

Testing for Genetic Syndromes Related to Autism Spectrum Disorder (ASD)

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INDICATIONS FOR TESTING
 ASD
WITH OR WITHOUT
 One or more comorbidities (eg, developmental delay/intellectual disability, dysmorphic features, seizures, congenital defects, psychiatric disorders, family history of recurrent miscarriages)

PERFORM
 Detailed behavioral, developmental, family, and medical histories; thorough neurologic and physical exams
AND OFFER
 Referral for pediatric medical genetics

Specific disorder suspected

No specific disorder suspected

ORDER
 Disorder-specific testing, such as:

- Testing for *PTEN*-related disorders
- X-linked disorder testing
- Testing for metabolic or mitochondrial disorders
- Other nonlaboratory assessments

Etiology consistent with patient phenotype identified

Etiology not identified

No further testing

ORDER
 CMA with fragile X testing
OR
 A panel that includes CMA, fragile X, and metabolic testing
AND CONSIDER
 Testing for Rett syndrome (*MECP2*) in female patients

First-Tier Testing

Etiology consistent with patient phenotype identified

Etiology not identified

No further testing

PROVIDE
 Medical genetics referral for additional testing, which may include whole exome sequencing

Second-Tier Testing

Abbreviations
 ASD Autism spectrum disorder
 CMA Chromosomal microarray

References
 • Hyman SL, Levy SE, Myers SM, et al. [Identification, evaluation, and management of children with autism spectrum disorder](#). *Pediatrics*. 2020;145(1):e20193447.
 • Schaefer GB, 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.