

# Neurological Disorders

## Patient Guide

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A Guide To Genetic Testing  
For Hereditary Neurological Disorders



Ambry Genetics®

# Understanding The Basics

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Nearly **1 in 6** individuals in the world suffer from a neurological disorder<sup>1</sup>

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**1-2%** of children in the U.S. have an autism spectrum disorder<sup>2</sup>

## WHAT IS A NEUROLOGICAL DISORDER?

A neurological disorder is any medical problem that affects the nervous system, which includes the 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.



**1 in 6** children have a neurological disorder<sup>3</sup>



**1 in 26**  
people have  
epilepsy<sup>4</sup>

**2-3%**  
of people have  
an intellectual  
disability<sup>5</sup>

## CAUSES FOR NEUROLOGICAL DISORDERS

There are many causes for neurological disorders, including infections, injuries, and environmental factors. Gene changes can also cause neurological disorders.

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.

Genetic testing can be helpful in identifying the cause of the neurological disorder in many different situations.

# Understanding Disease Better Through Quality Testing

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**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 you or 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?



Samples from the patient (and parents\*) sent to the lab



Lab performs testing



Results available in 1-8 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 or Your Child

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

## EPILEPSY

- EpiRapid®
  - EpilepsyNext®
  - EpilepsyNext-Expanded™
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## NEURODEVELOPMENTAL DISORDERS

- Fragile X DNA Analysis
- AutismNext®
- NeurodevelopmentNext™
- CustomNext-Neuro™

## GENOMICS

- SNP Array
  - ExomeNext®
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## NEURO CUTANEOUS/NEURO-ONCOLOGY DISORDERS

- Neurofibromatosis 1 (NF1)
  - Neurofibromatosis 2 (NF2)
  - Legius Syndrome
  - Schwannomatosis
  - Tuberous Sclerosis Complex
  - BrainTumorNext®
  - Ataxia-Telangiectasia
  - von Hippel-Lindau Disease
  - Li-Fraumeni Syndrome
  - Gorlin Syndrome
  - Hereditary Hemorrhagic Telangiectasia, HHTNext®
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## OTHER NEUROLOGICAL DISORDERS

- Familial Hemiplegic Migraine

## VISIT OUR WEBSITE

See updated information on which genes are included on the test your healthcare provider selected above: [ambrygen.com/patient/neurotest](https://ambrygen.com/patient/neurotest)

# How Genetic Testing Can Impact You and Your Family



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

Examples of what to expect can be progression of current symptoms or additional health screenings that can find new symptoms as early as possible



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

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



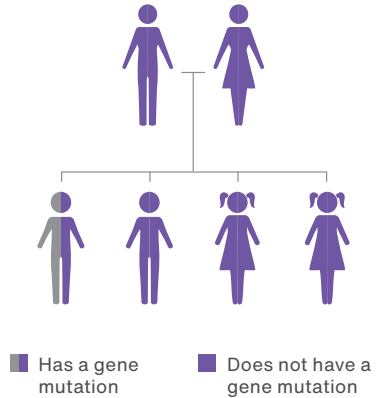
Your doctor can also identify and discuss other personalized medical management options that might be appropriate based on the genetic test results.

## AMBRY GENETICS OFFERS NO-COST TESTING TO RELATIVES

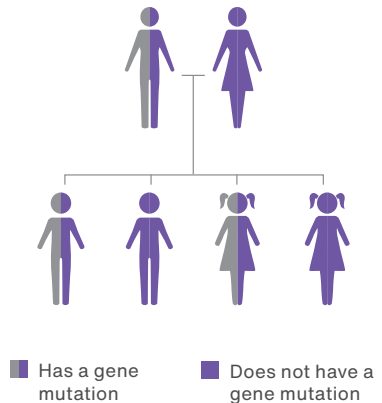
This is available for the specific genetic mutation identified in the first family member tested at Ambry within 90 days of the original report date.

### CAN NEUROLOGICAL DISORDERS RUN IN FAMILIES?

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

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## **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

Based on the results, genetic testing for certain family members may be recommended.

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## **NEGATIVE**

### **No genetic mutations 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.

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## **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.



# Resources For You and Your Family

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Ambry Patient Education  
Website

[ambrygen.com/patient](https://ambrygen.com/patient)

Children's Tumor Foundation

[ctf.org](https://ctf.org)

American Brain Tumor  
Association

[abta.org](https://abta.org)

Citizens United For Research  
In Epilepsy

[cureepilepsy.org](https://cureepilepsy.org)

American Epilepsy Society

[aesnet.org](https://aesnet.org)

Danny Did Foundation

[dannydid.org](https://dannydid.org)

Autism Speaks

[autismspeaks.org](https://autismspeaks.org)

Talk About Curing Autism (TACA)

[tacanow.org](https://tacanow.org)

Child Neurology Foundation

[childneurologyfoundation.org](https://childneurologyfoundation.org)

Tuberous Sclerosis Alliance

[tsalliance.org](https://tsalliance.org)



## FIND A GENETIC COUNSELOR

National Society of Genetic  
Counselors

[nsgc.org](https://nsgc.org)

Canadian Association of  
Genetic Counsellors

[cagc-accg.ca](https://cagc-accg.ca)

# Frequently Asked Questions

## 1 HOW IS GENETIC TESTING PERFORMED AND HOW LONG DOES IT TAKE?

Genetic testing requires a blood or saliva sample, which is collected using a special kit that is shipped overnight to Ambry Genetics by your healthcare provider. It takes between 1-8 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 THE RESULTS ARE READY?

Your healthcare provider will receive the results; they will not be sent directly to you. Every healthcare provider may have a different method and time frame for reviewing results with you, so it is important to discuss this process with them when the test is performed. Your healthcare provider will discuss recommended next steps based on the genetic test results.

## 3 DO GENETIC TEST RESULTS AFFECT 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](http://ginahelp.org) to learn more.

## 4 HOW WILL THE TEST RESULTS BE PROTECTED?

We are required by law to maintain the confidentiality of your protected health information in accordance with the Health Insurance Portability and Accountability Act (HIPAA). Visit [HHS.gov](http://HHS.gov) to learn more.

## **5 SHOULD I TELL MY FAMILY MEMBERS ABOUT THE GENETIC TEST RESULTS?**

It is important to share the results with your family members as they may provide additional information about their chance of having the same disorder. If you feel unsure about how to approach the subject, your healthcare provider may be able to offer some advice.

## **6 WILL GENETIC TESTING BE COVERED BY INSURANCE?**

Many insurance plans cover genetic testing, and Ambry Genetics is contracted with the majority of U.S. health plans. Your out-of-pocket cost may vary based on your individual plan. A team of dedicated specialists is available to help you get access to the genetic testing you need, and provide further details about our payment options. Please call or email our Billing department at +1.949.900.5795 or [billing@ambrygen.com](mailto:billing@ambrygen.com) with any questions. Visit [ambrygen.com/patientbilling](https://ambrygen.com/patientbilling) for more information.

## **7 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 Ambry Genetics genetic testing that appears on your EOB. It is important to remember that insurance companies can take several weeks or even a couple of months to process claims.

## **STILL HAVE QUESTIONS?**

Talk to your healthcare provider or visit our website: [ambrygen.com](https://ambrygen.com)



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#### References

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5. Bardoni, Barbara, et al. "Intellectual disabilities, neuronal posttranscriptional RNA metabolism, and RNA-binding proteins: three actors for a complex scenario." *Progress in brain research* 197 (2012): 29-51.