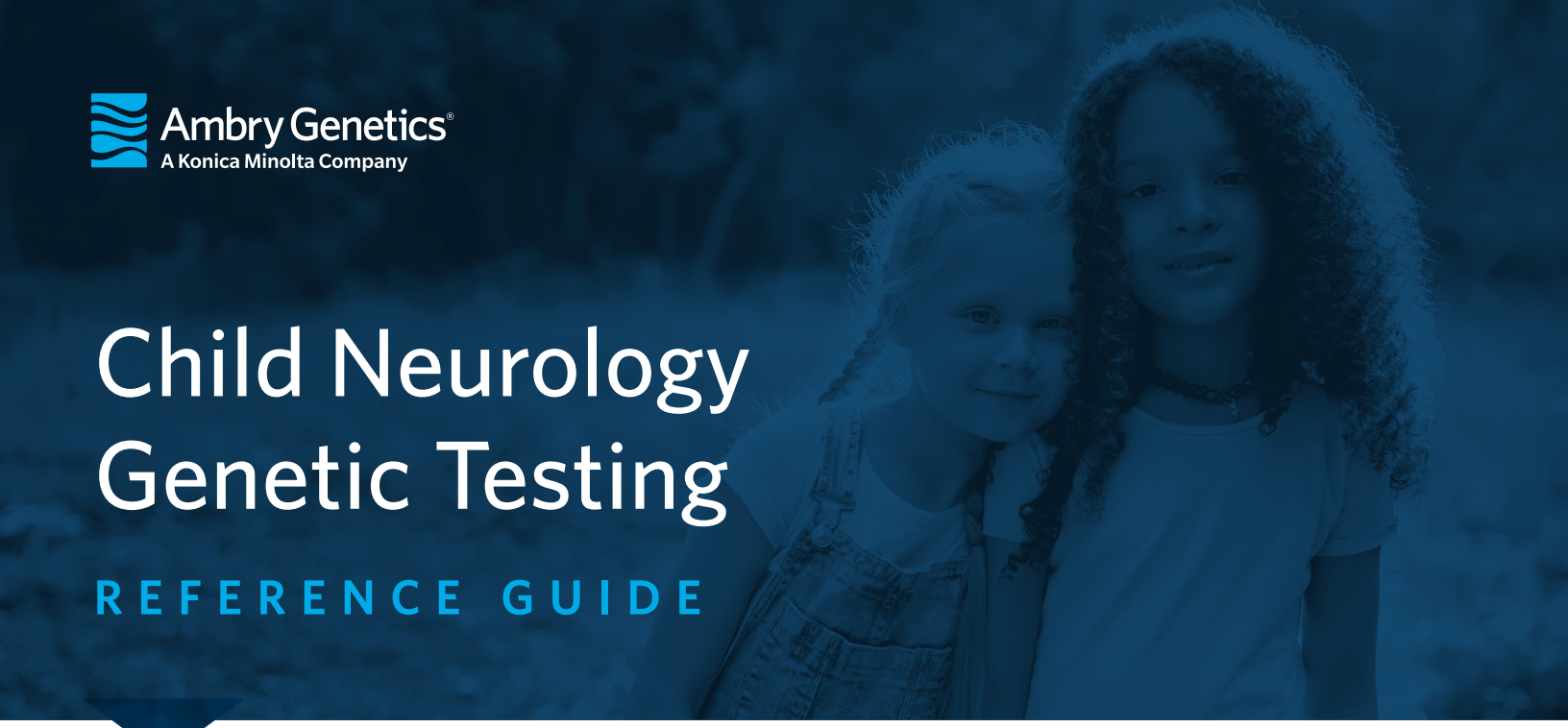
Ambry Genetics®
A Konica Minolta Company



Child Neurology Genetic Testing

REFERENCE GUIDE

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

- **American Academy of Neurology**[1]
- **American Academy of Pediatrics**[2]
- **American College of Medical Genetics and Genomics**[3]

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

AutismNext®

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

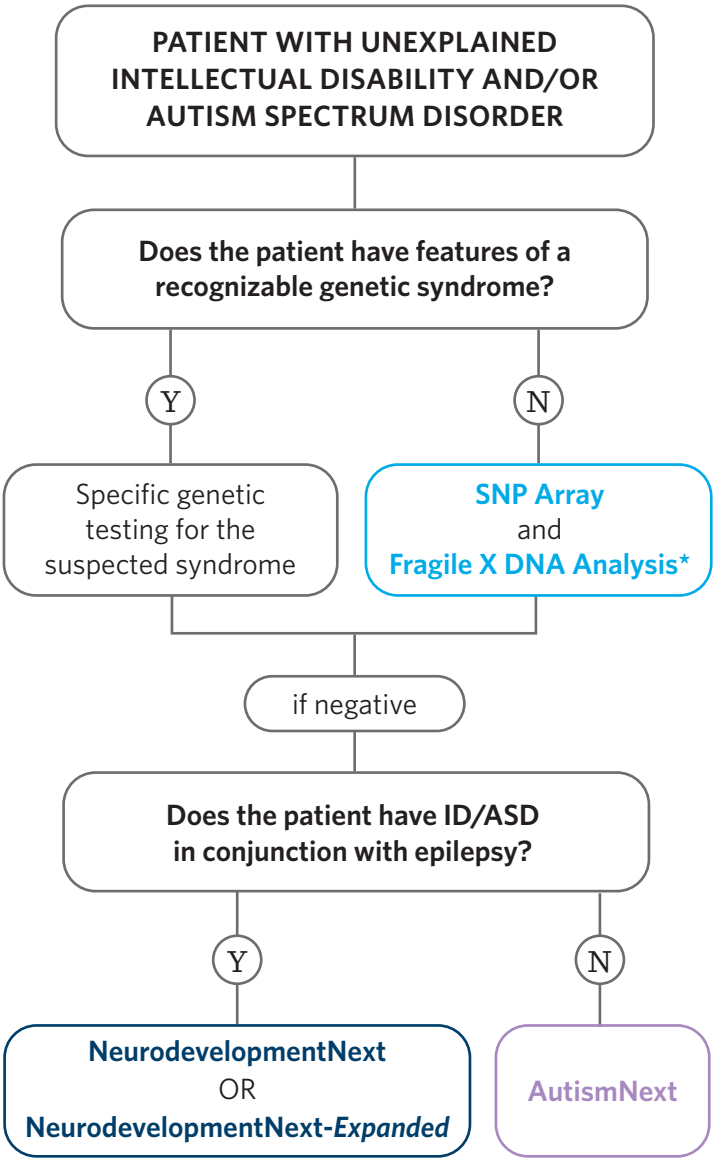
NeurodevelopmentNext™

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]

? DO YOU PREFER A BROADER PANEL WITH A HIGHER DIAGNOSTIC YIELD?

NeurodevelopmentNext-*Expanded*™

>1,400-gene panel including genes that are associated with neonatal to childhood onset developmental delay, seizures, ID, developmental regression, and/or ASDs with a 4-6 week TAT. Diagnostic rate is 24%[8]**



? DOES THE PATIENT HAVE ADDITIONAL CLINICAL FEATURES OR DO YOU WANT TO CAST A WIDER NET?

ExomeNext®

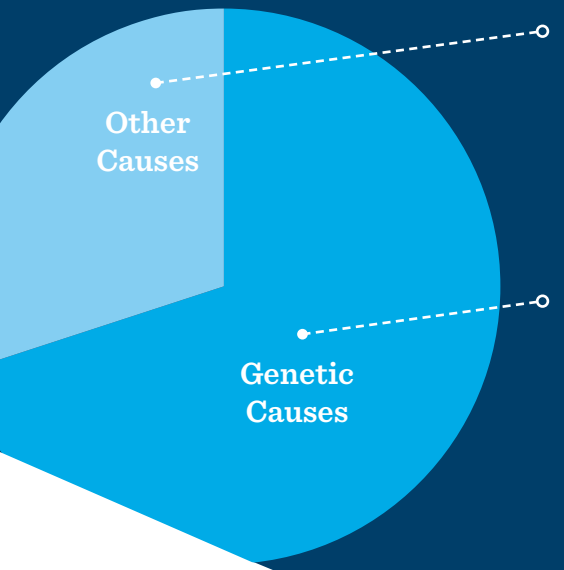
Analyzes all 20,000 genes and has been successful in ending the diagnostic odyssey for 30% of undiagnosed patients.

* Exome sequencing may be considered as a first tier test for patients with unexplained neurodevelopmental disorders.[9]

** Panel content is regularly updated due to proactive review of current literature using an internal, peer-reviewed clinical validity scheme[10]

Epilepsy

Causes of Epilepsy[11]



Structural/Acquired
› Stroke
› Trauma
› Congenital lesions
› Neoplasms
› Other

Genetic
› Single gene
› Chromosomal anomalies
› Complex inheritance
› Modifiers
› Susceptibility alleles

DID YOU KNOW?

A strong proportion of epilepsy may be due to an underlying genetic cause.[11,12]

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.[13]

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

EpiRapid®
22 genes
10-14 days

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*

EpilepsyNext®
124 genes
2-4 weeks

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]

EpilepsyNext-*Expanded*™
>890 genes
2-4 weeks

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^.

FOR COMPLETE GENE LISTS, VISIT
ambrygen.com/epilepsy

^ Panel content is regularly updated due to proactive review of current literature using internal, peer-reviewed clinical validity scheme.[10]

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*
Primary brain tumors (BrainTumorNext)	*29 genes*
Schwannomatosis	*SMARCB1*
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 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 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

The ATG lab provides 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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