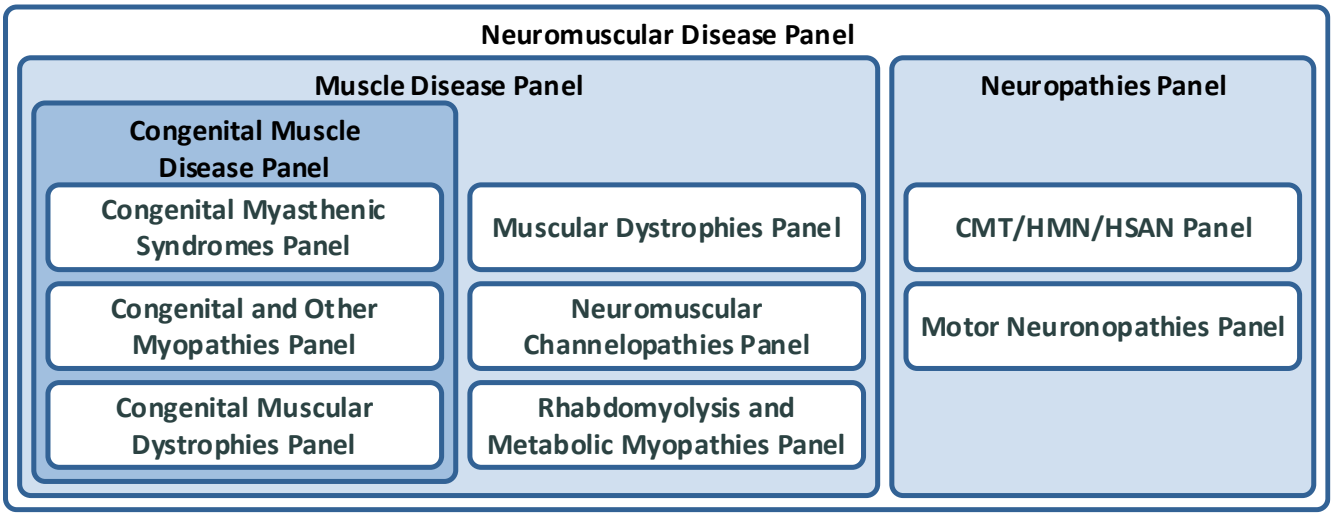
# Neuromuscular Disease Multigene Panels

The neuromuscular disease (NMD) panels (Figure 1) were curated to include relevant genes associated with inherited NMDs. Each panel aims to encompass genes associated with the likely differential diagnoses and be sufficiently comprehensive to include NMDs with substantial clinical overlap. The panels' structure and design should allow flexibility to the ordering clinician to select a more targeted or broad panel depending on the diagnostic certainty. The choice of panels should be driven by clinical judgement, informed by the patient phenotype, and dictated by disease certainty. For more information to guide test selection and establishing eligibility see the Neuromuscular Disease. Genetic Testing Recommendations.

**Figure 1. Neuromuscular Disease Multigene Panels**




CMT, Charcot-Marie-Tooth disease; HMN, hereditary motor neuropathy; HSAN, hereditary sensory and autonomic neuropathy.

**The panels capture the coding regions and flanking intron/exon boundaries and identify relevant copy number variants (CNVs) of all genes. Select relevant intronic variants are included for the genes listed in the panel.**

Note that some of the genetic variants underlying certain inherited NMDs (e.g., myotonic dystrophy, facioscapulohumeral muscular dystrophy (FSHD), oculopharyngeal muscular dystrophy (OPMD)) are not detectable via sequencing through the NMD panels. See Table 1 for CNV analysis exceptions. For availability of non-sequencing tests in Ontario see the NMD ordering guidance.

The Expert Group followed an evidence-based framework for each panel to achieve consensus on which genes to include on NMD panels. ClinGen, Genomic England PanelApp, and/or GeneTable of Neuromuscular Disorders curations were not available for all the disease entities included in the molecular panels.

## Evidence Framework for Gene Inclusion

The following constitutes the list of resources and evidence thresholds for inclusion:

- **Clinical Genome Resource (ClinGen):** Genes curated as Moderate, Strong, or Definitive for gene-disease validity in ClinGen.
- **Genomics England PanelApp:** Genes identified as Green using the Genomics England PanelApp.
- **GeneTable of Neuromuscular Disorders:** NMD-specific genetic information public database.
- **Expert Consensus:** Genes for which there is supportive evidence in the literature and vetted by the Expert Group members.

## Table 1. Gene Contents for Neuromuscular Disease Multigene Panels

| Genetic Test | Genes |
|---|---|
| **Neuromuscular Diseases Panel**<br>**(428 genes)** | *AARS1, ABCA1, ABHD12, ABHD5, ACAD9, ACADL, ACADM, ACADVL, ACTA1, ACTN2, ACVR1, ADSS1, AGL, AGRN, AGTPBP1, AIFM1, ALDOA, ALG14, ALG2, ANO5, APTX, ARHGEF10, ARSA, ASAH1, ASCC1, ASCC3, ATL1, ATL3, ATM, ATP1A1, ATP1A2, ATP2A1, ATP5F1D, ATP7A, B3GALNT2, B4GALNT1, B4GAT1, BAG3, BCKDHB, BICD2, BIN1, BSCL2, BVES, C1QBP, CACNA1A, CACNA1H, CACNA1S, CADM3, CAPN3, CASQ1, CAV3, CAVIN1, CCDC78, CCT5, CD59, CFAP276, CFL2, CHAT, CHCHD10, CHD8, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CLCN1, CLN3, CLTCL1, CNTN1, CNTNAP1, COA7, COL12A1, COL13A1, COL6A1, COL6A2, COL6A3, COLQ, COX6A1, CPOX, CPT1A, CPT2, CRPPA, CRYAB, CTDP1, CYP27A1, DAG1, DARS2, DCAF8, DCTN1, DEGS1, DES, DGAT2, DGUOK, DHTKD1, DMD, DNAJB2, DNAJB4, DNAJB6, DNM2, DNMT1, DNMT3B, DOK7, DOLK, DPAGT1, DPM1, DPM2, DPM3, DST, DYNC1H1, DYSF, ECEL1, EGR2, ELP1, EMD, ENO3, EPG5, ERCC6, ERCC8, ETFA, ETFB, ETFDH, EXOSC3, FAH, FBLN5, FBXO38, FDX2, FGD4, FHL1, FIG4, FKBP14, FKRP, FKTN, FLAD1, FLNC, FLVCR1, FXN[a], FXR1, GAA, GALC, GAN, GARS1, GATM, GBA2, GBE1, GBF1, GDAP1, GFPT1, GGPS1, GIPC1, GJB1, GJB3, GJC2, GLA, GMPPB, GNB4, GNE, GOLGA2, GOSR2, GYG1, GYS1, HACD1, HADHA, HADHB, HARS1, HINT1, HK1, HMBS, HNRNPA1, HNRNPA2B1, HNRNPDL, HOXD10, HRAS, HSPB1, HSPB3, HSPB8, HYCC1, IARS2, IGHMBP2, INF2, INPP5K, ISCU, ITGA7, ITPR3, JAG1, JAG2, KARS1, KBTBD13, KCNA1, KCNA2, KCNE3, KCNJ2, KIF1A, KIF1B, KIF5A, KLHL40, KLHL41, KY, LAMA2, LAMA5, LAMB2, LAMP2, LARGE1, LDB3, LDHA, LIMS2, LITAF, LMNA, LMOD3, LPIN1, LRIF1, LRP12, LRP4, LRSAM1, LYST, MAP3K20, MARS1, MB, MCM3AP, MCOLN1, MEGF10, MFN2, MGME1, MICU1, MLIP, MMACHC, MME, MORC2, MPDU1, MPV17, MPZ, MSTN, MSTO1, MT-ATP6, MTM1, MTMR2, MTRFR, MT-RNR1, MT-TL1, MTTP, MUSK, MYBPC1, MYBPC3, MYH2, MYH3, MYH7, MYL1, MYL2, MYMK, MYO18B, MYO9A, MYOT, MYPN, NAGA, NAGLU, NARS1, NDRG1, NEB, NEFH, NEFL, NGF, NHERF1, NMNAT2, NTRK1, OPA1, OPA3, ORAI1, PABPN1[a], PAX7, PCK2, PDHA1, PDK3, PDSS1, PDSS2, PEX10, PEX7, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PHYH, PIEZO2, PLEC, PLEKHG5, PMM2, PMP2, PMP22, PNKP, PNPLA2, PNPLA8, POGLUT1, POLG, POLG2, POLR3A, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, PPOX, PRDM12, PREPL, PRKAG2, PRNP, PRPS1, PRX, PTPN11, PURA, PUS1, PYGM, PYROXD1, RAB7A, RAPSN, RBCK1, REEP1, RETREG1, RFC1, RILPL1, RNASEH1, RPH3A, RRM2B, RXYLT1, RYR1, RYR3, SACS, SBF1, SBF2, SCN10A, SCN11A, SCN4A, SCN9A, SCO2, SELENON, SEPTIN9, SETX, SGCA, SGCB, SGCD, SGCG, SGPL1, SH3TC2, SIGMAR1, SIL1, SLC12A3, SLC12A6, SLC16A1, SLC18A3, SLC22A5, SLC25A1, SLC25A19, SLC25A20, SLC25A3, SLC25A32, SLC25A4, SLC25A42, SLC25A46, SLC52A2, SLC52A3, SLC5A7, SMCHD1, SMN1[b], SNAP25, SORD, SOX10, SPAST, SPEG, SPG11, SPTBN4, SPTLC1, SPTLC2, STAC3, STIM1, SUCLA2, SURF1, SVIL, SYNE1, SYNE2, SYT2, TAFAZZIN, TANGO2, TCAP, TFG, TK2, TMEM43, TNNC2, TNNI2, TNNT1, TNNT3, TNPO3, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRIM2, TRIM32, TRIM54, TRIM63, TRIP4, TRMT5, TRPA1, TRPV4, TSFM, TTN, TTPA, TTR, TUBB3, TYMP, UBA1, UNC13A, UNC45B, VAMP1, VAPB, VCP, VMA21, VPS13A, VRK1, VWA1, WARS1, WNK1, XK, XPA, YARS1, YARS2, ZFHX2, ZFYVE26* |

| Genetic Test | Genes |
|---|---|
| **Muscle Diseases Panel**<br>**(252 genes)** | *ABHD5, ACAD9, ACADL, ACADM, ACADVL, ACTA1, ACTN2, ACVR1, ADSS1, AGL, AGRN, ALDOA, ALG14, ALG2, ANO5, ASCC3, ATP1A2, ATP2A1, ATP5F1D, B3GALNT2, B4GAT1, BAG3, BICD2, BIN1, BVES, C1QBP, CACNA1A, CACNA1H, CACNA1S, CAPN3, CASQ1, CAV3, CAVIN1, CCDC78, CFL2, CHAT, CHD8, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CLCN1, CLN3, CNTN1, COL12A1, COL13A1, COL6A1, COL6A2, COL6A3, COLQ, CPT1A, CPT2, CRPPA, CRYAB, DAG1, DES, DGUOK, DMD, DNAJB4, DNAJB6, DNM2, DNMT3B, DOK7, DOLK, DPAGT1, DPM1, DPM2, DPM3, DYSF, ECEL1, EMD, ENO3, EPG5, ETFA, ETFB, ETFDH, FDX2, FHL1, FKBP14, FKRP, FKTN, FLAD1, FLNC, FXR1, GAA, GATM, GBE1, GFPT1, GGPS1, GIPC1, GMPPB, GNE, GOLGA2, GOSR2, GYG1, GYS1, HACD1, HADHA, HADHB, HNRNPA1, HNRNPA2B1, HNRNPDL, HRAS, IGHMBP2, INPP5K, ISCU, ITGA7, JAG2, KBTBD13, KCNA1, KCNE3, KCNJ2, KLHL40, KLHL41, KY, LAMA2, LAMA5, LAMB2, LAMP2, LARGE1, LDB3, LDHA, LIMS2, LMNA, LMOD3, LPIN1, LRIF1, LRP12, LRP4, MAP3K20, MB, MCOLN1, MEGF10, MGME1, MICU1, MLIP, MPDU1, MSTN, MSTO1, MTM1, MUSK, MYBPC1, MYBPC3, MYH2, MYH3, MYH7, MYL1, MYL2, MYMK, MYO18B, MYO9A, MYOT, MYPN, NEB, ORAI1, PABPN1[Error! Bookmark not defined.], PAX7, PDSS1, PDSS2, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PIEZO2, PLEC, PNPLA2, PNPLA8, POGLUT1, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, PREPL, PRKAG2, PURA, PUS1, PYGM, PYROXD1, RAPSN, RBCK1, RILPL1, RNASEH1, RPH3A, RRM2B, RXYLT1, RYR1, RYR3, SCN4A, SELENON, SGCA, SGCB, SGCD, SGCG, SIL1, SLC12A3, SLC16A1, SLC18A3, SLC22A5, SLC25A1, SLC25A20, SLC25A3, SLC25A32, SLC25A4, SLC25A42, SLC5A7, SMCHD1, SNAP25, SPEG, SPTBN4, STAC3, STIM1, SUCLA2, SVIL, SYNE1, SYNE2, SYT2, TAFAZZIN, TANGO2, TCAP, TK2, TMEM43, TNNC2, TNNI2, TNNT1, TNNT3, TNPO3, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRIM32, TRIM54, TRIM63, TRIP4, TRMT5, TSFM, TTN, TYMP, UNC13A, UNC45B, VAMP1, VCP, VMA21, YARS2* |
| **Congenital Muscle Diseases Panel**<br>**(165 genes)** | *ACTA1, ACTN2, ACVR1, ADSS1, AGRN, ALG14, ALG2, ASCC3, ATP2A1, B3GALNT2, B4GAT1, BAG3, BICD2, BIN1, CACNA1H, CACNA1S, CASQ1, CAV3, CCDC78, CFL2, CHAT, CHD8, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CLN3, CNTN1, COL12A1, COL13A1, COL6A1, COL6A2, COL6A3, COLQ, CRPPA, CRYAB, DAG1, DES, DMD, DNAJB4, DNM2, DOK7, DOLK, DPAGT1, DPM1, DPM2, ECEL1, EPG5, FHL1, FKBP14, FKRP, FKTN, FLNC, FXR1, GAA, GATM, GFPT1, GIPC1, GMPPB, GOLGA2, GOSR2, HACD1, HNRNPA1, HNRNPA2B1, HRAS, IGHMBP2, INPP5K, ISCU, ITGA7, KBTBD13, KLHL40, KLHL41, KY, LAMA2, LAMA5, LAMB2, LAMP2, LARGE1, LDB3, LMNA, LMOD3, LRP12, LRP4, MAP3K20, MB, MCOLN1, MEGF10, MICU1, MPDU1, MSTN, MSTO1, MTM1, MUSK, MYBPC1, MYBPC3, MYH2, MYH3, MYH7, MYL1, MYL2, MYMK, MYO18B, MYO9A, MYOT, MYPN, NEB, PABPN1[Error! Bookmark not defined.], PAX7, PIEZO2, PLEC, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PREPL, PURA, PYROXD1, RAPSN, RILPL1, RPH3A, RXYLT1, RYR1, RYR3, SCN4A, SELENON, SGCA, SGCB, SGCD, SGCG, SIL1, SLC18A3, SLC25A1, SLC25A4, SLC5A7, SNAP25, SPEG, SPTBN4, STAC3, STIM1, SVIL, SYNE1, SYT2, TCAP, TNNC2, TNNI2, TNNT1, TNNT3, TNPO3, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRIM32, TRIM54, TRIM63, TRIP4, TTN, UNC13A, UNC45B, VAMP1, VCP, VMA21* |
| **Congenital Myasthenic Syndromes Panel**<br>**(36 genes)** | *AGRN, ALG14, ALG2, CHAT, CHD8, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, COL13A1, COLQ, DOK7, DPAGT1, GFPT1, GMPPB, LAMA5, LAMB2, LRP4, MUSK, MYO9A, PLEC, PREPL, PURA, RAPSN, RPH3A, RYR1, SCN4A, SLC18A3, SLC25A1, SLC5A7, SNAP25, SYT2, TOR1AIP1, UNC13A, VAMP1* |

| Genetic Test | Genes |
|---|---|
| **Congenital and Other Myopathies Panel**<br>**(98 genes)** | *ACTA1*, *ACTN2*, *ACVR1*, *ADSS1*, *ASCC3*, *ATP2A1*, *BAG3*, *BICD2*, *BIN1*, *CACNA1H*, *CACNA1S*, *CASQ1*, *CAV3*, *CCDC78*, *CFL2*, *CHKB*, *CLN3*, *CNTN1*, *COL12A1*, *CRYAB*, *DES*, *DNAJB4*, *DNM2*, *DOK7*, *ECEL1*, *EPG5*, *FHL1*, *FKBP14*, *FLNC*, *FXR1*, *GATM*, *GIPC1*, *HACD1*, *HNRNPA1*, *HNRNPA2B1*, *HRAS*, *IGHMBP2*, *ISCU*, *KBTBD13*, *KLHL40*, *KLHL41*, *KY*, *LAMA2*, *LAMP2*, *LDB3*, *LMNA*, *LMOD3*, *LRP12*, *MAP3K20*, *MB*, *MCOLN1*, *MEGF10*, *MICU1*, *MSTN*, *MTM1*, *MYBPC1*, *MYBPC3*, *MYH2*, *MYH3*, *MYH7*, *MYL1*, *MYL2*, *MYMK*, *MYO18B*, *MYOT*, *MYPN*, *NEB*, *PABPN1*[Error! Bookmark not defined.], *PAX7*, *PIEZO2*, *PLEC*, *PYROXD1*, *RILPL1*, *RYR1*, *RYR3*, *SCN4A*, *SELENON*, *SLC25A4*, *SPEG*, *SPTBN4*, *STAC3*, *STIM1*, *SVIL*, *TNNC2*, *TNNI2*, *TNNT1*, *TNNT3*, *TNPO3*, *TPM2*, *TPM3*, *TRIM32*, *TRIM54*, *TRIM63*, *TRIP4*, *TTN*, *UNC45B*, *VCP*, *VMA21* |
| **Congenital Muscular Dystrophies Panel**<br>**(48 genes)** | *ACTA1*, *B3GALNT2*, *B4GAT1*, *CHKB*, *COL12A1*, *COL6A1*, *COL6A2*, *COL6A3*, *CRPPA*, *DAG1*, *DMD*, *DNM2*, *DOLK*, *DPM1*, *DPM2*, *FHL1*, *FKRP*, *FKTN*, *GAA*, *GMPPB*, *GOLGA2*, *GOSR2*, *INPP5K*, *ITGA7*, *LAMA2*, *LARGE1*, *LMNA*, *MICU1*, *MPDU1*, *MSTO1*, *PLEC*, *POMGNT1*, *POMGNT2*, *POMK*, *POMT1*, *POMT2*, *RXYLT1*, *RYR1*, *SELENON*, *SGCA*, *SGCB*, *SGCD*, *SGCG*, *SIL1*, *SYNE1*, *TCAP*, *TRAPPC11*, *TRIP4* |
| **Muscular Dystrophies Panel**<br>**(79 genes)** | *ACADVL*, *ANO5*, *ATP2A1*, *BAG3*, *BVES*, *CAPN3*, *CAV3*, *CAVIN1*, *COL6A1*, *COL6A2*, *COL6A3*, *CPT2*, *CRPPA*, *CRYAB*, *DAG1*, *DES*, *DMD*, *DNAJB6*, *DNMT3B*, *DOK7*, *DPM3*, *DYSF*, *EMD*, *FHL1*, *FKRP*, *FKTN*, *FLNC*, *GAA*, *GGPS1*, *GMPPB*, *GNE*, *GOSR2*, *HNRNPDL*, *JAG2*, *KBTBD13*, *LAMA2*, *LAMP2*, *LARGE1*, *LIMS2*, *LMNA*, *LPIN1*, *LRIF1*, *MTM1*, *MYH7*, *MYOT*, *ORAI1*, *PFKM*, *PHKA1*, *PLEC*, *POGLUT1*, *POMGNT1*, *POMGNT2*, *POMK*, *POMT1*, *POMT2*, *POPDC3*, *PYGM*, *PYROXD1*, *RYR1*, *SELENON*, *SGCA*, *SGCB*, *SGCD*, *SGCG*, *SMCHD1*, *STIM1*, *SYNE1*, *SYNE2*, *TAFAZZIN*, *TCAP*, *TK2*, *TMEM43*, *TNPO3*, *TOR1AIP1*, *TRAPPC11*, *TRIM32*, *TTN*, *VCP*, *VMA21* |
| **Neuromuscular Channelopathies Panel**<br>**(9 genes)** | *ATP1A2*, *CACNA1A*, *CACNA1S*, *CLCN1*, *KCNA1*, *KCNE3*, *KCNJ2*, *SCN4A*, *SLC12A3* |
| **Rhabdomyolysis and Metabolic Myopathies Panel**<br>**(100 genes)** | *ABHD5*, *ACAD9*, *ACADL*, *ACADM*, *ACADVL*, *AGL*, *ALDOA*, *ANO5*, *ATP2A1*, *ATP5F1D*, *C1QBP*, *CACNA1S*, *CAPN3*, *CASQ1*, *CAV3*, *CHKB*, *CPT1A*, *CPT2*, *CRPPA*, *DAG1*, *DGUOK*, *DMD*, *DNAJB6*, *DYSF*, *EMD*, *ENO3*, *ETFA*, *ETFB*, *ETFDH*, *FDX2*, *FHL1*, *FKRP*, *FKTN*, *FLAD1*, *GAA*, *GATM*, *GBE1*, *GMPPB*, *GYG1*, *GYS1*, *HADHA*, *HADHB*, *ISCU*, *ITGA7*, *LAMA2*, *LAMP2*, *LARGE1*, *LDHA*, *LPIN1*, *MGME1*, *MLIP*, *PDSS1*, *PDSS2*, *PFKM*, *PGAM2*, *PGK1*, *PGM1*, *PHKA1*, *PHKB*, *PNPLA2*, *PNPLA8*, *POLG*, *POLG2*, *POMGNT1*, *POMGNT2*, *POMK*, *POMT1*, *POMT2*, *PRKAG2*, *PUS1*, *PYGM*, *RBCK1*, *RNASEH1*, *RRM2B*, *RYR1*, *SCN4A*, *SGCA*, *SGCB*, *SGCD*, *SGCG*, *SIL1*, *SLC16A1*, *SLC22A5*, *SLC25A20*, *SLC25A3*, *SLC25A32*, *SLC25A4*, *SLC25A42*, *STAC3*, *SUCLA2*, *TAFAZZIN*, *TANGO2*, *TCAP*, *TK2*, *TNPO3*, *TRIM32*, *TRMT5*, *TSFM*, *TYMP*, *YARS2* |

| Genetic Test | Genes |
|---|---|
| **Neuropathies Panel**<br>**(193 genes)** | *AARS1, ABCA1, ABHD12, AGTPBP1, AIFM1, APTX, ARHGEF10, ARSA, ASAH1, ASCC1, ATL1, ATL3, ATM, ATP1A1, ATP7A, B4GALNT1, BAG3, BCKDHB, BICD2, BSCL2, CADM3, CCT5, CD59, CFAP276, CHCHD10, CLTCL1, CNTNAP1, COA7, COX6A1, CPOX, CTDP1, CYP27A1, DARS2, DCAF8, DCTN1, DEGS1, DGAT2, DHTKD1, DNAJB2, DNM2, DNMT1, DST, DYNC1H1, EGR2, ELP1, ERCC6, ERCC8, EXOSC3, FAH, FBLN5, FBXO38, FGD4, FIG4, FLVCR1, FXN*[Error! Bookmark not defined.]*, GALC, GAN, GARS1, GBA2, GBF1, GDAP1, GJB1, GJB3, GJC2, GLA, GNB4, HADHA, HADHB, HARS1, HINT1, HK1, HMBS, HOXD10, HSPB1, HSPB3, HSPB8, HYCC1, IARS2, IGHMBP2, INF2, ITPR3, JAG1, KARS1, KCNA2, KIF1A, KIF1B, KIF5A, LAMP2, LDB3, LITAF, LMNA, LRSAM1, LYST, MARS1, MCM3AP, MEGF10, MFN2, MMACHC, MME, MORC2, MPV17, MPZ, MT-ATP6, MTMR2, MTRFR, MT-RNR1, MT-TL1, MTTP, NAGA, NAGLU, NARS1, NDRG1, NEFH, NEFL, NGF, NHERF1, NMNAT2, NTRK1, OPA1, OPA3, PCK2, PDHA1, PDK3, PEX10, PEX7, PHYH, PLEKHG5, PMM2, PMP2, PMP22, PNKP, POLG, POLR3A, PPOX, PRDM12, PRNP, PRPS1, PRX, PTPN11, RAB7A, REEP1, RETREG1, RFC1, SACS, SBF1, SBF2, SCN10A, SCN11A, SCN9A, SCO2, SEPTIN9, SETX, SGPL1, SH3TC2, SIGMAR1, SLC12A6, SLC25A19, SLC25A46, SLC52A2, SLC52A3, SLC5A7, SMN1*[b]*, SORD, SOX10, SPAST, SPG11, SPTBN4, SPTLC1, SPTLC2, SURF1, SYT2, TFG, TRIM2, TRIP4, TRPA1, TRPV4, TTPA, TTR, TUBB3, TYMP, UBA1, VAPB, VCP, VPS13A, VRK1, VWA1, WARS1, WNK1, XK, XPA, YARS1, ZFHX2, ZFYVE26* |
| **CMT/HMN/HSAN Panel**<br>**(188 genes)** | *AARS1, ABCA1, ABHD12, AGTPBP1, AIFM1, APTX, ARHGEF10, ARSA, ATL1, ATL3, ATM, ATP1A1, ATP7A, B4GALNT1, BAG3, BCKDHB, BICD2, BSCL2, CADM3, CCT5, CD59, CFAP276, CHCHD10, CLTCL1, CNTNAP1, COA7, COX6A1, CPOX, CTDP1, CYP27A1, DARS2, DCAF8, DCTN1, DEGS1, DGAT2, DHTKD1, DNAJB2, DNM2, DNMT1, DST, DYNC1H1, EGR2, ELP1, ERCC6, ERCC8, FAH, FBLN5, FBXO38, FGD4, FIG4, FLVCR1, FXN*[Error! Bookmark not defined.]*, GALC, GAN, GARS1, GBA2, GBF1, GDAP1, GJB1, GJB3, GJC2, GLA, GNB4, HADHA, HADHB, HARS1, HINT1, HK1, HMBS, HOXD10, HSPB1, HSPB3, HSPB8, HYCC1, IARS2, IGHMBP2, INF2, ITPR3, JAG1, KARS1, KCNA2, KIF1A, KIF1B, KIF5A, LAMP2, LDB3, LITAF, LMNA, LRSAM1, LYST, MARS1, MCM3AP, MEGF10, MFN2, MMACHC, MME, MORC2, MPV17, MPZ, MT-ATP6, MTMR2, MTRFR, MT-RNR1, MT-TL1, MTTP, NAGA, NAGLU, NARS1, NDRG1, NEFH, NEFL, NGF, NHERF1, NMNAT2, NTRK1, OPA1, OPA3, PCK2, PDHA1, PDK3, PEX10, PEX7, PHYH, PLEKHG5, PMM2, PMP2, PMP22, PNKP, POLG, POLR3A, PPOX, PRDM12, PRNP, PRPS1, PRX, PTPN11, RAB7A, REEP1, RETREG1, RFC1, SACS, SBF1, SBF2, SCN10A, SCN11A, SCN9A, SCO2, SEPTIN9, SETX, SGPL1, SH3TC2, SIGMAR1, SLC12A6, SLC25A19, SLC25A46, SLC52A2, SLC52A3, SLC5A7, SMN1*[b]*, SORD, SOX10, SPAST, SPG11, SPTBN4, SPTLC1, SPTLC2, SURF1, SYT2, TFG, TRIM2, TRPA1, TRPV4, TTPA, TTR, TUBB3, TYMP, UBA1, VCP, VPS13A, VRK1, VWA1, WARS1, WNK1, XK, XPA, YARS1, ZFHX2, ZFYVE26* |
| **Motor Neuronopathies Panel**<br>**(33 genes)** | *ASAH1, BICD2, BSCL2, CHCHD10, DCTN1, DYNC1H1, EXOSC3, GARS1, HINT1, HSPB3, HSPB8, IGHMBP2, REEP1, SLC52A2, SLC52A3, SLC5A7, SMN1*[b]*, SPG11, TRIP4, TRPV4, UBA1, VRK1, WARS1, AARS1, ASCC1, DNAJB2, FBXO38, HSPB1, PLEKHG5, SETX, SIGMAR1, SYT2, VAPB* |

[a] Repeat expansion analysis is not available

[b] Deletion/duplication analysis is not available