Supplementary Figure 1. List of 4 likely pathogenic germline variants.

| Chr | Position | Reference Allele | Sample Allele | Gene Region | Gene Symbol | Protein Variant | Read Depth | Allele Fraction | Translation Impact | SIFT Function Prediction | PolyPhen-2 Function Prediction | CADD Score | dbSNP ID | HGMD | CLINVAR ID | COSMIC ID |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 2 | 97241317 | T | G | Exonic | ANKRD36 | p.L1382* | 110 | 34.55 | stop gain | | | 29.5 | | | | |
| 3 | 75737676 | A | T | Promoter; Exonic; Intronic | ZNF717; MIR4273 | p.C599*; p.C649* | 534 | 48.31 | stop gain | | | 32 | 79635065 | | | 5021420 |
| 6 | 32040723 | G | A | Exonic | CYP21A2 | p.A362T; p.A257T; p.A392T | 572 | 20.28 | missense | Damaging | Probably Damaging | 23.9 | 202242769 | CM071683 (DM) | RCV000490344.5; RCV001357310.6 | 6353585 |
| 10 | 1.25E+08 | C | T | Exonic | CTBP2 | p.R103Q; p.R643Q; p.R171Q | 421 | 37.77 | missense | Tolerated | Possibly Damaging | 30 | 760489730 | | | 5620575 |