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

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

No further testing

Etiology not identified

Etiology not identified

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

No further testing

Etiology not identified

Second-Tier Testing

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

Abbreviations
ASD Autism spectrum disorder
CMA Chromosomal microarray

References

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