TEST ID: PNPN
PERIPHERAL NEUROPATHY EXPANDED PANEL BY NEXT-GENERATION SEQUENCING (NGS)

USEFUL FOR
- Diagnosis of inherited peripheral neuropathies with isolated nerve involvement or associated with other organ system or associated with metabolic syndromes with known causal genes
- A second-tier test for patients in which previous targeted gene mutation analyses for specific inherited peripheral neuropathy-related genes were negative
- Identifying mutations within genes known to be associated with inherited peripheral neuropathy, allowing for predictive testing of at-risk family members

GENETICS INFORMATION
Next-generation sequencing and/or Sanger sequencing is performed to test for the presence of a mutation in the following genes: AARS, ABCD1, ARHGEF10, ARSA, ATL1, ATM, ATP7A, BICD2, BSCL2, CCT5, CTDP1, CYP27A1, CYP7B1, DCAF8, DCTN1, DHTKD1, DNAJB2, DNMT1, DST, DYNC1H1, EGR2, FAM134B, FBLN5, FGD4, FIG4, GALC, GAN, GARS, GBE1, GDAP1, GJB1, GJB3, GLA, GNB4, HARS, HINT1, HINT2, HK1, HMBS, HSPB1, HSPB3, HSPB8, HSPD1, IGHMBP2, IKBKAP, INF2, KARS, KIAA0196, KIF1A, KIF1B, KIF5A, L1CAM, LITAF, LMNA, LRSAM1, MED25, MFN2, MPZ, MTMR2, NDRG1, NEFL, NGF, NIPA1, NTRK1, PDK3, PHYH, PLEKHG5, PLP1, PMP22, PRPS1, PRX, RAB7A, REEP1, RIM2, SBF2, SCN9A, SETX, SH3TC2, SLC12A6, SLC16A2, SLC5A7, SOD1, SPAST, SPG11, SPG20, SPG21, SPG7, SPTLC1, SPTLC2, SPTLC3, TFG, TRIM2, TRPV4, TTR, TUBB3, TYMP, WNK1/HSN2, XPC, YARS, ZFYVE26, and ZFYVE27.

CLINICAL INFORMATION
Inherited peripheral neuropathies are a relatively common diverse group of disorders with heterogeneous genetic causes. Due to the considerable overlap in the clinical phenotypes of various neuropathies, it is often difficult to distinguish these specific inherited disorders from sporadic, idiopathic, or acquired forms of neuropathy without genetic testing. Based on the pattern of inheritance and nerve conduction studies, there are 3 major categories of inherited peripheral neuropathies with isolated nerve involvement:
- Hereditary motor and sensory neuropathy (HMSN), also referred as Charcot Marie Tooth (CMT)
- Hereditary sensory and autonomic neuropathy (HSAN), or hereditary sensory neuropathy (HSN), if autonomic dysfunction is absent
- Distal hereditary motor neuropathy (dHMSN)

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

03/2016
Inherited peripheral neuropathies may also show involvement of the central nervous system (brain or spinal cord), as in hereditary spastic paraplegia (HSP) with neuropathy (complicated form, also referred to as HSMN type 5) or be part of a systemic syndromic or metabolic disorder.

Given the considerable phenotypic overlap and the broad genetic heterogeneity of inherited peripheral neuropathies a comprehensive diagnostic genetic test is useful to establish the genetic cause in these clinical groups.

**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**