Learning Objectives
At the end of this presentation, the participant should be able to:
- Cite pertinent clinical information regarding the definition/classification, diagnosis, and treatment of cognitive impairment, (Fetal alcohol, Fragile X, Down syndrome), autism, cerebral palsy, myelomeningocele, and attention deficit hyperactivity disorder
- We’ll focus on CI and Autism during this time

Cognitive Impairment - Definition
- IQ score that is more than 2 standard deviations below the mean for age (i.e. IQ of 70) on a standardized IQ test, plus
- Poor performance in daily adaptive behavior (must have significant limitation in adaptive functioning in at least 2 of these areas: communication, self-care, social skills, self-direction, academic skills, work, leisure activities, health, and safety).
- These limitations must have developed before 18 years of age to meet the old definition of "mental retardation".
Old MR/ Cognitive Impairment - Classification

- 70-85= Borderline normal intelligence
- 55 to 69= Mild MR (85% of all cases)
- 40 to 54= Moderate MR
- 25 to 39= Severe MR
- <24= Profound MR

Cognitive Impairment - Presentation

- The most common presenting symptom of Cognitive Impairment is:
  - Language delay, if identified after age 2 years
  - Gross motor delay, if identified before 18 months

Cognitive Impairment - Presentation

- The age at which delay in cognitive development first becomes apparent usually correlates with degree of cognitive impairment
  - <1 year of age predicts severe or profound MR/CI
  - After 2 years of age predicts mild MR/CI
  - Mild MR/CI and borderline intelligence may not be recognized until the child enters school
Cognitive Impairment- Comorbidities

- Important because they may be the presenting complaint
- Neurobehavioral disturbance
- Psychiatric problems
- Speech/language
- Hearing/vision
- Seizures
- Motor coordination problems
- Sleep disturbance

Cognitive Impairment- Cause

- The more severe the CI (especially severe to profound), the more likely a cause will be identified
- Specific genetic cause is found in 47% of severe MR, but 5% of mild MR
- Fetal alcohol spectrum disorder (FASD) is the leading preventable cause
- Fragile X and Down syndrome are the leading gene-related causes
- 1/3 of children with identifiable causes of severe intellectual impairment have Down syndrome, Fragile X, or FASD
- 7-9% chance of recurrence in a second child if there is one severely MR child of unidentifiable etiology

Cognitive Impairment- Evaluation

- Etiology is often difficult to determine
- Medical assessment
  - clinical history, three-generation family history, dysmorphologic examination, neurologic examination
  - Fragile X molecular genetic testing/ fluorescent in-situ hybridization (FISH) studies for subtelomere chromosome rearrangements/ molecular genetic testing
  - Consider microarray for single gene disorders
  - Magnetic resonance brain imaging as indicated
  - Targeted studies for metabolic disorders
  - Also consider: EEG and serum lead levels
CI – Treatment and Management

- Educational Program: IEP age 3-21
- Behavior Problems
- Foster Independence
- Transitions: plan for adulthood
- Family Support – Informal/Formal
- Impacts to Siblings

Cognitive Impairment-Prognosis

- Children with mild CI can be expected to live independently, read at a 4th to 6th grade level, hold down a competitive semi-skilled job, and be physically and psychologically capable of parenthood
- Children who have moderate mental retardation may require some degree of support and supervision as adults. They will have pre-kindergarten academic skills.
- Individuals with severe to profound MR generally require substantial support, live in a group home setting or with family, and are incapable of managing their own affairs
- Adult outcome cannot be predicted by IQ alone, because it does not account for social adaptation and functioning

Cognitive Impairment: Summary

- IQ less than 70 plus adaptive behaviors that are delayed.
- Severity of impairment helps to predict likelihood of finding a specific genetic cause or definable etiology
- Both nature and nature contribute: Mom’s who did not complete high school are 4x more likely to have children with mild intellectual impairment
- Comorbidity is often medically treatable: 20% of severe CI also have cp, or seizures, and 50% have mood, autism spectrum or self injurious behaviors
- Education and behavioral support, and connections to leisure and social and family supports are embraced in Medical Home provision
Fetal Alcohol Spectrum Disorder

- 5 categories in FASD
  - Fetal Alcohol Syndrome with confirmed alcohol exposure
  - FAS without confirmed exposure
  - Partial FAS with confirmed exposure
  - Alcohol related birth defects (AFBD)
  - Alcohol related neurodevelopmental disorder (ARND)
- Effects most likely with early exposure, and as a result of binge drinking (4 or more drinks)
- Prevalence is approximately 1 in 100 live births (Burbacher & Grant 2006)

FASD

- FAS with confirmed alcohol exposure
  - A. Confirmed maternal alcohol exposure
  - B. Evidence of characteristic facial anomalies
    - Short palpebral fissures
    - Flat upper lip
    - Flattened philtrum and midface
  - C. Evidence of growth retardation
    - Low Birth Weight
    - Decelerating weight gain over time not related to nutrition
    - Disproportionate low weight to height ratio
  - D. Evidence of CNS neurodevelopmental abnormalities

FASD Criteria (continued)

- D. CNS neurodevelopmental abnormalities
  - Decreased cranial size at birth
  - Structural brain abnormalities
    - Microcephaly
    - Agenesis of the corpus callosum
    - Cerebellar hypoplasia
  - Hard or soft neurologic signs
    - Hearing loss
    - Fine motor coordination
    - Difficulty with tandem gait
    - Eye hand coordination difficulty
### Partial FAS

- Has confirmed exposure
- Some components of B: facial anomalies
- Either C or D or E

- E. Evidence of complex pattern of behavioral or cognitive abnormalities
  - Early impairments in cognition and language
  - Later appearing learning, behavioral and social problems
  - Poor impulse control
  - Poor social perception
  - Poor capacity for abstraction
  - Specific deficits in math, memory, attention or judgment

### Alcohol-related Birth Defects (ARBD)

- Confirmed alcohol exposure AND
- Cardiac: ASD, VSD, Tetralogy of Fallot
- Skeletal: hypoplastic nails, clinodactyly, pectus, radial-ulnar stenosis, flexion contractures, scoliosis
- Renal: aplastic, dysplastic, hypoplastic, horseshoe, hydronephrosis
- Ocular: strabismus, refractive problems
- Auditory: neurosensory or conductive loss

### Alcohol-related Neurodevelopmental Disorder (ARND)

- Confirmed alcohol exposure AND either
- Evidence of CNS neurodevelopmental abnormalities (D.)
- Evidence of complex pattern of behavioral or cognitive abnormalities (E.)
- Or both

- ARND and ARBD may exist separately or may coexist
FAS in Adolescents and Adults
Clinical Implications

- Poor judgement - Easily Victimized
- Attention deficits - Unfocused/distractible
- Arithmetic disability - Can't handle money
- Memory problems - Doesn't learn from experience
- Difficulty abstracting - Doesn't understand consequences
- Disoriented in time and space - Fails to perceive social cues
- Poor frustration tolerance - Quick to anger

Potential “Secondary Disabilities”
- Early school drop-out
- Alcohol and drug abuse problems
- Having children they can't care for
- Joblessness
- Homelessness
- Trouble with law
- Mental health problems
- Premature death

Fetal Alcohol Spectrum Disorder Summary
- More dysmorphic features notable at re-exam at 12 years
- Head and general growth affected
- Dysmorphology of heart, joints, face
- Neurobehavioral features include cognitive impairment, but also poor judgment and lack of abstraction and learning from experience makes these a vulnerable group of adolescents and adults
Fragile X Syndrome

- FMR1 gene carried on the X chromosome
  - repeating units of CGG in the gene
  - >200 Fragile X
  - 60-200 Premutation
- Males more severely affected (IQ 50-70) but females may have borderline normal
- IQMost common heritable cause of CI
- Premutation females may have early menopause or fertility challenges.

Fragile X- PE features

- Large ears
- Soft skin
- Low muscle tone
- Flexible joints
- Long face
- Larger testicles (after puberty)
- Seizures (20%)

Fragile X- Behavioral

- Speech and language delay
- Delayed crawling, walking, toileting
- Tactile defensiveness
- Impulsivity
- Autism spectrum disorders
- Cognitive impairments
  - Borderline to Mild MR
- Family hx of CI or LD or ASD’s
- Family hx of adult onset ataxia, tremor
Summary Fragile X

- FMR1 gene
- Males and Females are fertile
- Premutational carriers have challenges to fertility (shorter follicular stage, FSH abnormalities)
- Autism common, seizures 20%
- Connective tissue differences: soft skin, flat feet, pectus excavatum, high palate

Down Syndrome

- Trisomy 21- 95% sporadic
- Dysmorphology
  - Small brachycephalic heads
  - Epicanthal folds, upslanting palpebral
  - Flat nasal bridge
  - Single transverse palmar crease, wide space between great and second toes
  - Clinodactyly
- Congenital malformations
- Cognitive impairments

Down syndrome- Med conditions

- Cognitive Impairments – variable
- Hearing loss risk- 75%
- Obstructive sleep apnea – 50-79%
- Eye disease – 60% (cataract, refractive err)
- Congenital heart defects – 50%
- Neurologic dysfunction – 1-13%
- GI atresias – 12%
- Hip dislocations – 6%
- Thyroid dysfunction – 4-18%
Down Syndrome: med conditions

- Transient myeloproliferative disorder 4-10%
- Leukemia 1%
- Autism 1%
- Hirschprung Disease <1%
- Delayed dental eruption 25%
- Celiac disease 5%

Down syndrome management

- Fish studies detect an extra 21 but don’t identify translocations that may have genetic implications for other progeny
- Private room for informing re diagnosis
- Echocardiogram
- Feeding problems: swallowing imaging
- Eval for cataracts
- Hearing screening: follow up by 3 months
- Car seat challenge for hypotonia related airway obstruction

Down syndrome management

- Constipation: consider restricted diet or limited fluid intake, hypothyroidism, gi tract malformations, Hirschprung’s disease
- GERD: diagnosed clinically and managed medically unless contributing to cardiorespiratory problems, or failure to thrive when subspecialist assessments
- Stridor: consider airway anomalies
- Hematologic: leukemoid reactions usually regress in 3 months
Down syndrome guidelines

- New Health Supervision Guidelines
- Changes: sleep study for OSA by 4 y
- Atlanto-axial cervical spine films only for symptomatic children
- Use regular World Health Organization growth curves and monitor for appropriate growth for height
- AAP guidelines published in Pediatrics July 2011 online

Autism/PDD

- Autism diagnostic criteria will be changing with the DSM-5
- Autism spectrum disorder will encompass Autism, Aspergers, PDD and Rett’s and Child Disintegrative Disorder moved
- Language impairment will be combined with social impairments
- Rigidity and stereotypies will be the second component

Autism/PDD-Classification

- As defined by the Diagnostic and Statistical Manual- 4th Edition (DSM-IV), there are 5 diagnoses under this category
  - Autistic Disorder
  - Asperger Disorder
  - Pervasive Developmental Disorder- Not Otherwise Specified (PDD-NOS)
  - Rett Syndrome *
  - Childhood Disintegrative Disorder *
Rett’s Disorder

- Females (few males, but questionable)
- Normal head circumference at birth
- Normal development to 6 months
- Between 6m and 30m, head growth decelerates and loss of language and social skills
- Purposeful hand movement is lost
- Stereotyped hand movements develop
  Classically hand wringing or washing

Rett’s Disorder

- Neuropathology shows failure of neuronal maturation (small neurons with too few dendritic arbors) but NO evidence of a progressive degenerative process
- Mutations in MECP2 gene in more than 80% of females
  - Seizures and Severe MR
  - Breathing disturbances (apnea, hyperventilation, breath-holding, air swallowing
  - Loss of Motor skills but ambulatory until latest stages
  - Risk of sudden death, life expectancy shortened

Childhood Disintegrative Disorder

- Normal development for at least 2 years (age appropriate communication, social relatedness, play and adaptive behaviors)
- Clinically significant loss of at least 2 skills in the areas of receptive or expressive language, social skills, toileting, play, or motor skills
- Present with behavioral features of autism but delayed onset
### Autism/PDD-Diagnosis

- By DSM-IV criteria, symptoms of these conditions fall into three categories:
  - Qualitative impairment in social interaction
  - Qualitative impairment in verbal and nonverbal communication
  - Restrictive, repetitive, stereotypic patterns of behavior, interests, and activities
- Each of the 5 conditions listed under “Classification” requires differing numbers and types of each of the twelve total symptoms listed

### DSM IV Criteria Autistic Disorder

- At least 2 “Impaired social interaction”
  - Marked impairment in the use of nonverbal behaviors, such as eye contact, gestures to regulate social interaction, and facial expression
  - Failure to establish developmentally appropriate peer relationships
  - Lack of spontaneous sharing of enjoyment or interests with others
  - Lack of social or emotional reciprocity

### DSM IV Criteria: Autistic Disorder

- At least 1 “impairment in communication”
  - Delayed or absent speech, not accompanied by compensatory nonverbal communication
  - If speech is present, inability to initiate or sustain a conversation with others impaired
  - Stereotyped and repetitive language, echolalia, or idiosyncratic language
  - Lack of developmentally appropriate imaginary play or social imitative play
DSM IV Criteria: Autistic Disorder

- At least 1 “restricted behavior and interest”
  - Restricted pattern of interest that is abnormal in focus or intensity (such as learning numerous facts about dinosaurs or trains)
  - Rigid adherence to nonfunctional routines and/or rituals (such as same sequence of putting on clothes, or path followed to school)
  - Repetitive and stereotyped motor movements, such as hand flapping or body rocking, that are inappropriate for activities in which the child is engaged
  - Preoccupation with parts of objects, often overlooking the function or purpose of the total object (such as the wheels of a toy car or train, or buttons on clothing)

Autism Spectrum Disorders

- Onset of symptoms prior to 3 years, and not accounted for by Rett’s disorder or CDD
  - Autism
    - 6 symptoms, at least 2 socialization and at least 1 from communication and 1 from restricted interests/activities
  - Asperger’s
    - Language typical or advanced (2 word phrases by 2 years and 3 word phrases by 3), but 2 symptoms from social reciprocity, and one from habitual behaviors
  - PDD-NOS
    - Symptoms on the spectrum but do not have at least 6 or the correct distribution to meet autism criteria

Autism Diagnosis

- Currently, most children diagnosed 3y and over
- Parents bring to attention: absent or delayed speech
- Social skill deficits and lack of preverbal gestures are present before 18 months of age
- Using early “red flags” one study suggests 50% of children can be identified by 14 months of age
- Newer AAP Guidelines suggest screening children twice before 2 years of age for signs and symptoms of autism
Modified Checklist for Autism in Toddlers (M-CHAT)

1. Does your child enjoy being swung, bounced on your knee, etc.? Yes No
2. Does your child take an interest in other children? Yes No
3. Does your child like climbing on things, such as up stairs? Yes No
4. Does your child enjoy playing peek-a-boo/hide-and-seek? Yes No
5. Does your child ever pretend, for example, to talk on the phone or take care of dolls, or pretend other things? Yes No
6. Does your child ever use his/her index finger to point, to ask for something? Yes No
7. Does your child ever use his/her index finger to point, to indicate interest in something? Yes No
8. Can your child play properly with small toys (e.g. cars or bricks) without just mouthing, fiddling, or dropping them? Yes No
9. Does your child ever bring objects over to you (parent) to show you something? Yes No
10. Does your child look you in the eye for more than a second or two? Yes No
11. Does your child ever seem oversensitive to noise? (e.g., plugging ears) Yes No
12. Does your child smile in response to your face or your smile? Yes No
13. Does your child imitate you? (e.g., you make a face—will your child imitate it?) Yes No
14. Does your child respond to his/her name when you call? Yes No
15. If you point at a toy across the room, does your child look at it? Yes No
16. Does your child walk? Yes No
17. Does your child look at things you are looking at? Yes No
18. Does your child make unusual finger movements near his/her face? Yes No
19. Does your child try to attract your attention to his/her own activity? Yes No
20. Have you ever wondered if your child is deaf? Yes No
21. Does your child understand what people say? Yes No
22. Does your child sometimes stare at nothing or wander with no purpose? Yes No
23. Does your child look at your face to check your reaction when faced with something unfamiliar? Yes No

Autism Diagnosis Prior to Age 3

- Stone and associates have proposed applying just 4 of the possible DSM-IV criteria to children younger than 3 years:
  - Lack of spontaneous seeking to share enjoyment, interests, or achievements with other people (e.g. by lack of showing, bringing, or pointing out objects of interest)
  - Lack of social and emotional reciprocity
  - Marked impairment in the use of multiple nonverbal behaviors, such as eye-to-eye gaze, facial expression, body postures, and gestures to regulate social interaction
  - Delay in or total lack of the development of spoken language (not accompanied by an attempt to compensate through alternative modes of communication such as gesture or mime).

- Stone’s group propose that if all 4 of these criteria are present, that we make a “provisional diagnosis of autism” in the child under 2 years of age, with the recommendation for a re-evaluation using the full DSM-IV criteria after the 3rd birthday.

RED FLAGS for ASD’s before 2 yrs

- Are social and language development comparable to gross and fine motor, self care and cognitive skills?
- Does the child have “joint attention skills?”
  - Gaze averting 8-10 months
  - Follow a point 10-12 months
  - Point to request 12-14 months
  - Point to comment 14-16 months
RED FLAGS for autism before 2yrs

- Shared joint attention, and pointing to share interest in an object and then looking back to re-establish eye contact with the adult is a skill that should be present by 18 months of age (AKA protodeclarative pointing)
- Does the child recognize his name and respond by turning his head toward the speaker? Should be present by 10 mos.
- Does the child have simple pretend play? Should be present by 16 mos.

Autism- Epidemiology/ Etiology

- Incidence/ Prevalence increasing
  - Prevalence: 1:88
  - Male: Female 4:1
- Strongly genetic
  - Possibly partly causal in 95% of cases
  - Candidate genes: X, 7p, 7q, 15q 11-13 , PTEN, 22q

Autism/PDD-Presentation

- Key features are impaired social relatedness and language delay or lack of pragmatic language
- Presenting signs are listed on page 320
- Also may be seen along with various syndromes (FXS, Down Syndrome, Tuberous Sclerosis, CHARGE, TORCH, others), and with other CNS abnormalities
Autism/PDD-Recommended Workup

- "Medical" Evaluation
  - Physical exam for dysmorphic or neurocutaneous clues to genetics
  - History for regressions, seizures, sleep or dietary problems
  - Fragile X and microarray for single gene deletions
  - Targeted metabolic testing
  - Audiology

- Diagnostic Testing (Gold Standard)
  - ADI-R (Autism Diagnostic Interview) and ADOS (Autism Diagnostic Observation Schedule)
  - Formal Speech and language assessment
  - IQ testing

Autism/PDD-Treatment

- Genetic counseling: 3-6% recurrence risk
- Parent counseling/education
- Coordinated educational plan
- Behavioral counseling/therapy
- Speech/Occupational/Social Skills Therapy
- Pharmacologic therapy
  - Medication can be a beneficial adjunctive therapy, especially when it is used to treat such comorbidities as anxiety and depression, seizures, aggression, attention, or self-injurious behavior.

Cerebral Palsy-General Issues

- Definition: Cerebral palsy is persistent impaired motor function or posture that is evident at birth or becomes evident during early infancy, is nonprogressive (i.e., is a static encephalopathy), and is usually not genetic, although it may be part of a genetic syndrome.
  - If it is not clear that a child’s condition is nonprogressive, do not label it CP
- Incidence: 2 per 1,000 live births
  - In preterm infants incidence is inversely proportional to gestational age
- Prevalence: 1.2 to 2.5 children per 1,000 by early school age
Cerebral Palsy - Risk Factors

- Antenatal: Brain malformations, chromosomal abnormalities, multiple gestations, placental insufficiency, intrauterine infection, prematurity
- Perinatal: Intrapartum asphyxia, birth injury
- Postnatal: Intracerebral hemorrhage, cerebral infection, traumatic brain injury, hypoxia
- Intrapartum asphyxia leads to less than 10% of cases
- Etiology is unknown in the vast majority of cases

Cerebral Palsy: presentation

- Gross motor delays
- Upper motor neuron physical findings:
  - brisk deep tendon reflexes,
  - increased distal muscle tone,
  - diminished trunkal tone
- Persistent primitive reflexes (Moro, ATNR, tonic labyrinthian)
- Delayed acquisition of protective reflexes (head righting, parachute)

Cerebral Palsy - Classification

- Four subtypes
  - Spastic or “Pyramidal” (65%)
  - Dyskinetic or "Extrapyramidal" (25%)
  - Ataxic (5%)
  - Hypotonic (<5%)
  - (Mixed – 20%)
Cerebral Palsy - Spastic Subtype

- Involves injury of upper motor neurons
- Characterized by weakness, increased muscle tone, increased deep tendon reflexes, clonus, "clasp-knife" tightness or spasticity, extended "scissored" (crossed) legs, and a tendency toward contractures

Subtypes
- Quadriplegia (AKA quadraparesis)- All 4 limbs
- Diplegia- Legs> arms
- Hemiplegia- Both limbs, same side, UE worse

Cerebral Palsy - Diagnosis

- Gross motor findings emerge over the first year of life: floppy, irritable, poor feeders should raise red flags
- Tone, primitive reflexes, and deep tendon reflexes are the mainstays of physical diagnosis for CP
- Central nervous system still maturing during the first year of life: allow for corrected gestational age in premature babies, and allow for "transient dystonia"

Cerebral Palsy - Comorbidities

- Mental retardation (50-60%)
  - 15% of affected children have mild MR, 35% with moderate, and 50% with severe
- Hearing deficits (30%)
- Visual impairment (esp strabismus)
- Seizures (30-50%, most often with hemiplegic form)
- Skeletal deformities
- Oral-motor dysfunction
- Poor growth, nutritional difficulties
- Drooling
## Cerebral Palsy - Treatment

- **Mainstay:** Improving facilitating functional outcomes
  - Mobility
  - Communication
  - Correct visual deficits/ strabismus
  - Seizure control
  - Obtain educational assistance
  - Prevent secondary physical/ mental comorbidities

## Management of Spasticity

- Neurosurgical techniques
  - Selective dorsal root rhizotomy
  - Baclofen - Oral, intrathecal by implantable pumps. Dantrolene and diazepam are also used but are sedating.
  - Botulinum toxin to overactive muscle groups
  - Physical therapy/ Positioning devices
  - Orthopedic interventions of tendon lengthening or realignments

## Myelomeningocele

- **Definition:** A form of neural tube defect in which there is lack of fusion of the posterior bony arch(es) of various spinal cord segments, and which may be accompanied by protrusion of a meningeal sac containing part of the spinal cord itself
Myelomeningocele - **Etiology/ Risk Factors**

- Incidence (U.S.): 0.41 to 1.43/1,000 live births
- Risk of recurrence is 3-4% 
  - Two gene abnormalities, PAX3 and 5,10-methylene tetrahydrofolate reductase, have been shown to increase risk
- Folic acid at a dose of 400 mcg/day given to women pre-conceptively seems to reduce the risk of first occurrence cases
  - Doses of 4mg/day reduce the risk of recurrent neural tube defects
- There seems to be an increased incidence with maternal use of valproate, and possibly also with carbamazepine

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Myelomeningocele - **Diagnosis**

- Elevated maternal serum alpha-feto protein level 
  - Done between weeks 15-22
- Prenatal ultrasound
- Physical exam at birth

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**Myelodyplasia: overall pediatric issues to be aware of**

- Most children with have shunts for hydrocephalus.
  - Shunt infections or malfunctions are potentially life threatening.
- Almost all children will have continence issues. (Sacral nerves control sphincters)
  - Urosepsis risks
  - High pressure bladders can produce hydronephrosis and renal failure: yearly RUS
Myelodysplasia: pediatric issues

- Monitor for precocious puberty
- Prevent obesity
- Skin examination
- Know the level of neurosensory deficit:
  - Thoracic level insensate children may not have abdominal pain with intraabdominal processes
  - Broken bones in non-weightbearing extremities may produce fever and mild swelling but cause no pain
  - Decubiti, and sources of infection from lesions to insensate skin

Myelomeningocele- Areas of Clinical Concern

- Neurological Issues
  - Hydrocephalus- Seen in more than 90% of patients, with or without an Arnold-Chiari (Chiari II) malformation
  - Seizures- 20%
  - Tethered cord- High incidence at birth (all?) and in adolescence
  - IQ can be within normal range but is skewed by profound hydrocephalus, recurrent shunt malfunctions, and shunt infections.

Myelomeningocele- Areas of Clinical Concern

- Mobility
  - Ambulation
    - 1/3 ambulate without assistance, 1/3 ambulate with assistance, and 1/3 require a wheelchair for mobility
    - Level of the lesion primarily determines ambulation ability
      - With low lumbar lesions, 95% of adolescents and 40% of younger children will ambulate with assistance
      - With high lumbar or thoracic lesions only 30% of adolescents ambulate with assistance
ADHD-Classification

- **Classification** varies somewhat, but DSM-IV breaks it down into two primary categories, those being predominately inattentive and those being predominately hyperactive-impulsive (there is also a combined type)
  - There are 9 symptoms for each subtype, and 6 symptoms must be present for that subtype to be diagnosed.
  - If 6 symptoms are present for both subtypes, the diagnosis is **combined** type.
- Prevalence is 8-10%
- Male: Female 3:1

ADHD-Classification

- **Combined Type**
  - Most common, studied, and well understood form
  - More commonly seen in males
- **Predominantly Inattentive Type**
  - 2nd most common subtype
  - M:F ratio is lower than for other subtypes (i.e. the M:F ratio is nearly equal)
  - Generally diagnosed later than the other 2 subtypes
  - Has fewer disruptive associated behaviors
- **Predominantly Hyperactive/ Impulsive Type**
  - Most often diagnosed in younger children who have not reached the age where attention deficits are significant (i.e. may be a precursor to the combined type)

ADHD-Diagnosis

- No one single test should be considered diagnostic
- In addition, for a child to meet diagnostic criteria:
  - Symptoms must begin before seven years of age, and have persisted for at least 6 months
  - Symptoms must be present in at least two settings (e.g. home and school)
  - There must be clear evidence of clinically significant impairment in social, academic, or occupational functioning.
  - The symptoms must not be better accounted for as being part of another diagnosis (e.g. mental retardation)
- A positive response to stimulants is NOT diagnostic
### AD/HD - Epidemiology/ Etiology
- **Hereditary** - most important etiologic factor
  - Sibs of affected children are 5-7 times more likely to be affected than controls
  - If one parent has AD/HD, risk is greater than 50% that their children will be affected
  - 3 Implicated genes: D2 and D4 dopamine receptor genes, and DAT1 (dopamine transporter gene)
- **Brain injury, teratogen or toxin exposure, syndromes, and abnormalities of brain structure** all confer increased risk

### ADHD - Other Clinical Issues
- **Comorbidities** are very common and problematic
  - Learning Disabilities
  - Tic Disorders
  - Obsessive Compulsive Disorders
  - Bipolar Disorder
  - Depression
  - Other developmental disabilities, especially speech delay
  - Deficits in motor planning and execution
  - Sleep disruption
  - Accidental and nonaccidental injuries

### ADHD - Treatment
- **First line**: methylphenidate or dextroamphetamine salts
  - Short (4h) or long acting (8h) oral
  - Bimodal oral release (8 h with 2 peaks am and after lunch)
  - Skin patch
  - Prodrug for dextroamphetamine (lysdexamfetamine dimesylate)
  - Active enantiomer for methylphenidate (dexmethylphenidate)
- **Non-stimulant medication** (e.g. atomoxetine) which have an indication for first line treatment
- **Second line**: non-stimulants are also used (antidepressants- SSRI’s- and alpha-2 adrenergic drugs- clonidine, guanfacine)
ADHD-Treatment

- Stimulants- act on dopamine, norepinephrine, and serotonin transporters- primarily dopamine
  - Methylphenidate- Block the dopamine transporter, and slightly increase dopamine release
  - Amphetamines- Greater effect on stored dopamine release
- Atomoxetine- Probably works by blocking reabsorption of norepinephrine
- The neurotransmitters dopamine, norepinephrine, and serotonin all affect attention and behavior to varying degrees, and their roles in ADHD are being studied

Cardiovascular evaluations before use of medication

- AAP policy statement Peds 122(2):451-453 August 2008
- Before starting stimulants:
  - Targeted cardiac history in patient for previously detected cardiac disease, palpitations, syncope, seizures
  - Cardiac family history for sudden death in children or young adults: hypertrophic cardiomyopathy, long QT syndrome
  - Physical examination including careful cardiac exam
- AAP does not recommend routine ECG or routine subspecialty cardiac evaluation.

LAWS GOVERNING ALTERNATIVE EDUCATIONAL PLACEMENT

- Review this in the book (Page 333). (3 questions on this material each year)
- Free and appropriate public education is available to every child now under federal law Individuals with Disabilities Education Act (IDEA)
- Assessment for specialized educational services is called: multifactored educational assessment (MFE)
- Specialized plan is called: individualized educational plan (IEP)
DEVELOPMENTAL SCREENING/ ASSESSMENT TESTS

- Review this in the book (Page 332-333). There are generally 3 questions on this material each year.

Early Intervention

- Created and revised by a series of Federal initiatives starting with Public Law 94-142 in 1975 (see page 310 of the book) and now are included in IDEA
- Provides services including PT/OT/ST, service coordination, audiology and vision services, diagnostic testing, etc
- Serves children with known delays or conditions which predispose for delays from Birth until the 3rd birthday
- Parents may self refer for services, but
- You play a critical role in linking children to these services

Learning Disabilities- Basics

- Diagnosed in 6% of US children
  - Criteria for diagnosis vary in each state, so know those for life but not for Boards
- Reading disability is the most common type
- Vast majority of affected children have average to above average intelligence
- Most common times of diagnosis are during third grade and in junior high school
Learning Disabilities - Basics

- Diagnosis requires Intelligence Testing and Achievement Testing
- IQ: Discrepancy between performance IQ and verbal IQ = at risk for LD
- Discrepancy between Overall IQ (potential) and achievement scores (actual performance) defines a learning disability

Review Questions

Question 1

The diagnosis of mental retardation requires:
1. An IQ score that is more than 2 standard deviations below the mean for age
2. Poor performance in daily adaptive behavior
3. Symptoms must develop before 7 years of age

A. All of the above
B. 1 and 2
C. 1 and 3
D. None of the above
Answer: B

- Point 3 is not true because symptoms must develop before 18 years of age for mental retardation. Symptoms must develop before 7 years of age for Attention Deficit/Hyperactivity Disorder.

Question 2

True statements regarding cerebral palsy include:
1. The etiology is known in most cases
2. The dyskinetic subtype is most common
3. Most involved children do not have mental retardation
4. The condition is often progressive

A. All of the above
B. 1 and 2
C. 1 and 3
D. None of the above

Answer: D

In the case of cerebral palsy, the etiology is unknown in most cases, the spastic diplegic subtype is the most common, 50-60% of involved children also have mental retardation, and the condition must be nonprogressive.
Question 3

True statements regarding myelomeningocele include:
1. Urosepsis is a common complication
2. A majority of patients have a Chiari II malformation
3. The condition is suggested by an elevated prenatal AFP level
4. Folic acid has been shown to be protective

A. All of the above
B. 1 and 2
C. 1 and 3
D. None of the above

Answer: A

All are true statements

Question 4

Autistic Disorder: Language development is disproportionately impaired compared to gross motor, fine motor, and cognitive milestones

A. True
B. False
Answer: A

Autistic Disorder has social and language impairments delayed out of proportion to other developmental achievements, EVEN in children who have both global developmental delay and cognitive impairment in addition to their autism.

Question 5

- The following is NOT a criterion for making the diagnosis of ADHD combined type
  - A. Condition present in more than 1 setting
  - B. Symptom onset before 7 years of age
  - C. Symptoms present for more than 6 months
  - D. Condition results in functional impairment
  - E. Have at least 6 symptoms in each set of Inattention/distractibility, Hyperactivity, and Impulsivity groupings

Answer: E

- At least 6 symptoms from the Inattention/Distractibility AND Hyperactivity
- Plus all the other 4 criteria a-d, and lack of other medical, psychiatric or social condition
- Are required to make the diagnosis of combined type ADHD, which is the most common subgroup