child with diurnal enuresis
12. Know the pharmacologic interventions for nocturnal enuresis
13. Know the epidemiology of enuresis
14. Know the potential side effects of medications used in the treatment of nocturnal enuresis

19. **Elimination Disorders**

A. Normal development of continence

1. Know the sex related variability in the development of urinary daytime and night time continence
   - Typical ages for developing continence differ by resource but based on data in U.S., girls gain skills earlier than boys.

	Daytime Stool	Night time stool
Girls	31.5 [30.0-33.3]	22.1 [20.7-24.3]
Boys	34.7 [33.6-36.5]	24.7 [23.0-25.5]

	Daytime urine	Night time urine
Girls	32.5 [30.9-33.7]	34.1 [31.9-35.8]
Boys	35.0 [33.3-36.7]	35.8 [33.5-37.8]

2. Know the typical developmental course for acquiring stool continence in American children
   - Sequence of acquisition to remember is: **BM night – BM day – Urine day – Urine night.**
3. Know the cultural variability in the development of urinary continence
   - Infant training:
     - In the late 19th century, infant training was more the norm in U.S.
     - Today, there is cultural variability regarding infant training. In the Digo culture in Kenya, infants are more likely to achieve urine continence by 6 mo.
     - Infant training involves: close parent observations of infant's cues; parent availability to get child to the right location in time to evacuate; pairing of a sound with the activity of urination or stooling to create a conditioned cue.
   - In the U.S., the age of toilet training drifted later over time; by the 1930s later toilet training (>18 mo) came into favor.
   - Switzerland longitudinal cohort 1950-60s: day urine continence 90% by 4 y and 97% by 6 y; night time urine continence at 6 y – 90% boys and 94% girls.
   - 1960-70s Brazelton emphasized a "child-centered" approach; when the child demonstrates readiness skills such as 2-hr periods of dryness; ability to communicate the need to go; ability to get to the toilet, etc.
   - Several other cultures today train earlier than U.S. including Asian, South American and African.

B. Encopresis

1. Know the epidemiology of encopresis
   - Rome V Criteria for Functional Constipation
     - ≥2 at least once/week, for at least 1 month (and does not meet IBS criteria)
       - ≤ 2 defecations in toilet / week **in child at least developmental age 4 y**
       - ≥ 1 fecal incontinence episode / week
       - History of retentive posturing or excessive volitional stool retention
       - History of painful or hard BMs
       - Presence of large fecal mass in rectum
       - History of large diameter stools that can obstruct toilet
       - Not better explained by another medical condition
     - Irritable Bowel Syndrome includes all of the following:
       - Abdominal pain ≥4 day/mo associated with one or more: related to defecation; change in stool frequency, change in appearance of stool
       - If constipation, pain no resolve with resolution of constipation

        - After appropriate evaluation, symptoms not fully explained by another medical condition.
- Definitions of Encopresis (see also #10 below):
    - DSM 5 Criteria for Encopresis
        - Repeated stool passage into inappropriate places
        - At least once/month x 3 months
        - Chronologic (or developmental) age ≥ 4 yr
        - Not explained by medication effects nor medical condition (except constipation)
        - Specify: with or without constipation and overflow incontinence
    - Primary encopresis: never achieved stool continence
    - Secondary encopresis: was continent for stool and now is not. This should raise concern about constipation or another problem.
- Constipation prevalence: 10-30% of children
    - 3-5% of Primary Care Pediatrician visits and 25% of GI visits
    - Peak incidence of problem onset is at toilet training age
    - No sex differences for constipation occurrence
    - ~95% of constipation cases are ultimately determined to be "functional" in origin
- Encopresis: 1-3 % of children; ~ 3% at age 4 years and 1.6% at age 10 years
    - More common in boys; affects 3-6 males for every affected girl.
    - Fecal soiling occurs in association with constipation ~ 80- 90% of the time.

2. Know the medical and psychosocial factors that may predispose to encopresis
- Overall: 3 ages/developmental periods when risks for developing problem is greater
    - 6-12 months – around the introduction of cereal
    - 2-3 years around the time of toilet training
    - 3-5 years at the start of school
- Predisposing medical factors:
    - **Uncomfortable stool passage**
    - Constipation
    - Stool impaction
    - Anatomic, metabolic, neuromuscular, trauma, other
    - Medication side effects
- Other factors:
    - Developmental delay, Cognitive Impairment
    - ADHD, oppositional behavior, Autism Spectrum Disorder
    - Anxiety, neglect, abuse
    - Usually there are not significant co-occurring psychiatric disorders
    - Family history of constipation
    - Note that volitional non-retentive encopresis is far less common

3. Know the psychological and behavioral complications of encopresis
- Child
    - Low self-esteem
    - ADHD, anxiety, oppositional behavior, obsessive compulsive behaviors
    - Bullying activity (victim or aggressor)
    - However, many mental health issues improve with management
- Family
    - Ashamed
    - Feelings of isolation

- Higher financial costs OTC
- Factors NOT associated with encopresis risk: SES, family size, parent age

4. Understand the pathophysiology of encopresis
- Normally, the colon absorbs water from waste traveling from the small intestine. A typical transit time is ~30-45 hr.
- For often common reasons, a child has an uncomfortable stool passage and begins a cycle of withholding of stool and the longer stool is withheld, the stool becomes more firm and of greater volume
- This leads to a vicious cycle of pain at passage → withholding of future stools → even more pain and larger stools.
- While a painful passage may be recalled, often the onset is subtle.
- As a consequence of increasing stool volumes at the rectum, physiologic changes occur and include:
  - Altered sensory threshold such that standard stool volumes are not perceived and larger volumes are required before the need to go is recognized.
  - Accommodation to the feeling of fullness, soiling and the smell.

5. Understand the role of initial catharsis in the treatment of encopresis
- Disimpaction: Clean out to relieve stool build up/impaction before beginning maintenance therapy
  - Oral - Osmotic, stimulants, high dose mineral oil
    - PEG 3350 recommended by NASPGHAN 1-1.5 g/kg/d for up to 6 days.
    - May require NG administration – GoLYTELY (PEG 3350 with electrolytes)
  - Rectal – enema, suppository
  - Combination – multimodal therapy helpful for very large stools
  - Tailor to constipation severity

6. Know how to evaluate a child with encopresis
- History
  - Time of meconium passage
  - Infant-toddler stool pattern – did the problems start early suggesting anatomic or other organic factors (slow transit, DM, thyroid dysfunction)
  - History of pain at defecation
  - Current stool pattern – note, ask caretakers and the child as toileting details may have been missed
  - Diet factors in past and present
  - Past evaluations for this problem or other conditions.
  - Past interventions for this problem and how that went
  - Medications tried
  - Behavioral approaches tried
- Other past medical history, social history and psychosocial factors, family history of this problem or related disorders of elimination.
- Physical exam
  - Growth
  - Vital signs
  - Abdomen
  - Back/spine
  - Anal placement and ability to squeeze and relax

    - Neurologic
- Other studies
    - Abdominal film if uncertain about stool burden after H&P
    - No other recommended studies unless indicated by H&P

7. Know the treatment of a child with encopresis
- Education
    - The GI system and how it functions
    - Prevalence of the constipation and encopresis problems and what the common risk factors are (and why it is almost always no one's fault).
    - Physiologic changes occur at the rectum that make the need for sit times so necessary.
    - Cleanouts are needed to reduce stool burden and set the stage for the colon to begin recovery
    - Chronic nature of this problem; this is a chronic illness and recovery requires time and adherence to the medical and behavioral program.
- Behavioral Program
    - Set realistic goals
    - Reward the work (the sits) not the outcome (the stool production)
    - Toilet sits: schedule and method
    - Collect data and review; Revise the plan
- Clean out to relieve stool build up/impaction
    - Oral - Osmotic, stimulants
    - Rectal – enema, suppository
    - Combination – oral and rectal
- Maintenance to reduce re-accumulation
    - Osmotic or fiber/fluid to soften stool
    - Stimulant to improve transit times
    - Regular follow up assessments

8. Understand the time course of recovery of normal bowel function during treatment for encopresis
- Recovery rates: 30-50% after one year; 48-75% after 5 years from diagnosis
    - Studies have been unable to identify factors that predict failure in treatment
    - However, severe constipation, abnormal Anorectal manometry (ARM) and/or balloon expulsion test are significantly related to treatment failure.
- In a Netherlands study of 401 children followed for about 11 years; 25% had symptoms in adulthood (Bongers ME et al, Pediatrics 2010).
    - Risk factors for poor outcomes: older age at onset, longer delay before effective treatment (seeing GI doc), fewer BM/wk at presentation
- Overall, long-term follow up studies suggest 25% still have symptoms in teen and adult years.

9. Formulate the differential diagnosis for encopresis
- GI and Metabolic
    - Irritable bowel syndrome
    - Celiac disease
    - Cystic fibrosis
    - Diabetes mellitus
    - Hypothyroidism
    - Hypocalcemia
- Anatomic

    - Anteriorly-displaced anus
    - Imperforate anus
    - Anal stenosis
    - Pelvic mass, abscess, fissure
- Neuromuscular
    - Hirschsprung Disease
    - Abdominal muscle abnormality: Downs Syndrome, Gastroschisis
    - Spinal cord: agenesis, tethered cord, tumor, myelomeningocele
- Mental Health
    - ADHD, ASD, ODD, CI
    - Eating disorder
    - Pharmacologic: iron, lead, chemotherapy, opiates, alpha-adrenergic, anticholinergics, antidepressants
- Other
    - Genetic, Dietary
    - protein allergy

10. Know the diagnostic criteria for encopresis
- DSM 5 Criteria for Encopresis
    - Repeated stool passage into inappropriate places
    - At least once/month x 3 months
    - Chronologic (or developmental) age ≥ 4 yr
    - Not explained by medication effects nor medical condition (except constipation)
    - Specify: with or without constipation and overflow incontinence

C. Enuresis

1. Recognize the medical disorders that can present with urinary incontinence
- Children could have one or several overlapping categories of disorders resulting in daytime urine incontinence.
- Overactive bladder
    - Abnormal bladder contraction during filling
    - Symptoms: urgency; holding behaviors (squatting / legs crossed) and are more likely to have other symptoms of constipation, history of UTI.
- Voiding postponement / underactive bladder
    - Habit of avoiding micturition leading to low voiding frequency and holding.
    - Overtime, the increased bladder capacity leads to stretch and weaker detrusor muscle, leading to weak contraction and emptying seen with an underactive bladder.
    - UTI is a risk of having an underactive bladder due to post-void residual.
- Dysfunctional voiding
    - Detrusor contracts against a closed sphincter (external urinary) due to inability to relax sphincter or pelvic floor during voiding.
    - If there isn't a neurologic reason, then it is called non-neurogenic dysfunctional voiding and the most severe form is Hinman-Allen syndrome.
    - Individuals with this problem are more likely to also have constipation, UTI, and vesicoureteral reflux (VUR).
- Other
    - Giggle incontinence
    - Vaginal voiding (due to legs too close during voiding and urine entrapment)

- Primary neck bladder dysfunction – delayed or incomplete opening of the bladder neck during voiding. Symptoms: hesitancy, urgency, weak urine stream, frequency, pelvic pain during voiding.
- UTI – always consider UA-UC
- Vesicoureteral Reflux (VUR) is associated with bladder dysfunction
- Constipation – 30 to >80% of children with bladder dysfunction have associated constipation.

2. Know the pathophysiologic theories of primary enuresis
- Voiding dysfunction involves problems filling or emptying the bladder and may be a part of the presentation of "bowel and bladder dysfunction" in children having both problems.
- Normal voiding:
  - Involuntary voiding in infancy
  - Progressive maturation and becoming aware of
    - bladder filling
    - increase bladder capacity
    - voluntary control of detrusor and urethra sphincters.
  - Bladder capacity: Age in years + 2 x 30 = # ml
- Dysfunction may result from any problem with: innervation of bladder or sphincter; bladder capacity; structure of outlet; or function of detrusor. Categories:
  - Neurogenic reasons: disruption of the external sphincter or bladder innervation that may have occurred in early development or as a result of trauma.
  - Anatomic reasons: obstruction of outlet or abnormal placement of tract relative to bladder and sphincters. VUR is a risk factor for bowel and bladder dysfunction.
  - Functional reasons: idiopathic – no known cause but thought to be due to delayed maturation of normal bladder function and toileting habits.

3. Know that genetic factors, including sex, may predispose a child to nocturnal enuresis
- 68% of monozygotic twins and 36% of dizygotic twins
- 44% of offspring are affected if one parent reports a history of nocturnal enuresis
- 77% of offspring are affected if both parents report a history of nocturnal enuresis
- Males are twice as likely to suffer from enuresis

4. Understand the natural history, including spontaneous remission, of nocturnal enuresis.
- In almost all cases nocturnal enuresis resolves spontaneously when there are no other urinary tract symptoms nor bladder dysfunction issues.
- T11-L2 sympathetic nerves relax the detrusor muscle so the bladder can fill and contract the bladder neck to contain the urine. S2-S4 parasympathetic nerves activate the detrusor muscle to increase pressure and relax the bladder neck to empty the bladder. Pontine micturition center and sacral nerves communicate to activate voluntary control and inhibit bladder emptying.
- Bladder capacity increases between ages 2-4, which is also necessary to make it through the night. Children are aroused by a full bladder during the night. They also need to concentrate their urine at night and not overcome their bladder capacity. Delay of any of these elements result in enuresis, and maturation improves for a majority of patients over time.
- Children with developmental delays of language/gross motor have an increased risk of nocturnal enuresis.
- Children with ADHD frequently experience incomplete emptying. Children with ADHD have a higher risk of bedwetting and urinary dysfunction, they are also less successful with bed wetting treatments by alarm or medications which may be due to compliance.

5. Differentiate between primary and secondary enuresis and their etiologies
- Daytime incontinence is intermittent urine leakage generally after age 5 years.
- Primary enuresis: not having attained at least 6 months daytime continence after age 5 y
- Secondary enuresis: onset of daytime incontinence after attaining at least 6 months of daytime continence after age 5 years.

6. Know how to evaluate a child with nocturnal enuresis
- History (urologic and neurologic disorders are more common in children with daytime symptoms)
  -urgency
  -holding maneuvers
  -interrupted micturition
  -weak stream
  -straining
  -prolonged periods of dryness (6 months)
  -frequency and trend of nocturnal enuresis
  -fluid intake diary (majority of fluids in the afternoon/evening have a greater risk of nocturnal enuresis, fluid intake that exceeds the maintenance requirements may indicate DM, DI, primary polydipsia)
  -stool history (constipation or fecal incontinence)
  -prior interventions tried
  -associations with sleep, or other medical concerns such as UTI, sickle cell disease, DM, gait or neurological challenges
  -family history of nocturnal enuresis
  -social/emotional concerns that could be associated with a secondary enuresis
  -readiness for treatment
  -screen for developmental and behavioral comorbidities
- Physical exam
  -poor growth/hypertension- concerns for kidney pathology
  -tonsillar hypertrophy- OSA
  -stool palpable on abdominal exam- constipation
  -perianal excoriation- vulvovaginitis or pinworms
  -abnormal lumbosacral area- spinal cord abnormality
- Urinalysis
  -first morning void is best to differentiate from water intoxication and DI. Reflex urine culture if concerning for UTI
- Voiding diary
  -include timing of daytime voids, volume of voids, lower urinary tract symptoms (difficulty stopping or starting stream, dribbling, sensation of incomplete emptying)
- Additional studies include: renal ultrasound, voiding cystourethrogram, ultrasound for postvoid residual volume, urine culture
  -Reserved for those who will be referred for daytime incontinence, increased or decreased voiding frequency suggesting bladder dysfunction, weak stream and use of abdominal pressure, proteinuria, nausea, weight loss, fatigue (suggesting kidney disease), excessive thirst, nighttime drinking (polydipsia or kidney disease), abnormalities of the spine
- Referrals: sleep medicine, nephrology, urology, neurosurgery, child psychologist

7. Know how to treat a child with nocturnal enuresis

- Appropriate referrals depending on the comorbidities.
- Make sure the family and patient are motivated to start treatment.
- Discuss parent and child short term goals prior to treatment- desire to attend a sleepover, etc
- Discuss treatment can be prolonged, need frequent follow up, is associated with relapses, and need full cooperation from the family and patient.
- Educate parents that this is a common condition, it is not the child nor the caregiver's fault; use neutral communication when accidents occur and have the child participate as it is possible with the cleanup process
- Keep a calendar of dry and wet days to track progress
- Sit times of 4-7 times per day, with first sit upon waking up and a sit prior to going to bed
- Avoid caffeinated and high sugar drinks
- Try to drink 40% of fluid intake in the morning
- Try to avoid diapers/pull ups at night, protection measures for the mattress are ok to use
- Rewards should be given for complying with requests to sit, participation with medications/routine, and not for dryness. Bigger rewards can be offered for long periods of dryness or continuing with the program for a long period of time. It is not recommended to use negative reinforcement. (sticker charts, favorite breakfast in the morning, a new book, are some examples)
- Consider psychotherapy to help with self-esteem issues or discouragement due to the activities they miss related to this problem (sleepovers, etc) if there is no improvement after 3-6 months
- Enuresis alarm- lower relapse rates; needs family cooperation as a team for up to 3-4 months. Parents often hear the alarm and need to help the child wake at the beginning of treatment. 30% of families discontinue the alarm usually due to disturbance to others in the family, and failure to wake the child.
- Desmopressin –higher relapse rate, more short term relief, useful for families with more negative reactivity from parents for bedwetting, more helpful for children with nocturnal polyuria. Avoid if history of hyponatremia. Avoid excess fluid intake in the evening especially after taking the DDAVP.
- Waking the child to use the bathroom after they have fallen asleep is not recommended by some authors as it does not condition the child to wake from the sensation of a full bladder.
- Bladder training to ask the child to hold their urine and increase their intervals is also not recommended. Increased bladder capacity does not necessarily improve enuresis rates.
- Electrical stimulation therapy (neuromodulation or neurostimulation) an implantation device to stimulate pelvic muscles to contract or modulate detrusor contractions is not routinely recommended.
- Hypnosis, psychotherapy, chiropractic approaches have limited evidence. Acupuncture may have a growing body of literature supporting some benefit.

8. Understand the theoretical psychological explanation for the efficacy of urinary alarms.
    - The alarm conditions the child to wake or inhibit the bladder contracting in response to the physiologic conditions present before wetting.
    - General tips for alarm use
        - Use the alarm every night.
        - When the alarm goes off, have the child turn off the alarm, finish voiding in the toilet if possible, change bedding and wet clothes, reattach the alarm, go back to bed (child can practice this routine and/or visualize the steps prior to going to bed to help with the process)
        - Positively reinforce if the child complies with this routine and later, for dry nights.

    - Follow up 2 weeks after alarm initiation to help adherence to this approach and continue for 3 months.
    - Watch for and point out early positive signs such as smaller wetting accidents, accidents occurring later in the sleep period, fewer alarms, child waking to the alarm
    - Continue the alarm if there are improvements, even if they are not dry for >14 consecutive days. Restart alarm for any relapses.
- Alarms cure 2/3 of affected children with a low relapse rate

9. Know the indications for pharmacologic management of nocturnal enuresis
    - Offer the choice of desmopressin for families who want more immediate results, or after 3 months without improvement of the alarm. Can take the medication concurrently with the alarm, or restart the alarm in 6-12 months.
    - Use the oral form one hour prior to bedtime.
    - Titrate from 0.2 mg tab to most effective dose (max 0.4-0.6 [depending on resource]).
    - Try a trial of medication if plan to use if for sleepovers only
    - Drink no more than 6 oz of fluids one hour prior to bed, and for 8 hours after administration
    - There is generally no need to follow labs on this medication but be watchful for electrolyte disturbance if there is a co-occurring risk factor (renal impairment, dehydration). Consider discontinuation during illnesses.
    - Continue for 3 months. Taper and withhold for 1 week to see if it can be discontinued. Taper dose by cutting it in half for 2 weeks as this has shown better success than stopping abruptly.
    - Failure may be due to reduced bladder capacity
    - DDAVP is more affective for nocturnal polyuria and lasts 7 hours

10. Know how to evaluate a child with diurnal enuresis.
    - History
        - Age of onset of concern
        - Frequency of voids and incontinence episodes and volume estimates
        - Symptoms of urgency, pain, holding, stream strength, straining
        - History of UTI
        - History of Constipation
        - Diet
        - History of spinal cord trauma or other neurologic dysfunction
        - Toilet training history
        - Primary vs secondary daytime incontinence
        - History of sexual abuse
    - PMH
        - Evidence of perinatal or neonatal insult – anoxia or infection.
        - Diabetes
        - Neurodevelopmental delay
        - Behavioral disorders – 25-50% prevalence of associated ADHD, anxiety, depression.
        - Family history of urologic/incontinence problems
    - Physical
        - Examine lower back for signs of occult spine problem – patch of hair or a dimple, gluteal fold difference or lipoma
        - Neurologic assessment of lower extremity strength and DTRs, gait, anal wink and tone/strength
        - Abdominal exam for stool / colonic distension.
        - Inspect genitals for stenosis of meatus, labial adhesions, fissures, rash.

    - Observe urine stream
- Tests
    - Urinalysis (preferably first morning void) to screen for ability to concentrate, glucose spilling, leukocyte esterase and nitrate for UTI screen, hematuria
    - Follow along culture if indicated
    - Other as indicated by history and consider evaluation by Urologist.

11. Know how to treat a child with diurnal enuresis.
    - Treat underlying or co-occurring factors including UTI
    - Education about normal urinary tract and bladder function and expectations by age
    - Adequate fluids & adjust timing such that 2/3 occur in earlier part of day
    - Regular toilet opportunities, check sitting position and sit duration to encourage complete evacuation
    - Avoid caffeine

12. Know the pharmacologic interventions for nocturnal enuresis
    - Anticholinergics such as oxybutynin is used to relax the detrusor muscle for children with small bladder capacity and uninhibited detrusor contractions. It may be helpful for daytime symptoms or in combination with desmopressin to increase bladder capacity during sleep.
    - TCAs such as imipramine stimulates vasopressin secretion and relaxes the detrusor muscle. Review cardiac history prior to starting imipramine. Child takes this orally one hour before bedtime. The initial dose is 10 to 25 mg; it may be increased by 25 mg if there is no response after one week (maximum dose 50 mg for children 6 to 12 years of age; maximum dose 75 mg for children ≥12 years of age). Imipramine can be held for 2 weeks every three months to decrease the risk of tolerance
    - Discontinuing DDAVP and imipramine is associated with a relapse rate of 60%

13. Know the epidemiology of enuresis.
    - At 4-6 years, about 20% have occasional daytime wetting and about 3 percent wet 2-3x/wk.
    - Daytime urine incontinence defined as accident at least once/2 weeks:
        - 10% 3-5 yo
        - 5% 6-12 yo
        - 4% 12-18 yo
    - Studies across cultures also demonstrate prevalence of daytime incontinence:
        - In Australia – 17% of parents noted wetting for their 7 yo children
        - In Korea – 17% of school-age children had urgency with or without urine accidents and the incidence declines with age.
    - In general, boys > girls at younger ages but the gender difference declines with age.

14. Know the potential side effects of medications used in the treatment of nocturnal enuresis.
    - The most concerning adverse reaction for imipramine is cardiotoxicity with cardiac conduction disturbances and myocardial depression, particularly in cases of overdose. Other less common side effects are neurologic symptoms, including nervousness, personality change, and disordered sleep. They also carry a boxed warning regarding the possibility of increased suicidality, particularly in individuals with preexisting depressive symptoms
    - Hyponatremia and seizures can occur with desmopressin

- Oxybutynin side effects are constipation, sweating less, dry mouth, fatigue, headache, blurry vision.

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- *KG Nepple, MD, CS Cooper, MD, Evaluation and diagnosis of bladder dysfunction in children.*
- *ND Tu, MD, LS Baskin, MD Nocturnal Enuresis in Children: Etiology and Evaluation.*
- *ND Tu, MD, LS Baskin, MD Nocturnal Enuresis in Children: Management.*

Content Category 19- Elimination Disorders

ABP Content Specs for Developmental-Behavioral Pediatrics Fellow Collaboration Project

Prepared by: Beatrice Egboh, MD, University of Nebraska, DBP Fellow

Reviewed by: Howard Needelman, MD, University of Nebraska, DBP Fellowship Director

Final Version/Faculty Reviewed

Elimination Disorders

A. Normal development of continence

1.Know the sex related variability in the development of urinary daytime and nighttime continence
2.Know the typical developmental course for acquiring stool continence in American children
3.Know the cultural variability in the development of urinary continence

B. Encopresis

1.Know the epidemiology of encopresis
2.Know the medical and psychosocial factors that may predispose to encopresis
3.Know the psychological and behavioral complications of encopresis
4.Understand the pathophysiology of encopresis
5.Understand the role of initial catharsis in the treatment of encopresis
6.Know how to evaluate a child with encopresis
7.Know the treatment of a child with encopresis
8.Understand the time course of recovery of normal bowel function during treatment for encopresis
9.Formulate the differential diagnosis for encopresis
10.Know the diagnostic criteria for encopresis

C. Enuresis

1.Recognize the medical disorders that can present with urinary incontinence
2.Know the pathophysiologic theories of primary enuresis
3.Know that genetic factors, including sex, may predispose a child to nocturnal enuresis
4.Understand the natural history, including spontaneous remission, of nocturnal enuresis
5.Differentiate between primary and secondary enuresis and their etiologies
6.Know how to evaluate a child with nocturnal enuresis
7.Know how to treat a child with nocturnal enuresis
8.Understand the theoretical psychological explanation for the efficacy of urinary alarms
9.Know the indications for pharmacologic management of nocturnal enuresis
10.Know how to evaluate a child with diurnal enuresis
11.Know how to treat a child with diurnal enuresis
12.Know the pharmacologic interventions for nocturnal enuresis
13.Know the epidemiology of enuresis
14.Know the potential side effects of medications used in the treatment of nocturnal enuresis

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Normal Development of Continence

- Bowel maturation typically precedes bladder maturation.
- Continence proceeds from night time stool bowel continence, then daytime bowel continence, then daytime urine continence, then nighttime urine continence
- 15% to 20% of children will become partially toilet trained but continue to have wetting accidents after age 5.
- Infant voiding is a spinal reflex.
- Voluntary control over bowel and bladder reflex actions (expected to emerge at age 9 months), ability to cooperate with training (expected to emerge at age 18–24 months).
- Stool is held in the rectum by the internal anal sphincter, innervated by ganglion cells, and the external anal sphincter and puborectalis muscle, innervated by the pudendal nerve (S2-4).
- The distention of the rectosigmoid results in stimulation of the recto-rectal reflex (contraction of the rectum and relaxation of the internal anal sphincter). The child then has the urge to defecate.
- Voluntary control of defecation is by contracting the external anal sphincter and puborectalis (part of the pelvic floor), reducing the recto-rectal reflex
- The ability to voluntarily hold urine is by detrusor relaxation (mediated by sympathetic fibers T10-L2), internal urinary sphincter (which is continuous with the detrusor muscle), and external urinary sphincter contraction (mediated by the pudendal nerves S2-4).
- Bladder emptying is by detrusor contraction (mediated by parasympathetic nerves S2-4) and relaxation of the external urinary sphincter.
- By age 2, 30% of children detect urge to void urine.
- AAP text says by the 1990's average age for toilet training 35-39 months of age, increased over the years with girls 2-3 months earlier than boys
- By age 4, all typical children detect urge to void urine.
- Daytime Urinary Continence: 5yrs
- Night time Urinary Continence: within 1 year of daytime
- The International Children's Continence Society recommends that at age 5, daytime urinary incontinence should be evaluated.

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URINARY CONTINENCE.

Sex Related Variability in The Development of Urinary Daytime and Nighttime Continence.

- Girls are about twice more likely to have daytime symptoms than boys are
- Nocturnal incontinence is more common in boys

Daytime Incontinence/Diurnal Enuresis: repeated voiding of urine into clothes during daytime hours that occurs at least twice a week for at least 3 consecutive months or that causes clinically significant distress or impairment of function in children 5 years of age chronologically or developmentally.

Night time incontinence/Nocturnal Enuresis: Discrete episodes of urinary incontinence during sleep after 5 years chronologic or mental age.
Any wetting that occurs in discrete amounts at night is termed enuresis regardless of whether it is associated with daytime symptoms.

Primary nocturnal enuresis is defined as nocturnal wetting in a child who has never been dry on consecutive nights for longer than 6 months.

- Types:
    - **Monosymptomatic enuresis** – Enuresis in children without any other lower urinary tract symptoms.
    - **Non-monosymptomatic enuresis –** Enuresis in children with other lower urinary tract symptoms (e.g., increased frequency, daytime incontinence, urgency, genital or lower urinary tract pain).

Secondary enuresis is defined as new-onset nighttime wetting on consecutive nights after a 6-month or greater period of dryness.

Epidemiology of Daytime Incontinence.

- Repeated voiding of urine into clothes during daytime hours that occurs at least twice a week for at least 3 consecutive months or that causes clinically significant distress or impairment of function in children 5 years of age chronologically or developmentally.
- Prevalence: 8%
- More common in Girls.
- Includes: overactive bladder, voiding postponement, underactive bladder, dysfunctional voiding, stress incontinence, vesicovaginal reflux

Pathophysiology:

- Loss of cortical control: Brain lesion; seizures

- Spinal cord lesions/ tethered cord
- Neural:
    - Peripheral Nerve lesions e.g. to pudendal nerve injury; peripheral neuropathies
- Congenital anatomic anomalies: Posterior urethral valve; urethral stenosis, ectopic ureter
- Trauma to urethral sphincter.
- Labial adhesion
- Vaginal reflux
- Stress Incontinence
- Bladder wall irritation
- Constipation
- Emotional Stress (PTSD; Sexual Abuse)
- Excessive urine production: diabetes mellitus, diabetes insipidus, sickle cell disease with reduced urine concentration, volume overload, and diuretic agents
- Hinman syndrome /**non-neurogenic, neurogenic bladder** (is a voiding dysfunction of the bladder of neuropsychological origin that is characterized by functional bladder outlet obstruction in the absence of neurologic deficits).
- Voiding postponement
- **Functional voiding disorders:** heterogeneous group of disorders that account for **daytime** incontinence in most children. Physical and neurologic examinations are normal.
    - Overactive bladder or urge incontinence
    - Giggle incontinence
    - Underactive bladder

Evaluation of a Child with Daytime Incontinence.

History:

- A voiding diary with timing.
- Irritative urinary symptoms: Urgency, frequency, and dysuria
- Obstructive symptoms: Hesitancy, straining, and a weak urinary stream (urethral obstruction)
- Children with ectopic ureter or posterior urethral valves may be constantly wet.
- Back pain and gait abnormalities.
- Triggering psychological events.
- Family response to a child's incontinence
- Emotional impact of incontinence on the child.
- Sexual or physical abuse.

Physical Exam:

- Growth parameters
- Vital signs including blood pressure (sign of renal disease).
- Abdominal: palpable constipated stool masses, bladder
- Back/Spine: vertebral anomalies or cutaneous markers of spinal dysraphism, Neurologic: anal wink and cremasteric reflexes in boys.

Initial Diagnostic Test:

All children with daytime incontinence require **Urinalysis.**

A random specific gravity of less than 1.002 (or less than 1.025 after 12-hour fluid restriction) → inability to concentrate urine.

Glucosuria → diabetes mellitus.

Nitrites or white blood cells → urinary tract infection.

Treatment.
Treat underlying cause e.g. constipation may resolve symptoms.
For children with concomitant daytime and nighttime symptoms, address daytime symptoms first and then the night symptoms, which may remit after daytime symptoms are treated.

Epidemiology of Nighttime incontinence/ Enuresis

- 15% to 20% of 5-year-olds.
- 15% annual spontaneous remission rate.
- 0.5% to 3% remain enuretic as adolescents and adults.

Enuresis DSM 5 Diagnostic Criteria for Enuresis.

A. Repeated voiding of urine into bed or clothes (whether involuntary or intentional)
B. Behavior must be clinically significant as manifested by either a frequency of twice a week for at least 3 consecutive months or the presence of clinically significant distress or impairment in social, academic (occupational), or other important areas of functioning.
C. Chronological age is at least 5 years of age (or equivalent developmental level).
D. The behavior is not attributable to the physiological effect of a substance (such as a diuretic, antipsychotic or SSRI) or to another medical condition (e.g., diabetes mellitus, seizure disorder, spina bifida).

Pathophysiologic Theories of Primary Enuresis:

Not completely understood. Several theories:

I. Association with Obstructive sleep apnea;
↑ atrial natriuretic factor→ inhibits the renin-angiotensin-aldosterone pathway→ ↑ diuresis
Nocturnal polyuria due to alterations in vasopressin release has been shown to be a factor in nocturnal enuresis; children with nocturnal polyuria have abnormal circadian release of antidiuretic hormone (ADH).
II. Smaller-than-normal functional bladder capacities at night, and urodynamic studies have demonstrated higher bladder instability at night compared with during the day.
III. Genetics: strong familial predisposition; twin concordance. Parental age of resolution often predicts when child's enuresis will resolve.
IV. Neurodevelopmental factors: ↑ in children with ADHD, Autism Spectrum Disorder, Intellectual Disability.
V. Maturation Delay hypothesis: commoner in children with more fine and gross motor clumsiness, perceptual dysfunction, and speech defects

Pathophysiology of secondary enuresis:

- Usually not related to an organic cause.
- Underlying stressful event, such as the birth of a sibling, a move, or the death of a parent or grandparent
- Should be evaluated and treated like primary enuresis.

Evaluation of Enuresis.

History:

- Determine whether the enuresis is primary or secondary.
- Pattern of enuresis: the number of nights per week and the number of episodes per night.
- Pattern of nighttime fluid intake.
- Caffeine intake.
- Other symptoms: polyuria, polydipsia, urgency, frequency, dysuria, abnormal urine stream, history of urinary tract infection, constant wetness, and bowel complaints (15% of children who have enuresis also have encopresis).
- History of sleep disorders such as sleep apnea or insomnia
- Neurologic and developmental history
- Rule out secondary causes, including:
    - Chronic Kidney Disease
    - Seizures
    - Diabetes Mellitus
    - Obstructive sleep apnea
    - Pin worms
    - Psychogenic polydipsia
    - Urinary tract infection
    - Bladder Dysfunction
    - Posterior Ureteral valves
    - Ectopic ureter in girls
    - Fecal incontinence
    - Sickle cell disease

Examination/ Test:

- Focus on the gastrointestinal (GI), urogenital, and neurologic systems.
- Most children who have PNE have normal exam.
- Urinalysis.

Treatment:

Goal:

- Stay dry on particular occasions.
- Reduce number of wet nights.
- Avoid Recurrence.
- Reduce emotional effects on child and family.

Interventions:

Watchful waiting and education: high rate of spontaneous resolution

Behavioral Modification

- Ensure child is well hydrated during day.
- limiting nighttime fluid intake 2 hours before bedtime, limiting dairy products 4 hours before bedtime (to decrease urine output from osmotic diuresis)
- voiding prior to going to sleep.

Motivational therapy

Enuresis alarms

- Success rates as high as 66% to 70%.

- It is the most difficult method to employ.
- Its mechanism is unknown, ??? a conditioned response.
- The alarm must be used every night for success and may require 3 to 4 months for results.
- Child is considered cured if he or she has worn the alarm for 1 month and it is not triggered because he or she remains dry.
    - Treatment duration 2-3 months

Medications

Desmopressin- synthetic analog of ADH.

- Reduces urine output overnight
- Response rate (50% reduction in wet nights) is 60-70%.
- Side Effects of Desmopressin.
    - Patients should be cautious of fluid intake when taking medication to prevent potential water intoxication and hyponatremia and should have their therapy interrupted during acute illnesses that may lead to fluid and/or electrolyte imbalance.
    - Increased thirst.
    - Confusion; Headache dizziness
    - Decreased urine output
    - Dizziness
    - Irregular heartbeat
    - Nausea and vomiting
    - Increased thirst
    - Muscle pain or cramps
    - Seizures
    - Edema
    - Unusual tiredness or weakness

Imipramine (a tricyclic anticholinergic agent)

- Exact action unknown; a weak anticholinergic effect as well as an antispasmodic effect on the detrusor muscle; increase ADH release.
- *The lack of bladder selectivity and the effects on other organ/system reduces its use.*
- Side Effects of Imipramine.
    - As with other antipsychotics, increased the risk of suicidal thinking and behavior in children, adolescents, and young adults in short-term studies with major depressive disorder (MDD) and other psychiatric disorders.
    - Tardive Dyskinesia
    - Renal /liver disease
    - Elevate blood glucose
    - Bone marrow suppression
    - Anticholinergic effects
    - Lower seizure threshold

- Long QT – significant side effect which limits the use in the pediatric population

Oxybutynin – antispasmodic, anticholinergic
- represses detrusor activity;
- side effects of constipation, dry mouth, fatigue, mood changes
- Used for overactive bladder

ENCOPRESIS.

Constipation: is defined as a delay or difficulty in defecation, present for 2 or more weeks, and sufficient to cause significant distress to the patient (NASPGHAN).

ROME IV Criteria for Functional Constipation.
Must include 2 or more of the following occurring at least once per week for a minimum of 1 month with insufficient criteria for a diagnosis of irritable bowel syndrome in infants up to 4 years of age:

1. 2 or fewer defecations per week
2. History of excessive stool retention
3. History of painful or hard bowel movements
4. History of large-diameter stools
5. Presence of a large fecal mass in the rectum

In toilet-trained children, the following additional criteria may be used:

6. At least 1 episode/week of incontinence after the acquisition of toileting skills
7. History of large-diameter stools that may obstruct the toilet

Fecal continence: is the ability to recognize when the rectum is full, discriminate between solid, liquid stool or gas and the ability to retain the content until emptying is convenient.

Encopresis is repetitive voluntary or involuntary fecal soiling at age 4 and older in inappropriate places.
Primary cause is a physical predisposition to constipation, reduced bowel regularity, and ineffective evacuation.

DSM-5 Diagnostic Criteria.

A. Repeated passage of feces into inappropriate places, whether involuntary or intentional.
B. At least one such event occurs each month for at least 3 months.
C. Occurs in children at least age 4 years (or of equivalent developmental level).
D. The behavior is not attributable to the effects of a substance, e.g., laxative, or another medical condition, with the exception of a mechanism involving constipation.

Specify whether:

- With constipation and overflow incontinence: through physical examination or medical history, there is evidence of constipation.
- Without constipation and overflow incontinence: through physical examination or medical history, there is no evidence of constipation.

Types of Encopresis.
Retentive: encopresis associated with functional constipation
Non-retentive: encopresis without constipation; it is usually a behavioral problem as a method of control. There is no association of non-retentive encopresis with other behavioral disorders.

Epidemiology of Encopresis.
- 3% of 4-year-old and 1.6% of 10-year-old children
- More common in the 5- to 10-year-old group
- More common in boys (3-6X) than in girls
- No correlation with socioeconomic status, family size, child's position in family, or parental age
- The time of symptom onset to the time of diagnosis ranges from 1 to 5 years

Medical and Psychosocial Factors That May Predispose to Encopresis.
- Functional: Over 90% of encopresis is functional (not caused by organic defect or illness).
    - Passage of a painful stool → fear of defecation → withholding behaviors
    - Negative toilet training practices → associate using the potty with punishment
    - Unsubstantiated fears e.g. fear of falling into the potty; be flushed away
    - Shy about using public toilets/ finding public toilets unsanitary.
    - lack of access to the toilet.
    - disorganized home setting.

- Organic: 5-10%
    - Anatomic: corrected imperforate anus, ectopic anus, anal stenosis, Hirschsprung's disease, Carcinoma.
    - Neurologic: Hirschsprung disease, neuronal intestinal dysplasia, Chagas disease, Spinal cord damage due to spinal dysraphism, tumors, trauma , Visceral myopathy, visceral neuropathy

- Metabolic: Celiac disease, Endocrine tumors, including pheochromocytoma, cause diarrhea, which may result in non-retentive encopresis.
- (No report of Hypothyroidism and hypoparathyroidism leading to encopresis).
- Iatrogenic: Laxative abuse → severe diarrhea and fecal incontinence

Psychological Triggers of Encopresis.

- Toilet training
- Separation (e.g. starting school or divorce)
- Change in the child's daily schedule
- Troublesome, threatening, or confusing event associated with certain individuals (family, peers, caregivers, teachers) or places (school bathroom)
- Sexual abuse
- Exposure to trauma or violence
- Children with attentional deficits, obsessions and compulsions, and oppositional behavior are more prone to encopresis
- Behavioral rigidity decreased social and communication skills as seen in ASD
- Inability to communicate the urge to defecate

Psychological and Behavioral Complications of Encopresis.

- Fear or aversion to use of the toilet.
- Anxiety around toileting
- Disrupted family dynamics and struggles between parent and child around use of the toilet
- Smell of their fecal soiling → devastating effects on school and social interactions
- Sadness, feeling alone, being upset, guilty, embarrassed, low self-esteem,
- Effects on family: isolation, worry, stress, time and financial burden.

Pathophysiology of Encopresis.

Retentive Encopresis (same pathophysiology as functional constipation).
Dietary changes, separation, changes in schedules or other psychosocial stressors that result in distressing experience associated with defecation
(3 major periods: Introduction of cereal or solids; Toilet training and start of School);
↓
Child wishes to avoid these experiences resulting in 'Stool withholding.'
↓
Further contributes to constipation.
It is unclear why some children with constipation develop encopresis and others don't.

Non-retentive: Pathophysiology is unclear, there are some association with psychological and behavioral symptoms, including attention problems and anxiety and mood disorders.

How to Evaluate a Child with Encopresis.

History:

- Age meconium passed
- Stool characteristics
- Symptom onset/ triggering event.
- Frequency and consistency
- Stools Passage problems: pain, withholding, fissures bloody diarrhea
- Other symptoms include appetite, abdominal , fatigue, fever, bilious vomiting, rash
- Past /current treatment
- Poor adherence/ Good adherence to treatment
- Diet: fiber, fluid, dairy intake
- Development
- Delayed toilet training
- Developmental disability/delay
- Behavior
- Medication
- Family history

Behavior rating Scales assessing for Psychological Factors:

- Pediatric Symptom Questionnaire
- Child behavior Checklist
- Behavior Assessment Scale for Children
- Child Depression Checklist.

Physical Exam:

NASPGHAN (North American Society for Pediatric Gastroenterology, Hepatology and Nutrition) recommend examination should include **Back, Perineum and Anal region.**

General: Growth parameters

Abdomen: Distention; palpable stool

Back: skin/spine: look out for sacral agenesis, pilonidal dimple, hair tuft, gluteal cleft deviation

Anal: Stool in anus, clothes, fissures/ scars, location of anus (normal vs anteriorly displaced).

Evidence does not support digital rectal examination.

Neurologic: Present anal wink; gait, tone strength, and reflexes of lower limbs

Extreme fear during anal inspection.

Further Tests:

Guided by index of suspicion.

Poor growth/ family history: metabolic studies to rule out hypothyroidism and celiac disease.

Laxative abuse: basic metabolic panel along with assessment of serum
History of enuresis/ symptoms suggestive of UTI: Urinalysis and culture.
Abdominal plain radiography: according to NASPGHAN, evidence supports not using an abdominal radiography to diagnose functional constipation.
Biopsy: to rule out Hirschsprung disease

Treatment of a Child with Encopresis.

Medical management of the underlying constipation, if present, and behavioral intervention to encourage the child to use the toilet regularly.
Education is very important as this can be emotionally overwhelming for a child and family.
It should include information on

- GI system and function
- Physiologic changes
- Common condition
- Common risk factors
- Importance of fecal dis-impaction/ clean out.
- Chronic nature – recovery
- Commitment required

Fecal Dis-impaction:

- According to NASPGHAN, Polyethylene glycol (PEG) and enemas are equally effective for fecal dis-impaction.

Maintenance Therapy:

- PEG is more effective compared with lactulose, milk of magnesia, mineral oil, or placebo. Use of lactulose as the first-line maintenance treatment is recommended, if PEG is not available.
- Addition of enemas to the chronic use of PEG is not recommended.
- Use of milk of magnesia, mineral oil, and stimulant laxatives may be considered as an additional or second-line treatment.
- Maintenance treatment should continue for at least 2 months.
- All symptoms of constipation symptoms should be resolved for at least 1 month before discontinuation of treatment.
- Treatment should be decreased gradually.
- In the developmental stage of toilet training, medication should only be stopped once toilet training is achieved.

Behavior Modification.

- Toilet Sitting
- Reward System
- Monitoring

Differential diagnosis for encopresis.

- Chronic Diarrhea
- Anal Stenosis
- Spina Bifida

TOILET TRAINING APPROACHES

Contemporary toilet training is derived from 2 models of toilet training

1. child-oriented gradual training: a parent systematically responds to a child's signals of toilet "readiness,"
2. structured-behavioral, endpoint-oriented training: a process of eliciting a specific chain of independent toileting behaviors.

Behavioral Approach:

- Positive reinforcement encourages motivation, sticker charts.
- A wrist alarm watch that sounds at set intervals minimizes child-parent conflict, optimizes a child's sense of achievement, and may serve as an additional reward
- Regular voids every 2-3 hours regardless of sensation of the need to go; can be scheduled around natural breaks in the day, such as meals, recess, and snack
- Family education: Instruct families not to punish the child/ no negative remarks; the disorder is neither the fault of the child nor under his or her voluntary control.
- The child taking responsibility to change his or her clothes after wetting.

Social learning theory approach

- Learning occurs by observing the behaviors of others as well as the consequences of others' behavior.
- Social learning theory – used in a variety of toilet training approaches:
    - Examples: use of a potty doll (Imaginary play), reading of potty books with a "social story" approach, observing peers at school or older siblings using the toilet at home.

Child-oriented approach (Brazelton)

- Based on the abilities and skills of the individual child when they show signs of readiness (usually beginning around 18 months old)
- Brazelton identified 3 factors necessary to accomplish continence:
    - Physical ability (ability to sit, pull up/down pants, awareness of internal needs for elimination)
    - Response to external feedback (understanding and following directions)
    - Response to internal feedback (desire for independence)

Psychoanalytic theory (Freud)

- Freud theorized that personality development occurred in stages through which an individual must resolve conflicts to achieve maturity in adulthood.
- During the anal phase, around 1 to 3 years, the parents and child work together to accomplish bladder and bowel continence.
- Overly controlling or authoritarian parent actions during this period could result in their child growing to be an anal-retentive adult (perfectionistic, obsessively clean, and legalistic)

- Overly liberal parent actions could result in an anal-expulsive adult (messy, disorganized, inconsiderate, and rebellious).
- Other than emphasizing the important role parents play in toilet training, psychoanalytic theory does **not directly inform its teaching methods.**

Content Category 20- Sexuality

ABP Content Specs for Developmental-Behavioral Pediatrics Fellow Collaboration Project

Prepared by: Erika Nishiguchi, Seattle Children's/UW DBP Fellow

Reviewed by Sam Zinner, MD, DBP Program Director, SCH/UW & Emily Myers, MD, Associate PD for DBP at UW/SCH

20. Sexuality

A. Stages of development

1. Know how children of various ages understand sexuality
2. Understand the typical development of sexual behaviors during childhood and adolescence
3. Know the appropriate content of school-based programs on sexuality, at different grade levels

B. Gender dysphoria in children and adolescents

1. Gender dysphoria
   a. Know the diagnostic criteria for gender dysphoria in children
   b. Know the variable manifestations of gender dysphoria in prepubertal females and males
   c. Know the diagnostic criteria for gender dysphoria in adolescents
   d. Know the variable manifestation of gender dysphoria in adolescent females and males
   e. Know the comorbidities of gender dysphoria in children and adolescents
2. Sexual deviance
   a. Recognize children whose sexual interests or activities suggest a history of sexual abuse
   b. Recognize symptoms of pathological sexual development
   c. Know approaches to management of sexual deviance in children
3. Sexual orientation
   a. Understand that sexual orientation ranges from exclusively homosexual to exclusively heterosexual
   b. Know the typical stages of development of sexual orientation in males and females
   c. Understand the difficulties resulting from the isolation and stigmatization of homosexual individuals
   d. Know the problems faced by adolescents who are or think they may be gay or lesbian
   e. Know the range of responses and needs of parents who have a homosexual child
   f. Know the typical age of self-identification of sexual orientation in the U.S.
   g. Know that gay and lesbian adolescents have increased risk of tobacco, alcohol, and other substance abuse
   h. Know the emotional and behavioral disorders for which gay and lesbian youth are at increased risk (eg, restrictive eating disorders, depression, and suicide)

C. Sexuality in developmental disorders

1. Recognize the healthy drive for sexuality among individuals with developmental disabilities
2. Know the challenges to sexuality that face individuals with developmental disorders
3. Be able to develop a plan for sexuality counseling directed to individuals with developmental disabilities

Sexuality

Definition: There is no single, specific definition of sexuality. One definition is: "…a central aspect of being human throughout life [encompassing] sex, gender identities and roles, sexual orientation, eroticism, pleasure, intimacy and reproduction. Sexuality is experienced and expressed in thoughts, fantasies, desires, beliefs, attitudes, values, behaviors, practices, roles, and relationships. While sexuality can include all of these dimensions, not all of them are always experienced or expressed. Sexuality is influenced by the interaction of biological, psychological, social, economic, political, cultural, legal, historical, religious, and spiritual factors."i

A. Stages of development

1. Know how children of various ages understand sexuality
    a. "Developing a healthy sexuality is a key developmental milestone for all children and adolescents that depends on acquiring information and forming attitudes, beliefs and values about consent, sexual orientation, gender identity, relationships, and intimacy"ii.

    b. Infant: Bonding with caretaker, touch and being touched, builds foundation for ability to receive and give physical affection later in life. As infants achieve cognitive and motor milestones, including recognizing self vs. other, stabilized core, and purposeful reaching, they begin exploring their bodies and genitalia, including self-stimulatory behaviors (masturbation) with attendant physiological changes, including tachycardia, rigidity, and sweating. Male infants have erections frequently and female infants can produce vaginal lubrication. Circumcision is an important aspect of male sexuality that occurs in the first days of life.
    c. Toddler: Learn gendered expectations, names of body parts, curious about body & differences between girls and boys, self genital stimulation for pleasure is common. Exploration involves dress-up and make-believe play.
    d. Preschool: Peak in outward expression of sexual behaviors 3-5 years. Further interest in differences between sexes, children's sexual curiosity and exploration are normal; parents should teach socially acceptable touching and boundaries (ie one's own privates are not to be touched without permission)
        i. Developmentally common behaviors: touching/rubbing one's own genitalia, attempting to touch a woman's breast, showing of one's own genitalia, looking at another child's genitalia.
        ii. Less common/red flag behaviors: putting a finger into another's vagina or anus, putting mouth to another's (or a doll's) genitalia or anus, inserting penis into vagina, asking for genitalia to be touched.
    e. School age (5/6-puberty): Establish gender identity, tend to have same-sex friends (may change to same-gender friends if there is incongruence in assigned sex and

gender identity), sexuality expressed by major interest in sexual jokes, stories, or songs but generally masturbation decreases or still occurs, just more discretely. As children enter puberty, their experience with sexuality begins to mature as their bodies mature. Girls experience menses and boys experience nocturnal emissions/ "wet dreams."

f. Adolescent: Marked by significant body changes with development of secondary sexual characteristics; increasing involvement in peer and social relationships including those of the opposite sex, values and norms are paramount. Issues of sexual identity and orientation can be a cause of anxiety and insecurity. More than 50% of adolescents report initiation of sexual activity by age 18iii.

g. For more information in table format, see Figures 1 and 2, attached.

2. **Understand the typical development of sexual behaviors during childhood and adolescence**

a. See text from #1 and tables/figures.

b. Normal childhood sexual play and exploration has the following characteristics: spontaneous, intermittent, mutual, non-coercive, does not cause emotional distress, involves peers of similar age and developmental stage.

c. It is important to consider cultural context when discussing normal sexual and sexuality development.

3. Know the appropriate content of school-based programs on sexuality, at different grade levels.

i. a. Sexuality education is defined as "teaching about human sexuality, including intimate relationships, human sexual anatomy, sexual reproduction, sexually transmitted diseases, sexual activity, sexual orientation, gender identity, abstinence, contraception, and reproductive rights and responsibilities." Sexuality education involves:Dimensions:

1. Biological
2. Sociocultural
3. Psychological
4. Relational
5. Spiritual

ii. Domains of learning:

1. Cognitive (information)
2. Affective (feelings, values, attitudes)
3. Behavioral (communication, decision-making, other skills)

b. Educators

i. Parents:

1. Infants: Parents should know typical infant natural sexual behaviors so that they do not scold, punish, or ridicule
2. Toddlers/Preschoolers: Parents should use anatomically correct terms in developmentally appropriate manner to respond to children's many questions (AAP recommendation 2001), and use teachable moments such as birth of a sibling to help children understand how babies come into the world. Parents should teach socially acceptable behavior in a nurturing, coaching manner.

3. School age: Important for parents to have ongoing conversations about sexuality and appropriate vs. inappropriate touch. Discussions about the timing and progression of puberty can help children understand the changes and lay foundation for trusting discussions to come. Discussions about sex and conception should begin when children begin asking questions, and mature as the child matures.
4. Pre-teen: Discuss intercourse, how sexual diseases are transmitted, and how pregnancy occurs and can be prevented. Children should have a clear understanding about what constitutes abuse.
5. Adolescent: Child/parent conflict about initiation of sexual activity may occur. As teens become more independent they may disclose less to their parents.

ii. Schools:

1. Sexual education programs should include functional knowledge, support for developing personal and cultural values and beliefs about healthy behaviors, help shape group norms regarding healthy behaviors, and facilitate the development of skills necessary to adopt, practice, and maintain healthy behaviors.
2. Structured Recommendations:
3. Both the Independent and Government recommendations outlined below follow the CDC National Health Education Standardsiv, a ladder of eight core "Healthy Behavior Outcomes":

    a. Independent: The Sexuality Information Education Council of the United States (SIECUS): Sex Ed for Social Changev recommends seven evidence-based essential curricular topics to be included at every age:

        i. Consent and Healthy Relationships
        ii. Anatomy and Physiology
        iii. Puberty and Adolescent Sexual Development
        iv. Gender Identity and Expression
        v. Sexual Orientation and Identity
        vi. Sexual Health
        vii. Interpersonal violence

    These seven topics are organized into eight strands recommended by the CDC's National Health Education Standards:

        i. Core concept
        ii. Analyzing influencers
        iii. Assessing information
        iv. Interpersonal communication
        v. Decision making
        vi. Goal setting

vii. Self-management

viii. Advocacy

b. Government: The CDCvi outlines the following 8 core "Healthy Behavior Outcomes":

:

i. Establish and maintain healthy relationships
ii. Be sexually abstinent
iii. Engage in behaviors that prevent or reduce sexually transmitted disease (STD), including HIV
iv. Engage in behaviors that prevent or reduce unintended pregnancy
v. Avoid pressuring others to engage in sexual behaviors
vi. Support others to avoid or reduce sexual risk behaviors
vii. Treat others with courtesy and respect without regard to their sexuality.
viii. Use appropriate health services to promote sexual health.

A curricular guide can be found at the following link: https://www.cdc.gov/healthyyouth/hecat/pdf/hecat_module_sh.pdf

iii. Health Care Providers:

1. AAP Policy Revision "Sexuality Education in Children and Adolescents, 2016)vii

2. Figure 3, attached.

B. Gender dysphoria in children and adolescents viii

Added Content Definitions. See Figure 4 for an image to help understand these terms, the "Genderbread Person".

- **Sex.** A biological construct: maleness or femaleness (e.g., chromosomes, sex-determining genes, hormones, anatomy).
- **Gender**. A social construct: attitudes, feelings, and behaviors developed within any given culture that associates with biological sex, including:
  - expression (clothing, hairstyle, name, pronouns),
  - roles and behaviors,
  - identity (personal sense of gender—can be binary or on a spectrum).
  - Definitions:

        - **Cis-gender:** Gender identity and assigned/assumed gender (based on sex) are congruent.
        - **Transgender**: Gender identity and assigned/assumed gender (based on sex) are incongruent.
    - Gender development
        - 18-24 months: Develop ability to label and assign gender.
        - 2-4 years: Recognize gender differences and use gender pronouns.
        - 5-6 years: Most children declare a gender identity of male or female.
- **Gender nonconformity**: Behaviors not matching the cultured gender norms or stereotypes of sex or assigned/assumed gender.
- **Gender dysphoria**: An individual's internal/affective/cognitive discontent experienced due to discordance between sex and gender identity. Not all individuals who have an incongruence between sex and gender will experience gender dysphoria. Added content: Dysphoria is due to societal expectations / social conditioning more than internal pathology as evidenced by improved health outcomes in children met with supportive (wait and see) or affirming approaches compared to corrective approach by family.
- **Transitioning:** Process by which an individual identifying as transgender begin living/externally expressing more permanently their gender identity. This may or may not include pubertal blockers and/or hormonal treatment.

1. Know the diagnostic criteria for gender dysphoria in children (DSM-5)

A. A marked incongruence between one's experienced/ expressed gender and assigned gender; of at least 6 months' duration, as manifested by at least six of the following (one of which must be criterion A1).

1. A strong desire to be of the other gender or an insistence that one is the other gender (or some alternative gender different from one's assigned gender.)

2. In boys (assigned gender), a strong preference for cross-dressing or simulating female attire; or in girls (assigned gender), a strong preference for wearing only typical masculine clothing and a strong resistance to the wearing of typical female clothing.

3. A strong preference for cross-gender roles in make-believe or fantasy play.

4. A strong preference for the toys, games, or activities stereotypically used or engaged in by the other gender.

5. A strong preference for playmates of the other gender.

6. In boys (assigned gender), a strong rejection of typically masculine toys, games, and activities and a strong avoidance of rough-and-tumble play; or in girls (assigned gender), a strong rejection of typically feminine toys, games, and activities.

7. A strong dislike of one's sexual anatomy.

8. A strong desire for the primary and/or secondary sex characteristics that match one's experienced gender.

B. The condition is associated with clinically significant distress or impairment in social, school, or other important areas of functioning.

Specify if : With a disorder of sex development (e.g., congenital adrenogenital disorder, congenital adrenal hyperplasia, or androgen insensitivity syndrome)

2.Know the variable manifestations of gender dysphoria in prepubertal females and males

a) Epidemiology:

a. The minority (6-27%) of prepubertal children with gender dysphoria experience persistence of dysphoria into adulthood.
b. Boys with pre-pubertal gender dysphoria are more likely to identify as gay in adulthood than as transgender
c. Male:female ratio range from 6:1 to 3:1.

b) Cross-gender behaviors may be evident between 2 and 4 years of age. Gender-atypical behaviors can be considered a normal part of development.

c) Children with more intense symptoms and distress, with more persistence of these feelings over time, more insistence with their cross-gender statements and behaviors, and declarative statements such as "I am a boy/girl" compared to "I want to be a boy/girl" are more likely to have persistent gender dysphoria through puberty and into adulthood. ixx

3.Know the diagnostic criteria for gender dysphoria in adolescents:

A. A marked incongruence between one's experienced/expressed gender and assigned gender, of at least 6 months' duration, as manifested by at least two of the following:

1. A marked incongruence between one's experienced/expressed gender and primary and/or secondary sex characteristics (or in young adolescents, the anticipated secondary sex characteristics).
2. A strong desire to be rid of one's primary and/or secondary sex characteristics because of a marked incongruence with one's experienced/expressed gender (or in young adolescents, a desire to prevent the development of the anticipated secondary sex characteristics).
3. A strong desire for the primary and/or secondary sex characteristics of the other gender.
4. A strong desire to be of the other gender (or some alternative gender different from one's assigned gender).
5. A strong desire to be treated as the other gender (or some alternative gender different from one's assigned gender).
6. A strong conviction that one has the typical feelings and reactions of the other gender (or some alternative gender different from one's assigned gender).

B. The condition is associated with clinically significant distress or impairment in social, occupational, or other important areas of functioning.

Specify if:

With a disorder of sex development (e.g., a congenital adrenogenital disorder such as ICD-9/-10 code 255.2 [E25.0] congenital adrenal hyperplasia or 259.50 [E34.50] androgen insensitivity syndrome).

Coding note: Code the disorder of sex development as well as gender dysphoria.

Specify if:

Post-transition: The individual has transitioned to full-time living in the desired gender (with or without legalization of gender change) and has undergone (or is preparing to have) at least one cross-sex medical procedure or treatment regimen—namely, regular cross-sex hormone treatment or gender reassignment surgery confirming the desired gender (e.g., penectomy, vaginoplasty in a natal male; mastectomy or phalloplasty in a natal female).

4.Know the variable manifestation of gender dysphoria in adolescent females and males

a) Epidemiology:

a. No prospective studies, but appear the majority of individuals experiencing post-pubertalgender dysphoria have this dysphoria persist into adulthood.
b. Male:female ratio 1:1

5.Know the comorbidities of gender dysphoria in children and adolescents

a) Personal Social

a. Early childhood: Isolation from peer groups
b. School age: School refusal, bullied
c. Adolescence: Relationship difficulties

b) High levels of stigmatization, discrimination, and victimization that lead to:

a. Negative self-concept
b. Depression
c. Anxiety
d. Disruptive behaviors
e. Impulse-control disorders
f. School drop-out
g. Economic marginalization
h. Run-away / homelessness

c) Co-occurring

a. Autism spectrum disorder is more prevalent in clinically referred children with gender dysphoria than in the general populationThe prevalence of mental health problems differs among cultures; these differences may also be related to differences in attitudes toward gender variance in children. However, also in some non-Western cultures, anxiety has been found to be relatively common in individuals with gender dysphoria, even in cultures with accepting attitudes toward gender-variant behavior.

C. Sexual deviance

1. Recognize children whose sexual interests or activities suggest a history of sexual abuse

a. Epidemiology:

i. By age 18, 1 in 4 girls and 1 in 6 boys will have been sexually assaulted

ii. Most offenders are known to the children

iii. Children exhibit variable reactions to the assault and often do not tell parents right away

b. Preschool Red flag behaviors:

i. putting a finger into another's vagina or anus,

ii. putting mouth to another's (or a doll's) genitalia or anus,

iii. inserting penis into vagina,

iv. asking for genitalia to be touched.

c. Older ages Red flag behaviors:

i. Excessive preoccupation with sexual words, sexual body parts, sexual activity

ii. Repeated display of sexual body parts in public

iii. Persistent, secretive sex play with other children after being told not to

iv. Putting objects into sexual body parts

v. Imitating or trying to have sexual intercourse with toys, pets, or other children

vi. Doing sexual things with much younger or much older children

vii. Pressuring or forcing others into sexual activity of any kind

2. Recognize symptoms of pathological sexual development

a. Definition: Children 12 years of age and younger who initiate behaviors involving sexual body parts (ie. genitals, anus, buttocks, or breasts) that are developmentally inappropriate or potentially harmful to themselves or others.

i. The intentions or motivations of these behaviors may or may not be related to sexual gratification or stimulation.

ii. May be related to curiosity, anxiety, imitation, attention-seeking, self-calming, or other reasons

b. Epidemiology:

i. No population-based data for incidence or prevalence

ii. There has been increase in reporting of these behaviors over time to professional organizations (child protective services, juvenile services, clinical settings)

iii. Children who have been abused have a higher frequency of developing problematic sexual behaviors

iv. Many children with problematic sexual behaviors do not have a history of abuse

c. Considerations:

i. Child's developmental stage, culture, frequency of behaviors, extent to which the behavior has become a preoccupation, whether the child responds to parental intervention/correction.

ii. Coercion, intimidation, force, physical injury

d. Risk factors:

i. Sexual abuse, neglect, "substandard parenting practices", exposure to sexually explicit media, highly sexualized environment, family violence

ii. Overall pattern of developmental or behavioral differences

iii. Genetic predisposition (eg to co-occuring psychiatric disorders that decrease impulse control or social awareness)

3. Know approaches to management of sexual deviance in childrenxi

Note: the most up-to-date information I could find was published in 2006.

a. Survivors of sexual abuse

i. Gradual, exposure-based Cognitive-Behavioral Therapy (CBT) with parent/caregiver involvement is superior to non-specific supportive therapy

ii. CBT-based group therapy has shown effectiveness in preschool children

b. Problematic sexual behavior / deviance

i. Cognitive-behavioral based approaches have been shown to reduce occurrence of sex offense arrests and reports from child welfare sexual offense perpetration in adulthood

ii. Relapse-prevention training adapted from adult programs may be useful but is no better at ensuring lasting effects than CBT treatment without a focus on identifying relapse risk factors

c. Overall

i. Generally, improvement does occur over time, as evidenced by wait-list arms in studies

ii. Treatment is beneficial in both less aggressive and highly aggressive behaviors

iii. Benefits are reported among populations with significant trauma, co-morbid disorders, and varying level of family function.

iv. Parents and Caregivers should be involved in treatment for maximal effect

v. Considerations such as Group vs. Individual therapy and sexual behavior (details below) vs. trauma-focused CBT should be individualized and evidence-based

vi. Resources for selecting empirically supported intervention models are available from a number of registries including the Substance Abuse and Mental Health Services Administration (www.modelprograms.samhsa.gov), the American Psychological Association's Division 53 on Child and Adolescent Clinical Psychology (http://www.wjh.harvard.edu/%7Enock/ Div53/EST/index.htm), the Cochrane Collaborative (www.cochrane.org), the Crime Victims Research and Treatment Center (http://www.musc.edu/cvc/guide1.htm) or other repositories.

d. Components of Problematic Sexual Behavior CBT

i. Child

1. Identifying, recognizing, and apologizing for rule-violating sexual behaviors

2. Learning and practicing rules about sexual behaviors and physical boundaries.

3. Age-appropriate sex education

4. Coping and self-control strategies

5. Sexual abuse prevention skills

6. Social Skills

ii. Parent/Caregiver

1. Developing and implementing a safety plan

2. Information about sexual development, normal sexual play and exploration, and how these differ from problematic behaviors

3. Strategies to encourage children to follow basic privacy and behavior rules

4. Factors that contribute to the development and maintenance of problem behaviors

5. Sex education and how to listen and talk with children about sexual matters

6. Parenting strategies to build positive relationships with children and address behavior problems

7. Supporting children's use of self-support strategies that they have learned

8. Relationship building and appropriate physical affection with children

9. How to guide the child toward positive peer groups

D. Sexual orientation

1. Understand that sexual orientation ranges from exclusively homosexual to exclusively heterosexual

a. Traditionally, sexual orientation has been defined as attraction to the opposite sex (heterosexual), attraction to the same sex (homosexual), and attraction to both sexes (bisexual).

b. Increasingly, sexual orientation is recognized as more continuous on a spectrum than neatly categorical.

2. Know the typical stages of development of sexual orientation in males and femalesxii

a. Research in recent decades has explored the developmental nature of sexual orientation and attempted to define milestones according to individuals' reported experiences and recognizing the continuous/spectrum experience of sexual orientation.

b. Gender differences and types of sexual minority (eg gay/lesbian vs. bisexual vs. other identities) may affect timing and order of sexual orientation developmental milestones.

c. Sexual orientation developmental milestones:

i. Same-gender attraction

ii. Same-gender sexual experience

iii. Sexual minority identity

iv. Other oft-experienced milestones:

1. Other-gender attraction

2. Other-gender sexual experience

d. In the “Growing Up Today Study (GUTS),” males reached sexual minority milestones earlier than females.

3. Understand the difficulties resulting from the isolation and stigmatization of homosexual individuals

a. Life course theory examines sexual orientation/identity development within the dominant culture in which an individual is living. In the US, the dominant culture is heteronormative. Individuals undergoing sexual minority identity development in the US experience stigma that may affect their process and timing.

b. Large population based studies:

i. USxiii

1. Homosexual women have higher prevalence of obesity, stroke, and functional limitation than heterosexual women.

2. Homosexual men have higher prevalence of hypertension and heart disease than heterosexual men, and men of any sexual minority have more functional limitation than heterosexual men.

3. Bisexual women have higher prevalence of injury/poisoning and feeling depressed than other sexual minority groups sand heterosexual individuals.

4. There are differences in healthcare utilization between heterosexual individuals and sexual minority individuals, with sexual minorities having similar access but delayed seeking of care.

5. Sexual minority individuals report higher rates of smoking and alcohol use than their heterosexual peers.

ii. UKxiv

1. Hetereosexual individuals report the best general health compared to sexual minorities, and bisexual individuals report the worst overall.

2. All sexual minority groups have worse mental health functioning and minor psychological distress than heterosexual individuals.

3. Bisexual, “other,” and “prefer not to say” individuals have worse physical functioning than heterosexual individuals.

4. Gay/lesbian and bisexual individuals are more likely to report a chronic health conditioning affecting function.

5. Bisexual individuals are more likely than heterosexual individuals to report having one or more disabilities.

4. Know the problems faced by adolescents who are or think they may be gay or lesbian

a. Societal stigma, manifesting in difficulty with own sexual minority identity development, and fears about family and peer acceptance.

b. The Growing Up Today Study found the following factors were associated with more stress related to sexual minority identity:

i. Reaching sexual minority milestones earlier in adolescence

ii. Sexual orientation mobility, or change in sexual orientation over time

iii. Bullied

5. Know the range of responses and needs of parents who have a homosexual child

a. Typical responses include:

i. Corrective: view of sexual minority status as a psychopathology

ii. Supportive: “wait and see” approach

iii. Affirming: supportive of sexual minority status

b. Family acceptance is associated with increased self-esteem, social support and overall health, and protection against depression, substance use, and suicidal ideation.

c. Family rejection is associated with much higher rates of depression, attempted suicide, substance use, and unprotected sexual intercourse.

6. Know the typical age of self-identification of sexual orientation in the U.S.

a. Average age of attainment of sexual orientation developmental milestones (age attainment endorsed by GUTS study; Figure 6):

i. Same-gender attraction: Males age 14.9; Females 16.8

ii. Same-gender sexual experience: Males 15.2; Females 18.1

iii. Sexual minority identity: Males 16.4; Females 17.5

iv. Other oft-experienced milestones:

1. Other-gender attraction: Males 9.6; Females 9.8

2. Other-gender sexual experience: Males 16.6; Females 16.2

7. Know that gay and lesbian adolescents have increased risk of tobacco, alcohol, and other substance abuse

a. See #8, below.

8. Know the emotional and behavioral disorders for which gay and lesbian youth are at increased risk (eg, restrictive eating disorders, depression, and suicide)

a. “Minority Stress Model” (Ilan Meyer, 2003) has been used by many researchers to conceptualize the compounding mental and physical health effects of sexual minority status (Fig 5).

b. The Institute of Medicine:

i. Report released in 2011 calling for increased attention to sexual minority health using the life-course framework, minority stress model, intersectionality of an individual’s multiple cultural and social identities, and social ecology perspective.xv

ii. IOM health status findings of sexual minority youth:

1. Increased risk of suicidal ideation and attempts

2. Increased rates of smoking, alcohol consumption, and substance use

3. Disproportionate representation of sexual minority youth in the homeless youth population

4. Elevated experienced of violence, victimization and harassment

5. HIV disproportionately affects young men who have sex with men, particularly young black men

c. CDC health status findings of sexual minority youth:xvi

i. Bullied at school

ii. Seriously consider suicide

iii. Feel sad or hopeless

iv. Use illicit drugs

v. Misuse prescription drugs

vi. Forced into sexual encounters

9. Added content: Protective factors and what health professionals can do.

a. Acceptance and support from families, peers, and health professionals.

b. Promote positive youth development by identifying and strengthening protective factors

c. Advocate for inclusive sexual education.

d. Support gay-straight alliances at schools.

e. Promote family acceptance when discussing with parents.

C. Sexuality in developmental disorders

1. Recognize the healthy drive for sexuality among individuals with developmental disabilities

a. Many adolescents with disabilities express desire and hope for marriage, children and typical adult sex lives.

b. Adolescents with physical disabilities are as sexually experienced as their peers.

c. People without disabilities are more willing to accept people with disabilities as fellow employees or casual friends and less willing to accept them as dating, sexual, or marriage partners.

d. Societal and psychosocial barriers may be more of a hindrance to an adolescent's sexual development than the limitations of the disability itself.

2. Know the challenges to sexuality that face individuals with developmental disorder

a. History: The Eugenic movement in the US and Germany in the early 20th Century forced thousands of persons with disabilities to undergo involuntary sterilization. Society has long not perceived individuals with disabilities as sexual beings, asexual, or hypersexual.

b. Parents/caregivers tend to under-estimate the sexual interest and activity of their child with Intellectual Disability.

c. Parents often express hesitation about discussing sexual health, fearing that they encourage inappropriate sexual behavior, or that their child does not have the coping skills to deal with sexuality.

d. Conversations about only the negative aspects of sex can lead to negative feelings about sexuality and internalized negative self-concept or inability to accept pleasure.

e. Individuals with intellectual disabilities are at much greater risk of sexual abuse (2.2x increased risk)

i. The US Department of Justice reports that 68% to 83% of women with developmental disabilities will be sexually assaulted in their lifetimes and less than half of them will seek assistance from legal or treatment services.

f. Adolescents with myelomeningocele and spinal cord injury have unique educational and medical needs that must be addressed to enjoy safe and satisfying sexual lives.

3. Be able to develop a plan for sexuality counseling directed to individuals with developmental disabilities

a. Excellent list of compiled resources on this topic:
http://www.med.umich.edu/yourchild/topics/disabsex.htm

i. Policy statements

ii. Parent/caregiver factsheets

iii. Recommended reading for parents/caregivers

b. Parent and educator resources for sexual education for children with disabilities:
https://www.parentcenterhub.org/sexed/

c. Suggested modifications:

i. Simplify information,

ii. Teach in a special needs rather than a general education setting,

iii. Special teaching materials such as anatomically correct dolls, role playing, and frequently reviewing and reinforcing the material may be required.

d. Individualized education programs (IEPs) should include the provision of sexuality education for children with disabilities. An appropriate program for children with disabilities includes the following topics:

i. body parts,

ii. pubertal changes,

iii. personal care and hygiene,

iv. medical examinations,

v. social skills,

vi. sexual expression,

vii. contraception strategies,

viii. rights and responsibilities of sexual behavior.

4. Added content: Considerations for sexual and menstrual care.

a. Puberty progresses at the biological/chronological age and work-up should be indicated for delayed menstruation as per typically developing girls.

b. Menstrual symptoms

i. Menstrual related pain (cramps, migraines) should be first managed with pain control (NSAIDs).

ii. Cyclical behavioral effects may be related to hormones or pain.

iii. There are now many menstrual underwear products that may assist with hygiene concerns (e.g., Thinx underwear)

c. Long Acting Reversible Contraception (LARC) are excellent tools for many families to ensure predictable menstrual cycles with light flow.

i. Hormonal IUDs, although invasive, result in more predictable menstrual bleeding than hormonal implants (e.g., Nexplanon).

ii. Adolescents who are already at risk of osteopenia from chronic medical conditions may be at even greater risk of bone mineral density loss from depot medroxyprogesterone acetate use.

iii. Be alert for pharmacological interactions when providing contraception. For example, some anti-epileptic medications reduce hormonal based contraceptives, including implanted devices.

d. Pelvic exams are rarely indicated if a child or adolescent is not sexually active. Provide appropriate level of respect, guidance, and reassurance if a pelvic exam is indicated.

e. Historically, sterilization of minors with developmental disabilities was performed without appropriate regard for their decision-making capacities, abilities to care for children, feelings, or interests. Such decisions should be made only in the context of the individual's capacity to make decisions, the consequences of reproduction for the person and any children that might be born, and applicable local, state, and federal laws.

f. Boxes, in attachment, detail the following considerationsxvii:

i. Box 1: Components of sexuality education program for youth with disabilities.

ii. Box 2: Tips for managing menstruation in girls with intellectual disabilities.

iii. Box 3: Supporting healthy sexual development in adolescents with developmental disabilities; the role of the medical provider.

iv. Box 4: Supporting healthy sexual development in adolescents with developmental disabilities; anticipatory guidance for caregivers.

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Figures

1. Common Sexual Behaviors of Children and Adolescents
2. Common Sexual Behaviors in Childhood
3. Clinical Guidance for Pediatricians
4. The Genderbread Person (Sexual Orientation and Identity Terminology)
5. The Minority Stress Model
6. Endorsement and Age Reached Sexual Orientation Developmental Milestones
7. Health Disparities Among LGBTQ Youth

Box 1: Components of sexuality education program for youth with disabilities.

Box 2: Tips for managing menstruation in girls with intellectual disabilities.

Box 3: Supporting healthy sexual development in adolescents with developmental disabilities; the role of the medical provider.

Box 4: Supporting healthy sexual development in adolescents with developmental disabilities; anticipatory guidance for caregivers.

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Figure 1: Common Sexual Behaviors of Children and Adolescents[1]

Age	Sexual Behaviors
Infancy (birth through 1 year)	Exploration of genital area during diaper changes Erections in boys Masturbation
Toddlers and preschoolers (ages 1 to 4 years)	Masturbation Touching mother's breasts or other body parts Taking clothes off Showing genitals to other children or adults
School-age to early adolescence (ages 5 to 11 years)	Masturbation (public displays decrease) Sex play between peers, such as playing "house" or "doctor," in which children explore sexuality through looking at and touching of genitals Asking questions and talking about sex
Adolescence	Dressing up as the opposite sex as dramatic play Continued masturbation, increasingly private Exploration of sexual intimacy with the same or opposite sex Awareness and questioning of sexual orientation Initiation of sexual intercourse

Figure 2: Common Sexual Behaviors in Childhood[2]

Preschool children (less than 4 years)	■ Exploring and touching private parts, in public and in private ■ Rubbing private parts (with hand or against objects) ■ Showing private parts to others ■ Trying to touch mother's or other women's breasts ■ Removing clothes and wanting to be naked ■ Attempting to see other people when they are naked or undressing (such as in the bathroom) ■ Asking questions about their own—and others'—bodies and bodily functions ■ Talking to children their own age about bodily functions such as "poop" and "pee"
Young Children (approximately 4-6 years)	■ Purposefully touching private parts (masturbation), occasionally in the presence of others ■ Attempting to see other people when they are naked or undressing ■ Mimicking dating behavior (such as kissing, or holding hands) ■ Talking about private parts and using "naughty" words, even when they don't understand the meaning ■ Exploring private parts with children their own age (such as "playing doctor", "I'll show you mine if you show me yours," etc.)
School-Aged Children (approximately 7-12 years)	■ Purposefully touching private parts (masturbation), usually in private ■ Playing games with children their own age that involve sexual behavior (such as "truth or dare", "playing family," or "boyfriend/girlfriend") ■ Attempting to see other people naked or undressing ■ Looking at pictures of naked or partially naked people ■ Viewing/listening to sexual content in media (television, movies, games, the Internet, music, etc.) ■ Wanting more privacy (for example, not wanting to undress in front of other people) and being reluctant to talk to adults about sexual issues ■ Beginnings of sexual attraction to/interest in peers

[1] From Chapter 45 "Sexuality" in Developmental and Behavioral Pediatrics Textbook

[2] http://ncsby.org/sites/default/files/resources/Sexual%20Development%20and%20Behavior%20in%20Children%20--%20NCTSN%20NCSBY.pdf

Figure 3: Clinical Guidance for Pediatricians[3]

1. The pediatrician should encourage early parental discussion with children at home about sexuality, contraception, and Internet and social media use that is consistent with the child's and family's attitudes, values, beliefs, and circumstances.

2. Diverse family circumstances, such as families with same-sex parents or children who identify as lesbian, gay, bisexual, transgender, or questioning, create unique guidance needs regarding sexuality education.

3. Modeling ways to initiate talks about sexuality with children at pertinent opportunities, such as the birth of a sibling can encourage parents to answer children's questions fully and accurately.

4. Parents and adolescents are encouraged to receive information from multiple sources, including health care providers and sexuality educators, about circumstances that are associated with earlier sexual activity. Adolescents are encouraged to feel empowered through discussing strategies that allow for practicing social skills, assertiveness, control, and rejection of unwanted sexual advances and cessation of sexual activity when the partner does not consent.

5. Discussions regarding healthy relationships and intimate partner violence can be effectively included in health care visits.

6. Pediatricians are encouraged to acknowledge that sexual activity may be pleasurable but also must be engaged in responsibly.

7. Specific components of sexuality education offered in schools, religious institutions, parent organizations, and other community agencies vary based on many factors. The pediatrician can serve as a resource to each.

8. School-based comprehensive sexuality education that emphasizes prevention of unintended pregnancy and STIs should be encouraged.

9. The discussion of methods of contraception and STI and HPV cancer prevention with male and female adolescents is encouraged before the onset of sexual intercourse (see the AAP statement "Contraception and Adolescents"). It is also important to discuss consistent use of safer sex precautions with sexually active teens. Bright Futures recommendations can be used.

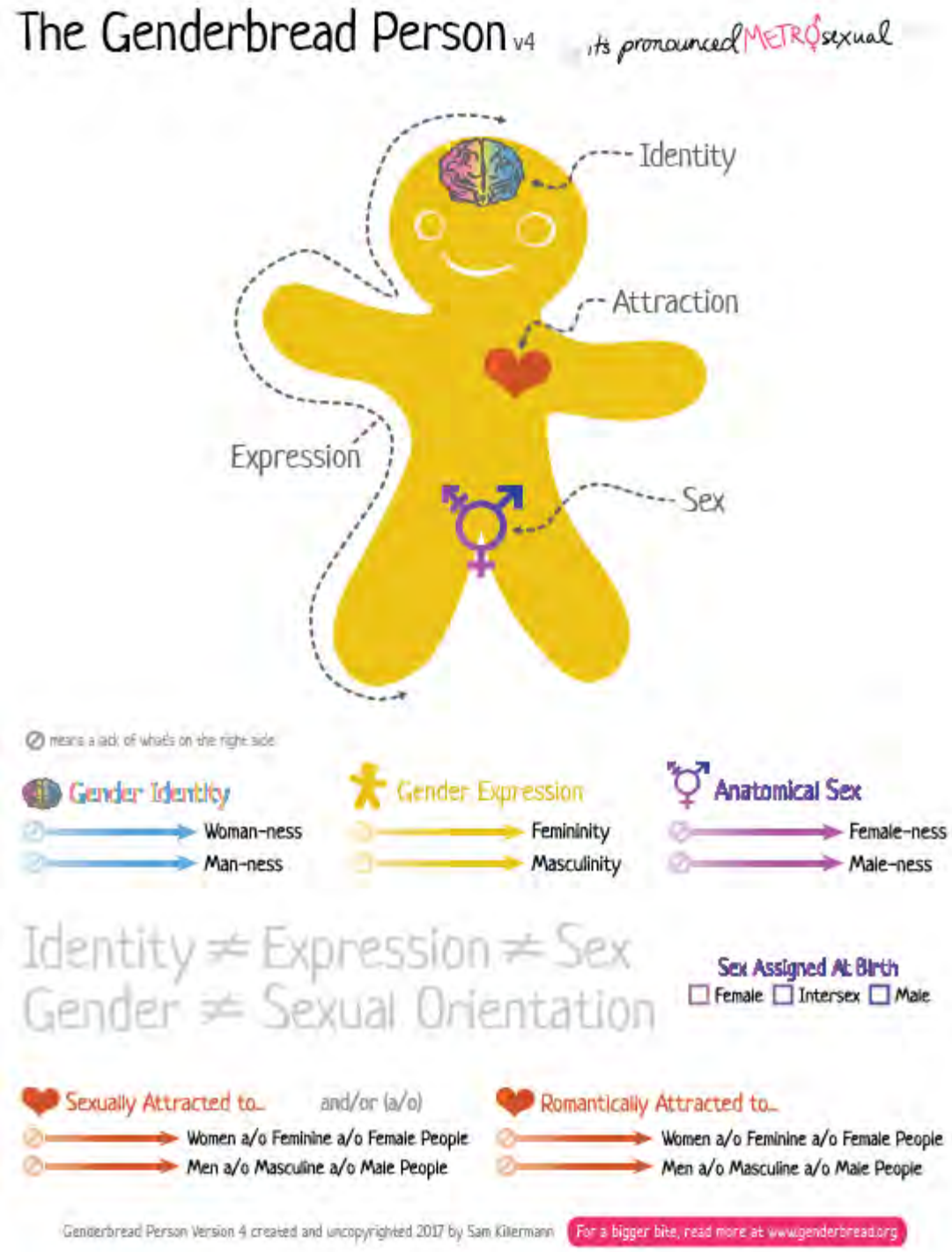
10. Abstinence is the most effective strategy for preventing HIV infection and other STIs, as well as for prevention of pregnancy.

11. Preparation for college entry is an excellent opportunity for pediatricians to address issues such as the effects of alcohol, marijuana, and other drug consumption on decisions about safe, consensual sexual practices.

12. Children and adolescents with special issues and disabilities may benefit from additional counseling, referrals, and sharing of online resources listed at the end of this report.

[3] https://pediatrics.aappublications.org/content/138/2/e20161348

Figure 4: The Genderbread Person (Sexual Orientation and Identity Terminology)[4]



[4] https://www.genderbread.org/

Figure 5: The Minority Stress Model[5]

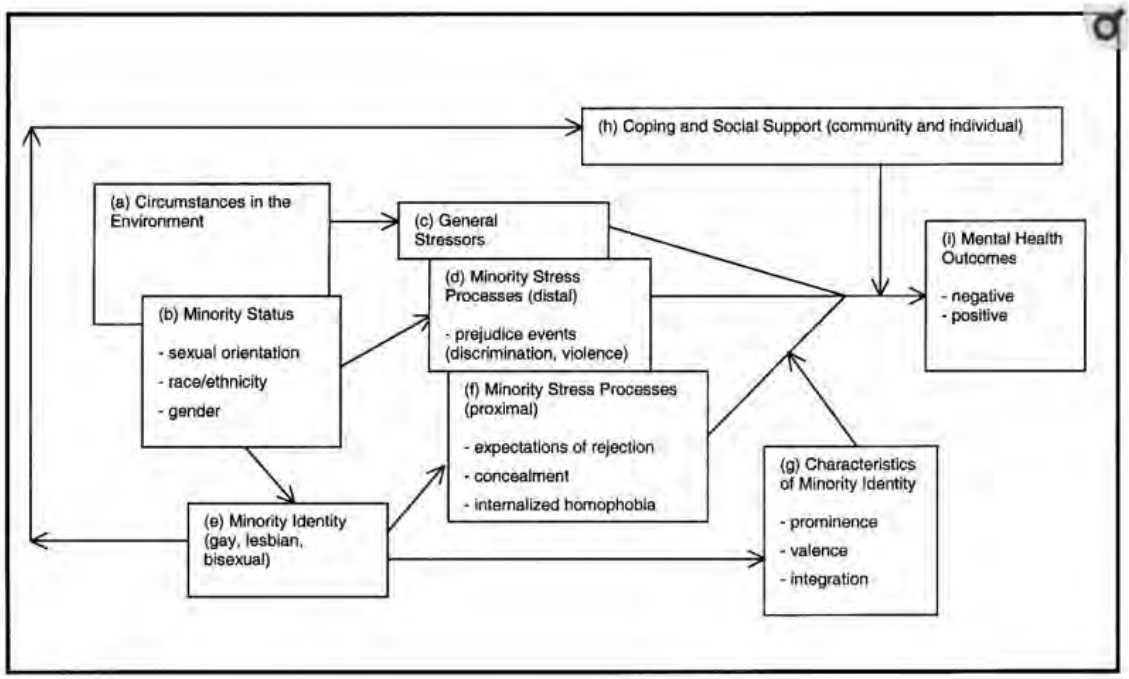


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Figure 6: Endorsement and Age Reached Sexual Orientation Developmental Milestones[6]

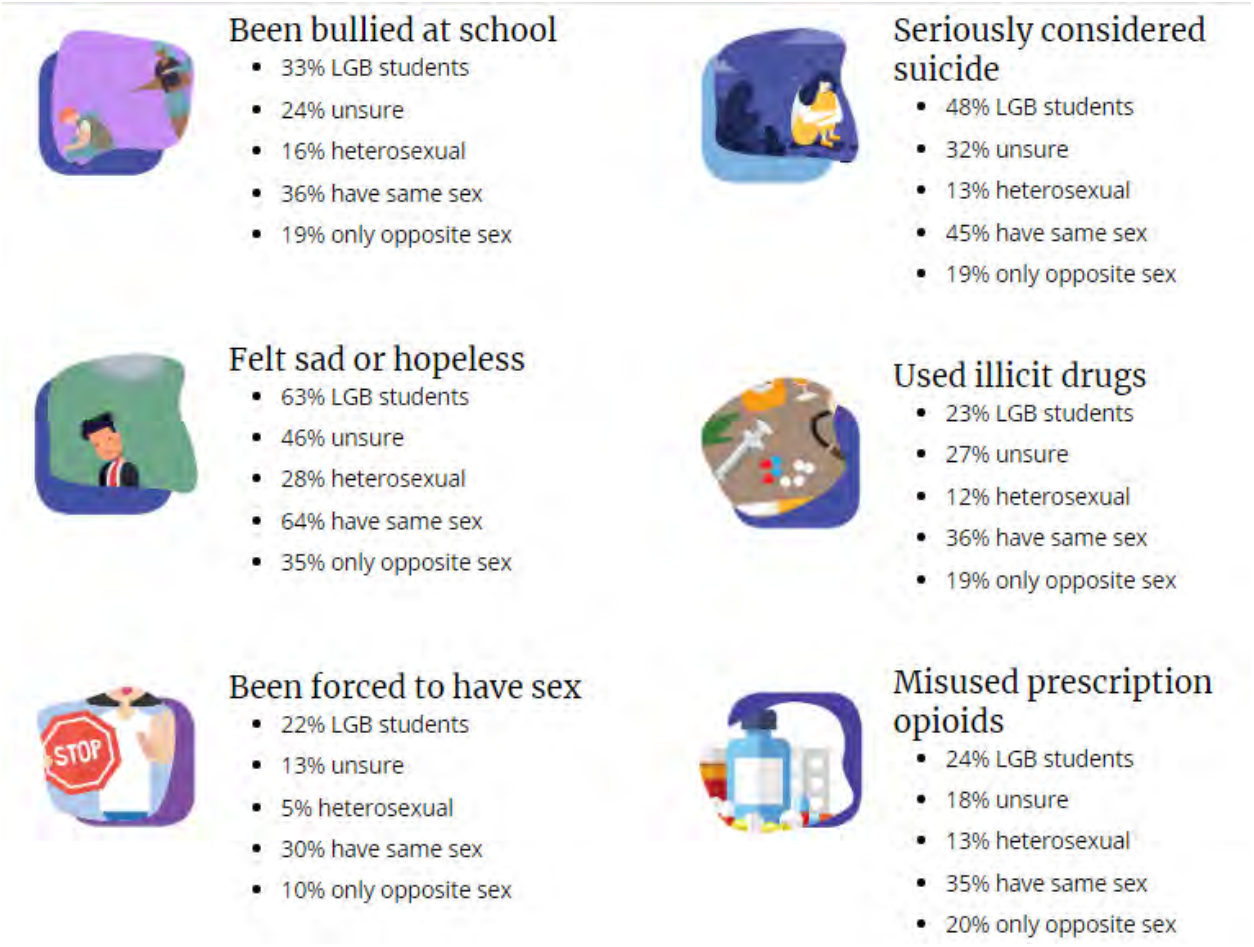
Endorsement and Age Reached Sexual Orientation Developmental Milestones by Gender for Female (N=1235) and Male (N=398) Young Adults, Ages 22 to 29 Years, in the Growing Up Today Study who Reported Any Same-Gender Orientation in 2010

	Endorsement of Milestones (%, *n*)			
Milestones	**Females**	**Males**	**Females vs. Males (*p*-value)**	**Females vs. Males (Cramer's *v*)**
Same-gender Attraction	80.8 (995)	62.3 (246)	<.001	0.19
Other-gender attraction	97.9 (1201)	85.8 (339)	<.001	0.24
Same-gender sexual experience	61.5 (759)	64.9 (257)	0.23	0.03
Other-gender sexual experience	99.2 (1182)	84.4 (335)	<.001	0.31
Sexual minority identity	63.2 (777)	60.4 (239)	0.31	0.03
	Age Reached Milestones (age in years, *M, SD*)			
Milestones	Females	Males	Females vs. Males (*p*-value)	Females vs. Males (Cohen's *d*)
Same-gender attraction	16.8 (4.0)	14.9 (5.0)	<.001	0.42
Other-gender attraction	9.8 (3.4)	9.6 (3.4)	0.24	0.06
Same-gender sexual experience	18.1 (4.2)	15.2 (5.5)	<.001	0.59
Other-gender sexual experience	16.2 (2.7)	16.6 (3.4)	0.03	−0.13
Sexual minority identity	17.5 (3.9)	16.5 (4.3)	0.001	0.24

Note. Timing of sexual orientation developmental milestones is measured by age in years. Age range for same-gender attraction: 3–27 years for females, 0–27 years for males. Age range for other gender attraction: 0–26 years for females, 1–24 years for males. Age range for same-gender sexual experience: 2–27 years for females, 3–26 years for males. Age range for other gender sexual experience: 1–28 years for females, 5–26 years for males. Age range for sexual minority identity: 0–28 years for females, 4–27 years for males. Reported *p*-values are from chi-square tests for endorsement and t-tests for timing.

[6] https://www.ncbi.nlm.nih.gov/pubmed/27148762 Growing up Today Study

Figure 7: Health Disparities Among LGBTQ Youth[7]



[7] https://www.cdc.gov/healthyyouth/disparities/health-disparities-among-lgbtq-youth.htm

Boxes 1-4[8]:

Box 1
Components of a sexuality education program for youth with disabilities

- Content
  - Simple but accurate terms for anatomy
  - Physical boundaries
  - Negotiating sexual situations
  - Understanding and avoidance of sexual abuse and exploitation
  - Same-sex and opposite-sex attraction
  - Healthy sexual interactions (intercourse and noncoital alternatives)
  - Assertiveness training (saying "No")
  - Safer sexual practices
  - Pregnancy prevention
- Educational approach
  - Strength-based versus deficit-based approach
  - Simple, explicit, concrete language
  - Use of pictures, anatomically correct dolls
  - Frequent repetition
  - Adaptability for developmental levels and health literacy
  - Practice and role playing
  - Incorporate experiential learning
  - Avoidance of heteronormative approach

Box 2
Tips for managing menstruation in girls with intellectual disabilities

- Avoid negative references to menstruation; discuss menstruation as a normal part of growing up.
- Discuss and practice pad use *prior to* menstruation.
- Use an alarm or reminder system to signal time for a pad change.
- Create a checklist for steps involved in changing pads (eg, remove, wrap up, and discard used pad; wash hands, and so forth).
- Consider the use of a sticker chart or reward system for good hygiene practices.
- For patients using a diaper, a pad placed within the diaper may facilitate easier changes and save money.
- Consider allowing patient to observe a relative or close female friend managing menstrual hygiene, if comfortable.
- Track periods on a calendar to anticipate next menses.

[8] Holland-Hall and Quint. 2016. Sexuality and Disability in Adolescents.

Box 3
Supporting healthy sexual development in adolescents with developmental disabilities: the role of the medical provider

- Emphasize sexual development as a normative experience
  - Discuss both positive and negative aspects
  - Approach as you would with all patients at a similar developmental level
- Include external genital examinations in routine physicals
  - Demonstrate respect for privacy and dignity
  - Use as "teachable moment" for appropriate and inappropriate touch
  - Set stage for reproductive care in adulthood
- Discuss behavioral and hormonal approaches to managing menstruation
- For sexually active patients
  - Provide or refer for contraception
  - Perform STI testing using urine or vaginal swab
  - Refer for genetic counseling if indicated
  - Folic acid supplementation for adolescent girl (4 mg daily for adolescents with myelomeningocele; 0.4–1 mg daily for others)
- Screen for sexual abuse and consider in patients presenting with behavior changes or genitourinary/bowel symptoms
- Vaccinate against HPV
- Screen for depression, anxiety, and substance use
- Provide anticipatory guidance on healthy sexual development throughout childhood and adolescence (see Box 4)

Box 4
Supporting healthy sexual development in adolescents with developmental disabilities: anticipatory guidance for caregivers

- Begin in early childhood
  - Role model appropriate degrees of modesty, privacy
  - "Public" versus "private" behaviors
  - Role model healthy sexual expression
- Start conversations early and repeat often
  - Approach sequentially (eg, body parts → boundaries → puberty → sexual behaviors) at the pace that is right for your child
  - Speak frankly and concretely
  - Ask about questions or concerns
- Respond calmly to questions about sex
  - Do not demonstrate anger or shock
  - Use as an opportunity to explore the adolescent's thoughts and experiences
  - "What makes you ask that question?"
  - "Where did you learn that word?"
- Use teachable moments (real life and media) to facilitate discussion and reinforce appropriate behavior
- Prepare female children for menstruation
  - Discuss ahead of time
  - Distinguish menstrual blood from bleeding caused by injury
  - Model menstrual hygiene if comfortable doing so
  - Consider use of reminders, sticker charts, rewards to support menstrual hygiene practices
- Acknowledge that many with intellectual disability ultimately have the desire and decisional capacity to choose to engage in sexual activity
- Provide opportunities for healthy sexual development while limiting risk of harm
  - Promote normal teen activities and interactions
  - Respect need for privacy
  - Teach appropriate setting and context for masturbation
  - Provide experiential learning opportunities
- Monitor use of social media and place limits as needed
- Include sex education and social skills training in Individualized Education Program

Content Category 21- Atypical Behaviors

ABP Content Specs for Developmental-Behavioral Pediatrics Fellow Collaboration Project

Prepared by: Jonathan Chooey, DO, Madigan Army Medical Center DBP Fellow & Preeya Desai, MD, UCLA DBP Fellow

Reviewed by Irene Koolwijk, MD, UCLA, DBP Fellowship Director

21. Atypical Behaviors

A. Repetitive behaviors and habits
   1. Differentiate between normal variations in repetitive behaviors and stereotypic movement disorder
   2. Know the potential medical complications of repetitive behaviors such as nail-biting, nose-picking, and hair-pulling
   3. Know how to evaluate a child with repetitive behaviors such as nail-biting, nose-picking, and hair-pulling
   4. Know how to plan the treatment for a child with repetitive behaviors such as nail-biting, nose-picking, and hair-pulling
   5. Know the theories related to the etiology of repetitive behaviors

B. Tics
   1. Differentiate tics from voluntary and other involuntary movements, such as chorea, athetosis, and ballismus
   2. Know the epidemiology of tics and tic disorders
   3. Know the diagnostic criteria for tic disorders, including provisional tic disorder, persistant motor or vocal tic disorder, and Tourette disorder
   4. Describe the natural history of tics and tic disorders
   5. Know the conditions commonly associated with Tourette disorder (eg, ADHD and OCD)
   6. Understand the pathophysiology of Tourette disorder
   7. Plan the evaluation of a child with a tic disorder
   8. Plan the treatment for a child with a tic disorder
   9. Know the pharmacologic interventions that can be helpful in management of tics
   10. Recognize the behavioral and developmental complications of Tourette disorder
   11. Understand the genetics of Tourette disorder

C. Self-injurious behaviors without developmental disabilities
   1. Know the epidemiology of self-injurious behavior in children without developmental disabilities
   2. Understand the natural history of self-injurious behavior in children without developmental disabilities
   3. Recognize the signs of self-injurious behaviors that occur in adolescents without developmental disabilities
   4. Distinguish between self-injurious behaviors that are suicidal in intent, and those that are not
   5. Know how to evaluate an adolescent without disabilities who engages in self-injurious behaviors
   6. Know how to plan the treatment of an adolescent without disabilities who engages in non-suicidal self-injurious behaviors
   7. Know the potential causes and associated conditions of self injurious behavior in children without developmental disabilities
   8. Know the conditions commonly associated with self-injurious behavior in children without developmental disabilities

D. Repetitive/disruptive/self-injurious behavior in dev. disabilities
    1. Know the epidemiology of self-injurious behavior among children with developmental disabilities
    2. Recognize the specific genetic disorders that are associated with an increased risk of self-injurious behavior
    3. Describe the complications that may result from self-injurious behavior in children with developmental disabilities
    4. Understand the etiologies of self-injurious, repetitive, and disruptive behavior among children with developmental disabilities
    5. Understand the role of functional behavioral analysis in the evaluation of self-injurious, repetitive, and disruptive behaviors in children with developmental disabilities
    6. Know how to plan the management for patients with developmental disabilities who engage in self-injurious, repetitive, or disruptive behavior
    7. Know the pharmacologic treatment of self-injurious behaviors
    8. Recognize the spectrum of repetitive behaviors that may be seen in children with autism spectrum disorder
    9. Know that some self-injurious, disruptive, or repetitive behaviors may be the result of an unrecognized medical disorder in patients with developmental disabilities
    10. Know how to evaluate a child with a developmental disability who engages in repetitive, disruptive, or self-injurious behaviors
    11. Know the pharmacologic management of repetitive behaviors in individuals with developmental disabilities
    12. Know the pharmacologic management of disruptive behaviors in individuals with developmental disabilities
    13. Know the behavioral treatments that may be used for self-injurious, repetitive, or disruptive behaviors in children with developmental disabilities
E. Alterations in mental status
    1. Recognize the signs and symptoms of delirium
    2. Recognize the signs and symptoms of dementia
    3. Understand the heightened risk for delirium or dementia in some disorders causing developmental disabilities
    4. Know the differential diagnosis for delirium and dementia
    5. Plan the evaluation of a child with delirium or dementia

21. **Atypical Behaviors**

A. Repetitive behaviors and habits

1. Differentiate between normal variations in repetitive behaviors and stereotypic movement disorder

   Repetitive behaviors are part of normal child development. Body rocking, head banging, or digit-sucking occur in most infants during the first year of life. Usually these behaviors decrease in frequency during the toddler and preschool years. When these repetitive, seemingly driven, and nonfunctional motor behaviors impair function (i.e. interfering normal activities or result in bodily injury), they can be classified as a stereotypic movement disorder.

   Diagnostic and Statistical Manual of Mental Disorders: DSM-5. Washington, DC; American Psychiatric Publishing; 2013.

   Voigt RG, Macias MM, Myers SM, Tapia CD. *Developmental and Behavioral Pediatrics.* Itasca, IL: American Academy of Pediatrics; 2018.

2. Know the potential medical complications of repetitive behaviors such as nail-biting, nose-picking, and hair-pulling

   Nail-biting may lead to paronychia or other infections.
   Nose-picking may lead to epistaxis.
   Hair-pulling may lead to alopecia.

3. Know how to evaluate a child with repetitive behaviors such as nail-biting, nose-picking, and hair-pulling

   DSM-5 Diagnostic Criteria: Stereotypic movement disorder

   A. Repetitive, seemingly driven, and apparently purposeless motor behavior (e.g., hand shaking or waving, body rocking, head banging, self-biting, hitting own body).
   B. The repetitive motor behavior interferes with social, academic, or other activities and may result in self-injury.
   C. Onset is the early developmental period.
   D. The repetitive motor behavior is not attributable to the physiological effects of a substance or neurological condition and is not better explained by another neurodevelopmental or mental disorder (e.g., trichotillomania, obsessive-compulsive disorder).

   Specify if:
   With self-injurious behavior (or behavior that would result in an injury if preventative measures were not used)
   Without self-injurious behavior

   Specify if:
   Associated with a known medical or genetic condition, neurodevelopmental disorder, or environmental factor (e.g., Lesch-Nyhan syndrome, intellectual disability, intrauterine alcohol exposure)
   Coding note: use additional code to identify the associated medical or genetic condition, or neurodevelopmental disorder

   Specify current severity:
   Mild: symptoms are easily suppressed by sensory stimulus or distraction.
   Moderate: symptoms require explicit protective measures and behavioral modification.
   Severe: continuous monitoring and protective measures are required to prevent injury.

Diagnostic and Statistical Manual of Mental Disorders: DSM-5. Washington, DC; American Psychiatric Publishing; 2013.

DSM-5 Diagnostic Criteria: Trichotillomania (Hair pulling disorder)

A. Recurrent pulling out of one's hair, resulting in hair loss.
B. Repeated attempts to decrease or stop hair pulling.
C. The hair pulling causes clinically significant distress or impairment in social, occupational, or other important areas of functioning.
D. The hair pulling or hair loss is not attributable to another medical condition (e.g., a dermatological condition)
E. The hair pulling is not better explained by the symptoms of another mental disorder (e.g., attempts improve a perceived defect or flaw in appearance in body dysmorphic disorder).

Diagnostic and Statistical Manual of Mental Disorders: DSM-5. Washington, DC; American Psychiatric Publishing; 2013.

4. Know how to plan the treatment for a child with repetitive behaviors such as nail-biting, nose-picking, and hair-pulling

When the repetitive behavior (habit) causes the child or family substantial distress, social isolation, or physical injury, a therapeutic intervention may be required. If the physical examination reveals bodily damage from a habit behavior, focus on treating the specific injury and reducing or eliminating the immediate physical harm the child may be inflicting on themselves. Dental splints may be considered for bruxism, helmets for severe and persistent head banging, bitter nail polish and shortening of nails for nail biting. Cognitive behavioral therapy components to reduce anxiety. Medication management is only considered after behavioral interventions have been tried. Some medication considerations include Naltrexone, Clomipramine (TCA), and SSRIs.

Medscape article: Childhood Habit Behaviors and Stereotypic Movement Disorder.

5. Know the theories related to the etiology of repetitive behaviors

Psychogenic hypotheses:

- In response to a lack of external stimuli the stereotypic movements help animals maintain a state of arousal. In support of this view, stereotypies are more common in children with sensory deprivation due to blindness or deafness and conditions where there is less interaction with the external environment (i.e. Autism).
- Stereotyped movements are a method of expending excess energy or attention. Attending to the movements may help to diminish other unwanted or unpleasant stimuli, as form of negative reinforcement. From a related perspective, sensory stimulation in various stereotypies is proposed as automatic positive reinforcement that causes the actions to persist.

Neurobiologic hypotheses:

- Anatomically, the basal ganglia is implicated in stereotypic disorders. There are case reports of stereotyped movements emerging after lesions of the putamen, orbitofrontal cortex, or thalamus. A volumetric MRI study in children with complex motor stereotypies demonstrated a reduction in the size of the caudate nuclei and also in frontal white matter. Precise localization within the basal ganglia and corticostriatal circuitry is not yet determined.

- Dopaminergic pathways appear to mediate complex motor stereotypies. Administration of high doses of levodopa in Parkinson's patients and dopamine in lab rodents have been shown to reduce stereotypies.

Medscape article: Childhood Habit Behaviors and Stereotypic Movement Disorder.

B. Tics

1. Differentiate tics from voluntary and other involuntary movements, such as chorea, athetosis, and ballismus

- Tics- Stereotyped, intermittent, sudden, discrete, repetitive, nonrhythmic movements, most frequently involving head and upper body.
- Stereotypy- Patterned, episodic, repetitive, purposeless, rhythmic movements.
- Chorea- Chaotic, random, repetitive, brief, purposeless movements that are rapid but not as rapid as myoclonus.
- Dystonia- Repetitive, sustained, abnormal postures and movements; abnormal postures typically have a twisting quality.
- Myoclonus- Sudden, brief, shock-like movements that may be repetitive or rhythmic.
- Tremor- Rhythmic oscillation about a central point or position involving any one or more body parts.
- Parkinsonism- Hypokinetic syndrome characterized by rest tremor, slow movement (bradykinesia), rigidity, and postural instability.
- Athetosis- slow, involuntary, convoluted, writhing movements of the fingers, hands, toes, and feet and in some cases arms, legs, neck and tongue.
- Ballismus- a type of involuntary movement affecting the proximal limb musculature, manifested as jerking, flinging movements of the extremity; caused a lesion of or near the contralateral subthalamic nucleus.

Although tics may resemble chorea or myoclonus in some cases, the presence of a premonitory sensory urge that is relieved by tics and the ability to suppress tics voluntarily distinguishes tics from true involuntary movement disorders.

Zinner SH, Mink JW. Movement Disorders I: Tics and Stereotypies. *Pediatrics in Review.* 2010; 31(6): 223-233

2. Know the epidemiology of tics and tic disorders

Simple tics are very common in childhood, as 6% to 13% of all children will experience a transient tic at some time in childhood. The childhood incidence of chronic tic disorder is around 1% to 2%, with approximate 3:1 ratio of boys to girls.

Tourette syndrome has a prevalence of about 0.3% and it is more common in teenagers than in preteens and in white children than black or Hispanic children.

Voigt RG, Macias MM, Myers SM, Tapia CD. *Developmental and Behavioral Pediatrics.* Itasca, IL: American Academy of Pediatrics; 2018.

3. Know the diagnostic criteria for tic disorders, including provisional tic disorder, persistent motor or vocal tic disorder, and Tourette disorder

<u>DSM-5 Diagnostic Criteria for Tic Disorders</u>

Note: A tic is a sudden, rapid, recurrent, nonrhythmic motor movement or vocalization.

Tourette's Disorder

A. Both multiple motor and one or more vocal tics have been present at some time during the illness, although not necessarily concurrently.
B. The tics may wax and wane in frequency but have persisted for more than 1 year since first tic onset.
C. Onset is before age 18 years.
D. The disturbance is not attributable to the physiologic effects of a substance (e.g. cocaine) or another medical condition (e.g., Huntington's disease, postviral encephalitis).

Persistent (Chronic) Motor or Vocal Tic Disorder

A. Single or multiple motor or vocal tics have been present during the illness, but not both motor and vocal.
B. The tics may wax and wane in frequency but have persistent for more than 1 year since first tic onset.
C. Onset is before age 18 years.
D. The disturbance is not attributable to the physiologic effects of a substance (e.g. cocaine) or another medical condition (e.g., Huntington's disease, postviral encephalitis).
E. Criteria have never been met for Tourette's disorder.

Specify if:

With motor tics only
With vocal tics only

Provisional Tic Disorder

A. Single or multiple motor and/or vocal tics.
B. The tics have been present for less than 1 year since first tic onset.
C. Onset is before age 18 years
D. The disturbance is not attributable to the physiologic effects of a substance (e.g. cocaine) or another medical condition (e.g., Huntington's disease, postviral encephalitis).
E. Criteria have never been met for Tourette's disorder or persistent (chronic) motor or vocal tic disorder

Diagnostic and Statistical Manual of Mental Disorders: DSM-5. Washington, DC; American Psychiatric Publishing; 2013.

4. Describe the natural history of tics and tic disorders

Transient tic disorders can last between 4 weeks and 1 year, include single or multiple motor and/or vocal tics, and most are simple rather than complex and do not usually cause great distress. However a child with complex and distressing motor and vocal tics lasting a few months may be at risk for developing Tourette syndrome.

Onset of tics is typically between 4 and 6 years. Peak severity occurs between ages 10 and 12 years, with decline in severity during adolescence. Many adults with tic disorders experience diminished symptoms. A small percentage of individuals will have persistently severe or worsening symptoms in adulthood.

Tic symptoms manifest similarly in all age groups and across the lifespan. Tics wax and wane in severity and change in affected muscle groups and vocalizations over time. As children get older, they begin to report their tics being associated with a premonitory urge—a somatic sensation that

precedes the tic-and a feeling of tension reduction following the expression of the tic. Tics associated with a premonitory urge may be experienced as not completely "involuntary" in that the urge and the tic can be resisted. An individual may also feel the need to perform a tic in a specific way or repeat it until he or she achieves the feeling that the tic has been done "just right."

The vulnerability toward developing co-occurring conditions changes as individuals pass through the age of risk for various co-occurring conditions. For example, prepubertal children with tic disorders are more likely to experience ADHD, OCD, and separation anxiety disorder than are adolescents and adults, who are more likely to experience the new onset of major depressive disorder, substance use disorder, or bipolar disorder.

Voigt RG, Macias MM, Myers SM, Tapia CD. *Developmental and Behavioral Pediatrics.* Itasca, IL: American Academy of Pediatrics; 2018.

Diagnostic and Statistical Manual of Mental Disorders: DSM-5. Washington, DC; American Psychiatric Publishing; 2013.

5. Know the conditions commonly associated with Tourette disorder (eg, ADHD and OCD)

ADHD
OCD
Learning disabilities/learning difficulties
Anxiety disorders
Mood disorders
Oppositional defiant disorder
Self-injurious behaviors
Speech and language disorders (eg, hesitations, disfluency)
Intermittent explosive disorder/anger dysregulation

Zinner SH, Mink JW. Movement Disorders I: Tics and Stereotypies. *Pediatrics in Review.* 2010; 31(6): 223-233

Forty percent of children with TS also meet criteria for OCD and more than 20% of children with tic disorder have OCD. Sixty-four percent of children with TS have been diagnosed with ADHD and half of all children with tic disorders have ADHD. Clinical depression (impacting 36% of individuals with TS), anxiety (40%), developmental problems (28%), and learning difficulties (80%) are also common comorbidities of TS.

Voigt RG, Macias MM, Myers SM, Tapia CD. *Developmental and Behavioral Pediatrics.* Itasca, IL: American Academy of Pediatrics; 2018.

6. Understand the pathophysiology of Tourette disorder

The pathogenesis of tic disorders is complicated and not well understood, although a wealth of biomedical research strongly supports a defective filtering, or "sensorimotor gating," mechanism, resulting in urges to perform elements of otherwise purposeful activity at inappropriate times, intensities, and frequencies. The central concept focuses on the basal ganglia and their role within brain-based circuits that also include the neocortex and thalamus. Other key brain regions, such as the midbrain, may be involved as well. When functioning properly, the basal ganglia assist in the execution of desired behaviors expressed by these circuits, but the basal ganglia also function to prevent the completion of undesired behaviors.

Many desired behaviors, when learned through repetition, may become automatic or sequenced as "prepackaged and ready-to-use," perhaps stored and reinforced in these circuits. Imprecise regulation results in the expression of bits of these behaviors, such as tics. This model also helps to explain related or comorbid conditions that, like tics, share features of loss of behavioral inhibition, such as compulsions or impulsive behaviors, and to explain why these behaviors often mimic purposeful activities. Chronic primary tic disorders and many of their associated conditions are strongly influenced genetically, although the inheritance pattern is unclear. Environmental factors, such as sleep insufficiency, stress, and possibly in utero exposure to maternal smoking, also are important as mediators of symptom severity.

Zinner SH, Mink JW. Movement Disorders I: Tics and Stereotypies. *Pediatrics in Review.* 2010; 31(6): 223-233

7. Plan the evaluation of a child with a tic disorder

   Does not require laboratory or imaging studies. Comprehensive history (including family history of tics and associated problems) and physical examination can rule out possible causes of secondary tics (infections, post infections, drugs, toxins, neurodevelopmental disorders, chromosomal disorders, stroke, neoplasm, heredodegenerative disorders [Huntington's, neuroacanthocytosis, Pantothenate kinase-associated neurodegeneration-PKAN], neurocutaneous disorders, head trauma, seizure disorders, and psychogenic factors). If diagnostic uncertainty, referral to a neurologic is indicated.

   Zinner SH, Mink JW. Movement Disorders I: Tics and Stereotypies. *Pediatrics in Review.* 2010; 31(6): 223-233

8. Plan the treatment for a child with a tic disorder

   The first approach to managing tic disorders should be providing education and reassurance. Education should focus on tic disorders as neurologically based, which often includes reversing long-held misconceptions and myths, and providing educational resources.

   When tics are chronic, anticipatory guidance can help avert needless interventions and the basis of management should focus on forming an identity separate from the tic disorder and maintaining and supplementing the development of academic, organizational, and interpersonal skills with an eye to ensuring a satisfying, independent adulthood. When tics cause impairment, specific treatments may be indicated. It is important to establish clear treatment goals, with the expected outcome being the reduction rather than the eradication of tics. Ongoing monitoring and reassuring periodic reminders that chronic tics usually improve during adolescence are adequate tic management. Children should also be screened for comorbid conditions above.

   One behavioral treatment is comprehensive behavioral intervention for tics (CBIT) that includes "awareness training" and "competing-response training (CRT)". It employs an individualized program, including recognition training of premonitory urges, followed by a competing response with "functional analysis" to disrupt the urge-tic-relief cycle that, when successful, seems ultimately to weaken the urge.

   Zinner SH, Mink JW. Movement Disorders I: Tics and Stereotypies. *Pediatrics in Review.* 2010; 31(6): 223-233

9. Know the pharmacologic interventions that can be helpful in management of tics

No medication has been designed specifically for tic reduction, and none eliminates tics entirely. A reasonable goal is to reduce tic severity to a state of tolerable functional outcomes that has acceptable adverse effects. The most consistently effective available tic-reducing agents work by blocking dopamine neurotransmission within regions of the basal ganglia. A conservative medication approach for moderate-to-severe tics begins with an alph-2-adrenergic agonist (clonidine, guanfacine) with the antipsychotics/neuroleptic agents (atypical-risperidone, ziprasidone, typical-haloperidol, pimozide) as the next option. Stimulants for comorbid ADHD and SSRIs for comorbid OCD.

Zinner SH, Mink JW. Movement Disorders I: Tics and Stereotypies. *Pediatrics in Review.* 2010; 31(6): 223-233

Voigt RG, Macias MM, Myers SM, Tapia CD. *Developmental and Behavioral Pediatrics.* Itasca, IL: American Academy of Pediatrics; 2018.

10. Recognize the behavioral and developmental complications of Tourette disorder

Learning disabilities (LDs) are another common co-occurring problem, although children who have TS may face obstacles to effective learning due to reasons other than LD, including ADHD, obsessive thoughts or compulsions, and complications due to other comorbid conditions, such as anxiety, poor frustration tolerance, insufficient sleep, and executive function difficulties such as poor organizational skills. Tics themselves can interfere with operations such as reading, writing, listening, and speaking. In addition, tics can be distracting and sometimes physically challenging, further jeopardizing the child's ability to attend.

Explosive anger and aggression, or episodic "rage attacks," are seen in 25% or more of referred patients who have TS, and parents overwhelmingly regard these events as the most problematic feature. Their origin likely is multifactorial, related both to primary disorders and secondary consequences of having a chronic disorder. These attacks may be related to comorbid conditions, including aggressive obsessions or other anxieties, loss of impulse control or other disinhibited urges, and hyperarousal. Although tics often improve or resolve in adolescence and young adulthood, it is not yet possible to predict a tic course for individual patients. A growing identification of endophenotypes, such as fine motor skills deficits, is emerging that may suggest degrees of basal ganglia dysfunction that could help serve as clues to predict greater tic severity in adulthood.

Zinner SH, Mink JW. Movement Disorders I: Tics and Stereotypies. *Pediatrics in Review.* 2010; 31(6): 223-233

11. Understand the genetics of Tourette disorder

In 2005, the first reported gene associated with TS was identified on chromosome 13 in 3 of 174 patients, although subsequent studies of this gene's association with TS have yielded mixed findings. In 2007, strong linkage to chromosome 2p was reported in a large genome scan. It is likely that the genetics of TS are more complex than previously believed. An autosomal dominant mode of transmission with incomplete penetrance and possible polygenic and additive factors and environmental influences seems likely.

Zinner SH, Mink JW. Movement Disorders I: Tics and Stereotypies. *Pediatrics in Review.* 2010; 31(6): 223-233

C. Self-injurious behaviors without developmental disabilities

1. Know the epidemiology of self-injurious behavior in children without developmental disabilities

- Typically begins between ages 12-14 years old
- Can occur prior to age 12 but rarely in children as young as 5-7 years old
- Prevalence
    - In the general population, the lifetime prevalence of at least one episode of non-suicidal self-injury (NSSI) is approximately 17-18%, though can range between 14-20%
    - Among adolescent patients in a clinic setting, the lifetime prevalence is 60-80%
    - There are not yet definitive studies indicating whether there is a difference in prevalence based on gender or race/ethnicity in community samples.
        - There is a higher prevalence of NSSI among females in clinical samples
    - There is a higher prevalence of NSSI among adolescents who identify as gay, lesbian or bisexual compared to adolescents who identify as heterosexual

Glenn C, Nock K. "Nonsuicidal ideation and behavior in children and adolescents: Evaluation and management." *UpToDate.* https://www.uptodate.com/contents/nonsuicidal-self-injury-in-children-and-adolescents-epidemiology-and-risk-factors?search=self%20injurious%20behavior&topicRef=116734&source=see_link
Heath N, Toste JR, Moore TR, Symons F. "Self-Harm" *Textbook of Pediatric Care. https://pediatriccare.solutions.aap.org/chapter.aspx?sectionid=109663654&bookid=1626*

2. Understand the natural history of self-injurious behavior in children without developmental disabilities
- Typically begins in early adolescence
- Thoughts of NSSI precede NSSI behavior by 4-6 months on average
- According to UpToDate, in a study of a random sample of adolescents followed over 13 years, most adolescents who engaged in NSSI during adolescence stopped by the end of adolescence/beginning of young adulthood.
    - Females were more likely to continue self-injury into adulthood than males
    - Self-reinforcing properties may be a reinforcing factor in those who continue self-injury
- Adolescents who discontinue self-injury may continue to use maladaptive strategies, such as substance use, to regulate negative emotions

Glenn C, Nock M. "Nonsuicidal self-injury in children and adolescents: Clinical features and proposed diagnostic criteria." *UpToDate*. https://www.uptodate.com/contents/nonsuicidal-self-injury-in-children-and-adolescents-clinical-features-and-proposed-diagnostic-criteria?search=self%20injurious%20behavior&topicRef=116730&source=see_link

3. Recognize the signs of self-injurious behaviors that occur in adolescents without developmental disabilities
- NSSI should be suspected in children/adolescents with
    - Frequent unexplained injuries or "accidents" to forearms, wrists, hands, or other body parts, particularly injuries that lead to scarring on the nondominant arm
    - Unusual or inappropriate dress:
        - Multiple bracelets, wristbands, or other jewelry that covers large areas of the forearm
        - Long sleeves in warmer temperatures
        - Not participating in activities that require less clothing (ex: gym class or swimming)
    - Prior history of NSSI
- Physical exam may reveal the following:
    - Fresh injuries, scars, burns or unexplained bruises
    - Pin or razor blade scratches (that could be explained as "cat scratches")
- Risk factors for NSSI
    - Hopelessness

- Psychopathology such as depression, disordered eating, emotional/internalizing problems, behavioral/externalizing problems,
- Sleep problems
- Affect dysregulation
- Distress
- Impulsivity
- History of childhood maltreatment
- Negative life events or stressors, bullying, peer victimization
- Prior suicidal thoughts and/or behaviors
- Exposure to peer NSSI behaviors
- Self-reported likelihood of engaging in future NSSI
- Parental psychopathology
- Impaired family functioning

Hornor G. "Nonsuicidal Self-Injury" *Journal of Pediatric Health Care*; 2016; 30(3):261-267

4. Distinguish between self-injurious behaviors that are suicidal in intent, and those that are not

- According to UpToDate, "By definition, NSSI is distinguished from suicidal behavior; socially accepted practices such as tattoos, piercings, and religious rituals; accidental self-harm; and indirect self-injury through behaviors such as disordered eating or substance use disorders."
- NSSI and suicidal behavior differ by motivation
  - NSSI does not represent a failed suicide attempt
  - NSSI occur most commonly to regulate negative emotions
- NSSI typically occurs more often than suicidal attempts
- Medical severity/lethality is usually greater with attempted suicide than NSSI
- The methods of non-suicidal self-injury and suicidal behavior differ
  - Most common methods of NSSI
    - Cutting of skin
    - Burning
    - Severe scratching
  - Other methods of NSSI
    - Carving words or symbols into the skin
    - Hitting or banging body parts
    - Breaking bones
    - Biting self to point of bleeding
    - Rubbing skin against rough surfaces
    - Inserting needles or other objects under the skin
    - Wound picking and interfering with wound healing
    - Pinching skin to point of bleeding
    - Pulling out hair (excluding trichotillomania)
    - Swallowing dangerous chemicals to cause tissue damage
  - Most common methods of suicide attempts
    - Poisoning (overdose)
    - Suffocation (hanging)
    - Firearms

Glenn C, Nock M. "Nonsuicidal self-injury in children and adolescents: Clinical features and proposed diagnostic criteria." *UpToDate*. https://www.uptodate.com/contents/nonsuicidal-self-injury-in-children-and-adolescents-clinical-features-and-proposed-diagnostic-criteria?search=self%20injurious%20behavior&topicRef=116730&source=see_link

5. Know how to evaluate an adolescent without disabilities who engages in self-injurious behaviors

- History
    - Interview the patient and parent separately
    - Ask directly about NSSI behaviors. If behaviors are endorsed, should assess suicide risk
    - Examiner should assess
        - Relationships with family, friends, and peers
        - Interpersonal stressors
        - For other psychiatric symptoms and disorders
        - Support from family, friends, teachers, etc.
        - Whether patient is already receiving treatment
    - Identify the following during the evaluation:
        - Onset
        - Past and current frequency of behavior
        - Past and current methods of self-injury
        - Medical severity/lethality of self-injury
        - Location of injuries on the body
        - Context (i.e. alone or around others)
        - Duration of an episode
        - Antecedents and consequents
        - Function of behavior
        - Protective factors
        - Impact on functioning
        - Past attempts to stop the behavior and current motivation for stopping
- Assessment instruments (can help inform treatment)
    - Functional Assessment of Self-Mutilation
    - Self-Injurious Thoughts and Behaviors Interview
- Physical Exam
- Identify comorbid psychiatric illness
- Evaluate risk factors

Peterson J, Freedenthal S, Sheldon C, Andersen R. "Nonsuicidal Self injury in Adolescents." *Pschiatry*; 2008; 5(11):20-26
Glenn C, Nock M. "Nonsuicidal self-injury in children and adolescents: Assessment." *UpToDate.* https://www.uptodate.com/contents/nonsuicidal-self-injury-in-children-and-adolescents-assessment?search=self%20injurious%20behavior&topicRef=116734&source=see_link

6. Know how to plan the treatment of an adolescent without disabilities who engages in non-suicidal self-injurious behaviors

- Treatment
    - First line – dialectical behavior therapy adapted for adolescents
    - Second line – cognitive behavior therapy
    - Third line – Other psychotherapy, such as psychodynamic psychotherapy with a family component, interpersonal psychotherapy, family therapy, or emotion regulations group therapy
- When to refer
    - Following initial risk assessment
    - Emotional regulation difficulties or poor coping strategies
- When to Admit
    - Threat of suicide

- Serious physical injury
- Underlying psychiatric conditions

Peterson J, Freedenthal S, Sheldon C, Andersen R. "Nonsuicidal Self injury in Adolescents." *Pschiatry*; 2008; 5(11):20-26
Heath N, Toste JR, Moore TR, Symons F. "Self-Harm" *Textbook of Pediatric Care.*
https://pediatriccare.solutions.aap.org/chapter.aspx?sectionid=109663654&bookid=1626
Glenn C, Nock M. "Nonsuicidal self-injury in children and adolescents: Prevention and choosing treatment." *UpToDate.*
https://www.uptodate.com/contents/nonsuicidal-self-injury-in-children-and-adolescents-prevention-and-choosing-treatment?search=self%20injurious%20behavior&topicRef=116738&source=see_link

7. Know the potential causes and associated conditions of self-injurious behavior in children without developmental disabilities

- Pathogenesis largely unknown, though likely due to a combination of psychological, social, and biological factors
- Psychological factors
  - Functions of behavior can help understand how NSSI are initiated and continue. Functions can be categorized as internal (for automatic purposes) or external (for social purposes):
    - Intrapersonal negative reinforcement – the behavior reduces negative emotions or thoughts, such as anger, sadness, and anxiety
    - Intrapersonal positive reinforcement – the behavior generates a desired feeling or thought
    - Social negative reinforcement – behavior facilitates an escape from undesired social demands or intolerable social situations
    - Social positive reinforcement – the behavior elicits a positive response from others, such as attention
- Social factors
  - Childhood neglect or abuse
  - Peer influences
  - Interpersonal factors, such as:
    - Peer victimization
    - Loneliness and social isolation
    - Interpersonal loss
    - Perceived and actual parental criticism
    - Difficulty communicating with family members
  - NSSI can be socially reinforced if behaviors help the adolescent obtain something they want in their environment or removes something negative from their environment
- Biological factors
  - Studies have looked at the role of genetics, endogenous opioids, altered functioning of the HPA axis, altered neural circuitry

Glenn C, Nock M. "Nonsuicidal self-injury in children and adolescents: Pathogenesis." *UpToDate.*
https://www.uptodate.com/contents/nonsuicidal-self-injury-in-children-and-adolescents-prevention-and-choosing-treatment?search=self%20injurious%20behavior&topicRef=116738&source=see_link
Hornor G. "Nonsuicidal Self-Injury" *Journal of Pediatric Health Care*; 2016; 30(3):261-267 Heath N, Toste
Heath N, Toste JR, Moore TR, Symons F. "Self-Harm" *Textbook of Pediatric Care.*
https://pediatriccare.solutions.aap.org/chapter.aspx?sectionid=109663654&bookid=1626

8. Know the conditions commonly associated with self-injurious behavior in children without developmental disabilities

- Depressive disorders

- Anxiety disorders
- Disordered eating
- Conduct disorder/Oppositional defiant disorder
- Personality disorders
- Posttraumatic stress disorder
- Substance use disorders
- Sleep disorders
- Affect dysregulation
- Risky sexual behavior

Hornor G. "Nonsuicidal Self-Injury" *Journal of Pediatric Health Care*; 2016; 30(3):261-267
Glenn C, Nock M. "Nonsuicidal self-injury in children and adolescents: Clinical features and proposed diagnostic criteria." *UpToDate.* https://www.uptodate.com/contents/nonsuicidal-self-injury-in-children-and-adolescents-clinical-features-and-proposed-diagnostic-criteria?search=self%20injurious%20behavior&topicRef=116730&source=see_link

D. Repetitive/disruptive/self-injurious behavior in dev. disabilities

1. Know the epidemiology of self-injurious behavior among children with developmental disabilities
    - Prevalence reports are heterogeneous ranging from 4-53% in children with developmental disabilities

Richman DM. "Early intervention and prevention of self-injurious behavior exhibited by young children with developmental disabilities." *Journal of Intellectual Disability Research*. 2008; 52: 3-17

2. Recognize the specific genetic disorders that are associated with an increased risk of self-injurious behavior
    - Lesch-Neyhan
        - X-linked recessive condition due to mutation in gene coding for the enzyme hypoxanthine-guanin phosphoribosyltransferase (HPRT)
        - Behaviors include self-mutilating behavior
    - Rett syndrome
        - Due to mutation in MECP2 gene on X chromosome
        - Behaviors include finger kneading and rubbing, hand clapping and washing, wringing, squeezing, twisting, and pill rolling
    - Cornelia de Lange
        - Due to mutation of 5p13.1
        - Behaviors may include self-injurious behaviors, hyperactivity, repetitive behaviors, anxiety, social impairments, low mood
    - Prader Willi
        - Due to paternal deletion on chromosome 15
        - Behaviors may include hyperphagia, temper tantrums, impulsivity, skin picking, repetitive speech, stubbornness, and aggression
    - Angelman Syndrome
        - Due to absence of maternally derived genetic maternal on chromosome 15
        - Behaviors may include strong drive for adult attention, high levels of laughing and smiling, sleep difficulties, hyperactivity and inattention
    - Smith Magenis Syndrome
        - Typically caused by de novo deletion of 17p11.2, but is due to mutation of retinoic acid-induced 1 gene on chromosome 17 in 10% of cases

    - Behaviors may include self-injurious behaviors, sleep difficulties, aggressive behavior, restlessness, distractibility, hyperactivity, unique "self-hug"
- Fragile X
    - Due to expansion of trinucleotide repeat sequence, CCGG, in promoter region of FMR1 located at Xq17.3 on the long arm of the X chromosome
    - Behaviors may include aggression, inattention, hyperactivity, and ASD-like behaviors
    - Most common inherited form of intellectual disability
- Cri du Chat:
    - Caused by deletion of 5p15 region on short arm of chromosome 5
    - Behaviors may include self-injurious behavior, repetitive behavior, obsessive attachment to objects, sleep problems, hypersensitivity to sensory stimuli, and destructive behavior

Powis L, Oliver C. "The prevalence of aggression in genetic syndromes: A review." *Research in Developmental Disabilities*. 2014; 35: 1051-1071
Jankovic J. "Hyperkinetic movement disorders in children." *UpToDate*. https://www.uptodate.com/contents/hyperkinetic-movement-disorders-in-children?search=lesch%20nyhan%20syndrome&sectionRank=1&usage_type=default&anchor=H25&source=machineLearning&selectedTitle=1~36&display_rank=1#H25
Schultz R, Glaze D. "Rett syndrome: Genetics, clinical features, and diagnosis." *UpToDate*. https://www.uptodate.com/contents/rett-syndrome-genetics-clinical-features-and-diagnosis?search=rett%20syndrome&source=search_result&selectedTitle=1~34&usage_type=default&display_rank=1

3. Describe the complications that may result from self-injurious behavior in children with developmental disabilities

Type of Self-Injury	Complication
Head Banging	Detached retina, abrasions, contusions
Head hitting or slapping	Fracture of bones in hand, detached retina, abrasions, contusions
Eye poking	Eye abrasions
Gum or tooth digging or banging	Gum injury, tooth auto extraction, tooth fracture
Scratching or skin picking	Infection, scarring
Finger and toenail biting or picking	Infection, nail removal, ingrown nails, paronychia
Kicking or stomping	Bruises, fracture
Rumination	Esophageal ulceration and bleeding, dental damage, nutritional compromise, precancerous lesions or esophagus

Hyman SL, Levy SE, Myers SM. "Identification, Evaluation, and Management of Children with Autism Spectrum Disorder." *Pediatrics*. 2020; 145(1): e20193447

4. Understand the etiologies of self-injurious, repetitive, and disruptive behavior among children with developmental disabilities

- The following models have been developed to explain the cause of self-injurious behavior in children with developmental disabilities
    - Theoretical Model – early rhythmic stereotypies that are characteristic in typical and delayed development are the precursor to self-injurious behavior, and are reinforced by social contingencies
    - Conceptual Model – neurobiological and genetic factors lead to the initial emergence of self-injurious behaviors, which are reinforced and maintained by caregiver implemented contingencies
- Self-injurious behaviors are likely multifactorial

- Physiological Causes
  - Biochemical – some research has suggested that self-injurious behavior may lead to the release of beta-endorphins, which can provide a euphoric-like feeling
  - Seizures in the frontal or temporal lobes can lead to seizure activity leading to headbanging, slapping ears/head, hand biting, chin hitting, scratching of arms and face. Functional behavioral analysis will not show relationship between the individual's behavior and environment, though stressor can lead to seizure activity
  - Hypo- or hyper-arousal
  - Some genetic conditions (see #2 above) are associated with self-injurious behavior
  - Pain – for instance, head banging or hitting may be a response to ear infection or migraine
  - Sensory
- Social Causes
  - Communication
  - Attention-seeking
  - Avoidance
  - To obtain a tangible

Hagopian LP, Leoni M. "Self-injurious Behavior among Individuals with Intellectual and Developmental Disabilities." *Acta Psychopathologica*. 2017; 3(5): 1-9.
Edelson, S. "Understanding and Treating Self-Injurious Behavior." Autism Research Institute. https://www.autism.org/self-injury/#references5. Understand the role of functional behavioral analysis in the evaluation of self-injurious, repetitive, and disruptive behaviors in children with developmental disabilities

5. Understand the role of functional behavioral analysis in the evaluation of self-injurious, repetitive, and disruptive behaviors in children with developmental disabilities

- Frontline approach to understanding challenging behaviors, including self-injurious behaviors
- Functional behavior assessment (FBA) identifies environmental events that influence the individual's responses. Looks systematically at Antecedents, Behavior and Consequences.
- Treatments are based on information obtained from FBA, and are focused on the modification of the contingencies suggested as directly responsible for behavioral maintenance
- Involves systematically exposing the individual to controlled conditions, where the antecedent and consequent events are manipulated while changes in self-injurious behavior are recorded
- FBA can help categorize self-injurious behaviors as socially-mediated and non-socially mediated
  - Socially-mediated
    - Attention
    - Access to tangible
    - Escape from demand
  - Non-socially mediated: the behavior itself produces reinforcement through biologically mediated processes

Hagopian LP, Leoni M. "Self-injurious Behavior among Individuals with Intellectual and Developmental Disabilities." *Acta Psychopathologica*. 2017; 3(5): 1-9.

6. Know how to plan the management for patients with developmental disabilities who engage in self-injurious, repetitive, or disruptive behavior

- Behavioral therapy based on the results of the functional behavioral assessment is first line treatment

- Consider pharmacological intervention when behavioral treatment alone does not address the behavior. Pharmacological treatment should be implemented in conjunction with behavioral treatment

Hagopian LP, Leoni M. "Self-injurious Behavior among Individuals with Intellectual and Developmental Disabilities." *Acta Psychopathologica*. 2017; 3(5): 1-9.

7. Know the pharmacologic treatment of self-injurious behaviors

- Atypical antipsychotics – risperidone, aripiprazole
    - Most effective if combined with behavioral strategies
    - Adverse effects: weight gain, increased appetite, dyslipidemia, fatigue, drowsiness, dizziness, drooling
    - Monitor for: extrapyramidal symptoms; weight, height, BMI; glucose and lipid levels.
- Alpha-2 adrenergic agonists (clonidine, guanfacine)
    - Small studies documenting beneficial effects
    - May have better adverse effect profiles than antipsychotics

Sabus A, Feinstein J, Romani P, Goldson E, Blackmer A. "Management of Self-injurious Behaviors in Children with Neurodevelopmental Disorders: A Pharmacotherapy Overview." *Pharmacotherapy*. 2019; 39(6): 645-664.
Hyman SL, Levy SE, Myers SM. "Identification, Evaluation, and Management of Children with Autism Spectrum Disorder." *Pediatrics*. 2020; 145(1): e20193447

8. Recognize the spectrum of repetitive behaviors that may be seen in children with autism spectrum disorder

- The repetitive behaviors and restricted interests of children with autism spectrum disorder can be divided into the following categories
    - Repetitive use of objects
    - Unusual sensory interests
    - Hand/finger mannerisms
    - Complex mannerisms
    - Compulsions and rituals
    - Difficulties with changes in routine
    - Resistance to change
    - Unusual preoccupations
    - Unusual attachment to objects
    - Sensitivity to noise
    - Abnormal/idiosyncratic response to sensory stimuli
    - Circumscribed interests
    - Self-injurious behaviors

Richler J, Bishop S, Kleinke J, Lord C. "Restricted and Repetitve Behaviors in Young Children with Autism Spectrum Disorders." *Journal of Autism and Developmental Disorders*. 2007; 37: 73-85.
Bishop SL, Hus V, Duncan A, Huerta M, Gotham K, Pickles A, Kreiger A, Buja A, Lund S, Lord C. "Subcategories of restricted and repetitive behaviors in children with autism spectrum disorders." *Journal of Autism and Developmental Disorders*. 2013; 43(6): 1287-1297

9. Know that some self-injurious, disruptive, or repetitive behaviors may be the result of an unrecognized medical disorder in patients with developmental disabilities

Type of Self-Injury	Potential Associated Conditions
Head Banging	Headache, toothache, sinus infection, ear infection
Head hitting or slapping	Headache, toothache, sinus infection, ear infection
Eye poking	Vision loss, eye pain

Gum or tooth digging or banging	Dental pain, gingivitis
Scratching or skin picking	Allergy, eczema, drug reaction, skin infection or infestation (fleas, scabies)
Finger and toenail biting or picking	Pain
Kicking or stomping	Restless leg syndrome, leg pain
Rumination	Gastroesophageal reflux, eosinophilic esophagitis

Hyman SL, Levy SE, Myers SM. "Identification, Evaluation, and Management of Children with Autism Spectrum Disorder." *Pediatrics*. 2020; 145(1): e20193447

10. Know how to evaluate a child with a developmental disability who engages in repetitive, disruptive, or self-injurious behaviors

- The evaluation of a child with a developmental disability who engages in repetitive, disruptive or self-injurious behaviors includes the following:
    - History and physical exam
        - The following checklists and questionnaires can help in obtaining the history:
            - Child Behavior Checklist
            - Aberrant Behavior Checklist
            - Functional Analysis Screening Tool
            - Problem Behavior Questionnaire
            - Functional Assessment for Multiple Causality
            - Questions About Behavioral Function
            - Functional Assessment Interview
            - Behavior Problems Inventory
        - Assess for unrecognized medical disorders
    - Functional Behavior Analysis

Clay C, Jorgenson C, Kahng S. "Self-Injurious Behavior in Children with Intellectual and Developmental Disabilities: Current Practices in Assessment and Treatment." *Handbook of Childhood Psychopathology and Developmental Disabilities Treatment*. 2018; 269-285

Vollmer T, Sloman K, Borrero C. "Behavioral Assessment of Self-Injury." *Assessing Childhood Psychopathology and Developmental Disabilities*. 2009; 341-369.

11. Know the pharmacologic management of repetitive behaviors in individuals with developmental disabilities

- Atypical antipsychotics (risperidone, aripiprazole)
    - There have been multiple double blinded placebo-controlled studies documenting improvement in repetitive behavior for short-term use
- Anticonvulsants
    - Modest improvement has been reported with divalproex sodium treatment
    - Topiramate may lead to improvement when combined with risperidone
    - Adverse effects: sedation
- Hyman SL, Levy SE, Myers SM. "Identification, Evaluation, and Management of Children with Autism Spectrum Disorder." *Pediatrics*. 2020; 145(1): e20193447

12. Know the pharmacologic management of disruptive behaviors in individuals with developmental disabilities

- Atypical antipsychotics – risperidone, aripiprazole
    - Most effective if combined with behavioral strategies

    - Adverse effects: weight gain, increased appetite, dyslipidemia, fatigue, drowsiness, dizziness, drooling
    - Monitor for: extrapyramidal symptoms; weight, height, BMI; glucose and lipid levels.
- Alpha-2 adrenergic agonists (clonidine, guanfacine)
    - Small studies documenting beneficial effects
    - May have better adverse effect profiles than antipsychotics

Hyman SL, Levy SE, Myers SM. "Identification, Evaluation, and Management of Children with Autism Spectrum Disorder." *Pediatrics*. 2020; 145(1): e20193447

13. Know the behavioral treatments that may be used for self-injurious, repetitive, or disruptive behaviors in children with developmental disabilities

- ABA
- Function-Based Interventions
    - Differential reinforcement of alternative behavior: reinforcing a behavior that differs from the behavior that is targeted to be reduced ex: Functional Communication Training
    - Differential reinforcement of other behavior: reinforcing the absence of the target behavior
        - Reinforcement is provided after a predetermined level of time if the target behavior does not occur
    - Noncontingent reinforcement: provide reinforcement continuously or at predetermined intervals regardless of behavior

Hagopian LP, Leoni M. "Self-injurious Behavior among Individuals with Intellectual and Developmental Disabilities." *Acta Psychopathologica*. 2017; 3(5): 1-9.
Richman DM. "Early intervention and prevention of self-injurious behavior exhibited by young children with developmental disabilities." *Journal of Intellectual Disability Research*. 2008; 52: 3-17

E. Alterations in mental status

1. Recognize the signs and symptoms of delirium

    As in the adult population, delirium in the pediatric population can be classified based on the psychomotor state into hyperactive, hypoactive, or mixed delirium. While many of the clinical features of adult delirium can be applied to children, certain features are more prominent in children, which necessitate a unique approach to the pediatric delirium examination. For example, in a preverbal child the examiner might forgo formal bedside tests of attention and instead assess inattentiveness by observing poor eye contact or difficulty with engagement. Caregiver involvement can prove very helpful in making this diagnosis.

    Features of delirium that are particularly prominent in the pediatric population include irritability, affective lability, agitation, sleep-wake disturbance, and fluctuations of symptoms. In contrast, delusions, hallucinations, speech disturbances, and memory deficits are less commonly seen in children. Unique features of pediatric delirium include developmental regression with loss of previously acquired skills, inability of the usual caregiver to console the child, and reduced eye contact with the usual caregiver. Schieveld and Janssen also described the "inconsolable child" as a red flag for delirium. A child who is agitated, breathing against the ventilator, and receiving escalating doses of sedating medications, should be considered to be delirious until proven otherwise.

Thom RP. Pediatric Delirium. *American Journal of Psychiatry.* 2017; 12(2): 6-8

Disturbance in behavior (aggression, agitation, fidgeting, increased activity level, restlessness)
Disturbance in circadian rhythm (insomnia, sleep-wake cycle disturbance)
Disturbance in cognition and mood (anxiety, depression, disorientation, impaired short-term memory, irritability, language disturbance, memory deficit, pressured speech)
Disturbance in consciousness (confusion, disorientation)
Disturbance in perception (apprehension, hallucinations, poor judgement)

Holly. C, Porter S, Echevarria M, et al. Recognizing Delirium in Hospitalized Children: A Systemic Review of the Evidence of Risk Factors and Characteristics. *The American Journal of Nursing*. 2018; 118(4): 24-36

2. Recognize the signs and symptoms of dementia

   Loss of memory, difficulty in language, decline in intellectual skills, changes in personality.

   https://www.news-medical.net/health/Childhood-Dementia-Signs-and-Symptoms.aspx

3. Understand the heightened risk for delirium or dementia in some disorders causing developmental disabilities

   Developmental delay is a risk factor for the development of delirium in hospitalized children.

   Holly. C, Porter S, Echevarria M, et al. Recognizing Delirium in Hospitalized Children: A Systemic Review of the Evidence of Risk Factors and Characteristics. *The American Journal of Nursing*. 2018; 118(4): 24-36

   Dementia in children and young adults is most frequently caused by neuronal ceroidlipofuscinoses (NCL) is a group for incurable lysosomal storage disorders linked by the accumulation of a characteristic intracellular storage material and progressive deterioration, usually in combination with visual loss, epilepsy, and motor decline.

   Schulz A, Kohlschutter A. NCL Disorders: Frequent Causes of Childhod Demential. *Iran J Child Neurol.* 2013; 7(1): 1-8.

   Other conditions associated with increased risk of dementia include Gaucher's Disease, Tay-Sach's Disease, Niemann-Pick Disese type C, Myotonic dystrophy, Fabry's disease, Lesch-Nyhan syndrome, mitochondrial disorders, Wilson's disease, Down's Syndrome, Fucosidosis (a-Fucosidase Deficiency), and Schindler Disease (alpha-N-Acetylgalactosamindase Deficiency).

   Kline M. *Rudolphs Pediatrics.* New York; McGraw-Hill Education; 2018.

4. Know the differential diagnosis for delirium and dementia

   Delirium differential diagnosis: psychotic disorder and bipolar and depressive disorders with psychotic features, acute stress disorder, malingering and factitious disorder, and other neurocognitive disorders.

   Dementia differential diagnosis: delirium, major depressive disorder, specific learning disorder and other neurodevelopmental disorders.

   Diagnostic and Statistical Manual of Mental Disorders: DSM-5. Washington, DC; American Psychiatric Publishing; 2013.

5. Plan the evaluation of a child with delirium or dementia

Delirium: A fundamentally clinical diagnosis. Because of the inherent communication limitations in evaluating preverbal or nonverbal children, the diagnosis is contingent on close observation of behavioral symptoms. Symptoms of pediatric delirium can be subtle, can vary depending on developmental stage, and are complicated by developmental variability. Involvement of the caregiver, who may not be easily accessible, is necessary to make a diagnosis. Many symptoms overlap with other conditions such as pain, distress, and drug withdrawal. Several validated delirium rating/screening tools are available (The Pediatric Anesthesia Emergence Delirium Scale, the Pediatric Confusion Assessment Method for the ICU, the Cornell Assessment of Pediatric Delirium, and the Sophia Observation Withdrawal Symptoms-Pediatric Delirium Scale).

Thom RP. Pediatric Delirium. *American Journal of Psychiatry.* 2017; 12(2): 6-8

Dementia: The clinical assessment of a patient with cognitive impairment should be the same regardless of age but with breadth of the differential diagnosis in younger patients, which include many rare disease, demands a structured approach. The first objective is to determine the pattern of cognitive and behavioral deficit, and will include cognitive and behavioral/psychiatric assessments. The second is to determine the involvement of the nervous system more generally, which includes a neurologic exam. Finally, a physical exam which may show clues to the cause of cognitive dysfunction that lie outside the nervous system. All patients with young-onset dementia should have structural neuroimaging and CSF examination as recommended by the American Academy of Neurology and European Federation of Neurological Societies guidelines. Decisions about whether to undertake tissue biopsies are based on the clinical phenotype and usually confined to the dementia plus syndromes. When a diagnosis cannot be established, observation and repeat investigations are often informative.

Rossor MN, Fox NC, Mummery CJ et al. The diagnosis of young-onset dementia. Lancet Neurol. 2010; 9(8): 793-806.

Content Category 22- Law, Policy, & Ethics- A-C

ABP Content Specs for Developmental-Behavioral Pediatrics Fellow Collaboration Project

Prepared by: Christine McGivney, Mediatrix Mbamalu, Ana Treadwell, DBP Boston Medical Center

Reviewed by Naomi Steiner, Boston Medical Center

Final Version/Faculty Reviewed

22.Law, Policy, and Ethics

A. Legal rights and processes
   1. Individuals with disabilities
      a. Know the criteria for early intervention or special education for children from birth through 21 years of age
      b. Understand the different implications of the Americans with Disabilities Act for public, private, and parochial schools
      c. Know the components of an Individual Family Service Plan (IFSP)
      d. Understand parents' rights to participate in special education decisions as described in the Individuals with Disabilities Education Act (IDEA)
      e. Understand how to apply the concept of least restrictive environment to designing an educational program for a child with a disability
      f. Understand the importance to special education of the related concepts of natural environments, inclusion, and mainstreaming
      g. Appreciate the right to a publicly funded evaluation and special education services for all children in need, including those attending private and parochial schools
      h. Recognize the necessity to provide medical services within the school setting if required by a child in order to participate in an educational program
      i. Understand the components of the Individuals with Disabilities Education Act (IDEA)
      j. Describe the eligibility criteria for services described in the Individuals with Disabilities Education Act (IDEA)
      k. Know the guidelines for school disciplinary procedures described in the Individuals with Disabilities Education Act (IDEA)
      l. Know the application of the Americans with Disabilities Act and special education law to higher education

B. Educational administration and processes
   1. Understand the role of a pediatrician in initiating and participating within a school evaluation of a child with learning problems
   2. Know the purpose and limitations of an individualized education program
   3. Know the differences between special education services provided under an individualized education program and those provided under Section 504 of the Rehabilitation Act

C. Health care structures and processes
   1. Understand the impact of various healthcare financing arrangements on the quality of services for children with special healthcare needs
   2. Appreciate the financial incentive for insurance companies to discourage enrollment of children with special healthcare needs
   3. Know the ethical implications of financial disincentives to specialty referrals within managed care systems and its differential impact on children with special healthcare needs
   4. Be familiar with issues relating to the impact of mental health carve-outs on the quality of and access to mental health services for children

DBP BOSTON MEDICAL CENTER
CHRISTINE MCGIVNEY
MEDIATRIX MBAMALU
ANA TREADAWAY

22. Law, Policy, and Ethics

A. Legal rights and processes - Individuals with disabilities

a. Know the criteria for early intervention or special education for children from birth through 21 years of age
https://www.understood.org

- Early intervention is for children birth to age three with a developmental delay or specific condition predisposing the child to delay. Specific eligibility criteria vary by state. Select states provide services for at risk children (e.g. low birth weight, substance exposure).
- Children age 3-21 years qualify for special education if they are determined to have a disability and require special education to progress in school as a result of that disability.
- IDEA defines 13 categories of disabilities. Categories of disability are: autism, deaf-blindness, deafness, emotional disturbance, hearing impairment, multiple disabilities, orthopedic impairment, other health impairment (ADHD included), specific learning disability, speech or language impairment, traumatic brain injury, visual impairment (including blindness).

b. Understand the different implications of the Americans with Disabilities Act (ADA) for public, private, and parochial schools
https://www.understood.org

- Child Find is a federal law requiring public school districts to identify and evaluate students suspected of having disabilities. Child Find applies to all children; including those who attend charter school, private school or who are homeschooled.
- The evaluation is paid for by the public school district
- “Equitable services” refers to services provided by public funding set aside specifically for students with disabilities whose parents put them in private school.
- An Individual Service Plan may be developed. This is similar to an IEP, potentially less extensive.
- Teacher qualifications and hiring standards can differ in private schools.

c. Know the components of an Individual Family Service

Plan (IFSP)

https://www.specialeducationguide.com/

- People and organizations involved (name of service coordinator)
- Current levels of functioning and need
- Family information
- Services that will be provided
- Outcome goals
- When and where the services will be provided
- Duration and number of sessions
- Who will pay
- Transition plan out of early intervention

d. Understand parents' rights to participate in Special Education decisions as described in the Individuals with Disabilities Act (IDEA)

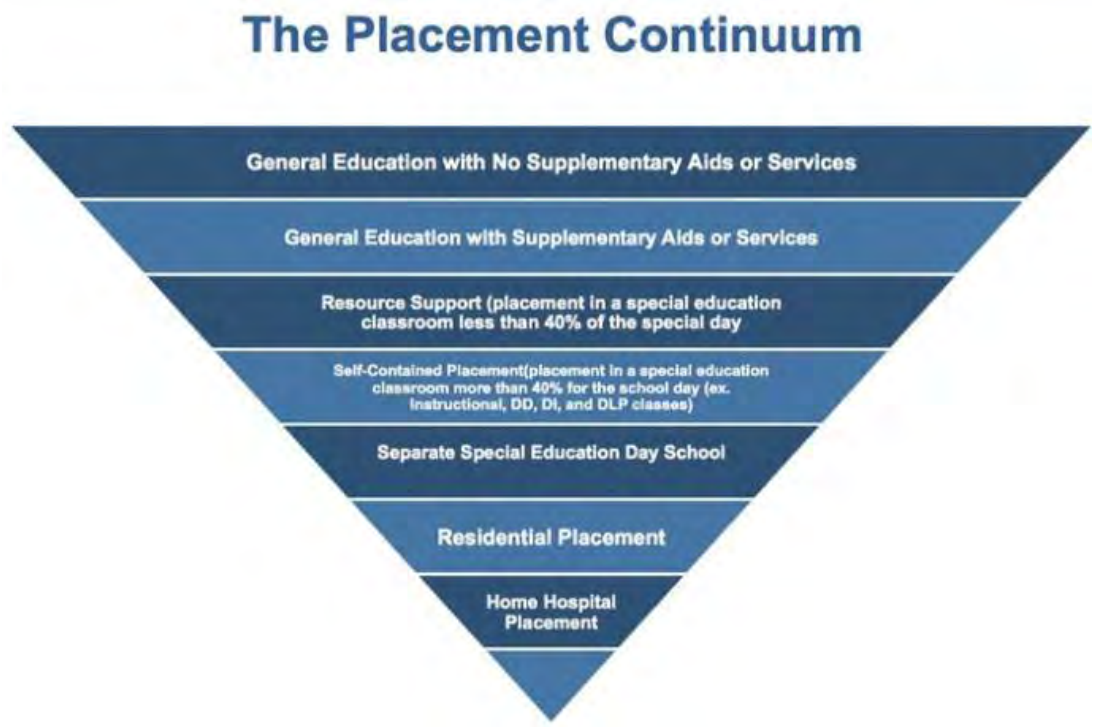
http://www.ldonline.org/article/6086/

- IDEA requires the public school to provide impartial hearings when a parent disagrees with the identification, evaluation, or placement of the student
- Written parental consent is required before a special education evaluation can occur
- Stay-put provisions dictate that the student's current IEP and placement continue to be implemented until all proceedings are resolved
- Parents are required to receive 10 days notice prior to any change in placement

e. Understand how to apply the concept of least restrictive environment (LRE) to designing an educational program for a child with a disability

http://www.fortelawgroup.com/least-restrictive-environment-lre-mean/

- "To the maximum extent appropriate, children with disabilities, including children in public or private institutions or other care facilities, are educated with children who are not disabled, and special classes, separate schooling, or other removal of children with disabilities from the regular educational environment occurs only when the nature or severity of the disability of a child is such that education in regular classes with the use of supplementary aids and services cannot be achieved satisfactorily."
- Intent of the LRE provision is to ensure that a child receiving special education be included in the general education classroom environment as often and frequent as possible.



f. Understand the importance to special education of the related concepts: natural environments, inclusion, and mainstreaming
https://www.parentcenterhub.org/naturalenvironments/

https://www.specialeducationguide.com/

https://www.theedadvocate.org/

- *Natural environments* are defined as settings that are natural or typical for a same-aged infant or toddler without a disability. Natural environments may include the home or community settings, and must be consistent with the provisions of §303.126.
- EI services should be provided in natural environments or in settings other than the natural environment that are most appropriate, as determined by the parent and the IFSP Team, only when early intervention services cannot be achieved satisfactorily in a natural environment.
- IDEA dictates that "students with special needs have the right to receive necessary curricular adaptations. Adaptations include accommodations and modifications. Students who receive accommodations are held to the same academic expectations as their general ed classmates. Alternatively, modifications entail making changes that lower these expectations. Curricular adaptations vary based upon each learner's individual needs."
- The inclusion model is a classroom setting in which students with disabilities are taught alongside their non-disabled peers in a general

education classroom. The student is included in the general education class for the entire day. A special education teacher works with the special needs children in the classroom during the day.

- In mainstreaming, students with special needs are taught in a special education classroom and join a general education classroom for specific classes as skills permit.

g. Appreciate the right to a publicly funded evaluation and special education services for all children in need, including those attending private and parochial schools

- See section b
- An Independent Educational Evaluation (IEE) completed by a qualified examiner who is not employed by the school district.
- Parents have the right to request an IEE at public expense if they disagree with the school district's evaluation of their child.
- Parents can request an IEE even without a school evaluation.
- If parents request an IEE, the school district must provide information on where to obtain an IEE and state requirements regarding IEEs.

h. Recognize the necessity to provide medical services within the school setting if required by a child in order to participate in an educational program

https://www.parentcenterhub.org/

- Medical services are defined in IDEA as services provided by a licensed physician to determine a child's medically related disability that results in the child's need for special education and related services.
- FAPE includes special education and related services. Medical services fall under the category of related services when:
  - provided by a licensed medical provider
  - for diagnostic or evaluation purposes only

i. Understand the components of IDEA

https://www.smartkidswithld.org/

- Individualized Education Plan (IEP)
  - The IEP is an education document for students from age 3 - 22 years detailing special education and related services in schools
- Free and Appropriate Public Education (FAPE)
  - FAPE is defined as special education and related services. Special education is "specially designed instruction at no cost to

the parents, to meet the unique needs of the child with a disability..." and related services are services provided if required for the student to benefit from the specifically designed instruction.

- Least Restrictive Environment (LRE)
  - “To the maximum extent appropriate, children with disabilities, including children in public or private institutions or other care facilities, are educated with children who are not disabled, and special classes, separate schooling, or other removal of children with disabilities from the regular educational environment occurs only when the nature or severity of the disability of a child is such that education in regular classes with the use of supplementary aids and services cannot be achieved satisfactorily.”
- Appropriate Evaluation
  - A multidisciplinary team evaluates, documents and jointly considers placement of the student only after parental consent is provided. Reevaluations are conducted at least every 3 years. An IEP meeting is required before any change in placement can be made.
- Parent and Teacher Participation
- Procedural Safeguards

j. Describe the eligibility criteria for services described in IDEA

- See section a
- A child should be eligible for Special Education Services, if they have a disability and the disability causes the child to be unable to progress effectively in regular education. In other words, the child requires specially designed instruction to make progress or requires services in order to access the general curriculum?
- At age 3, supports and services change as eligible children transition from part C (early intervention) to Part B (special education)

k. Know the guidelines for school disciplinary procedures described in IDEA

http://www.uky.edu/

- 9 mandates detailed in Part B of IDEA from §§300.530 through 300.536
- What is the authority of school personnel regarding the discipline of students with disabilities?
  - School personnel can discipline students with disabilities for the same amount of time and with the same disciplinary methods as students without disabilities so long as a change of placement

does not occur and the behavior is not a manifestation of their disability. A student may be removed for up to 10 school days for each separate act of misconduct without a change of placement occurring.

    - A student can also be placed in an alternative educational setting for up to 45 days for violations related to possessionof weapons, possession or use of illegal drugs, or the sale or solicitation of a controlled substance on school grounds or at school functions. The alternative educational setting is selected by the IEP Team so that the student can continue to progress in the general curriculum. In addition, the alternative educational setting should be selected so that the student can receive services and modifications required to meet goals stated in the student's IEP as well as to address the behavior that resulted in the disciplinary action.
- What is the authority of a hearing officer in the placement of a student in an alternative educational settings?
    - At the request of a school, a hearing officer can remove a student from his or her current placement if remaining in the setting would likely result in harm to the student or others. The student can be removed for up to 45 days; however, the school can request an extension of this placement if the return of the student to the educational placement is still considered a danger.
    - The school must present substantial evidence of possible injury to the student or others and of reasonable efforts to minimize this harm. The hearing officer must consider the appropriateness of the student's current placement prior to removal of the student to an alternative education setting selected by the student's IEP Team.
    - The alternative educational setting selected must allow the student to continue to make progress in the general curriculum and to receive services and modifications required to achieve goals stated in the student's IEP as well as to address the behavior that resulted in the disciplinary action.
- When does a change of placement occur?
    - A student's educational placement is considered changed for disciplinary reason when the student is removed for more than 10 consecutive school days for a violation of school rules.
    - A change in placement also occurs when the student is removed for several incidents that constitute a pattern based on the total number of days - more than 10 school days in a year - and the length of each removal, the amount of time the student has been removed, or the proximity of the removals.
    - For example, a student suspended and/or placed in in-school suspension for disrespect to school personnel on 4 occasions for 12 school days within a 60 day time period would be a pattern that constitutes a change in placement.

- When is the school required to provide services to a student removed for disciplinary reasons?
    - The school is not required to provide services for the first 10 days that a student is removed in a school year.
    - During any subsequent removals for 10 days or less, schools must provide the services required to enable a student to make progress in the general curriculum and on goals stated in the student's IEP. The decision on required services is made by the school personnel in consultation with the student's special education teacher.
    - During any long-term removal for behavior that is not a manifestation of the student's disability, schools must provide the services required to enable a student to make progress in the general curriculum and achieve goals stated in the student's IEP. The decision on required services is made by the student's IEP Team.
- When must a functional assessment be conducted and a behavioral intervention plan developed?
    - A functional assessment is required for possession of weapons, possession or use of illegal drugs, or the sale or solicitation of a controlled substance on school grounds or at school functions. It also is required the first time a student is removed from his or her current placement for more than 10 school days in a school year or when a removal will result in a change in placement.
    - A meeting of the student's IEP Team must be convened no later than 10 business days after the disciplinary action to develop a behavior assessment plan. The IEP Team must conduct a functional assessment and develop a behavior intervention plan to reduce the likelihood of the reoccurrence of the student's behavior that resulted in disciplinary actions.
    - For a student with a behavior intervention plan, the IEP Team must have a meeting to review the plan only if it is the first time the student is removed for more than 10 days in a school year or when the removal will result in a change of placement. For other disciplinary actions, the IEP Team must review the plan and its implementation to determine if modifications are necessary. If one or more members of the team desires changes in the behavior intervention plan, the IEP Team must have a meeting to modify it.
- When and how is a manifestation determination review conducted?
    - A manifestation determination review is conducted to determine whether the behavior that resulted in the disciplinary action is a manifestation of the student's disability. A review must be conducted for the removal of a student that constitutes a change of placement. The parent must receive notification of this decision no later than the date the decision to take that action is

made. The review must be conducted no later than 10 school days after the decision to take the action is made.
    - The review is conducted by the IEP Team and other qualified personnel. As part of the review, the IEP Team must use evaluation and diagnostic results, information from the parents, observations of the student, and the student's IEP and current placement to determine whether the behavior subject to disciplinary action is a manifestation of the student's disability.
    - The team also must determine whether the student's IEP and current placement were appropriate and whether services and intervention strategies were implemented consistently with the student's IEP and placement. They must determine whether the student's disability impaired his or her ability to understand the impact and consequences of the behavior and control the behavior.
- What happens if a parent appeals the decision of the school personnel?
    - A parent can request a hearing if he or she disagrees with the decision regarding placement or a manifestation determination review. The state or school district must arrange a due process hearing with a hearing officer.
    - If the parent disagrees about a manifestation determination, a hearing officer must determine whether the behavior is a manifestation of the student's disability using the same procedures and information used by the IEP Team during the manifestation determination review.
    - If the parent disagrees with a placement decision, a hearing officer must be presented with substantial evidence of possible injury to the student or others and of reasonable efforts to minimize this harm. The appropriateness of the student's current placement must be evaluated as well as the alternative educational setting selected to allow the student to continue in the general curriculum and achieve goals stated in the student's IEP.
    - During the process, the student is to remain in the alternative education setting until a decision is made or expiration of the time period for removal. If the time period expires, the student is to be returned to the current placement stated in the student's IEP. If the current placement is deemed dangerous, the school district can request an expedited hearing to be conducted within 45 days.
- What protection do children who are not eligible for special education have under IDEA?
    - A student who is not eligible for special education and has violated school conduct can claim protection under IDEA if the school district had knowledge that child had a disability prior to

the behavior that resulted in the disciplinary action. A school district is considered knowledgeable if the parent expressed concern in writing (or orally if the parent does not know how to write or has a disability that prevents a written statement) to school personnel that the child needed special education and related services; the behavior of the child demonstrated the need for these services; the parent has requested evaluation; or school personnel have expressed concerns about the behavior of the child to personnel involved in the established child find or special education referral system.

- If an evaluation has been conducted and it was determined that the student was not eligible for special education or it has been determined that an evaluation is not warranted, a parent cannot claim protection for their child under IDEA.
- If a request is made for an evaluation to determine eligibility for special education services during the time period of the disciplinary action, the evaluation is conducted in an expedited manner. During the evaluation, the student remains in the educational setting selected by the school. [For more information on evaluation and eligibility, view the IDEA tutorial on evaluation.]

- What should school personnel do if a student with disabilities commits a crime?
  - If a student with a disability commits a crime, school personnel should report it to the appropriate authorities. The law enforcement and judicial authorities should apply federal and state law to these crimes. Copies of the student's special education and disciplinary records should be transmitted, to the extent possible under the Family Educational Rights and Privacy Act, for consideration to the appropriate authorities when reporting a crime.

I. Know the application of ADA and special education law to higher education

- https://www.pacer.org/ Title II of the ADA covers state-funded programs such as universities, community colleges, and career and technical education programs. Title III of the ADA covers private colleges, and vocational programs. If a postsecondary education program receives federal funding, regardless of whether it is a private or public program, it is also covered by the regulations of Section 504 of the Rehabilitation Act which requires the school to make programs accessible to qualified students with disabilities.

- The ADA is a civil rights law giving protections to all individuals with disabilities in the US in various aspects of life. Title II of the ADA prohibits discrimination by state and local governments, including public schools

B. Educational administration and processes

1. Understand the role of a pediatrician in initiating and participating within a school evaluation of a child with learning problems

https://pediatrics.aappublications.org/content/144/4/e20192520

https://www.aappublications.org/news/2017/01/09/IEP010917

https://pediatrics.aappublications.org/content/104/1/124.long

- Pediatrician's office often is the first stop for families seeking help with school issues. Navigating the special education system, understanding their rights and collaborating with school officials to come up with a plan can be a confusing for parents.
- The most important and best role for the pediatrician is to be an advocate for the child.
- Several roles for the pediatrician exist under IDEA. All pediatricians should ensure that in their practices, every child with a disability has access to the following services:
    - A medical home with care that is accessible, continuous, comprehensive, family-centered, coordinated, and compassionate.
    - Screening, surveillance, and diagnosis. The pediatrician should screen all children from the first encounter, checking for risk or existence of a disability or developmental delay.
    - Pediatricians are in key positions to identify at the earliest possible age those children who may benefit from services under IDEA.
    - Pediatricians should provide screening and surveillance using a combination of methods best designed to take advantage of multiple sources of information.
    - Referral. The pediatrician should be knowledgeable about the referral process to early intervention, parents' right for multidisciplinary team evaluation by the school- or state-designated agency if a disabling condition may be present.
    - Emotional and social support and practical advice to parents.
    - Diagnosis and eligibility. For early intervention, the pediatrician has an important role in the identification of children with established delays and in the diagnosis of conditions with a high probability of developmental delay, which will qualify a child for this program.
    - Participation in assessment. A child identified through screening or

observation as meeting the definition for developmental delay should receive a comprehensive multidisciplinary assessment. The pediatrician has an important role as a referral source or, if more extensive participation is elected, as a member of the multidisciplinary team.

- Counsel and advice. During the assessment process, families will need a knowledgeable person for medical advice and counsel. Pediatricians can alert parents to the benefits of a pre-IEP conference; of their right to sign the IEP only when they are comfortable with the recommendations.
- Creation of the IEP. Pediatricians should be consulted by the assessment team when these documents are created. Such consultation is vital to preparing an appropriate and effective plan. The pediatrician should review the plan developed, counsel the family, and comment on health-related issues as needed.
- The pediatrician should determine if the health-related services proposed are appropriate and sufficiently comprehensive.
- Coordinated medical services. When medical services are part of the IEP they should be conducted by the primary care pediatrician or an appropriate pediatric subspecialist.
- Medical services and communication should be coordinated by the primary care pediatrician or his or her designee in those cases in which the children have complex medical needs involving several physicians or centers.
- Special education personnel should be made aware of the restrictions of health care insurance including limited referral options and the role of the primary physician as “the gatekeeper” in some programs.
- Advocacy. Pediatricians have many local and state opportunities to serve as knowledgeable, thoughtful advocates for improved community and educational services for children with disabilities.
- Active participation in the local or state early intervention interagency council, consulting with the local school system or state department of education, or becoming a school board member.

2. Know the purpose and limitations of an individualized education program

- When child turns 3 years of age, services and supports change from Early Intervention to Special Education as per IDEA.
- The Individualized Family Service Plan (IFSP) used by Early Intervention is then replaced by an Individualized Education Program or IEP.
- To start the process:
  - Children enrolled in Early Intervention services, the EI caseworker will contact the school at least 3 months prior to the child turning 3 years old.
  - Children older than 3 y/o will require a letter to the Special Education Department of their School District requesting evaluation for special education.
  - Some School Districts require the family to register the child prior to start the IEP process while others require the family to go through the eligibility determination process before registering the child. Contact your district

and ask.

- The IEP is a multidisciplinary, team-developed plan required for every child receiving special education services under part B of the IDEA.
    - As per Public Law 108-144, legally required components of the IEP includeChild's present level of performance
    - Measurable annual goals
    - How the child's progress will be measured and when said progress should be reported
    - Special education and related services and supplementary aids and services, to be provided to the child
    - Program modifications or supports for school personnel that will be provided to the child
    - The extent to which the child will not participate with nondisabled children in the regular classroom (meaning the time he spends in a regular classroom)
    - Individual appropriate accommodations that are necessary to measure the academic achievement and functional performance of the child on state and district wide assessments.
- IEP should address students' individual strengths and needs and include a timeline for meeting objectives. However, research indicates that sometimes:
    - IEPs may be lacking in their consistency with recommended practice. In particular, IEPs have tended to include inadequate descriptions of present performance, goals that were not specific and expectations that were unrealistic and misaligned with children's abilities.
    - Contained placement recommendations that appeared to be based upon eligibility criteria rather than performance.
- Additionally, common concerns from parents include difficulties in:
    - Being viewed as equals in making educational decisions regarding their children
    - IEP objectives' being properly followed in the classroom
    - Being fully informed about special education law and their rights
    - Classroom practices such as ineffective discipline programs and inappropriate placement decisions.

3. Know the differences between special education services provided under an individualized education program and those provided under Section 504 of the Rehabilitation Act

https://www.understood.org/en/school-learning/special-services/504-plan/the-difference-between-ieps-and-504-plans

http://www.clcm.org/504_Plans.pdf

- IEPs and 504 plans are free to families and provide a blueprint of formal help for a child but in different ways.
- An IEP falls under IDEA, a federal special education law. IEPs are available to children whose educational performance and/or ability to learn in a general K-12 curriculum are affected by at least one of 12 disabilities.
- An IEP provides individualized special education and related services. It may include accommodations (changes to the learning environment) and modifications to what the child is expected to know. It also outlines how a child will participate in standardized testing and will be included in general education classes and school activities. A team of school staff, parents and possibly others create the written plan that includes goals, progress tracking and services from specialists like occupational and speech therapists.
- A 504 plan is covered by Section 504 of the federal civil rights law to stop discrimination against public school students with a disability. Eligibility is based on a broader definition of disability than IDEA, and the disability must substantially interfere with a student's ability to learn in a general education classroom. The plan does not have to be written and does not include special education services, but unlike an IEP does involve all levels of education including college. It typically includes accommodations such as extended time on tests, extra textbooks for home, preferred seating in a classroom and breaks during class.
- Unlike a special education student, a student with a 504 plan **is able to make effective progress in school without the need for <u>specialized instruction and/or related services</u>**. However, he or she requires accommodations in order to gain equal access to the instruction and/or the facility.
- A student's 504 plan will provide accommodations that allow a student with an impaired major life activity to have the same level of access to the instruction, school activities, and the school building as students without disabilities.
- Some of these concepts are necessary to understand the process of accessing special education or a 504 plan:

    - Individualized Education Plan: The term individualized education program or IEP means a written statement for each child with a disability that is developed, reviewed, and revised in a meeting in accordance with Sec. 300.320 through 300.324
    - Special Education: means specially designed instruction to meet the unique needs of the eligible student or related services necessary to access the general curriculum.
    - Free and Appropriate Public Education: Once child is eligible, s/he is entitled to FAPE which includes:
        - Specially designed instruction
        - Accommodations
        - Related services – like Counseling, Behavioral consult, OT, Transition and transportation.
        - Placement

        - "Appropriate" education means an education that is comparable to the education provided to students without disabilities. This may include regular or special education services. Students can receive related services under Section 504 even if they are not provided with special education services.
        - Least Restrictive Environment: is part of the Individuals with Disabilities Education Act (IDEA). IDEA says that children who receive special education should learn in the **least restrictive environment**. This means they should spend as much time as possible with peers who do not receive special education.
    - "Stay put rights": Protect the student while a dispute regarding placement or total rejection of an IEP is solved. Student continues receiving services as listed in previous IEP. If placement is the issue, student will continue placed in their previous school/classroom.

Similarities between 504 Plan and IEP	Differences between 504 Plan and IEP
• Requires an evaluation	• Does not require progress reporting
• Accommodation on standard testing	• Limited discipline protections
• Related services to assist in accessing regular education	• Does not apply for "Stay Put rights"
• Both can technically provide specialized instruction, but because no federal funding accompanies a 504, in practice schools use a 504 only for accommodations, modifications, (not for specialized instruction, related services, etc.)	• Section 504 protections follows the child after s/he leaves the public school system. IDEA (IEP) does not. (504's are written per environment. Colleges and secondary school write the 504 plan, not the high school. They can use 504/IEP as input.) • 504 is a Civil rights law, under ADA (American with Disabilities Act). An IEP (Individual education Program) falls under IDEA (Individual with Disabilities Education Act) and is an Educational Law.

Differences between Section 504 and IDEA Laws
• Section 504 does not require written plans (most school have created their own forms).
• The school does not have to invite the parent to the meeting when the 504 plan is developed. The school must notify the parent that a 504 plan was developed
• Section 504 has fewer procedural safeguards to protect the parent and child.

C. Health care structures and processes

1. Understand the impact of various healthcare financing arrangements on the quality of services for children with special healthcare needs

- CYSHCN have had better access to public insurance, especially because more are enrolled in SSI[1]
- CYSHCN have higher rates of utilization and expenditures than other children do[1]
- In general, compared with most private health insurance plans, Medicaid offers relatively generous benefits for people with disabilities, including much better long-term care benefits. [1]
- Medicaid contracts and managed care plans may ration the provision of specialized therapies for people with disabilities (e.g., physical and occupational therapy, respiratory therapy, speech and language services), given the limited evidence for their efficacy[1].
- In Medicaid and private insurance contracts, “medical necessity”—the basis for determining whether to pay for a particular service—also may limit access to certain specialized therapies, especially when no trials of their efficacy have been conducted[1]

2. Appreciate the financial incentive for insurance companies to discourage enrollment of children with special healthcare needs

- CSHCN are at increased risk for chronic physical, developmental, behavioral or emotional conditions and who also require health and related services of a type or amount beyond that required by children generally.”[2]
- Nearly 20% of U.S. children under age 18 years of age have a special health care need.

- One in five U.S. families have a child with a special health care need.

3. Know the ethical implications of financial disincentives to specialty referrals within managed care systems and its differential impact on children with special healthcare needs

- Impacts access to referral services; particularly for CSHCN whose needs exceed their use of services due to the high need for multiple specialty care
- They are twice as likely to delay care because of cost and to have an unmet health need, most of which are for referral services
- Children and youth with special health care needs and their families often need services from multiple systems – health care, public health, education, mental health, and social services.

4. Be familiar with issues relating to the impact of mental health carve-outs on the quality of and access to mental health services for children

- The carve-out policy increased the number of children receiving Title V-authorized services although cost intensity per Title V recipient generally declined[4]
- The carve-out policy increased identification of children with special health care needs.

[1]https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2690116/, Accessed June/2020

[2]https://www.cdc.gov/childrenindisasters/children-with-special-healthcare-needs.html,June/2020

[3]E.J. Silver, R.E. Stein: Access to care, unmet health needs, and poverty status among children with and without chronic conditions. Ambul Pediatr. 1:314-320 2001 11888421

[4] Moira Inkelas: Incentives in a Medicaid Carve-Out: Impact on Children with Special Health Care Needs. Health Serv Res. 2005 Feb; 40(1): 79–100

Content Category 22- Law, Policy, & Ethics- Parts D&E

ABP Content Specs for Developmental-Behavioral Pediatrics Fellow Collaboration Project

Prepared by: Beatrice Egboh, MD, University of Nebraska, DBP Fellow

Reviewed by Howard Needelman, MD, University of Nebraska, DBP Fellowship Director

Final Version/Faculty Reviewed

22. Law, Policy, and Ethics.

D. Advocacy (See also XVIII.D.)
1. Know how to advocate within a school system for a child with learning problems, utilizing both informal means and due process
2. Know how to advocate for children with disabilities at the local, state, and federal level

E. Ethics
1. Treatment
a. Understand the ethics of participation of the competent adolescent patient in decisions to withhold treatment in serious, life- threatening medical conditions
b. Understand the appropriateness of psychiatric hospitalization of a young adolescent who the clinician feels is at serious risk of self-harm when the parent and child do not agree to hospitalization
c. Understand the legal and ethical implications of drug screening in adolescents
d. Understand the ethical implications of potential financial conflicts of interest of treating physicians and how to avoid and manage such potential conflicts
e. Know the limits of confidentiality of disclosures of child patients to physicians

ADVOCACY.

1. **How to Advocate Within A School System for A Child with Learning Problems, Utilizing Both Informal Means and Due Process.**
    - Learning problems in school-age children may be due to a specific learning disorder or associated with other disabilities.
    - Access to private (independent) psychoeducational testing is often hindered by the family's financial limitations, lack of eligibility in most health care insurance plans, or the pediatrician's lack of familiarity with available community resources.
    - The IDEA Act states that 'A free appropriate public education is available to all children with disabilities residing in the state between the ages of 3 and 21, including children with disabilities who are suspended or expelled from school' .[1]
    - All states are required to conduct "Child Find" activities to identify and evaluate all children with disabilities who may need special education whether the children are in private, parochial, or public schools.
    - Parents are encouraged to be advocates for their children. When they have concerns about their child having a learning disability, they can make a verbal or written request for an evaluation for special education through the child's teacher, school administrator, or the school district office.
    - A teacher can also request for an IEP evaluation but, Parental consent is needed before a child can be evaluated.[2]
    - Parents should receive a response within 15 days of the request, and the school has 50 days to complete an assessment and hold a meeting to go over the results.
    - The minimum districts will assess for is academic achievement and cognition. A provider can ask for an assessment in other potential areas of need such as language, motor, sensory processing, social/emotional, and health.
    - Parents and providers can informally advocate for the child by communicating in a positive tone to teachers and school to ensure the child succeeds.
    - Due process: is a formal way of resolving a dispute related specifically to a student's education at school, is a part of IDEA. This process begins with a parent's submission of a written complaint to the school, after which a resolution session occurs within 15 days. This session involves the individualized education program team, the parent, and a school district representative who has decision-making authority working together to find a resolution. Either party may waive this meeting, or mediation may be sought. If no resolution is found, then a due process hearing will be scheduled. Each party provides evidence and witnesses, and a hearing officer offers a solution, which may be further disputed in court. Should the parent be the prevailing party, he or she may recoup payment for attorneys' fees, within reasonable limits.
    - Parents are advised to review any document before signing. They can ask for modifications, if an agreement cannot be reached, they can request for a mediator.
    - Parents who need help navigating the school system bureaucracy and obtaining appropriate services may wish to turn to an education lawyer or, when that's not an option, an advocacy agency.[3]

- As advocates for children, physicians can join with parents to monitor their state education laws and policies to see that these conform to IDEA.[4]
- IEP should have measurable goals and state what the child should accomplish in one year. This provides a way to determine if the child is receiving sufficient services and making progress.
- Progress monitoring is used to assess a child's academic progress on IEP goals and evaluate the effectiveness of instruction. The teacher evaluates a child's progress in each goal and tells what the child has learned.
- Parents can request an IEP meeting at any time to review goals, set new goals, or discuss new evaluations. The entire IEP team should meet and review goals at least once a year.

1. **How to Advocate for Children with Disabilities at The Local, State, And Federal Level.**

   A pediatrician can advocate for children with disabilities at the local, state, and federal levels.

   This process starts with identifying issues that affect the patient, connecting families with organizations that share a common interest, setting realistic goals, making the case with state and federal elected officials, and informing them about issues important to children with disabilities.

   - Advocacy, broadly, is speaking out on behalf of one's patients. There are four levels of advocacy: individual, community, state, and federal. Advocacy at the community, state, and federal levels allows pediatricians to expand their efforts beyond treating individual children to support the health and well-being of all children. Often, advocacy starts when a pediatrician identifies an issue that affects his or her patients.
   - The most effective strategy pediatricians can use to advocate for early intervention funding is to meet with legislators directly to share personal experiences of how early intervention has benefited their patients. Effective advocacy methods combine personal experiences with a direct, specific contact. Personal contact and communications, including visits, public testimony, letters, phone calls, e-mails, and petitions, can influence decision-makers. The more direct and personal the contact, the more meaningful and potentially effective it will be. For example, a handwritten letter is more meaningful than is a petition or form letter. A letter to the editor may highlight the importance of an issue to the community, but it is less direct than contacting legislators personally.
   - The advocacy process will be different according to the scope of the issue, but there are some basic steps:
   - Identifying an issue: Individual patients' problems are often part of a broader issue that could be addressed with policy changes.
   - Finding partners: Connecting with organizations with similar interests and goals can broaden support for the issue.
   - Setting goals: Goals should be realistic and attainable; for large-scale objectives, focusing on incremental, short-term goals may be a more appropriate way of making strides toward the broader goal

- Finding the decision-makers: This may include community leaders and elected or appointed officials.
- Making the case: Personal communication puts a face on the issue; it is vital to include a direct appeal so that decision-makers know how they can support the issue
- Sustaining efforts across time: It can take time to change policy; repeated communications can inspire the decision-maker to act.

The American Academy of Pediatrics (AAP) has a long history of advocating on behalf of all children through its work with governments, communities, and other national organizations. Extensive resources are available online through the AAP to support individual providers in their advocacy efforts.

ETHICS.

Ethics of Participation of The Competent Adolescent Patient in Decisions to Withhold Treatment in Serious, Life-Threatening Medical Conditions.

Individuals that have attained their age of majority, usually at 18 years, are in most countries legally considered as competent (unless they suffer from severe psychological disturbance or intellectual impairments). Through statute or common law, however, most states permit mature minors to consent to some treatments. Minor adolescents or even children can be considered competent, if, in a given situation, their health care provider and health care team deem them so.[5]

The capacity for autonomous decision-making is not a fixed, predetermined state but is mainly dependent on how the adults and professionals in charge address the various aspects of the adolescent's condition, development, and circumstances.[5]

In general, adolescents should not be allowed to refuse life-saving treatment for conditions with a good prognosis, even when the parents agree.

Refusal of life-sustaining therapy by an adolescent with a life-threatening illness should be seriously considered by both the parents and the health care team when the likelihood of a good outcome is low. Adolescents in such situations often have an enhanced capacity for decision-making.

If the adolescent's decision is an ethically permissible treatment option, the pediatrician should advocate for it in collaboration with the health care team and the family to resolve any conflicts.

Components to consider:

- Legal – consent/ assent: Even when the child is not legally able to give consent, his assent should be obtained.
- Align with the United Nation's convention on the rights of a child (right of adolescent to be heard) [6]
- Specific autonomy to treatment (sexual health/ reproductive health)
- Developmental view in decision making- recognizing adolescent decision making differs from adult (development of prefrontal cortex)
- The capacity to understand the short- and long-term aspects and consequences of a decision depends on the situation.[5]
- Adolescent reacting in different manners depending on their emotional state
- Establishing trust with the adolescent.

- Adolescent's experience and understanding of the condition, options of treatment, and implications of withholding treatment.
- Empathetic and respectful evaluation adolescent and assessment of the adolescent's understanding.
- The input of relatives to guide adolescent's decision capabilities.
- The urgency of the decision.
- Escalation: involving stakeholders; parents/ relatives, ethics committee, use of courts.
- Three situations in which a minor has the legal authority to make medical decisions for himself or herself are emancipation, the mature minor exception, and exceptions based on specific medical conditions. Laws vary among states as to when a minor has the authority to consent to or refuse treatment.
- Legally emancipated: the right to give consent or refuse medical treatment. The definition of an emancipated minor varies according to state. In most states, minors who are married, economically self-supporting, and living separately from their parents, as well as those on active duty in the military, are considered emancipated. In some states, a pregnant minor or a minor who has a child may be considered emancipated. Some states might require a court order to declare a minor emancipated.
- A mature minor is one who has sufficient maturity and intelligence to understand the benefits, risks, and alternatives of the proposed treatment and to use that information to make a voluntary medical decision. A mature minor is usually 14 years old or older (depending on the individual's state law, a mature minor is between 14 and 16). In some states, a physician can determine that the minor has this capacity; however, in most states, a judicial determination is required to allow an adolescent to make medical decisions as a mature minor.
- In most states, minors can consent to treatment for specific medical conditions without a parent or guardian. However, specific conditions and ages vary according to state. All states allow adolescents the ability to give consent regarding health care needs related to sexual activity, including contraceptive services, prenatal care, and treatment of sexually transmitted infections. In many states, adolescents can also give consent for mental health and substance abuse treatment services. However, not all states protect the confidentiality of adolescents who consent to these treatments.

Appropriateness of Psychiatric Hospitalization of a Young Adolescent Whom the Clinician Feels Is at Serious Risk of Self-Harm When the Parent and Child Do Not Agree to Hospitalization.

According to the Center for Disease Control (CDC), suicide is the 2nd leading cause of death among 15-19-year-olds. Thus, inpatient hospitalization may be necessary to provide one on one monitoring and ensure a safe environment without access to self-harm means while providing therapies and treatment.

Legal and Ethical Implications of Drug Screening in Adolescents.

- The AAP believes that adolescents should not be drug tested without their consent and has strong reservations about testing at school and home.
- Drug testing may be invasive, yield only limited information, and results are easily misinterpreted, and a negative test would not eliminate concern for substance use problems.
- Adolescents are less likely to seek care if they perceive that health care services are not confidential. This may also result in mistrust and conflict within the family.
- Some state laws prohibit physician disclosure of drug screens to parents without consent from the adolescent.
- Random drug screening is sometimes used as a deterrent for juveniles in the probation system.

Limits of Confidentiality of Disclosures of Child Patients to Physicians.
Physicians should encourage minors to involve parents when they request for confidentiality.
These can be breached when-

- Reporting child abuse.
- Threatens to inflict harm to self or others.
- Diagnosis of Sexually transmitted diseases such as gonorrhea, chlamydia, syphilis, and HIV that are in the category of reportable diseases. It is required that positive results are reported to local departments of health and, ultimately, the centers for disease control and prevention (CDC) and informing sexual partners.
- Gunshot wounds and knife wounds should be reported as required by appropriate authorities.

Exceptions for certain conditions regarding when adolescents can consent for care and thus control their own medical information (but is encouraged to involve parents).

- STI, birth control, pregnancy, substance abuse treatment.

Ethical Implications of Potential Financial Conflicts of Interest of Treating Physicians and How to Avoid and Manage Such Potential Conflicts.
Professional courtesy (no charge to patient or insurance for care) is a long-standing tradition in medicine, but it is not an ethical requirement. Physicians should use their judgment in deciding to reduce or wave payment. This tradition has virtually disappeared with the growth of 3rd party paying for most services with required copays.
Accepting insurance and waving copayment may violate some insurance policies.
(Code of medical ethics 6:13).

- A 2009 Institute of Medicine report defined conflict of interest (COI) as "A set of circumstances that create a risk that professional judgment or action regarding a primary interest will be unduly influenced by a secondary interest." Primary interests refer to research activity or reporting, patient welfare, and educational efforts. Secondary interests refer to influences that may bias a physician and therefore compromise the integrity of the primary interest. Financial incentives or influences were the first of the secondary interests addressed. However, influences such as promotion, reputation, position or employment, and even

personal opinion or a family member or friend close to the individual can introduce bias and cloud judgment.

- Variability exists in the interpretation of COI, and policies for documenting, reporting, or reviewing potential COI are still being established. Two different strategies for the management of COI are proffered: a review of declared COI by a committee with authority to limit the scope of activity or influence of the individual who disclosed such a conflict, or review of potential COI in advance to circumvent the opportunity for conflict. In the latter scenario, committed individuals such as institutional review board members may review proposed studies for recruitment, methods, and biostatistical protocols to point out elements that may introduce bias and require changes to the protocol before it is approved. Similarly, educational committees may review industry-sponsored activities that may introduce bias.

Strategies to avoid the ethical conundrum of COI have been suggested by the Institute of Medicine and other authorities; these strategies suggest institutional, individual, and federal procedures for identifying and managing potential COIs.

At the institutional level:

- Creation of an institutional committee to outline guidelines for declaring, reviewing, and mediating COI
- Creation of committees to review study protocols submitted for institutional review board approval for potential bias in the ascertainment of study participants, study methods, data collection, and data interpretation
- Avoidance of industry provision of continuing medical education, samples, and other "gifts" to the institution and the clinicians with privileges at the institution
- Restriction of patient care and research activities of physicians with defined COI

At the individual level:

- Physicians should recuse themselves from any situation (clinical, research, or educational) in which they know they may have a bias.
- Physicians should provide open, honest disclosure whenever COI is appreciated.
- When offered by interested parties, clinicians should not accept any monetary, material, or personally beneficial gift in any form.
- Researchers should never accept the contributions of ghostwriters for research or educational publications

At the federal government level:

- Mandate open access reporting of individuals and industries that provide in-kind compensation for activities
- Revise legislation for COI reporting
- Review industrial contribution to continuing medical education activity

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Content Category 23- Core Knowledge in Scholarly Activities

ABP Content Specs for Developmental-Behavioral Pediatrics Fellow Collaboration Project

Prepared by: Petrina Kaluzhny, MD, Rosa Denisse Rodriguez, MD, & Ruchi Punatar, MD- DBP Fellows, UC Davis

Reviewed by: Kathleen Angkustsiri, MD, UC Davis DBP Fellowship Director

23. Core Knowledge in Scholarly Activities

A. Principles of Use of Biostatistics in Research

1. Types of variables
    a. Distinguish types of variables (eg, continuous, categorical, ordinal, nominal)
    b. Understand how the type of variable (eg, continuous, categorical, nominal) affects the choice of statistical test
2. Distribution of data
    a. Understand how distribution of data affects the choice of statistical test
    b. Differentiate normal from skewed distribution of data
    c. Understand the appropriate use of the mean, median, and mode
    d. Understand the appropriate use of standard deviation
    e. Understand the appropriate use of standard error
3. Hypothesis testing
    a. Distinguish the null hypothesis from an alternative hypothesis
    b. Interpret the results of hypothesis testing
4. Statistical tests
    a. Understand the appropriate use of the chi-square test versus a t-test
    b. Understand the appropriate use of analysis of variance (ANOVA)
    c. Understand the appropriate use of parametric (eg, t-test, ANOVA) versus non-parametric (eg, Mann-Whitney U, Wilcoxon) statistical tests
    d. Interpret the results of chi-square tests
    e. Interpret the results of t-tests
    f. Understand the appropriate use of a paired and non-paired t-test
    g. Determine the appropriate use of a 1- versus 2-tailed test of significance
    h. Interpret a p-value
    i. Interpret a p-value when multiple comparisons have been made
    j. Interpret a confidence interval
    k. Identify a type I error
    l. Identify a type II error
5. Measurement of association
    a. Differentiate relative risk reduction from absolute risk reduction
    b. Calculate and interpret a relative risk
    c. Calculate and interpret an odds ratio
    d. Interpret a hazard ratio
    e. Understand the uses and limitations of a correlation coefficient
6. Regression
    a. Identify when to apply regression analysis (eg, linear, logistic)
    b. Interpret a regression analysis (eg, linear, logistic)
    c. Identify when to apply survival analysis (eg, Kaplan-Meier)

d. Interpret a survival analysis (eg, Kaplan-Meier)
e. Distinguish between regression and correlation

7. Diagnostic tests
   a. Recognize the importance of an independent "gold standard" in evaluating a diagnostic test
   b. Calculate and interpret sensitivity and specificity
   c. Calculate and interpret positive and negative predictive values
   d. Understand how disease prevalence affects the positive and negative predictive value of a test
   e. Calculate and interpret likelihood ratios
   f. Interpret a receiver operator characteristic curve
   g. Interpret and apply a clinical prediction rule
8. Systematic reviews and meta-analysis
   a. Understand the purpose of a systematic review
   b. Understand the advantages of adding a meta-analysis to a systematic review
   c. Interpret the results of a meta-analysis
   d. Identify the limitations of a systematic review
   e. Identify the limitations of a meta-analysis

B. Principles of Epidemiology and Clinical Research Design

1. Study types
   a. Distinguish between Phase I, II, III, and IV clinical trials
   b. Recognize a retrospective study
   c. Understand the strengths and limitations of retrospective studies
   d. Recognize a case series
   e. Understand the strengths and limitations of case series
   f. Recognize a cross-sectional study
   g. Understand the strengths and limitations of cross-sectional studies
   h. Recognize a case-control study
   i. Understand the strengths and limitations of case-control studies
   j. Recognize a longitudinal study
   k. Understand the strengths and limitations of longitudinal studies
   l. Recognize a cohort study
   m. Understand the strengths and limitations of cohort studies
   n. Recognize a randomized-controlled study
   o. Understand the strengths and limitations of randomized-controlled studies
   p. Recognize a before-after study
   q. Understand the strengths and limitations of before-after studies
   r. Recognize a crossover study
   s. Understand the strengths and limitations of crossover studies
   t. Recognize an open-label study
   u. Understand the strengths and limitations of open-label studies
   v. Recognize a post-hoc analysis

w. Understand the strengths and limitations of post-hoc analyses
x. Recognize a subgroup analysis
y. Understand the strengths and limitations of subgroup analyses

2. Bias and Confounding
    a. Understand how bias affects the validity of results
    b. Understand how confounding affects the validity of results
    c. Identify common strategies in study design to avoid or reduce bias
    d. Identify common strategies in study design to avoid or reduce confounding
    e. Understand how study results may differ between distinct sub-populations (effect modification)
3. Causation
    a. Understand the difference between association and causation
    b. Identify factors that strengthen causal inference in observational studies (eg, temporal sequence, dose response, repetition in a different population, consistency with other studies, biologic plausibility)
4. Incidence and Prevalence
    a. Distinguish disease incidence from disease prevalence
5. Screening
    a. Understand factors that affect the rationale for screening for a condition or disease (eg, prevalence, test accuracy, risk-benefit, disease burden, presence of a presymptomatic state)
6. Decision analysis
    a. Understand the strengths and limitations of decision analyses
    b. Interpret a decision analysis
7. Cost-benefit, cost-effectiveness, and outcomes
    a. Differentiate cost-benefit from cost-effectiveness analysis
    b. Understand how quality-adjusted life years are used in cost analyses
    c. Understand the multiple perspectives (eg, of an individual, payor, society) that influence interpretation of cost-benefit and cost-effectiveness analyses
8. Sensitivity analysis
    a. Understand the strengths and limitations of sensitivity analysis
    b. Interpret the results of sensitivity analysis
9. Measurement
    a. Understand the types of validity that relate to measurement (eg, face, construct, criterion, predictive, content)
    b. Distinguish validity from reliability
    c. Distinguish internal from external validity
    d. Distinguish accuracy from precision
    e. Understand and interpret measurements of interobserver reliability (eg, kappa)
    f. Understand and interpret Cronbach's alpha

C. Applying Research to Clinical Practice

1. Assessment of study design, performance, & analysis (internal validity)
    a. Recognize when appropriate control groups have been selected for a case-control study
    b. Recognize when appropriate control groups have been selected for a cohort study
    c. Recognize the use and limitations of surrogate endpoints
    d. Understand the use of intent-to-treat analysis
    e. Understand how sample size affects the power of a study
    f. Understand how sample size may limit the ability to detect adverse events
    g. Understand how to calculate an adequate sample size for a controlled trial (ie, clinically meaningful difference, variability in measurement, choice of alpha and beta)
2. Assessment of generalizability (external validity)
    a. Identify factors that contribute to or jeopardize generalizability
    b. Understand how non-representative samples can bias results
    c. Assess how the data source (eg, diaries, billing data, discharge diagnostic code) may affect study results
3. Application of information for patient care
    a. Estimate the post-test probability of a disease, given the pretest probability of the disease and the likelihood ratio for the test
    b. Calculate absolute risk reduction
    c. Calculate and interpret the number-needed-to treat
    d. Distinguish statistical significance from clinical importance
4. Using the medical literature
    a. Given the need for specific clinical information, identify a clear, structured, searchable clinical question
    b. Identify the study design most likely to yield valid information about the accuracy of a diagnostic test
    c. Identify the study design most likely to yield valid information about the benefits and/or harms of an intervention
    d. Identify the study design most likely to yield valid information about the prognosis of a condition

D. Principles of Teaching and Learning

1. Educational theory
    a. Understand the basic principles of adult learning theory (eg, adult learners are self-directed, goal-oriented, practical; need to feel respected, build on life experiences; learn best when learning is based on an existing framework)
    b. Understand the attributes of an effective learning environment
    c. Understand the importance of "reflective practice" in teaching and learning
    d. Identify strategies that motivate learners

e. Recognize the impact of the "hidden curriculum" on learning

2 Feedback and Evaluation

a. Identify components of effective feedback

b. Distinguish between formative and summative feedback

c. Distinguish between evaluation and feedback

d. Understand the strengths and weaknesses of various methods to evaluate learners

3. Teaching Methods

a. Understand the strengths and weaknesses of various teaching methods (eg, lecture, small group discussion, bedside teaching, simulation)

b. Understand that individuals may learn more effectively with certain teaching methods (eg, reading, hearing, doing) than with others

4. Educational Planning

a. Understand the role of needs assessment in educational planning

b. Distinguish between goals and learning objectives

c. Identify components of well-formulated learning objectives

d. Recognize the strengths and weaknesses of various educational outcome measures (eg, participant satisfaction, acquisition of knowledge and skills, behavioral change, patient outcomes)

E. Ethics in Research

1. Conflicts of Interest and Commitment

a. Evaluate whether an investigator has a conflict of interest during the course of a study

b. Understand ways to manage a conflict of interest

c. Understand what constitutes a conflict of commitment

2. Professionalism and Misconduct in Research

a. Identify forms of research misconduct (eg, plagiarism, fabrication, falsification)

b. Differentiate honest error and differences of opinion from research misconduct

c. Understand the criteria for authorship of clinical research publications

3. Principles of Research with Human Subjects

a. Understand and apply the three main principles of research ethics articulated in the Belmont Report (ie, respect for persons, beneficence, and justice)

b. Understand the role of analysis of risks and benefits in the ethical conduct of research

c. Understand the federal regulatory definitions regarding which activities are considered research

d. Understand the federal regulatory definitions regarding when research includes the use of human subjects

e. Understand the federal regulatory definition of minimal risk

f. Understand the functions of an Institutional Review Board
g. Understand when an exemption from review by the Institutional Review Board is permissible
h. Understand the functions of a Data Safety Monitoring Board
i. Understand the importance of clinical equipoise in research with human subjects
j. Understand the impact of "therapeutic fallacy" on clinical research with human subjects
k. Understand the ethical considerations of study design (eg, placebo, harm of intervention, deception, flawed design)
l. Understand the privacy rules regarding recruitment and participation of subjects in a research study and reporting the results of that study

4. Principles of Consent and Assent
   a. Understand what constitutes informed consent in research
   b. Understand when an exemption from review by the Institutional Review Board is permissible (eg, medical record review of de-identified data
   c. Understand how undue influence can affect obtaining consent for research
   d. Understand how coercion can affect obtaining consent for research
   e. Understand the special ethical considerations related to research utilizing children because of their inability to give informed consent
   f. Distinguish among consent, assent, and permission in research involving children
5. Vulnerable Populations
   a. Recognize that the definition of "children" is related to the underlying clinical intervention in the jurisdiction in which the child is located rather than a fixed nationwide notion of age
   b. Recognize the types of protections that might be accorded to vulnerable populations (eg, incarcerated individuals, pregnant women, fetuses, children, mentally disabled individuals, educationally or economically disadvantaged individuals)
   c. Understand the concept of minimal risk as it applies to research involving children
   d. Understand the circumstances under which research that involves children and that entails greater than minimal risk may be permissible

Core Knowledge in Scholarly Activities

A. Principles of Use of Biostatistics in Research

1.Types of variables

a. Distinguish types of variables (eg, continuous, categorical, ordinal, nominal)

- Continuous
  - Numeric data that is measurable in some way.
    - e.g., height, weight, or time.
- Categorical = Nominal:
  - Data that can be placed into categories with no inherent ordering or quantitative value
    - e.g., sex, race, marital status, primary language
- Ordinal:
  - Data that has some inherent ordering of values but without equal intervals
    - e.g., Likert scales, age group: infant/toddler/preschool/school-age/adolescent/adult)

b. Understand how the type of variable (eg, continuous, categorical, nominal) affects the choice of statistical test

- Continuous analyzed by parametric tests
  - Independent t-test, paired t-test, one way anova (uses the mean)
- Nominal and ordinal data analyzed using Non-parametric
  - Chi-square, Mann-Whitney Test, Wilcoxon signed rank (uses rank order or median)

2.Distribution of data

a. Understand how distribution of data affects the choice of statistical test

o Parametric assumes normal distribution

- 4 main assumptions of parametric analyses to be valid = acronym LINE:

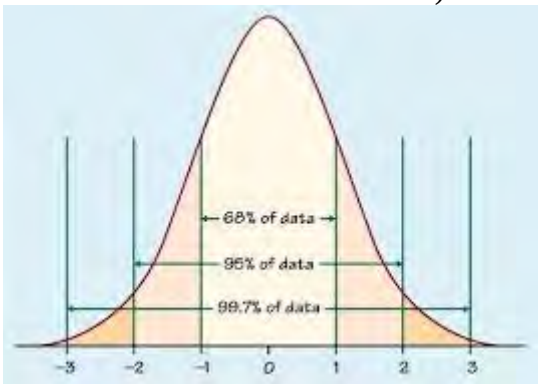
1. Linearity/linear relationship (between independent and dependent variables),
2. Independence (each observation is independent of the others),
3. Normality/normal distribution of the variables,
4. Equal variance/homogeneity of the variances in the groups being compared.

b. Differentiate normal from skewed distribution of data

Normal Distribution – Gaussian

- Bell shaped curve based on the dispersion of values of a variable.
- The highest point is the mean of the variable
- Standard deviation determines the width of the curve, or the dispersion of the data.
- The mean, median and mode are identical in a true normal distribution.
- The probability of an individual observation falling within one standard deviation (SD) of the mean is 68.3%, within two SD's 95.4% and within 3 SD's 99.7%.

- If the group truly has a normal distribution, an individual observation will fall randomly more than 2 SD's from the mean only about 5% of the time (2.3% more than 2 SD above, and 2.3% more than 2 SD below).



Skewness

- Distribution of the observations is not symmetric
- The mean, median and mode are not the same
- Direction of the skewness is based on the tail,
  - "right skewed"- Tail is skewed toward the right (typically indicating large values)
  - "left skewed." - Tail is skewed toward the left (typically toward smaller values)

c. Understand the appropriate use of the mean, median, and mode
   - Mean is equal to the sum of all the values in the data set divided by the number of values in the data set
   - Median – middle score for a set of data that has been arranged in order of magnitude. It is possible for none of the observed values to equal the median value – e.g., if the observed values are (1, 1, 3, 3), the median is the average of the two middle values ie 2. The median is less affected by outliers and skewed data
   - Mode – most frequently occurring value. There can be more than 1 mode – if the observed values are again (1, 1, 3, 3), there are two modes (bimodal distribution): 1 and 3.

d. Understand the appropriate use of standard deviation
   - A measure to describe the spread of individual observations around the observed mean

- 68% of the population is within 1 SD of the mean, 95% are within 2 SD
- Square root of the variance; absolute value of the average difference of individual values from the mean.

e. Understand the appropriate use of standard error of the mean
- Describes how far the sample mean is from the true population mean (many samples make up a population); related to sample size
- SD divided by square root of the sample size.

3. Hypothesis testing

a. Distinguish the null hypothesis from an alternative hypothesis
- Null Hypothesis- This is the "usual view of the world" and suggests that there is no difference between the group/treatment/exposure being studied and the general population.
- Alternate Hypothesis- there is a chance of finding a difference between the groups based on theory or prior studies.

b. Interpret the results of hypothesis testing
- If a difference is found, the null hypothesis is rejected.
- If there is no difference, then the null hypothesis is confirmed.

4.Statistical tests

a. Understand when to use and how to interpret the chi square test
- Chi-square tests are a non-parametric method used to compare the difference between groups on the frequency of a nominal or ordinal variable (e.g., the frequency of short stature in groups taking/not-taking psychostimulants).
- The results of a Chi-squared test indicate whether the frequency of a nominal or ordinal variable is the same in two groups. E.g., is the frequency of short stature the same in children taking stimulant medication compared with children who aren't

b. Understand when to use and how to interpret tests comparing continuous variables between two groups (eg, t test, Mann Whitney U)
- T-tests are a **parametric** method used to compare the **differences of the means** between 2 groups on a continuous variable (e.g., comparing height in children taking/not taking psychostimulants)
- Mann Whitney U test is a **nonparametric** test of **ordinal or continuous** values that allows two groups or conditions or treatments to be compared without making the assumption that values are normally distributed. Used for independent samples – can compare medians

c. Understand when to use and how to interpret tests comparing continuous variables between three or more groups (eg, ANOVA, Kruskal-Wallis)
- ANOVA tests are a parametric method used to determine statistically significant differences **between the means** of three or more independent (unrelated) groups. The dependent variable is continuous. Multiple independent variables, whether nominal, ordinal, ratio, or interval, can be used in the analysis. ANOVA is an **omnibus** test statistic and cannot tell you which specific groups were statistically significantly different from each other, only that at least two groups were different. E.g., ANOVA can be used to compare height by age, sex and medication dose.

d. Understand when to use paired tests

- "Paired" statistical testing refers to situations where certain observations are
matched with each other.
- One example would be observations before & after treatment in a single individual
- Another example would be multiple repeated measures on an individual (before, during, and after treatment)
- Paired/matched statistical tests have greater statistical power than non-paired tests (see type II error, below).

e. Understand the appropriate use of parametric versus nonparametric tests
- Continuous analyzed by parametric tests
    i. Independent t-test, paired t-test, one way anova
- Nominal and ordinal data analyzed using Non-parametric
    i. Chi-square, Mann-Whitney Test, Wilcoxin signed rank

f. Interpret a p value
- P-values describe the probability that the result of the statistical test) could arise when the null hypothesis is true.
- i.e. the chance that the difference between comparison groups could have arisen by chance alone, when the groups are actually no different from each other.

g. Interpret a p value when multiple comparisons have been made
- p-values should be adjusted when multiple statistical tests are being carried out as when more than 1 test is being performed, there is a greater likelihood that one of the comparisons will have a nominally significant result by chance alone.

h. Interpret a confidence interval
- A confidence interval indicates a range of values (upper and lower) that would be likely to be found if an experiment were repeated.

i. Identify a type I error
- Type I Error – The mistake made when the researcher rejects the null hypothesis when the null hypothesis is actually true.
- i.e. a false positive result
- The p-value is used to estimate the chance of a Type 1 error (i.e., we usually demand less than 5%).

j. Identify a type II error
- Type II Error – The mistake made when the researcher does not reject the null hypothesis when the null hypothesis is actually false.
- i.e. a false positive result.

5.Measurement of association and effect

a. Understand how to interpret relative risk and absolute risk
- Absolute risk of a disease is the risk of developing the disease over a time period.
    - AR (**absolute risk**) = the number of events in treated or control groups, divided by the number of people in that group.
- Relative risk is used to compare the risk in two different groups of people.

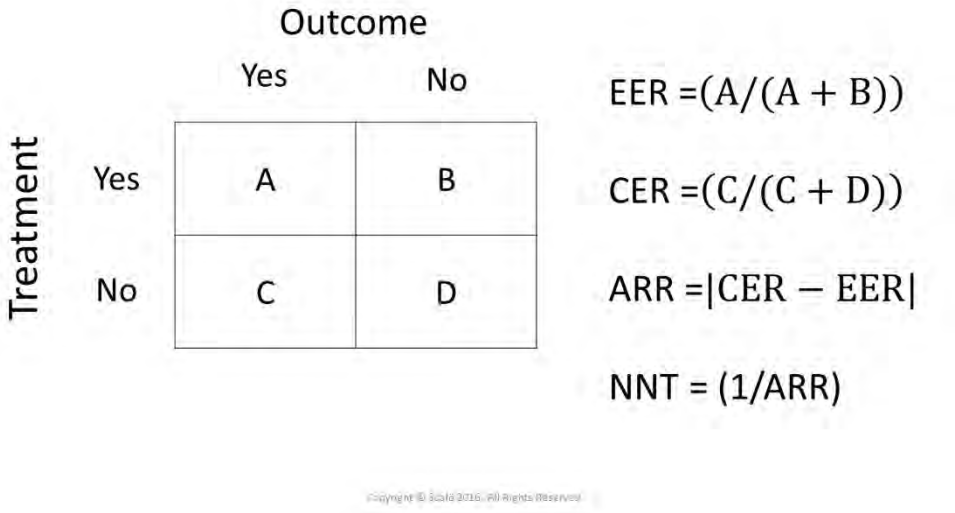
Exposure Status	Event Occurred	
	Yes	No
Exposed	a	b
Not Exposed	c	d

$$\text{Relative Risk} = \frac{a/(a+b)}{c/(c+d)}$$

$$\text{Odds Ratio} = \frac{a/b}{c/d} = \frac{ad}{cb}$$

b. Understand how to interpret odds ratio
   - The odds ratio describes how strongly having 1 characteristic is associated with having another characteristic.

c. Understand how to interpret number needed to treat or harm
   - The NNT is the average number of patients who need to be treated to prevent one additional bad outcome



d. Understand how to interpret hazard ratio
   - Hazard ratios are used in survival analysis.
   - They describe the likelihood of some event occurring under one condition compared to another.

e. Understand when to use and how to interpret correlation coefficient
   - "Correlation" refers to a broad class of statistical relationships between **2 continuous variables. when one changes by a certain amount the other changes on an average by a certain amount.**
   - Correlation coefficients typically are quantified in manner that they may range from 0 (no correlation) to 1 (complete correlation between 2 variables).
   - Complete correlation does not mean that the 2 variables are identical

6.Regression

a. Understand when to use and how to interpret regression analysis (eg, linear, logistic)
    - Regression refers to a group of statistical methods to estimate the relationships among variables.
    - In general, one or more independent variables are used to predict the value of a dependent variable.
    - Linear regression, including multiple linear regression is encountered most commonly – in these analyses, the relationship between the independent and dependent variables is linear.
    - When the dependent variable is categorical (e.g., nominal data), then logistic regression is used.
    - In a typical regression analysis, multiple independent variables are tested to see which, if any, are related to the dependent variable.

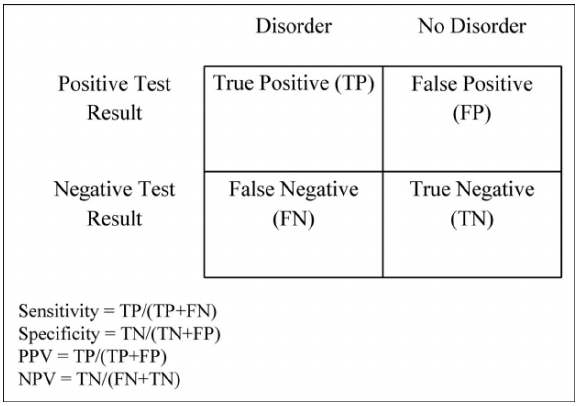
b. Understand when to use and how to interpret survival analysis (eg, Kaplan Meier)
    - Survival analysis tells us the probability of an event at a certain time interval (such as survival).

7.Diagnostic tests

a. Recognize the importance of an independent "gold standard" in evaluating a diagnostic test
    - A “gold standard” is the best assessment of the truth but is not always practical/possible to perform.
    - Other assessments are typically compared to the gold standard to determine their utility.

b. Interpret sensitivity and specificity
    - Sensitivity is the proportion of subjects with a disorder who have a true positive test result
        - High sensitivity is most important in screening for severe, treatable conditions (e.g., PKU), so that no cases are missed.
    - Specificity: the proportion of subjects without a disorder who have a true negative test result
        - High specificity is most important when false positive results could be associated with high burdens (physically, emotionally or financially)

c. Interpret positive and negative predictive values
    - Predictive Value: The predictive value of a test result (either positive or negative) depends on its sensitivity and specificity, and also on the prevalence of the disorder under investigation
    - Positive Predictive Value (+PV) – probability of actually having a condition when the test result is positive (abnormal): true positives / all positives.
        - Higher specificity is associated with higher +PV.
    - Negative Predictive Value (–PV) – probability of not having a disorder when the test result is negative (normal).
        - Higher sensitivity is associated with higher –PV



d. Understand how disease prevalence affects the positive and negative predictive value of a test

- Note that changing the prevalence in the tested population does not change the sensitivity or specificity
- The PPV will increase with increasing prevalence; and NPV decreases with increase in prevalence

e. Interpret a receiver operating characteristic curve

- An ROC curve shows the relationship of test sensitivity to specificity, and possibly showing multiple curves corresponding to various "cut-off" (threshold) levels used in interpreting the test.
- Plot of sensitivity vs. 1-sensitivity
- Test accuracy; the closer the graph is to the top and left-hand borders, the more accurate the test. Likewise, the closer the graph to the diagonal, the less accurate the test. A perfect test would go straight from zero up the top-left corner and then straight across the horizontal.
- The greater the area under the curve, the more accurate the test.

8. Systematic reviews and meta-analysis

a. Understand the purpose of a systematic review

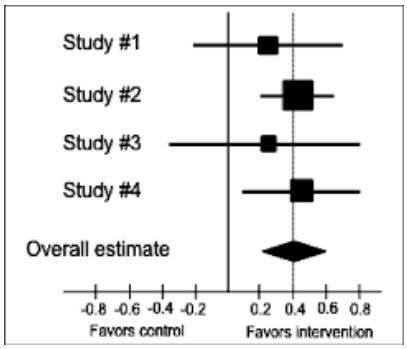
- Systematic reviews utilize concrete, reproducible methods to identify, appraise, select and synthesize the available research on a specific question.

b. Understand the advantages of adding a meta-analysis to a systematic review

- The largest potential advantage of a meta- analysis is the increased statistical power provided by combining data across multiple study populations.

c. Interpret the results of a meta-analysis

- A meta-analysis is a formal, mathematical/statistical analysis of data that were collected across multiple studies.
- The findings of a meta-analysis are commonly presented in a format known as "forest plot." These plots typically depict the key result and the 95% confidence interval for that result, from each study. At the bottom of the forest plot, the authors of the meta-analysis typically present the result of their overall statistical synthesis. The size of the symbols in the forest plot sometimes reflects the weighting given to each study relative to others, which may reflect the sample size of each study.

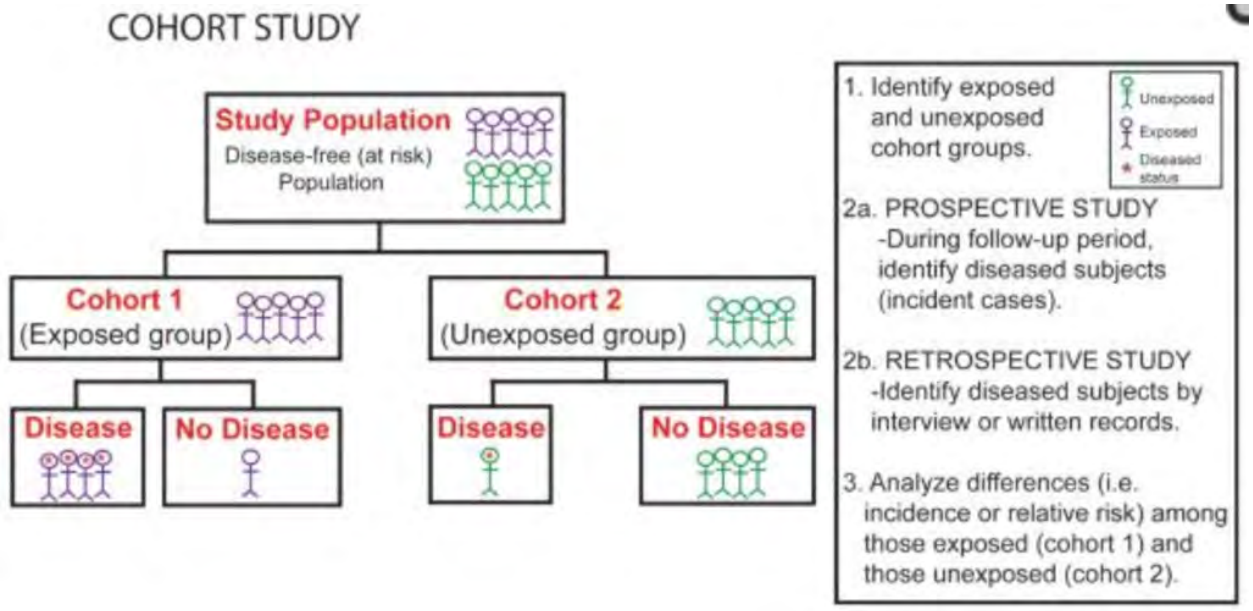


B. Principles of Epidemiology and Clinical Research Design

1. Assessment of study design, performance and analysis (internal validity)
Internal validity: how sound the results are; how well the study is done. achieved by adequate sample size, avoiding selection biases in recruiting the study sample, measuring the exposures and outcomes accurately, controlling for confounding, and performing appropriate analyses.

a. Recognize and understand the strengths and limitations of a cohort study, case control study, and randomized controlled clinical trial

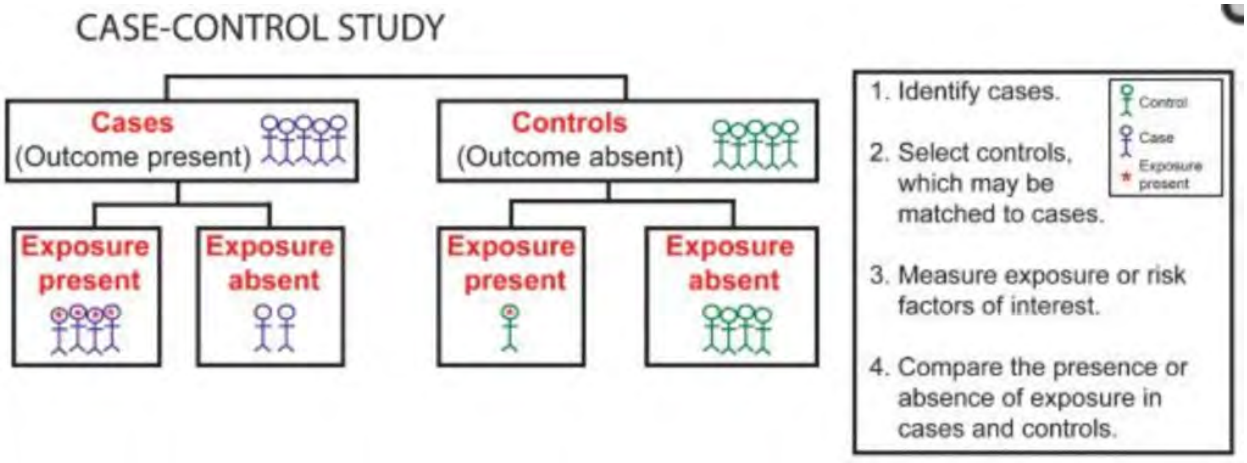
- Cohort Studies: an outcome or disease-free study population is first identified by the exposure or event of interest and followed in time until the disease or outcome of interest occurs (prospective). They can be prospective or retrospective. Because exposure is identified before the outcome, cohort studies have a temporal framework to assess causality and thus have the potential to provide the strongest scientific evidence.



- Strengths:
- Investigator can examine multiple outcomes simultaneously.

- Can calculate rate of disease in exposed and unexposed individuals over time (relative risk, incidence)
- Limitations:
- Susceptible to selection bias.
- Large numbers are required to study rare exposures.
- Susceptible to loss to follow up or withdrawals.
- Susceptible to recall bias.
- Less control over variables.

- Case-Control Studies: Case-control studies identify subjects by outcome status at the outset of the investigation (retrospective). Outcomes of interest may be whether the subject has undergone a specific type of surgery, experienced a complication, or is diagnosed with a disease. Once outcome status is identified and subjects are categorized as cases, controls (subjects without the outcome but from the same source population) are selected.



- Strengths:
- Good examining rare outcomes or outcomes with long latency.
- Relatively quick to conduct.
- Relatively inexpensive.
- Multiple exposures or risk factors can be examined.
- Requires comparatively few subjects.
- Limitations:
- Difficult to validate information.
- Control of extraneous variables may be incomplete.
- Selection of an appropriate comparison group may be difficult.
- Susceptible to recall or information bias.

- Randomized controlled trials (RCTs): are the gold standard for ascertaining the efficacy and safety of a treatment. RCTs can demonstrate the superiority of a new treatment over an existing standard treatment or a placebo. In RCTs the patients are randomly assigned to the different study groups. This is intended to ensure that all potential confounding factors are divided equally among the groups that

will later be compared. The assignment to study groups must not be in any way predictable (random).

- Strengths:
- Best studies to determine causal relationship.
- Eliminates conscious and unconscious bias
- Population is clearly identified
- Limitations:
- Relatively expensive in time and money.
- Need for rapid enrollment (new advances may cause the trial to be obsolete or unethical)
- Intervention fidelity may not be maintained.
- Large number of participants with specific characteristics (may affect generalization).

	Objective	Typical No. of Patients
Phase I	To explore possible toxic effects and determine tolerance of the intervention (and tolerated dose, if a drug study).	10 to 30
Phase II	To determine if treatment has a therapeutic effect or if there is any hope for benefits to outweigh the risks.	20 to 50
Phase III	To compare new treatment to the standard therapy or a control or placebo (if no standard therapy exists).	100 to 1000
Phase IV	To obtain long-term, large-scale information on morbidity and late effects (postmarketing study).	Hundreds or thousands

b. Recognize the use and limitations of surrogate endpoints

- *End Point*: is the outcome measure used to make the decision on the overall result of the study and serves as the basis to determine the number of patients needed for the study. In clinical trials, an event or outcome that can be measured objectively to determine whether the intervention being studied is beneficial. Some examples of endpoints are survival, improvements in quality of life, relief of symptoms, and disappearance of the tumor.
- A surrogate endpoint is a measure that is predictive of the clinical event but takes a shorter time to observe. The definitive endpoint often measures clinical benefit whereas the surrogate endpoint tracks the progress or extent of disease. Surrogate endpoints could also be used when the clinical endpoint is too expensive or difficult to measure, or not ethical to measure.

c. Understand the use of intent-to-treat analysis

- *Intention to treat (ITT)*: primary population for analysis, this includes all patients who were randomized, the data for analysis include some patients whose treatment was interrupted, prematurely discontinued, or did not take place at all.

d. Understand how sample size affects the power of a study

- Sample size is an important element of trial design because too large of a sample size is waste of resources but too small of a sample size could result in inconclusive results. Calculation of the sample size requires a clearly defined objective. The sample size is based on the planned analyses. In order to find the sample size you must:

1. Formulate null and alternative hypotheses.
2. Select the Type I error (a error) rate. Type I error is the probability of incorrectly rejecting the null hypothesis when the null hypothesis is true. Type I error often implies that you incorrectly conclude that an intervention is effective.
3. Select the Type II error (b error) rate. Type II error is the probability of incorrectly failing to reject the null hypothesis when the null hypothesis should be rejected. The implication of a Type II error in the example above is that an effective intervention is not identified as effective.
4. Obtain estimates of quantities that may be needed. This may require searching the literature for prior data or running pilot studies.
5. Sample size calculations may also need to be adjusted for the possibility of a lack of adherence or participant drop-out.

The following increases the required sample size: lower Type I error, lower Type II error, larger variation, and the desire to detect a smaller effect size or have greater precision.

2. Assessment of generalizability (external validity)

- External validity describes the extent to which the results of a trial can be generalized into clinical practice and the general population.

a. Understand how nonrepresentative samples can bias results
b. Assess how the data source (eg, diaries, billing data, discharge diagnostic code) may affect study results
A sampling method is called biased if it systematically favors some outcomes over others. Sampling bias is sometimes called ascertainment bias (especially in biological fields) or systematic bias. Bias can be intentional, but often it is not.

3. Bias and confounding

- The four main sources of bias in clinical trials are:

1. Selection bias usually controlled by randomization. The main goal of randomized trials is therefore to assure that each individual has an equal probability to be assigned to one or the other treatment.
2. Performance bias: insufficient adherence to the study protocol either by the participant or the investigator.
3. Information bias refers to systematically different outcome assessments among study groups.
4. Attrition bias occurs if there are systematic differences in the number of participants dropping out of the study among the study groups.

- Confounding: the effect of the exposure of interest on the outcome is confused with the effect of another risk or protective factor for the outcome.

a. Identify common strategies in study design to avoid or reduce bias

- Randomization
- Blinding (single, double blind)
- Allocation concealment

b. Identify common strategies in study design to avoid or reduce confounding

- *Matching or standardization* is a method used in an attempt to ensure comparability between cases and controls and reduces variability and systematic differences due to background variables that are not of interest to the investigator. Age, sex, and race are often used to match cases and controls because they are typically strong confounders of disease.[2]
- *Stratified randomization or restriction* intended to ensure that all potential confounding factors are divided equally among the groups that will later be compared (structural equivalence). [4] examples of stratification: stratified by center (multicenter trials), stratified by sex (male, female), age <18 years, >18 years).

4. Causation

a. Understand the difference between association and causation

- *Association:* Is a specified health outcome more likely in people with a particular "exposure"? Is there a link? Association is a statistical relationship between two variables.
- *Causation*: means that the exposure produces *t*he effect. Causative factors can also be the absence of a preventive exposure, such as not wearing a seatbelt or not exercising.

5. Incidence and prevalence

a. Distinguish disease incidence from disease prevalence

- *Incidence* is a measure of the number of **new cases** of a characteristic that develop in a population in a specified time period; whereas *prevalence* is the proportion of a population who have a specific characteristic in a given time period, regardless of when they first developed the characteristic.
- Prevalence may be reported as a percentage (5%, or 5 people out of 100), or as the number of cases per 10,000 or 100,000 people. The way prevalence is reported depends on how common the characteristic is in the population.

There are several ways to measure and report prevalence depending on the timeframe of the estimate.

- Point prevalence is the proportion of a population that has the characteristic at a specific point in time.
- Period prevalence is the proportion of a population that has the characteristic at any point during a given time period of interest. “Past 12 months” is a commonly used period.

- Lifetime prevalence is the proportion of a population who, at some point in life has ever had the characteristic.

6. Screening

a. Understand factors that affect the rationale for screening for a condition or disease (eg, prevalence, test accuracy, risk benefit, disease burden, presence of a presymptomatic state)

- *Screening* is defined as the presumptive identification of unrecognized disease in an apparently healthy, asymptomatic population by means of tests, examinations or other procedures that can be applied rapidly and easily to the target population.
- Screening programs should be undertaken only when their effectiveness has been demonstrated, when resources are sufficient to cover nearly all of the target group, when facilities exist for follow-up of those with abnormal results to confirm diagnoses and ensure treatment and when prevalence of the disease is high enough to justify the effort and costs of screening.

7. Cost benefit, cost effectiveness, and outcomes

- In *cost-benefit analyses,* both costs and outcomes are measured in monetary units. When all outcomes have been valued in monetary terms, all of the costs are added up and subtracted from the dollar value of the outcomes. If the resulting total is positive, the outcomes outweigh the costs, and the intervention is favored. The difficulty in performing cost-benefit analyses is in valuing the outcomes, such as extending life, in monetary terms. This is usually performed using either the human capital approach or the willingness to pay approach. The human capital approach values an improvement in health on the basis of future productive worth from being able to return to work.8The willingness to pay approach values an improvement in health on the basis of how much people are willing to pay for the improvement.

a. Interpret cost-effectiveness ratios

- *Cost-effectiveness analyses* measure outcomes (effectiveness) in naturally occurring, health-related units, such as lives saved, life years gained, or cases of ventilator-acquired pneumonia prevented. When the outcomes are combined with costs, a ratio (known as a cost-effectiveness ratio) of the net change in costs (between two interventions) divided by the net change in outcomes is produced. The net change in costs reflects the extra cost required to achieve the difference in outcome (such as life years gained), with the ratio expressed as the cost per outcome gained, such as the cost per life year gained or the cost per life saved.

b. Distinguish costs from charges

- The true measure of the *cost* of a resource is what could have been achieved with that resource in its next best alternative use (known as opportunity cost). In practice this is valued as the financial cost of the resource although there may be cases in which prices are distorted and this needs to be amended. Costs are typically measured using bottom-up microcosting methods, more aggregated bundled grouped costs such as diagnosis-related grouped costs, or charges.

Microcosting involves measuring the quantity of resources used for each patient and attaching a unit cost to each of the resources. Examples of resources include personnel (nursing, medical, allied health) time, medications, diagnostic tests, and transport costs.

- *Charges* are values that use the market or administered price for treatment. The use of charges is suboptimal, as they may not reflect actual costs and are very specific to given departments and institutions. As charges do not reflect actual costs, a cost-to-charge index (which is publicly available) is often used to adjust the charges; however, it is unclear what the resultant cost figure represents.

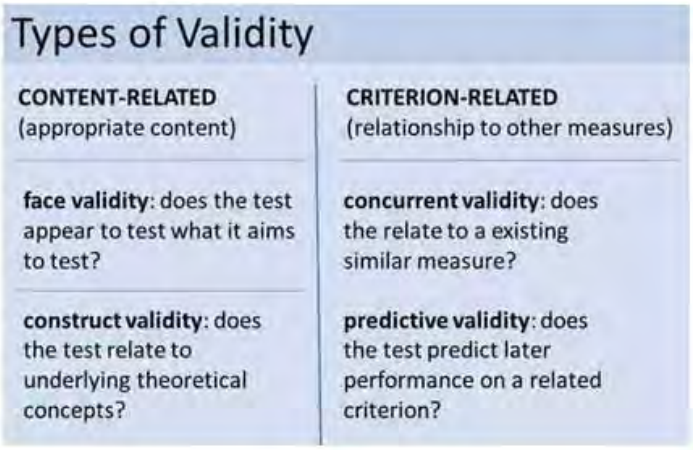
c. Understand quality-adjusted life years

- The *quality-adjusted life year* (QALY) is a unit of measurement for valuing health outcomes. It is designed to capture in one single measure an individual's gain in utility from improvement in both quality of life and length of life. Cost-effectiveness ratios using QALYs allow comparisons of value for money of different interventions in different areas of medicine.

8. Measurement

a. Understand the types of validity that relate to measurement (eg, face, construct, criterion, predictive, content)

The concept of validity was formulated by Kelly who stated that a test is valid if it measures what it claims to measure. For example, a test of intelligence should measure intelligence and not something else (such as memory).



- *Content validity* is a theoretical concept which focuses on the extent to which the instrument of measurement shows evidence of fairly and comprehensive coverage of the domain of items that it purports to cover. Factor analysis is used to determine whether items in the instrument fit into conceptual domain
- *Face validity* is simply whether the test appears (at face value) to measure what it claims to. This is the least sophisticated measure of validity. The face validity of a test can be considered a robust construct only if a reasonable level of agreement exists among raters. Having face validity does not mean that a test really measures what the researcher intends to measure, but only in the judgment of raters that it appears to do so. Consequently, it is a crude and basic measure of validity.
- *Construct validity* is a device commonly used in educational research. It is based on the logical relationships among variables.

- Construct validity refers to whether the operational definition of a variable actually reflects the theoretical meanings of a concept. In other words, construct validity shows the degree to which inferences are legitimately made from the operationalization's in one's study to the theoretical constructs on which those operationalization's are based.
- A criterion is a standard of judgment or an established standard against which other measure is compared. Therefore, *criterion-related validity* covers correlations of the measure with another criterion measure, which is accepted as valid. In other words, criterion-related validity is where a high correlation coefficient exists between the scores on a measuring instrument and the scores on other existing instrument which is accepted as valid.
- *Predictive Validity* operationalization's ability to predict what it is theoretically able to predict. The extent to which a measure predicts expected outcomes. Concurrent Validity operationalization's ability to distinguish between groups it theoretically should be able to.

b. Distinguish accuracy from precision

- *Accuracy* is how close you are to the true value, i.e. how well you are measuring a construct.
- *Precision* is how close two or more measurements are to each other.

If you are precise, that doesn't necessarily mean you are accurate. However, if you are consistently accurate, you are also precise.

c. Understand when to use and how to interpret a kappa coefficient

- The kappa statistic is frequently used to test interrater reliability. The importance of rater reliability lies in the fact that it represents the extent to which the data collected in the study are correct representations of the variables measured. There are actually two categories of reliability with respect to data collectors: reliability across multiple data collectors, which is *interrater* reliability, and reliability of a single data collector, which is termed *intrarater* reliability. Cohen's kappa, is a robust statistic useful for either interrater or intrarater reliability testing. It can range from −1 to +1, where 0 represents the amount of agreement that can be expected from random chance, and 1 represents perfect agreement between the raters.

Interpretation of Cohen's kappa.

Value of Kappa	Level of Agreement	% of Data that are Reliable
0–.20	None	0–4%
.21–.39	Minimal	4–15%
.40–.59	Weak	15–35%
.60–.79	Moderate	35–63%
.80–.90	Strong	64–81%
Above.90	Almost Perfect	82–100%

C. Ethics in Research

1. Professionalism and misconduct in research

a. Identify and manage potential conflicts of interest in the funding, design, and/or execution of a research study

- Conflict of interest = "circumstances that create a risk that professional judgement or actions regarding a primary interest will be unduly influenced by a secondary interest." (B Low & MJ Fields, Conflict of Interest in Medical Research, 2009)
    - Primary interest includes integrity of research, quality of medical education welfare of patients
    - Secondary interests include financial, professional advancement, recognition
- How to manage potential conflict of interest:
    - Removal of the conflict
        - For example, placing financial investments in a blind trust
    - Disclosure of the conflict
        - This will mitigate conflict, but does not remove it
    - Recusal from research study
    - Third party mediation

b. Identify various forms of research misconduct (eg, plagiarism, fabrication, falsification)

- Research Misconduct = fabrication, falsification, or plagiarism in proposing, performing, or reviewing research, or in reporting research results
    - Deliberate or deceptive action, deviation from accepted norms
    - Misconduct may be intentional (falsifying results) or negligent (not monitoring safety of subject)
- Types of research misconduct
    - Plagiarism = the appropriation of another person's idea, processes, results, or words without giving appropriate credit
    - Fabrication = making up data or results and reporting them
    - Falsification = manipulating research material, equipment, or processes, or changing or omitting data such that the research is not accurately represented in the research record

c. Know how, and to whom, to report concerns of research misconduct

- If there is concern for research misconduct
    - Be aware of policy establish by an individual's institution for making and responding to allegations
    - Institutional policy should address
        - What information should be reported
        - Whom the allegation should be reported to
        - What protections are provided
        - What role the individual will play in the proceedings
    - Allegations should be made to the institutional official whose role it is to receive such allegations
        - Allegations can also be made to the Office of Research Integrity (part of the U.S. Department of Health & Human Services)
    - The Public Health Service Policy on Research Misconduct obligates institutions to provide confidentiality to all respondents, complainants, and research subjects
        - The complainant should be protected from retaliation
    - It is the role of the investigative body or Office of Research Integrity to ensure the allegation is thoroughly and completely investigated

2. Principles of research with human subjects

- Three core principals of research ethics from The Belmont Report
    - Respect for person: An individual's right to make decision for and about themselves without influence or coercions from someone else
    - Beneficence: The principle of doing no harm and minimizing risk, the intention of the research is for good
    - Justice: obligation to distribute benefits and risks equally without prejudice to particular individuals or groups

a. Understand and contrast the functions of an Institutional Review Board and a Data Safety Monitoring Board

- Institutional Review Board (IRB)
    - A group of experts and community members responsible for reviewing, approving the initiation of, and conducting periodic review of research involving human subject
    - Primary function is to protect the rights and welfare of human subjects
- Data Safety Monitoring Board (DSMB)
    - Committee responsible for periodically monitoring the data that are gathered to assess the emerging evidence on safety and effectiveness of the investigational treatment, the study conduct, and monitoring of external data that may have implications for the study
    - Evaluates information to determine whether the study should continue to be conducted as originally planned, if modification to study conduct should be made, or if study should be terminated (such as for safety risk).

b. Recognize the types of protections in designing research that might be afforded to children and other vulnerable populations

- For any vulnerable population special considerations should be made to ensure that risk of participating is acceptable, selection is fair, consent information is as basic as possible, and adequate follow up is provided
    - Vulnerable populations = incarceration individuals, pregnant women, fetuses, children, individuals with intellectual disability, educationally or economically disadvantaged individuals
- Clinical research with children requires more careful ethical analysis and oversight
    - Federal regulation mandates that clinical research with children is acceptable if it involves only minimal risk. Research that subjects children to greater than minimal risk may be approved under certain circumstances (see section 2d)

c. Understand the federal regulatory definitions regarding which activities are considered research and what constitutes human subjects research

- Research
    - systematic investigation, including development, testing, and evaluation designed to develop or contribute to generalizable knowledge
- Human subject
    - living individuals about whom an investigator conducting research obtains:
        - Data through intervention or interaction with the individual
        - Identifiable private information

d. Understand the federal regulatory definition of minimal risk and apply this to research involving children

- Minimal Risk
    - The probability and magnitude of harm or discomfort anticipated in the proposed research are not greater, in and of themselves, than those ordinarily encountered in daily life or during performance of routine physical or psychological examinations or tests
- There are three categories of research involving children which Health and Human Services permits IRB to approve
    - Research not involving greater than minimal risk to children
        - Research presents no greater than minimal risk to children AND
        - Adequate provisions are made for soliciting the assent of children and the permission of guardian
        - Example: Blood draws are considered minimal risk
    - Research involving greater than minimal risk but there is prospect of direct benefit to the individual child subjects involved in the research
        - The risk is justified by the anticipated benefits to the subject
        - The relationship of the anticipated benefit to the risk presented by the study is at least as favorable to the subjects as that provided by available alternative approaches
        - Adequate provisions are made for soliciting the assent of the children and the permission of their guardians

    - Research involving greater than minimal risk and no prospect of direct benefit to the individual child subjects involved in the research, but likely to yield generalizable knowledge about the subject's disorder or condition
        - The risk of the research represents a minor increase over minimal risk
        - The intervention or procedure is likely to yield generalizable knowledge about the subject's disorder or condition which is of vital importance for the understanding or amelioration of the disorder or condition
        - Adequate provisions are made for soliciting the assent of the child and the permission of their guardian
- A fourth category of research requires a review by the Department of Health and Human Services
    - There is no direct benefit to the child or generalizable knowledge, but the research presents reasonable opportunity to further understanding, prevention, or alleviation of a serious problem affecting the health or welfare of children

e. Understand the ethical considerations of study design (eg, placebo, harm of intervention, deception, flawed design)

- The principle of beneficence requires minimizing the risk to study participants. Ethical considerations must be taken into account when designing a study
    - Placebo is an inactive version of a treatment identical in appearance to the real treatment.
        - The Declaration of Helsinki states "The benefits, risks, burdens and effectiveness of a new method should be tested against those of the best current prophylactic, diagnostic, and therapeutic methods. This does not exclude the use of a placebo, or no treatment, in studies where no proven prophylactic, diagnostic or therapeutic method exist"
        - Placebo controlled trials should only be used in the absence of existing proven therapy, except if there is a compelling and scientifically sound reason to use a placebo or when the placebo is for a minor condition and the patient will not be subject to additional risks of serious harm
    - Harm of intervention
        - Research design need to balance between patient benefit and harm of intervention
        - Research protocol must be designed to minimize harm caused by an intervention
    - Deception
        - Deception is a practice where investigators provide false of incomplete information to participants for the purpose of misleading research subjects
        - Rarely used in medical research, but is used in psychological research

        - Deception should not be used if the non-deceptive methods can be used to obtain same information
        - If deception is employed, post procedure debriefing with disclosure of the deception is expected
    - Flawed design
        - Poorly designed and executed studies can influence practice and policy
        - Elements that make for a flawed design include
            - Excessive risk compared to benefit
            - Inadequate power
            - Inappropriate allocation of dosages in comparison trial
            - Poor selection and misallocation of participants
            - Midstream changes to protocol
            - Failure to monitor or record significant adverse events

3. Principles of consent and assent

a. Understand what constitutes informed consent in research

- Informed consent = the process of providing information about the research study to an individual so that he or she can make an informed decision about whether to participate in research. This is a process, which occurs throughout the length of the research study, begins with recruitment and occurs periodically throughout the study.
- Informed consent must have
    - Information
        - Explanation of the research in understandable language, including nature of proposed study, probability of success for outcome, risks to subject, costs, benefits and drawback of participation, and alternatives to participation
    - Comprehension
        - All information must be comprehensible to the prospective subject. The investigator must assess the subject's understand, as well as capacity of subject or surrogate to make decision
    - Voluntariness
        - Assurance that the subject knows that he or she has the freedom to choose to participate or not, without coercion or manipulation. Participant can withdraw at any point

b. Distinguish between consent and assent in research involving children

- Consent is an agreement made by an individual or his/her authorized representative who has legal capacity to give consent
- Assent is the willingness to participate in research by a person who is not able to legally consent to participate in the activity
    - The individual should have an understanding of the research and what it means to participate
- Assent in researching involving children
    - A child's affirmative agreement to participate in research differs by age
        - Ages 2-7: request for assent should be kept simple and direct

        - Ages 6-14: the request should include a general description of the purpose of the child's participation, brief description of the experimental task, an assurance that the child's participation is voluntary and that he may withdraw from the study at any point, and an offer to answer any questions
        - Ages 12-17: the request for assent should include the elements of informed consent presented to adults but presented in language appropriate to the child's level of comprehension
- Parental consent must be obtained prior to seeking child's assent
    - Consent of the parent/legal guardian and assent of the subject is needed in research

D. Quality Improvement

- Quality improvement is a systematic, formal approach to the analysis of practice performance and efforts to improve performance

1. Design of a Project

a. Understand various models of quality improvement and recognize that all utilize a data-informed, iterative process using tests of change to achieve a stated aim

- A variety of approaches exist to help collect data, analyze the data, and test change
    - Model for Improvement (Plan-Do-Study-Act cycles)
        - The Institute for Healthcare model combines two popular QI models: Total Quality Management (all members of an organization participate in improving processes) and Rapid-Cycle Improvement (implementation and measurement of changes made to improve system, usually over a period of three months of less)
            - This framework is used to test intervention on small scale in multiple progressive cycles
    - LEAN: continual improvement of processes by increasing value-adding activities and reducing non-value adding activities, variation, and bad work conditions.
        - There is an emphasis on cutting out unnecessary and wasteful steps in the creation of a product or the delivery of a service so that only steps that directly add value are taken
        - Also called "Toyota Production System"
    - Six Sigma: eliminate defects and waste, thereby improving quality and efficiency by streamlining and improving all business processes
        - Improves quality of process output by identifying and correcting the root cause of variation
        - DMAIC methodology = Define, Measure, Analyze, Improve, and Control

b. Understand that the aim of any quality improvement project should be specific, measurable, achievable, realistic, and time-limited

- When planning a QI project, the aim statement should reflect what will be accomplished with the project using SMART.
    - SMART

- S= specific
- M = measurable
- A = achievable
- R = realistic
- T = time limited

Specific	Is the statement precise about what the team hopes to achieve?
Measurable	Are the objectives measurable? Will you know whether the changes resulted in improvement?
Achievable	Is this doable in the time you have? Are you attempting too much? Could you do more?
Realistic / Relevant	Do you have the resources needed (people, time, support?) Aligns with mission
Timely	Do you identify the timeline for the project – when will you accomplish each part?

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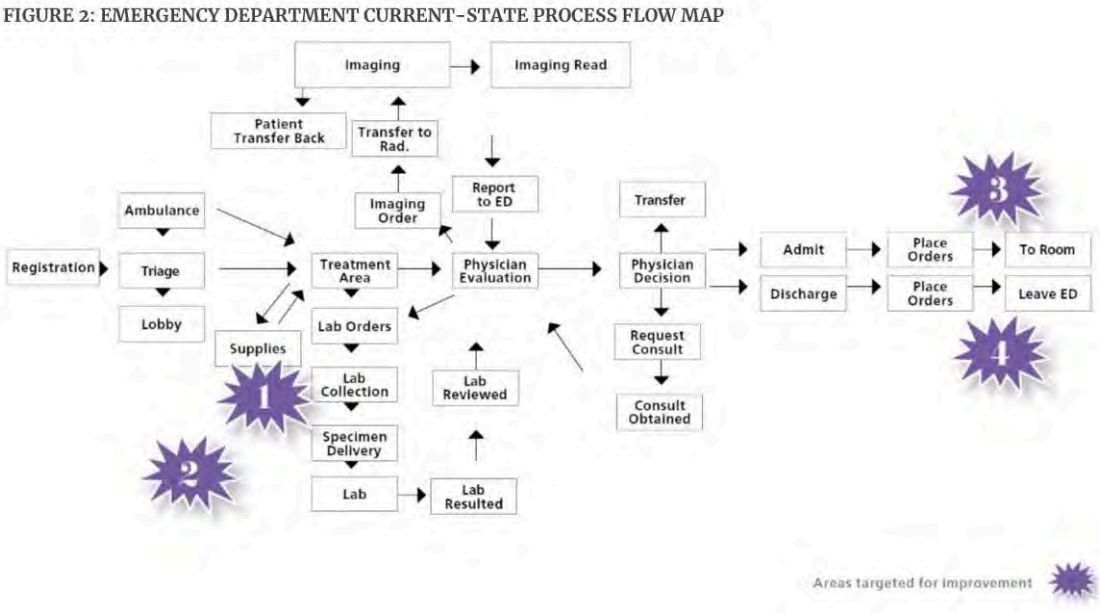
c. Understand strategies to optimize identification of key drivers and interventions to achieve a specific aim

- To increase the odds of a successful quality improvement process, aims and drivers should be identified
- Aim
  - o clearly stated goal or objective of the work describing the desired outcome
- Primary drivers
  - o Key driver, or system components, which contribute directly to achieving the aim
- Secondary drivers
  - o Elements of the associated primary drivers that help create change
  - o Secondary drivers are actions, interventions, subcomponents which are expected to affect primary driver and thus the aim
- Primary drivers and interventions are identified through experts, evidence, and team members involved in the process

d. Understand tools to facilitate completion of quality improvement work, including key driver diagrams and process maps

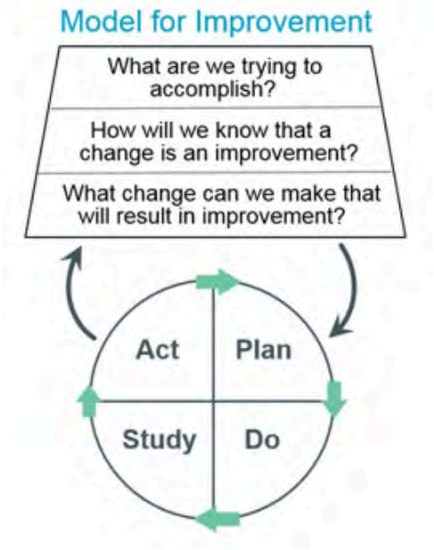
- Key Driver Diagram is a visual display which shows the causal relationship between the secondary driver, primary driver, and the aim
  - o Tool for building a hypothesis that can be tested
  - o Systematically lays out aspects of an improvement project
  - o Developed by team member consensus

- Process Mapping is a visual illustration of the entire process
  - Provides insight into systems and processes in which improvement interventions are introduced
  - Used to help identify where problems are and identify area for improvement
  - Example of a process map:



e. Understand each phase of a Plan-Do-Study-Act (PDSA) cycle

- The Plan-Do-Study-Act Cycle is a method for testing a change in the real work setting
  - It is the scientific method used for action-oriented learning
  - Step 1 Plan: Plan the test or observation, including a plan for data collection

        - State objective of test
        - Make predictions
        - Have a plan to test the change
    - Step 2 Do: Try out the test on a small scale
        - Carry out test
        - Document problems and unexpected observation
        - Begin analysis of the data
    - Step 3 Study: Analyze data and study results
        - Complete the analysis of the data
        - Compare results to predictions
        - Reflect on what you learned
    - Step 4 Act: Based on what you learned from the test refine the change
        - Determine what modifications should be made
        - Prepare a plan for next test
    - 

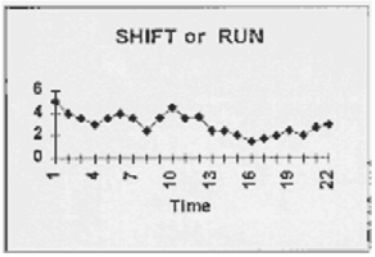
2. Data and Measurement

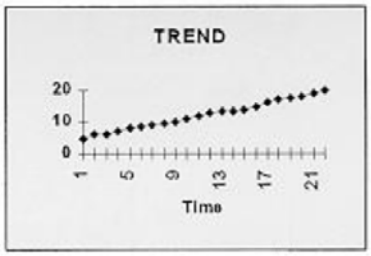
a. Differentiate between process, outcome, and balancing measures

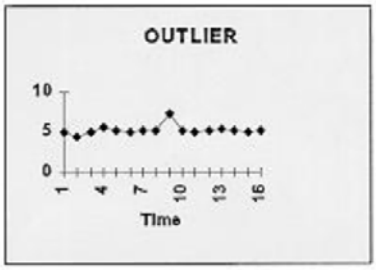
- Different types of measures are used to determine whether QI projects had the desired impact
    - Process Measure: the way the system and processes work to deliver the desired outcome
        - Ex: percentage of patients with diabetes whose A1c was measured twice in the last year, percentage of clinicians washing hands with each new visit
    - Outcome Measure: reflects the impact on the patient and demonstrate the end result of your improvement work and whether it has ultimately achieved the aim set.
        - Ex: reduced mortality, reduced hospital acquired infection
    - Balancing measures: recognition of unintended and or wider consequences of change
        - Ex: reducing patient length of stay in hospital after tonsillectomy and making sure readmissions rates are not increasing

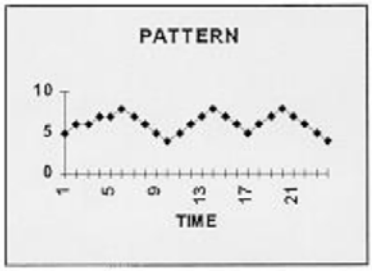
b. Interpret a run chart and identify shifts, trends, and outliers in data

- Data collected about a work process can be illustrated as a run chart
  - Run chart is used to see if the work performed is in a consistent way or if there are changes as the work progresses over time
  - Data for a measured process is plotted against work done over a period of time
    - Y-axis is the item being measured, X-axis is time
  - Run charts are used to identify shifts, trends, patterns, and outliers
    - Shift or Run = process change in which the average or center line shifts
    - Trend = change in the process where valves move in the same direction over time
    - Outlier = a value that lies significantly outside the range of the rest of the data
    - Pattern = any non-random result that repeats over time



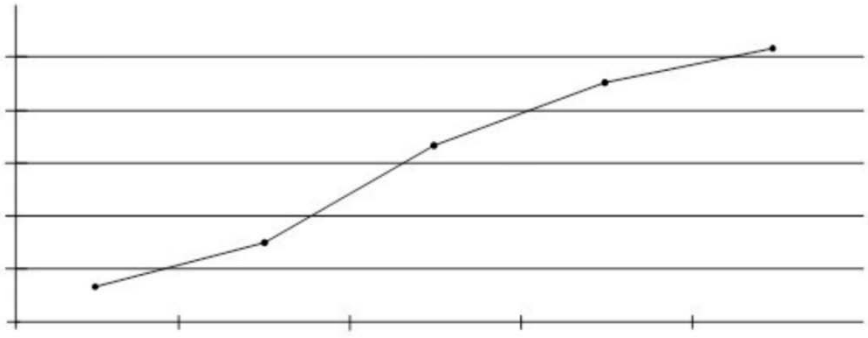




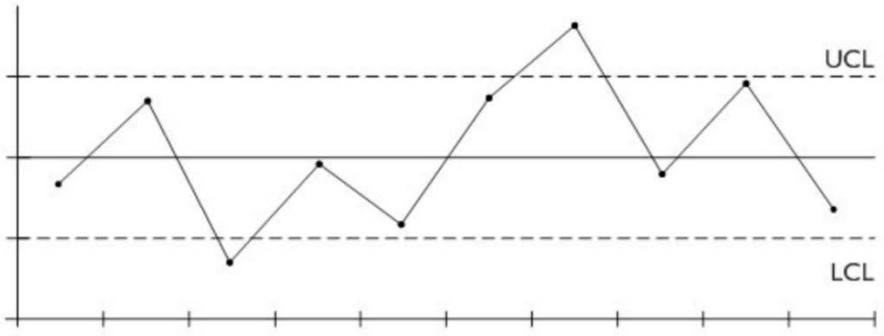


c. Differentiate between a run chart and a control chart

- Run chart is a way to illustrate a work process over a period of time
  - Shows data points over time, which helps to identify runs, trends, outliers, and patterns
  - Shows the general picture of a process



- Control chart
    - Used to determine whether a process is stable or have predictable performance
    - Has an upper and lower limit to determine acceptable range of the test result
    - Illustrates how a process behaves over time and defines the acceptable range of results
        - If a data point is outside the control limit lines, system changes may be required
    - Control charts are designed to prevent
        - Adjusting a process when it should be left alone
        - Ignoring the process when it may need to be adjusted



    - UCL = upper control limit, LCL = lower control limit

d. Differentiate between common cause and special cause variation

- Variance indicates how data is distributed compared to an expected value or mean
- Common cause and special cause variation can be identified on control charts
    - Common cause variation = natural or random variations in a process, “noise.” It is ongoing, consistent, and predictable
        - These variations usually lie within three standard deviations of the mean
        - On control charts they are indicated by a few random points that are within the control limits
    - Special cause variations = unexpected and unpredictable variations that results from an unusual occurrence
        - The cause is usually related to some defect in the system or method
        - On a control chart the points lie beyond the control limit or a non-random pattern
        - The issue which caused the special cause variation should be identified and corrected
    - Common cause variation should not be corrected for, while special cause variation should be corrected for

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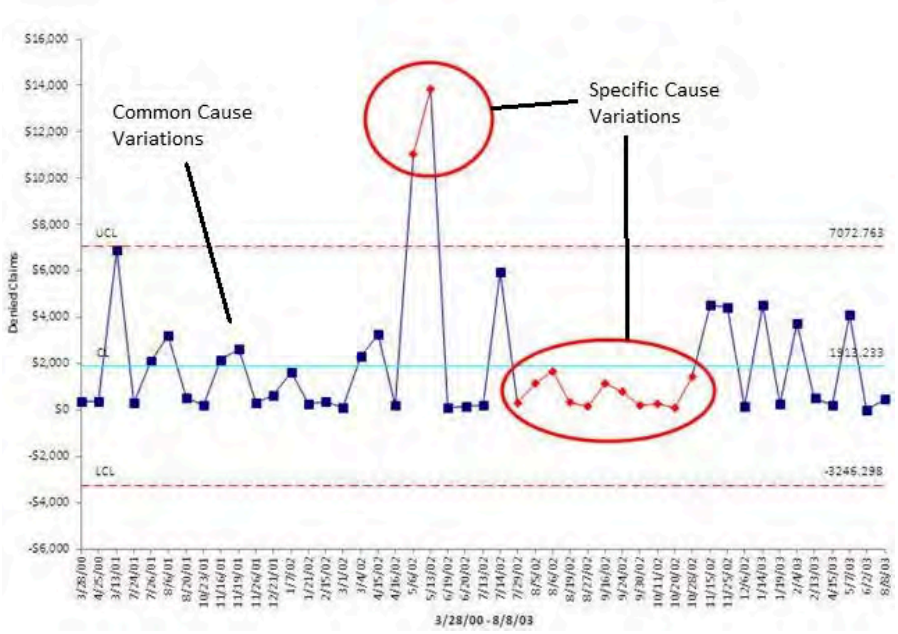
Common Cause Variations
Specific Cause Variations
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Denied Claims
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KEEP
CALM
AND
DEVO ON!