

Supplemental Table S1: Classification of novel *BEST1* gene mutations

Variant (NM_004183.4)	Protein	MAF (gnomAD v2.1.1)	Bioinformatics	Classification
c.199_200del	p.(Leu67Valfs*164)	-	-	5
c.287_298del	p.(Gln96_Asn99del)	-	-	4
c.454C>T	p.(Pro152Ser)	-	Align GVGD (v2007): C0 (GV: 353.86 - GD: 0.00) SIFT (v6.2.0): Deleterious (score: 0, median: 2.92) MutationTaster (v2013): disease causing (prob: 1) PP2 HumVar: probably damaging with a score of 1.000	4
c.524del	p.(Ser175Thrfs*19)	-	-	5
c.590_615del	p.(Leu197Profs*26)	-	-	5
c.620T>A	p.(Leu207His)	-	Align GVGD (v2007): C0 (GV: 248.74 - GD: 67.97) SIFT (v6.2.0): Deleterious (score: 0.02, median: 2.92) MutationTaster (v2013): disease causing (prob: 1) PP2 HumVar: probably damaging with a score of 0.997	4