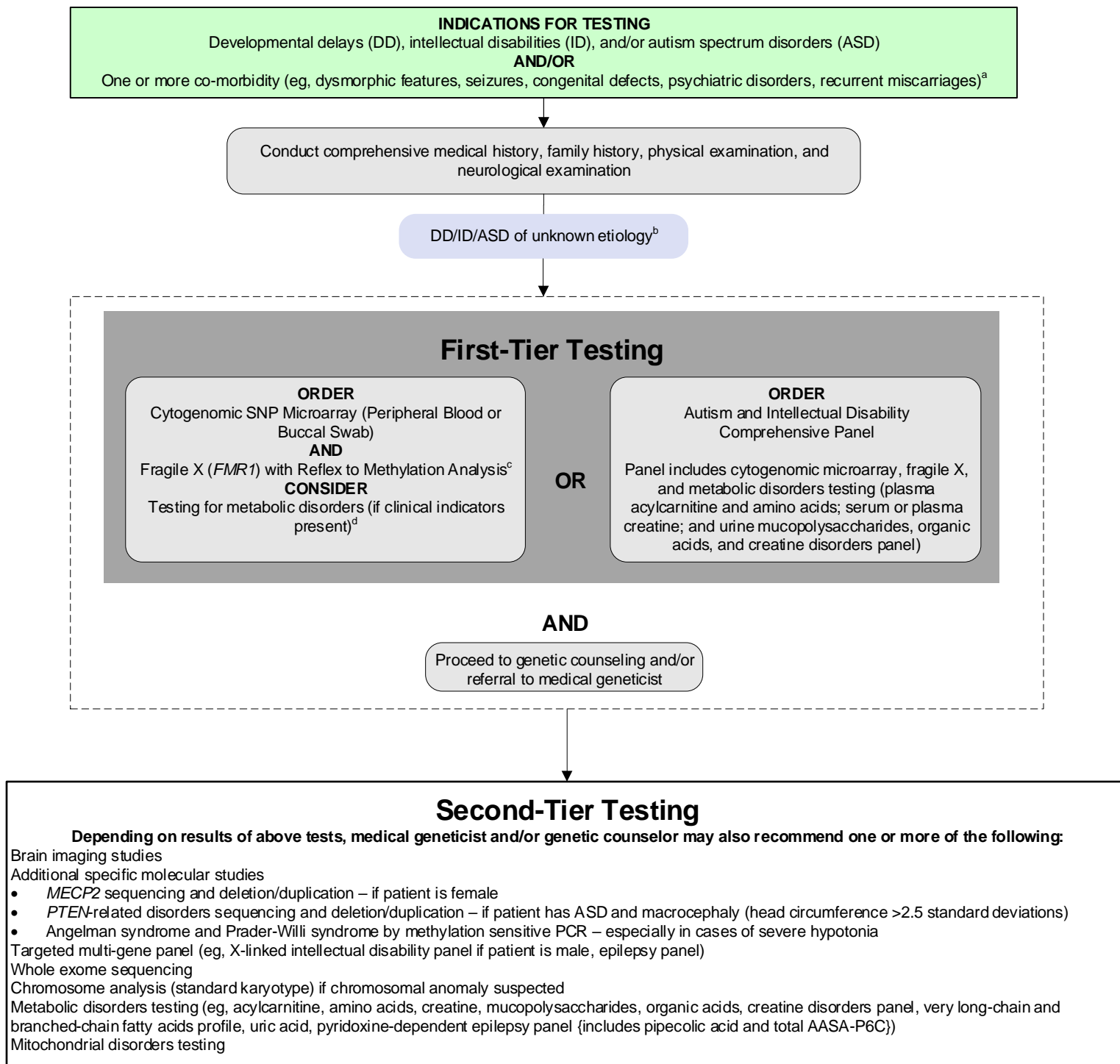


Testing for Genetic Syndromes Related to Developmental Delay (DD), Intellectual Disability (ID), and Autism Spectrum Disorder (ASD)

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^aFamily history of DD, ID, or ASD with co-morbidities generally increases diagnostic yield of testing.

^bIf specific disorder(s) suspected, order disorder-specific testing and/or consult with medical geneticist.

^cACMG suggests fragile X syndrome screening for male patients.

^dConsider metabolic testing 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

- Battaglia A, Doccini V. Confirmation of chromosomal microarray as a first-tier clinical diagnostic test for individuals with developmental delay, intellectual disability, autism spectrum disorders and dysmorphic features. *Eur J Paediatr Neurol*. 2013; 17(6):589-599. PubMed
- 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
- Schaefer B, Mendelsohn NJ, Professional Practice and Guidelines Committee. Clinical genetics evaluation in identifying the etiology of autism spectrum disorders: 2013 guideline revisions. *Genet Med*. 2013; 15(5): 399-407. PubMed