TEST ID: MSNP
METABOLIC / SYNDROMIC NEUROPATHY PANEL BY NEXT-GENERATION SEQUENCING (NGS)

USEFUL FOR
- Diagnosis of an inherited metabolic or syndromic neuropathy associated with known causal genes
- Second-tier testing for patients in whom previous targeted gene mutation analyses for specific inherited metabolic or syndromic neuropathy genes where negative
- Identifying mutations within genes known to be associated with inherited metabolic or syndromic neuropathy, allowing for predictive testing of at-risk family members

GENETICS INFORMATION
Next-generation sequencing is performed to test for the presence of a mutation in the following genes: ABCD1, ARSA, ATM, CTDP1, CYP27A1, FBLN5, GALC, GAN, GBE1, GJB3, GLA, HMBS, L1CAM, PHYH, PRPS1, TTR, TUBB3, TYMP, and XPC.

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. Additionally, peripheral neuropathy may be part of an inherited systemic syndromic or metabolic disorder caused by genes in metabolic pathways.

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 this group of inherited diseases.

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
**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**