REVIEW OF DEVELOPMENTAL PEDIATRICS
2012
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Rose F. Kennedy UCEDD
Children’s Hospital at Montefiore
• Know Developmental Milestones
• Understand Developmental Disorders
  • Disorders affecting motor development
  • Intellectual Disabilities
  • Communication Disorders
  • Pervasive Developmental Disorders
  • Learning Disorders
  • Associated Behavioral Disorders
Part I: Know the milestones!
BRIGHT FUTURES
Guidelines for Health Supervision of Infants, Children, and Adolescents
THIRD EDITION
A baby is pulled to sit with no head lag, grasps a rattle, and follows an object visually 180 degrees. These milestones are typical for:

1. 2 months
2. 4 months
3. 6 months
4. 8 months
<table>
<thead>
<tr>
<th>Age</th>
<th>Gross Motor</th>
<th>Fine Motor</th>
<th>Cognitive, Linguistic, and Communication</th>
<th>Social-Emotional</th>
</tr>
</thead>
<tbody>
<tr>
<td>2 Months</td>
<td>Head up 45°</td>
<td>Follow past midline</td>
<td>Laugh</td>
<td>Smile spontaneously</td>
</tr>
<tr>
<td></td>
<td>Lift head</td>
<td>Follow to midline</td>
<td>Vocalize</td>
<td>Smile responsively</td>
</tr>
<tr>
<td>4 Months</td>
<td>Roll over</td>
<td>Follow to 180°</td>
<td>Turn to rattling sound</td>
<td>Regard own hand</td>
</tr>
<tr>
<td></td>
<td>Sit—head steady</td>
<td>Grasp rattle</td>
<td>Laugh</td>
<td></td>
</tr>
<tr>
<td>6 Months</td>
<td>Sit—no support</td>
<td>Look for dropped yarn</td>
<td>Turn to voice</td>
<td>Feed self</td>
</tr>
<tr>
<td></td>
<td>Roll over</td>
<td>Reach</td>
<td>Turn to rattling sound</td>
<td>Work for toy (out of reach)</td>
</tr>
<tr>
<td>9 Months</td>
<td>Pull to stand</td>
<td>Take 2 cubes</td>
<td>Dada/Mama, nonspecific</td>
<td>Wave bye-bye</td>
</tr>
<tr>
<td></td>
<td>Stand holding on</td>
<td>Pass cube (transfer)</td>
<td>Single syllables</td>
<td>Feed self</td>
</tr>
</tbody>
</table>

**KEY**

Black Color: 50% to 90% of children pass this item.
Green Color: More than 90% of children pass this item.

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Tanya is now walking well, and can stoop to the floor and get back up. She generally points to indicate what she wants, but can ask for her “bottle”, a “cookie” and her “blankie”. She drinks from a sippy cup and feeds herself cheerios. She places a toy bottle in her doll’s mouth. Tanya is most likely a typically developing:

1. 12 month old
2. 15 month old
3. 18 month old
4. 24 month old
<table>
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<tr>
<th>Age</th>
<th>Gross Motor</th>
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<th>Social-Emotional</th>
</tr>
</thead>
<tbody>
<tr>
<td>1 Year</td>
<td>*Stand alone</td>
<td>*Put block in cup</td>
<td>*Imitate vocalizations and sounds</td>
<td><em>Protodeclarative pointing</em></td>
</tr>
<tr>
<td></td>
<td>*Pull to stand</td>
<td>*Bang 2 cubes held in hands</td>
<td><em>Babbling</em></td>
<td>*Wave bye-bye</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>*1 word</td>
<td>*Imitate activities</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>*Play pat-a-cake</td>
</tr>
<tr>
<td>15 Months</td>
<td>*Walk backwards</td>
<td>*Scribble</td>
<td><em>1 word</em></td>
<td>*Drink from cup</td>
</tr>
<tr>
<td></td>
<td>*Stoop and recover</td>
<td>*Put block in cup</td>
<td>*3 words</td>
<td>*Wave bye-bye</td>
</tr>
<tr>
<td></td>
<td>*Walk well</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>18 Months</td>
<td>*Walk up steps</td>
<td>*Dump raisin, demonstrated</td>
<td>*Point to at least 1 body part</td>
<td>*Remove garment</td>
</tr>
<tr>
<td></td>
<td>*Run</td>
<td>*Tower of 2 cubes</td>
<td>*6 words</td>
<td></td>
</tr>
<tr>
<td></td>
<td>*Walk backwards</td>
<td></td>
<td>*3 words</td>
<td></td>
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- *Absence of these milestones should trigger screening for autism.

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Of the following motor milestones, the one that is MOST typical of a 2 ½ year old child is:

1. Walking up steps
2. Building a tower of 2 cubes
3. Scribbling
4. Imitating a vertical line

![Bar graph showing the percentages for each milestone:]

- Walking up steps: 20%
- Building a tower of 2 cubes: 20%
- Scribbling: 33%
- Imitating a vertical line: 27%
A 3 year old boy should have mastered each of the following except:

A. Naming a red truck
B. Towering 6 cubes
C. Stating his name and gender
D. Hopping on one foot
A 3 year old boy should have mastered each of the following except:

A. Naming a red truck (50%ile ~ 30 mos)
B. Towering 6 cubes (50%ile ~ 20 mos)
C. Stating his name and gender (50%ile ~ 3 yrs)
D. *Hopping on one foot* (50%ile ~ 4 yrs)
On a pre-kindergarten screening a school official is most concerned about a 5 year old boy who cannot:

A. Draw a Person with 6 parts
B. Copy a Square
C. Name 4 colors
D. Tandem Walk
On a pre-kindergarten screening a school official is most concerned about a 5 year old boy who cannot:

A. Draw a Person with 6 parts (50%ile ~4 ½ yrs)
B. Copy a Square (50%ile ~ 5 yrs)
C. *Name 4 colors* (50%ile ~ 3 ¾ yrs)
D. Tandem Walk (50%ile ~ 4 ½ yrs)
<table>
<thead>
<tr>
<th>2 Years</th>
<th>2 1/2 Years</th>
<th>3 Years</th>
<th>4 Years</th>
</tr>
</thead>
</table>
| • Throw ball overhand  
• Jump up  
• Kick ball forward  
• Walk up steps  | • Throw ball overhand  
• Jump up  | • Balance on each foot 1 second  
• Broad jump  
• Throw ball overhand  | • Hop  
• Balance on each foot 2 seconds  |
| • Tower of 6 cubes  | • Imitate vertical line  
• Tower of 8 cubes  
• Tower of 6 cubes  | • Thumb wiggle  
• Imitate vertical line  
• Tower of 8 cubes  
• Tower of 6 cubes  | • Draw a person with 3 parts  
• Tower of 8 cubes  |
| • Name 1 picture  
• Combine words  
• Point to 2 pictures  | • Know 2 actions  
• Speech half understandable  
• Point to 6 body parts  
• Name 1 picture  | • Speech all understandable  
• Name 1 color  
• Know 2 adjectives  
• Name 4 pictures  | • Define 5 words  
• Name 4 colors  
• Speech all understandable  |
| • Put on clothing  
• Remove garment  | • Wash and dry hands  
• Put on clothing  | **Name friend**  
• Brush teeth with help  | • Copy a cross (+)  
• Copy a circle  |

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You would be most concerned about:

A. A one year old who doesn’t stand alone

B. A 15 month old who doesn’t stoop and recover

C. A four year old who cannot hop on each foot

D. A two year old who cannot jump
You would be most concerned about:

A. A one year old who doesn’t stand alone  
   (50-90% of 1 year olds)

B. A 15 month old who can’t stoop and recover  
   (>90% of 15 month olds)

C. A four year old who cannot hop on each foot  
   (50-90% of 4 yr olds)

D. A two year old who cannot jump  
   (50-90% of 2 yr olds)
You would be less concerned about:

A. A 3 year old who cannot answer a “why” question
B. An 18 month old who uses 2 words
C. A one year old who doesn’t point
D. A 9 month old who doesn’t babble
You would be less concerned about:

A. A 3 year old who cannot answer a “why” question (50% ile ~4-5 yrs)
B. An 18 month old who uses 2 words (over 90% of 15 mo olds)
C. A one year old who doesn’t point (over 90% of 1 yr olds)
D. A 9 month old who doesn’t babble (over 90% of 9 mo olds)
### Social and Emotional Development in Middle Childhood

#### Topics

<table>
<thead>
<tr>
<th>Key Areas</th>
</tr>
</thead>
<tbody>
<tr>
<td>(Key areas in italics are especially important for children with special health care needs.)</td>
</tr>
</tbody>
</table>

**Self**

- Self-esteem:
  - Experiences of success
  - Reasonable risk-taking behavior
  - Resilience and ability to handle failure
  - Supportive family and peer relationships

- Self-image:
  - Body image, *celebrating different body images*
  - Prepubertal changes; initiating discussion about sexuality and reproduction; *prepubertal changes related to physical care issues*

**Family**

- What matters at home:
  - Expectation and limit setting
  - Family times together
  - Communication
  - Family responsibilities
  - Family transitions
  - Sibling relationships
  - Caregiver relationships

**Friends**

- Friendships:
  - Making friends, *friendships with peers with and without special health care needs*
  - Family support of friendships, *family support to have typical friendship activities, as appropriate*
<table>
<thead>
<tr>
<th>Topics</th>
<th>Key Areas</th>
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<tbody>
<tr>
<td></td>
<td><em>(Key areas in italics are especially important for children with special health care needs.)</em></td>
</tr>
<tr>
<td></td>
<td>• Expectation for school performance, <em>school performance defined in the Individualized Education Program (IEP)</em></td>
</tr>
<tr>
<td></td>
<td>• Homework</td>
</tr>
<tr>
<td></td>
<td>• Child-teacher conflicts, <em>building relationships with teachers</em></td>
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<tr>
<td></td>
<td>• <em>Parent-teacher communication</em></td>
</tr>
<tr>
<td></td>
<td>• Ability of schools to address the needs of children from diverse backgrounds</td>
</tr>
<tr>
<td></td>
<td>• Awareness of aggression, bullying, and victimization</td>
</tr>
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<td></td>
<td>• Absenteeism</td>
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<tr>
<td>Community</td>
<td><strong>Community strengths:</strong></td>
</tr>
<tr>
<td></td>
<td>• Community organizations</td>
</tr>
<tr>
<td></td>
<td>• Religious groups</td>
</tr>
<tr>
<td></td>
<td>• Cultural groups</td>
</tr>
<tr>
<td>High-risk behaviors</td>
<td><strong>High-risk behaviors and environments:</strong></td>
</tr>
<tr>
<td></td>
<td>• Substance use</td>
</tr>
<tr>
<td></td>
<td>• Unsafe friendships</td>
</tr>
<tr>
<td></td>
<td>• Unsafe community environments</td>
</tr>
<tr>
<td></td>
<td>• <em>Particular awareness of risk-taking behaviors and unsafe environments, because children may be easily victimized</em></td>
</tr>
<tr>
<td>Domain</td>
<td>Early Adolescence (11 to 14 Years)</td>
</tr>
<tr>
<td>-----------------</td>
<td>--------------------------------------------------------------------------------------------------</td>
</tr>
<tr>
<td>Physiological</td>
<td>Onset of puberty, growth spurt, menarche (females)</td>
</tr>
<tr>
<td>Psychological</td>
<td>Concrete thought, preoccupation with rapid body changes, sexual identity, questioning independence, parental controls remain strong</td>
</tr>
<tr>
<td>Social</td>
<td>Search for same-sex peer affiliation, good parental relationships, other adults as role models; transition to middle school, involvement in extracurricular activities; sensitivity to differences between home culture and culture of others</td>
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<td>Domains of Adolescent Development</td>
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The Individuals with Disabilities Education Act (IDEA) requires states to provide:

A. Evaluations of “at risk” children ages 0-3
B. Services in the natural environment
C. Family-centered services to infants and toddlers
D. A “child-find” component
E. All of the above
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E. All of the above
ENTITLEMENTS

1975: PL 94-142, Education for All Handicapped Children Act: Free and appropriate public education to all students with disabilities, ages 6-17

1986: PL 99-457, Part B (ages 3-5), Part H (ages 0-3)

1991: IDEA, Individuals with Disabilities Education Act

PART C: “Early Intervention”

- Birth to age 3
- Child Find
- Evaluation and therapeutic services for children with developmental delays
- Evaluation and follow-up of “at risk” children
- Multidisciplinary coordinated interagency model
- IFSP: family is central focus of service
- Services to be provided in “natural environment”
• **IDEA- Part B (to age 21)**
  • Provides special services for children whose disabilities severely affect their educational performance
    – specific LD, serious emotional disturbance, other health impaired…
    – Individual Educational Plan (IEP)
    – Least restrictive environment

• **Section 504 of Vocational Rehabilitation Act of 1973**: Provides accommodations for students with a physical or mental impairment that substantially limits one or more major life activity
PART 2: Know the Presentation of Developmental Disorders, and their differential diagnoses

- Infant/toddlers with motor delay
- Toddler/preschoolers with language delay
- School age children with school failure
You are evaluating a 9 month old baby who is not yet sitting without support. She is a former 26 week premature infant. Brain MRI reveals periventricular leukomalacia. Of the following findings, which would you most likely expect to see:

1. Increased tone in all 4 extremities, UE>LE
2. Equally increased tone in all 4 extremities
3. Dyskinetic, choreoathetoid movements
4. Increased tone in all 4 extremities, LE>UE
5. Increased tone in the right upper extremities compared with the left

![Bar chart showing percentages]

- 1: 23%
- 2: 27%
- 3: 13%
- 4: 13%
- 5: 23%
Motor Delay in Infancy

Rule out:

- Neurological Disorders
- Genetic Disorders
- Metabolic Disorders
- Systemic Illness
Cerebral Palsy

- Disorder of Movement and Posture
- Results from a nonprogressive brain injury or developmental deficit of brain
- Injury occurs during the period of brain growth
- Diagnosis of exclusion
- Associated with conditions reflecting CNS insult: MR, Seizures, Sensory Deficits
- Classified by type
  - Spastic: most common: sub-classified by distribution
  - Dyskinetic, Hypotonic, Mixed
### Selected Clinical Findings or Laboratory Abnormalities Suggesting a Metabolic Disorder


<table>
<thead>
<tr>
<th>Clinical Finding or Laboratory Abnormality</th>
<th>Suggested Disorder</th>
</tr>
</thead>
<tbody>
<tr>
<td>Failure of appropriate growth</td>
<td>Arachnodactyly</td>
</tr>
<tr>
<td>Recurrent unexplained illness</td>
<td>Hepatosplenomegaly</td>
</tr>
<tr>
<td>Seizures</td>
<td>Metabolic/lactic acidosis</td>
</tr>
<tr>
<td>Ataxia</td>
<td>Hyperuricemia</td>
</tr>
<tr>
<td>Loss of psychomotor skills</td>
<td>Hyperammononemia</td>
</tr>
<tr>
<td>Hypotonia</td>
<td>Low cholesterol</td>
</tr>
<tr>
<td>“Coarse” appearance</td>
<td>Structural hair abnormalities</td>
</tr>
<tr>
<td>Eye abnormalities (cataracts, ophthalmoplegia, corneal clouding, abnormal retina)</td>
<td>Bone abnormalities (dysostosis, occipital horns, punctuate calcifications)</td>
</tr>
<tr>
<td>Recurrent somnolence/coma</td>
<td>Skin abnormalities (angiokeratoma, “orange-peel” skin, ichthyosis)</td>
</tr>
<tr>
<td>Abnormal sexual differentiation</td>
<td></td>
</tr>
</tbody>
</table>
Parents of a 3 year old girl present with concerns about speech and language delays. Their daughter has a vocabulary of about 10 words, and she recently began pointing to body parts and following single un-gestured commands. She can imitate a vertical line, jump in place, and broad jump. She is able to wash and dry her hands, and put on a t-shirt. In your office, she points to your stethoscope, and when you hand it to her she smiles at you and places it on her father’s chest.
You most strongly suspect:

A. Mental Retardation
B. Autistic Spectrum Disorder
C. Mixed receptive/repressive language disorder
D. Hearing Impairment
E. Environmental understimulation

Your first referral is to:

A. Social service
B. Audiology
C. Psychology
D. Speech and Language Pathology
You most strongly suspect:

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Language Delay in a Toddler/Preschooler

CONSIDER:

• Hearing Impairment
• Global Developmental Delay: Intellectual Disability
• Communication Disorders
• Pervasive Developmental Disorders
• Environmental Factors
• General Health
Language Delay in a Toddler/Preschooler

CONSIDER:

• **Hearing Impairment**
  • Global Developmental Delay: Intellectual Disability
  • Communication Disorders
  • Pervasive Developmental Disorders
  • Environmental Factors
  • General Health
HEARING IMPAIRMENT:

Key Points

• 1-6/1000 newborns

• 50% genetic
  30% syndromic (e.g. Waardenburg, Pendred, Usher)
  70% non-syndromic, (e.g. connexin 26/GJB2)
    -77% AR, 22% AD, 1% X-linked or mitoch.

• 50% Non-genetic:
  TORCH infection
  Ear/craniofacial anomalies
  Birth Weight<1500 g.
  Low Apgar Scores (0-3 at 5 min, 0-6 at 10 min)
  Respiratory Distress/ Prolonged mechanical ventilation, hyperbilirubinemia requiring exchng transfusion
  Bacterial meningitis/ Ototoxic meds
HEARING LOSS:
Post-newborn

- Recurrent or persistent OME (at least 3 mo)
- Head trauma with fracture of temporal bone
- Congenital CMV (often asymptomatic, hl may show up in later childhood- median age 44 months)
- Childhood infectious diseases (eg, meningitis, mumps, measles)
- Chemotherapy
- Structural anomalies: (e.g. Mondini malformation, enlarged vestibular aqueduct)
- Neurodegenerative disorders (eg, Hunter syndrome), demyelinating diseases (eg, Friedreich ataxia, Charcot-Marie-Tooth)
Hearing Loss

Mild  25-39 dBHL
Moderate  40-68 dBHL
Severe  70-94 dBHL

FS Audiometrics
P.O. Box 36423
Louisville, Ky., 40233-6423
Tools for Hearing Screening

- Auditory Brainstem Response (ABR), Evoked Otoacoustic Emissions (OAE)
  - Tests of Auditory Pathway Structural Integrity
  - Newborn Screening
- Visual Reinforcement Audiometry (VRA)
  - 9-12 months+
- Play Audiometry
  - 2-4 years+
- Conventional Audiometry
  - 4 years +
Visual Impairment

- Overall prevalence: 12.2 per 1,000 under age 18
- Severe Visual Impairment: .06 per 1,000
  - legally blind: less than 20/200 vision in the better eye or a very limited field of vision (20 degrees at its widest point)
  - or totally blind
- The effect of visual problems on a child's development depends on severity, type of loss, age of loss, and overall level of the child’s functioning
- Lack of visual input limits imitation and exposure to non-verbal cues affecting social skill development; milestones in all areas can be delayed

» NICHCY
P.O. Box 1492
Washington, DC 20013
A 5 year old boy presents for health maintenance. Developmental surveillance reveals that he can copy a circle, knows the adjectives “tired” and “hungry” and can broad jump, but cannot hop in place, draw a person in 3 parts or name 4 colors. You suspect:

1. Learning Disability
2. Mild Intellectual Disability (Mental Retardation)
3. Cerebral palsy
4. Autistic Spectrum Disorder
5. Severe Intellectual Disability
Language Delay in a Preschooler

CONSIDER:

• Hearing Impairment

• **Global Developmental Delay:** *Intellectual Disability (Mental Retardation)*
  
  • Communication Disorders
  • Pervasive Developmental Disorders
  • Environmental Factors
  • General Health
INTELLECTUAL DISABILITY
(Mental Retardation)

2010 definition (AAIDD):

Disability characterized by:

• Significant limitations in intellectual functioning (IQ >2SD below mean)
• Significant limitations in adaptive behavior (covers many everyday social and practical skills)
• Originates before age 18
Mild ID/MR: IQ level 50-55 to approx. 70

Moderate ID/MR: IQ level 35-40 to 50-55

Severe ID/MR: IQ level 20-25 to 35-40

Profound ID/MR: IQ level below 20 or 25

DSM 4: American Psychiatric Association
ID Level Predicts Rate of Learning and Ultimate Prognosis**

• **Mild ID/MR:**
  Reads at grade 3-6 level, lives independently, holds job, has children

• **Moderate ID/MR:**
  Reads at 1st to 3rd grade level, community living, supportive work environment

• **Severe ID/MR:**
  Some sight words ("stop", "exit"), needs supervision/assistance in ADL

**In absence of significant behavioral disturbance**
Intellectual Disability-Key Points

• **Assessment:**
  – Standardized psychological testing → IQ
    • IQ = developmental age divided by chronological age
  – Test of adaptive functioning, e.g. Vineland Adaptive Behavior Scales

• **Earlier presentation = more severe degree**
  – Moderate to Severe: Presents infancy to age 2
  – Mild: Presents preschool to early school age

• **2-3% of population, majority mild ID/MR**
ID/MR- Etiology

- **Prenatal (50-70%)**: genetic, CNS malformations, fetal compromise, infection, teratogens
- **Perinatal (<10%)**: HIE, prematurity
- **Postnatal**: Trauma, asphyxia, infection, toxins, vascular malformations, tumors, degenerative disease
- **Environmental (additive)**: Deprivation/malnutrition
- *More severe forms, more likely to find definitive etiology*
ID/MR: Associated Conditions

- Behavior Disorders
- Seizures
- Motor Impairments/CP
- Syndromes and their associated medical conditions
- More common with more severe intellectual disability
A stranger should be able to understand half of a child’s speech at age:

1. 12 months
2. 18 months
3. 24 months
4. 36 months
Language Delay in a Preschooler

CONSIDER:

• Hearing Impairment
• Global Developmental Delay: Intellectual Disability

• Communication Disorders
  • Pervasive Developmental Disorders
  • Environmental Factors
  • General Health
Communication Disorders

- Expressive Language Disorders
- Mixed Expressive/Receptive Disorders
- Phonological Disorders
• **Expressive Disorders**
  Disorders of morphology (form), semantics (word meaning), syntax (grammar), pragmatics (social use of language)

• **Mixed Expressive/Receptive Disorders:**
  – Above plus comprehension deficits

• **Phonological Disorders**
  – Disorders of articulation (motor movements), dyspraxias (motor planning)
  – Disorders of fluency (flow, rhythm)
  – Disorders of voice/resonance
Communication Disorders- Key Points

• 3-5% of Preschoolers
  – Estimated 30-50% go on to develop reading disorders

• Genetic Basis: May be seen in 30-40% of first degree relatives

• Developmental Language Disorder alone vs. communication deficit associated with:
  – Cognitive delay
  – PDD
  – Hearing impairments
  – Social/environmental factors
“Stuttering”

- Disturbance in fluency and time patterning of speech
- Begins age 2 ½ to 4, peak age 5
- Male:female 3-4: 1
- 75% of preschoolers will stop
- Indications for evaluation:
  - Family history of stuttering
  - Persists 6 months or more
  - Presence of concomitant speech or language disorders
  - Secondary emotional distress
All of the following observations are considered risk factors for Autistic Spectrum Disorders except:

A. Lack of protodeclarative pointing at 16 months
B. Lack of babbling at one year
C. Lack of gaze monitoring at 10 months
D. Echoing phrases at 18 months
All of the following observations are considered risk factors for Autistic Spectrum Disorders except:

A. Lack of protodeclarative pointing at 16 months
B. Lack of babbling at one year
C. Lack of gaze monitoring at 10 months
D. *Echoing phrases at 18 months*
Language Delay in a Toddler/Preschooler

CONSIDER:

• Hearing Impairment
• Global Developmental Delay: Intellectual Disability
• Communication Disorders
• Pervasive Developmental ("Autistic Spectrum") Disorders
• Environmental Factors
• General Health
Pervasive Developmental Disorders- DSM IV

- Autistic Disorder
- PDD NOS
- Asperger’s Disorder
- Rett’s Syndrome
- Childhood Disintegrative Disorder
AUTISTIC DISORDER*

1. QUALITATIVE IMPAIRMENT IN SOCIAL INTERACTION

a) marked impairment in nonverbal behaviors (eye contact, facial expressions, gestures)
b) failure to develop peer relationships
c) lack of spontaneous seeking to share enjoyment, interests, or achievements with other people
d) lack of social or emotional reciprocal

*[Six or more criteria, at least two from (1), one each from (2) and (3)]
2. QUALITATIVE IMPAIRMENT IN COMMUNICATION

(a) delay in, or total lack of, the development of spoken language
(b) marked impairment in the ability to initiate or sustain a conversation
(c) stereotyped and repetitive use of language or idiosyncratic language
(d) lack of varied, spontaneous make-believe play or social imitative play appropriate to developmental level
3. RESTRICTED REPETITIVE AND STEREOTYPED PATTERNS OF BEHAVIORS, INTERESTS AND ACTIVITIES

(a) encompassing preoccupation with stereotyped and restricted patterns of interest
(b) inflexible adherence to specific, nonfunctional routines or rituals
(c) stereotyped and repetitive motor mannerisms
(d) persistent preoccupation with parts of objects
Autistic Spectrum Disorders
Key Points

• **Prevalence** *(Pediatrics 2009:124): ~ 1/100*

• Male: Female 4:1

• **Seen in association with:**
  – Seizure disorders, congenital infection, metabolic abn (PKU)
  – Neurocutaneous disorders (TS, NF)
  – Genetic Disorders (Fra X, Angelman’s, Smith-Lemli Opitz )
  – No proven ass’n with vaccines (MMR, thimerosol)

• **Genetic Basis:** concordance in monozygotic (60-80%) vs. dizygotic twins, sibs (3-7%)
Autistic Disorder

• Presents prior to age 3 with:
  Absent or delayed joint attention
  Lack of:
  – gaze monitoring (8-10 mos),
  – proto-imperative pointing(12-14 mos),
  – proto-declarative pointing(14-16 mos)

• Delayed language
• Delayed play skills
• Language regression between 12-24 months in 1/3 of cases
PDD NOS

• Presentations that do not meet criteria for autism
  – late age of onset
  – atypical symptomatology
  – subthreshold symptomatology

• Autistic *Spectrum* Disorders
  – Clinical presentation varies with degree of severity and cognitive level
Asperger’s Disorder

• Qualitative impairment in social interaction
• No clinically significant general delay in language
  – Impaired pragmatics
  – “Little professors”
• No clinically significant delay in cognitive development or in the development of age-appropriate self-help skills
  – Motor coordination difficulties
• **Childhood Disintegrative Disorder**
  – Normal development first 2 years
  – Loss of previously acquired skills by age 10

• **Rett’s Syndrome**
  – Occurs in females
  – Normal development first 5 months
  – Deceleration in head growth between 5-48 m.
  – Stereotypic hand movements
  – Impaired communication, socialization; severe MR
  – MECP2 mutation
Autism Screening Instruments:
  e.g. CHAT, m-CHAT
Diagnostic Instruments:
  e.g. CARS, ADOS, ADIR
ASD’s: Treatment

• Special Education
  – ABA, TEACCH, DIR

• Psychopharmacology for targeted behaviors
  – Hyperactivity, aggression, self-injury

• CAM considered by a majority of families
  – Antifungals, IVIG, megavitamins, chelation, hyperbaric oxygen, secretin, gluten or casein-free diets
  – Few are evidence-based
An 8 year old second grade boy was referred for evaluation due to academic difficulties. His psychological and psychoeducational evaluations revealed:

WISC 4: Full Scale IQ = 99, Verbal Comprehension = 85, Perceptual Reasoning = 105, working memory = 110, Processing Speed = 108;
WIAT 2: Word Reading = 92, Reading comprehension = 81, Numerical operations = 98, Math reasoning = 79.

This child’s likely diagnosis is:
A. Borderline Intellectual Functioning
B. Reading Disability
C. Attention Deficit Hyperactivity Disorder
D. Nonverbal Learning Disability
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A. Borderline Intellectual Functioning  

B. **Reading Disability**  

C. Attention Deficit Hyperactivity Disorder  

D. Nonverbal Learning Disability
School Failure

• “Slow Learner”: Borderline Intelligence
• Learning Disorders: Average Intelligence
• Attention Deficit and Disruptive Behavior Disorders
  
  Oppositional Defiant Disorder, Conduct Disorder

• Mood and Anxiety Disorders
• Chronic Medical Illness
• Psychosocial stressors
Learning Disorders

Disabilities in:

• receptive language, expressive language
• basic reading skills, reading comprehension
• written expression
• mathematics calculation, mathematical reasoning

IDEA (2004): Intelligence-achievement discrepancy ("wait to fail", "test to diagnose") approach replaced by response to intervention (RTI) approach.
Learning Disorders: Key Points

- Discrepancy between intellectual potential and achievement
- Associated Self-esteem and Behavioral Issues
- “Dyslexia” = Reading Disability
  - Majority = Language-based, disordered phonemic processing
  - Familial: Up to 60% of parents also have reading difficulties, 68% concordance in monozygotic twins, 38% in dizygotic
  - Structural and functional CNS abnormalities
- 50% Of Children with Reading Disability meet criteria for ADHD
Learning Disabilities: Evaluation

• Cognitive Assessment
  • Full scale IQ, Verbal IQ, Performance IQ
    – 2 SD discrepancy is significant
    – PIQ>VIQ: Language-based LD: difficulties in phonemic processing, reading, spelling
    – VIQ>PIQ: Nonverbal LD: perceptual problems, motor impairments, slow processing speed, dyscalculia

• Educational (achievement) testing
  • Specific academic abilities
A 9 year old third grade boy is brought to your office by his mother who is distraught about his report card. He is below average in reading and spelling and his teacher states that he does not complete assignments and is distractible in class. He is not a management problem at home other than when it’s time to do his homework. He has friends and excels on the baseball field. An appropriate next step would be:

A. Request completion of parent and teacher Vanderbilt Questionnaires
B. Initiate a trial of methylphenidate
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Attention-Deficit/Hyperactivity Disorder

- A. Symptoms of Inattention, Impulsivity, Hyperactivity
- B. Some symptoms present before age 7 years.
- C. Impairment from the symptoms is present in two or more settings.
- D. Clear evidence of clinically significant impairment in social, academic, or occupational functioning.
Attention-Deficit/Hyperactivity Disorder: Inattention, Hyperactivity/Impulsivity

• **INATTENTION:**

Six (or more) for at least 6 months: often

- (a) does not attend to details, makes careless mistakes
- (b) has difficulty sustaining attention in tasks or play
- (c) does not listen when spoken to directly
- (d) does not follow through on instructions
- (e) has difficulty with organization
- (f) avoids tasks that require sustained mental effort
- (g) loses things
- (h) easily distracted
- (i) forgetful
• **HYPERACTIVITY/IMPULSIVITY**: 

• Six (or more) for at least 6 months: often  
  (a) fidgets with hands or feet or squirms in seat  
  (b) leaves seat when remaining seated is expected  
  (c) runs about or climbs excessively (in adolescents or adults, feelings of restlessness)  
  (d) has difficulty playing or engaging in leisure activities quietly  
  (e) "on the go" or acts as if "driven by a motor“  
  (f) talks excessively  
  (g) blurts out answers before questions have been completed  
  (h) has difficulty awaiting turn  
  (i) interrupts or intrudes on others
• ADHD Subtypes:

Combined Type (80%*)

Predominantly Inattentive Type (10-15%*)

Predominantly Hyperactive-Impulsive Type (5%*)

* in school-age children
ADHD: Key Points

- Disorder of dopamine and norepinephrine systems in frontostriatal circuitry
- 3-7% of school age children
- Male: female (6:1-3:1)
- Genetic Predisposition: 5-6 fold increase in first degree relatives
- Environmental Factors: e.g. head trauma, lead exposure, VLBW, prenatal teratogens
- Symptoms Persist into Adulthood in 60-80%
ADHD- Key Points cont’d

- Symptoms in two or more settings
  - Rating scales
- Consider co-morbid conditions
- Rule out Medical Conditions (e.g. thyroid disorder, toxic exposure, medications, CNS disease)
ADHD- Key points (cont’d)

• Co-morbid Conditions:
  – Learning Disorders
  – Anxiety Disorders
  – Oppositional Defiant Disorder
  – Conduct Disorder
  – Tic Disorders
  – Mood Disorders
  – Substance abuse disorders (adolescents)
ADHD- Treatment

• **Psychopharmacologic: stimulants= first line**
  – Inhibit reuptake of dopamine and norepinephrine
  – Stimulant Side effects: appetite suppression, headache, abdominal pain, growth suppression, irritability, onset/exacerbation of tics
  – “Black box” warnings

• **Behavioral Interventions**
MTA Study (Pediatrics 113:754-761, 2004):

Behavior Therapy alone not as effective as medication alone
597 children ages 7-9.9 at 6 centers, treated for 14 months
- 68% improved with combination of medication and intensive behavioral treatment
- 56% improved with medication alone
- 34% improved with behavioral treatment alone
## Medications for ADHD

<table>
<thead>
<tr>
<th>STIMULANTS</th>
<th>Brand Name</th>
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<tbody>
<tr>
<td>Methylphenidate-</td>
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<tr>
<td>short-acting</td>
<td>Ritalin</td>
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<tr>
<td>Intermediate-acting</td>
<td>Metadate ER, Metylin ER</td>
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<tr>
<td>long-acting</td>
<td>Concerta, Ritalin LA, Metadate CD</td>
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<tr>
<td>transdermal</td>
<td>Daytrana</td>
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<tr>
<td>Dexmethylphenidate</td>
<td>Focalin</td>
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<tr>
<td>Dextroamphetamine sulfate-</td>
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<tr>
<td>short-acting</td>
<td>Dexedrine</td>
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<tr>
<td>Intermediate-acting</td>
<td>Dexedrine spansules</td>
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<tr>
<td>Lisdexamfetamine</td>
<td>Vyvanase</td>
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<tr>
<td>long-acting</td>
<td>Adderall</td>
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<tr>
<td>Mixed amphetamines</td>
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<tr>
<td>Long-acting</td>
<td>Adderall XR</td>
</tr>
<tr>
<td>NOREPINEPHRINE REUPTAKE INHIBITOR</td>
<td>Brand Name</td>
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<tr>
<td>Atomoxetine</td>
<td>Strattera</td>
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<th>ALPHA 2 AGONISTS</th>
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<tbody>
<tr>
<td>Clonidine</td>
<td>Catapres</td>
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<td>Long-acting: kapvey</td>
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<td>Guanfacine</td>
<td>Tenex</td>
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A. Johnny’s behavior is most consistent with the lack of impulse control associated with ADHD.
B. Johnny’s behavior is likely to meet criteria for a disorder often co-morbid with ADHD, but not consistent with ADHD alone.
C. Johnny is also likely to be cruel to animals, to steal and to run away from home.
D. Johnny’s behavior is consistent with the general class of “internalizing” behaviors.
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D. Johnny’s behavior is consistent with the general class of “internalizing” behaviors.
“Externalizing Disorders”

- ADHD
- Oppositional-Defiant Disorder
- Conduct Disorder
Oppositional Defiant Disorder

- Actively does not follow adults' requests
- Angry and resentful of others
- Argues with adults
- Blames others for own mistakes
- Has few or no friends or has lost friends
- Is in constant trouble in school
- Loses temper
- Spiteful or seeks revenge
- Touchy or easily annoyed
- Disturbance causes clinically significant impairment in social, academic or occupational functioning,
- Behaviors do not occur exclusively during the course of a Psychotic or Mood Disorder
- Criteria are not met for Conduct Disorder
Conduct Disorder

- Antisocial behaviors, such as bullying and fighting
- Breaking rules without apparent reason
- Cruel or aggressive behavior toward people and animals
- Destruction of property
- Heavy drinking and/or heavy illicit drug use
- deceitfulness or theft
- Running away
- Truancy (beginning before age 13)
- *Disturbance causes clinically significant impairment in social, academic or occupational functioning*,
- *Childhood Onset type: onset of at least one criterion prior to age 10*
- *Adolescent Onset type: absence of any criteria prior to age 10*
The mother of your 14 year old patient, Rachel, confides in you that her daughter has changed. Now finishing 8th grade, Rachel tends to come home from school, get into bed and turn on the television most every day. Over a year ago she quit the Field Hockey team and the Key Club, activities she enjoyed in middle school stating she “wasn't good enough”. Previously an A-/B+ student, she now has a B-/C average. Her physical examination is normal although you note a significant increase in her BMI in the past year. You are concerned that Rachel might be exhibiting symptoms of:

A. Major Depression
B. Generalized Anxiety Disorder
C. School Phobia
D. Dysthymic Disorder
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“Internalizing Disorders”

Mood Disorders: e.g. Major Depressive Disorder, Dysthymic Disorder, Bipolar Disorder

Anxiety Disorders: e.g. Generalized Anxiety Disorder, Separation Anxiety Disorder, Panic Disorder, Social Anxiety Disorder, School Phobia, Obsessive-Compulsive Disorder, Post-traumatic Stress Disorder
Major Depressive Disorder

- Five or more of the following (nearly every day):
  - Depressed more most of the day
  - Diminished interest or pleasure
  - Significant weight loss or gain
  - Insomnia or hypersomnia
  - Psychomotor retardation
  - Fatigue or loss of energy
  - Feelings of worthlessness or excessive guilt
  - Diminished ability to think or concentrate/indecisiveness
  - Recurrent thoughts of death/suicidal ideation

- Symptoms do not meet criteria for a mixed episode, they cause significant distress or impairment in social, occupational or other important areas of functioning, and are not due to substance abuse, not better accounted for by bereavement.
Dysthymic Disorder

- Depressed mood for most of the day, for more days than not, for at least two years. In children and adolescents, mood can be irritable and duration must be at least one year.

- Presence of two or more:
  - Poor appetite or overeating
  - Insomnia or hypersomnia
  - Low energy or fatigue
  - Low self-esteem
  - Poor concentration
  - Feelings of hopelessness
  - Person has not been without the symptoms for at least 2 months at a time
Generalized Anxiety Disorder

• Excessive anxiety and worry, occurring more days than not for at least 6 months, about a number of events or activities. Worry is difficult to control, and is associated with 3 or more of the following
  • Restlessness or feeling keyed up or on the edge
  • Easily fatigued
  • Difficulty concentrating or mind going blank
  • Irritability
  • Muscle tension
  • Sleep disturbance
  • *Disturbance is not due to specific features of other anxiety disorders, causes significant distress or impairment in functioning, not due to direct physiological effects of a substance or general medical condition.*
In Summary......

1. Know developmental milestones!
2. Know how developmental/behavioral disorders present.
3. Know the differential diagnoses of developmental delays and behavior disorders:
   - Disorders affecting motor development
   - Intellectual Disabilities
   - Language Disorders
   - Autistic Spectrum Disorders
   - Learning Disabilities
   - Externalizing Disorders
   - Internalizing Disorders
Good Luck!!!