WHY SHOULD I ORDER A COMPREHENSIVE EVALUATION?

Hereditary peripheral neuropathies are quite varied in their presentations and even more diverse in terms of their specific genetic causes. While they can sometimes be framed in terms of the extent of weakness, sensory involvement, and/or autonomic involvements, it is difficult to discern the appropriate test for many patients. A comprehensive testing approach with next-generation sequencing has been studied to show greater value in testing, and is therefore recommended.

Furthermore, previous testing approaches required testing a huge list of etiologies for acquired possibilities, while at the same time, trying to guess what individual gene may be most likely responsible. Those methods are extremely challenging, provide a low diagnostic yield, and result in high costs.

A simplified algorithm of evaluations (see insert) can be considered, especially among those younger than 40 years of age or with a family history.

CONSULT WITH MAYO CLINIC GENETIC COUNSELORS ABOUT TESTING OPTIONS

Genetic counselors bring real value to proper test utilization by ensuring the most appropriate test has been ordered. On average, Mayo Clinic genetic counselors modify 8% of all reviewed genetic test orders1, reducing health care costs and benefiting patient care.

Mayo Clinic genetic counselors are available to:
- Support the ordering process.
- Provide additional information about testing options.
- Offer results interpretation.
- Assist with case review and coordination.

WHICH TEST SHOULD I ORDER?

- **Peripheral Neuropathy Expanded Panel by Next-Generation Sequencing (Mayo ID: PNPAN)**

**USEFUL FOR**
- Diagnosis of hereditary peripheral neuropathies with isolated nerve involvement or associated with other organ system or associated with metabolic syndromes with known causal genes.
- Serving as a second-tier test for patients in which previous targeted gene mutation analyses for specific hereditary peripheral neuropathy-related genes were negative.
- Identifying mutations within genes known to be associated with hereditary peripheral neuropathy, allowing for predictive testing of at-risk family members.

**EXTENT OF DISEASE AND OVERLAP**

**BENEFITS OF A DEFINITIVE DIAGNOSIS**

Based on age and family history and specific phenotype, this testing can now be harnessed in an algorithmic approach, to:

- Establish treatment and surveillance options.
- Avoid unnecessary diagnostic procedures.
- Eliminate expensive and unnecessary treatments such as IVIG.
- Provide prognosis and counseling.
- Accurately assess risk for family members.

**ADDITIONAL TESTING USEFUL FOR THE DIAGNOSIS OF HEREDITARY NEUROPATHIES**

- **PMP22**, Peripheral Neuropathy, FISH (Deletion/Duplication) (Mayo ID: PMP22)
- Hereditary Motor and Sensory Neuropathy Panel by Next-Generation Sequencing (NGS) (Mayo ID: HMSNP)
- Hereditary Motor Neuropathy Panel by Next-Generation Sequencing (NGS) (Mayo ID: HMNP)
- Hereditary Sensory/Autonomic Neuropathy Panel by Next-Generation Sequencing (NGS) (Mayo ID: HSNP)
- Hereditary Spastic Paraplegia Neuropathy Panel by Next-Generation Sequencing (NGS) (Mayo ID: HSPP)
- Metabolic or Syndromic Neuropathy Panel by Next-Generation Sequencing (NGS) (Mayo ID: MSNP)
EXPERTISE AT MAYO CLINIC

Our board-certified laboratory directors in collaboration with clinical neurologists and genetic counselors at Mayo Clinic provide expertise in molecular genetic testing for inherited peripheral neuropathies.

FOR MORE INFORMATION ABOUT HEREDITARY PERIPHERAL NEUROPATHIES, VISIT

MayoMedicalLaboratories.com/HPN