Global Developmental Delay

Global Developmental Delay (GDD) = when children **< 5 years old** experience a **significant delay** in at least **2 domains** of **development**

Developmental Domains:
- Gross Motor
- Fine Motor
- Speech/Language
- Cognitive
- Social

Children with **GDD** often go on to meet the criteria for **intellectual disability**

PRESENTATION

HISTORY

- **Developmental Milestones:** parental concerns, milestones met in each domain, regression
- **Birth:** prematurity, growth parameters, NICU – length of stay, complications, APGAR
- **Prenatal:** screening results, antenatal care, maternal medications, substance use, diabetes, HTN, infection
- **Family:** infant deaths, birth defects, neuro + genetic conditions, GDD, neurodevelopmental disability
- **Psychosocial:** abuse, neglect, parental substance use, involvement of children's services

PHYSICAL EXAM

- **Growth Parameters:** height, weight, head circumference (micro or macrocephaly)
- **Appearance:** dysmorphic features
- **Skin:** cutaneous stigmata
- **Head:** shape, fontanelles
- **Cardio:** murmurs, other abnormalities
- **GI:** hepatosplenomegaly
- **GU:** genital abnormalities
- **MSK:** spinal + limb abnormalities
- **Neuro:** tone, strength, reflexes, persistence or absence of primitive reflexes relative to age

INITIAL EVALUATION

- ❑ History and physical examination
- ❑ Audiology referral – if concerns in speech/language domain
- ❑ Ophthalmology/Optometry referral – deficits will guide management
- ❑ EEG – if suspect seizures on history

ADDITIONAL INVESTIGATIONS

- CBC, AST, ALT, glucose, urea – basic lab work
- Blood gases, electrolytes – to evaluate for acidosis, anion gap
- TSH – hypothyroidism; creatinine kinase – for muscular dystrophy
- Ammonia, lactate, homocysteine, carnitine – for metabolic testing
- Chromosomal microarray, fragile x testing – for genetic testing
- Brain MRI – if abnormal neuro exam, seizures, micro/macrocephaly

CAUSES

Genetic Causes	▪ Chromosomal disorders (e.g. trisomies), single-gene disorders (e.g. Fragile X, Rett syndrome) ▪ Inborn errors of metabolism
Prenatal Causes	▪ Toxins/Teratogens – alcohol, lead, radiation ▪ Congenital (STORCH) infections
Perinatal Causes	▪ Asphyxia, hypoxia, preterm birth ▪ Neonatal complications (infection, trauma, neonatal stroke)
Postnatal Causes	▪ Neglect/malnutrition, infections, trauma, toxins

MANAGEMENT

- Referral to subspecialist – developmental pediatrician, neurologist, genetics
- Early interventions – OT, PT, hearing, vision, SLP, social work, functional living skills
- Family support + education
- Management of underlying cause + comorbidities if applicable/if treatment available

June 2022

Dirusha Moodley (Medical Student, Western University), Dr. Hema Gangam (Pediatric Neurologist, Western University) for www.pedscases.com