MAYO CLINIC LABORATORIES

HEREDITARY PERIPHERAL NEUROPATHY
SIMPLIFY AND IMPROVE DIAGNOSIS THROUGH PHENOTYPIC-FOCUSED TESTING

Hereditary peripheral neuropathies are a diverse group of disorders with varying presentations and genetic causes. Although these disorders can sometimes be categorized by extent of weakness, sensory involvement, and autonomic involvement, overlapping phenotypes can make it difficult to distinguish inherited disorders from acquired or idiopathic forms.

A comprehensive testing approach using next-generation sequencing (NGS) can help establish whether a peripheral neuropathy is genetic in nature, enabling greater value in testing. Genetic diagnosis not only helps direct gene-specific therapies, it's critical to advance pathologic understanding and development of RNA and gene therapies.[1]

Mayo Clinic Laboratories' hereditary peripheral neuropathy test menu includes both comprehensive and targeted NGS panels to assist in identifying genetic causes across the spectrum of peripheral neuropathies. Our comprehensive panel simplifies the diagnostic process by evaluating 186 genes with associations to peripheral neuropathies. Comprised only of clinically relevant genes, our panel was carefully vetted by a multidisciplinary team of neurologists and geneticists and includes 60 unique genes that enable an enhanced diagnostic yield over traditional testing approaches.

Key testing

PEPAN | Comprehensive Peripheral Neuropathy Gene Panel, Varies

Advantages

- Uses next-generation sequencing to evaluate 186 genes with known associations to hereditary peripheral neuropathy.
- Establishes a molecular diagnosis for patients with peripheral neuropathy.
- Identifies variants within genes known to be associated with peripheral neuropathy, allowing for predictive testing of at-risk family members.
- Offers improved sensitivity of more than 99.9% for small and mutations and copy number variants combined on all genes.

60+

60+ UNIQUE GENES OFFERED IN MAYO CLINIC LABORATORIES' HEREDITARY NEUROPATHY PANEL

30

30 GENES INCLUDED IN OUR HEREDITARY NEUROPATHY PANEL HAVE COMPLEMENTARY FUNCTIONAL TESTING

10%+

10%+ MORE CASES IDENTIFIED BASED ON EVALUATION OF ADDITIONAL 60 GENES[2]

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Comprehensive, clinically supported testing

Mayo Clinic Laboratories' robust menu of hereditary neuropathy testing covers all major categories of inherited peripheral neuropathies, including:

- Hereditary motor sensory neuropathy (HMSN) or Charcot-Marie-Tooth (CMT)
- Hereditary sensory neuropathy (HSN)
- Hereditary sensory autonomic neuropathy (HSAN)
- Brachial plexus
- Distal hereditary neuropathy (dHMN)

Among our offerings are targeted evaluations for:

- Parkinson's disease
- Frontotemporal dementia (FTD)
- Amyotrophic lateral sclerosis (ALS)
- Ataxia
- Spastic paraplegia
- Huntington disease

Meaningful results through an integrated approach

Mayo Clinic Laboratories' neurogenetic testing is backed by a multidisciplinary team of clinical peripheral neuropathy experts, laboratory scientists, and genetic counselors to provide the highest standard for variant detection.

Using NGS techniques and unique reporting methods, our approach allows for optimal mutation detection. This process eliminates the risk of false negative results compared to whole exome sequencing and other capture techniques, providing physicians confidence that disease-specific variants were not missed.

In addition, our custom gene ordering tool allows physicians to choose what genes they want and build custom panels to fit each patient's unique needs.

CGPH | Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies

Advantages

- Customizable and combinable.
- Ability to design a patient-specific genetic panel.
- Useful for single gene analysis.
- Ability to add specific genes to existing panels.
- Gene list IDs can be reused for multiple patients.

[1]Klein, C., Charcot-Marie-Tooth Disease and Other Hereditary Neuropathies. American Academy of Neurology. October 2020. [2]Internal Mayo Clinic data

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SCAN to learn more

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