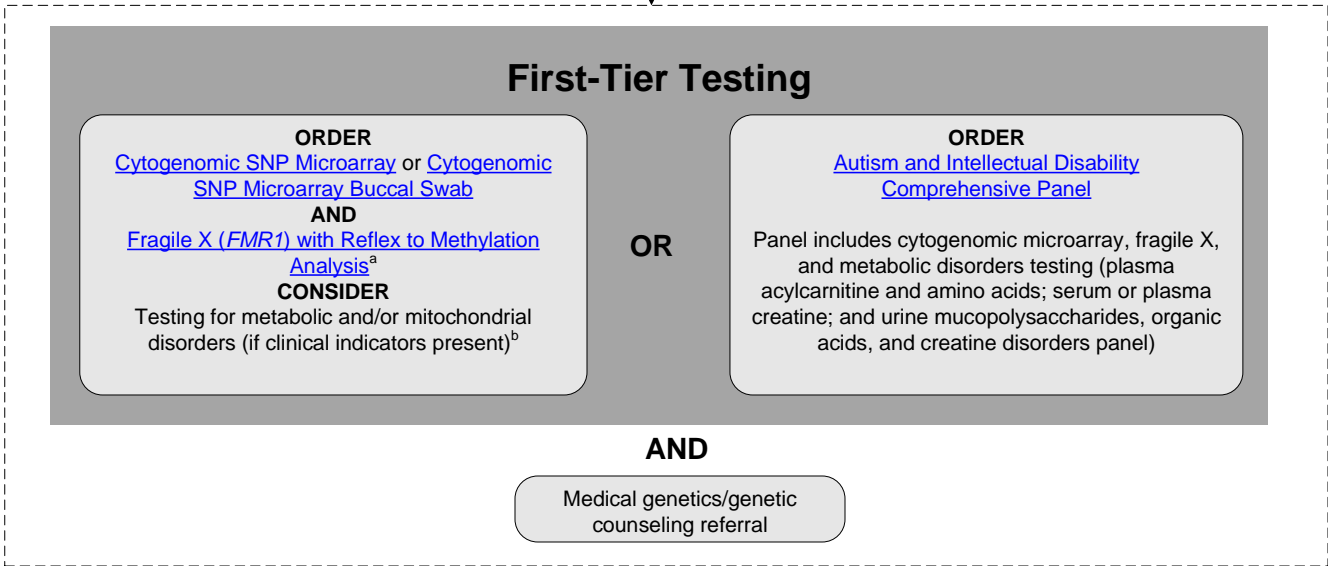
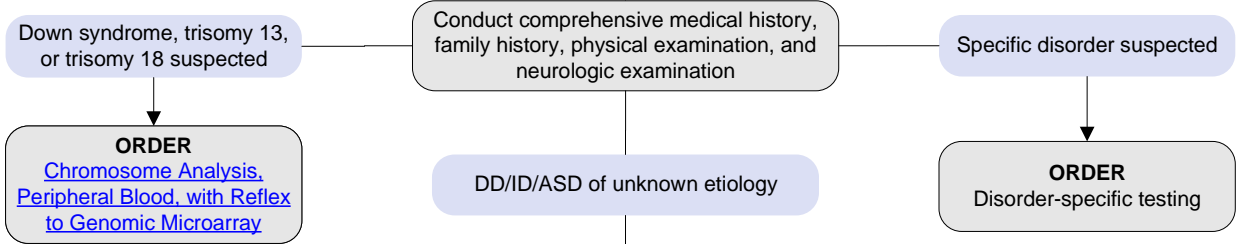


Testing for Genetic Syndromes Related to Developmental Delay (DD), Intellectual Disability (ID), and Autism Spectrum Disorder (ASD)

[Click here for topics associated with this algorithm](#)

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



Second-Tier Testing

- Consider additional specific molecular studies to assess for
 - Rett syndrome if patient is female
 - *PTEN*-related disorders 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

^aThe American College of Medical Genetics and Genomics suggests fragile X syndrome screening for males and females with compatible clinical features and family history. The American Academy of Pediatrics and American Academy of Neurology recommend screening in all cases.
^bConsider metabolic testing (eg, acylcarnitine, amino acids, creatine, mucopolysaccharides, organic acids, creatine disorders panel, very long-chain and branched-chain fatty acids profile, uric acid, pyridoxine-dependent epilepsy panel [includes pipercolic acid and total AASA-P6C]) and review of newborn screening results even if low incidence/suspicion because intervention may be possible. Clinical indicators may include poor feeding, vomiting, dehydration, lethargy, hypotonia, and seizures in the newborn period. Clinical indicators outside of the newborn period exhibit marked variability and may be episodic and/or progressive.

References
 Michelson DJ, Shevell MI, Sherr EH, Moeschler JB, Gropman AL, Ashwal S. Evidence report: Genetic and metabolic testing on children with global developmental delay: report of the Quality Standards Subcommittee of the American Academy of Neurology and the Practice Committee of the Child Neurology Society. *Neurology*. 2011; 77(17): 1629-35.
 Moeschler J, Shevell M, Committee on Genetics. Comprehensive Evaluation of the Child with Intellectual Disability or Global Developmental Delays. *Pediatrics*. 2014; 134(3):903-18. PubMed
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