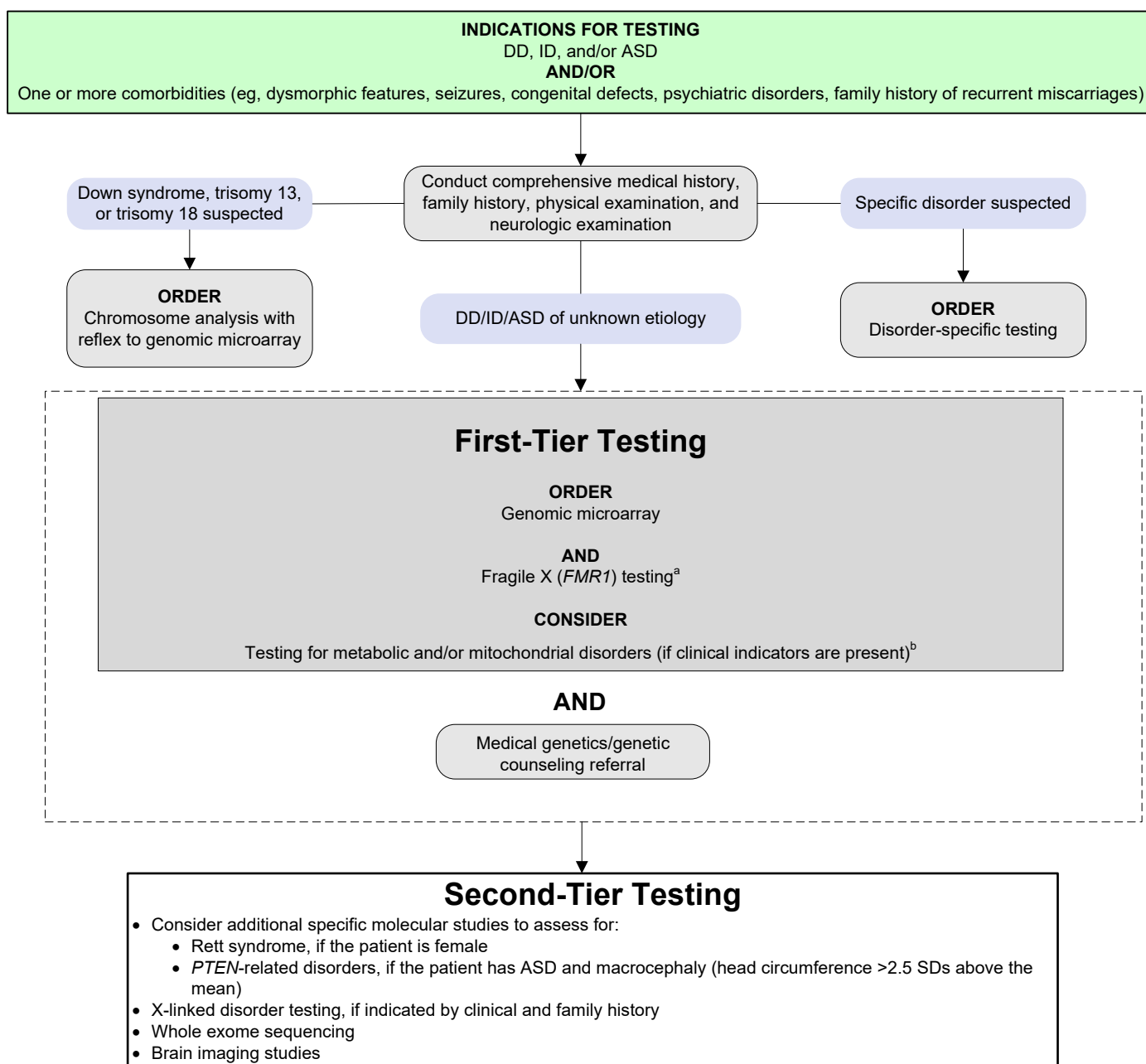


# Testing for Genetic Syndromes Related to Developmental Delay, Intellectual Disability, and Autism Spectrum Disorder

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## Abbreviations

ASD	Autism spectrum disorder
DD	Developmental delay
ID	Intellectual disability
SD	Standard deviation

<sup>a</sup>The American College of Medical Genetics and Genomics suggests fragile X syndrome screening for male and female patients with compatible clinical features and family history. The American Academy of Pediatrics and the American Academy of Neurology recommend screening in all cases.

<sup>b</sup>Consider 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 there is 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

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