

Table S1. Summary of deleteriousness prediction methods used in our study.

Name	Category	Threshold
SIFT	Function prediction	< 0.05
Polyphen-2	Function prediction	> 0.85
Mutation Taster	Function prediction	Disease causing o Disease causing automatic
PhyloP	Conservation score	> 0.7
CADD	Ensembl score	> 10
Grantham Score	Conservation score	> 100
PhastCons	Conservation score	> 0.7
GERP++	Conservation score	> 3
LRT	Function prediction	Deleterious
Mutation Assessor	Function prediction	High o Medium
LR	Ensembl score	Deleterious
FATHMM	Function prediction	Deleterious
RadialSVM	Ensembl score	Deleterious
SSF	Effect on splicing	> 70
MaxEnt	Effect on splicing	> 0
NNSPLICE	Effect on splicing	> 0.4
GeneSplicer	Effect on splicing	> 0
HSF	Effect on splicing	> 65
dbscSNV_RF_Score	Effect on splicing	> 0.6
dbscSNV_Ada_Score	Effect on splicing	> 0.6