Patients	hg19	NM_000214.3	NP_000205.1	gnomAD v2.1.1 global MAF	MetaDome*	CADD phred	Eigen phred	MPA score**	SPiP	spliceAI max score†	SIFT***	HumDiv****	Polyphen 2 HumVar*****	Fathmm******	REVEL*******	ClinPred********	Meta SVM*********	Meta LR**********	Mistic***********	ACMG	References
10	chr20:g.10622218A>C	c.2806T>G	p.(Cys936Gly)	n/a	0,58	25	6,84	9,0	No effect on splicing	0	0,004	0,6	0,3	-3,81	0,902	1,000	1.0071 (10)	0.9001 (10)	0,94	LPat (PM2, PP3, PS2)	novel

* The closer to 0, the more intolerant to variation
** Raw score [0;10], 10: high impact
† Thresholds ≥ 0.2|0.5|0.8 for impact
*** Threshold < 0.05 for Damaging
**** Thresholds ≥ 0.454|0.957 for Possibly and Probably Damaging
***** Thresholds ≥ 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 (reliabilty index: 0-10), 10:high
********* Threshold ≥ 0.5 for Damaging (reliabilty index: 0-10), 10:high
********** Threshold ≥ 0.5 for Damaging