

Supplementary Table 2. Bioinformatic analyses performed on the missense *TBX3* variant p.S190R identified in Family B. All algorithms predicted a deleterious impact of the variant.

Chr	Start	End	Ref	Alt	Func.refGene	Gene.refGene	ExonicFunc.refGene	AAChange.refGene
chr12	115118771	115118771	G	T	exonic	TBX3	nonsynonymous SNV	TBX3:NM_005996:exon2:c.C570A:p.S190R , TBX3:NM_016569:exon2:c.C570A:p.S190R

SIFT_score	SIFT_converted_rankscore	SIFT_prediction	Polyphen2_HDIV_score	Polyphen2_HDIV_rankscore	Polyphen2_HDIV_prediction	Polyphen2_HVAR_score	Polyphen2_HVAR_rankscore	Polyphen2_HVAR_prediction
0	0,912	Deleterious	0,999	0,899	Deleterious	0,996	0,971	Deleterious
LRT_score	LRT_converted_Rank score	LRT_prediction	MutationTaster_score	MutationTaster_converted_Rank score	MutationTaster_prediction	MutationAssessor_score	MutationAssessor_rankscore	MutationAssessor_prediction
0	0,843	Deleterious	1	0,81	Deleterious	3,9	0,962	Deleterious
FATHMM_M_score	FATHMM_converted_rank score	FATHMM_prediction	PROVEAN_score	PROVEAN_converted_rankscore	PROVEAN_prediction	CADD_raw	CADD_raw_rankscore	CADD_prediction
-2,76	0,909	Deleterious	-4,98	0,821	Deleterious	5,372	0,728	25,9 Deleterious