Supplement Table S2. Novel disease-causing variant identified in *TTLL5* (NM\_015072.4) by whole-exome sequencing after filtering. HOM\_mut, homozygous mutated; HTZ, heterozygous mutated.

|  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Sample** | **Gene** | **refseq** | **ProteinVariation** | **HGVSc** | **Impact** | **Consequence** | **EVS Variant Freq (%)** | **1000G EURVariant Freq (%)** | **Depth****Used** | **FrequencyMutant Allele** |
| I-1 | *TTLL5* | NM\_015072.4 | p.(Glu61Aspfs\*19) | c.182-3\_182-1delinsAA | HIGH | Splice AcceptorVariant | 0 | 0 | 124 | 48.39 |
| I-2 | 132 | 40.15 |
| II-3 | 141 | 98.58 |