MAYO CLINIC LABORATORIES | *Targeted Genes and Methodology Details for Peripheral Neuropathy Gene Panels*

Reference transcripts based on build GRCh37 (hg19) interrogated by Peripheral Neuropathy Gene Panels

Peripheral Neuropathy Expanded Panel	
Gene	**GenBank Accession Number**
AAAS	NM_015665
AARS	NM_001605
ABCA1	NM_005502
ABCD1	NM_000033
ADCY6	NM_015270
AIFM1	NM_004208
AMACR	NM_014324
AP1S1	NM_001283
AP4B1	NM_006594
AP4E1	NM_007347
AP4M1	NM_004722
AP4S1	NM_007077
AP5Z1	NM_014855
APOA1	NM_000039
APTX	NM_175073
ARHGEF10	NM_014629
ARSA	NM_000487
ATL1	NM_015915
ATM	NM_000051
ATP7A	NM_000052
B2M	NM_004048
B4GALNT1	NM_001478
BAG3	NM_004281
BCKDHB	NM_183050
BICD2	NM_001003800
BSCL2	NM_032667
C12orf65	NM_152269
CCT5	NM_012073
CLCF1	NM_013246
CNTNAP1	NM_003632
COX10	NM_001303
CPOX	NM_000097
CRLF1	NM_004750
CTDP1	NM_004715
CTSA	NM_000308
CYP27A1	NM_000784
CYP2U1	NM_183075
CYP7B1	NM_004820

Peripheral Neuropathy Expanded Panel	
Gene	**GenBank Accession Number**
DARS2	NM_018122
DCAF8	NM_015726
DCTN1	NM_004082
DDHD1	NM_001160147
DDHD2	NM_015214
DGUOK	NM_080916
DHH	NM_021044
DHTKD1	NM_018706
DNAJB2	NM_001039550
DNM2	NM_001005360
DNMT1	NM_001130823
DST	NM_001723
DYNC1H1	NM_001376
EGR2	NM_000399
ERBB3	NM_001982
ERCC6	NM_000124
ERCC8	NM_000082
ERLIN2	NM_007175
FA2H	NM_024306
FAH	NM_000137
FAM126A	NM_032581
FAM134B	NM_001034850
FBLN5	NM_006329
FBXO38	NM_030793
FGD4	NM_139241
FGF14	NM_004115
FIG4	NM_014845
FLVCR1	NM_014053
FMR1	NM_002024
GALC	NM_000153
GAN	NM_022041
GARS	NM_002047
GBA2	NM_020944
GBE1	NM_000158
GDAP1	NM_018972
GJB1	NM_000166
GJB3	NM_024009
GJC2	NM_020435

Peripheral Neuropathy Expanded Panel	
Gene	**GenBank Accession Number**
GLA	NM_000169
GNB4	NM_021629
GSN	NM_000177
HADHA	NM_000182
HADHB	NM_000183
HARS	NM_002109
HINT1	NM_005340
HK1	NM_000188
HMBS	NM_000190
HSPB1	NM_001540
HSPB3	NM_006308
HSPB8	NM_014365
HSPD1	NM_002156
IGHMBP2	NM_002180
IKBKAP	NM_003640
INF2	NM_022489
KARS	NM_001130089
KIF1A	NM_004321
KIF1B	NM_015074
KIF5A	NM_004984
L1CAM	NM_000425
LAMA2	NM_000426
LITAF	NM_004862
LMNA	NM_170707
LRSAM1	NM_138361
LYST	NM_000081
MAF	NM_005360
MARS	NM_004990
MED25	NM_030973
MFN2	NM_014874
MMACHC	NM_015506
MPV17	NM_002437
MPZ	NM_000530
MTMR2	NM_016156
MTTP	NM_000253
MYH14	NM_024729
NAGA	NM_000262
NAGLU	NM_000263
NDRG1	NM_006096
NEFL	NM_006158
NF2	NM_000268
NGF	NM_002506
NIPA1	NM_144599

Peripheral Neuropathy Expanded Panel	
Gene	**GenBank Accession Number**
NTRK1	NM_002529
OAT	NM_000274
OPA1	NM_015560
PANK2	NM_153638
PDHA1	NM_000284
PDK3	NM_001142386
PDYN	NM_024411
PEX10	NM_153818
PEX7	NM_000288
PHYH	NM_006214
PLA2G6	NM_003560
PLEKHG5	NM_198681
PLOD1	NM_000302
PLP1	NM_000533
PMM2	NM_000303
PMP2	NM_002677
PMP22	NM_000304
PNKP	NM_007254
PNPLA6	NM_006702
POLG	NM_002693
PPOX	NM_000309
PRNP	NM_000311
PRPS1	NM_002764
PRX	NM_181882
RAB7A	NM_004637
REEP1	NM_022912
RRM2B	NM_015713
RTN2	NM_005619
SACS	NM_014363
SBF1	NM_002972
SBF2	NM_030962
SCN10A	NM_006514
SCN11A	NM_014139
SCN9A	NM_002977
SCO2	NM_005138
SCP2	NM_002979
SETX	NM_015046
SH3TC2	NM_024577
SLC12A6	NM_133647
SLC16A2	NM_006517
SLC25A19	NM_021734
SLC25A46	NM_138773
SLC33A1	NM_004733

Peripheral Neuropathy Expanded Panel	
Gene	**GenBank Accession Number**
SLC52A2	NM_024531
SLC5A7	NM_021815
SNAP29	NM_004782
SOD1	NM_000454
SOX10	NM_006941
SPAST	NM_014946
SPG11	NM_025137
SPG20	NM_015087
SPG21	NM_016630
SPG7	NM_003119
SPTLC1	NM_006415
SPTLC2	NM_004863
SURF1	NM_003172
TDP1	NM_018319
TECPR2	NM_014844
TFG	NM_006070
TRIM2	NM_015271
TRPA1	NM_007332
TRPV4	NM_021625
TTPA	NM_000370
TTR	NM_000371
TUBB3	NM_006086
TWNK	NM_021830
TYMP	NM_001953
VPS37A	NM_152415
WASHC5	NM_014846
WNK1	NM_018979
XPA	NM_000380
XPC	NM_004628
YARS	NM_003680
ZFYVE26	NM_015346

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of a mutation in these genes.

There are regions of the genes *CRLF1*, *DNMT1*, *GJC2*, *INF2*, *MAF*, and *PNKP* that cannot not be effectively amplified and sequenced as a result of technical limitations of the assay, including regions of homology, high GC-rich content, and repetitive sequences.

Additionally, NGS is used to test for the presence of large deletions and duplications in the *GDAP1*, *GLA*, *MFN2*, *MPZ*, *MTTP*, *PMP22*, *PNKP*, *POLG*, and *SPG7* genes.

Multiplex Ligation-Dependent Probe Amplification (MLPA), PCR, and/or Sanger sequencing is used to confirm alterations detected by NGS when appropriate. (Unpublished Mayo method)

Motor and Sensory Neuropathy Panel	
Gene	**GenBank Accession Number**
AARS	NM_001605
ABCD1	NM_000033
ADCY6	NM_015270
AIFM1	NM_004208
ARHGEF10	NM_014629
ARSA	NM_000487
ATP7A	NM_000052
CNTNAP1	NM_003632
COX10	NM_001303
CTDP1	NM_004715
DCAF8	NM_015726
DHH	NM_021044
DHTKD1	NM_018706
DNM2	NM_001005360
DYNC1H1	NM_001376
EGR2	NM_000399
ERBB3	NM_001982
ERCC6	NM_000124
ERCC8	NM_000082
FAM126A	NM_032581
FBLN5	NM_006329
FGD4	NM_139241
FIG4	NM_014845
FMR1	NM_002024
GALC	NM_000153
GAN	NM_022041
GARS	NM_002047
GDAP1	NM_018972
GJB1	NM_000166
GLA	NM_000169
GNB4	NM_021629
HARS	NM_002109
HINT1	NM_005340
HK1	NM_000188
HSPB1	NM_001540
HSPB8	NM_014365
HSPD1	NM_002156
IGHMBP2	NM_002180
INF2	NM_022489
KARS	NM_001130089
KIF1B	NM_015074
LAMA2	NM_000426
LITAF	NM_004862
LMNA	NM_170707
LRSAM1	NM_138361
MARS	NM_004990
MED25	NM_030973
MFN2	NM_014874

Motor and Sensory Neuropathy Panel	
Gene	**GenBank Accession Number**
MPZ	NM_000530
MTMR2	NM_016156
NDRG1	NM_006096
NEFL	NM_006158
PDHA1	NM_000284
PDK3	NM_001142386
PEX7	NM_000288
PHYH	NM_006214
PLEKHG5	NM_198681
PLP1	NM_000533
PMM2	NM_000303
PMP2	NM_002677
PMP22	NM_000304
POLG	NM_002693
PRNP	NM_000311
PRPS1	NM_002764
PRX	NM_181882
RAB7A	NM_004637
SACS	NM_014363
SBF1	NM_002972
SBF2	NM_030962
SH3TC2	NM_024577
SLC12A6	NM_133647
SLC25A46	NM_138773
SOX10	NM_006941
SURF1	NM_003172
TDP1	NM_018319
TFG	NM_006070
TRIM2	NM_015271
TRPV4	NM_021625
TTR	NM_000371
TUBB3	NM_006086
TYMP	NM_001953
YARS	NM_003680

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of a mutation in these genes.

There are regions of the gene *INF2* that cannot be effectively amplified and sequenced as a result of technical limitations of the assay, including regions of homology, high GC-rich content, or repetitive sequences may not provide accurate sequence.

Additionally, NGS is used to test for the presence of large deletions and duplications in the *GDAP1*, *GLA*, *MFN2*, *MPZ*, *PMP22*, and *POLG* genes.

Multiplex Ligation-Dependent Probe Amplification (MLPA), PCR, and/or Sanger sequencing is used to confirm alterations detected by NGS when appropriate. (Unpublished Mayo method)

Hereditary Sensory Neuropathy Panel	
Gene	**GenBank Accession Number**
ATL1	NM_015915
CCT5	NM_012073
CLCF1	NM_013246
CRLF1	NM_004750
DNMT1	NM_001130823
DST	NM_001723
FAM134B	NM_001034850
IKBKAP	NM_003640
KIF1A	NM_004321
NGF	NM_002506
NTRK1	NM_002529
SCN10A	NM_006514
SCN11A	NM_014139
SCN9A	NM_002977
SPTLC1	NM_006415
SPTLC2	NM_004863
TRPA1	NM_007332
WNK1	NM_018979

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of a mutation in these genes.

There are regions of the genes *CRLF1* and *DNMT1* that cannot be effectively amplified and sequenced as a result of technical limitations of the assay, including regions of homology, high GC-rich content, and repetitive sequences may not provide accurate sequence.

Sanger sequencing is used to confirm alterations detected by NGS when appropriate. (Unpublished Mayo method)

Hereditary Motor Neuropathy Panel	
Gene	**GenBank Accession Number**
ATP7A	NM_000052
BICD2	NM_001003800
BSCL2	NM_032667
DCTN1	NM_004082
DNAJB2	NM_001039550
DYNC1H1	NM_001376
FBLN5	NM_006329
FBXO38	NM_030793
GARS	NM_002047
GJB1	NM_000166
HARS	NM_002109
HSPB1	NM_001540
HSPB3	NM_006308
HSPB8	NM_014365
IGHMBP2	NM_002180
PDK3	NM_001142386
PLEKHG5	NM_198681
REEP1	NM_022912
SCP2	NM_002979
SETX	NM_015046
SLC5A7	NM_021815
SOD1	NM_000454
TRPV4	NM_021625

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of a mutation in these genes.

Regions of homology, high GC-rich content, and repetitive sequences may not provide accurate sequence. Therefore, all reported alterations detected by NGS are confirmed by an independent reference method based on laboratory developed criteria. However, this does not rule out the possibility of a false-negative result in these regions.

Sanger sequencing is used to confirm alterations detected by NGS when appropriate. (Unpublished Mayo method)

Spastic Paraplegia Neuropathy Panel	
Gene	**GenBank Accession Number**
ABCD1	NM_000033
AP4B1	NM_006594
AP4E1	NM_007347
AP4M1	NM_004722
AP4S1	NM_007077
AP5Z1	NM_014855
ATL1	NM_015915
B4GALNT1	NM_001478
BSCL2	NM_032667
C12orf65	NM_152269
CCT5	NM_012073
CYP2U1	NM_183075
CYP7B1	NM_004820
DDHD1	NM_001160147
DDHD2	NM_015214
ERLIN2	NM_007175
FA2H	NM_024306
GBA2	NM_020944
GJC2	NM_020435
HSPD1	NM_002156
KIF1A	NM_004321
KIF5A	NM_004984
L1CAM	NM_000425
NIPA1	NM_144599
PLP1	NM_000533
PNPLA6	NM_006702
REEP1	NM_022912
RTN2	NM_005619
SLC12A6	NM_133647
SLC16A2	NM_006517
SLC33A1	NM_004733
SPAST	NM_014946
SPG11	NM_025137
SPG20	NM_015087
SPG21	NM_016630
SPG7	NM_003119
TECPR2	NM_014844
TFG	NM_006070
VPS37A	NM_152415
WASHC5	NM_014846
ZFYVE26	NM_015346

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of a mutation in these genes.

There are regions of the gene *GJC2* that cannot be effectively amplified and sequenced as a result of technical limitations of the assay, including regions of homology, high GC-rich content, and repetitive sequences.

Additionally, NGS is used to test for the presence of large deletions and duplications in the *SPG7* gene.

PCR and/or Sanger sequencing is used to confirm alterations detected by NGS when appropriate. (Unpublished Mayo method)

Metabolic or Syndromic Neuropathies	
Gene	**GenBank Accession Number**
AAAS	NM_015665
ABCA1	NM_005502
ABCD1	NM_000033
AIFM1	NM_004208
AMACR	NM_014324
AP1S1	NM_001283
APOA1	NM_000039
APTX	NM_175073
ARSA	NM_000487
ATM	NM_000051
B2M	NM_004048
BAG3	NM_004281
BCKDHB	NM_183050
CPOX	NM_000097
CTDP1	NM_004715
CTSA	NM_000308
CYP27A1	NM_000784
DARS2	NM_018122
DGUOK	NM_080916
FAH	NM_000137
FBLN5	NM_006329
FGF14	NM_004115
FLVCR1	NM_014053
GALC	NM_000153
GAN	NM_022041
GBE1	NM_000158
GJB3	NM_024009
GLA	NM_000169
GSN	NM_000177
HADHA	NM_000182
HADHB	NM_000183
HMBS	NM_000190
L1CAM	NM_000425
LMNA	NM_170707
LYST	NM_000081
MAF	NM_005360
MMACHC	NM_015506
MPV17	NM_002437
MTTP	NM_000253
MYH14	NM_024729
NAGA	NM_000262
NAGLU	NM_000263
NF2	NM_000268

Metabolic or Syndromic Neuropathies	
Gene	**GenBank Accession Number**
OAT	NM_000274
OPA1	NM_015560
PANK2	NM_153638
PDHA1	NM_000284
PDYN	NM_024411
PEX10	NM_153818
PHYH	NM_006214
PLA2G6	NM_003560
PLOD1	NM_000302
PNKP	NM_007254
POLG	NM_002693
PPOX	NM_000309
PRNP	NM_000311
PRPS1	NM_002764
RRM2B	NM_015713
SACS	NM_014363
SCO2	NM_005138
SETX	NM_015046
SLC25A19	NM_021734
SLC52A2	NM_024531
SNAP29	NM_004782
SPG20	NM_015087
SPG7	NM_003119
TDP1	NM_018319
TTPA	NM_000370
TTR	NM_000371
TUBB3	NM_006086
TYMP	NM_001953
XPA	NM_000380
XPC	NM_004628

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of a mutation in these genes.

There are regions of the genes *MAF* and *PNKP* that cannot be effectively amplified and sequenced as a result of technical limitations of the assay, including regions of homology, high GC-rich content, and repetitive sequences.

Additionally, NGS is used to test for the presence of large deletions and duplications in the *GLA*, *MTTP*, *PNKP*, *POLG*, and *SPG7* genes.

PCR and/or Sanger sequencing is used to confirm alterations detected by NGS when appropriate. (Unpublished Mayo method)

SEPT9 Gene, Full Gene Analysis	
Gene	**GenBank Accession Number**
SEPT9	NM_006640

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of a mutation in these genes.

Regions of homology, high GC-rich content, and repetitive sequences may not provide accurate sequence. Therefore, all reported alterations detected by next generation sequencing are confirmed by an independent reference method based on laboratory developed criteria. However, this does not rule out the possibility of a false-negative result in these regions.

Additionally, NGS is used to test for the presence of large deletions and duplications in the *SEPT9* gene.

Multiplex Ligation-Dependent Probe Amplification (MLPA), PCR, and/or Sanger sequencing is used to confirm alterations detected by NGS when appropriate. (Unpublished Mayo method)