**RISK FACTORS**
- Family history of genetic disorders/ID
- Neurocognitive dysfunction
- Cerebral palsy and static encephalopathy
- Hypotonia
- Seizure disorder
- Birth defects (e.g., 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

- Episodic deterioration
  - Yes: Metabolic testing
    - Refer for metabolic consult
  - No: Cytogenomic SNP Microarray or Cytogenomic SNP Microarray Buccal Swab – first line testing for most developmental delay syndromes
  - OR
    - Cytogenomic SNP Microarray with Five-Cell Chromosome Study, Peripheral Blood – useful if chromosome and array tests would otherwise have been ordered concurrently

**Cyto genetic and/or molecular testing**
- May want to consider
  - Cytogenetic and/or molecular testing based on clinical presentation
  - Genetics consultation

**Cyto genetic and/or molecular testing**
- 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

**Order**
- MRI/CT

- Presence of
  - Microcephaly
  - Macrocephaly
  - Focal findings on neurologic exam
  - Cerebral palsy
  - Hypotonia
  - Seizures
  - Autism/ASD

**Metabolic testing**
- Refer for metabolic consult

**Abnormal**
- Genetics consultation
- Treat symptomatically