Hereditary Neuropathies

Hereditary neuropathies are genetic disorders that affect the peripheral nervous system. Symptoms of these conditions vary depending on which nerves are involved, but the arms and legs are often affected. Other symptoms include, but are not limited to, progressive muscle weakness, impaired motor skills, tingling in the hands and/or feet, and differences in physical appearance. Some neuropathies also feature abnormal sweating and/or insensitivity to pain. The symptoms of hereditary neuropathies may begin during infancy, but the onset may be as late as middle age.

WHAT CAUSES HEREDITARY NEUROPATHIES?

Hereditary neuropathies are caused by pathogenic (disease-causing) variants in genes that affect the processing and signaling of nerve cells throughout the body. Abnormal neuron signaling and function may lead to symptoms involving the voluntary and/or autonomic nervous systems. More than 30 genes have been linked to hereditary neuropathies and genetic testing may identify up to 70% of individuals with a hereditary neuropathy condition.

TYPES OF HEREDITARY NEUROPATHIES

There are several types of hereditary neuropathies. Generally, they are categorized by the type of nervous system involvement: Motor (muscle involvement), Sensory (feeling/sensation), or Autonomic (automatic body controls). The three most common hereditary neuropathies are:

- Hereditary Motor and Sensory Neuropathy (HMSN)
- Hereditary Motor Neuropathy
- Hereditary Sensory and Autonomic Neuropathy (HSAN)

WHO IS THIS TEST FOR?

This panel may be appropriate for anyone with a personal or family history of sensory, autonomic, or motor neuropathy. Additionally, those who have had abnormal neurological testing (Nerve conduction velocity, electromyography, or nerve biopsy), may also benefit from testing. There are many different types of hereditary neuropathies, so any patient experiencing abnormalities in the peripheral nerves may benefit from this test.
BENEFITS OF GENETIC TESTING

Genetic testing for hereditary neuropathies can:

- Establish or confirm the appropriate diagnosis
- Identify risks for additional health-related symptoms
- Assist in modifying lifestyle changes, including diet and exercise
- Result in more personalized treatment and symptom management
- Inform family members about their own risk factors
- Provide options for family planning

TEST SPECIFICATIONS

Acceptable Sample Requirements
- Blood, two 4-mL EDTA tubes (lavender top)
- Extracted DNA, 3 μg in EB buffer
- Buccal swab or saliva

Turnaround Time 3-5 weeks

Coverage ≥96% at 20x

Reporting
VUS, likely pathogenic, and pathogenic variants

Customization
Customizable Gene list, VUS opt-out

RELATED TESTS
Visit Fulgent website for most up-to-date list

Hereditary Neuropathies NGS Panel
Distal Hereditary Motor Neuropathy NGS Panel
Charcot Marie Tooth Disease Extended NGS Panel

GET CONNECTED

The Foundation for Peripheral Neuropathy - foundationforpn.org
National Organization for Rare Disorders - rarediseases.org
Global Genes - globalgenes.org

REFERENCES
- ncbi.nlm.nih.gov/pmc/articles/PMC2812746
- ninds.nih.gov/Disorders/All-Disorders/Hereditary-Neuropathies-Information-Page

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A Patient’s Guide to Genetic Testing

What does a genetic test check for?

Genetic testing checks the order of one’s DNA sequence (coded by the letters A, T, G, C) in specific genes linked to genetic conditions. Letters that were added, missing, or changed, are known as variants and can sometimes be harmful to one’s health, increasing the risk for a genetic condition.

What are the potential results?

There are three possible results from genetic testing:

Positive
A pathogenic/likely pathogenic variant is detected in one’s DNA. This type of variant is known to increase one’s risk of a genetic condition. Identifying the specific gene involved can help confirm a diagnosis, inform screening and management, and reveal risk factors for an individual and/or their family.

Negative
No variation known to be associated with a genetic condition was detected in one’s DNA. While a result may not show an increased risk for the condition(s) tested for, one can still be at risk for disease, especially if there is a family history.

Variant of Uncertain Significance (VUS)
A variant was detected in one’s DNA, however, not enough information is known about this variant to determine whether or not it is known cause the condition(s) tested for. More research is needed to better understand this variant.

What about family members?

Children, siblings, and parents of individuals who have a variant(s) identified in genetic testing could carry the same variant(s) and benefit from testing. Regardless of whether or not a variant was identified, individuals can still be at an increased risk for a genetic condition, especially with a family history.

Do genetic test results affect health insurance or employment?

No, the Genetic Information Nondiscrimination Act (GINA) was signed into law in 2008. It protects individuals from discrimination by an employer or a health insurance company based on genetic testing results and genetic information. GINA does not protect against life and disability insurance discrimination. For more information on GINA, go to www.ginahelp.org.

Where can I learn more?

Medline Plus/Genetics Home Reference – medlineplus.gov/genetics/understanding
National Society of Genetic Counselors – nsgc.org
Fulgent Genetics – fulgentgenetics.com/products/carrierscreening/learning.html