

Child Neurology Genetic Testing

Reference Guide



**Genetic Testing
For Children With
Intellectual Disability
and/or Autism
Spectrum Disorders
is Recommended By:**

**American Academy
of Neurology¹**

**American Academy
of Pediatrics²**

**American College of
Medical Genetics and Genomics³**

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 for neurological disorders may include:



**Availability of Tailored Treatment
Options For Some Conditions**

For example, TOR inhibitors for *TSC1/TSC2*⁴



**Inform Personalized Medical
Management and Additional
Specialty Referrals**

For example, ECG monitoring for *MECP2*⁵



**Improved Understanding of
Diagnosis and Prognosis**



**Identification of At-Risk
Family Members**

Neurodevelopmental Disorders

2% of school-aged children in the U.S. are diagnosed with intellectual disability (ID)⁶ and 2-3% of children in the U.S. are found to have an autism spectrum disorder (ASD).⁷

SNP Array

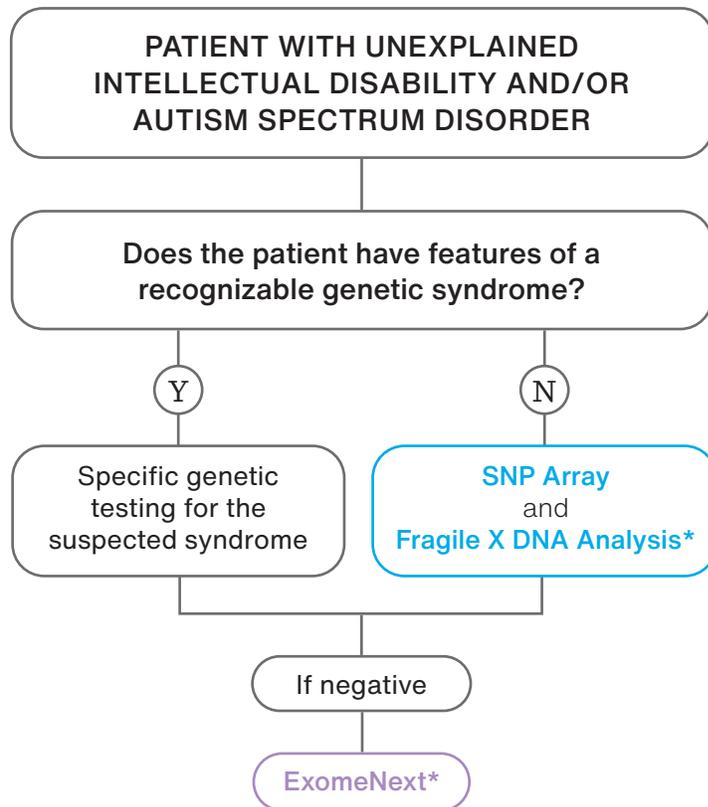
Ambry microarray with a 2-3 week turnaround time (TAT)

Fragile X DNA Analysis

FMR1 repeat expansion analysis with a 1-2 week TAT

ExomeNext[®]

Analyzes all 20,000 genes and has been successful in ending the diagnostic odyssey for 30% of undiagnosed patients⁹



The American College of Medical Genetics and Genomics (ACMG) strongly recommends that exome sequencing be considered as a first- or second- tier test for patients with congenital anomalies, developmental delay, and/or or intellectual disability.¹⁰

? Do you prefer to order panels before ExomeNext?

AutismNext[®]

72-gene panel including genes associated with non-syndromic ASDs and/or ID with a 3-4 week TAT.

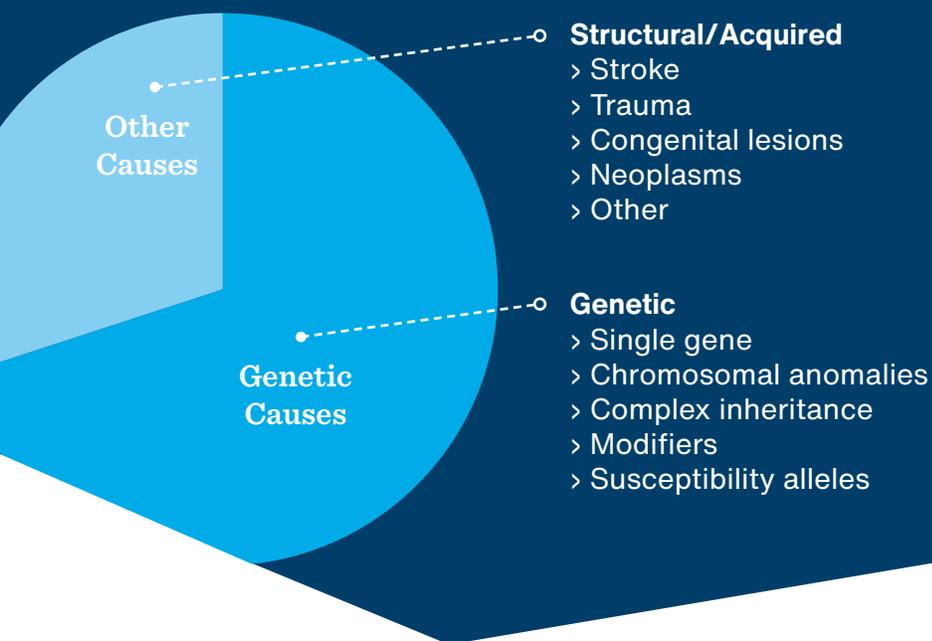
NeurodevelopmentNext[™]

202-gene panel including genes accounting for >60% of patients identified to have a genetic cause for a neurodevelopmental disorder including developmental delay, ID, and/or ASDs with a 3-4 week TAT. Diagnostic rate is 17%.⁸

* Exome sequencing may be considered as a first tier test for patients with unexplained neurodevelopmental disorders.^{9,10}

Epilepsy

Causes of Epilepsy¹²



DID YOU KNOW?

A strong proportion of epilepsy may be due to an underlying genetic cause.^{12,13}

Epilepsy is a common condition that affects about **1 in every 26 people**, with approximately 150,000 new cases diagnosed in the U.S. per year.¹⁴

Ambry Offers a Range of Epilepsy Genetic Testing Options to Help Identify an Underlying Cause and Inform Management

Genes associated with epilepsy that may have implications for seizure management.

Gene List: *ALDH7A1, AMT, DDC, FOLR1, GLDC, KCNQ2, KCNQ3, KCNT1, MECP2, PCDH19, PNPO, POLG, PRRT2, SCN1A, SCN2A, SCN8A, SLC19A3, SLC2A1, SLC6A8, STXBP1, TSC1, TSC2*

Includes genes accounting for approximately 60% of patients identified to have genetic epilepsies such as Dravet syndrome, epileptic encephalopathy, non-lesional focal epilepsy, and febrile-related seizures.⁸

Designed to identify causes of seizures primarily with neonatal to childhood onset including epilepsy-only disorders, syndromic conditions in which seizures have been reported, as well as treatable metabolic conditions that can include seizures when undiagnosed and/or untreated[^].

EpiRapid®

22 genes
10-14 days

EpilepsyNext®

124 genes
2-4 weeks

EpilepsyNext-Expanded™[^]

>960 genes
2-4 weeks



FOR COMPLETE GENE LISTS, VISIT
ambrygen.com/epilepsy

[^] Panel content is annually updated due to proactive review of current literature using internal, peer-reviewed clinical validity scheme.¹¹

Neurocutaneous/ Neuro-Oncology Disorders

These disorders cause tumors that can be benign or malignant and often require medical or surgical intervention. Accurate diagnosis involves a combination of clinical assessment and diagnostic testing. Ambry offers a variety of tests with a 2-3 week turnaround time to aid in the diagnosis of these conditions.

CONDITION NAME	GENE(S)
Ataxia-telangiectasia	ATM
Legius syndrome	SPRED1
Neurofibromatosis 1	NF1
Neurofibromatosis 2	NF2
Brain/Other nervous system tumors (BrainTumorNext)	29 genes
Schwannomatosis	SMARCB1, LZTR1, NF2
Tuberous sclerosis complex	TSC1, TSC2

Ambry Genetics Offers Additional Services at No Cost to Help Find Answers for Your Patients



PATIENT FOR LIFE

Ambry continually reviews data for patients who have been tested with our exome and neurology panels for potential pathogenic or likely pathogenic variants in newly added genes and proactively issues reclassification reports, as applicable.



PARENTAL SAMPLE ANALYSIS

Reduce follow-up time and uncertain results by submitting parental samples (biological mother and father) along with the patient sample. Co-segregation studies will be performed as needed prior to results being released.



RNA STUDIES IN AMBRY'S TRANSLATIONAL GENOMICS (ATG) LAB

In some cases, the ATG lab can provide additional genomic analysis to potentially bring clarity to certain variants of unknown significance (VUS). This can drive down the VUS rate to give you more actionable data that may inform medical management.



NO-COST TESTING FOR FAMILY MEMBERS

We offer specific site analysis at no additional cost for family members following single gene or multigene panel testing of the first family member (proband) within 90 days of the original Ambry report date.

References

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