Intellectual Developmental Disorders

Laura Carpenter, PhD

Additional lecture credits:
Shawna McCafferty, MD & Jane Charles, MD

Objectives

- Understand the prevalence of developmental disabilities (DD)
- Differentiate global developmental delay (GDD) and intellectual disability (ID)
- Define common syndrome specific causes of ID
- Compare and Contrast IEPs and 504 plans

Definitions

- **Neurodevelopmental Disorders (DSM-5):** Disorders of brain development with onset in childhood, affecting cognition, behavior, or motor function.
- **Developmental Disabilities (Legal/Service Term):** Chronic conditions due to physical and/or mental impairments, emerging before age 22, likely lifelong.

Overlapping Terminology

- Developmental Disability (legal/educational term)
  - Intellectual disability
  - Autism spectrum disorder
  - Cerebral palsy
  - Epilepsy
  - Genetic conditions (e.g., Rett syndrome, spina bifida)
- Neurodevelopmental Disorder (DSM-5 Term)
  - Intellectual disability
  - Autism spectrum disorder
  - ADHD
  - Specific learning disorder
  - Communication disorders
  - Motor disorders
  - Global developmental delay

Developmental disabilities are common

CDC, Boyle et al., 2011; Zablotsky et al., 2019; National Health Interview Survey data

Developmental Disability	% Prevalence	Notes
Any developmental disability	~17%	About 1 in 6 U.S. children
ADHD	9.8%	Most common NDD
Learning Disability	7.7%	Includes reading, writing, and math disorders
Speech or Language Disorder	7.0–8.0%	Common in early childhood
Developmental Delay (unspecified)	3.5–4.0%	Used when child is too young for specific diagnoses
Autism Spectrum Disorder (ASD)	~3.2%	1 in 31 children (age 8); prevalence continues to rise
Intellectual Disability (ID)	1.2–1.5%	May co-occur with other conditions
Epilepsy/Seizure Disorder	0.7%	Often co-occurs with ASD, ID, or CP
Cerebral Palsy (CP)	0.3%	Most common motor disability in children
Vision Impairment	0.14%	Not correctable with glasses
Hearing Loss	0.12%	Permanent hearing loss; more common in NICU graduates

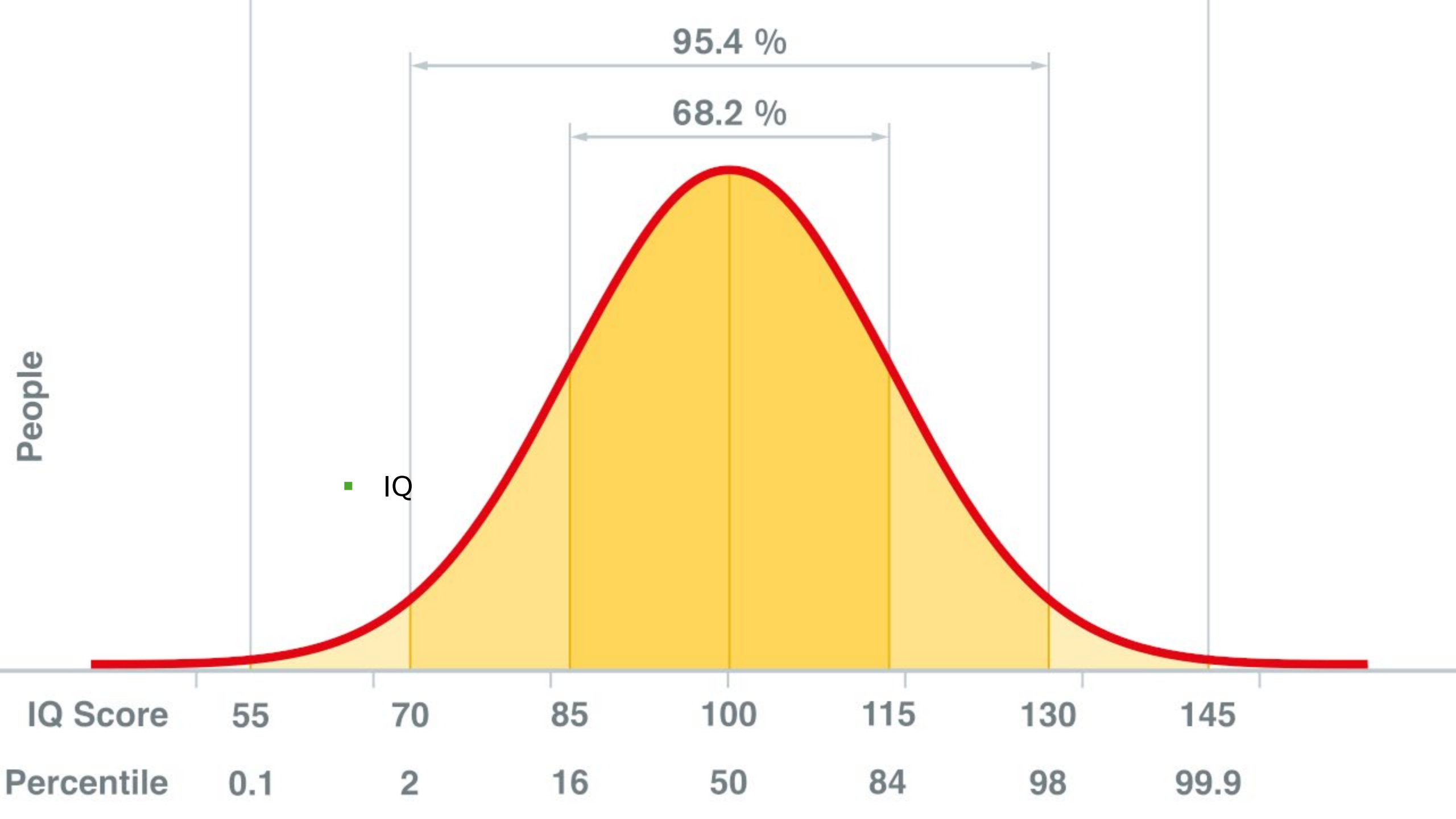
Global Developmental Delay and Intellectual Disability

GLOBAL DEVELOPMENTAL DELAY

- Diagnosis for children aged 0 to 5 years with significant delays in 2 or more areas of development
- May be transient
- 30-60% of children with GDD will transition to a diagnosis of ID after 5 years of age.
- Worse outcomes for those with:
  - More severe delays
  - Delays in multiple areas
  - Known genetic/neurological cause

INTELLECTUAL DISABILITY

- Onset before the age of 18
- Deficits in cognitive functioning (reasoning, learning, problem solving)
- Deficits in adaptive behavior which covers a range of everyday social and practical skills


95.4 %
68.2 %
People
IQ
IQ Score 55 70 85 100 115 130 145
Percentile 0.1 2 16 50 84 98 99.9

Cognitive Functioning

- IQ testing measures intellectual functioning, the capacity for learning, reasoning, problem solving, memory, processing speed
- Average IQ Score is 100
- Scores > 2 SD below average (Full Scale IQ <70) → ID
- Full Scale IQ 70-84 = Borderline Intelligence / Slow Learner

Adaptive Behavior

- **Conceptual**
  - Memory, language, academics, judgement
- **Social**
  - Social responsibility, self-esteem, gullibility, naïveté (i.e., wariness), social problem solving, and the ability to follow rules, obey laws, and avoid being victimized
- **Practical**
  - Activities of daily living (personal care), occupational skills, healthcare, travel/transportation, schedules/routines, safety, use of money, use of the telephone

- Impairment in ONE domain is sufficient to meet DSMV definition of Intellectual Disability

Level of Severity

ID Level	IQ	Level of Support
Mild	55 to 70	Intermittent
Moderate	40 to 55	Limited
Severe	25 to 40	Extensive
Profound	<25	Pervasive

Level of severity is primarily based on level of adaptive functioning and support required

	Mild	Moderate	Severe	Profound
Conceptual Skills	Ψ some learning difficulties; Ψ might require some support in academic skills	Ψ slow in learning with difficulties using language Ψ might require more support in academic skills	Ψ limited understanding of language and/ or concept	Ψ unable to understand conceptual skills or follow instructions
Social Skills	Ψ some difficulties perceiving social cues; Ψ some social immaturity with some difficulties regulating emotions and behaviours; Ψ might be at risk of being manipulated by others	Ψ some difficulties with communication; Ψ difficulties perceiving and interpreting social cues; Ψ limited ability to participate in social activities Ψ might be at a higher risk of being manipulated by others	Ψ limited communication skills; might use single words or phrases with gestures; Ψ limited ability to perceive and interpret social cues Ψ might not be able to participate in social activities	Ψ rudimentary communication skills; might use non-verbal communication; Ψ unable to perceive or interpret social cues; Ψ unable to participate in social activities
Practical Skills	Ψ might be fully independent with early intervention; Ψ might require some support with complex daily living tasks;	Ψ might not be fully independent Ψ might be able to get around to familiar places	Ψ unable to be fully independent; Ψ need regular support and supervision for daily activities Ψ some motor impairments	Ψ unable to be fully independent; Ψ need constant help and supervision at all times Ψ may be incontinent or immobile

Causes of Intellectual Disability

Genetic Causes

- Chromosomal Disorders
  - Down syndrome (Trisomy 21) – Most common genetic cause
  - Trisomy 18 (Edwards), Trisomy 13 (Patau)
  - Turner syndrome (45,X) – Learning disabilities
  - Klinefelter syndrome (47,XXY)
  - Cri du chat syndrome (5p deletion)
  - Williams syndrome (7q11.23 deletion)
  - Wolf-Hirschhorn syndrome (4p deletion)
  - 22q11.2 deletion syndrome (DiGeorge)
- X-linked
  - Fragile X syndrome – Most common inherited cause
  - Rett syndrome – Females; regression and seizures
  - Lesch-Nyhan syndrome – Self-injury, dystonia
  - MECP2 duplication
- Autosomal Dominant
  - Tuberous sclerosis complex – Seizures, autism
  - Neurofibromatosis type 1 – Learning issues
  - Noonan syndrome, CHARGE syndrome
- Autosomal Recessive:
  - Phenylketonuria (PKU) – Preventable ID
  - Sanfilippo syndrome, Canavan disease
  - Smith-Lemli-Opitz, Tay-Sachs disease
- Microdeletion / Imprinting Disorders
  - Angelman syndrome – Happy demeanor, seizures
  - Prader-Willi syndrome – Hypotonia, hyperphagia
  - Smith-Magenis syndrome – Sleep, behavior issues
  - Phelan-McDermid syndrome – SHANK3-related

Acquired Causes

- Prenatal Causes
  - Congenital infections
    - TORCH, *Rubella, CMV, Herpes*
  - Maternal substance use
  - Maternal illnesses
  - Radiation or toxin exposure
- Perinatal Causes
  - Birth asphyxia / hypoxic-ischemic encephalopathy (HIE)
  - Extreme prematurity
  - Complicated labor/delivery
- Postnatal Causes
  - Severe infections
    - *Meningitis, encephalitis, sepsis*
  - Traumatic brain injury (TBI)
  - Severe malnutrition – especially during critical brain growth
  - Environmental deprivation / neglect
  - Lead poisoning
  - Near-drowning, stroke, or cardiac arrest

Most common causes

- Down Syndrome
- Fragile X Syndrome
- Fetal Alcohol Syndrome

DOWN SYNDROME

Most common genetic cause of ID

Occurs in about 1/800 live births in all ethnic groups

DOWN SYNDROME: CYTOGENETIC CAUSES

Trisomy 21: 95%

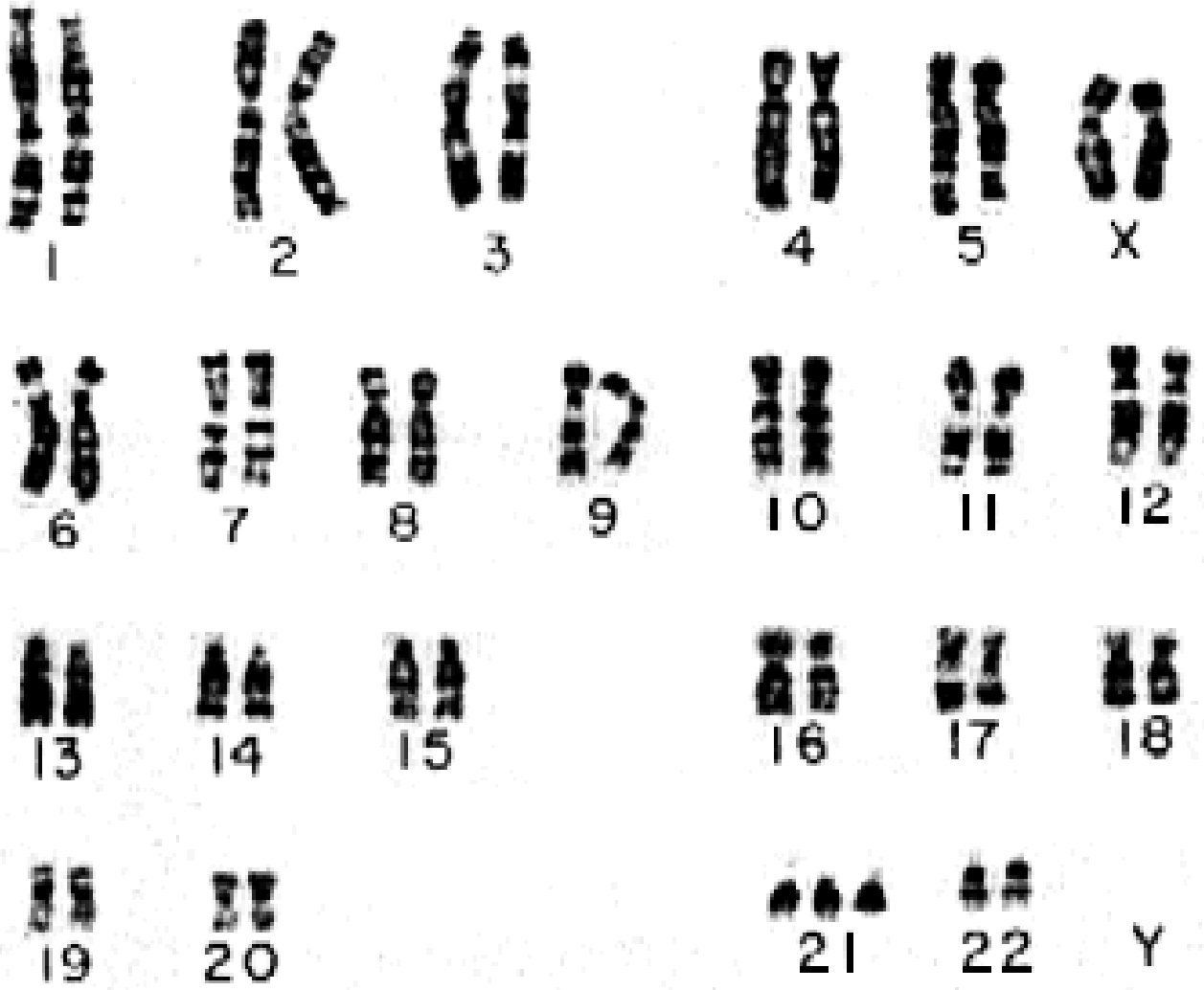
- Non-disjunction results from unequal chromosome division, usually in the mother's egg production

Translocation: 3-4%

- The extra chromosome 21 is permanently attached to another chromosome causing a translocation

Mosaicism: 1%

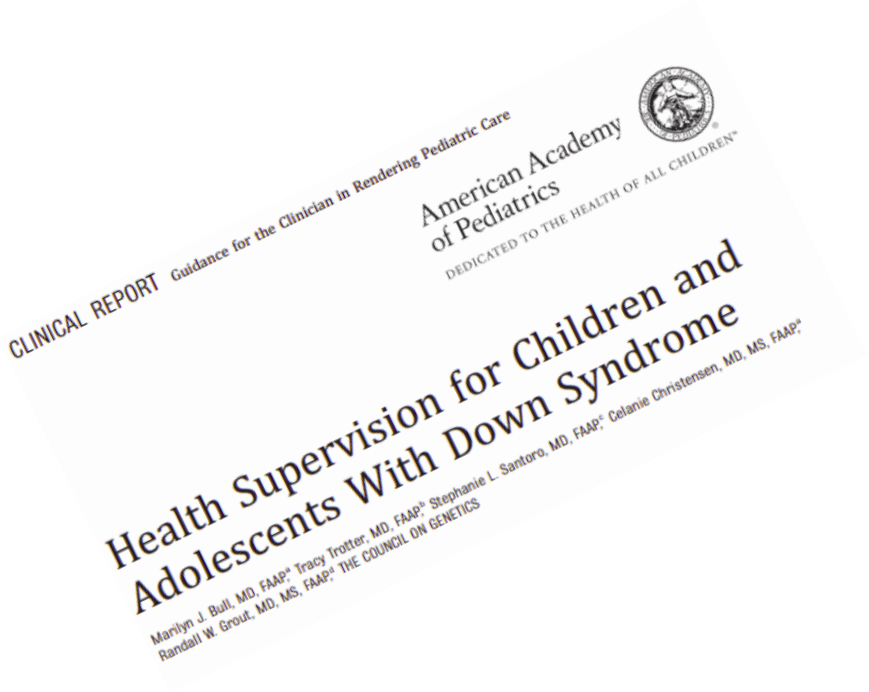
- Two populations of cells, the trisomy 21 cells, and a second cell line, usually normal



Intellectual Disability in Down Syndrome

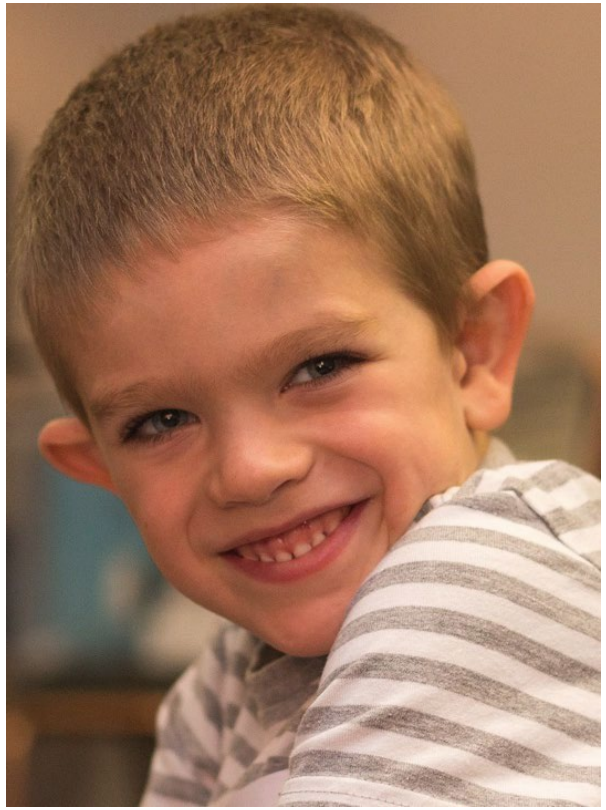
- Majority have mild to moderate ID
- Starts in the first year of life
- Average age of sitting (11 mo) and walking (26 mo) is twice the typical age
- First words at 18 months (compared to 12-15 months)
- IQ declines through the first 10 years of age, reaching a plateau in adolescence that continues into adulthood

System	Common Conditions in Down Syndrome
Cardiac	- Congenital heart defects (40–50%)
Neurological & Developmental	- Intellectual disability (100%) - Early-onset Alzheimer's disease - Hypotonia
Musculoskeletal	- Atlantoaxial instability (up to 20%) - Joint laxity - Ligamentous hypermobility
ENT / Sensory	- Hearing loss (up to 75%) - Vision problems (strabismus, cataracts, refractive errors) - Chronic ear infections - Obstructive sleep apnea
Endocrine	- Hypothyroidism (10–15%) - Obesity - Diabetes
Hematologic / Immunologic	- Increased risk of leukemia (AMKL, TAM)
Gastrointestinal	- GI anomalies (e.g., duodenal atresia, Hirschsprung disease) - Feeding difficulties - Constipation



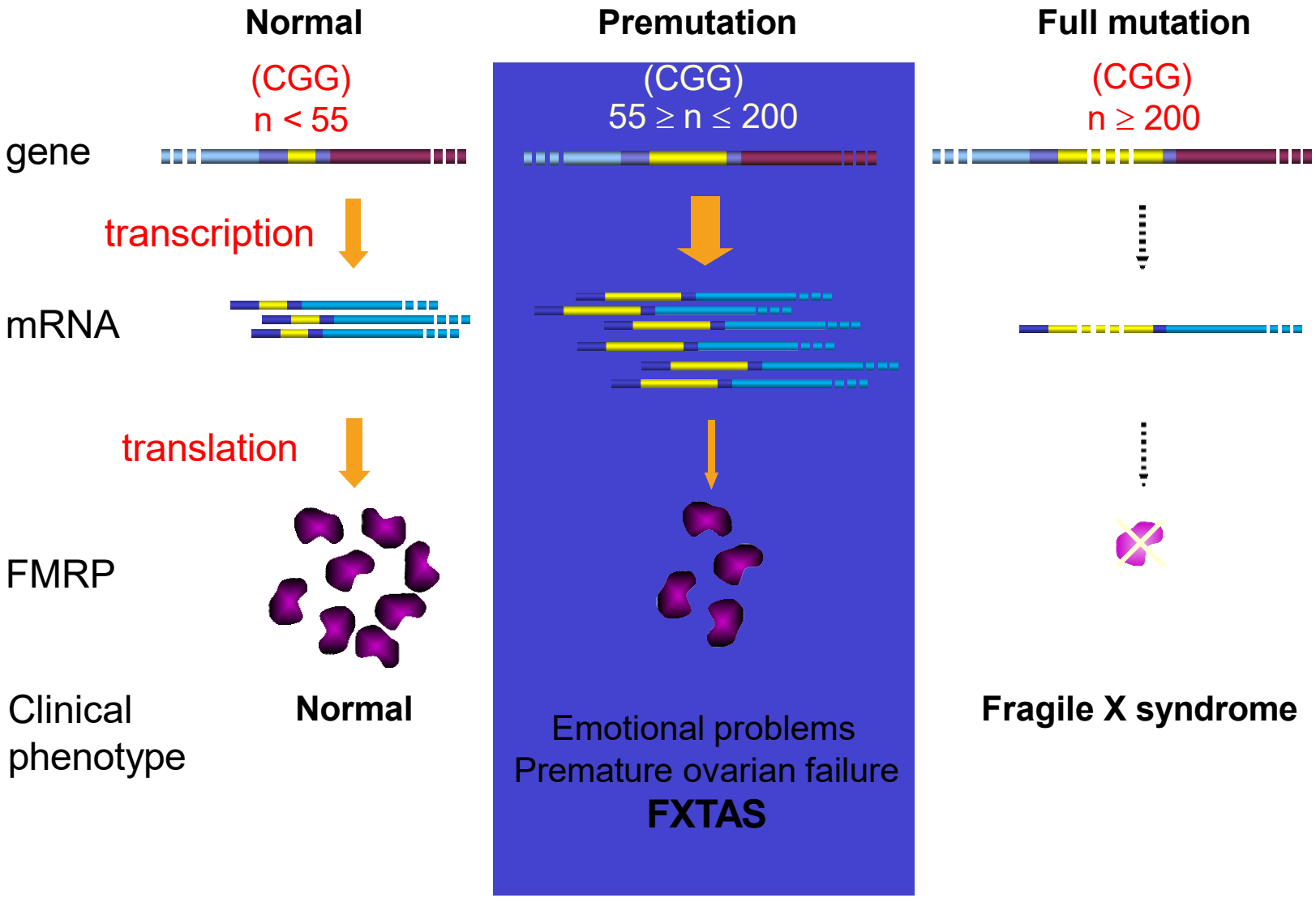
Come visit our Down Syndrome Clinic in Developmental Behavioral Pediatrics led by Dr. Angela LaRosa!

Fragile X



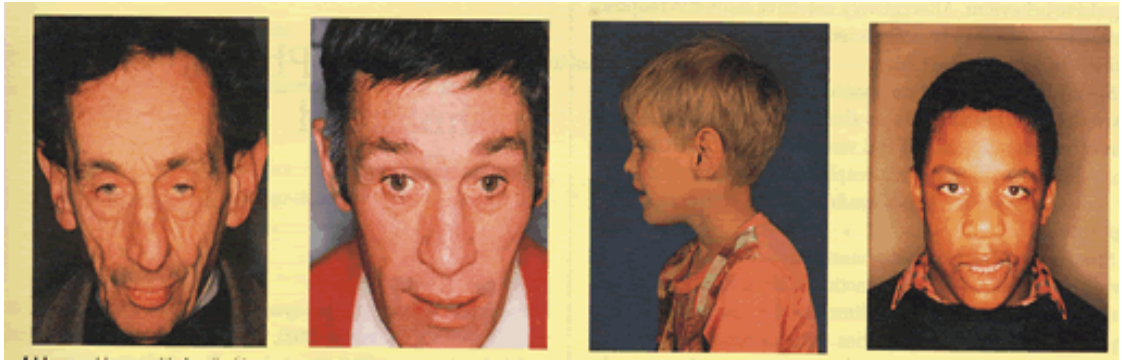
- Most common inherited cause of intellectual disability
- X-linked dominant disorder caused by FMR1 gene mutation
- Full mutation ~1 in 4,000 males and ~1 in 8,000 females
- More severe in males
- Intellectual disability (mild to severe)

Expression of the FMR1 Gene

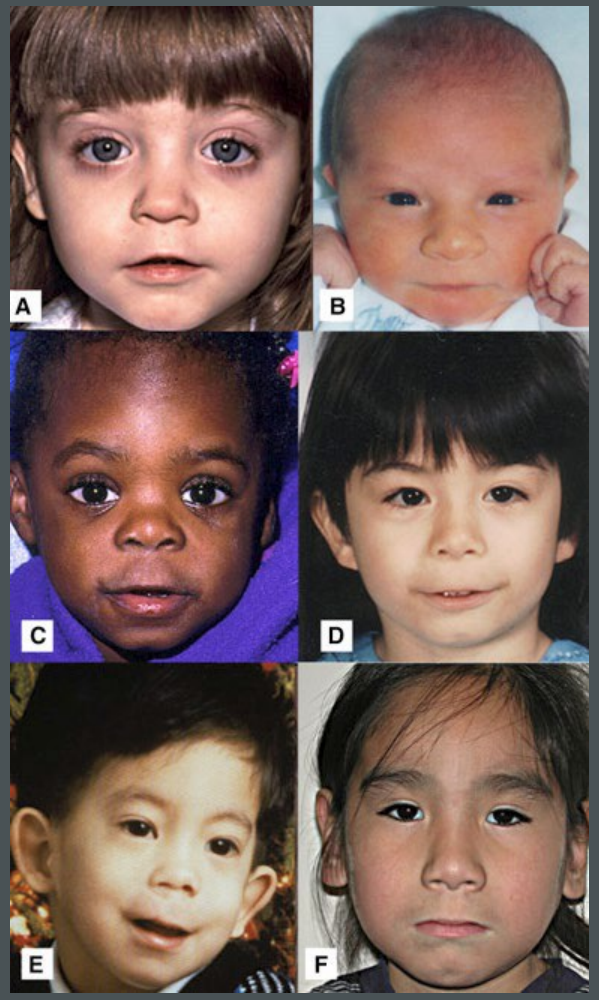


Features of Fragile X

- **Physical Features**
  - Long face, large ears, prominent jaw and forehead
  - Macroorchidism (post-pubertal males)
  - Connective tissue signs: hyperextensible joints, flat feet
- **Behavioral & Developmental Features**
  - Poor eye contact, social anxiety, hand flapping
  - Sensory sensitivities
  - Delays in motor and adaptive skills
- **Associated Conditions**
  - Autism spectrum disorder
  - Seizures (~15–20%)
  - Mitral valve prolapse, strabismus



FETAL ALCOHOL SYNDROME



- Leading Cause of Acquired and Preventable ID
- Prevalence: .3-.8% (1-5% for Fetal Alcohol Spectrum Disorders)
- Dose-dependent effect
- Phenotype: microcephaly and growth retardation, facial features such as short palpebral fissures, thin upper lip,smooth philtrum
- Mild to Moderate ID
- Associated with ADHD and Behavioral problems

High Risk Population

Risks

Lack of awareness of risk and danger	Lack of communication skills	Social problems
• accidental injury	• Untreated medical conditions • disruptive behaviors	• Gullibility or naiveté in social situations • Exploitation (fraud, unintentional criminal involvement, false confessions) • Risk for abuse.

Carolina Autism Transition Study (CATS)

- 4,988 children identified through South Carolina Autism and Developmental Disabilities Network (SC-ADDM)
  - ASD = 607
  - ID = 1,280
  - Population control = 3,101

DSS
SOUTH CAROLINA
DEPARTMENT of SOCIAL SERVICES

dss.sc.gov

ADDM NET

CDC

Young adults with ID were…

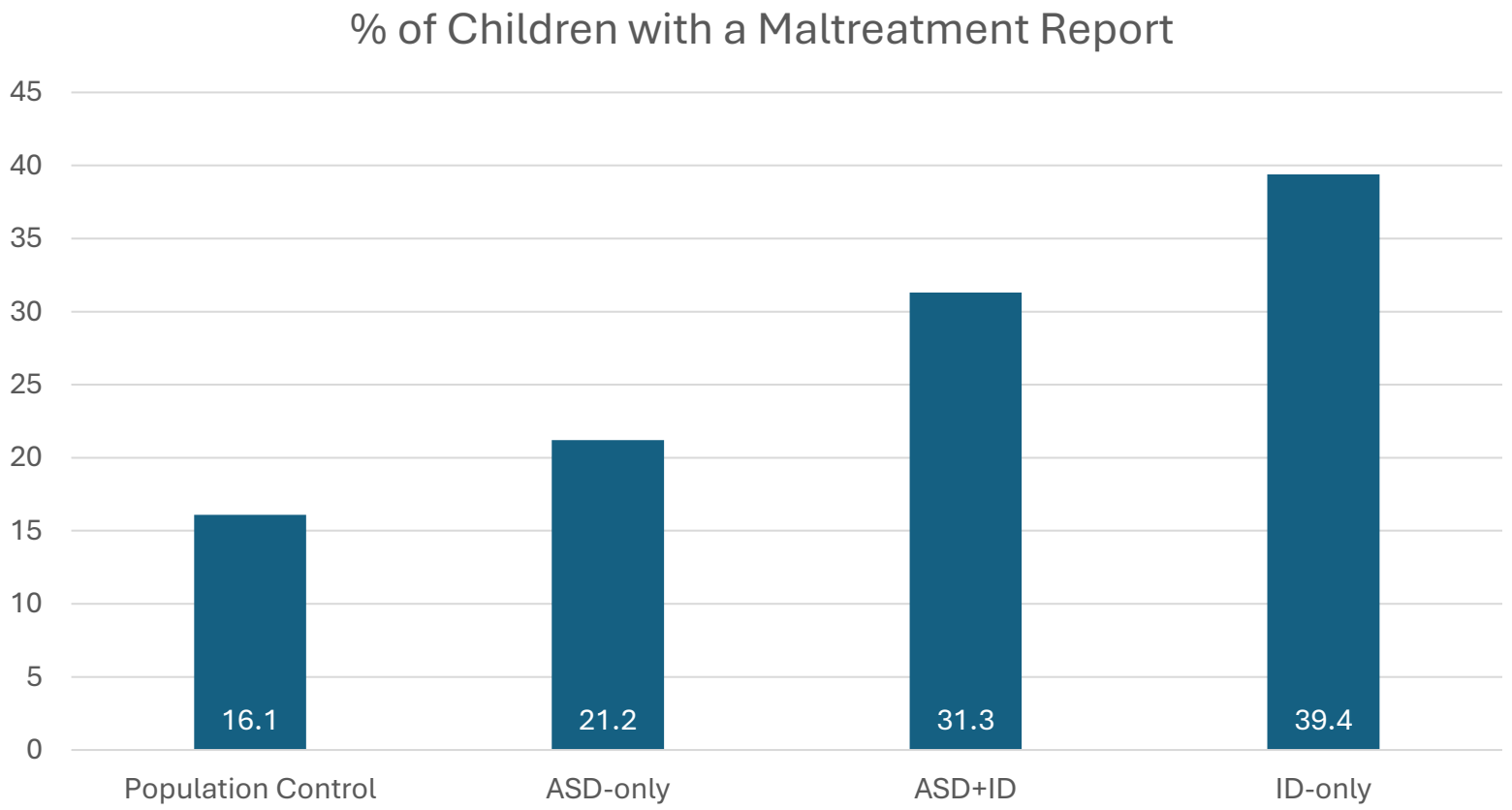
More likely to have reported and substantiated maltreatment

More likely to have passed away before reaching young adulthood (chronic disease vs. accidental injury)

More like to have been treated in the ED (and had more ED encounters overall)

Hand, BN., Boan, A. D., Bradley, C. C., Charles, J. M. and Carpenter, L. A (2018

Elevated Rates of Abuse and Neglect in ASD and ID



McDonnell et al. (2019), *Journal of Child Psychology & Psychiatry*

ADULTS WITH ID/DD ARE LESS LIKELY TO…

Receive cancer screenings:

Pap Smears, Mammograms, PSA

Receive dental, hearing and vision screening

ADULTS WITH ID/DD HAVE…

GREATER NUMBER OF HEALTH PROBLEMS

GREATER RISK FOR NEUROLOGICAL PROBLEMS, SUCH AS EPILEPSY, EARLY DEMENTIA.

GREATER RISK FOR OBESITY DUE TO SEDENTARY LIFESTYLE, SOME MEDICATIONS, LACK OF ACCESS TO RECREATIONAL AND FITNESS ACTIVITIES

GREATER RISK FOR PSYCHIATRIC DISORDERS

GREATER RISK OF GI D/O

GREATER RISK DENTAL DISEASE

Why the drop-off in care for adults with ID?

- No standard clinical guidelines exist
- Physicians caring for adults may lack exposure to community resources
- Not enough time for complex patient visits in current primary care setting
- Physical barriers
  - accessing exam room
  - sensory issues in office
- Communication barriers
  - Patient not always included in discussion of healthcare
- Behavioral barriers

Interventions and Support

Interventions

- Infants and Toddlers <3y
  - Individualized Family Service Plan (IFSP) through statewide Early Intervention (called BabyNet in SC)
- School Age (3-21)
  - Individualized Education Plan (IEP) or 504 Plan through public school
- Young Adulthood
  - Individualized Transition Plan (ITP) at 16y

	IDEA (Individuals with Disabilities Education Act)	Section 504 (Rehab Act of 1973)
Purpose	Special education services	Equal access and accommodations
Who Qualifies?	Children with specific disabilities impacting learning	Broader—any disability that limits a major life activity
Ages Covered	3–21 years	All ages, including college
Plan Type	Individualized Education Program (**IEP**)	504 Plan
Services Provided	Specialized instruction, related services	Classroom and test accommodations
Legal Mandate	Federal special education law	Civil rights law
Funding	Federally funded	Unfunded mandate (local responsibility)

IEP

Legal document

Provides necessary specialized education and services to children with disabilities

- Educational Environment
- Therapies: SLP/OT/PT
- Behavioral Interventions
- Counseling
- Adaptive Communication Devices
- Classroom Modifications / Accommodations

Renewed annually with updated and individualized goals

Applicable through high school (Or 21yo)

504 Plan

- Covered by section 504 of the federal civil rights law, under the Americans with Disabilities Act (ADA)
- Provides accommodations/modifications based on medical diagnosis
    - Extended time on tests
    - Extra textbooks for home
    - Preferred seating
- Does not include funding for special services or instruction.
- Unlike the IEP, accommodations extend all levels of education, including college

Questions?