TEST ID: HMNP

HEREDITARY MOTOR NEUROPATHY PANEL BY NEXT-GENERATION SEQUENCING (NGS)

USEFUL FOR

- Diagnosing distal hereditary motor neuropathy (dHMN) associated with known causal genes
- Second-tier testing for patients in whom previous targeted gene mutation analyses for specific inherited hereditary motor neuropathy-related genes were negative
- Identifying mutations within genes known to be associated with inherited hereditary motor neuropathy, allowing for predictive testing of at-risk family members

GENETICS INFORMATION

Next-generation sequencing and/or Sanger sequencing are performed to test for the presence of a mutation in the following genes: ATP7A, BICD2, BSCL2, DCTN1, DNAJB2, DYNC1H1, GARS, GJB1, HARS, HSPB1, HSPB3, HSPB8, IGHMBP2, PDK3, PLEKHG5, REEP1, SETX, SLC5A7, SOD1, and TRPV4.

CLINICAL INFORMATION

Inherited peripheral neuropathies are a relatively common, diverse group of disorders with heterogeneous genetic causes. Based on the pattern of inheritance and nerve conduction studies, inherited peripheral neuropathies with isolated nerve involvement can be divided into major categories. Distal hereditary motor neuropathies (dHMN) are one of the major categories of peripheral inherited neuropathies and are characterized by length-dependent, slowly progressive motor neuropathies with variable nerve conduction velocities. The clinical phenotype is variable, but includes progressive weakness and atrophy of the distal muscles, foot deformities, and decreased reflexes. There is significant phenotypic overlap with hereditary motor sensory neuropathy (HMSN), also known as Charcot-Marie-Tooth (CMT); however, sensory loss is usually absent in dHMN.

dHMN are subdivided into 11 subtypes based on inheritance pattern and clinical features and include types 1-7, dHMN plus pyramidal signs, X-linked, congenital distal SMA, and Jerash type.

Given the considerable phenotypic and genetic heterogeneity of dHMN, a comprehensive diagnostic genetic test is helpful to establish the genetic cause in this group of inherited neuropathies.

REFERENCE VALUES

An interpretive report will be provided.

ANALYTIC TIME

10 weeks
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SPECIMEN REQUIRED

Specimen Type
Whole blood

Container/Tube
Preferred: Lavender top (EDTA) or yellow top (ACD)
Acceptable: Any anticoagulant

Specimen Volume
3 mL

INTERPRETATION

All detected alterations are evaluated according to American College of Medical Genetics and Genomics recommendations.1 Variants are classified based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential or known significance.

CLINICAL REFERENCE