**Supplementary Table 2.** Functionally significant variants in myopathy-related genes

| Gene | Map | Ref Seq* | Variants | | dbSNP138 | 1000G (2014 Sep) | SIFT† | PolyPhen2† | Description |
|---|---|---|---|---|---|---|---|---|---|
| | | | Nucleotides | Amino acids | | | | | |
| *AGRN1* | 1p36.33 | NM_198576.3 | c.4516G>A | p.A1506T | rs200182409 | 0.000 | 0.018 | 0.183 | Likely benign |
| *DCTN1* | 2p13 | NM_004082.4 | c.2054T>G | p.V685G | - | - | 0.000 | 0.989 | Uncertain significance |
| *NEB* | 2q22 | NM_001271208.1 | c.3412A>G | p.N1138D | rs117048449 | 0.019 | 0.196 | 0.946 | Likely benign |
| | | | c.5102T>C | p.V1701A | rs117271684 | 0.019 | 0.29 | 0.621 | Likely benign |
| | | | c.8189A>G | p.D2730G | rs76767949 | 0.020 | 0.02 | 0.973 | Uncertain significance |
| *TTN* | 2q31 | NM_001267550.1 | c.22012G>A | p.D7338N | - | - | 0.05 | 0.801 | Likely benign |
| | | | c.82855A>G | p.T27619A | - | - | 0.375 | 0.334 | Likely benign |
| | | | c.10931G>A | p.S3644N | rs78535378 | 0.016 | 0.636 | 0.000 | Likely benign |
| | | | c.21044C>T | p.A7015V | rs72648960 | 0.036 | 0.074 | 0.096 | Likely benign |
| | | | c.39704C>G | p.P13235R | rs72650066 | 0.043 | 0.220 | 0.010 | Likely benign |
| | | | c.98164A>T | p.I32722F | rs72648270 | 0.019 | 0.182 | 0.997 | Likely benign |
| *DAG1* | 3p21 | NM_001165928.3 | c.331G>A | p.D111N | rs117209107 | 0.004 | 0.284 | 0.162 | Likely benign |
| *FAT1* | 4q35.2 | NM_005245.3 | c.13652C>G | p.A551G | rs148468016 | 0.004 | 0.009 | 1.000 | Uncertain significance |
| *MYOT* | 5q31.2 | NM_006790.2 | c.220A>C | p.K74Q | rs6890689 | - | 1.000 | 1.000 | Uncertain significance |
| *LAMA2* | 6q22-q23 | NM_000426.3 | c.2438T>C | p.I813T | - | - | 0.461 | 0.001 | Likely benign |
| *ANO5* | 11p14.3 | NM_213599.2 | c.966A>T | p.L322F | rs7481951 | - | 0.136 | 0.011 | Likely benign |
| *TRPV4* | 12q24.11 | NM_021625.4 | c.55C>T | p.P19S | rs3742030 | 0.044 | 0.209 | 0.000 | Likely benign |
| *ADSSL1* | 14q32.33 | NM_199165.1 | c.910G>A | p.D304N | rs140614802 | - | 0.000 | 1.000 | Pathogenic |
| | | | c.1048delA | p.I350fs | rs559454746 | 0.001 | - | - | Pathogenic |
| *SYNE2* | 14p23.2 | NM_182914.2 | c.10567A>C | p.K3523Q | rs35203186 | 0.039 | 0.618 | 0.006 | Likely benign |
| | | | c.11935C>G | p.L3979V | rs76576553 | 0.016 | 0.107 | 0.010 | Likely benign |
| *CHRNB1* | 17p12-p11 | NM_000747.2 | c.725G>A | p.R242H | rs79220301 | 0.001 | 0.006 | 0.984 | Uncertain significance |
| *COL6A1* | 21q22.3 | NM_001848.2 | c.2548C>A | p.R850S | rs566453317 | 0.000 | 0.449 | 0.005 | Likely benign |
| *DMD* | Xp21.2 | NM_004006.2 | c.3083G>A | p.R1028H | - | - | 0.134 | 0.003 | Likely benign |
| | | | c.8729A>T | p.E2910V | rs41305353 | 0.036 | 0.003 | 0.119 | Likely benign |
| | | | c.8734A>G | p.N2912D | rs1800278 | 0.037 | 0.472 | 0.281 | Likely benign |

*GenBank registration number of reference sequence, †SIFT: <0.05 predicted to be deleterious, †PolyPhen2: –1 indicates prediction of pathogenicit.