**Table 4. Novel variants identified in the *MTMR13* gene.**

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| **Nucleotide variant** | **Protein variant** | **Patient ID** | **Patient Origin** | **Haplotype** | **GnomAD****Allele Frequency (AF)** | **Provean** | **Polyphen-2** |
| c.16\_19del | p.D6Tfs\*5 | 2-A | Turkish\* | Homoz | 0 |  |  |
| c.184C>T | p.Q62\* | 2-G | Italian | Homoz | 0 |  |  |
| c.1066 C>T | p.R356\* | 2-S | Albanian | Biallelic, segregation N/A | 8.041e-6 |  |  |
| c.3800 G>A | p.W1267\* | 2-S | Albanian | Biallelic, segregation N/A | 0 |  |  |
| c.3563C>A | p.S1188\* | 2-B | Maltese | Homoz | 0 |  |  |
| c.4430A>C | p.D1477G | 2-J | Maghreb | Biallelic, *in trans* | 0 | Deleterious-6.351 | Probably damaging 1.000 |
| c.862-1G>A | Splicing | 2-J | Maghreb | Biallelic, *in trans* | 0 |  |  |
| c.4499delT | p.L1500Wfs\*22 | 2-C, 2-D | Italian | Homoz | 0 |  |  |
| c.620G>T | p.G207V | 2-R | British | Biallelic, *in trans* | 0 | Deleterious-7.556 | Probably damaging 1.000 |
| c.2536+1G>A | Splicing | 2-R | British | Biallelic, *in trans* | 0 |  |  |
| c.161G>A | p.W54\* | 2-I | British | Biallelic, *in trans* | 3.987e-6 |  |  |
| c.1718delC | p.P537Lfs\* | 2-I | British | Biallelic, *in trans* | 0 |  |  |
| c.2457delT | p.T820Pfs\*24 | 2-K | USA | Biallelic, *in trans* | 0 |  |  |
| [hg19] 11p15.4(9,829,246 - 9,917,849) x3 | Ex 17-32dup | 2-K | USA | Biallelic, *in trans* | 0 |  |  |
| c.2215G>A | p.E739K | 2-O | British | Biallelic, *in trans* | 0 | Deleterious-3.797 | Probably damaging 0.972 |
| [hg19] 11p15.4 (9,853,777 – 10,003,829) | Ex 14-27del | 2-O | British | Biallelic, *in trans* | 0 |  |  |
| c. 4009\_4016del | p.E1337Sfs\*3 | 2-Q | Pakistani | Homoz | 0 |  |  |
| c.5041C>T | p.Q1681\* | 2-N | British | Biallelic, *in trans* | 3.191e-5 |  |  |
| c.5037+1G>C | Splicing | 2-N | British | Biallelic, *in trans* | 0 |  |  |

*Legend*: Novel variants (AF=0, not reported) and novel disease association of known rare variants (AF<0.01), which have not previously associated with CMT4B and identified in the *MTMR13* gene. **\***Details pertaining to patient 2-A will be included in a manuscript in preparation. Biallelic, *in trans,* was inferred on the basis of the segregation analysis from the parents. N/A= Not assessed.