**Table S1 -** Potentially damaging variants present in affected patients, but not in the healthy ones.

| Gene | Chr | Position | DNA & AA change | gnomAD MAF (%) | *In silico* prediction tools | CADD score |
|---|---|---|---|---|---|---|
| *LINGO4* | 1 | 151774279 | NM_001004432:c.902G>A:p.(Arg301Gln) | 0,026 | 10/18 damaging | 28,5 |
| *RNF149* | 2 | 101911601 | NM_173647:c.503G>T:p.(Gly168Val) | 0,001 | 9/12 damaging | 29,8 |
| *C2orf40* | 2 | 106690387 | NM_032411:exon3:c.C173T:p.A58V | 0,0004 | 9/11 damaging | 24,6 |
| *DBI* | 2 | 120125869 | NM_001079862:c.115G>A:p.(Asp39Asn) | 0,466 | . | 24,4 |
| *COL23A1* | 5 | 177673296 | NM_173465:c.C1372C>T:p.(Pro458Ser) | 0,062 | 8/12 damaging | 23,9 |
| *JARID2* | 6 | 15496817 | NM_004973:c.1361A>C:p.(Gln454Pro) | . | 8/12 damaging | 22,7 |
| *MDC1* | 6 | 30681083 | NM_014641:c.618_636del:p.(Gly207ProfsTer3) | 0,21 | 1/1 damaging | 24 |
| *UPK2* | 11 | 118827665 | NM_006760:c.149C>A:p.(Pro50His) | 0,049 | 12/20 damaging | 26,3 |
| *KIFC3* | 16 | 57803855 | NM_005550:c.952G>A:p.(Glu318Lys) | 0,024 | 10/12 damaging | 27,7 |
| *ABCA9* | 17 | 66981048 | NM_080283:c.4357C>A:p.(Pro1453Thr) | 0,41 | 8/11 damaging | 24 |
| *GIPR* | 19 | 46181024 | NM_000164:c.911T>G:p.(Leu304Arg) | 0,016 | 8/12 damaging | 28,2 |
| *EHD2* | 19 | 48239657 | NM_014601:c.947A>G:p.(Lys316Arg) | 0,011 | 14/19 damaging | 22,8 |
| *EMILIN3* | 20 | 39990792 | NM_052846:c.1417C>T:p.(Arg473Cys) | 0,006 | 11/19 damaging | 26,3 |
| *PREX1* | 20 | 47307634 | NM_020820:c.1037C>T:p.(Ala346Val) | 0,001 | 10/19 damaging | 28,1 |
| *GATD3* | 21 | 45560145 | NM_004649:c.442G>A:p.(Val148Met) | 0,95 | 6/9 damaging | 23,5 |
| *HDAC10* | 22 | 50686379 | NM_032019:c.1277C>T:p.(Pro426Leu) | . | . | 24 |

*Chr = Chromosome*

*AA change = Aminoacid change*

*GnomAD MAF = Minor allele frequency of the identified variant in the population reported by the GnomAD database*

*CADD score = deleteriousness score of single nucleotide variants and insertion/deletion variants in the human genome*