MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Inherited Sensory Neuropathy Gene Panel

The following applies to ISNP / Inherited Sensory Neuropathy Gene Panel. Testing is performed to evaluate for the presence of variants in coding regions and extending to +/- 10 base pairs of adjacent intronic sequence on either side of the coding exons of the genes analyzed. In addition, the analysis will cover select non-coding variants. Next-generation sequencing and/or a polymerase chain reaction-based quantitative method is performed to test for the presence of copy number variants in the genes analyzed. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

This list is current from December 2022 to the present. This document is intended to highlight additional evaluations for variants of high clinical interest as well as technical limitations. However, this document does not comprehensively reflect all genomic regions covered by this test. For questions regarding transcripts, or genes or regions covered, contact the laboratory at 800-533-1710.

Genomic Build: GRCh37 (hg19) unless otherwise specified

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
AIFM1	NM_004208.4	c.697-44T>G	-
ATL1	NM_015915.4	-	-
ATL3	NM_015459.5	-	CNV in exon 5 may not be detected or reported
CLCF1	NM_013246.3	-	-
CLTCL1	NM_007098.4	-	-
COX20	NM_198076.6	-	-
CRLF1	NM_004750.5	-	-
DNMT1	NM_001130823.3	-	CNV in exon 5 may not be detected or reported
DST	NM_015548.5	-	-
DST	NM_001144769.3	-	-
ELP1	NM_003640.5	-	-
GLA	NM_000169.2	c.640-801C>T c.640-859G>A	-
KIF1A	NM_004321.7	-	-
NGF	NM_002506.3	-	-
NTRK1	NM_001012331.1	c.851-33T>A	-
PRDM12	NM_021619.3	-	-
PRKCG	NM_002739.5	-	-
RETREG1	NM_001034850.2	-	-
SCN10A	NM_006514.3	-	-
SCN11A	NM_014139.2	-	-
SCN9A	NM_002977.3	-	-
SPTLC1	NM_006415.4	-	-
SPTLC2	NM_004863.3	-	-
WNK1	NM_213655.4	-	-