MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Neurologic Disorders Custom Gene Panel

The following applies to CGPH / Neurologic Disorders Custom 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. A polymerase chain reaction-based assay is performed to test for the presence of C9orf72 GGGGCC hexanucleotide repeat expansions. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

This list is current from August 2024 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
AAAS	NM_015665.6	-	-
AARS1	NM_001605.2	-	-
ABCA1	NM_005502.4	c.4176-11T>G	-
ABCB7	NM_004299.6	-	-
ABCD1	NM_000033.4	c.-16_10del	-
ABHD12	NM_001042472.3	-	-
ABHD5	NM_016006.6	-	-
ACAD9	NM_014049.5	-	-
ACADM	NM_000016.5	c.388-19T>A c.388-14A>G c.600-18G>A	CNV in exon 2 may not be detected or reported
ACADS	NM_000017.4	-	-
ACADVL	NM_000018.4	c.1183-15A>G	-
ACO2	NM_001098.3	-	-
ACTA1	NM_001100.4	-	-
ADORA1	NM_000674.3	-	-
ADPRHL2 (ADPRS)	NM_017825.3	-	-
ADSS1	NM_199165.2	-	-
AFG3L2	NM_006796.3	-	-
AGK	NM_018238.4	-	-
AGL	NM_000642.3	c.4260-12A>G	-
AGRN	NM_198576.4	-	-
AHSA1	NM_012111.3	-	-
AIFM1	NM_004208.4	c.697-44T>G	-
ALDH18A1	NM_002860.4	-	-
ALDH5A1	NM_001080.3	-	-
ALDOA	NM_000034.3	-	-
ALG14	NM_144988.4	-	-
ALG2	NM_033087.4	-	-
ALG6	NM_013339.4	-	-

Targeted Genes and Methodology Details for Neurologic Disorders Custom Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
ALS2	NM_020919.4	-	-
AMACR	NM_014324.6	-	-
AMPD2	NM_001257360.1	-	-
ANG	NM_001145.4	-	-
ANO10	NM_018075.5	-	-
ANO3	NM_031418.4	-	-
ANO5	NM_213599.2	-	CNV in exon 2 may not be detected or reported
ANXA11	NM_001157.3	-	-
AP4B1	NM_006594.5	-	-
AP4E1	NM_007347.5	-	-
AP4M1	NM_004722.4	-	-
AP4S1	NM_007077.4	-	-
AP5Z1	NM_014855.3	-	-
APOA1	NM_000039.2	-	-
APOA2	NM_001643.2	-	-
APOPT1 (COA8)	NM_032374.4	-	-
APP	NM_000484.4	-	-
APTX	NM_175073.2	-	-
ARG1	NM_000045.4	-	-
ARL6IP1	NM_015161.3	-	-
ARSA	NM_000487.6	-	-
ASAH1	NM_177924.5	-	CNV in exon 4 may not be detected or reported
ASCC1	NM_001198800.3	-	-
ASNS	NM_133436.3	-	-
ATCAY	NM_033064.5	-	-
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	-
ATP13A2	NM_022089.4	-	-
ATP1A1	NM_000701.8	-	-
ATP1A2	NM_000702.4	-	-
ATP1A3	NM_152296.5	-	-
ATP2A1	NM_173201.4	-	-
ATP6AP2	NM_005765.3	c.301-11_301-10del	-
ATP7A	NM_000052.7	-	-

Targeted Genes and Methodology Details for Neurologic Disorders Custom Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
ATP7B	NM_000053.4	c.-676A>G c.-460G>A c.-447C>T c.-442G>A c.-436_-422del15 c.-388C>T c.-210A>T c.-133A>C c.-128A>C c.-123C>A c.-128_-124delAGCCG c.1947-19T>A c.3061-12T>A	-
ATP8A2	NM_016529.6	-	CNV in exon 6 may not be detected or reported
AUH	NM_001698.2	-	-
B2M	NM_004048.3	-	-
B3GALNT2	NM_152490.5	-	-
B4GALNT1	NM_001478.5	-	-
B4GAT1	NM_006876.3	-	-
BAG3	NM_004281.3	-	-
BICD2	NM_001003800.2	-	-
BIN1	NM_139343.3	-	-
BOLA3	NM_212552.3	-	-
BSCL2	NM_032667.6	-	-
BVES	NM_007073.4	-	-
C1orf194	NM_001122961.2	-	-
C9orf72	NM_001256054.3	GGGGCC hexanucleotide repeat expansion	Analyses for sequence variants and CNV will not be performed
C12orf65 (MTRFR)	NM_152269.5	-	-
C19orf12	NM_001031726.3	-	CNV in exon 1 may not be detected or reported
CA8	NM_004056.6	-	-
CACNA1A	NM_001127221.1	-	-
CACNA1G	NM_018896.5	-	-
CACNA1S	NM_000069.3	-	-
CAMTA1	NM_015215.4	-	-
CAPN1	NM_001198868.2	-	-
CAPN3	NM_000070.3	c.380-13T>A c.2184+21G>A c.2185-16A>G	-
CASQ1	NM_001231.5	-	-
CAV3	NM_033337.3	-	-
CAVIN1	NM_012232.6	-	-
CC2D2A	NM_001080522.2	-	-
CCDC88C	NM_001080414.4	-	-

Targeted Genes and Methodology Details for Neurologic Disorders Custom Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
CCNF	NM_001761.3	-	-
CFL2	NM_021914.7	-	-
CHAT	NM_020549.4	-	-
CHCHD10	NM_213720.3	-	-
CHCHD2	NM_016139.4	-	-
CHKB	NM_005198.4	-	-
CHMP2B	NM_014043.4	-	-
CHRNA1	NM_000079.4	-	-
CHRNB1	NM_000747.3	-	-
CHRND	NM_000751.3	-	-
CHRNE	NM_000080.4	c.501-16G>A c.-94G>A c.-95G>A c.-96C>T	-
CLCF1	NM_013246.3	-	-
CLCN1	NM_000083.3	-	-
CLCN2	NM_004366.6	-	-
CLN3	NM_001042432.1	-	-
CLN5	NM_006493.4	-	-
CLPP	NM_006012.4	-	-
CLTCL1	NM_007098.4	-	-
CNTNAP1	NM_003632.3	-	-
COL12A1	NM_004370.6	-	-
COL13A1	NM_001130103.2	-	CNV in exons 3 and 33 may not be detected or reported
COL6A1	NM_001848.3	c.930+189C>T	-
COL6A2	NM_001849.4	-	-
COL6A3	NM_004369.4	-	-
COLQ	NM_005677.4	-	-
COQ2	NM_015697.8	-	-
COQ4	NM_016035.5	-	-
COQ6	NM_182476.3	-	-
COQ7	NM_016138.5	-	-
COQ8A	NM_020247.5	-	-
COQ9	NM_020312.4	-	-
COX10	NM_001303.4	-	CNV in exon 6 may not be detected or reported
COX20	NM_198076.6	-	-
COX6A1	NM_004373.4	-	-
CP	NM_000096.4	-	CNV in exon 19 may not be detected or reported
CPOX	NM_000097.7	-	-
CPT1C	NM_001136052.2	-	CNV in exons 6-7 may not be detected or reported

Targeted Genes and Methodology Details for Neurologic Disorders Custom Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
CPT2	NM_000098.3	-	-
CRLF1	NM_004750.5	-	-
CRPPA	NM_001101426.4	-	-
CRYAB	NM_001885.3	-	-
CSF1R	NM_005211.3	c.1969+115_1969+116del c.1859-119G>A	
CST3	NM_000099.4	-	-
CTBP1	NM_001328.3	-	CNV in exon 1 may not be detected or reported
CTC1	NM_025099.6	-	-
CTDP1	NM_004715.4	c.863+389C>T	-
CTSD	NM_001909.5	-	-
CWF19L1	NM_018294.6	-	-
CYP27A1	NM_000784.4	-	-
CYP2U1	NM_183075.3	-	-
CYP7B1	NM_004820.5	-	-
DAG1	NM_004393.6	-	-
DARS1	NM_001349.4	-	CNV in exons 2–3, 5–10, 12–14 will not be detected or reported
DARS2	NM_018122.5	c.228-33 to c.228-11 c.1345-17_1345-5del	-
DCAF17	NM_025000.4	-	-
DCTN1	NM_004082.4	-	-
DDC	NM_000790.4	-	-
DDHD1	NM_001160147.2	-	-
DDHD2	NM_015214.3	-	-
DES	NM_001927.4	-	-
DGAT2	NM_032564.5	-	-
DGUOK	NM_080916.3	c.444-62C>A c.444-11C>G	-
DHH	NM_021044.4	-	-
DHTKD1	NM_018706.7	-	-
DLD	NM_000108.5	-	-
DMD	NM_004006.2	c.-54T>A c.31+36947G>A c.3408+2036A>G c.9225-648A>G c.9225-647A>G c.9225-287C>A c.9225-285A>G	-
DNA2	NM_001080449.3	-	-
DNAJB2	NM_001039550.2	-	-
DNAJB6	NM_058246.4	-	-
DNAJC12	NM_021800.3	-	-
DNAJC13	NM_015268.4	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
DNAJC19	NM_145261.4	-	-
DNAJC3	NM_006260.5	-	-
DNAJC6	NM_001256864.2	-	-
DNM1L	NM_012062.5	c.251-1532dup	-
DNM2	NM_001005360.2	-	-
DNMT1	NM_001130823.3	-	CNV in exon 5 may not be detected or reported
DOK7	NM_173660.5	c.54+25_55-38del	-
DPAGT1	NM_001382.4	-	-
DPM1	NM_003859.2	-	-
DPM2	NM_003863.3	-	-
DPM3	NM_153741.2	-	-
DST	NM_015548.5	-	-
DST	NM_001144769.3	-	-
DYNC1H1	NM_001376.5	-	-
DYSF	NM_003494.4	c.1054-43_1059delinsA	-
EARS2	NM_001083614.2	-	-
EBF3	NM_001005463.3	-	-
EGR2	NM_000399.5	-	-
EIF2B1	NM_001414.4	-	-
EIF2B2	NM_014239.4	-	-
EIF2B3	NM_020365.5	-	-
EIF2B4	NM_015636.3	-	-
EIF2B5	NM_003907.3	-	-
EIF4G1	NM_198241.3	-	-
ELOVL4	NM_022726.4	-	-
ELOVL5	NM_021814.5	-	-
ELP1	NM_003640.5	-	-
EMD	NM_000117.3	-	-
ENO3	NM_053013.4	-	-
ENTPD1	NM_001776.6	-	-
EPM2A	NM_005670.4	-	-
ERBB4	NM_005235.3	-	-
ERCC8	NM_000082.3	-	CNV in exon 5 may not be detected or reported
ERLIN1	NM_006459.4	-	-
ERLIN2	NM_007175.8	-	-
ETFA	NM_000126.4	-	CNV in exon 12 may not be detected or reported
ETFB	NM_001985.3	-	-
ETFDH	NM_004453.4	c.-75A>G	-
EXOSC3	NM_016042.4	-	-

Targeted Genes and Methodology Details for Neurologic Disorders Custom Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
FA2H	NM_024306.5	-	-
FAM111B	NM_198947.4	-	-
FAM126A	NM_032581.4	-	-
FAR1	NM_032228.6	-	-
FARS2	NM_006567.5	-	-
FBLN5	NM_006329.3	-	-
FBXL4	NM_012160.4	-	-
FBXO38	NM_030793.5	-	-
FBXO7	NM_012179.4	-	-
FDX2	NM_001031734.4	-	-
FGA	NM_021871.4	-	-
FGD4	NM_139241.3	-	-
FGF14	NM_004115.3	-	-
FHL1	NM_001449.5	-	-
FIG4	NM_014845.5	-	CNV in exon 17 may not be detected or reported
FKBP14	NM_017946.4	-	-
FKRP	NM_024301.5	c.-272G>A c.-253+4A>G	-
FKTN	NM_001079802.1	c.648-1243G>T c.*4375_*4376ins3062	-
FLAD1	NM_025207.5	-	-
FLNC	NM_001458.4	-	-
FLVCR1	NM_014053.4	-	-
FMR1	NM_002024.5	-	CNV in exon 2 may not be detected or reported
FOLR1	NM_016725.3	-	-
FTL	NM_000146.4	5'UTR c.-200 to c.-143	-
FUCA1	NM_000147.4	-	-
FUS	NM_004960.3	c.*59G>A	-
FXN	NM_000144.5	-	-
GAA	NM_000152.5	c.-32-18 to c.-11 c.1076-22T>G c.2647-20T>G	-
GALC	NM_000153.4	c.-66G>C	CNV in exon 6 may not be detected or reported
GAMT	NM_000156.6	-	-
GAN	NM_022041.3	-	CNV in exon 2 may not be detected or reported
GARS1	NM_002047.4	-	-
GBA	NM_001005741.3	-	-
GBA2	NM_020944.3	-	-
GBE1	NM_000158.4	c.2053-3358_2053-3350delinsTGTTTTTTACATGACAGGT	-
GBF1	NM_004193.3	-	-

Targeted Genes and Methodology Details for Neurologic Disorders Custom Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
GCDH	NM_000159.4	-	-
GCH1	NM_000161.3	-	CNV in exon 4 may not be detected or reported
GDAP1	NM_018972.4	-	-
GDAP2	NM_017686.4	-	CNV in exon 12 may not be detected or reported
GFAP	NM_002055.5	c.1171+475_1171+482delinsATC c.1171+472G>A	-
GFER	NM_005262.3	c.259-25_259-24del	-
GFPT1	NM_001244710.1	-	CNV in exon 5 may not be detected or reported
GIGYF	NM_001103146.2	-	CNV in exons 8 and 18 may not be detected or reported
GJB1	NM_000166.6	c.-103C>T c.-17G>A c.-17+1G>T c.-17+2T>C	-
GJC2	NM_020435.4	-	-
GLA	NM_000169.2	c.640-801C>T c.640-859G>A	-
GLB1	NM_000404.4	-	-
GM2A	NM_000405.5	-	-
GMPPB	NM_013334.3	-	-
GNB4	NM_021629.4	-	-
GNE	NM_001128227.3	-	-
GOLGA2	NM_004486.5	-	-
GOSR2	NM_004287.4	-	CNV in exon 2 may not be detected or reported
GPT2	NM_133443.4	-	-
GRID2	NM_001510.4	-	-
GRM1	NM_001278064.2	-	-
GRN	NM_002087.3	c.-256 to c.-8+10	-
GSN	NM_000177.5	-	CNV in exon 16 may not be detected or reported
GYG1	NM_004130.3	-	-
GYS1	NM_002103.5	-	-
HACE1	NM_020771.4	-	CNV in exons 5 and 7 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	-	-
HEPACAM	NM_152722.5	-	-
HEXA	NM_000520.6	-	-
HEXB	NM_000521.4	c.1509-26G>A	CNV in exon 4 may not be detected or reported
HIBCH	NM_014362.4	-	-

Targeted Genes and Methodology Details for Neurologic Disorders Custom Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
HINT1	NM_005340.7	-	-
HK1	NM_000188.2	c.-40257G>C c.-40237G>C	-
HMBS	NM_000190.4	-	-
HNRNPA1	NM_031157.4	-	-
HNRNPA2B1	NM_031243.3	-	-
HNRNPDL	NM_031372.3	-	-
HRAS	NM_005343.4	-	-
HSPB1	NM_001540.5	-	-
HSPB8	NM_014365.2	-	-
HSPD1	NM_002156.5	-	-
HSPG2	NM_005529.7	-	CNV in exon 1 will not be detected or reported
HTRA1	NM_002775.5	-	-
HTRA2	NM_013247.4	-	-
IARS2	NM_018060.4	-	-
IBA57	NM_001010867.4	-	-
IFIH1	NM_022168.4	-	-
IGHMBP2	NM_002180.2	-	-
INF2	NM_022489.4	-	-
INPP5K	NM_016532.4	-	-
IRF2BPL	NM_024496.4	-	-
ISCA2	NM_194279.4	-	-
ISCU	NM_213595.3	c.343+382G>C	-
ITGA7	NM_002206.3	-	-
ITM2B	NM_021999.5	-	-
ITPR1	NM_002222.6	-	-
JAG2	NM_002226.5	-	Sequence variants in exon 1 may not be detected or reported, CNV in exon 1 will not be detected or reported
KARS1	NM_001130089.1	-	-
KBTBD13	NM_001101362.2	-	-
KCNA1	NM_000217.3	-	-
KCNA2	NM_004974.4	-	-
KCNC3	NM_004977.3	-	CNV in exon 4 may not be detected or reported
KCND3	NM_004980.4	-	-
KCNJ10	NM_002241.5	-	-
KCNJ2	NM_000891.3	-	-
KCTD7	NM_153033.4	-	-
KDM5C	NM_004187.4	-	-
KIDINS220	NM_020738.4	-	-

Targeted Genes and Methodology Details for Neurologic Disorders Custom Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
KIF1A	NM_004321.7	-	-
KIF1C	NM_006612.6	-	-
KIF5A	NM_004984.4	-	-
KLHL40	NM_152393.4	-	-
KLHL41	NM_006063.3	-	-
KY	NM_178554.6	-	-
L1CAM	NM_000425.5	c.2432-19A>C	-
L2HGDH	NM_024884.3	-	CNV in exon 6 may not be detected or reported
LAMA1	NM_005559.4	-	-
LAMA2	NM_000426.3	c.3556-13T>A	-
LAMB2	NM_002292.4	-	-
LAMP2	NM_013995.2	-	-
LARGE1	NM_004737.6	-	-
LDB3	NM_001080116.1	-	-
LDHA	NM_005566.4	-	-
LITAF	NM_004862.3	-	-
LMNA	NM_170707.4	c.1698+13C>A	-
LMOD3	NM_198271.4	-	-
LPIN1	NM_145693.4	-	-
LRP10	NM_014045.5	-	-
LRP4	NM_002334.4	-	-
LRRK2	NM_198578.4	-	-
LRSAM1	NM_138361.5	-	-
LYRM7	NM_181705.4	-	CNV in exon 3 may not be detected or reported
LYST	NM_000081.4	-	-
LYZ	NM_000239.3	-	-
MAG	NM_002361.4	-	-
MAP3K20	NM_016653.3	-	-
MAPT	NM_005910.5	c.823-15T>C c.915+11 to c.915+24	-
MARS1	NM_004990.4	-	-
MARS2	NM_138395.4	-	-
MATR3	NM_199189.2	-	Duplication analysis for CNV will not be performed
MB	NM_005368.3	-	-
MCM3AP	NM_003906.5	-	-
MECR	NM_016011.5	-	-
MED17	NM_004268.5	-	-
MEGF10	NM_032446.3	-	-
MFN2	NM_014874.4	-	-
MGME1	NM_052865.4	-	-

Targeted Genes and Methodology Details for Neurologic Disorders Custom Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
MICU1	NM_006077.3	-	-
MLC1	NM_015166.3	-	-
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	-	-
MRE11	NM_005591.3	-	-
MRPS25	NM_022497.5	-	-
MSTO1	NM_018116.3	-	Sequence variants and CNV in exons 1–7, 13 and 14 will not be detected or reported
MTFMT	NM_139242.4	-	-
MTM1	NM_000252.2	-	-
MTMR2	NM_016156.5	-	-
MTO1	NM_012123.4	-	-
MTTP	NM_000253.3	-	-
MUSK	NM_005592.4	-	-
MYBPC1	NM_002465.4	-	-
MYH14	NM_024729.3	-	-
MYH2	NM_017534.6	-	-
MYH7	NM_000257.4	-	CNV in exon 27 may not be detected or reported
MYMK	NM_001080483.3	-	-
MYOT	NM_006790.3	-	-
MYPN	NM_032578.3	-	-
NAGLU	NM_000263.4	-	-
NDRG1	NM_006096.4	-	-
NDUFAF2	NM_174889.5	-	-
NDUFAF6	NM_152416.4	c.420+784C>T	-
NDUFS1	NM_005006.7	-	-
NDUFS7	NM_024407.5	-	-
NDUFV1	NM_007103.4	-	-
NEB	NM_001271208.2	-	Sequence variants and CNV in exons 82–105 will not be detected or reported
NEFH	NM_021076.4	-	-
NEFL	NM_006158.4	-	-
NEU1	NM_000434.4	-	-
NF2	NM_000268.3	-	-
NGF	NM_002506.3	-	-
NHLRC1	NM_198586.3	-	-
NIPA1	NM_144599.5	-	-

Targeted Genes and Methodology Details for Neurologic Disorders Custom Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
NKX6-2	NM_177400.3	-	-
NOTCH3	NM_000435.3	c.341-26_341-24del	-
NPC1	NM_000271.5	c.882-28A>T/G c.1554-1009G>A c.3591+105A>T c.3591+121C>T	-
NPC2	NM_006432.4	-	-
NT5C2	NM_012229.4	-	-
NTRK1	NM_001012331.1	c.851-33T>A	-
NUBPL	NM_025152.3	c.815-27T>C	-
OPA1	NM_015560.2	c.625-5459G>A	-
OPA3	NM_025136.4	-	-
OPTN	NM_021980.4	-	-
ORAI1	NM_032790.3	-	-
OTC	NM_000531.6	-	-
PANK2	NM_153638.3	-	-
PARK7	NM_007262.5	-	CNV in exon 4 may not be detected or reported
PAX7	NM_002584.3	-	-
PCNA	NM_002592.2	-	-
PDE10A	NM_001130690.2	-	CNV in exons 1 and 10 may not be detected or reported
PDE8B	NM_003719.4	-	-
PDGFB	NM_002608.4	-	-
PDGFRB	NM_002609.4	-	-
PDHA1	NM_000284.4	c.419-17_419-14del c.511-30G>A c.759+26G>A	-
PDHX	NM_003477.3	c.816+11C>G	-
PDK3	NM_001142386.3	-	-
PDSS1	NM_014317.5	-	CNV in exons 2–3 may not be detected or reported
PDSS2	NM_020381.4	-	CNV in exon 7 may not be detected or reported
PDYN	NM_024411.5	-	-
PEX1	NM_000466.3	-	-
PEX10	NM_153818.1	-	-
PEX16	NM_004813.3	-	-
PEX7	NM_000288.4	c.-45C>T	-
PFKM	NM_000289.6	-	-
PFN1	NM_005022.4	-	-
PGAM2	NM_000290.4	-	-
PGAP1	NM_024989.4	-	CNV in exons 9 and 17 may not be detected or reported
PGK1	NM_000291.4	-	-

Targeted Genes and Methodology Details for Neurologic Disorders Custom Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
PGM1	NM_002633.3	c.1145-222G>T c.1600-523G>A	-
PHKA1	NM_002637.4	-	-
PHYH	NM_006214.4	-	-
PINK1	NM_032409.3	-	-
PLA2G6	NM_003560.4	-	-
PLD3	NM_012268.4	-	-
PLEC	NM_000445.5	-	-
PLEC	NM_201378.4	-	-
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	-
PMM2	NM_000303.3	c.179-25A>G c.640-15479C>T c.640-23A>G	-
PMP2	NM_002677.5	-	-
PMP22	NM_000304.4	-	-
PMPCA	NM_015160.3	-	-
PNKP	NM_007254.4	c.1386+49_1387-33del	-
PNP	NM_000270.3	-	-
PNPLA2	NM_020376.4	-	-
PNPLA6	NM_006702.5	-	-
PNPLA8	NM_015723.5	-	-
PODXL	NM_005397.4	-	-
POGLUT1	NM_152305.3	-	CNV in exon 10 may not be detected or reported
POLG	NM_002693.2	-	-
POLG2	NM_007215.4	-	-
POLR1C	NM_203290.4	-	-
POLR3A	NM_007055.4	c.1909+18G>A c.1909+22G>A c.3337-11T>C c.*18C>T	-
POLR3B	NM_018082.6	-	-
POMGNT1	NM_017739.3	-	-
POMGNT2	NM_032806.6	-	-
POMK	NM_032237.5	-	-
POMT1	NM_007171.3	-	-
POMT2	NM_013382.5	c.1333-14G>A	-
PPOX	NM_000309.5	-	-
PRDM12	NM_021619.3	-	-

Targeted Genes and Methodology Details for Neurologic Disorders Custom Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
PREPL	NM_006036.4	-	-
PRF1	NM_001083116.3	-	-
PRKAG2	NM_016203.4	-	CNV in exon 13 may not be detected or reported
PRKAR1B	NM_001164761.1	-	-
PRKCG	NM_002739.5	-	-
PRKN	NM_004562.3	-	-
PRKRA	NM_003690.5	-	CNV analysis will not be performed
PRNP	NM_000311.5	-	-
PRPH	NM_006262.4	-	-
PRPS1	NM_002764.4	-	-
PRRT2	NM_145239.3	c.880-34G>A	-
PRUNE1	NM_021222.3	-	-
PRX	NM_181882.3	-	-
PSAP	NM_002778.4	c.777+1915C>A	-
PSEN1	NM_000021.4	-	-
PSEN2	NM_000447.3	-	-
PTRH2	NM_016077.4	-	-
PTRHD1	NM_001013663.1	-	-
PUM1	NM_001020658.2	-	-
PUS1	NM_025215.6	-	-
PYCR2	NM_013328.4	-	-
PYGM	NM_005609.4	c.425-26A>G	-
PYROXD1	NM_024854.5	-	-
RAB18	NM_021252.5	-	-
RAB29	NM_003929.3	-	-
RAB39B	NM_171998.4	-	-
RAB3GAP1	NM_012233.3	-	-
RAB3GAP2	NM_012414.4	-	CNV in exon 17 may not be detected or reported
RAB7A	NM_004637.6	-	-
RAPSN	NM_005055.5	c.-210A>G c.-199C>G	-
RARS1	NM_002887.4	-	-
RBCK1	NM_031229.4	-	-
REEP1	NM_022912.3	-	-
REEP2	NM_001271803.2	-	-
RETREG1	NM_001034850.2	-	-
RIC3	NM_001206671.4	-	-
RNASEH1	NM_002936.5	-	-
RNASEH2A	NM_006397.2	-	-

Targeted Genes and Methodology Details for Neurologic Disorders Custom Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
RNASEH2	NM_024570.4	-	CNV in exon 8 may not be detected or reported
RNASEH2C	NM_032193.4	-	-
RNF170	NM_001160223.1	-	-
RNF216	NM_207111.4	-	-
RPGRIP1L	NM_015272.5	-	CNV in exon 23 may not be detected or reported
RRM2B	NM_015713.5	-	-
RTN2	NM_005619.5	-	-
RUBCN	NM_001145642.4	-	-
RXYLT1	NM_014254.3	-	-
RYR1	NM_000540.3	-	-
SACS	NM_014363.6	-	-
SAMD9L	NM_152703.5	-	-
SAMHD1	NM_015474.3	-	-
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	-	-
SCN2A	NM_021007.3	-	-
SCN4A	NM_000334.4	-	-
SCN8A	NM_014191.4	-	-
SCN8A	NM_001330260.2	-	-
SCN9A	NM_002977.3	-	-
SCO2	NM_005138.2	-	-
SCYL1	NM_020680.4	-	-
SDHA	NM_004168.4	-	-
SDHAF1	NM_001042631.2	-	-
SELENON	NM_020451.3	c.-71 to c.-11	CNV in exon 3 may not be detected or reported
SEPTIN9	NM_006640.4	c.-134G>C	-
SERAC1	NM_032861.4	-	CNV in exon 3 may not be detected or reported
SETX	NM_015046.7	-	-
SGCA	NM_000023.4	-	-
SGCB	NM_000232.4	-	-
SGCD	NM_000337.5	-	-
SGCG	NM_000231.2	-	-
SH3TC2	NM_024577.4	-	-
SIGMAR1	NM_005866.4	-	-
SIL1	NM_022464.5	-	-

Targeted Genes and Methodology Details for Neurologic Disorders Custom Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
SLC12A6	NM_133647.1	-	CNV in exon 3 may not be detected or reported
SLC16A2	NM_006517.5	-	-
SLC17A5	NM_012434.5	-	-
SLC18A2	NM_003054.6	-	-
SLC18A3	NM_003055.3	-	-
SLC19A3	NM_025243.4	c.980-14A>G	-
SLC1A3	NM_004172.5	-	-
SLC20A2	NM_006749.5	-	-
SLC22A5	NM_003060.4	c.-149G>A c.394-16T>A	-
SLC25A1	NM_005984.5	-	-
SLC25A19	NM_021734.4	-	-
SLC25A20	NM_000387.6	-	-
SLC25A3	NM_005888.3	-	-
SLC25A4	NM_001151.4	-	-
SLC25A42	NM_178526.5	-	-
SLC25A46	NM_138773.4	-	-
SLC2A1	NM_006516.3	c.680-11G>A c.679+25_680-43del	-
SLC30A10	NM_018713.2	-	-
SLC33A1	NM_004733.4	-	-
SLC39A14	NM_015359.6	-	-
SLC44A1	NM_080546.5	-	-
SLC52A2	NM_024531.5	-	-
SLC52A3	NM_033409.4	-	-
SLC5A7	NM_021815.5	-	-
SLC6A19	NM_001003841.3	-	-
SLC6A3	NM_001044.5	-	-
SLC6A8	NM_005629.4	-	-
SLC9A1	NM_003047.5	-	-
SLC9A6	NM_006359.3	-	-
SMCHD1	NM_015295.2	-	-
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
SNAP29	NM_004782.4	-	-
SNCA	NM_000345.4	-	-
SNCB	NM_001001502.3	-	-
SNX14	NM_153816.6	-	-
SOD1	NM_000454.4	c.358-11A>G	-

Targeted Genes and Methodology Details for Neurologic Disorders Custom Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
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	-	-
SOX2	NM_003106.4	-	-
SPART	NM_015087.5	-	-
SPAST	NM_014946.3	-	-
SPEG	NM_005876.5	-	-
SPG11	NM_025137.4	-	-
SPG21	NM_016630.7	-	-
SPG7	NM_003119.4	-	-
SPR	NM_003124.5	-	-
SPTAN1	NM_001130438.3	-	-
SPTBN2	NM_006946.3	-	-
SPTLC1	NM_006415.4	-	-
SPTLC2	NM_004863.3	-	-
SQSTM1	NM_003900.5	-	-
SRD5A3	NM_024592.5	-	-
STAC3	NM_145064.3	-	-
STIM1	NM_003156.3	-	-
STUB1	NM_005861.4	-	-
SUCLA2	NM_003850.2	-	-
SUCLG1	NM_003849.4	-	-
SUMF1	NM_182760.4	-	-
SUN1	NM_001130965.3	-	-
SUN2	NM_015374.3	-	-
SURF1	NM_003172.4	-	-
SYNE1	NM_033071.3	c.15705-12A>G	-
SYNJ1	NM_003895.3	-	-
SYT2	NM_177402.5	-	-
TACO1	NM_016360.4	-	-
TAF1	NM_004606.4	-	-
TAF15	NM_139215.3	-	-
TANGO2	NM_152906.7	-	-
TARDBP	NM_007375.3	-	-
TAZ (TAFAZZIN)	NM_000116.5	c.284+110G>A	-
TBC1D20	NM_144628.4	-	-
TBCD	NM_005993.5	c.1564-12C>G	-
TBK1	NM_013254.4	-	CNV in exon 16 may not be detected or reported
TBP	NM_003194.5	-	-

Targeted Genes and Methodology Details for Neurologic Disorders Custom Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
TCAP	NM_003673.4	-	-
TDP1	NM_018319.4	-	-
TDP2	NM_016614.3	-	-
TECPR2	NM_014844.5	-	-
TENM4	NM_001098816.3	-	-
TFG	NM_006070.6	-	-
TGM6	NM_198994.3	-	-
TH	NM_199292.3	c.-71C>T c.-70G>A c.1198-24T>A	-
THAP1	NM_018105.3	-	-
THG1L	NM_017872.5	-	-
TIA1	NM_022173.4	-	-
TIMM8A	NM_004085.4	-	-
TK2	NM_004614.5	-	-
TMEM216	NM_001173990.3	-	-
TMEM230	NM_001009923.2	-	CNV in exon 2 may not be detected or reported
TMEM240	NM_001114748.2	-	-
TMEM43	NM_024334.2	-	-
TMEM65	NM_194291.2	-	-
TMEM67	NM_153704.6	-	-
TMEM70	NM_017866.6	-	-
TNNT1	NM_003283.6	-	-
TNPO3	NM_012470.3	-	-
TOR1A	NM_000113.3	-	-
TOR1AIP1	NM_001267578.1	-	-
TPK1	NM_022445.4	-	-
TPM2	NM_003289.3	-	-
TPM3	NM_152263.4	-	-
TPP1	NM_000391.4	c.887-18A>G	-
TRAPPC11	NM_021942.6	-	-
TREM2	NM_018965.4	-	-
TREX1	NM_033629.6	-	-
TRIM2	NM_001130067.2	-	-
TRIM32	NM_012210.3	-	-
TRIP4	NM_016213.5	-	-
TRPV4	NM_021625.5	-	-
TSFM	NM_001172696.2	-	CNV in exon 5 may not be detected or reported
TTBK2	NM_173500.4	-	-
TTC19	NM_017775.4	-	-

Targeted Genes and Methodology Details for Neurologic Disorders Custom Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
TTN	NM_133378.4	-	Sequence variants and CNV in exons 153–155 will not be detected or reported
TTPA	NM_000370.3	-	-
TTR	NM_000371.3	-	-
TUBA4A	NM_006000.3	-	-
TUBB3	NM_006086.4	-	-
TUBB4A	NM_006087.4	-	-
TWNK	NM_021830.5	-	-
TYMP	NM_001953.5	-	-
TYROBP	NM_003332.4	-	-
UBA1	NM_003334.4	-	-
UBA5	NM_024818.4	-	CNV in exon 5 may not be detected or reported
UBAP1	NM_001171201.1	-	-
UBQLN2	NM_013444.3	-	-
UCHL1	NM_004181.5	-	-
UNC13A	NM_001080421.2	-	-
VAMP1	NM_014231.5	-	-
VAPB	NM_004738.5	-	-
VARS2	NM_001167734.1	-	-
VCP	NM_007126.5	-	-
VLDLR	NM_003383.5	-	-
VMA21	NM_001017980.3	c.54-27A>C/T c.54-16_54-8del	
VPS13A	NM_033305.3	-	-
VPS13C	NM_020821.3	-	CNV in exons 1–2 and 12 may not be detected or reported
VPS13D	NM_015378.4	-	-
VPS35	NM_018206.6	-	-
VRK1	NM_003384.3	-	-
VWA1	NM_022834.5	-	-
WARS1	NM_004184.4	-	-
WASHC5	NM_014846.4	-	-
WDR45	NM_007075.3	-	-
WDR73	NM_032856.4	-	-
WDR81	NM_001163809.1	-	-
WFS1	NM_006005.3	-	-
WNK1	NM_213655.4	-	-
XPR1	NM_004736.4	-	-
YARS1	NM_003680.3	-	-
YARS2	NM_001040436.3	-	-
ZFYVE26	NM_015346.4	-	-
ZFYVE27	NM_001002261.3	-	-

Targeted Genes and Methodology Details for Neurologic Disorders Custom Gene Panel (continued)

Available Neurologic Disorders Panels

Test ID	Test Name	Genes
AFTDP	Inherited Frontotemporal Dementia and ALS Gene Panel	*ALS2, ANG, ANXA11, APP, ASAH1, C9orf72, CCNF, CHCHD10, CHMP2B, CSF1R, DCTN1, ERBB4, FIG4, FUS, GRN, HEXB, HNRNPA1, HNRNPA2B1, ITM2B, KIF5A, MAPT, MATR3, NEFH, NOTCH3, NPC1, NPC2, OPTN, PANK2, PFN1, PRNP, PSEN1, PSEN2, SETX, SIGMAR1, SNCA, SOD1, SPG11, SPTLC1, SQSTM1, TAF15, TARDBP, TBK1, TBP, TIA1, TIMM8A, TREM2, TUBA4A, TYROBP, UBQLN2, VAPB, VCP, VRK1*
ATAXP	Inherited Ataxia Gene Panel	*AAAS, ABCB7, ABHD12, ACO2, ADPRHL2, AFG3L2, ALDH5A1, ALG6, ALS2, ANO10, APOPT1, APTX, ARSA, ATCAY, ATM, ATP1A3, ATP8A2, AUH, C12orf65, C19orf12, CA8, CACNA1A, CACNA1G, CAMTA1, CAPN1, CC2D2A, CCDC88C, CHCHD10, CLCN2, CLN5, CLPP, COQ2, COQ8A, COX20, CP, CTBP1, CWF19L1, CYP27A1, DARS2, DLD, DNAJC19, DNAJC3, DNMT1, EBF3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ELOVL5, EPM2A, FA2H, FBXL4, FGF14, FLVCR1, FMR1, FOLR1, FXN, GAMT, GBA2, GCDH, GDAP2, GFAP, GJC2, GOSR2, GRID2, GRM1, HEPACAM, HEXA, HIBCH, ITM2B, ITPR1, KCNA1, KCNA2, KCNC3, KCND3, KCNJ10, KCTD7, KIF1C, KIF5A, L2HGDH, LAMA1, LYRM7, MARS2, MECR, MLC1, MPV17, MRE11, MSTO1, MTFMT, MTO1, MTTP, NDUFAF2, NDUFAF6, NDUFS1, NDUFS7, NDUFV1, NEU1, NKX6-2, NOTCH3, NUBPL, OPA1, OPA3, OTC, PANK2, PCNA, PDHA1, PDSS2, PDYN, PEX10, PEX7, PHYH, PLA2G6, PLD3, PLP1, PMM2, PMPCA, PNKP, PNPLA6, POLG, POLR1C, POLR3A, POLR3B, PRKCG, PRNP, PRRT2, PSAP, PTRH2, PUM1, RARS1, RNASEH1, RNF170, RNF216, RPGRIP1L, RRM2B, RUBCN, SACS, SAMD9L, SCN2A, SCN8A, SCYL1, SDHA, SDHAF1, SETX, SIL1, SLC16A2, SLC17A5, SLC19A3, SLC1A3, SLC25A3, SLC25A46, SLC2A1, SLC44A1, SLC52A2, SLC52A3, SLC6A19, SLC9A1, SLC9A6, SNX14, SPAST, SPG11, SPG7, SPR, SPTBN2, SQSTM1, SRD5A3, STUB1, SUMF1, SURF1, SYNE1, TACO1, TBP, TDP1, TDP2, TGM6, THG1L, TIMM8A, TMEM216, TMEM240, TMEM67, TMEM70, TPK1, TPP1, TSFM, TTBK2, TTC19, TTPA, TUBB4A, TWNK, UBA5, VAMP1, VARS2, VLDLR, VPS13D, WDR73, WDR81, WFS1*
CMSP	Inherited Congenital Myasthenic Syndrome Gene Panel	*AGRN, ALG14, ALG2, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, COL13A1, COLQ, DNM2, DOK7, DPAGT1, GAA, GFPT1, GMPPB, LAMB2, LRP4, MUSK, PLEC, PREPL, RAPSN, SCN4A, SLC18A3, SLC25A1, SLC5A7, SYT2, VAMP1*
DMDZ	DMD Gene, Full Gene Analysis	*DMD*
DWPAN	Comprehensive Distal Weakness Gene Panel	*AAAS, AARS1, ABCA1, ABCD1, ACTA1, AIFM1, ALDH18A1, AMACR, ANO5, AP5Z1, APOA1, APTX, ARSA, ATL1, ATL3, ATM, ATP1A1, ATP7A, B4GALNT1, BAG3, BICD2, BIN1, BSCL2, C12orf65, C1orf194, CAV3, CHCHD10, CLCF1, CLTCL1, CNTNAP1, COQ4, COQ7, COX10, COX20, COX6A1, CPOX, CRLF1, CRYAB, CTDP1, CYP27A1, CYP2U1, CYP7B1, DCTN1, DDHD1, DES, DGAT2, DHH, DNAJB2, DNAJB6, DNM2, DNMT1, DST, DYNC1H1, DYSF, EGR2, ELP1, ERCC8, FA2H, FAM126A, FBLN5, FBXO38, FDX2, FGD4, FGF14, FHL1, FIG4, FLNC, FLVCR1, FMR1, FXN, GALC, GAN, GARS1, GBA2, GBE1, GBF1, GDAP1, GJB1, GLA, GM2A, GNB4, GNE, GSN, HADHA, HADHB, HARS1, HEXA, HEXB, HINT1, HK1, HMBS, HNRNPA1, HNRNPA2B1, HSPB1, HSPB8, HSPD1, IARS2, IBA57, IGHMBP2, INF2, KARS1, KIF1A, KIF5A, LAMA2, LDB3, LITAF, LMNA, LRSAM1, MARS1, MATR3, MCM3AP, MFN2, MME, MORC2, MPC1, MPV17, MPZ, MTMR2, MTTP, MYH14, MYH2, MYH7, MYOT, NAGLU, NDRG1, NEB, NEFH, NEFL, NF2, NGF, NIPA1, NTRK1, OPA1, PDK3, PDYN, PEX7, PHYH, PLA2G6, PLEKHG5, PLP1, PMP2, PMP22, PNKP, PNPLA6, POLG, PPOX, PRDM12, PRKCG, PRNP, PRPS1, PRX, PTRH2, RAB7A, REEP1, RETREG1, RNASEH1, RRM2B, RTN2, SACS, SBF1, SBF2, SCN10A, SCN11A, SCN9A, SCO2, SELENON, SEPTIN9, SETX, SH3TC2, SIGMAR1, SLC12A6, SLC25A19, SLC25A46, SLC52A2, SLC52A3, SLC5A7, SMN1, SMN2, SNAP29, SOD1, SORD, SOX10, SPAST, SPG11, SPG21, SPG7, SPTAN1, SPTLC1, SPTLC2, SQSTM1, SUCLA2, SURF1, TDP1, TFG, TIA1, TRIM2, TRPV4, TSFM, TTN, TTPA, TTR, TUBB3, TWNK, TYMP, UBA1, VCP, VPS13D, VRK1, VWA1, WARS1, WASHC5, WNK1, YARS1, ZFYVE26*
EDMDP	Inherited Emery-Dreifuss Gene Panel	*EMD, FHL1, LMNA, SUN1, SUN2, SYNE1, TMEM43*
IMNP	Inherited Motor Neuropathy Gene Panel	*ATP7A, BICD2, BSCL2, CPOX, DCTN1, DNAJB2, FBXO38, GARS1, GBF1, HINT1, HSPB1, HSPB8, IGHMBP2, PLEKHG5, SETX, SIGMAR1, SLC5A7, SMN1, SMN2, SOD1, SORD, SPTAN1, TRPV4, VRK1, VWA1, WARS1*

Targeted Genes and Methodology Details for Neurologic Disorders Custom Gene Panel (continued)

Available Neurologic Disorders Panels (continued)

Test ID	Test Name	Genes
IMSNP	Inherited Motor and Sensory Neuropathy Gene Panel	*AARS1, ABCD1, AIFM1, ARSA, ATP1A1, ATP7A, BAG3, BSCL2, C1orf194, CHCHD10, CNTNAP1, COX10, COX6A1, CTDP1, DGAT2, DHH, DNM2, DYNC1H1, EGR2, ERCC8, FAM126A, FBLN5, FGD4, FIG4, FMR1, GALC, GAN, GARS1, GBF1, GDAP1, GJB1, GLA, GNB4, HARS1, HINT1, HK1, HSPB1, HSPB8, IGHMBP2, INF2, KARS1, LAMA2, LITAF, LMNA, LRSAM1, MARS1, MCM3AP, MFN2, MORC2, MPV17, MPZ, MTMR2, NDRG1, NEFH, NEFL, PEX7, PHYH, PLEKHG5, PLP1, PMP2, PMP22, PNKP, POLG, PRPS1, PRX, RAB7A, SACS, SBF1, SBF2, SCO2, SH3TC2, SLC12A6, SLC25A46, SORD, SOX10, SURF1, TDP1, TFG, TRIM2, TRPV4, TSFM, TTR, TUBB3, TWNK, TYMP, VPS13D, YARS1*
ISNP	Inherited Sensory Neuropathy Gene Panel	*AIFM1, ATL1, ATL3, CLCF1, CLTCL1, COX20, CRLF1, DNMT1, DST, DST, ELP1, GLA, KIF1A, NGF, NTRK1, PRDM12, PRKCG, RETREG1, SCN10A, SCN11A, SCN9A, SPTLC1, SPTLC2, WNK1*
ISPP	Inherited Spastic Paraplegia Gene Panel	*ABCD1, ACO2, AFG3L2, ALDH18A1, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APOPT1, ARG1, ARL6IP1, ASNS, ATL1, AUH, B4GALNT1, BICD2, BOLA3, BSCL2, C12orf65, COQ7, CPT1C, CTC1, CTSD, CYP27A1, CYP2U1, CYP7B1, DARS1, DARS2, DDHD1, DDHD2, DLD, EARS2, ENTPD1, ERLIN1, ERLIN2, EXOSC3, FA2H, FAR1, FARS2, FUCA1, FXN, GALC, GBA2, GBE1, GJC2, GLB1, GM2A, GPT2, HACE1, HEXA, HIBCH, HSPD1, HTRA1, IBA57, IFIH1, IRF2BPL, ISCA2, KDM5C, KIDINS220, KIF1A, KIF5A, L1CAM, L2HGDH, LYRM7, MAG, MARS2, MED17, MTFMT, NIPA1, NT5C2, NUBPL, OPA3, PANK2, PDHX, PEX16, PGAP1, PLA2G6, PLP1, PNP, PNPLA6, PNPLA8, PRF1, PRUNE1, PSAP, PYCR2, RAB18, RAB3GAP1, RAB3GAP2, RARS1, REEP1, REEP2, RNASEH2A, RNASEH2B, RNASEH2C, RTN2, SACS, SAMHD1, SDHAF1, SERAC1, SLC12A6, SLC16A2, SLC19A3, SLC33A1, SLC6A8, SOX2, SPART, SPAST, SPG11, SPG21, SPG7, SPTAN1, SUMF1, TACO1, TBC1D20, TECPR2, TFG, TREX1, TTC19, TYROBP, UBAP1, UBQLN2, VAMP1, VPS13D, WASHC5, ZFYVE26, ZFYVE27*
LGCMP	Inherited Limb-Girdle Muscular Dystrophy and Congenital Myasthenic Syndrome Gene Panel	*AGRN, ALG14, ALG2, ANO5, BIN1, BVES, CAPN3, CAV3, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, COL13A1, COL6A1, COL6A2, COL6A3, COLQ, CRPPA, DAG1, DES, DNAJB6, DNM2, DOK7, DPAGT1, DPM3, DYSF, FKRP, FKTN, GAA, GFPT1, GMPPB, HNRNPDL, LAMA2, LAMB2, LMNA, LRP4, MUSK, MYOT, PLEC, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PREPL, RAPSN, SCN4A, SGCA, SGCB, SGCD, SGCG, SLC18A3, SLC25A1, SLC5A7, SYT2, TCAP, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN, VAMP1, VCP*
MDYSP	Inherited Muscular Dystrophy Gene Panel	*ACTA1, ANO5, B3GALNT2, B4GAT1, BAG3, BIN1, BVES, CAPN3, CAV3, CAVIN1, CHKB, COL12A1, COL6A1, COL6A2, COL6A3, CRPPA, CRYAB, DAG1, DES, DMD, DNAJB6, DNM2, DPM1, DPM2, DPM3, DYSF, EMD, FHL1, FKRP, FKTN, FLNC, GAA, GMPPB, GNE, GOSR2, HNRNPA1, HNRNPA2B1, HNRNPDL, INPP5K, ITGA7, JAG2, LAMA2, LARGE1, LDB3, LMNA, MATR3, MSTO1, MYH7, MYOT, PLEC, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1, SELENON, SGCA, SGCB, SGCD, SGCG, SQSTM1, SUN1, SUN2, SYNE1, TCAP, TIA1, TMEM43, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN, VCP*
MNDP	Inherited Motor Neuron Disease Gene Panel	*ALS2, ANG, ANXA11, ASAH1, C9orf72, CCNF, CHCHD10, CHMP2B, DCTN1, ERBB4, FIG4, FUS, HEXB, HNRNPA1, HNRNPA2B1, KIF5A, MATR3, NEFH, OPTN, PFN1, SETX, SIGMAR1, SOD1, SPG11, SPTLC1, SQSTM1, TAF15, TARDBP, TBK1, TIA1, TUBA4A, UBQLN2, VAPB, VCP, VRK1*
MUPAN	Comprehensive Neuromuscular Gene Panel	*ABHD5, ACAD9, ACADM, ACADS, ACADVL, ACTA1, ADSS1, AGK, AGL, AGRN, ALDOA, ALG14, ALG2, ANO5, ASAH1, ASCC1, ATP1A2, ATP2A1, B3GALNT2, B4GAT1, BAG3, BIN1, BVES, CACNA1S, CAPN3, CASQ1, CAV3, CAVIN1, CFL2, CHAT, CHCHD10, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CLCN1, COL12A1, COL13A1, COL6A1, COL6A2, COL6A3, COLQ, COQ2, COQ4, COQ6, COQ8A, COQ9, CPT2, CRPPA, CRYAB, CTDP1, DAG1, DES, DGUOK, DMD, DNA2, DNAJB6, DNM2, DOK7, DPAGT1, DPM1, DPM2, DPM3, DYSF, EMD, ENO3, ETFA, ETFB, ETFDH, FAM111B, FBXL4, FDX2, FHL1, FKBP14, FKRP, FKTN, FLAD1, FLNC, GAA, GBE1, GFER, GFPT1, GMPPB, GNE, GOSR2, GYG1, GYS1, HADHA, HADHB, HNRNPA1, HNRNPA2B1, HNRNPDL, HRAS, INPP5K, ISCU, ITGA7, JAG2, KBTBD13, KCNJ2, KLHL40, KLHL41, KY, LAMA2, LAMB2, LAMP2, LARGE1, LDB3, LDHA, LMNA, LMOD3, LPIN1, LRP4, MAP3K20, MATR3, MB, MEGF10, MGME1, MICU1, MRPS25, MSTO1, MTM1, MTO1, MUSK, MYBPC1, MYH2, MYH7, MYMK, MYOT, MYPN, NEB, OPA1, ORAI1, PAX7, PDSS1, PDSS2, PFKM, PGAM2, PGK1, PGM1, PHKA1, PLEC, PNPLA2, PNPLA8, POGLUT1, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PREPL, PRKAG2, PUS1, PYGM, PYROXD1, RAPSN, RBCK1, RNASEH1, RRM2B, RXYLT1, RYR1, SCN4A, SDHA, SELENON, SGCA, SGCB, SGCD, SGCG, SIL1, SLC18A3, SLC22A5, SLC25A1, SLC25A20, SLC25A3, SLC25A4, SLC25A42, SLC5A7, SMN1, SMN2, SPEG, SQSTM1, STAC3, STIM1, SUCLA2, SUCLG1, SUN1, SUN2, SYNE1, SYT2, TANGO2, TAZ, TBCD, TCAP, TFG, TIA1, TK2, TMEM43, TMEM65, TNNT1, TNPO3, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRIM32, TRIP4, TSFM, TTC19, TTN, TWNK, UBA1, VAMP1, VCP, VMA21, YARS2*

Targeted Genes and Methodology Details for Neurologic Disorders Custom Gene Panel (continued)

Available Neurologic Disorders Panels (continued)

Test ID	Test Name	Genes
PARDP	Inherited Parkinson Disease Gene Panel	*ADORA1, AHSA1, ANG, ANO3, APP, ATP13A2, ATP1A3, ATP6AP2, ATP7B, C19orf12, CHCHD2, CHMP2B, CLN3, CP, CSF1R, CYP27A1, DCAF17, DCTN1, DDC, DNAJB2, DNAJC12, DNAJC13, DNAJC6, DNM1L, EIF4G1, FBXO7, FTL, FUS, GBA, GCH1, GIGYF2, GRN, HTRA2, KIF5A, LRP10, LRRK2, LYST, MAPT, OPTN, PANK2, PARK7, PDE10A, PDE8B, PDGFB, PDGFRB, PEX1, PINK1, PLA2G6, PLD3, PODXL, POLG, POLG2, PRKAR1B, PRKN, PRKRA, PRRT2, PSEN1, PSEN2, PTRHD1, RAB29, RAB39B, RIC3, SIGMAR1, SLC18A2, SLC20A2, SLC30A10, SLC39A14, SLC6A3, SNCA, SNCB, SOD1, SPG11, SPR, SQSTM1, SYNJ1, TAF1, TAF15, TARDBP, TENM4, TH, THAP1, TMEM230, TOR1A, TUBA4A, TWNK, UBQLN2, UCHL1, UNC13A, VCP, VPS13A, VPS13C, VPS35, WDR45, XPR1*
PEPAN	Comprehensive Peripheral Neuropathy Gene Panel	*AAAS, AARS1, ABCA1, ABCD1, AIFM1, ALDH18A1, AMACR, AP5Z1, APOA1, APTX, ARSA, ATL1, ATL3, ATM, ATP1A1, ATP7A, B4GALNT1, BAG3, BICD2, BSCL2, C12orf65, C1orf194, CHCHD10, CLCF1, CLTCL1, CNTNAP1, COQ4, COQ7, COX10, COX20, COX6A1, CPOX, CRLF1, CTDP1, CYP27A1, CYP2U1, CYP7B1, DCTN1, DDHD1, DGAT2, DHH, DNAJB2, DNM2, DNMT1, DST, DYNC1H1, EGR2, ELP1, ERCC8, FA2H, FAM126A, FBLN5, FBXO38, FGD4, FGF14, FIG4, FLVCR1, FMR1, FXN, GALC, GAN, GARS1, GBA2, GBE1, GBF1, GDAP1, GJB1, GLA, GM2A, GNB4, GSN, HADHA, HADHB, HARS1, HEXA, HEXB, HINT1, HK1, HMBS, HSPB1, HSPB8, HSPD1, IARS2, IBA57, IGHMBP2, INF2, KARS1, KIF1A, KIF5A, LAMA2, LITAF, LMNA, LRSAM1, MARS1, MCM3AP, MFN2, MME, MORC2, MPC1, MPV17, MPZ, MTMR2, MTTP, MYH14, NAGLU, NDRG1, NEFH, NEFL, NF2, NGF, NIPA1, NTRK1, OPA1, PDK3, PDYN, PEX7, PHYH, PLA2G6, PLEKHG5, PLP1, PMP2, PMP22, PNKP, PNPLA6, POLG, PPOX, PRDM12, PRKCG, PRNP, PRPS1, PRX, PTRH2, RAB7A, REEP1, RETREG1, RNASEH1, RRM2B, RTN2, SACS, SBF1, SBF2, SCN10A, SCN11A, SCN9A, SCO2, SEPTIN9, SETX, SH3TC2, SIGMAR1, SLC12A6, SLC25A19, SLC25A46, SLC52A2, SLC52A3, SLC5A7, SMN1, SMN2, SNAP29, SOD1, SORD, SOX10, SPAST, SPG11, SPG21, SPG7, SPTAN1, SPTLC1, SPTLC2, SUCLA2, SURF1, TDP1, TFG, TRIM2, TRPV4, TSFM, TTPA, TTR, TUBB3, TWNK, TYMP, UBA1, VPS13D, VRK1, VWA1, WARS1, WASHC5, WNK1, YARS1, ZFYVE26*
RABMP	Inherited Rhabdomyolysis and Metabolic Myopathy Panel	*ABHD5, ACAD9, ACADM, ACADS, ACADVL, AGK, AGL, ALDOA, ANO5, ATP2A1, CASQ1, CAVIN1, CHCHD10, COQ2, COQ4, COQ6, COQ8A, COQ9, CPT2, CTDP1, DGUOK, DMD, DNA2, DYSF, ENO3, ETFA, ETFB, ETFDH, FBXL4, FDX2, FKRP, FKTN, FLAD1, GAA, GBE1, GFER, GYG1, GYS1, HADHA, HADHB, ISCU, LAMP2, LDHA, LPIN1, MGME1, MRPS25, MSTO1, OPA1, PDSS1, PDSS2, PFKM, PGAM2, PGK1, PGM1, PHKA1, PNPLA2, PNPLA8, POLG, POLG2, PRKAG2, PUS1, PYGM, RBCK1, RNASEH1, RRM2B, RYR1, SCN4A, SDHA, SLC22A5, SLC25A20, SLC25A4, SLC25A42, SUCLA2, SUCLG1, TANGO2, TK2, TMEM65, TRIM32, TSFM, TTC19, TWNK, VMA21, YARS2*
SEP9Z	SEPTIN9 Gene, Full Gene Analysis	*SEPTIN9*
SMCP	Inherited Skeletal Muscle Channelopathy Gene Panel	*ATP1A2, CACNA1S, CLCN1, KCNJ2, SCN4A*
SOD1Z	SOD1 Gene, Full Gene Analysis	*SOD1*
TTRZ	TTR Gene, Full Gene Analysis	*TTR*

Effective Date	Version	Synopsis of Test Change
12/20/2022	V2	Available Neurologic Disorders Panels, Test ID *PEPAN*: Added genes *HEXA* and *HEXB*
03/10/2023	V3	Updated *ATP7B* additional variants detected
08/07/2024	V4	Test ID AFTDP: Added gene SPTLC1 Test ID DWPAN: Added gene SEPTIN9 Test ID MNDP: Added gene SPTLC1 Test ID PEPAN: Added gene SEPTIN9