OBJECTIVES

“The most common presentation of a developmental disability is failure to achieve age-appropriate developmental milestones.”

- How common are developmental problems?
- How does one approach developmental delay?
- What are some “rules of thumb” for recognizing delay?
- What are the developmental implications of common newborn findings?
DEFINITIONS

- Delay vs. Deviance
- Surveillance vs. Screening
- Streams vs. Domains

LOOK FOR PATTERNS!

Streams of Development

- Gross motor
- Fine motor
- Language
  - Expressive
  - Receptive
- Problem solving
- Social

Patterns in Development

<table>
<thead>
<tr>
<th></th>
<th>MOTOR</th>
<th>PS</th>
<th>RL/EL</th>
<th>SOCIAL</th>
</tr>
</thead>
<tbody>
<tr>
<td>ID</td>
<td>V</td>
<td>D</td>
<td>D</td>
<td>D</td>
</tr>
<tr>
<td>CP</td>
<td>D</td>
<td>V-N</td>
<td>V-N</td>
<td>D</td>
</tr>
<tr>
<td>VI</td>
<td>D</td>
<td>D</td>
<td>N</td>
<td>D</td>
</tr>
<tr>
<td>HI</td>
<td>N</td>
<td>N</td>
<td>D</td>
<td>V-N</td>
</tr>
<tr>
<td>AUTISM</td>
<td>N</td>
<td>V-N</td>
<td>D</td>
<td>D</td>
</tr>
</tbody>
</table>
Prevalence of DD

<table>
<thead>
<tr>
<th>Dx</th>
<th>per 1000</th>
<th>age (mo)</th>
<th>MD 1st</th>
</tr>
</thead>
<tbody>
<tr>
<td>GDD</td>
<td>50</td>
<td>24</td>
<td>30%</td>
</tr>
<tr>
<td>ID (mild MR)</td>
<td>25</td>
<td>39</td>
<td>60%</td>
</tr>
<tr>
<td>ID (mod/severe MR)</td>
<td>5</td>
<td>12</td>
<td>90%</td>
</tr>
<tr>
<td>Learning disability</td>
<td>100</td>
<td>69</td>
<td>12%</td>
</tr>
<tr>
<td>ADHD</td>
<td>90</td>
<td>59</td>
<td>45%</td>
</tr>
<tr>
<td>CP</td>
<td>3</td>
<td>12-14</td>
<td>99%</td>
</tr>
<tr>
<td>AUTISM</td>
<td>8</td>
<td>40</td>
<td>30%</td>
</tr>
<tr>
<td>Visual impairment</td>
<td>.5</td>
<td>55</td>
<td>60%</td>
</tr>
<tr>
<td>Hearing Impairment</td>
<td>1-3</td>
<td>39</td>
<td>40%</td>
</tr>
</tbody>
</table>

What's the problem?

- 16% of preschool children have developmental or behavioral problems.
- <30% of MDs screen.

Use the parent!

<table>
<thead>
<tr>
<th>Parent concern</th>
<th>PPV</th>
</tr>
</thead>
<tbody>
<tr>
<td>Behavior-emotions</td>
<td>41%</td>
</tr>
<tr>
<td>Speech/language</td>
<td>55%</td>
</tr>
<tr>
<td>Social/self-help</td>
<td>NS</td>
</tr>
<tr>
<td>Fine motor</td>
<td>75%</td>
</tr>
<tr>
<td>School readiness</td>
<td>40%</td>
</tr>
<tr>
<td>Global concerns</td>
<td>80%</td>
</tr>
</tbody>
</table>
**Ages for early diagnosis**

<table>
<thead>
<tr>
<th></th>
<th>0-12 mo.</th>
<th>1-2 yr.</th>
<th>2-3 yr.</th>
<th>3-4 yr.</th>
</tr>
</thead>
<tbody>
<tr>
<td>ID, mod/sev.</td>
<td>ID, mod</td>
<td>ID, mild</td>
<td>ID, mild</td>
<td></td>
</tr>
<tr>
<td>VI/HI</td>
<td>HI</td>
<td>HI</td>
<td></td>
<td></td>
</tr>
<tr>
<td>CP</td>
<td>CP</td>
<td>CP mild</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Autism</td>
<td>Autism/LD</td>
<td>LD</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

**CHILD WITH DEVELOPMENTAL DELAY**

- **HISTORY**
  - NBN, VISION, HEARING, DEVELOPMENTAL SCREEN
- **PHYSICAL EXAM**
  - Identify and treat VI, HI, syndromes
- **ASSESS PATTERN OF DELAY**
  - SPECIFIC DELAY?
  - GLOBAL DELAY?

**RED FLAG**

Any time there is a history or exam consistent with LOSS of SKILLS (regression)

THINK......metabolic, genetics w/u, neuroimaging, seizures, hydrocephalus, toxin exposure, autism
**Developmental Quotient**

DQ = Developmental Age / Chronologic age X 100

- DQ <70  Delay
- DQ 70-85 Monitor
- DQ >85 Typical range

*Perform for each stream of development

---

**Gross motor milestones**

- **Prone**
  - Head up 1 mo
  - Chest up 2 mo
  - On elbows 3 mo
  - On hands 4 mo

- **Roll**
  - Front to back 4-5 mo
  - Back to front 5-6 mo

- **Sit**
  - With support 5 mo
  - Without support 6 mo

- **Crawl (1/3 don’t)** 8-9 mo
- **Pull to stand** 9 mo
- **Cruise** 9-10 mo
- **Walk alone** 12 mo
- **Run** 15 mo
- **Jump in place** 24 mo
- **Pedal trike** 36-36 mo
- **Gallops** 48 mo(4)
- **Skips** 60 mo(5)
- **Tandem**
  - Forward 5 yr
  - Backward 7-8 yr

---

**RED FLAG**
**RED FLAGS**

- Any boy not walking by 15 months should get a CPK to r/o MD
- The most common cause of delayed GM is global delay.

**MD = muscular dystrophy**

- 3/10,000 boys
- 30% have no Fam Hx
- Absent dystrophin
- Xp21.2
- CPK is >20x normal!
- DNA analysis of blood can make dx in 2/3's.

**Motor delay**

- Decreased TONE
- Increased STRENGTH
- Decreased REFLEXES
- Normal or increased
- Neurological Syndromes, DCD, LE
- Degenerative Disorders
Motor delay: Evaluation

- Central (CNS)
  - MRI
  - CHROMOSOMES
  - FRAGILE X
  - ? METABOLIC EVAL
  - ? TFT
  - Get PT or OT eval
- Neuromuscular
  - CPK
  - LFT'S
  - ESR
  - LACTATE/PYRUVATE
  - TFT
  - CARNITINE
  - EMG/NCV
  - MUSCLE BX

Definitions

- **Language**: a system of verbal, written, or gestured symbols used to communicate information or feelings.
  - Components: phonology, morphology, syntax, semantics, pragmatics
- **Speech**: The physical production of spoken language.
  - Components: articulation, phonology, voice quality, pitch, loudness, resonance, fluency, rate, rhythm

Identify early!

- 5-10% of all children have developmental language disorder (that is 2/day for PCM)
- Language is the BEST predictor of later cognitive function
- Delayed language may be a marker of other developmental disorders
- Early intervention yields best outcomes
Normal Language Development

- Expressive: two phases
  - Social smile 5 wk
  - Coos 6-8 wk
  - Laughs 3-4 mo
  - Rasberry 4-5 mo
  - Squeals 5 mo
  - Babbles 6 mo
  - UNTIL 6-8 mo!

- 1st word 11 mo
- Immature jargon 12 mo
- 4-6 words 15 mo
- 2 words 21 mo
- Pronouns indiscrim 2 y
- Tells stories 4 years, 100% intelligible

- Receptive – Infant
  - Alerts to voice 1 mo
  - Regards speaker 3 mo
  - Listen then vocalizes 5 mo
  - Enjoys gesture games 9 mo
  - Orients to name 10 mo
  - Understands “no” 11 mo
  - Command, with gesture 12mo

- Receptive – Toddler
  - 1 step command without gesture 14 mo
  - 1 body part 15 mo
  - Fetches on command 16 mo
  - Points to picture 18 mo
  - 6 body parts 20 mo
  - 2 step command 24 mo

Language delay

- Developmental Language Disorder 5-10%
- ID 3%
- Hearing Impairment .5-1%
- Autism .6-1%
All children with language delay should be referred for hearing assessment. Infants who are deaf may have normal pre-linguistic expressive language until 6-9 months of age.

Normal Pattern is for RL>EL. Expressive language that significantly exceeds receptive language is deviant. THINK:
1. AUTISM, with echolalia
2. Syndromes with “cocktail personalities”
3. Parent mis-interpretation

Red Flags of Autism Spectrum Disorder and Developmental Delays in the Second Year of Life

ASD Red Flags
- Lack of smiling
- Lack of coordination of nonverbal communication
- Lack of sharing interest or enjoyment
- Repetitive movements with objects
- Lack of appropriate gaze
- Lack of response to name
- Lack of warm, joyful expressions
- Unusual postures
- Repetitive movements or posturing of body

ASD & DD Red Flags
- Lack of pointing
- Lack of playing with a variety of toys
- Lack of response to contextual cues
- Lack of communicative vocalizations with consens

www.firstsigns.com
Joint Attention Skills*

<table>
<thead>
<tr>
<th></th>
<th>TYPICAL</th>
<th>ASD</th>
</tr>
</thead>
<tbody>
<tr>
<td>8-10 months</td>
<td>Gaze monitoring</td>
<td>No eye contact</td>
</tr>
<tr>
<td>10-12 months</td>
<td>Following a point</td>
<td>Does not respond to request “oh look!”</td>
</tr>
<tr>
<td>12-14 months</td>
<td>PIP</td>
<td>Develops advanced self help skills</td>
</tr>
<tr>
<td>14-16 months</td>
<td>PDP</td>
<td>* Consistently absent</td>
</tr>
<tr>
<td>14-18 months</td>
<td>Show and tell</td>
<td>Often brings to parent to obtain repeated action</td>
</tr>
</tbody>
</table>

*Joint attention deficits appear to be specific to ASD and reliably differentiate children with ASD from other developmental disabilities.

RED FLAG

A BIFID UVULA is evidence of a submucous cleft in the palate and warrants evaluation if associated with recurrent OM, speech delay, or VPI.
CONSIDER VCFS (del 22q.11)

Problem solving

- Problem solving milestones are evidence of cognitive abilities, or intelligence, without the use of language.
  - Patterns....
    - Normal PS=RL>EL...COMMON, often resolves
    - Normal PS>RL>EL...less common, often LD
    - Low PS, Low RL, Low EL = ID
Normal development

- Blocks
  - Regards 3 mo
  - Attains 6 mo
  - Takes 2nd 7-8 mo
  - Releases into cup 12 mo
  - Takes a 3rd 14 mo
  - Builds a tower of 2 15 months
  - Builds a tower of 4 18 months
  - Builds a tower of 6 24 months
  - Train 30 months

- Pencil
  - Imitates scribble 15 mo
  - Scribbles spontaneously 18 mo
  - Imitates vertical line 24 mo
  - Imitates horizontal line 28 mo
  - Circle 3 yr
  - Square 4 yr
  - Triangle 5 yr
  - Goodenough-Harris DAP see HLH

RED FLAG

- Early Handedness

Children with handedness before age 15 months usually have an abnormally weak upper extremity on the other side.
“Global developmental delay”
- A significant delay in 2 or more streams
- NOT a diagnosis
- NOT regression or loss of skills
- Can’t be used for services after age 6-9.

Intellectual Disabilities (Mental retardation)
- 2-3% of population
- Male 1.6 : Female 1
- 85% of ID is MILD category

Most common genetic cause: Down Syndrome
Most common inherited cause: Fragile X
Most common preventable cause: Fetal alcohol

MR = ID =
Cognitive /Adaptive Disability
- Significantly sub-average general intellectual functioning (IQ <70-75)
- Limited adaptive functioning in at least 2 of 10 skill areas
- Onset before 18 years of age
Levels of ID (CAD)

- Mild (Intermittent Support) IQ ~ 55-69
  - Vast majority 85%
  - More common in boys
- Moderate (Limited Support) IQ ~ 40-54
- Severe (Extensive Support) IQ ~ 25-39
  - Rare .5%
  - Ratio of boys to girls is equal
  - Think about Rett Syndrome in girls
- Profound (Pervasive Support) IQ < 24

Intellectual Disability: Known causes

- Prenatal (60-75%):
  - CNS malformation
  - Chromosomal abnormality
  - Genetic
  - Toxins
  - Infection
  - Neurocutaneous syndrome
  - Malnutrition
- Perinatal (10%):
  - Hypoxia
  - Neonatal seizures
- Postnatal (1-10%)
  - CNS infection
  - Stroke/Hemorrhage
  - Trauma/Abuse
  - Hypoxia
  - Degenerative
  - Epileptic encephalopathy
  - Metabolic
  - Complications of prematurity

ID: The Search

The more severe the ID, the more likely to find etiology.

- Chromosomes or CGH (40% + in SEVERE)
- DNA for Fragile X (2-6% +)
- ± Neuro-imaging (MRI study of choice)
  - IQ <50, micro/macrocephaly, abnormal neuro exam, seizures, loss of milestones
- ± Metabolic Studies
**Predictive value of a good exam**

The presence of three or more minor anomalies is highly predictive of a major malformation (19.6%)

- Examples: bossing, absent hair whorl, anteverted nostrils, epicanthal folds, preauricular tags, pits, abnormal pinna of ears, bifid uvula, extra nipples, single umbilical artery, umbilical hernia, dimple over sacrum, single palmar creases, syndactyly, overlapping toes, recessed toes.....

**Head Circumference**

- Normal growth in full term infants:
  - 2 cm/mo for 3 months
  - 1 cm/mo for 3 months
  - 1/2 cm/mo for 6 months
- 12 cm in first 12 months!
### Head Circumference

- **Rule of 3’s and 9’s**
  - Birth: 35 cm
  - 3 mo: 40 cm
  - 9 mo: 45 cm
  - 3 yrs: 50 cm
  - 9 yrs: 55 cm

### Pits and tags

- 1/100 – common!
- ALL need Hearing
- Renal US if:
  - Other malformation
  - FHx of deafness, ear or kidney issue
  - Gestational DM

### Single umbilical artery

- 6/1000
- More common in twins
- If exam normal, and mother did not have oligo/polyhydramnios, no further work up.
- Exam should r/o GI, renal, cardiac and CNS findings
Sacral lesions

- Dimple – need sacral neuro-imaging (US) if:
  - > 5 mm in size
  - > 2 cm away from anus
  - Associated with any other back lesion
    - Hemangioma, hairy patch, tail, masses
- All hemangiomas, hairy patches, masses, tails need US (36% +)

“Find the ability in disability”