





Supplementary Figure S2. Overall features of variants compiled in this study. A) Bar chart showing the percentage of protein-altering variants (PAVs) and protein-truncating variants (PTVs) and their types. Pie charts showing the percentage of variants according to their inheritance (B), detection method (C), and interpretation (D). CNV=Copy-number variant, ES=Exome sequencing, NGS=Next-generation sequencing, VUS= Variant of uncertain significance