Evaluation of global developmental delay

Global developmental delay is defined as significant delay in two or more of the following developmental domains: cognitive, fine/gross motor, speech/language, personal/social, and activities of daily living. The delay may be classed as mild if the functional age is < 33% less than chronological age, moderate if functional age is 34-66% of chronological age, and severe if functional age is > 66% below chronological age\(^1\). Approximately 1-3% of children under the age of 5 years are estimated to have global developmental delay\(^2\). Estimates of the diagnostic yield resulting in a definitive etiology for the developmental delay are highly variable, ranging from 10% to 81\%\(^{1,2,3,4}\).

History

- Detailed prenatal, perinatal, postnatal history. Pregnancy loss, threatened miscarriage, drug ingestion?
- Parental consanguinity.
- Family history of developmental disability or childhood death.
- Loss or regression of developmental milestones.

Examination

- Occipitofrontal circumference of child and parents, measured and plotted.
- Dysmorphic features
- Neurocutaneous lesions.
- Complete neurological examination
- Eyes (may need formal ophthalmology evaluation).

First Line Investigations\(^{5,6}\)

- 0-3 years of age: REFER TO REGIONAL CENTER FOR EVALUATION FOR EARLY INTERVENTION SEVICES!
- Over 3 years of age: REFER TO THE SCHOOL SYSTEM FOR EVALUATION FOR EARLY CHILDHOOD PROGRAM!
- Karyotype (Chromosomal microarray may replace karyotype as test of choice)
- DNA for Fragile X syndrome
- Lead level
- Thyroid function tests
- CBC (screening for iron deficiency)
- Comprehensive metabolic panel
- Uric acid (screen for purine disorders, which can cause isolated developmental delay; more stable than lactate and ammonia)
- Biotinidase
- Evaluations of vision and hearing
Second Line Investigations

- Neuroimaging (preferably MRI) if abnormal head size, seizures, focal neurological findings.
- EEG (sleep deprived) if speech regression, seizures, history suggestive of neurodegenerative disorder. Consider video EEG telemetry if frequent paroxysmal events, or speech regression.
- Genetics referral if dysmorphic features, family history.
- Metabolic workup if family history of metabolic disorders, consanguinity, regression, organomegaly, coarse facial features.
  - Blood: Lactate, amino acids, ammonia, very long chain fatty acids, carnitine, isoforms of transferrin, acylcarnitine profile
  - Urine: organic acids, oligosaccharides, glycosaminoglycans.

References: