Neurological Disorders

GENETIC TESTING

Finding answers quickly to help guide patient care
Know the Basics

Over 6 million people in the U.S. are affected by a neurological disorder.

1-2% of children in the U.S. have an autism spectrum disorder.

**WHAT IS A NEUROLOGICAL DISORDER?**

A neurological disorder is any medical problem that affects the nervous system. Your body’s nervous system includes your brain, spinal cord, and other nerves. Problems with the structure or function of any part of the nervous system can lead to a neurological disorder.

**WHAT ARE SYMPTOMS OF A NEUROLOGICAL DISORDER?**

Symptoms of a neurological disorder can be mild, such as muscle weakness, poor coordination, or changes in thinking patterns. Symptoms can also be more noticeable, such as seizures, pain, and intellectual disability. People with a neurological disorder can have just one or many of these symptoms.
There are many causes for neurological disorders, including infections, injuries, and environmental factors such as poor nutrition or exposure to heavy metals like lead. Gene changes can also cause neurological disorders. A change in a gene can change the instructions to our brain, spinal cord and other nerves, which could result in a neurological disorder.

Most neurological disorders that are caused by gene changes will begin affecting a person early in life, usually in childhood and sometimes even as early as birth. However, some neurological disorders caused by gene changes don’t affect a person until later in life. Any signs or symptoms may not be obvious until well into adulthood.

Genetic testing can be helpful in identifying the cause of the neurological disorder in many different situations.
YOUR GENES CARRY A STORY THAT IS UNIQUE TO YOU AND MAKES YOU WHO YOU ARE. GENETIC TESTING CAN HELP BETTER UNDERSTAND AND MANAGE THE NEUROLOGICAL DISORDER THAT MAY BE OCCURRING IN YOUR FAMILY.

Genetic testing for neurological disorders can include a variety of genes that are linked to the symptoms that your family member has. Based on the results, your healthcare provider may discuss more specific prognosis and treatment options for you and your family.

How is Genetic Testing Performed?

1. Samples from you (and parents*) sent to the lab
2. Lab performs testing
3. Results available in 1-10 weeks, depending on the test ordered

* Parent samples can be accepted with the patient sample for most tests to help clarify results
The Best Test for You

Your healthcare provider has ordered the following test(s) for you:

**EPILEPSY**
- □ EpiRapid
- □ EpiFirst-Neonate
- □ EpiFirst-Fever
- □ EpiFirst-IS
- □ EpiFirst-Focal
- □ PMEFirst
- □ PMENext
- □ EpilepsyNext
- □ CustomNext-Epilepsy
- □ NCLNext
- □ Familial hemiplegic migraine

**NEURODEVELOPMENTAL DISORDERS**
- □ Fragile X DNA analysis
- □ IDNext
- □ AutismFirst
- □ AutismNext
- □ RettAngelmanNext
- □ Neurodevelopment-Expanded

**GENOMICS**
- □ SNPArray
- □ ExomeNext

**NEUROCUTANEOUS/NEURO-ONCOLOGY DISORDERS**
- □ Neurofibromatosis 1
- □ Legius syndrome
- □ Neurofibromatosis 2
- □ Schwannomatosis
- □ Tuberous sclerosis complex
- □ BrainTumorNext
- □ Ataxia-telangiectasia
- □ von Hippel-Lindau disease
- □ Li-Fraumeni syndrome
- □ Gorlin syndrome
- □ HHTFirst
- □ HHTNext

**VISIT OUR WEBSITE**
See updated information on which genes are included on the test your healthcare provider selected above: ambrygen.com/patient/neurotest
What are the Benefits of Genetic Testing?

FOR YOU:

Your healthcare provider can give you better information about what to expect based on your genetic test results.

- Examples of what to expect can be worsening of current symptoms or screening tests that can find new symptoms as early as possible.

Your healthcare provider may discuss possible treatment options, such as specific medications or other interventions, based on your genetic test results.

- Examples are using a medical dietary therapy or certain types of anti-seizure medications.

Your doctor can discuss the possibility of your genetic test results helping you to avoid other, potentially invasive, testing.
Many people with a neurological disorder are the first person in their family to have it. Often, genetic testing can find a gene mutation for someone with a neurological disorder that is not found in other family members and was not passed down from a parent. This is called a de novo, or new, gene mutation in a family.

In other families, gene changes that cause neurological disorders can be inherited from earlier generations. Your doctor or genetic counselor can talk with you more about the inheritance pattern of the neurological disorder in your family.
Possible Genetic Test Results

**POSITIVE**

A mutation was found in at least one of the genes tested

There may be management recommendations specific to the gene that has a mutation

Genetic testing for certain family members may be recommended

**NEGATIVE**

No genetic changes were found in any of the genes tested

Management recommendations are based on personal and family history

Talk to your healthcare provider to find out if genetic testing should be considered for your family members

**VARIANT OF UNKNOWN SIGNIFICANCE (VUS)**

At least one genetic change was found, but it is unclear if this change causes the neurological disorder or not

Management recommendations are based on personal and family history

Talk to your healthcare provider to find out if genetic testing should be considered for your family members

*It is possible to have a combination of positive and VUS results, since multiple genes are tested.*
Resources For You

Ambry’s Patient Education Website
ambrygen.com/patient

American Brain Tumor Association
abta.org

American Epilepsy Society
aesnet.org

Autism Speaks
autismspeaks.org

Child Neurology Foundation
childneurologyfoundation.org

Children’s Tumor Foundation
ctf.org

Citizens United For Research In Epilepsy
cureepilepsy.org

Danny Did Foundation
dannydid.org

Talk About Curing Autism (TACA)
tacanow.org

Tuberous Sclerosis Alliance
tsalliance.org

FIND A GENETIC COUNSELOR

National Society of Genetic Counselors
nsgc.org

Canadian Association of Genetic Counsellors
cagc-accg.ca
1 HOW IS GENETIC TESTING PERFORMED AND HOW LONG DOES IT TAKE?

Genetic testing is done using a blood or saliva sample, which is collected using a special kit that is shipped overnight to Ambry (all coordinated by your healthcare provider). Testing looks for mutations that cause the neurological disorder in your family. It takes between 1-10 weeks for the testing to be completed depending on which test your provider orders. Results are sent to your healthcare provider.

2 WHAT WILL HAPPEN WHEN MY RESULTS ARE READY?

Your healthcare provider will receive your results; they will not be sent directly to you. Every healthcare provider may have a different method and time frame to contact you to discuss your results, so it is important to discuss this process with them. Based on your test results, your healthcare provider will discuss any next steps.

3 WILL MY GENETIC TEST RESULTS AFFECT MY INSURANCE COVERAGE?

In the U.S., the Genetic Information Nondiscrimination Act (2008) prohibits discrimination by health insurance companies and employers, based on genetic information. Depending on where you live in the world, you may have different (or fewer) laws in this area. Visit ginahelp.org to learn more.
4 SHOULD I TELL MY FAMILY MEMBERS ABOUT MY GENETIC TEST RESULTS?

It is important to share your results with your family members as they may provide additional information about their chance of having the same disorder. Your healthcare provider may be able to guide you on finding the best way to inform family members.

5 WILL GENETIC TESTING BE COVERED BY MY INSURANCE?

Many insurance plans cover genetic testing and Ambry is contracted with the majority of U.S. health plans. Your out-of-pocket cost may vary based on your individual plan; therefore, we offer personalized verification of insurance coverage and financial options for your genetic testing. A team of dedicated specialists is available to help you get access to the genetic testing you need and answer any questions you have about our payment options. Call or email our Billing department at +1.949.900.5795 or billing@ambrygen.com with any questions.

6 WHAT IS AN EXPLANATION OF BENEFITS (EOB)?

Your insurance company sends you an EOB to explain any services paid on your behalf. You can contact us directly to speak with a Billing specialist with any questions or concerns about your EOB. Some genetic tests take weeks to process in order to receive the best results. In addition, insurance companies can take several weeks or even a couple of months to process claims.

STILL HAVE QUESTIONS?
Talk to your doctor or visit our website: ambrygen.com
Finding Answers.