GLOBAL DEVELOPMENTAL DELAYS

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Behavioral Objectives

1. Identify children with global developmental delays
2. Initiate an evaluation to determine an underlying etiology of global developmental delays
3. Provide guidance and counseling to families of children with global developmental delays

Disclosures

• I have no conflicts of interest to disclose
• I will not be discussing the off-label use of any treatments or products

A 25 year old mother brings her 18 month old son, Jack, to the clinic for a routine check-up. His general health and physical examination have always been normal, but mother is concerned because his development has been “slow” compared to his older brother. He sits and crawls, but does not walk. He babbles and sometimes says “mama”, but does not use any other words.

Questions to be Asked

• Is there a problem?
• What is it called?
• What evaluation is needed?
• What do you tell the family?

Diagnostic Terms

• Global developmental delay
• Intellectual disability
• Mental retardation

As a parent, which term would you prefer to hear?
As a professional, which terms are you uncomfortable using?

- Global developmental delay
- Intellectual disability
- Mental retardation
- I’m comfortable with all of them

Which “diagnosis” has the worst prognosis in terms of outcome?

- Global developmental delay
- Intellectual disability
- Mental retardation
- They are all the same

Definitions

Global Developmental Delay (GDD)

- Significant delays in 2 or more developmental domains: gross/fine motor, speech/language, cognition, personal/social and activities of daily living

- Unless they are told otherwise, the term “delay” suggests to parents that the child is “slow” and will “catch up”

Definitions

Mental Retardation (MR)

- “Significantly sub-average general intellectual function accompanied by limitations in adaptive functioning”

- An older term that was typically applied to school aged children, but clearly implies that the deficits will be permanent

Which terms do you feel you understand well?

- Global developmental delay
- Intellectual disability
- Mental retardation
- None of them
Definitions

Intellectual Disability (ID)

- **The new “PC”** medical and educational term that has replaced mental retardation
- Defined the same as MR, but considered by many as less “harsh” or demeaning
- Focuses specifically on the impairment of intellectual function, independent of any other “mental” or emotional problems

GDD / ID / MR

- “Significant” typically defined as a level of functional impairment that is more than 2 SD below the mean, or a delay of more than 30%
- Since there is no single etiology for GDD/ID/MR, there is variability what is implied and understood with respect to “permanence” of the condition

Global Developmental Impairment

- GDD/GDI is typically applied to children under 5 years of age, although many professionals continue to use the term well beyond that age
- ID is a new term that is considered less stigmatizing, but may be confusing and misleading; never-the-less, it is becoming the accepted terminology

Severity of GDI / ID

- The severity of GDI / ID is determined “numerically” and “statistically” by the extent of the delay
- Mild – Moderate – Severe – Profound
- Each level represents statistical variation away from “average” performance

Developmental Quotient (DQ)

\[
\text{Age equivalent} \times 100 = \text{DQ} \\
\text{Mean} = 100 \\
\text{SD} = 15
\]
Severity of GDI / ID

**DSM-IV**

- **Mild**  
  DQ’s 55-70  
  (30-45% delayed)
- **Moderate**  
  DQ’s 40-54  
  (46-60% delayed)
- **Severe**  
  DQ’s 25-39  
  (61-75% delayed)
- **Profound**  
  DQ’s < 25  
  (>75% delayed)

Severity of GDI / ID

**The severity of GDI / ID is also defined by level of independence with the “gold standard” being full independence**

Severity of GDI / ID

**AAMR**

- **Mild**  
  51 to 75
- **Severe**  
  < 50

*Based on an increased likelihood of:*

- An identifiable cause
- Inability to benefit from formal academics
- Co-morbid health and behavioral disorders

Identifying Children with Global Developmental Impairment / Intellectual Disability

- Prevalence: 2 to 3% of children
- Up to 100,000 children in the USA
- Most (85%) will fall in the mild range
Identifying Children with GDI / ID

- Developmental Surveillance
- Developmental Screening
- Developmental Assessment
- Developmental Diagnosis

Developmental Surveillance

- Informal survey of development at each health maintenance visit
- Interview of parent impressions
- Brief review of milestones

Developmental Screening

- Standardized screen of development at specific intervals
- AAP recommends routine screening at 9 months and 24-30 months
- Screen any child who fails surveillance
- Ages and Stage vs. PEDS

Developmental Assessment

Step 1:

- Identify current functional skills in all areas of development including gross motor skills, non-language cognition (fine motor/visual motor/adaptive) and expressive and receptive language

Gross Motor Skills

- Sitting
- Crawling
- Cruising
- Walking
- Hop, skip, jump
**Non-language Cognition**

**Visual Motor / Adaptive Skills**
- Fine motor manipulation
- Cause and effect
- Spacial relationships
- Pencil/paper tasks
- ADL’s

**Expressive and Receptive Language Skills**
- Gestures
- Vocalization
- Identifying Objects
- Recognizing Pictures
- Following Commands

**Developmental Assessment**

**Step 2:**
- Identify level of performance / extent of deficit for age in each area using DQ’s

**Developmental Quotient (DQ)**

\[
\text{Age equivalent} \times \frac{100}{\text{Chronological age}} = \text{DQ}
\]

Mean = 100
SD = 15

**Levels of Performance**

- **Normal**  
  DQ’s 70-100  (no/minimal delay)
- **Mild**  
  DQ’s 55-70  (30-45% delayed)
- **Moderate**  
  DQ’s 40-54  (46-60% delayed)
- **Severe**  
  DQ’s 25-39  (61-75% delayed)
- **Profound**  
  DQ’s < 25  (>75% delayed)

**Developmental Diagnosis**

- Create and interpret a “DQ composite” that includes all aspects of development
- A diagnosis of GDI / ID should be applied only if ALL aspects of development are “significantly” impaired (2 SD below the mean, or DQ < 70)
**Developmental Composite #1**
Assessment at 18 months

- GM skills at 9 GM  
  \[ GM \text{ DQ} = 50 \]
- VM/Adaptive skills at 9 months  
  \[ VM/Adaptive \text{ DQ} = 50 \]
- Language skills at 6 months  
  \[ Language \text{ DQ} = 30 \]

**Developmental Composite #2**
Assessment at 18 months

- GM skills at 14 GM  
  \[ GM \text{ DQ} = 77 \]
- VM/Adaptive skills at 18 months  
  \[ VM/Adaptive \text{ DQ} = 100 \]
- Language skills at 9 months  
  \[ Language \text{ DQ} = 50 \]

**Coding for GDI / ID**

- Nonspecific encephalopathy (348.30)
- Alternative codes  
  - Delay of milestones (783.42)
  - Lack of normal physiological development (783.40)
- Do NOT use Developmental Delay (315)

**Developmental Composite #1**
Assessment at 18 months

- All DQ’s < 70
- Child *has* GDI / ID

**Developmental Composite #2**
Assessment at 18 months

- DQ’s variable, only language is < 70
- Child *does not have* GDI / ID

**What To Do Next?**
Medical Evaluation to Determine Etiology

- An etiology can be identified in approximately 50% of children with GDI / ID with a range from 10 to 80%
- Etiology established by history and physical examination in 15-30%

Medical Evaluation

- In children with mild GDI / ID, the most common etiology is non-specific inheritance
- If the family history is positive for mild GDI / ID, additional medical testing is not necessary unless there are clinical features to suggest a specific genetic disorder

Differential Diagnosis of GDI / ID

- Metabolic disorder
- Genetic disorder
- Congenital brain malformation
- Acquired brain injury
- Impaired processing / wiring

Medical Evaluation

- Lead screen
- Thyroid screen
- Metabolic studies
- EEG
- Genetic studies
- Brain imaging (MRI)

Medical Evaluation

- **Lead Screen**: Lead level >10ug/dL found in 10% of children living in endemic areas
- **Thyroid Screen**: Congenital hypothyroidism identified in up to 4% of children with cognitive impairment in the absence of neonatal screening (0% with neonatal screening)
Medical Evaluation

- **Metabolic Studies:**
  Less than 1% yield on routine metabolic screening for inborn errors of metabolism

- **EEG:**
  Less than 1% yield for specific etiology on EEG

Genetic Studies

Sub-telomeric deletions

- Prader Willi Syndrome
- Deletion on Chromosome #15

Special Studies

- Rett Syndrome
- MECP2

Genetic Studies

Chromosome Analysis

- Cri du chat
- 5p-

Special Studies

- Fragile X syndrome
- Multiple repeats in the FMR1 gene on the X chromosome

Medical Evaluation

- Chromosome analysis: yield 3.7%
- Subtelomeric deletions: yield 6.6%
- Fragile X (FMR1): yield 2.6%
- Rett syndrome (MECP2): yield ??
Medical Evaluation

*Brain Imaging Studies*

- Identifies congenital malformations and acquired brain injury
- Structural abnormalities are seen on 30 to 50% of MRI’s of children with GDI / ID; yield much lower with CT

Recommendations for Testing for GDI / ID

- MRI scan
- Genetic Micro-array (includes karyotype and analysis for subtelomeric and other DNA deletions)
- Separate chromosome analysis needed only to determine translocations

Structural Abnormalities

- Lissencephaly
- Cerebral Atrophy

Recommendations for Testing for GDI / ID

- DNA for Fragile-X in boys with no etiology
- DNA for Rett’s in girls with no clear etiology
- No indication for metabolic studies, lead screen, thyroid screen, or EEG

Additional Consultations

- Audiological evaluation??
- Vision evaluation??
- Neurology consultation??
- Genetic consultation??

What Do You Tell the Family??
Giving Bad News to Families

- Describe levels of performance
- If appropriate, introduce “mental retardation / ID”, and use the term “currently functioning”
- Indicate that the problem is in the brain – poor brain development or formation

Giving Bad News to Families

- Indicate that services and support are available
- Code as nonspecific encephalopathy (348.30)
- Be as positive as possible – indicate probabilities, but no “crystal ball”

Explaining the Diagnosis

- Testing may identify a specific etiology
- Literature regarding specific diagnoses is now readily available through the internet
- Many specific etiologies have a somewhat predictable pattern of development and a predictable outcome

Explaining the Diagnosis

- If all testing is “normal”, a clear etiology may not be identified, but the child still has an impairment of brain processing and brain function
- This is most likely caused by “faulty wiring” with inefficient synaptic connections that result from problems with neuronal arborization, myelination, and pruning

Dendritic Arborization

Synaptic Connections
Myelination

Pruning

Explaining the Diagnosis

• Children with even severe GDI / ID may have no clear etiology

• Families may have difficulty understanding and accepting that their child is severely impaired if all the tests are normal, since they expect that there is always an answer for everything

Predicting Outcome

• Contrary to many opinions, a the outcome of children with GD / ID / MR CAN be predictive in children under 5 years of age

• Reliability of outcome prediction depends on multiple factors including age of evaluation, severity of impairment, consistency of impairment, and the presence of a clear etiology or evidence of brain damage
Positive Predictive Factors

- Older age
- Greater severity of impairment
- Consistency across areas of development
- Consistency of impairment over time
- Clear etiology or evidence of brain damage

Summary

- Explore etiology through medical testing – does NOT require a specialist
- Provide sensitive guidance to families, but do not ignore difficult discussions about outcome
- Recognize that GDD / ID / MR are flip sides of the same coin – using more “sensitive” terms may confuse and mislead families

References