**Table e-4: Genetic features in 17 patients with variants of unknown significance (VUS)** M: men, F: women, y: years

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **ID** | **Sex/age** | **Gene** | **Mutations**  **cDNA level Protein level** | **Reference** |
| V1 | M/17y | *CAPN3* | c.1986\_1988del/- p.Gly663del/- | **This work** |
| V2 | F/68 y | *CAPN3* | c.755T>G/- p.Met252Arg/- | [1] |
| V3 | F/65 y | *LDB3*  *DNAJB6* | c.2107G>A/- p.Ala703Thr/-  c.116\_118del/- p.Glu41del/- | **This work**  **This work** |
| V4 | F/24y | *RYR1* | c.10747G>C/- p.Glu3583Gln/- | [2] |
| V5 | M/11 y | *CPT2* | c.614A>G/- p.Tyr205Cys/- | **This work** |
| V6 | M/35 | *DYSF* | c.383G>A/- p.Gly128Glu/- | [3] |
| V7 | F/53 y | *SLC22A5*  *ANO5* | c.364G>T/c.1535G>A p.Asp122Tyr/p.Arg512His  c.692G>T/- p.Gly231Val/- | [4]; **This work**  [5] |
| V8 | M/74 y | *PLEC* | c.1324G>A/c.92G>A p.Val442Met/p.Arg31Gln | **This work** |
| V9 | M/23 y | *TMEM43*  *TRAPPC11* | c.413A>G/- p.Gln138Arg/-  c.1702C>T/c.2388A>C p.Arg568\*/p.Gly796Gly | **This work**  **This work** |
| V10 | M/40 y | *CPT2* | c.338C>T p.Ser113Leu | [6] |
| V11 | M/16 y | *ACADS*  *SEPN1* | c.1231C>T/c.625G>A p.Arg411Trp/p.Gly209Ser  c.1520A>G/- p.Asn507Ser/- | [7]; [8]  **This work** |
| V12 | M/63 y | *ANO5* | c.155A>G/- p.Asn52Ser/- | [9] |
| V13 | M/62 y | *MYH7* | c.3133C>T/- p.Arg1045Cys/- | [10] |
| V14 | M/64 y | *CAPN3* | c.526G>A/- p.Val176Met/- | **This work** |
| V15 | M/22 y | *DYSF* | c.3023G>A/c.3950G>T p.Arg1008Gln/p.Arg1317Leu | **This work** |
| V16 | F/72 y | *RYR1* | c.4999C>T/- p.Arg1667Cys/- | dbSNP entry rs144157950 |
| V17 | M/17 y | *ANO5* | c.1519delT p.Phe507Serfs\*6 | [11] |

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