



Supplementary Figure 1. Flow diagram of pathogenicity annotation of rare germline variants.

MAF: minor allele frequency. MAF is less than 0.005 (0.5%) in the Japanese individuals in the 38KJPN of the jMorp (Tohoku medical megabank organization [ToMMo]) genome database. P: pathogenic. LP: likely pathogenic. VUS: variant of uncertain significance. CIP: conflicting interpretation of pathogenicity. LB: likely benign. B: benign. NR: not reported. The final variant classification was performed following the guidelines of American College of Medical Genetics (ACMG).