MAYO CLINIC
Mayo Medical Laboratories

TEST ID: NMPAN

NEUROMUSCULAR GENETIC PANELS BY NEXT-GENERATION SEQUENCING (NGS)

USEFUL FOR

- Establishing a diagnosis of a neuromuscular disorder associated with known causal genes
- Serving as a second-tier test for patients in whom previous targeted gene mutation analyses for specific inherited neuromuscular disorder-related genes were negative
- Identifying mutations within genes known to be associated with inherited neuromuscular disorders, allowing for predictive testing of at-risk family members

GENETICS TEST INFORMATION

This ordered service includes the option for one of several neuromuscular disease-related panel tests to be performed. Testing options include the following:

Myopathies

- Myopathy Expanded Panel (141 genes)
- Muscular Dystrophy Panel (77 genes)
- Congenital Myopathy Panel (36 genes)
- Metabolic Myopathy Panel (41 genes)
- Myofibrillar Myopathy Panel (12 genes)
- Distal Myopathy Panel (27 genes)
- Emery-Dreifuss Panel (5 genes)
- Rhabdomyolysis and Myopathy Panel (31 genes)

Distal Myopathy + Peripheral Neuropathy

- Distal Weakness Expanded Panel (217 genes)

(See below for additional peripheral neuropathy testing options available)

Motor Neuron Disease

- Motor Neuron Disease Panel (17 genes)

REFERENCE VALUES

An interpretative report will be provided.

ANALYTIC TIME

10 weeks

11/2017

Neuromuscular Junction

- Congenital Myasthenic Syndromes Panel (25 genes)

Hyperexcitable Muscle Disease

- Skeletal Muscle Channelopathy Panel (6 genes)

See **Targeted Genes and Methodology Details for Neuromuscular Genetic Panels** in Special Instructions for details regarding the targeted genes for each test.

Related Testing

Focused hereditary peripheral neuropathy testing is available separately. See below for the different test IDs and the number of genes for various hereditary peripheral neuropathies panels:

- **PMP22 / *PMP22*, Peripheral Neuropathy, FISH** (1 gene)
- **PNPAN / Peripheral Neuropathy Expanded Panel by Next-Generation Sequencing (NGS)** (103 genes)
- **HMSNP / Hereditary Motor and Sensory Neuropathy Panel by Next-Generation Sequencing (NGS)** (43 genes)
- **HMNP / Hereditary Motor Neuropathy Panel by Next-Generation Sequencing (NGS)** (20 genes)
- **HSNP / Hereditary Sensory/Autonomic Neuropathy Panel by Next-Generation Sequencing (NGS)** (15 genes)
- **HSPP / Hereditary Spastic Paraplegia Neuropathy Panel by Next-Generation Sequencing (NGS)** (18 genes)
- **MSNP / Metabolic/Syndromic Neuropathy Panel by Next-Generation Sequencing (NGS)** (19 genes)

CLINICAL INFORMATION

Inherited neuromuscular disorders are a diverse group of diseases with heterogeneous genetic causes that affect the peripheral nervous system. The age of onset for these disorders ranges from in utero to old age. Based on the pattern of inheritance; clinical presentation; nerve conductions including, electromyography (EMG) pattern, and muscle and nerve biopsy findings; inherited neuromuscular disorders can be divided into major categories. These categories include muscular dystrophies, congenital muscular dystrophies, congenital myopathies, distal myopathies, ion channel hyperexcitable muscle diseases, metabolic myopathies, congenital myasthenic syndromes, hereditary motor and sensory neuropathies, hereditary motor neuropathies, motor neuron disorders, hereditary spastic paraplegias, and hereditary sensory neuropathies. Due to the considerable overlap in the clinical phenotypes of various neuromuscular disorders, it is often difficult to distinguish these specific inherited disorders from acquired forms without genetic testing. Additionally, even though most myopathies present with proximal shoulder and girdle weaknesses, some forms may present with distal weakness and, thereby, mimic neuropathies. Therefore, genetic testing can be extremely helpful in making the diagnosis. This is especially true for some genetic forms where neurophysiology may be ambiguous, as both neuropathy and myopathy exist simultaneously.

INTERPRETATION

All detected alterations are evaluated according to American College of Medical Genetics and Genomics recommendations. Variants are classified based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential or known significance.