Diagnostic Approach to Developmental Delay

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What is development?

Young Baby ➔ Adulthood

- Wide variation between children
- Variation between domains in same child
Rule: Variable and Progressive

Developmental delay
An information guide for parents
Royal Children Hospital Melbourne
What is Developmental Delay?
What is Developmental Delay?

Development

Time (years)

substantial lag
Developmental Quotient (DQ)

Developmental age \times 100 = DQ \text{ Chronological age}

DQ < 70 indicates developmental delay
### Red flags in evaluation of children with Neuromotor delay

<table>
<thead>
<tr>
<th>Red Flags: Indications for Prompt Referral</th>
<th>Implications</th>
</tr>
</thead>
<tbody>
<tr>
<td>Elevated CK to greater than 3× normal values (boys and girls)</td>
<td>Muscle destruction, such as in DMD, Becker muscular dystrophy, other disorders of muscles</td>
</tr>
<tr>
<td>Fasciculations (most often but not exclusively seen in the tongue)</td>
<td>Lower motor neuron disorders (spinal muscular atrophy; risk of rapid deterioration in acute illness)</td>
</tr>
<tr>
<td>Facial dysmorphism, organomegaly, signs of heart failure, and early joint contractures</td>
<td>Glycogen storage diseases (mucopolysaccharidosis, Pompe disease may improve with early enzyme therapy)</td>
</tr>
<tr>
<td>Abnormalities on brain MRI</td>
<td>Neurosurgical consultation if hydrocephalus or another surgical condition is suspected</td>
</tr>
<tr>
<td>Respiratory insufficiency with generalized weakness</td>
<td>Neuromuscular disorders with high risk of respiratory failure during acute illness (consider inpatient evaluation)</td>
</tr>
<tr>
<td>Loss of motor milestones</td>
<td>Suggestive of neurodegenerative process</td>
</tr>
<tr>
<td>Motor delays present during minor acute illness</td>
<td>Mitochondrial myopathies often present during metabolic stress</td>
</tr>
</tbody>
</table>
## Referral Guidelines: Speech Delay

### TABLE 8 Guidelines for Children with Abnormal Speech Development

<table>
<thead>
<tr>
<th>Age, mo</th>
<th>Referral Guidelines for Children With “Speech” Delay</th>
</tr>
</thead>
<tbody>
<tr>
<td>12</td>
<td>No differentiated babbling or vocal imitation</td>
</tr>
<tr>
<td>18</td>
<td>No use of single words</td>
</tr>
<tr>
<td>24</td>
<td>Single-word vocabulary of ≤10 words</td>
</tr>
<tr>
<td>30</td>
<td>Fewer than 100 words; no evidence of 2-word combinations; unintelligible</td>
</tr>
<tr>
<td>36</td>
<td>Fewer than 200 words; no use of telegraphic sentences; clarity &lt;50%</td>
</tr>
<tr>
<td>48</td>
<td>Fewer than 600 words; no use of single sentences; clarity ≤80%</td>
</tr>
</tbody>
</table>

Guidelines on Identification of Children with ASD

Prespeech Deficits Common in Children with Autism Spectrum Disorders

- Decreased or absent use of prespeech gestures (e.g., waving, pointing)
- Delayed onset of babbling past nine months of age
- Disregard for vocalizations (i.e., lack of response to own name), yet awareness of environmental sounds
- Lack of appropriate gaze
- Lack of expressions such as “oh-oh” or “huh”
- Lack of interest or response to neutral statements
- Lack of recognition of mother’s (or father’s or consistent caregiver’s) voice
- Lack of the alternating pattern of vocalizations between infant and parent that usually occurs at approximately six months of age
- Lack of warm, joyful expressions with gaze
Developmental Trajectory
Developmental Trajectory

Developmental Delay

Transient Developmental delay

Persistent Developmental delay

Typical Developmental

Developmental Disability
1. Environmental causes of DD are real
Even infants and young children are affected adversely when significant stresses threaten their family and caregiving environments.
Stress

**POSITIVE**
Brief increases in heart rate, mild elevations in stress hormone levels.

**TOLERABLE**
Serious, temporary stress responses, buffered by supportive relationships.

**TOXIC**
Prolonged activation of stress response systems in the absence of protective relationships.
Nurturing & Stable & Engaging relationships

#2 Development is a highly interactive process, and life outcomes are not determined solely by genes.

Center on the Developing Child 哈佛大学
While attachments to their parents are primary, young children can also benefit significantly from relationships with other responsive caregivers both within and outside the family.
Unplug

Swapping screen time for active play:

- Make screen time a reward—not a routine.
- No screen time before age 2.
  and no more than two hours of screen time for kids older than 2.

NUH
National University Hospital
DDx of hypotonia in infancy

A schematic approach to hypotonia in infancy JoAnna Leyenaar, MD MPH, Peter Camfield, MD FRCPC, and Carol Camfield, MD FRCPC
Approach to Investigations

- **General Investigations**: TSH, free T4, electrolytes (& Ca)
- **CNS Dysfunction**: CT/MRI head, consider EEG, consult neurology and consider karyotype
- **Metabolic Disease**: urine and serum amino acids, urine organic acids, ammonia, liver function tests
- **Lower motor neuron disease**: creatine kinase, referral to neurology for specialised test
2. Motor Delay warrants closer attention
Fe def anemia affects development

Summary: Implications for Research and Programs Rebecca J. Stoltzfus Center for Human Nutrition, Department of International Health, School of Hygiene and Public Health, The Johns Hopkins University, Baltimore
Iron deficiency and impaired child development The relation may be causal, but it may not be a priority for intervention. Haroon Saloojee, senior lecturer and John M Pettifor, professor
Low vitamin D status & development

In utero  ➔  post natal  ➔  early childhood
## Alstrom Syndrome

<table>
<thead>
<tr>
<th>Feature</th>
<th>Age of Onset Range (Mean)</th>
<th>Incidence</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cone-rod dystrophy</td>
<td>Birth - 15 mos (5 mos)</td>
<td>100%</td>
</tr>
<tr>
<td>Obesity</td>
<td>Birth - 5 years (2.5 yrs)</td>
<td>98%</td>
</tr>
<tr>
<td>Progressive sensorineural hearing loss</td>
<td>2-25 yrs (9 yrs)</td>
<td>88%</td>
</tr>
<tr>
<td>Dilated cardiomyopathy</td>
<td>2 wks - 4 mos</td>
<td>42%</td>
</tr>
<tr>
<td>Restrictive cardiomyopathy</td>
<td>Juvenile - late 30s</td>
<td>18%</td>
</tr>
<tr>
<td>Insulin resistance / type 2 diabetes mellitus</td>
<td>4-30 yrs / 8-40 yrs (16 yrs)</td>
<td>92% / 68%</td>
</tr>
<tr>
<td>Developmental delay</td>
<td>Birth-adolescence</td>
<td>25%-30%</td>
</tr>
<tr>
<td>Short stature</td>
<td>Puberty - adult</td>
<td>98%</td>
</tr>
<tr>
<td>Hypogonadotropic hypogonadism</td>
<td>10+ yrs</td>
<td>78% of males</td>
</tr>
<tr>
<td>Urologic disease</td>
<td>Adolescence - adult</td>
<td>48%</td>
</tr>
<tr>
<td>Renal disease</td>
<td>Adolescence - adult</td>
<td>Variably progressive with age in all individuals</td>
</tr>
<tr>
<td>Hepatic disease</td>
<td>8-30 yrs</td>
<td>23%-92%</td>
</tr>
</tbody>
</table>

3. Hearing screen is important
(despite newborn hearing screen)
Hearing lost is difficult to diagnose clinically.
Use risk stratification instead.
Risk Indicators Associated With Permanent Congenital, Delayed-Onset, and/or Progressive Hearing Loss in Childhood

- Caregiver concerns regarding hearing,
- speech, language delay
- developmental delay
- Warrants objective hearing assessment

AAP Clinical Report—Hearing Assessment in Infants and Children: Recommendations Beyond Neonatal Screening *Pediatrics* 2009
Thank You.

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Questions?