**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
  - Negative

**Cyto genomic SNP Microarray or Cytogenomic SNP Microarray Buccal Swab** – first line testing for most developmental delay syndromes

**OR**

**Cyto genomic SNP Microarray with Five-Cell Chromosome Study, Peripheral Blood** – useful if chromosome and array tests would otherwise have been ordered concurrently

**OTHER AVAILABLE TESTING**
- 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
- 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
- PTEN-Related Disorders (PTEN) Sequencing and Deletion/Duplication
- Autism and Intellectual Disability Comprehensive Panel
- Autism and Intellectual Disability Metabolic Panel

**Abnormal**

- Genetics consultation
- Treat symptomatically

**IF male with neurocognitive dysfunction**

**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

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