Supplemental Data Table S1. Interpretation of three putative sequence variants based on the American College of Medical Genetics and Genomics and the Association for Molecular Pathology criteria

Sequence variant	Evidence of pathogenicity				Classification
	Very strong	Strong	Moderate	Supporting	
c.824G>A (p.Ala275Asp)		PS3	PM2, PM3	PP4, PP5	Pathogenic
c.135delC (p.Cys46Alafs*12)	PVS1		PM2, PM3	PP4, PP5	Pathogenic
c.337_353delinsG (p.Ile113Glyfs*4)	PVS1		PM2, PM3	PP4, PP5	Pathogenic