| Patient number | Sex | age onset / age at WES request | Reason | Disease causing gene(s) | Intragenic /# of genes | Variant^ | inheritance | Classification* | Likely diagnosis | OMIM disease |
|---|---|---|---|---|---|---|---|---|---|---|
| **Inheritance pattern of the involved variant/gene: AD** | | | | | | | | | | |
| CD1 | F | ? / 44 | neuropathy | *DCTN1* | > 20 | seq[GRCh37] 2p13.2p13.2(74300675_74909185)x3<br>NC_000002.11:g.(74273449_74300675)_(74909185_75059781)dup | nd | VOUS | uncertain | Distal hereditary motor neuronopathy type VIIB (#607641) |
| CD2 | M | ? / 72 | neuropathy | *PMP22* | *7* | seq[GRCh37] 17p12p12( 14095305)_(15477522)x1<br>NC_000017.10:g.(14005599_14095305)_(15477522_15492232)del | nd | P | y | Neuropathy, recurrent, with pressure palsies #162500) |
| CD3 | F | ? / 44 | HMSN / HNPP | *PMP22* | *7* | seq[GRCh37] 17p12p12( 14095305)_(15477522)x1<br>NC_000017.10:g.(14005599_14095305)_(15477522_15492232)del | nd | P | y | Neuropathy, recurrent, with pressure palsies #162500) |
| CD4 | F | <10 / 64 | diffuse paresis distal> proximal, pes cavus | *PMP22* | *7* | seq[GRCh37] 17p12p12( 14095305)_(15477522)x3<br>NC_000017.10:g.(14005599_14095305)_(15477522_15492232)dup | nd | P | y | Charcot-Marie-Tooth disease 1A (#1118220 ) |
| CD5 | F | ? / 60 | neuropathy | *PMP22* | *7* | seq[GRCh37] 17p12p12( 14095305)_(15477522)x3<br>NC_000017.10:g.(14005599_14095305)_(15477522_15492232)dup | nd | P | y | Charcot-Marie-Tooth disease 1A (#1118220 ) |
| CD6 | F | ? / 61 | Charcot-Marie-Tooth | *PMP22* | *7* | seq[GRCh37] 17p12p12( 14095305)_(15477522)x3<br>NC_000017.10:g.(14005599_14095305)_(15477522_15492232)dup | nd | P | y | Charcot-Marie-Tooth disease 1A (#1118220 ) |
| CD7 | F | 7 / 27 | HMSN type I | *PMP22* | *7* | seq[GRCh37] 17p12p12( 14095305)_(15477522)x3<br>NC_000017.10:g.(14005599_14095305)_(15477522_15492232)dup | nd | P | y | Charcot-Marie-Tooth disease 1A (#1118220 ) |
| CD8 | M | ? / 68 | HMSN type I | *PMP22* | *7* | seq[GRCh37] 17p12p12( 14095305)_(15477522)x3<br>NC_000017.10:g.(14005599_14095305)_(15477522_15492232)dup[c] | nd | P | y | Charcot-Marie-Tooth disease 1A (#1118220 ) |
| CD9 | M | 30 / 49 | neuropathy, muscle atrophy, distal muscle weakness, hollow feet | *PMP22* | *7* | seq[GRCh37] 17p12p12( 14095305)_(15477522)x3<br>NC_000017.10:g.(14005599_14095305)_(15477522_15492232)dup | nd | P | y | Charcot-Marie-Tooth disease 1A (#1118220 ) |
| CD10 | F | 26 / 60 | HMSN type II | *PMP22* | *7* | seq[GRCh37] 17p12p12( 14095305)_(15477522)x3<br>NC_000017.10:g.(14005599_14095305)_(15477522_15492232)dup | nd | P | y | Charcot-Marie-Tooth disease 1A (#1118220 ) |
| CD11 | F | ? / 50 | polyneuropathy | *PMP22* | *7* | seq[GRCh37] 17p12p12( 14095305)_(15477522)x3<br>NC_000017.10:g.(14005599_14095305)_(15477522_15492232)dup | nd | P | y | Charcot-Marie-Tooth disease 1A (#1118220 ) |
| CD12 | M | ? / 5 | HMSN type 1A, hypotonia, ptosis, ceroid lipofuscinosis | *PMP22* | *7* | seq[GRCh37] 17p12p12( 14095305)_(15477522)x3<br>NC_000017.10:g.(14005599_14095305)_(15477522_15492232)dup | nd | P | y | Charcot-Marie-Tooth disease 1A (#1118220 ) |
| CD13 | M | ? / 44 | HMSN type I | *PMP22* | *7* | seq[GRCh37] 17p12p12( 14095305)_(15477522)x3<br>NC_000017.10:g.(14005599_14095305)_(15477522_15492232)dup | nd | P | y | Charcot-Marie-Tooth disease 1A (#1118220 ) |
| CD14 | M | ? / 64 | neuropathy | *PMP22* | *7* | seq[GRCh37] 17p12p12( 14095305)_(15477522)x3<br>NC_000017.10:g.(14005599_14095305)_(15477522_15492232)dup | nd | P | y | Charcot-Marie-Tooth disease 1A (#1118220 ) |
| **Inheritance: AR** | | | | | | | | | | |
| CR1 | M | ? / 7 | neuropathy, motor developmental delay | *FARS2* | *2* | seq[GRCh37] 6p25.1p25.1(5216764_5545601)x1<br>NC_000006.11:g.(5144510_5216764)_(5545601_5613394)del<br>exon 1-5 (NM_006567.5)<br>*no second variant* | nd | LP | uncertain | autosomal recessive spastic paraplegia 77 (#617046) |

Genome assembly GRCh37 (hg19)

c= confirmed by MLPA/array/sequencing/deletion PCR, see suppl. File 2

*Based on ACMG guidelines. P=Pathogenic, LP=Likely Pathogenic, VOUS = Variant of Unknown Significance

# in some cases CNV's may extend into other sequences beyond the genes (i.e. intergenic or non-coding genes)