Supplementary table 1: Prediction of pathogenicity

Mutation	c.23G>C	c.28C>T
Chromosomal location	chr6:33679441C>G	chr6:33679436G>A
Protein change	p.Arg8Pro	p.Leu10Phe
Polyphen-2 score (max. 1)	0.991	0.934
Polyphen-2 prediction	Probably damaging	Possibly damaging
SIFT score (cutoff<0.050)	0.034	0.009
SIFT prediction	Damaging	Damaging
Provean score (cutoff<-2.5)	-4.33	-2.88
Provean prediction	Deleterious	Deleterious
MutationTaster score (max. 1)	0.999997704040623	0.996059894177104
MutationTaster prediction	Disease causing	Disease causing
CADD score	25.2	27.3