

**Table S3.** Molecular findings from next-generation sequencing

| Patient ID                  | NGS | Gene                                                                                          | Reference sequence | Nucleotide change | Protein change       | Zygosity              | Parental mutation status | MOI   | Publication                | ACMG classification <sup>a</sup> | AF in gnomAD (Total) | Result   |
|-----------------------------|-----|-----------------------------------------------------------------------------------------------|--------------------|-------------------|----------------------|-----------------------|--------------------------|-------|----------------------------|----------------------------------|----------------------|----------|
| Group 1: Inherited myopathy |     |                                                                                               |                    |                   |                      |                       |                          |       |                            |                                  |                      |          |
| CM1                         | ES  | <i>FHL1</i>                                                                                   | NM_001159702.3     | c.377G>A          | p.Cys126Tyr          | Hemizygous            | Maternal allele          | XR    | [PMID: 24928078]           | Likely pathogenic                | Variant not found    | Positive |
| CM2                         | GP  | <i>MYH7</i>                                                                                   | NM_000257.4        | c.2711G>A         | p.Arg904His          | Heterozygous          | De novo                  | AD    | [PMID: 22464770]           | Pathogenic                       | 0.0000006195         | Positive |
| CM3 <sup>b</sup>            | ES  | <i>MYH7</i>                                                                                   | NM_000257.4        | c.4807G>C         | p.Ala1603Pro         | Heterozygous          | Maternal allele          | AD    | [PMID: 27387980]           | Likely pathogenic                | Variant not found    | Positive |
| CM4 <sup>b</sup>            | ES  | <i>MYH7</i>                                                                                   | NM_000257.4        | c.4807G>C         | p.Ala1603Pro         | Heterozygous          | Maternal allele          | AD    | [PMID: 27387980]           | Likely pathogenic                | Variant not found    | Positive |
| CM5                         | GP  | <i>NEB</i>                                                                                    | NM_001164508.2     | c.133_146del      | p.Ser45Thrfs*48      | Heterozygous          | Maternal allele          | AR    | -                          | Pathogenic                       | 0.0000012410         | Probable |
| CM6                         | ES  | <i>RYR1</i>                                                                                   | NM_000540.3        | c.487C>T          | p.Arg163Cys          | Compound heterozygous | Paternal allele          | AR    | [PMID: 12124989]           | Pathogenic                       | 0.0000012390         | Positive |
|                             |     |                                                                                               | NM_000540.3        | c.15089G>A        | p.Arg5030His         |                       | Maternal allele          |       | [PMID: 36697461]           | Likely pathogenic                | 0.0000266400         |          |
| CM7                         | GP  | <i>RYR1</i>                                                                                   | NM_000540.3        | c.9499C>T         | p.Arg3167*           | Heterozygous          | Paternal allele          | AR    | [PMID: 28818389]           | Pathogenic                       | 0.0000043380         | Probable |
| CM8                         | ES  | <i>RYR1</i>                                                                                   | NM_000540.3        | c.10347+1G>A      | p.?                  | Compound heterozygous | Maternal allele          | AR    | [PMID: 32655342]           | Pathogenic                       | 0.0000080560         | Positive |
|                             |     |                                                                                               | NM_000540.3        | c.11314C>T        | p.Arg3772Trp         |                       | Paternal allele          |       | [PMID: 37996280]           | Pathogenic                       | 0.0000055770         |          |
| CM9                         | GP  | <i>RYR1</i>                                                                                   | NM_000540.3        | c.11321C>A        | p.Ala3774Glu         | Homozygous            | Not confirmed            | AR    | -                          | Likely pathogenic                | Variant not found    | Positive |
| CM10                        | GP  | <i>RYR1</i>                                                                                   | NM_000540.3        | c.13244_13264del  | p.Ala4415_Asp4421del | Heterozygous          | Maternal allele          | AD/AR | [PMID: 26994242]           | VUS                              | 0.0001139000         | Possible |
| CM11                        | ES  | <i>SELENON</i>                                                                                | NM_020451.3        | c.1406G>A         | p.Arg469Gln          | Homozygous            | Not confirmed            | AR    | [PMID: 30932294]           | Pathogenic                       | 0.0000123900         | Positive |
| CM12                        | ES  | <i>TTN</i>                                                                                    | NM_001267550.2     | c.2254C>A         | p.Arg752Ser          | Compound heterozygous | Maternal allele          | AR    | [PMID: 36697461]           | VUS                              | 0.0000006200         | Possible |
|                             |     | <i>TTN</i>                                                                                    | NM_001267550.2     | c.38876-2A>C      | p.?                  |                       | Paternal allele          |       | [PMID: 38937733, 36697461] | Pathogenic                       | 0.0000056110         |          |
| CM13                        | ES  | <i>TTN</i>                                                                                    | NM_001267550.2     | c.39109G>T        | p.Glu13037*          | Compound heterozygous | Paternal allele          | AR    | [PMID: 36697461]           | Pathogenic                       | 0.0000012390         | Positive |
|                             |     | <i>TTN</i>                                                                                    | NM_001267550.2     | c.53348T>C        | p.Leu17783Pro        |                       | Maternal allele          |       | [PMID: 36697461]           | Likely pathogenic                | Variant not found    |          |
| CM14                        | GP  | <i>TTN</i>                                                                                    | NM_001267550.2     | c.46966+5G>C      | p.?                  | Compound heterozygous | Paternal allele          | AR    | -                          | Likely pathogenic                | Variant not found    | Positive |
|                             |     | <i>TTN</i>                                                                                    | NM_001267550.2     | c.86188A>T        | p.Lys28730*          |                       | Maternal allele          |       | -                          | Likely pathogenic                | Variant not found    |          |
| CM15                        | ES  | 19q13.33q13.41 deletion (87 protein-encoding genes with possible NMD gene: <i>MYH14</i> gene) |                    |                   |                      |                       |                          | -     | -                          | VUS                              | -                    | Possible |
| DMD1                        | ES  | <i>DMD</i>                                                                                    | NM_004006.3        | c.515_516dup      | p.Ile173Serfs*36     | Hemizygous            | Not confirmed            | XR    | [PMID: 36697461]           | Pathogenic                       | Variant not found    | Positive |
| DMD2                        | GP  | <i>DMD</i>                                                                                    | NM_004006.3        | c.3277-1G>C       | p.?                  | Hemizygous            | Maternal allele          | XR    | [PMID: 27122458]           | Pathogenic                       | Variant not found    | Positive |
| DMD3                        | ES  | <i>DMD</i>                                                                                    | NM_004006.3        | c.3630del         | p.Glu1211Lysfs*4     | Hemizygous            | Not confirmed            | XR    | [PMID: 25007885, 16770791] | Pathogenic                       | Variant not found    | Positive |
| DMD4                        | GP  | <i>DMD</i>                                                                                    | NM_004006.3        | c.3917dup         | p.Asp1307Argfs*4     | Hemizygous            | Maternal allele          | XR    | -                          | Pathogenic                       | Variant not found    | Positive |
| DMD5                        | ES  | <i>DMD</i>                                                                                    | NM_004006.3        | c.5697del         | p.Lys1899Asnfs*2     | Hemizygous            | Not confirmed            | XR    | [PMID: 31727011]           | Pathogenic                       | 0.0000008270         | Positive |
| DMD6                        | ES  | <i>DMD</i>                                                                                    | NM_004006.3        | c.5932del         | p.Arg1978Valfs*5     | Hemizygous            | Not confirmed            | XR    | -                          | Pathogenic                       | Variant not found    | Positive |
| DMD7                        | ES  | <i>DMD</i>                                                                                    | NM_004006.3        | c.6614+2T>C       | p.?                  | Hemizygous            | Not confirmed            | XR    | [PMID: 25007885]           | Pathogenic                       | Variant not found    | Positive |
| DMD8                        | ES  | <i>DMD</i>                                                                                    | NM_004006.3        | c.6986del         | p.Lys2329Serfs*9     | Hemizygous            | Maternal allele          | XR    | [PMID: 26911353]           | Pathogenic                       | 0.0000000000         | Positive |
| DMD9                        | ES  | <i>DMD</i>                                                                                    | NM_004006.3        | c.7197dup         | p.Lys2400Glufs*10    | Hemizygous            | Maternal allele          | XR    | -                          | Likely pathogenic                | Variant not found    | Positive |

|                   |    |               |             |                  |                      |                       |                                |    |                            |                   |                   |          |
|-------------------|----|---------------|-------------|------------------|----------------------|-----------------------|--------------------------------|----|----------------------------|-------------------|-------------------|----------|
| DMD10             | ES | <i>DMD</i>    | NM_004006.3 | c.8086del        | p.Leu2696Trpfs*30    | Hemizygous            | Not confirmed                  | XR | [PMID: 25007885, 36315559] | Pathogenic        | Variant not found | Positive |
| DMD11             | ES | <i>DMD</i>    | NM_004006.3 | c.9100C>T        | p.Arg3034*           | Hemizygous            | Not confirmed                  | XR | [PMID: 32153624]           | Pathogenic        | 0.0000016540      | Positive |
| DMD12             | ES | <i>DMD</i>    | NM_004006.3 | c.9484G>T        | p.Glu3162*           | Hemizygous            | Maternal allele                | XR | [PMID: 36697461]           | Likely pathogenic | Variant not found | Positive |
| DMD13             | ES | <i>DMD</i>    | NM_004006.3 | c.9649+5G>T      | p.?                  | Hemizygous            | Not confirmed                  | XR | [PMID: 20485447]           | Likely pathogenic | Variant not found | Positive |
| DMD14             | ES | <i>DMD</i>    | NM_004006.3 | c.10087_10098del | p.Thr3363_Gly3366del | Hemizygous            | Not confirmed                  | XR | [PMID: 32962870]           | Likely pathogenic | Variant not found | Positive |
| DMD15             | ES | <i>DMD</i>    | NM_004006.3 | c.10097_10099del | p.Gly3366del         | Hemizygous            | Maternal allele                | XR | [PMID: 19937601, 28859693] | Likely pathogenic | Variant not found | Positive |
| DMD16             | ES | <i>DMD</i>    | NM_004006.3 | c.10108C>T       | p.Arg3370*           | Hemizygous            | Not confirmed                  | XR | [PMID: 18583217]           | Pathogenic        | Variant not found | Positive |
| DMD17             | ES | <i>DMD</i>    | NM_004006.3 | c.10108C>T       | p.Arg3370*           | Hemizygous            | Not confirmed                  | XR | [PMID: 18583217]           | Pathogenic        | Variant not found | Positive |
| MD1               | ES | <i>CHKB</i>   | NM_005198.5 | c.598del         | p.Gln200Argfs*11     | Homozygous            | Not confirmed                  | AR | [PMID: 31926838]           | Pathogenic        | 0.0000061960      | Positive |
| MD2               | GP | <i>CHKB</i>   | NM_005198.5 | c.598del         | p.Gln200Argfs*11     | Homozygous            | Not confirmed                  | AR | [PMID: 31926838]           | Pathogenic        | 0.0000061960      | Positive |
| MD3               | ES | <i>COL6A1</i> | NM_001848.3 | c.850G>A         | p.Gly284Arg          | Heterozygous          | De novo                        | AD | [PMID: 17785674]           | Pathogenic        | Variant not found | Positive |
| MD4               | ES | <i>COL6A1</i> | NM_001848.3 | c.868G>A         | p.Gly290Arg          | Heterozygous          | De novo                        | AD | [PMID: 30706156]           | Pathogenic        | Variant not found | Positive |
| MD5               | ES | <i>COL6A1</i> | NM_001848.3 | c.887G>A         | p.Gly296Glu          | Heterozygous          | Not confirmed                  | AD | -                          | Pathogenic        | Variant not found | Probable |
| MD6               | ES | <i>COL6A1</i> | NM_001848.3 | c.1056+1G>C      | p.?                  | Heterozygous          | Not confirmed                  | AD | [PMID: 25749816]           | Pathogenic        | Variant not found | Probable |
| MD7               | GP | <i>COL6A2</i> | NM_001849.4 | c.580C>T         | p.Gln194*            | Homozygous            | Paternal and maternal allele   | AR | -                          | Pathogenic        | Variant not found | Positive |
| MD8               | ES | <i>COL6A2</i> | NM_001849.4 | c.893G>A         | p.Gly298Glu          | Heterozygous          | Not confirmed                  | AD | -                          | Pathogenic        | Variant not found | Probable |
| MD9               | GP | <i>COL6A2</i> | NM_001849.4 | c.2995G>A        | p.Glu999Lys          | Heterozygous          | Not confirmed                  | AD | -                          | VUS               | 0.0000920800      | Possible |
| MD10              | GP | <i>COL6A3</i> | NM_004369.4 | c.6309G>A        | p.Lys2103=           | Heterozygous          | Not confirmed                  | AD | [PMID: 34167565]           | Pathogenic        | Variant not found | Positive |
| MD11              | ES | <i>LAMA2</i>  | NM_000426.4 | c.172T>C         | p.Cys58Arg           | Compound heterozygous | Paternal allele                | AR | -                          | Likely pathogenic | 0.0000006200      | Positive |
|                   |    | <i>LAMA2</i>  | NM_000426.4 | c.5085dup        | p.Ala1696Serfs*6     |                       | Maternal allele                |    | [PMID:32904964]            | Pathogenic        | 0.0000006200      |          |
| MD12              | ES | <i>LAMA2</i>  | NM_000426.4 | c.283+1G>C       | p.?                  | Heterozygous          | Paternal allele                | AR | -                          | Pathogenic        | Variant not found | Probable |
| MD13 <sup>b</sup> | GP | <i>LAMA2</i>  | NM_000426.4 | c.2049_2050del   | p.Arg683Serfs*21     | Heterozygous          | Not confirmed                  | AR | [PMID: 28804634]           | Pathogenic        | 0.0001450000      | Probable |
|                   |    | <i>LAMA2</i>  | NM_000426.4 | c.4714_4717del   | p.Val1572Phefs*22    | Heterozygous          | Not confirmed                  |    | -                          | Likely pathogenic | 0.0000006200      |          |
| MD14 <sup>b</sup> | GP | <i>LAMA2</i>  | NM_000426.4 | c.2049_2050del   | p.Arg683Serfs*21     | Heterozygous          | Not confirmed                  | AR | [PMID: 28804634]           | Pathogenic        | 0.0001450000      | Probable |
|                   |    | <i>LAMA2</i>  | NM_000426.4 | c.4714_4717del   | p.Val1572Phefs*22    | Heterozygous          | Not confirmed                  |    | -                          | Likely pathogenic | 0.0000006200      |          |
| MD15              | GP | <i>LAMA2</i>  | NM_000426.4 | c.2538-1G>C      | p.?                  | Heterozygous          | Not confirmed                  | AR | [PMID: 32028919]           | Likely pathogenic | Variant not found | Possible |
|                   | ES | <i>LAMA2</i>  | NM_000426.4 | c.2718del        | p.Phe906Leufs*169    | Heterozygous          | Not confirmed                  | AR | [PMID: 36697461]           | Pathogenic        | Variant not found | Probable |
|                   |    | <i>LAMA2</i>  | NM_000426.4 | c.7452-16T>G     | p.?                  | Heterozygous          | Not confirmed                  |    | [PMID: 36697461]           | Likely pathogenic | Variant not found |          |
| MD17              | GP | <i>LAMA2</i>  | NM_000426.4 | c.5476C>T        | p.Arg1826*           | Compound heterozygous | Paternal allele                | AR | [PMID: 32936536]           | Pathogenic        | 0.0000589600      | Positive |
|                   |    | <i>LAMA2</i>  | NM_000426.4 | c.9101_9104dup   | p.His3035Glnfs*5     |                       | Maternal allele                |    | [PMID: 32904964]           | Pathogenic        | 0.0000049570      |          |
| MD18 <sup>b</sup> | GP | <i>LMNA</i>   | NM_170707.4 | c.364A>G         | p.Lys122Glu          | Homozygous            | Maternal allele (heterozygous) | AR | -                          | Likely pathogenic | 0.0000018590      | Positive |
| MD19 <sup>b</sup> | GP | <i>LMNA</i>   | NM_170707.4 | c.364A>G         | p.Lys122Glu          | Homozygous            | Maternal allele (heterozygous) | AR | -                          | Likely pathogenic | 0.0000018590      | Positive |
| MD20              | ES | <i>LMNA</i>   | NM_170707.4 | c.1072G>A        | p.Glu358Lys          | Heterozygous          | De novo                        | AD | [PMID: 34487530]           | Pathogenic        | Variant not found | Positive |

|                               |    |                |             |                |                  |                       |                                      |       |                            |                   |                   |          |
|-------------------------------|----|----------------|-------------|----------------|------------------|-----------------------|--------------------------------------|-------|----------------------------|-------------------|-------------------|----------|
| MD21                          | ES | <i>LMNA</i>    | NM_170707.4 | c.1153_1155del | p.Glu385del      | Heterozygous          | De novo                              | AD    | [PMID: 36697461]           | Pathogenic        | Variant not found | Positive |
| MD22                          | ES | <i>LMNA</i>    | NM_170707.4 | c.1381-2A>C    | p.?              | Heterozygous          | De novo                              | AD    | [PMID: 36697461]           | Pathogenic        | Variant not found | Positive |
| MM1                           | ES | <i>GAA</i>     | NM_000152.5 | c.1556T>C      | p.Met519Thr      | Compound heterozygous | Maternal allele                      | AR    | [PMID: 33741225]           | Pathogenic        | 0.0000030980      | Positive |
|                               |    | <i>GAA</i>     | NM_000152.5 | c.1561G>A      | p.Glu521Lys      |                       | Paternal allele                      |       | [PMID: 26830551]           | Pathogenic        | 0.0000037180      |          |
| MM2                           | ES | <i>GBE1</i>    | NM_000158.4 | c.1496T>A      | p.Met499Lys      | Compound heterozygous | Maternal allele                      | AR    | [PMID: 36697461]           | Likely pathogenic | 0.0000006210      | Positive |
|                               |    | <i>GBE1</i>    | NM_000158.4 | c.1561A>T      | p.Lys521*        |                       | Paternal allele                      |       | [PMID: 20058079, 36697461] | Pathogenic        | 0.0000006210      |          |
| MC1                           | GP | <i>CLCN1</i>   | NM_000083.3 | c.871G>A       | p.Glu291Lys      | Heterozygous          | Not confirmed                        | AR    | [PMID: 27415035]           | Pathogenic        | 0.0000049570      | Probable |
|                               |    | <i>CLCN1</i>   | NM_000083.3 | c.991G>T       | p.Ala331Ser      | Heterozygous          | Not confirmed                        |       | [PMID: 34529042]           | Likely pathogenic | 0.0000037230      |          |
| MC2                           | ES | <i>CLCN1</i>   | NM_000083.3 | c.913G>A       | p.Gly305Arg      | Heterozygous          | Not confirmed                        | AD    | -                          | Likely pathogenic | Variant not found | Probable |
| Group 2: Inherited neuropathy |    |                |             |                |                  |                       |                                      |       |                            |                   |                   |          |
| CMT1                          | GP | <i>DYNC1H1</i> | NM_001376.5 | c.1756T>C      | p.Ser586Pro      | Heterozygous          | Assumed de novo (maternal confirmed) | AD    | -                          | Likely pathogenic | Variant not found | Probable |
| CMT2                          | ES | <i>EGR2</i>    | NM_000399.5 | c.925C>T       | p.Arg309Trp      | Heterozygous          | Not confirmed                        | AD    | [PMID:16198564]            | Pathogenic        | Variant not found | Positive |
| CMT3                          | ES | <i>EGR2</i>    | NM_000399.5 | c.1231G>A      | p.Asp411Asn      | Heterozygous          | Not confirmed                        | AD    | [PMID: 27549087]           | Likely pathogenic | Variant not found | Probable |
| CMT4                          | GP | <i>GAN</i>     | NM_022041.4 | c.27C>G        | p.Asp9Glu        | Compound heterozygous | Maternal allele                      | AR    | -                          | VUS               | 0.0000006520      | Possible |
|                               |    | <i>GAN</i>     | NM_022041.4 | c.1629C>A      | p.Tyr543*        |                       | Paternal allele                      |       | -                          | VUS               | Variant not found |          |
| CMT5                          | ES | <i>GDAP1</i>   | NM_018972.4 | c.368A>G       | p.His123Arg      | Heterozygous          | Assumed de novo (maternal confirmed) | AD    | [PMID: 26525999]           | Pathogenic        | 0.0000006200      | Probable |
| CMT6                          | ES | <i>GDAP1</i>   | NM_018972.4 | c.368A>G       | p.His123Arg      | Heterozygous          | Not confirmed                        | AD    | [PMID: 26525999]           | Pathogenic        | 0.0000006200      | Probable |
| CMT7                          | GP | <i>GDAP1</i>   | NM_018972.4 | c.445G>T       | p.Asp149Tyr      | Compound heterozygous | Maternal allele                      | AR    | [PMID: 15469949]           | Pathogenic        | 0.0000006200      | Positive |
|                               |    | <i>GDAP1</i>   | NM_018972.4 | c.482G>A       | p.Arg161His      |                       | Not confirmed                        |       | [PMID:37058526, 12843336]  | Pathogenic        | 0.0000024810      |          |
| CMT8                          | ES | <i>GDAP1</i>   | NM_018972.4 | c.533del       | p.Asn178Thrfs*28 | Heterozygous          | Not confirmed                        | AD/AR | -                          | Likely pathogenic | Variant not found | Possible |
| CMT9                          | ES | <i>GDAP1</i>   | NM_018972.4 | c.767A>G       | p.His256Arg      | Heterozygous          | Paternal allele                      | AD/AR | [PMID: 21692914]           | Pathogenic        | 0.0000037170      | Probable |
| CMT10                         | ES | <i>GJB1</i>    | NM_000166.6 | c.223C>T       | p.Arg75Trp       | Hemizygous            | Not confirmed                        | XR    | [PMID: 12460545]           | Pathogenic        | 0.0000000000      | Positive |
| CMT11                         | ES | <i>HK1</i>     | NM_000188.3 | c.1A>G         | p.Met1?          | Homozygous            | Not confirmed                        | AR    | -                          | Likely pathogenic | 0.0000006270      | Positive |
| CMT12                         | ES | <i>IGHMBP2</i> | NM_002180.3 | c.547+1G>A     | p.?              | Heterozygous          | Not confirmed                        | AR    | [PMID: 28902413]           | Pathogenic        | 0.0000093590      | Possible |
| CMT13                         | ES | <i>IGHMBP2</i> | NM_002180.3 | c.983_987del   | p.Lys328Thrfs*46 | Heterozygous          | Not confirmed                        | AR    | [PMID: 27450922]           | Pathogenic        | 0.0000316000      | Possible |
| CMT14                         | ES | <i>IGHMBP2</i> | NM_002180.3 | c.2773del      | p.His925Thrfs*53 | Compound heterozygous | Maternal allele                      | AR    | -                          | Pathogenic        | Variant not found | Positive |
|                               |    | <i>IGHMBP2</i> | NM_002180.3 | c.2362C>T      | p.Arg788*        |                       | Paternal allele                      |       | [PMID: 29761130]           | Pathogenic        | 0.0000247900      |          |
| CMT15                         | ES | <i>MFN2</i>    | NM_014874.4 | c.272T>G       | p.Val91Gly       | Heterozygous          | Assumed de novo (paternal confirmed) | AD    | -                          | Pathogenic        | Variant not found | Probable |
| CMT16                         | ES | <i>MFN2</i>    | NM_014874.4 | c.280C>T       | p.Arg94Trp       | Heterozygous          | Not confirmed                        | AD    | [PMID: 19889647]           | Pathogenic        | Variant not found | Probable |
| CMT17                         | ES | <i>MFN2</i>    | NM_014874.4 | c.430G>A       | p.Ala144Thr      | Compound heterozygous | Maternal allele                      | AR    | -                          | Likely pathogenic | Variant not found | Positive |
|                               |    | <i>MFN2</i>    | NM_014874.4 | c.707C>T       | p.Thr236Met      |                       | Paternal allele                      |       | [PMID: 30373780]           | Pathogenic        | 0.0000018590      |          |

|                                         |    |        |                |                      |                  |                       |                                      |    |                            |                   |                   |          |
|-----------------------------------------|----|--------|----------------|----------------------|------------------|-----------------------|--------------------------------------|----|----------------------------|-------------------|-------------------|----------|
| CMT18                                   | ES | MFN2   | NM_014874.4    | c.472A>C             | p.Lys158Gln      | Heterozygous          | Maternal allele                      | AD | -                          | VUS               | Variant not found | Possible |
| CMT19                                   | ES | MFN2   | NM_014874.4    | c.617C>T             | p.Thr206Ile      | Heterozygous          | Maternal allele                      | AD | [PMID: 16437557]           | Pathogenic        | Variant not found | Positive |
| CMT20                                   | ES | MFN2   | NM_014874.4    | c.817G>T             | p.Val273Leu      | Heterozygous          | Paternal allele                      | AD | [PMID: 18316077, 16043786] | Likely pathogenic | Variant not found | Positive |
| CMT21                                   | ES | MFN2   | NM_014874.4    | c.1071G>C            | p.Lys357Asn      | Heterozygous          | Not confirmed                        | AD | [PMID: 15549395]           | pathogenic        | Variant not found | Probable |
| CMT22                                   | ES | MFN2   | NM_014874.4    | c.1090C>T            | p.Arg364Trp      | Heterozygous          | De novo                              | AD | [PMID: 28063088]           | Pathogenic        | Variant not found | Positive |
| CMT23                                   | ES | MFN2   | NM_014874.4    | c.1090C>T            | p.Arg364Trp      | Heterozygous          | De novo                              | AD | [PMID: 28063088]           | pathogenic        | Variant not found | Positive |
| CMT24                                   | ES | MFN2   | NM_014874.4    | c.1091G>C            | p.Arg364Pro      | Heterozygous          | Not confirmed                        | AD | [PMID: 20008656]           | Pathogenic        | Variant not found | Probable |
| CMT25                                   | ES | MPZ    | NM_000530.8    | c.245A>G             | p.Tyr82Cys       | Heterozygous          | De novo                              | AD | [PMID: 9633821]            | Pathogenic        | Variant not found | Positive |
| CMT26                                   | GP | MPZ    | NM_000530.8    | c.292C>T             | p.Arg98Cys       | Heterozygous          | Assumed de novo (maternal confirmed) | AD | [PMID: 30785048]           | pathogenic        | 0.0000018590      | Probable |
| CMT27                                   | ES | MPZ    | NM_000530.8    | c.551del             | p.Leu184Hisfs*68 | Heterozygous          | De novo                              | AD | -                          | Pathogenic        | Variant not found | Positive |
| CMT28                                   | ES | NEFL   | NM_006158.5    | c.280C>T             | p.Leu94Phe       | Heterozygous          | De novo                              | AD | [PMID: 27862672]           | Likely pathogenic | Variant not found | Positive |
| CMT29                                   | ES | NEFL   | NM_006158.5    | c.280C>T             | p.Leu94Phe       | Heterozygous          | De novo                              | AD | [PMID: 27862672]           | Likely pathogenic | Variant not found | Positive |
| CMT30                                   | ES | NEFL   | NM_006158.5    | c.293A>G             | p.Asn98Ser       | Heterozygous          | De novo                              | AD | [PMID: 31393079]           | Pathogenic        | Variant not found | Positive |
| CMT31                                   | ES | PMP2   | NM_002677.5    | c.155T>C             | p.Ile52Thr       | Heterozygous          | Assume de novo (maternal confirmed)  | AD | [PMID: 30249361]           | Pathogenic        | Variant not found | Probable |
| CMT32                                   | ES | PMP22  | NM_000304.4    | c.215C>T             | p.Ser72Leu       | Heterozygous          | De novo                              | AD | [PMID: 37296061]           | Pathogenic        | Variant not found | Positive |
| CMT33                                   | ES | PMP22  | NM_000304.4    | c.251_253del         | p.Phe84del       | Heterozygous          | Not confirmed                        | AD | [PMID: 11355152]           | Pathogenic        | Variant not found | Probable |
| CMT34                                   | ES | SH3TC2 | NM_024577.4    | c.929dup             | p.Ser312Valfs*18 | Homozygous            | Paternal and maternal allele         | AR | -                          | Pathogenic        | Variant not found | Positive |
| HSN1                                    | GP | KIF1A  | NM_001244008.2 | c.2840del            | p.Leu947Argfs*4  | Homozygous            | Paternal and maternal allele         | AR | [PMID: 21820098]           | Pathogenic        | 0.0000129000      | Positive |
| HSN2                                    | ES | PRDM12 | NM_021619.3    | c.570+2T>G           | p.?              | Compound heterozygous | Paternal allele                      | AR | -                          | pathogenic        | 0.0000020080      | Positive |
|                                         |    | PRDM12 | NM_021619.3    | c.796A>C             | p.Thr266Pro      |                       | Maternal allele                      |    | -                          | Likely pathogenic | Variant not found |          |
| Group 3: Congenital myasthenic syndrome |    |        |                |                      |                  |                       |                                      |    |                            |                   |                   |          |
| CMS1                                    | GP | CHRNE  | NM_000080.4    | c.865C>T             | p.Leu289Phe      | Heterozygous          | De novo                              | AD | [PMID: 27779167]           | pathogenic        | Variant not found | Positive |
| CMS2                                    | ES | COLQ   | NM_005677.4    | c.393+1G>A           | p.?              | Homozygous            | Paternal and maternal allele         | AR | [PMID: 34912755]           | Pathogenic        | 0.0000006200      | Positive |
| CMS3                                    | ES | COLQ   | NM_005677.4    | c.393+1G>A           | p.?              | Homozygous            | Paternal and maternal allele         | AR | [PMID: 34912755]           | Pathogenic        | 0.0000006195      | Positive |
| CMS4                                    | ES | COLQ   | NM_005677.4    | c.393+1G>A           | p.?              | Homozygous            | Not confirmed                        | AR | [PMID: 34912755]           | Pathogenic        | 0.0000006195      | Positive |
| CMS5                                    | ES | COLQ   | NM_005677.4    | c.393+1G>A           | p.?              | Homozygous            | Not confirmed                        | AR | [PMID: 34912755]           | Pathogenic        | 0.0000006195      | Positive |
| CMS6                                    | GP | COLQ   | NM_005677.4    | c.393+1G>A           | p.?              | Heterozygous          | Not confirmed                        | AR | [PMID: 34912755]           | Pathogenic        | 0.0000006195      | Probable |
|                                         |    | COLQ   | NM_005677.4    | c.1281C>T            | p.Cys427=        | Heterozygous          | Not confirmed                        |    | [PMID: 38003406]           | Likely pathogenic | 0.0000055880      |          |
| CMS7                                    | GP | COLQ   | NM_005677.4    | deletion exons 14-15 |                  | Homozygous            | Not confirmed                        | AR | [PMID: 34912755]           | Pathogenic        | -                 | Positive |
| CMS8 <sup>b</sup>                       | GP | RAPSN  | NM_005055.5    | c.-210A>G            | p.?              | Heterozygous          | Not confirmed                        | AR | [PMID: 22326364]           | Pathogenic        | 0.0000182700      | Probable |
|                                         |    | RAPSN  | NM_005055.5    | c.484G>A             | p.Glu162Lys      | Heterozygous          | Not confirmed                        |    | [PMID: 28495245]           | Pathogenic        | 0.0000080580      |          |
| CMS9 <sup>b</sup>                       | ES | RAPSN  | NM_005055.5    | c.484G>A             | p.Glu162Lys      | Heterozygous          | Paternal allele                      | AR | [PMID: 28495245]           | Pathogenic        | 0.0000080580      | Probable |

|                               |    |              |             |              |             |                       |                 |    |                  |            |                   |          |
|-------------------------------|----|--------------|-------------|--------------|-------------|-----------------------|-----------------|----|------------------|------------|-------------------|----------|
| CMS10                         | ES | <i>RAPSN</i> | NM_005055.5 | c.752_754del | p.Cys251del | Compound heterozygous | Paternal allele | AR | -                | VUS        | Variant not found | Possible |
|                               |    | <i>RAPSN</i> | NM_005055.5 | c.844C>T     | p.Arg282Cys |                       | Maternal allele | AR | -                | VUS        | 0.0000037270      |          |
| Group 4: Motor neuron disease |    |              |             |              |             |                       |                 |    |                  |            |                   |          |
| MND1                          | ES | <i>ASAH1</i> | NM_177924.5 | c.1A>T       | p.Met1?     | Compound heterozygous | Paternal allele | AR | -                | Pathogenic | 0.0000112600      | Positive |
|                               |    | <i>ASAH1</i> | NM_177924.5 | c.770T>C     | p.Leu257Pro |                       | Maternal allele |    | [PMID: 30029679] | Pathogenic | Variant not found |          |

**Abbreviations:** AD, autosomal dominant; AF, allele frequency; AR, autosomal recessive; ES, exome sequencing; gnomAD, genome aggregation database (v4.1.0); GP, gene panel; MOI, mode of inheritance; VUS, variant of uncertain significance; XR, X-linked recessive

<sup>a</sup> According to the American College of Medical Genetics and Genomics interpretation guidelines [PMID: 25741868]

<sup>b</sup> Sibling relationship

In the patient-ID column, CM, DMD, MD, MM, MC, CMT, HSN, CMS and MND represent patients clinically diagnosed with congenital myopathies, Duchenne muscular dystrophy, other muscular dystrophies, metabolic/mitochondrial myopathies, muscle channelopathy, Charcot–Marie–Tooth disease, hereditary sensory neuropathy, congenital myasthenic syndrome, and motor neuron disease, respectively.