

Patients	hg38	NM_021930.6	NP_068749.3	gnomAD v4.1.0 global MAF
Proband	chr7:g.105551671G>C	c.1435G>C	p.(Ala479Pro)	No match in gnomADv4

‡ Thresholds 0.34|0.564 for Likely Benign, Ambiguous, Likely Pathogenic

* The closer to 0, the more intolerant to variation

** Raw score [0;10], 10: high impact

† Thresholds $\geq 0.2|0.5|0.8$ for impact

*** Threshold < 0.05 for Damaging

**** Thresholds $\geq 0.454|0.957$ for Possibly and Probably Damaging

***** Thresholds $\geq 0.447|0.909$ for Possibly and Probably Damaging

***** Threshold ≤ -1.5 for Damaging

***** Thresholds $0.2|0.5$ for Benign, Uncertain, Damaging

***** Threshold ≥ 0.5 for Damaging

***** Threshold ≥ 0 for Damaging (reliability index: 0-10), 10:high

***** Threshold ≥ 0.5 for Damaging (reliability index: 0-10), 10:high

***** Threshold ≥ 0.5 for Damaging

AlphaMissense‡	MetaDome*	CADD phred	Eigen phred	MPA score**	SPiP
0,985	0,40	28,2	10,62	7.0	No effect on splicing

spliceAI max score†	SIFT***	HumDiv****	Polyphen 2 HumVar*****	Fathmm*****	REVEL*****
0,00	0,006	1,0	0,995	0,75	0,714

ClinPred*****	Meta SVM*****	Meta LR*****	Mistic*****	ACMG
0,994	-0.354 (10)	0.3291 (10)	0,58	VUS (PM2, PP3)

References

novel