**Developmental Delay (DD) or Intellectual Disability (ID) Testing**

**RISK FACTORS**
- Family history of genetic disorders/ID
- Neurocognitive dysfunction
- Cerebral palsy and static encephalopathy
- Hypotonia
- Seizure disorder
- Birth defects (eg, cardiac defect, cleft palate, club feet)
- Growth abnormalities
- Nonfamilial dysmorphic features
- Family history of recurrent miscarriages

**Evaluation for DD/ID**
Identify family history of risk factors

- Sufficient minor or major dysmorphic (atypical) features
  - Yes: Refer for genetics consultation
  - No: Presence of
    - Microcephaly
    - Macrocephaly
    - Focal findings on neurologic exam
    - Cerebral palsy
    - Hypotonia
    - Seizures
    - Autism/ASD
      - Yes: Order MRI/CT
      - No: May want to consider
        - Cytogenetic and/or molecular testing based on clinical presentation
        - Genetics consultation

- Episodic deterioration
  - Yes: Metabolic testing
    - Refer for metabolic consult
  - No: Cytojenomic SNP Microarray or Cytojenomic SNP Microarray Buccal Swab – first line testing for most developmental delay syndromes
    - Abnormal: Genetics consultation
      - Treat symptomatically
    - Normal: Order Fragile X (FMR1) with Reflex to Methylation Analysis
      - Note: test more likely to be positive in the following cases:
        - Physical features characteristic of Fragile X
        - Family history supportive of X-linked ID
        - Maternal family history of premature ovarian failure, ataxia and/or tumor

- Family history of metabolic disorder
  - Yes: Order Fragile X (FMR1) with Reflex to Methylation Analysis
  - No: Order Fragile X (FMR1) with Reflex to Methylation Analysis

**ORDER**
- Fragile X (FMR1) with Reflex to Methylation Analysis

**OTHER AVAILABLE TESTING**
- X Chromosome Ultra-High Density Microarray (consider this test if FMR1 testing is negative)
- Chromosome Analysis, Peripheral Blood
- Chromosome FISH, Metaphase
- Fragile X (FMR1) with Reflex to Methylation Analysis
- Rett Syndrome (MECP2), Sequencing and Deletion/Duplication
- Rett Syndrome (MECP2), Full Gene Sequencing
- Rett Syndrome (MECP2), Deletion and Duplication
- Angelman Syndrome and Prader-Willi Syndrome by Methylation-Sensitive PCR
- Angelman Syndrome (UBE3A) Sequencing
- Angelman Syndrome and Prader-Willi Syndrome by Methylation-Sensitive PCR, Fetal
- CDKL5-Related Disorders (CDKL5) Sequencing and Deletion/Duplication
- CDKL5-Related Disorders (CDKL5) Sequencing
- CDKL5-Related Disorders (CDKL5) Deletion/Duplication
- PTEN-Related Disorders (PTEN) Sequencing and Deletion/Duplication
- X-Linked Intellectual Disability Panel, Sequencing, 76 Genes

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