MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Inherited Motor Neuropathy Gene Panel

The following applies to IMNP / Inherited Motor 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
ATP7A	NM_000052.7	-	-
BICD2	NM_001003800.2	-	-
BSCL2	NM_032667.6	-	-
CPOX	NM_000097.7	-	-
DCTN1	NM_004082.4	-	-
DNAJB2	NM_001039550.2	-	-
FBXO38	NM_030793.5	-	-
GARS1	NM_002047.4	-	-
GBF1	NM_004193.3	-	-
HINT1	NM_005340.7	-	-
HSPB1	NM_001540.5	-	-
HSPB8	NM_014365.2	-	-
IGHMBP2	NM_002180.2	-	-
PLEKHG5	NM_020631.5	-	-
SETX	NM_015046.7	-	-
SIGMAR1	NM_005866.4	-	-
SLC5A7	NM_021815.5	-	-
SMN1	NM_022874.2	g.27134T>G (NG_008691.1) Provided only upon request	Analyzed for the presence of exon 7 CNV only; analysis for other sequence variants and CNV will not be performed.
SMN2	NM_022876.2	-	Analyzed for the presence of exon 7 CNV only; analysis for other sequence variants and CNV will not be performed.
SOD1	NM_000454.4	c.358-11A>G	-
SORD	NM_003104.6	-	Sequence variants in exons 3 and 9 will not be detected or reported, CNV in exons 3 and 8–9 will not be detected or reported.
SPTAN1	NM_001130438.3	-	-
TRPV4	NM_021625.5	-	-
VRK1	NM_003384.3	-	-
VWA1	NM_022834.5	-	-
WARS1	NM_004184.4	-	-