Supplementary File 1

Class 5-Pathogenic: there is significant evidence to suggest that this variant is a dominant high-risk pathogenic variant with probability of pathogenicity >0.99. Class 4-Likely pathogenic: there is evidence that this variant is a dominant high-risk pathogenic variant with probability of pathogenicity between 0.95–0.99. Class 3-Uncertain: there is insufficient evidence to place this variant in Class 1, 2, 4 or 5 with probability of pathogenicity between 0.05–0.949. Class 2-Likely not pathogenic/little clinical significance: there is evidence against this variant being a dominant high-risk pathogenic variant with probability of pathogenicity between 0.001–0.049. Class1-Not pathogenic/low clinical significance: there is significant evidence against this variant being a dominant high-risk pathogenic variant with probability of pathogenicity <0.001. [1, 2]

1. Richards S, Aziz N, Bale S, Bick D, Das S, Gastier-Foster J, *et al*. ACMG Laboratory Quality Assurance Committee. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genet Med 2015;17:405-24. doi: 10.1038/gim.2015.30.
2. Richards CS, Bale S, Bellissimo DB, Das S, Grody WW, Hegde MR, *et al*. Molecular Subcommittee of the ACMG Laboratory Quality Assurance Committee. ACMG recommendations for standards for interpretation and reporting of sequence variations: Revisions 2007. Genet Med 2008;10:294-300. doi: 10.1097/GIM.0b013e31816b5cae.