Global Developmental Delay and Intellectual Disability: DD Plus Update

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Intellectual Disabilities*

- Definitions and terms have evolved
- Moving away from exclusively IQ-based
- More focus on skills and adaptive behavior
- Greater recognition of strengths
- Understanding levels of support to maximize function

AAIDD Definition 2013
- Significant limitations in intellectual functioning
- Significant limitations in adaptive behavior
- Onset in childhood
<table>
<thead>
<tr>
<th>Severity Category</th>
<th>Approximate Percent Distribution of Cases by Severity</th>
<th>DSM-IV Criteria (severity levels were based only on IQ categories)</th>
<th>DSM-5 Criteria (severity classified on the basis of daily skills)</th>
<th>AAIDD Criteria (severity classified on the basis of intensity of support needed)</th>
<th>SSI Listings Criteria (The SSI listings do not specify severity levels)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mild</td>
<td>85%</td>
<td>Approximate IQ range 50–69</td>
<td>Can live independently with minimum levels of support.</td>
<td>Intermittent support needed during transitions or periods of uncertainty.</td>
<td>IQ of 60 through 70 and a physical or other mental impairment imposing an additional and significant limitation of function</td>
</tr>
<tr>
<td>Moderate</td>
<td>10%</td>
<td>Approximate IQ range 36–49</td>
<td>Independent living may be achieved with moderate levels of support, such as those available in group homes.</td>
<td>Limited support needed in daily situations.</td>
<td>A valid verbal, performance, or full-scale IQ of 59 or less</td>
</tr>
<tr>
<td>Severe</td>
<td>3.5%</td>
<td>Approximate IQ range 20–35</td>
<td>Requires daily assistance with self-care activities and safety supervision.</td>
<td>Extensive support needed for daily activities.</td>
<td>A valid verbal, performance, or full-scale IQ of 59 or less</td>
</tr>
<tr>
<td>Profound</td>
<td>1.5%</td>
<td>IQ &lt;20</td>
<td>Requires 24-hour care.</td>
<td>Pervasive support needed for every aspect of daily routines.</td>
<td>A valid verbal, performance, or full-scale IQ of 59 or less</td>
</tr>
</tbody>
</table>
Outcome of child with mild ID*

- PCPs frequently have more pessimistic expectations of outcome
- Learns at ½ to 3/4 usual rate
- Adult reading 3rd to 7th grade level
- Vocational/occupational track in high school
- Usually lives independently, may marry and raise children
- Competitive employment, especially if good work habits, social skills and community support
Outcome of child with moderate ID*

• Learns at 1/3 to ½ usual rate
• Adult reading 1\textsuperscript{st} to 4\textsuperscript{th} grade level
• Vocational or Life Skills track in HS
• Needs formal teaching of ADL skills
• Lives in supervised group home
• Rarely marries or parents children
• Supportive or sheltered employment
Developmental Disability

- Severe chronic disability in child of 5 years or older
- Onset before age 22 years
- Results in substantial functional limitations
- Intellectual Disability is a subset of DD
- May include severe ADHD, LD, CP, ASD, etc
- Overall prevalence may be 1 in 6 – but when does a condition become a DD?
- ICF emphasizes activity and participation limitations and importance of environment

World Health Organization
International Classification of Functioning, Disability and Health (ICF)
Global Developmental Delay

- A precursor of developmental disabilities
- Implies likelihood of intellectual disability
- Significant delay in 2 or more domains:
  - motor, language, cognition, social, ADL
  - performance at least 2 SD below mean
- Distinguish from single-domain developmental delay
- Usually reserved for children < 5 years
- May be associated with other developmental disorders
- Common condition, prevalence approximately 5%
# Approximate Prevalence of Developmental Disorders

<table>
<thead>
<tr>
<th>Disorder</th>
<th>per 1000</th>
</tr>
</thead>
<tbody>
<tr>
<td>ADHD</td>
<td>70</td>
</tr>
<tr>
<td>LD</td>
<td>80</td>
</tr>
<tr>
<td>Global developmental delay</td>
<td>50</td>
</tr>
<tr>
<td>Developmental language disorder</td>
<td>50</td>
</tr>
<tr>
<td>Developmental coordination disorder</td>
<td>50</td>
</tr>
<tr>
<td>Mild intellectual disability (mild ID)</td>
<td>15</td>
</tr>
<tr>
<td>Autism spectrum disorders</td>
<td>15</td>
</tr>
<tr>
<td>Moderate-severe intellectual disability</td>
<td>5</td>
</tr>
<tr>
<td>Cerebral palsy</td>
<td>4</td>
</tr>
<tr>
<td>Fetal alcohol syndrome</td>
<td>3</td>
</tr>
<tr>
<td>Vision impairment or deafness</td>
<td>3</td>
</tr>
</tbody>
</table>
The etiology of Global Developmental Delay*

• Complex interplay of biological and environmental risk factors
  • Male gender
  • Low birth weight
  • Poverty, neglect, deprivation
  • Malnutrition
  • Low maternal education
  • Advanced maternal age

• Established causes of GDD/ID
  • Fetal alcohol exposure
  • Down syndrome and other genetic/chromosomal disorders
  • Neuro-Metabolic disorders and nutritional deficiencies
  • Congenital brain malformations
  • Lead and other environmental toxins
  • Brain injury – prematurity, asphyxia, trauma, abuse, other
Medical evaluation of child with global developmental delay*

• All children with GDD/ID merit comprehensive medical evaluation
• The value and limitations of etiologic diagnosis
• Review evidence:
  • Metabolic screening*
  • Genetics testing*
  • Neuroimaging*
  • EEG*
• Published guidelines

  Moeschler J, Shevell M. Pediatrics 2014;134:e903
Value of etiologic diagnosis

• Specific treatment implications
• Ongoing medical management of associated conditions
• Prognostic implications
• Assessment of recurrence risk
• Dispelling myths and pseudo-diagnoses
• Limiting further unnecessary testing
Limitations and drawbacks of etiologic search

• Many disorders have no specific treatment
• Enormous individual variation for most disorders
• Prognostic uncertainty of rare disorders
• Etiology often of more interest to physicians than to families
• “We’re not having any more children.”
• Costs of testing: $$$, pain, sedation
• False positives and parental anxiety
Recommendations: Metabolic and genetic tests*

• Routine metabolic screening not indicated, unless:
  • no newborn metabolic screening
  • indicated by history, physical or lab

• Routine CMA

• Fragile X DNA with clinical preselection

• MECP2 studies in females with moderate to severe ID/GDD

• PTEN screen in autism with macrocephaly (risk of tumors)
Fragile X Syndrome
<table>
<thead>
<tr>
<th>Associated conditions</th>
<th>Mild ID</th>
<th>Severe ID</th>
</tr>
</thead>
<tbody>
<tr>
<td>Seizures</td>
<td>10%</td>
<td>20%</td>
</tr>
<tr>
<td>Hearing impairment</td>
<td>7%</td>
<td>10%</td>
</tr>
<tr>
<td>Vision impairment</td>
<td>1%</td>
<td>15%</td>
</tr>
<tr>
<td>Cerebral palsy</td>
<td>10%</td>
<td>20%</td>
</tr>
<tr>
<td>Sleep disorders</td>
<td>30%</td>
<td>75%</td>
</tr>
<tr>
<td>Recurrent vomiting</td>
<td></td>
<td>10%</td>
</tr>
<tr>
<td>Autism</td>
<td>12%</td>
<td>30%</td>
</tr>
<tr>
<td><strong>Psychiatric disorders</strong></td>
<td><strong>30%</strong></td>
<td><strong>50%</strong></td>
</tr>
<tr>
<td>(&quot;Dual diagnosis&quot;)</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
Common psychiatric disorders in children with ID – the “DD Plus” population...

- ADHD
- Social anxiety, generalized anxiety, separation anxiety disorder, “sensory”
- Depression, Major depressive disorder
- OCD
- Tic disorders/Tourette
- PTSD (consequence of abuse, neglect)
- Mood dysregulation ("Disruptive mood dysregulation disorder", "Intermittent explosive disorder")
- Self injurious behavior
- Sleep disorders
Causes of Psychiatric Disorders in ID

Less common...
• Specific biochemical – Severe SIB in Lesch-Nyhan
• Other neurobiological – Neurotransmitter dysregulation, FAS, TBI
• Behavioral phenotypes – Fragile X, Prader-Willi, Smith-Magenis

More common...
• Genetic predisposition to psychiatric disorders
• Temperamental variation and lack of coping resources
• “Sensory” – hyperactivity, pica, stereotypy, self-injury, rumination
• Environmental factors – school failure, peer rejection, family dysfunction, fragmented community resources, poverty
• Abuse and neglect
Common problem behaviors

- Sleep problems
- Feeding problems
- Pica
- Toilet-training
- Overeating and obesity
- Temper outbursts
- Disruptive behavior, hyperactivity, impulsivity
- Aggression
- Self-injurious behavior
- Repetitive behaviors
- Sexual behavior
Understanding problem behaviors - Functional behavioral analysis

• ABCs: antecedents, behaviors, consequences
• The communicative function of behavior
• Common reasons for behavior problems
  • To get attention/what you want/preferred item
  • To get away/escape/avoid non-preferred activity
  • Overstimulation and anxiety/sensory overload
  • Sensation-seeking
  • Response to pain
Differential diagnosis of behavioral change

- Medication side effects
- Seizures
- Sleep disturbances
- Headaches
- Upper airway obstruction, chronic sinusitis
- Pain – GERD, facial, dental, musculoskeletal
- Chronic constipation
- Abuse or other family-related stress
- Educational mis-management
- Depression
Psychotropic Use and Polypharmacy in GDD/ID

- Psychotropic use common and increasing
- 64% of 33,600 with ASD on at least 1 medication
- 35% on 2 different classes of meds
- 15% on 3 or more
- Median length of polypharmacy 12 months
- Minimal evidence regarding multidrug combinations in children with GDD/ID

*Spencer et al, Pediatrics 2013;132:833-840*

*McGuire K et al. Pediatrics 2016;137 (S2):e20152851*
Good pediatric primary care of GDD/ID may prevent behavioral complications

- Breaking the news – the family conference
- Parent education and support
- Identify child and family strengths
- Building resilience, promoting independence
- Behavioral counseling
- Pharmacotherapy
- Health promotion and disease prevention
- Access to early intervention and therapy
- Educational care
- Community participation
- Transition to adult care
- Long term planning
Parent to Parent Support is key ingredient

• Acceptance of diagnosis
• Lived experience of Family Navigators
• Accessing community resources
• Especially important at transitions
• Early intervention to school
• Puberty and adolescence
• Pediatric to adult health care
• School to vocational preparation
• Group home, independent living