TEST ID: HSPP

HEREDITARY SPASTIC PARAPLEGIA NEUROPATHY PANEL BY NEXT-GENERATION SEQUENCING (NGS)

USEFUL FOR

- Diagnosis of inherited hereditary sensory (HSN) and autonomic neuropathy (HSAN) associated with known causal genes
- Second-tier testing for patients in whom previous targeted gene mutation analyses for specific inherited hereditary motor and sensory neuropathy-related genes were negative
- Identifying mutations within genes known to be associated with inherited hereditary motor and sensory 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: ATL1, BSCL2, CYP7B1, HSPD1, KIAA0196, KIF5A, NIPA1, PLP1, RTN2, SLC12A6, SLC16A2, SPAST, SPG11, SPG20, SPG21, SPG7, ZFYVE26, and ZFYVE27.

CLINICAL INFORMATION

Inherited peripheral neuropathies are a relatively common diverse group of disorders with heterogeneous genetic causes. Hereditary spastic paraplegia (HSP) is characterized by progressive lower extremity weakness and spasticity, and may present with prominent peripheral neuropathy as one of the complicated forms, also known as hereditary motor sensory neuropathy 5 (HMSN 5). The complicated forms are associated with a variety of other neurological systemic abnormalities and usually follow an autosomal recessive inheritance pattern. The uncomplicated or pure form presents with lower limb weakness and spasticity, and is predominantly characterized by an autosomal dominant inheritance pattern.

Given the considerable phenotypic and genetic heterogeneity of HSP, a comprehensive diagnostic genetic test is useful to establish the genetic cause in this HSP with neuropathy.

MOBILE APPS FROM MAYO MEDICAL LABORATORIES

Lab Catalog for iPad and Lab Reference for iPhone and iPod Touch
Requires iOS 5.1+

REFERENCE VALUES

An interpretive report will be provided.

ANALYTIC TIME

10 weeks

CONTENT AND VALUES SUBJECT TO CHANGE. SEE THE MAYO MEDICAL LABORATORIES TEST CATALOG FOR CURRENT INFORMATION.
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. Variants are classified based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential or known significance.

CLINICAL REFERENCE