Testing for Genetic Syndromes Related to Developmental Delay (DD), Intellectual Disability (ID), and Autism Spectrum Disorder (ASD)

**INDICATIONS FOR TESTING**

Developmental delay (DD), intellectual disability (ID), and/or autism spectrum disorder (ASD) AND/OR One or more comorbidity (eg, dysmorphic features, seizures, congenital defects, psychiatric disorders, family history of recurrent miscarriages)

- Down syndrome, trisomy 13, or trisomy 18 suspected
- Conduct comprehensive medical history, family history, physical examination, and neurologic examination
- Specific disorder suspected

**ORDER**

- Chromosome analysis (with reflex to CMA if results are normal)
- DD/ID/ASD of unknown etiology
- Disorder-specific testing

**First-Tier Testing**

**ORDER**

- Cytogenomic SNP Microarray (Peripheral Blood or Buccal Swab) AND
  - Fragile X (FMR1) with Reflex to Methylation Analysis
  - Testing for metabolic and/or mitochondrial disorders (if clinical indicators present)

**CONSIDER**

- Medical genetics/genetic counseling referral

**Second-Tier Testing**

Consider additional specific molecular studies:
- MECP2 sequencing and deletion/duplication if patient is female
- PTEN-related disorders sequencing and deletion/duplication if patient has ASD and macrocephaly (head circumference >2.5 standard deviations)

X-linked disorder testing if indicated by clinical and family history

Whole exome sequencing

Brain imaging studies

**References**

