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

ARUP Consult

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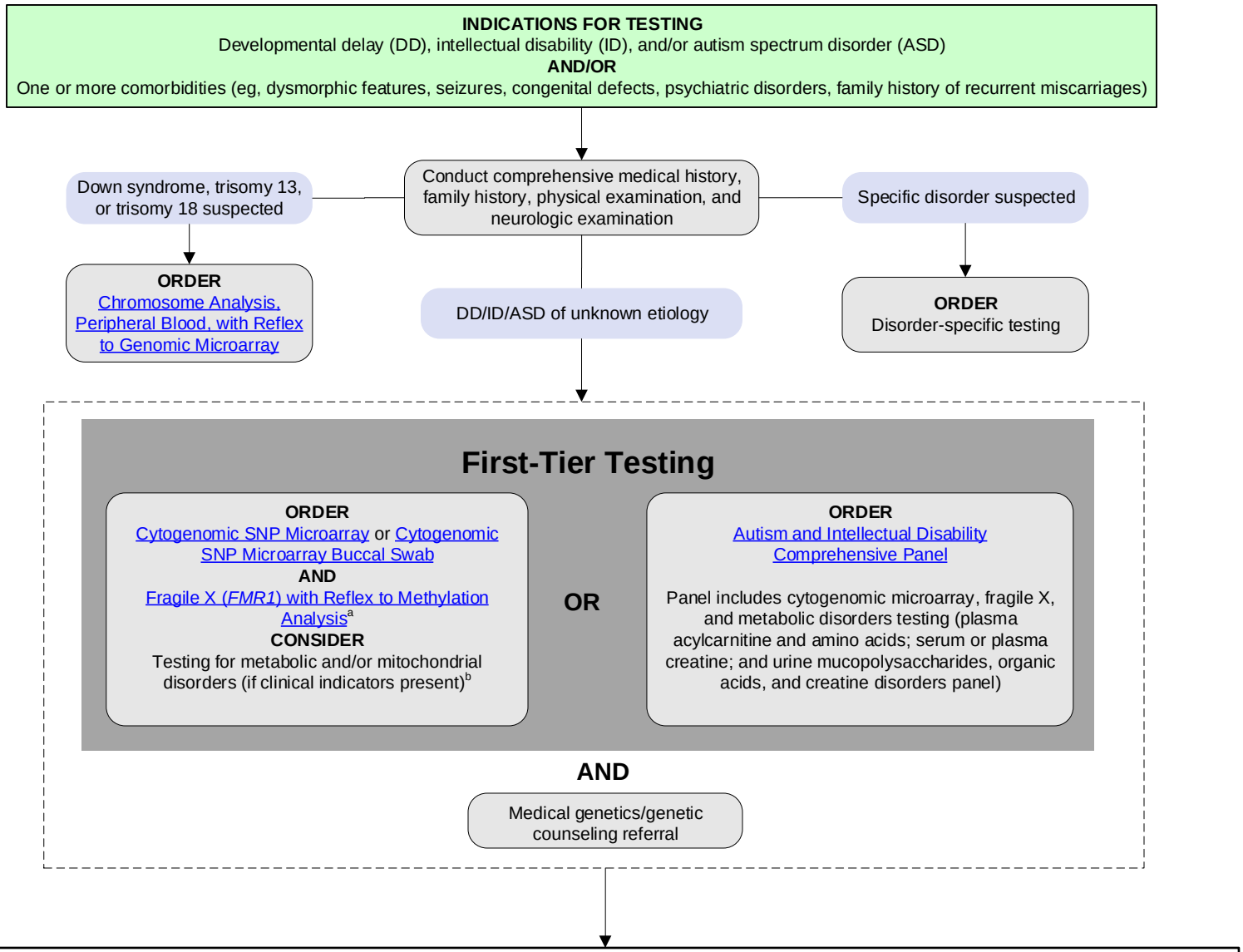

Second-Tier Testing

Consider additional specific molecular studies:

- Rett Syndrome (*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

[a]The 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.
[b]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 pipecolic 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

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