REFERENCE GUIDE

Ambry Genetics®

Genetic Testing for Neurological Disorders

A Practical Guide for Pediatric Neurologists and Developmental Pediatricians

Genetic Testing for Children With Neurological Disorders is Recommended By:

- **American Academy of Pediatrics**[1]
- **American College of Medical Genetics and Genomics**[2]
- **National Society of Genetic Counselors**[3]
- **American Epilepsy Society**[3]

Establishing an Etiology Provides Guidance for the Family

Identifying patients with a genetic cause for their neurological disorder can clarify a diagnosis and inform recommendations for personalized medical management. Benefits of genetic testing may include:[1–3]

Tailoring Medical Care

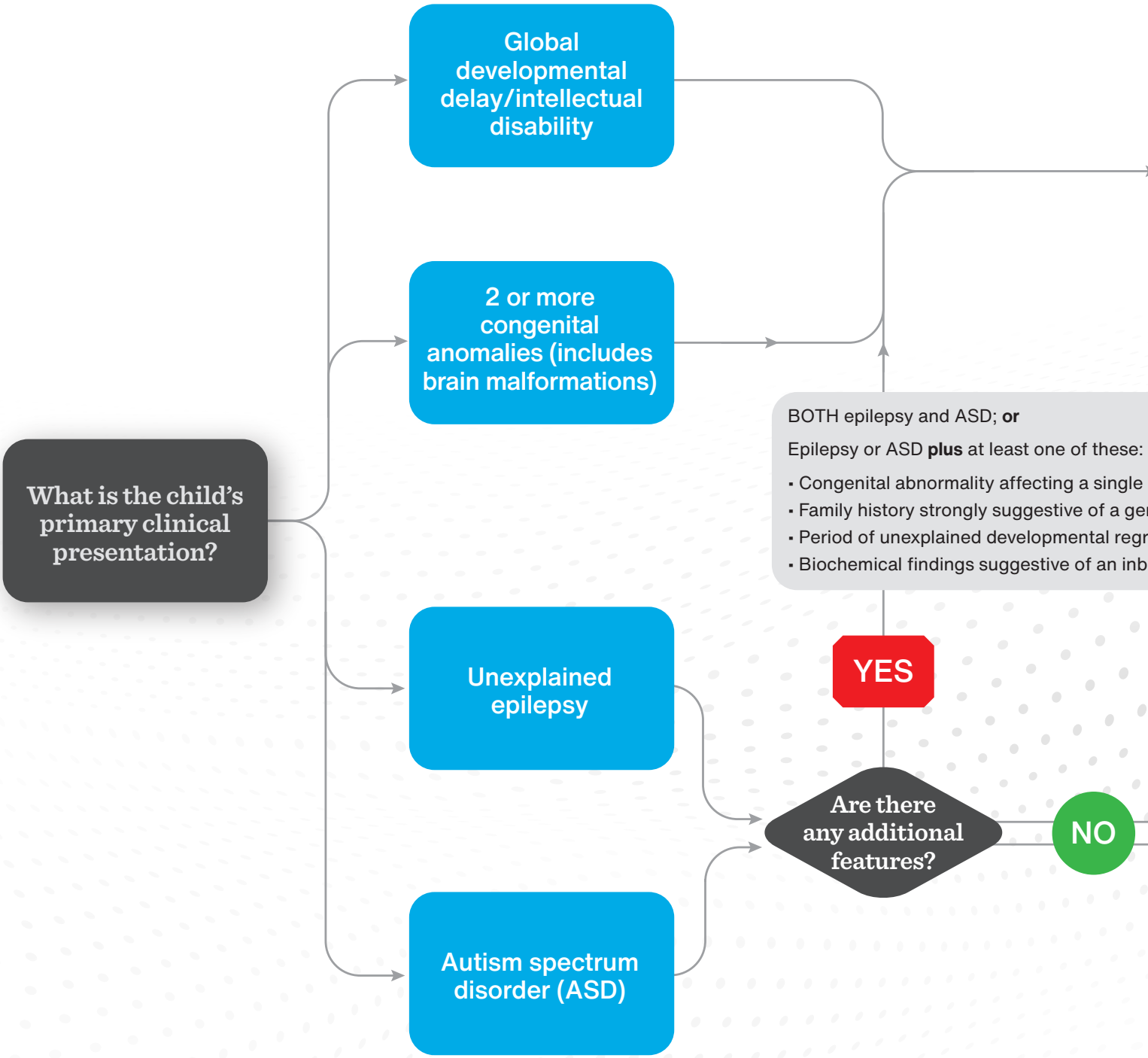
Guiding Therapy Selection

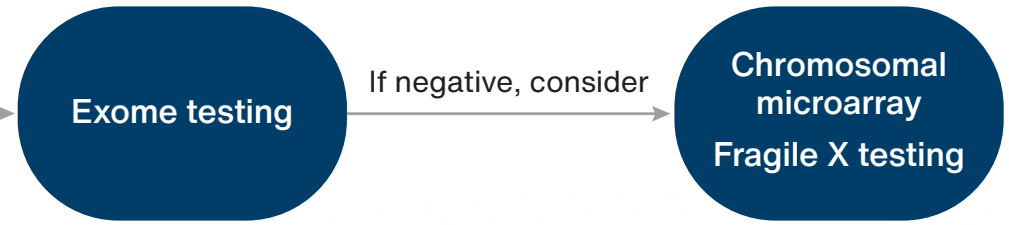
Creating Opportunities for Community

Informing Family Decisions

Clinical Testing Workflow Based on Professional Guideline Recommendations

This suggested clinical workflow factors in recommendations from professional society guidelines and typical medical coverage criteria. Other testing not captured here may be appropriate for individual patients.





Find More Answers with Exome Testing at Ambry

Our latest offering, ExomeReveal,™ offers enhanced analysis with RNA for higher diagnostic yield and lower VUS rates.

In addition, our Patient for Life™ program provides proactive, continuous reanalysis. This unique Ambry service yields answers for 1 in 20 patients who initially have negative or uninformative results.[4]

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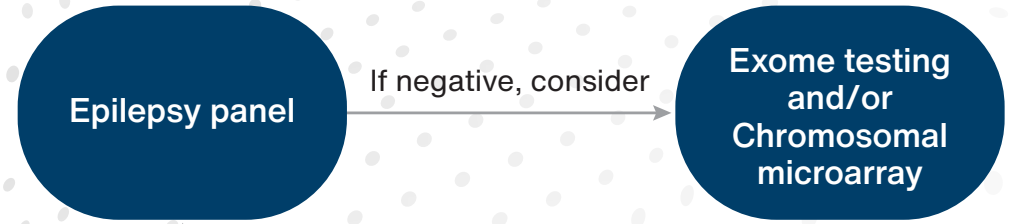

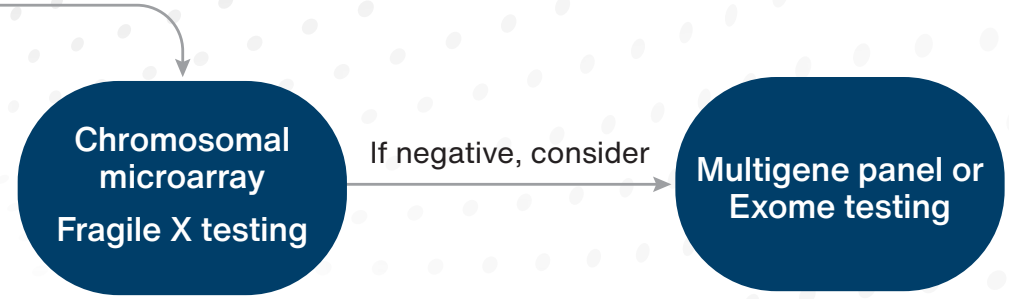

Expert Consultation Available

Commercial payor coverage can be more variable for second-tier testing, and the best clinical choice can also depend on the child's presentation.

Our Genomic Science Liaisons are happy to support your practice and are available for consultation.

We Offer a Comprehensive Test Portfolio for Your Practice

Product	Description	TAT	Specimen Types
ExomeNext®*	Sequences the coding regions of all 20,000 genes for variants that explain the phenotype	6–8 weeks	Blood (EDTA tube) Saliva Buccal
ExomeReveal™*	Includes RNA analysis for qualified variants	Additional 3–4 weeks	Blood (EDTA and PAXgene tubes)
Chromosomal Microarray (SNP Array)	Detects genome-wide copy number variants (CNVs) – extra or missing sections of genetic material	2–3 weeks	Blood (EDTA tube) Saliva Buccal
Fragile X Testing	Tests specifically for fragile X syndrome, the most common inherited cause of intellectual disability	1–2 weeks	
EpilepsyNext®*	124 gene panel	2–4 weeks	
AutismNext®*	72 gene panel	2–4 weeks	
NeurodevelopmentNext®*	202 gene panel	2–4 weeks	

*Recommend these tests performed as a family Trio when possible.
Biological parents' samples can be submitted with the patient's sample.

References

1. Hyman SL, Levy SE, Myers SM; Council on Children with Disabilities, Section on Developmental and Behavioral Pediatrics. Identification, Evaluation, and Management of Children With Autism Spectrum Disorder. *Pediatrics.* 2020;145(1):e20193447. doi:10.1542/peds.2019-3447
2. Manickam K, McClain MR, Demmer LA, et al. Exome and genome sequencing for pediatric patients with congenital anomalies or intellectual disability: an evidence-based clinical guideline of the American College of Medical Genetics and Genomics (ACMG). *Genet Med.* 2021;23(11):2029-2037. doi:10.1038/s41436-021-01242-6
3. Smith L, Malinowski J, Ceulemans S, et al. Genetic testing and counseling for the unexplained epilepsies: An evidence-based practice guideline of the National Society of Genetic Counselors. *J Genet Couns.* 2023;32(2):266-280. doi:10.1002/jgc4.1646
4. Smith ED, Radtke K, Rossi M, et al. Classification of genes: Standardized clinical validity assessment of gene-disease associations aids diagnostic exome analysis and reclassifications. *Hum Mutat.* 2017;38(5):600-608. doi:10.1002/humu.23183

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