TEST ID: HSNP
HEREDITARY SENSORY / AUTONOMIC 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, CCT5, DNMT1, DST, FAM134B, IKBKAP, KIF1A, NGF, NTRK1, SCN9A, SPTLC1, SPTLC2, SPTLC3, and WINK1/HSN2.

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. Hereditary sensory and autonomic neuropathies (HSAN), or hereditary sensory neuropathies (HSN) if autonomic dysfunction is absent, is one of these major categories of inherited peripheral neuropathies. They affect sensory and autonomic nerves and the hallmark feature is the presence of prominent small-fiber involvement. HSAN are subdivided into 5 groups based on age of onset, inheritance pattern, and clinical features:
- HSAN 1 varieties (HSAN 1A-E) follow an autosomal dominant inheritance pattern with juvenile or adult onset, and severe sensory loss and autonomic dysfunction
- HSAN 2-5 have an autosomal recessive inheritance pattern and are usually congenital
- HSAN3, also known as familial dysautonomia or Rilay-Day syndrome, is characterized by prominent autonomic and small-fiber sensory involvement
- HSAN 4 and 5 are characterized by insensitivity to pain and widespread autonomic disturbance, with HSAN 4 also featuring mental retardation

Given the considerable phenotypic and genetic heterogeneity of HSAN/HSN, a comprehensive diagnostic genetic test is useful to establish the genetic cause in this group of inherited neuropathies.
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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. Variants are classified based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential or known significance.

CLINICAL REFERENCE