

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 Pediatrics¹

American College
of Medical Genetics
and Genomics²

National Society of
Genetic Counselors³

American Epilepsy
Society³

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



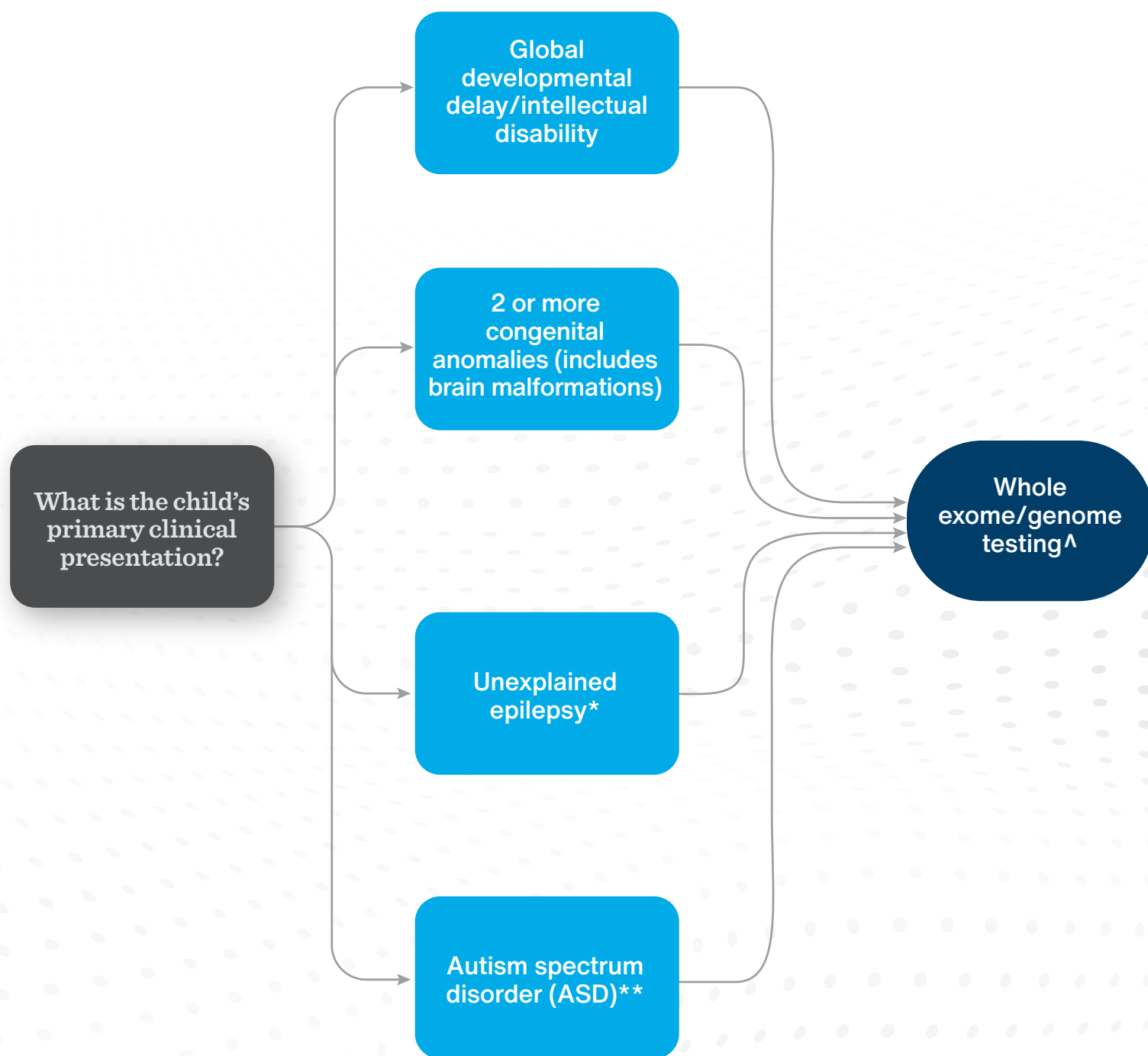
Creating Opportunities
for Community



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. Individual health plan coverage may differ. Other testing not captured here may be appropriate for individual patients.*



* Some payors require the age of onset to be under three years and/or an additional clinical feature.

** Some payors require an additional clinical feature.

^ The clinical guidelines and recommendations provided are for informational and navigational purposes only and do not guarantee insurance coverage. Individual insurance policies, benefit structures, and medical necessity criteria vary significantly by payer and plan.

If exome sequencing
is negative, consider

Chromosomal
microarray
Fragile X testing

Find More Answers with Testing at Ambry

ExomeReveal® and GenomeReveal™ offer 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

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	3–6 weeks	Blood (EDTA tube) Saliva Buccal
ExomeReveal^{®*}	Includes RNA analysis for qualified variants	Additional 3–4 weeks	Blood (EDTA and PAXgene tubes)
GenomeNext^{™*}	Comprehensive analysis of coding and noncoding genes. Built on a pangenome-enabled reference to reduce bias and improve accuracy across populations. Includes mitochondrial analysis.	5–7 weeks	Blood (EDTA tube) Saliva Buccal
GenomeReveal^{™*}	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 genetic 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

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