INDICATIONS FOR TESTING
DD and/or ID
WITH OR WITHOUT
One or more comorbidities (eg, dysmorphic features, seizures, congenital defects, psychiatric disorders, family history of recurrent miscarriages)

PERFORM
Comprehensive medical history, family history, physical examination, and neurologic examination

ORDER
Chromosome analysis

ORDER DISORDER-SPECIFIC TESTING
(depending on clinical indications):
- Testing for Rett syndrome
- Testing for PTEN-related disorders
- Testing for Angelman syndrome/Prader-Willi syndrome
- X-linked disorder testing
- Other nonlaboratory assessments

ORDER
CMA with fragile X testing
OR
WGS or WES with fragile X testing and consideration of microarray
OR
A panel test that includes genetic, fragile X, and metabolic testing
AND
Refer to medical genetics/genetic counseling

AND CONSIDER
Testing for metabolic and/or mitochondrial disorders

Etiology not identified
Etiology identified
Etiology not identified
Etiology not identified
Etiology consistent with patient phenotype identified
No further testing

First-Tier Testing

Second-Tier Testing

Etiology not identified
Etiology consistent with patient phenotype identified
No further testing

References