

Supplementary material 1A: Myopathy genes included in the initial next generation sequencing panel.

ACTA1, ANO5, ATP2A1, B3GNT1, B3GALNT2, BAG3, BICD2, BIN1, CACNA1S, CAPN3, CAV3, CFL2, CHKB, CLCN1, CNTN1, COL6A1, COL6A2, COL6A3, CRYAB, DAG1, DES, DMD, DNAJB6, DNMT2, DPM1, DPM2, DPM3, DYSF, EMD, FHL1, FKRP, FKTN, FLNC, GAA, GMPPB, GNE, ISPD, ITGA7, KBTBD13, KLHL40, LAMA2, LAMP2, LARGE, LDB3, LMNA, MEGF10, MTM1, MYH7, MYOT, NEB, PHKA1, PLEC, POMGNT1, POMT1, POMT2, PYGM, RYR1, SCN4A, SEPNI, SGCA, SGCB, SGCD, SGCG, SIL1, SYNE1, TCAP, TMEM5, TTN, TNNT2, TNNT1, TNPO3, TPM2, TPM3, and TRIM32

Supplementary material 1B: Additional next generation sequencing panel obtained mainly to search for deleterious variants in genes known to cause myopathy with rimmed vacuoles.

AAAS, AARS, ABCA1, ABCD1, ACTA1, ADCY6, AIFM1, AMACR, ANO5, AP1S1, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APOA1, APTX, ARHGEF10, ARSA, ATLI, ATM, ATP7A, B2M, B4GALNT1, BAG3, BCKDHB, BICD2, BIN1, BSCL2, C12orf65, CAV3, CCT5, CLCF1, CNTNAP1, COX10, CPOX, CRLF1, CRYAB, CTDP1, CTSA, CYP27A1, CYP2UI, CYP7B1, DARS2, DCAF8, DCTN1, DDHD1, DDHD2, DES, DGUOK, DHH, DHTKD1, DNAJB2, DNAJB6, DNMT2, DNMT1, DST, DYNC1H1, DYSF, EGR2, ERBB3, ERCC6, ERCC8, ERLIN2, FA2H, FAH, FAM126A, FAM134B, FBLN5, FBXO38, FGD4, FGF14, FHL1, FIG4, FLNC, FLVCR1, FMRI, GALC, GAN, GARS, GBA2, GBE1, GDAP1, GJB1, GJB3, GJC2, GLA, GNB4, GNE, GSN, HADHA, HADHB, HARS, HINT1, HK1, HMBS, HNRNPA1, HNRNPA2B1, HSPB1, HSPB3, HSPB8, HSPD1, IGHMBP2, IKBKAP, INF2, KARS, KIF1A, KIF1B, KIF5A, LICAM, LAMA2, LDB3, LITAF, LMNA, LRSAM1, LYST, MAF, MARS, MATR3, MED25, MFN2, MMACHC, MPV17, MPZ, MTMR2, MTPP, MYH14, MYH2, MYH7, MYOT, NAGA, NAGLU, NDRG1, NEB, NEFL, NF2, NGF, NIPA1, NTRK1, OAT, OPA1, PANK2, PDHA1, PDK3, PDYN, PEX10, PEX7, PHYH, PLA2G6, PLEKHG5, PLOD1, PLP1, PMM2, PMP2, PMP22, PNKP, PNPLA6, POLG, PPOX, PRNP, PRPS1, PRX, RAB7A, REEP1, RRM2B, RTN2, SACS, SBF1, SBF2, SCN10A, SCN11A, SCN9A, SCO2, SCP2, SELENON, SETX, SH3TC2, SLC12A6, SLC16A2, SLC25A19, SLC25A46, SLC33A1, SLC52A2, SLC5A7, SNAP29, SOD1, SOX10, SPAST, SPG11, SPG20, SPG21, SPG7, SPTLC1, SPTLC2, SQSTM1, SURF1, TDPI, TECPR2, TFG, TIA1, TRIM2, TRPA1, TRPV4, TTN, TTPA, TTR, TUBB3, TWNK, TYMP, VCP, VPS37A, WASHC5, WNK1, XPA, XPC, YARS, and ZFYVE26