MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Comprehensive Peripheral Neuropathy Gene Panel

The following applies to PEPAN / Comprehensive Peripheral 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 test was updated August 2024. 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
AAAS	NM_015665.6	-	-
AARS1	NM_001605.2	-	-
ABCA1	NM_005502.4	c.4176-11T>G	-
ABCD1	NM_000033.4	c.-16_10del	-
AIFM1	NM_004208.4	c.697-44T>G	-
ALDH18A1	NM_002860.4	-	-
AMACR	NM_014324.6	-	-
AP5Z1	NM_014855.3	-	-
APOA1	NM_000039.2	-	-
APTX	NM_175073.2	-	-
ARSA	NM_000487.6	-	-
ATL1	NM_015915.4	-	-
ATL3	NM_015459.5	-	CNV in exon 5 may not be detected or reported
ATM	NM_000051.3	c.4612-12A>G c.6573-12C>A	-
ATP1A1	NM_000701.8	-	-
ATP7A	NM_000052.7	-	-
B4GALNT1	NM_001478.5	-	-
BAG3	NM_004281.3	-	-
BICD2	NM_001003800.2	-	-
BSCL2	NM_032667.6	-	-
C12orf65 (MTRFR)	NM_152269.5	-	-
C1orf194	NM_001122961.2	-	-
CHCHD10	NM_213720.3	-	-
CLCF1	NM_013246.3	-	-
CLTCL1	NM_007098.4	-	-
CNTNAP1	NM_003632.3	-	-

Targeted Genes and Methodology Details for Comprehensive Peripheral Neuropathy Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
COQ4	NM_016035.5	-	-
COQ7	NM_016138.5	-	-
COX10	NM_001303.4	-	CNV in exon 6 may not be detected or reported
COX20	NM_198076.6	-	-
COX6A1	NM_004373.4	-	-
CPOX	NM_000097.7	-	-
CRLF1	NM_004750.5	-	-
CTDP1	NM_004715.4	c.863+389C>T	-
CYP27A1	NM_000784.4	-	-
CYP2U1	NM_183075.3	-	-
CYP7B1	NM_004820.5	-	-
DCTN1	NM_004082.4	-	-
DDHD1	NM_001160147.2	-	-
DGAT2	NM_032564.5	-	-
DHH	NM_021044.4	-	-
DNAJB2	NM_001039550.2	-	-
DNM2	NM_001005360.2	-	-
DNMT1	NM_001130823.3	c.251-1532dup	-
DST	NM_015548.5	-	-
DST	NM_001144769.3	-	-
DYNC1H1	NM_001376.5	-	-
EGR2	NM_000399.5	-	-
ELP1	NM_003640.5	-	-
ERCC8	NM_000082.3	-	CNV in exon 5 may not be detected or reported
FA2H	NM_024306.5	-	-
FAM126A	NM_032581.4	-	-
FBLN5	NM_006329.3	-	-
FBXO38	NM_030793.5	-	-
FGD4	NM_139241.3	-	-
FGF14	NM_004115.3	-	-
FIG4	NM_014845.5	-	CNV in exon 17 may not be detected or reported
FLVCR1	NM_014053.4	-	-
FMR1	NM_002024.5	-	CNV in exon 2 may not be detected or reported
FXN	NM_000144.5	-	-

Targeted Genes and Methodology Details for Comprehensive Peripheral Neuropathy Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
GALC	NM_000153.4	c.-66G>C	CNV in exon 6 may not be detected or reported
GAN	NM_022041.3	-	CNV in exon 2 may not be detected or reported
GARS1	NM_002047.4	-	-
GBA2	NM_020944.3	-	-
GBE1	NM_000158.4	c.2053-3358_2053-3350delinsTGTTTTTTACATGACAGGT	-
GBF1	NM_004193.3	-	-
GDAP1	NM_018972.4	-	-
GJB1	NM_000166.6	c.-103C>T c.-17G>A c.-17+1G>T c.-17+2T>C	-
GLA	NM_000169.2	c.640-801C>T c.640-859G>A	-
GM2A	NM_000405.5	-	-
GNB4	NM_021629.4	-	-
GSN	NM_000177.5	-	CNV in exon 16 may not be detected or reported
HADHA	NM_000182.5	-	CNV in exon 14 may not be detected or reported
HADHB	NM_000183.3	-	-
HARS1	NM_002109.6	-	-
HEXA	NM_000520.6	-	-
HEXB	NM_000521.4	c.1509-26G>A	CNV in exon 4 may not be detected or reported
HINT1	NM_005340.7	-	-
HK1	NM_000188.2	c.-40257G>C c.-40237G>C	-
HMBS	NM_000190.4	-	-
HSPB1	NM_001540.5	-	-
HSPB8	NM_014365.2	-	-
HSPD1	NM_002156.5	-	-
IARS2	NM_018060.4	-	-
IBA57	NM_001010867.4	-	-
IGHMBP2	NM_002180.2	-	-
INF2	NM_022489.4	-	-
KARS1	NM_001130089.1	-	-
KIF1A	NM_004321.7	-	-
KIF5A	NM_004984.4	-	-

Targeted Genes and Methodology Details for Comprehensive Peripheral Neuropathy Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
LAMA2	NM_000426.3	c.3556-13T>A	-
LITAF	NM_004862.3	-	-
LMNA	NM_170707.4	c.1698+13C>A	-
LRSAM1	NM_138361.5	-	-
MARS1	NM_004990.4	-	-
MCM3AP	NM_003906.5	-	-
MFN2	NM_014874.4	-	-
MME	NM_007289.3	-	-
MORC2	NM_001303256.3	-	-
MPC1	NM_016098.4	-	CNV in exon 2 may not be detected or reported
MPV17	NM_002437.5	-	-
MPZ	NM_000530.8	-	-
MTMR2	NM_016156.5	-	-
MTTP	NM_000253.3	-	-
MYH14	NM_024729.3	-	-
NAGLU	NM_000263.4	-	-
NDRG1	NM_006096.4	-	-
NEFH	NM_021076.4	-	-
NEFL	NM_006158.4	-	-
NF2	NM_000268.3	-	-
NGF	NM_002506.3	-	-
NIPA1	NM_144599.5	-	-
NTRK1	NM_001012331.1	c.851-33T>A	-
OPA1	NM_015560.2	c.625-5459G>A	-
PDK3	NM_001142386.3	-	-
PDYN	NM_024411.5	-	-
PEX7	NM_000288.4	c.-45C>T	-
PHYH	NM_006214.4	-	-
PLA2G6	NM_003560.4	-	-
PLEKHG5	NM_020631.5	-	-
PLP1	NM_000533.5	c.453+28_453+46del c.453+159G>A c.453+164G>A c.454-322G>A c.454-314T>A/G c.454-312C>G	-
PMP2	NM_002677.5	-	-
PMP22	NM_000304.4	-	-

Targeted Genes and Methodology Details for Comprehensive Peripheral Neuropathy Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
PNKP	NM_007254.4	c.1386+49_1387-33del	-
PNPLA6	NM_006702.5	-	-
POLG	NM_002693.2	-	-
PPOX	NM_000309.5	-	-
PRDM12	NM_021619.3	-	-
PRKCG	NM_002739.5	-	-
PRNP	NM_000311.5	-	-
PRPS1	NM_002764.4	-	-
PRX	NM_181882.3	-	-
PTRH2	NM_016077.4	-	-
RAB7A	NM_004637.6	-	-
REEP1	NM_022912.3	-	-
RETREG1	NM_001034850.2	-	-
RNASEH1	NM_002936.5	-	-
RRM2B	NM_015713.5	-	-
RTN2	NM_005619.5	-	-
SACS	NM_014363.6	-	-
SBF1	NM_002972.4	-	-
SBF2	NM_030962.3	-	CNV in exon 10 may not be detected or reported
SCN10A	NM_006514.3	-	-
SCN11A	NM_014139.2	-	-
SCN9A	NM_002977.3	-	-
SCO2	NM_005138.2	-	-
SEPTIN9	NM_006640.4	c.-134G>C	-
SETX	NM_015046.7	-	-
SH3TC2	NM_024577.4	-	-
SIGMAR1	NM_005866.4	-	-
SLC12A6	NM_133647.1	-	CNV in exon 3 may not be detected or reported
SLC25A19	NM_021734.4	-	-
SLC25A46	NM_138773.4	-	-
SLC52A2	NM_024531.5	-	-
SLC52A3	NM_033409.4	-	-
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

Targeted Genes and Methodology Details for Comprehensive Peripheral Neuropathy Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
SMN2	NM_022876.2	-	Analyzed for the presence of exon 7 CNV only; analysis for other sequence variants and CNV will not be performed
SNAP29	NM_004782.4	-	-
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
SOX10	NM_006941.4	-	-
SPAST	NM_014946.3	-	-
SPG11	NM_025137.4	-	-
SPG21	NM_016630.7	-	-
SPG7	NM_003119.4	-	-
SPTAN1	NM_001130438.3	-	-
SPTLC1	NM_006415.4	-	-
SPTLC2	NM_004863.3	-	-
SUCLA2	NM_003850.2	-	-
SURF1	NM_003172.4	-	-
TDP1	NM_018319.4	-	-
TFG	NM_006070.6	-	-
TRIM2	NM_001130067.2	-	-
TRPV4	NM_021625.5	-	-
TSFM	NM_001172696.2	-	CNV in exon 5 may not be detected or reported
TTPA	NM_000370.3	-	-
TTR	NM_000371.3	-	-
TUBB3	NM_006086.4	-	-
TWNK	NM_021830.5	-	-
TYMP	NM_001953.5	-	-
UBA1	NM_003334.4	-	-
VPS13D	NM_015378.4	-	-
VRK1	NM_003384.3	-	-
VWA1	NM_022834.5	-	-
WARS1	NM_004184.4	-	-
WASHC5	NM_014846.4	-	-
WNK1	NM_213655.4	-	-
YARS1	NM_003680.3	-	-

Targeted Genes and Methodology Details for Comprehensive Peripheral Neuropathy Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
ZFYVE26	NM_015346.4	-	-

Effective Date	Version	Synopsis of Test Change
December 14, 2022	V2	Added genes *HEXA* and *HEXB*
August 6, 2024	V3	Added gene *SEPTIN9*