**Supplementary Table 1.** Variants located in homozygous stretches detected in one of the patients. We filtered for variants with a high or moderate impact on the encoded protein and allele frequency below 0.01 in gnomAD database.

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| **Chromosome** | **Gene** | **Transcript ID** | **Impact** | **Variant** | **Amino Acid Change** | **gnomAD (allele frequency)** | **Number of Homozygotes** |
| 2 | *LRP2* | ENST00000263816 | MODERATE | c.4351G>T | p.Val1451Phe | 0.0019903 | 0 |
| 6 | *F13A1* | ENST00000264870 | MODERATE | c.1766T>A | p.Leu589Gln | 0.00377346 | 4 |
| 10 | *SEC31B* | ENST00000370345 | MODERATE | c.2041G>A | p.Glu681Lys | 0.00031705 | 0 |
| 10 | *PPRC1* | ENST00000278070 | MODERATE | c.2501C>G | p.Pro834Arg | 0.00219339 | 0 |
| 10 | *CCDC186* | ENST00000369287 | HIGH | c.2215C>T | p.Arg739Ter | 0.00000406 | 0 |
| 10 | *TDRD1* | ENST00000251864 | MODERATE | c.134G>A | p.Gly45Glu | 0.00004062 | 0 |
| 12 | *RERG* | ENST00000537647 | MODERATE | c.196A>G | p.Asn66Asp | 0.00311562 | 2 |
| 17 | *KRT39* | ENST00000355612 | MODERATE | c.1414A>G | p.Lys472Glu | 0.00029270 | 0 |
| 17 | *KRTAP2-3* | ENST00000391418 | MODERATE | c.275G>A | p.Cys92Tyr | 0 | 0 |
| 17 | *HIGD1B* | ENST00000253410 | MODERATE | c.19T>C | p.Trp7Arg | 0.0021698 | 10 |
| X | *DUSP9* | ENST00000342782 | MODERATE | c.455C>T | p.Pro152Leu | 0.00035897 | 36 |