

Case	Zigosity	gDNA ⁽¹⁾	cDNA	protein	CADD	DANN	SIFT (Score)	PolyPhen2 Score	ClinVar (ID)	GnomAD ⁽²⁾	Other variants in same residue ⁽³⁾	PMID	ACMD Classification
A1	Homozygous (paternal/maternal)	2:32144980	c.1660A>G	p.(Lys554Glu)	23.6	0.9983	D (0.002)	PD (0.613)	Absent	Absent	Lys554_Ala556delinsThr	30476002	LP (PS3, PM1, PM2, PP3)
B1	Heterozygous (maternal)	2:32136880	c.1325A>T	p.(Glu442Val)	32	0.9935	D(0)	PD (1)	LP (945131)	Absent	Glu442Ala; Asp; Lys; STOP	16832076; 31157359; 17957230; 16240363; 25525159	LP (PM1, PM2, PM5, PP3, PP5)
	Heterozygous (<i>de novo</i>)	2:32154425	c.1780C>T	p.(Arg594Cys)	23.8	0.9954	B (0.054)	PD(0.906)	VUS (1417996)	0.0000075	Arg594fs	34008892; 26671083	LP (PS3, PM1, PM2, PP3)
C1	Heteroz	2:32136591	c.1274C>T	p.(Ala425Val)	29	0.9991	D (0.001)	PD (1)	Absent	Absent	Ala425Pro; fs	15841487; 30489674; 31594988; 34782662	LP (PM1, PM2, PM5, PP3)
	Heteroz	2:32136925	c.1370C>T	p.(Ala457Val)	31	0.9991	D (0.003)	PD (1)	Absent	Absent		25577683	LP (PM1, PM2, PP3, PP5)

(1) References to hg38; (2) gromAD 4.0 Accessed April 2025 (3) According to HGMD® Professional 2025.1 accesed April 2025