

Supplemental Table 1. List of Genes for Targeted NGS Sequencing (579)**Con genital myopathy (30):**

ACTA1, ACTG2, ATP2A1, BIN1, CCDC78, CFL2, CLCN1, CNTN1, DNM2, FAM123B, FLNC, GNE, HOXD10, HRAS, KBTBD13, LDB3, MEGF10, MTM1, MTMR14, MYF6, MYH2, MYH7, NEB, TIA1, TNNT1, TPM2, TPM3, TTN, VCP, VMA21

Muscular dystrophy (48):

ANO5, BAG3, CAPN3, CAV3, CHKB, CNBP, COL6A1, COL6A2, COL6A3, CRYAB, DAG1, DES, DMD, DMPK, DNAJB6, DPM2, DPM3, DYSF, EMD, FHL1, FKBP14, FKRP, FKTN, GTDC2, ISPD, ITGA7, ITGA9, KLHL9, LAMA2, LARGE, LMNA, MTAP, MYOT, PABPN1, PLEC, PLOD3, POMGNT1, POMT1, POMT2, SECISBP2, SGCA, SGCB, SGCD, SGCG, SPP1, SYNE2, TCAP, TRIM32

Congenital myopathy or Muscular dystrophy (2):

RYR1, SEPN1

Metabolic myopathy (202):

AARS2, ACACA, ACAD9, ACADM, ACADVL, ACSF3, ACY1, ADCK3, AGK, AGL, AIFM1, ALDH1B1, ALDOA, ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, ALPL, AMACR, AMPD1, ATP5A1, ATP5E, ATP5G3, ATPAF2, B4GALT1, BCS1L, BRP44L, C10orf2, C12orf65, CACNA1S, COA5, COG1, COG4, COG5, COG6, COG7, COG8, COQ2, COQ4, COQ6, COQ9, COX10, COX14, COX15, COX20, COX4I2, COX6B1, COX7B, CPT1A, CPT1B, CPT2, CYC1, CYCS, D2HGDH, DARS2, DGUOK, DMGDH, DNA2, DOLK, DPM1, EARS2, ENO3, ETFA, ETFB, ETFDH, FARS2, FASTKD2, FOXRED1, GAA, GALC, GBE1, GFER, GFM1, GPR172B, GYG1, GYS1, HADH, HADHA, HADHB, HARS2, IARS, ISCU, LAMP2, LARS2, LDHA, LDHB, LIAS, LPIN1, LRPPRC, LYRM4, LYRM7, MARS, MATR3, MGAT2, MGME1, MPV17, MRPL12, MRPL3, MRPL44, MRPS16, MRPS22, MTFMT, MTHFD1, MTHFR, MTO1, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA4, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEU1, NFU1, NUBPL, OAT, OPA1, OXCT1, PDSS1, PDSS2, PEX12, PEX14, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PHYH, PMM2, PNPLA2, PNPT1, POLG, POLG2, PSAP, PTEN, PTRF, PUS1, PYGL, PYGM, RMND1, RRM2B, RSPH9, SCO1, SCO2, SDHA, SDHAF1, SDHAF2, SDHB, SDHC, SDHD, SLC22A5, SLC25A20, SLC25A3, SLC25A4, SUCLA2, SUCLG1, SURF1, TACO1, TARS2, TAZ, TIMM44, TK2, TMEM165, TMEM70, TSFM, TTC19, TYMP, UQCRB, UQCRC2, UQCRCQ, VARS2, YARS2

Motor neuron or Peripheral nerve disease (75):

AARS, AFG3L2, APTX, ARHGEF10, ATLL1, ATXN2, BSCL2, CCT5, COLQ, DCTN1, DHTKD1, DNAJB2, DNMT1, DYNC1H1, EGR2, ELAVL4, ERLIN2, FAM134B, FGD4, FOXE1, GAN, GARS, GDAP1, GJB1, HARS, HINT1, HOXB1, HSPB1, HSPB3, HSPB8, HSPG2, IGHMBP2, IKBKAP, INF2, KARS, KIF1B, KIF5A, LITAF, MED25, MFN2, MPZ, MTMR2, MYH14, NAGA, NDRG1, NEFL, NGF, NIPA1, PDK3, PLEKHG5, PLP1, PMP22, PNPLA6, PRX, RAB7A, REEP1, SACS, SBF2, SH3TC2, SLC12A6, SLC5A7, SMN1, SMN2, SNX25, SPTLC1, SPTLC2, SYNE1, TDP1, TFG, TGM6, TRPV4, UBA1, VRK1, WNK1, YARS

Myasthenic syndrome (12):

AGRN, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, DOK7, DPAGT1, GFPT1, LAMB2, MUSK, SCN4A

Cardiomyopathy (3):

MYBPC3, MYLK2, RYR2

Candidate genes (207):

ACAD10, ACAD11, AIFM2, ATP5B, ATP5C1, ATP5D, ATP5F1, ATP5G1, ATP5G2, ATP5H, ATP5I, ATP5J, ATP5J2, ATP5L, ATP5O, ATP5S, ATPAF1, ATPIF1, C14orf2, CARS, CARS2, CD36, CHCHD1, CHCHD7, CMC1, CMC2, COA1, COA3, COA6, COQ10A, COQ10B, COQ3, COQ5, COQ7, COX11, COX16, COX17, COX18, COX19, COX4I1, COX5A, COX5B, COX6A1, COX6A2, COX6B2, COX6C, COX7A1, COX7A2, COX7A2L, COX7B2, COX7C, COX8A, COX8C, ECi1, ECSIT, EPRS, FARSA, FARSB, FRG1, FUNDC2, HINT3, IARS2, ICT1, LACTB, LYRM1, MCAT, MDH2, METTL17, MNF1, MRP63, MRPL1, MRPL10, MRPL11, MRPL13, MRPL14, MRPL15, MRPL16, MRPL17, MRPL18, MRPL19, MRPL2, MRPL20, MRPL21, MRPL22, MRPL23, MRPL24, MRPL27, MRPL28, MRPL30, MRPL32, MRPL33, MRPL34, MRPL35, MRPL36, MRPL37, MRPL38, MRPL39, MRPL4, MRPL40, MRPL41, MRPL42, MRPL43, MRPL45, MRPL46, MRPL47, MRPL48, MRPL49, MRPL50, MRPL51, MRPL52, MRPL53, MRPL54, MRPL55, MRPL9, MRPS10, MRPS11, MRPS12, MRPS14, MRPS15, MRPS17, MRPS18A, MRPS18B, MRPS18C, MRPS2, MRPS21, MRPS23, MRPS24, MRPS25, MRPS26, MRPS27, MRPS28, MRPS30, MRPS31, MRPS33, MRPS34, MRPS35, MRPS36, MRPS5, MRPS6, MRPS7, MRPS9, MRS2, MTCH1, MTERF, MTG1, MTHFD1L, MTHFS, MTIF3, MTRF1L, NARS, NARS2, NDUFA3, NDUFA4L2, NDUFA5, NDUFA6, NDUFA7, NDUFA8, NDUFAB1, NDUFB1, NDUFB10, NDUFB11, NDUFB2, NDUFB4, NDUFB5, NDUFB6, NDUFB7, NDUFB8, NDUFC1, NDUFC2, NDUFS5, NDUFV3, NIPSNAP3A, NRF1, OXA1L, OXSM, PARS2, PGAM1, POLRMT, PRELID1, PRELID2, PTC1, PTC2, PTC3, PTRH2, QARS, RARS, SLIRP, SMCR7, SMCR7L, SUCLG2, TARS, TARSL2, TIMM21, TIMM23, TOMM70A, TOP1MT, TYMS, UQCR10, UQCR11, UQCRC1, UQCRCFS1, UQCRH, USMG5, VARS, WARS, WARS2, YME1L1