MAYO CLINIC LABORATORIES

HEREDITARY NEUROMUSCULAR DISORDERS ENHANCE DIAGNOSIS THROUGH PHENOTYPIC-DIRECTED TESTING

The emergence of advanced molecular testing technologies has improved detection of inherited neuromuscular disorders. However, diverse presentation and varying genetic causes of these conditions often lead to lengthy diagnostic journeys when traditional testing strategies are used. Genetic testing to achieve a molecular diagnosis of neuromuscular disease is integral to optimizing patient care.[1]

Mayo Clinic Laboratories' distinctive approach to testing for genetic neuromuscular disorders uses phenotypic information to streamline diagnosis. Phenotype-specific panels enable targeted testing, improving diagnosis and minimizing uncertain results.

Our robust suite of genetic testing for inherited neuromuscular disorders includes both comprehensive and targeted next-generation sequencing (NGS) panels. Developed by an integrated team of clinicians, geneticists, and laboratory testing experts, our panels are carefully vetted to include only clinically relevant genes identified in literature and recommended in numerous professional clinical treatment guidelines.

Key testing

MUPAN | Comprehensive Neuromuscular Gene Panel, Varies

Advantages

- Uses NGS to analyze 215 genes associated with inherited neuromuscular diseases.
- Establishes diagnoses of neuromuscular disorders associated with known causal genes.
- Offers full overage on all included genes and provides high sensitivity for the detection of both small mutations and copy number variants.
- Serves as a second-tier test in patients for whom previous targeted gene mutation analyses for specific inherited neuromuscular disorder-related genes were negative.
- Identifies mutations within genes known to be associated with inherited neuromuscular disorders, allowing for predictive testing of at-risk family members.
- Aligns with American College of Medical Genetics and Genomics clinical testing recommendations.
- Includes results report with interpretive comments detailing the potential or known significance of detected variations.

215

215 GENES IN OUR COMPREHENSIVE PANEL

16

16 FOCUSED NEUROMUSCULAR GENETIC PANELS

44

44 GENETIC COUNSELORS SUPPORT GENETIC TESTING AT MAYO CLINIC LABORATORIES

MAYO CLINIC LABORATORIES

Gain personalized insights through precision testing

Mayo Clinic Laboratories' robust suite of focused, neuromuscular genetic testing spans the range of neuromuscular illnesses. Developed by our laboratory and genetic testing experts to identify genes with known associations to neuromuscular disease, our next-generation sequencing panels clarify and confirm genetic variation to place patients on the correct diagnostic pathway.

- Congenital myasthenic syndromes
- Myopathies
  - Congenital myopathy
  - Emery-Dreifuss
  - Metabolic myopathy
  - Muscular dystrophy
  - Myofibrillar myopathy
  - Rhabdomyolysis and myopathy
- Neuromuscular genetics
- Hyperexcitable muscle disease
- Motor neuron disease
- Biochemical genetic testing

Optimizing outcomes through an integrated approach

Mayo Clinic's board-certified laboratory directors, in collaboration with clinical neurologists, pathologists, and genetic counselors, provide the highest standard of testing for hereditary neuromuscular disorders. Genetic counselors add value to proper test utilization by assisting with case review and coordination to ensure the most appropriate test is ordered. On average, genetic counselors modify 8% of all reviewed genetic test orders,[2] reducing costs and enhancing patient care. Our genetic counselors are available to:

- Support the ordering process.
- Provide additional information about testing options.
- Offer results interpretation.

In addition, we offer a custom gene ordering tool, allowing 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

- 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]Kassardjian, c., amato, a., boon, a., childers, m., klien, c. aanem professional practice committee. The Utility of Genetic Testing in Neuromuscular Disease: A Consensus Statement from the AANEM on the Clinical Utility of Genetic Testing in Diagnosis of Neuromuscular disease. Policy Department, American Association of Neuromuscular & Electrodiagnostic Medicine, Rochester, Minnesota, USA. Accepted 22 August 2016 [2]Kotzer KE, Riley JD, Conta JH, et al. Genetic testing utilization and the role of the laboratory genetic counselor. *Clin Chem Acta.* 2014;427:193-195. doi:10.1016/j.cca.2013.09.033

FOR CLINICAL OR TECHNICAL SUPPORT CONTACT OUR SPECIALISTS: **855-516-8404 | +1-855-379-3115** (INTERNATIONAL)
NEWS.MAYOCLINICLABS.COM/NEUROGENETICS

SCAN to learn more

MC2775-647