Global Developmental Delay and Intellectual Disability: DD Plus Update

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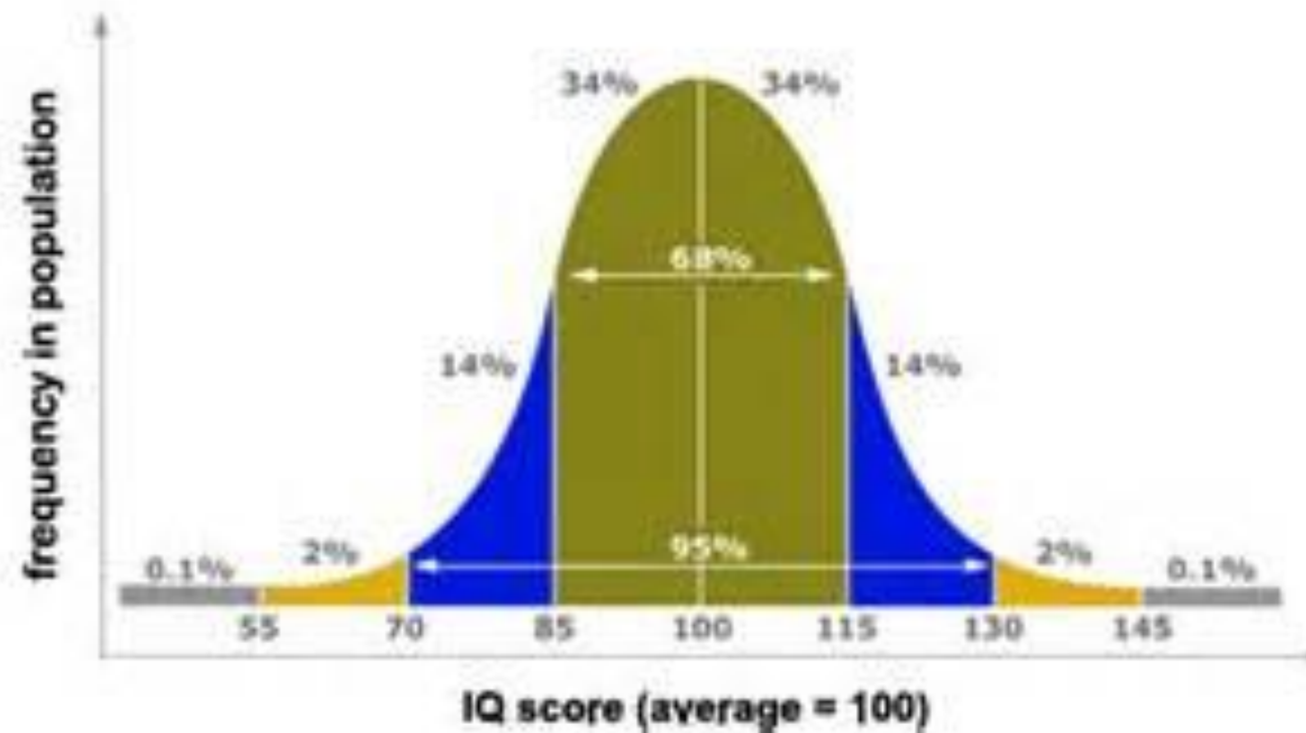
Intellectual Disabilities*

- 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



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

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

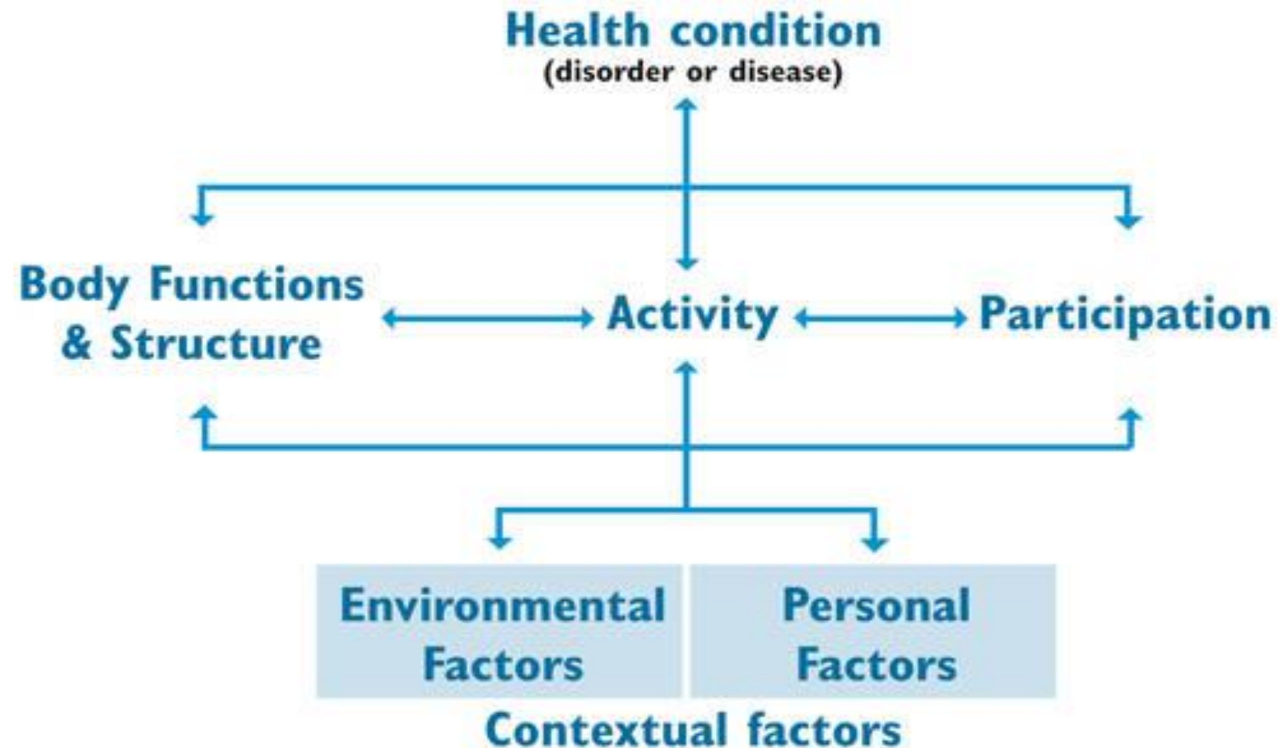
Outcome of child with moderate ID*

- Learns at 1/3 to ½ usual rate
- Adult reading 1st to 4th 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

	per 1000
ADHD	70
LD	80
Global developmental delay	50
Developmental language disorder	50
Developmental coordination disorder	50
Mild intellectual disability (mild ID)	15
Autism spectrum disorders	15
Moderate-severe intellectual disability	5
Cerebral palsy	4
Fetal alcohol syndrome	3
Vision impairment or deafness	3

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

  Shevell M. et al. Practice parameter (AAN). Neurology 2003;60:370-380

  Moeschler J, Shevell M. Pediatrics 2014;134:e903

Value of etiologic diagnosis

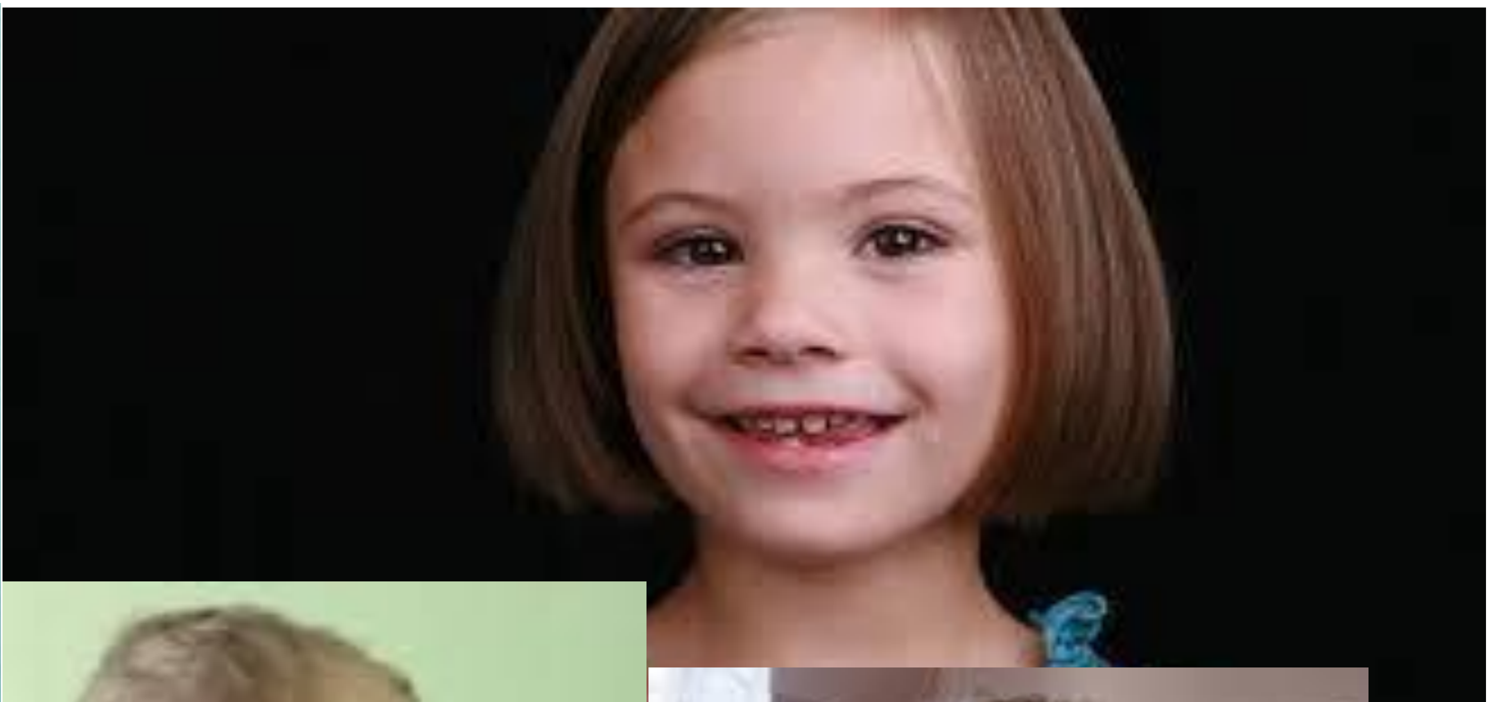
- Specific treatment implications
- Ongoing medical management of associated conditions
- Prognostic implications
- Assessment of recurrence risk
- Dispelling myths and pseudo-diagnoses
- Limiting further unnecessary testing

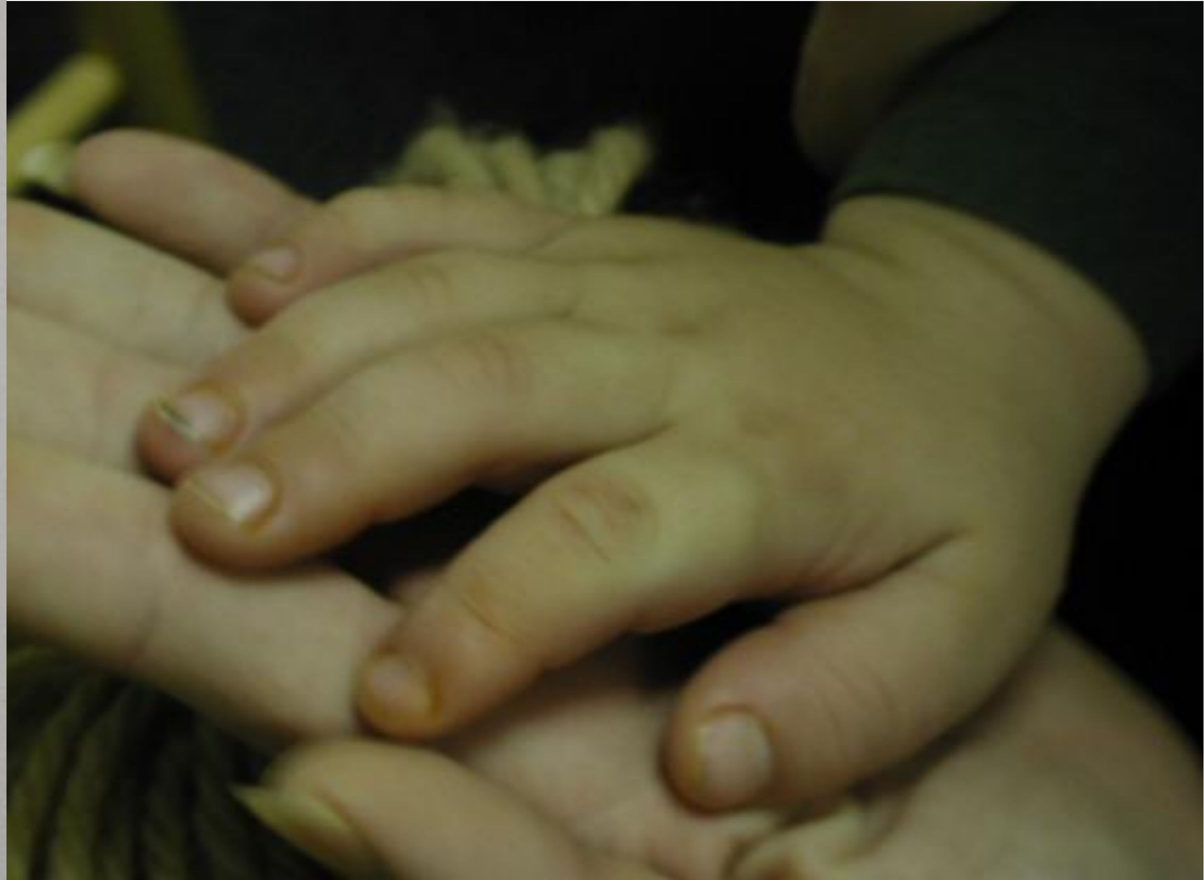
Limitations and drawbacks of etiologic search

- 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

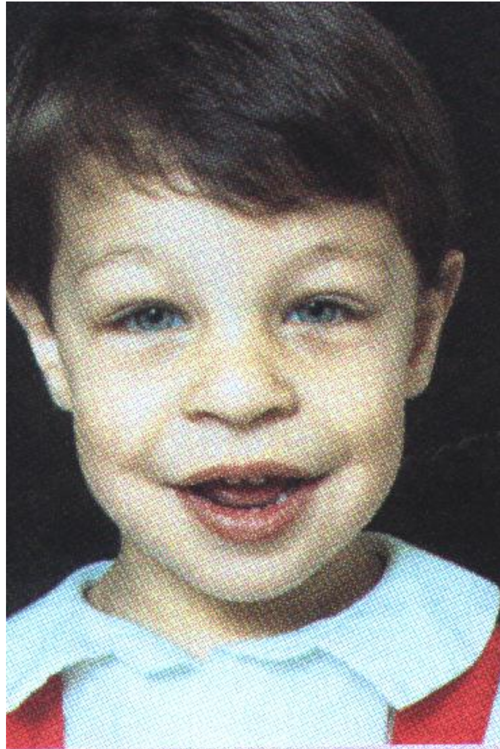
Recommendations: Metabolic and genetic tests*

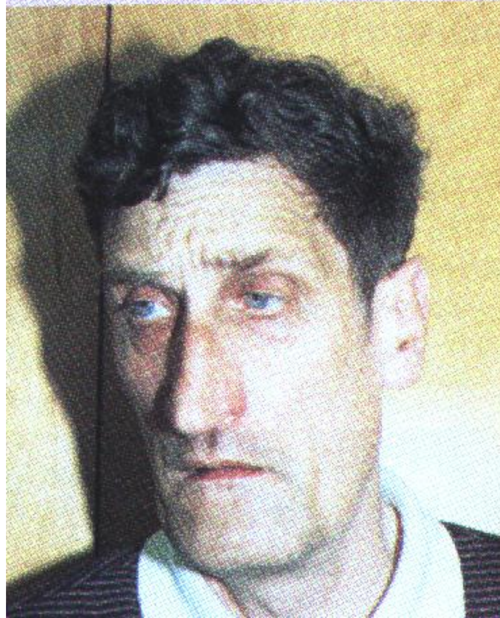
- Routine metabolic screening not indicated, unless:
  - no newborn metabolic screening
  - indicated by history, physical or lab
- Routine CMA
- Fragile X DNA with clinical preselection
- MECP2 studies in females with moderate to severe ID/GDD
- PTEN screen in autism with macrocephaly (risk of tumors)





Fragile X Syndrome





ID - Associated conditions

	Mild ID	Severe ID
Seizures	10%	20%
Hearing impairment	7%	10%
Vision impairment	1%	15%
Cerebral palsy	10%	20%
Sleep disorders	30%	75%
Recurrent vomiting		10%
Autism	12%	30%
Psychiatric disorders	**30%**	**50%**
("Dual diagnosis")		

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
- Use of the FAST http://adapt-fl.com/files/FAST.pdf

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