Ambry Genetics®
A Konica Minolta Company

Child Neurology Genetic Testing

REFERENCE GUIDE

June 2018

Genetic testing can help you predict medical risks that may be associated with childhood neurological disorders and may offer a chance to implement early intervention. The sooner we can uncover the source of the condition, the sooner we can work towards a treatment.

Our ongoing participation in numerous research studies and collaborations helps us better assess clinical validity of gene-disease relationships. This is to ensure that the results you receive are as accurate and informative as possible.

1 in 6 children have a neurological disorder

Practice Guideline

American Academy of Neurology (AAN)

American Academy of Pediatrics (AAP)

American College of Medical Genetics and Genomics (ACMG)

The AAN, AAP, and ACMG recommend chromosomal microarray (CMA) and fragile X DNA analysis as first-tier genetic tests in the evaluation of individuals with ID and/or ASD.

Michelson (2011) Neurology

Moeschler (2014) Pediatrics

Miller DT (2010) Am J Hum Genet

Why Is Genetic Testing Important?

KEY BENEFITS

Identifying patients with a genetic cause for their neurological disorder can allow informed recommendations and personalized medical management treatments for improved patient outcomes.

- Availability of tailored treatment options (*e.g.* mTOR inhibitors for *TSC1/TSC2*, avoid sodium channel blockers for *SCN1A*)
- Avoid alternative, potentially invasive testing
- Identification of at-risk family members
- Improved understanding of prognosis and additional screening recommendations (*e.g.* ECG monitoring for *MECP2*)

Choose the Right Test

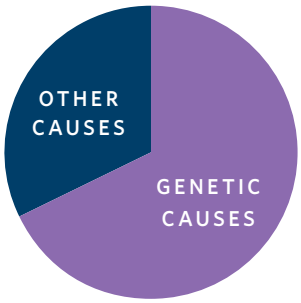
Our flexible range of neurological genetic tests can help end the diagnostic odyssey.

- Targeted panels minimize cost, turnaround time, and reduce the potential for variants of uncertain clinical significance
- Larger, broad panels may increase detection rates
- Reflex options allow you to start small and test in a methodical fashion
- Chromosomal microarray and whole exome sequencing can supplement panel testing
- Send in parental samples with the patient sample for up-front family studies that decrease the VUS rate

Epilepsy

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.[1]

Causes of Epilepsy[2]



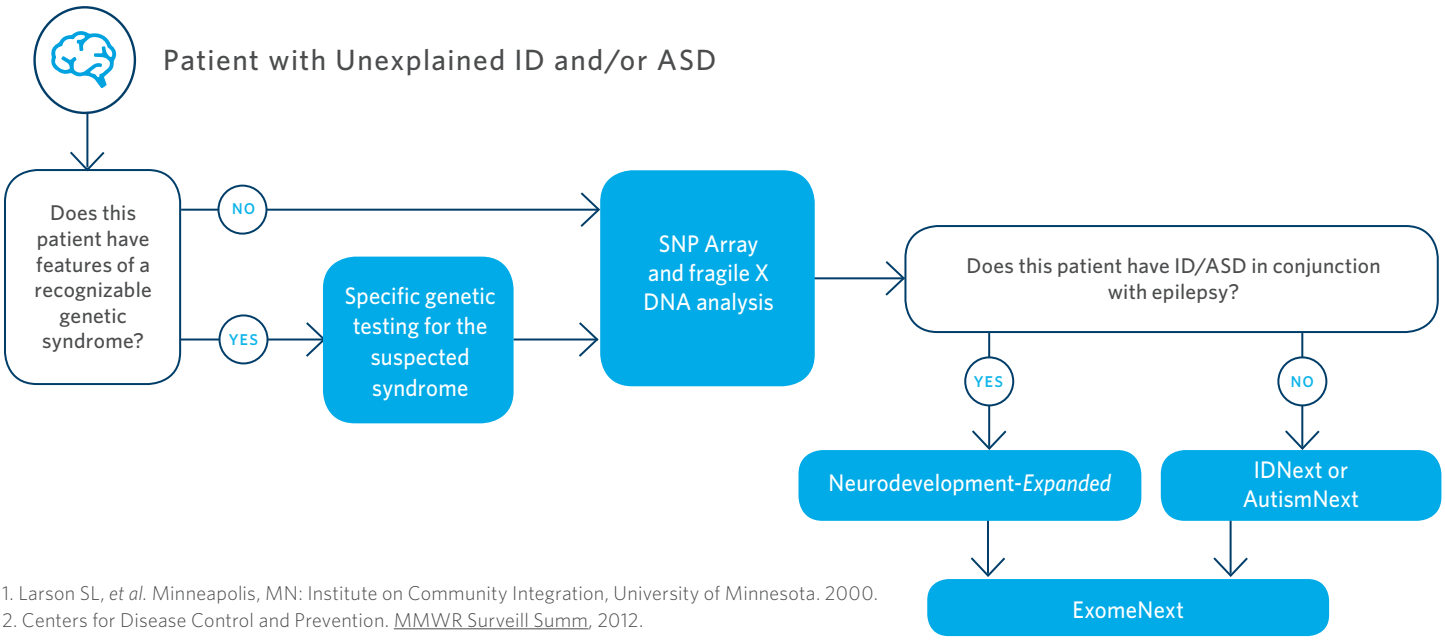
TEST	DESCRIPTION	# OF GENES	TAT (WEEKS)
TARGETED PANEL TESTING			
EpiRapid	Genes that could have immediate implications for treatment	16	10-14 days
EpiFirst	Phenotype-specific epilepsy panels	*Fever*: 13	2-3
		Focal: 11	2-3
		IS (infantile spasms): 17	2-3
BROAD PANEL TESTING			
EpilepsyNext	Broad epilepsy panel	EpilepsyNext: 100	2-3
Neurodevelopment-*Expanded*	Epilepsy with ID and/or ASD	196	4-6
CustomNext-*Neuro*	Comprehensive neurology testing	Up to 196	2-3*
GENOMIC TESTING			
SNP Array	Chromosomal microarray	Whole genome	2-3
ExomeNext	Whole exome sequencing	Whole exome	6-8

* Orders with >140 genes will have a TAT of 4-6 weeks.

1. Institute of Medicine of the National Academies. March 20, 2012.
2. Thomas RH and Berkovic SF, Nat Rev Neurol., 2014.

Neurodevelopmental Disorders

2-3% of individuals in the U.S. are diagnosed with intellectual disability (ID)[1] and 1-2% of children in the U.S. are found to have an autism spectrum disorder (ASD).[2]



1. Larson SL, *et al.* Minneapolis, MN: Institute on Community Integration, University of Minnesota. 2000.
2. Centers for Disease Control and Prevention. MMWR Surveill Summ, 2012.

TEST	DESCRIPTION	# OF GENES	TAT (WEEKS)
Fragile X DNA Analysis	*FMR1* repeat expansion analysis	1	1-2
SNP Array	Chromosomal microarray	Whole genome	2-3
AutismNext	Syndromic and non-syndromic autism spectrum disorders	48	2-3
IDNext	Intellectual disability	140	2-3
Neurodevelopment-*Expanded*	Genes that cause a combination of epilepsy, intellectual disability and autism spectrum disorders	196	4-6
ExomeNext	Diagnostic exome sequencing	Whole exome	6-8

Neurocutaneous Disorders

These disorders cause tumors that can be benign or malignant and often require medical or surgical intervention. Accurate diagnosis often involves a combination of clinical assessment and diagnostic testing.

CONDITION NAME	GENE(S)	TAT (WEEKS)
Ataxia-telangiectasia	*ATM*	2-3
Legius syndrome	*SPRED1*	2-3
Neurofibromatosis 1	*NF1*	2-3
Neurofibromatosis 2	*NF2*	2-3

CONDITION NAME	GENE(S)	TAT (WEEKS)
Primary brain tumors (BrainTumorNext)	27 genes	2-3
Schwannomatosis	*SMARCB1*	2-3
Tuberous sclerosis complex	*TSC1, TSC2*	2-3

Over 1 Million Tests Completed

MOVING SCIENCE FORWARD

Purposeful Confirmatory Testing

Many labs validate their tests based on certain limited studies. That's why we participated and led the largest study of its kind (20,000 cases) guiding us to utilize confirmatory testing when we see specific well-defined thresholds. Our mission is to get it right the first time.

Understanding Disease Better Through Free Data Sharing

Identifying an individual's genetic information is nothing new—it's what we do with it that is unique. When labs share genomic information, we can together accelerate the understanding of human disease. Through AmbryShare, we leverage de-identified information to collaborate with others and help people everywhere find answers.

SuperLab

Our 65,000 square foot highly-automated CLIA/CAP certified lab produces some of the fastest turnaround times in the industry, without compromising testing accuracy or specificity.

Ambry's Translational Genomics (ATG) Lab

As an advanced diagnostic lab, it's our responsibility to ensure the results you get from us are accurate and that classification is as complete and robust as possible. Our ATG lab is a unique laboratory that provides an additional service at no additional cost for you and your patients to generate more precise data potentially bringing clarity to some variants of unknown significance (VUS). This helps to actively drive down the rate of VUS results and can give you an increased understanding of your patient's results, so you can better provide medical management recommendations and improve health outcomes.

About Ambry

Just as no two fingerprints are alike, the way disease presents itself in every individual is different. Since 1999, our mission has always been about understanding disease better, so treatments and cures can be found faster. Every sample that arrives in our lab is viewed as a person with a life and a story that is unique to only them. By providing advanced confirmation genetic testing for inherited and non-inherited diseases, we can help you make more informed and responsible treatment decisions with your patients.

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