Mutation	Inheritance	Phenotype	Reference
Cys221Phe	AD	VMD	[1, 2]
Cys221Trp	AD	VMD	[1, 3]
Gly222Glu	AD	VMD	[1, 4, 5]
Gly222Val	AR	ARB	[6]
Gly222Val	AD	VMD	[7, 8]
Leu224Met	AD	VMD	[1, 9, 10]
Leu224Pro	AD	VMD	[1, 7, 11]
Leu224Gln	AR	ARB	This study
Tyr227Cys	AD	VMD	[6, 7, 10, 12-14]
Tyr227Phe	AD	VMD	[1, 15]
Tyr227Asn	AD	VMD	[1, 6, 7, 10, 13, 16-19]
Asp228Glu	AR	ARB	[6]
Asp228Glu	AD	VMD	[6, 11]
Trp229Gly	AD	VMD	[20]
Ile230Asn	AD	VMD	[1]
Ile230Thr	AD	VMD	[21]
Ser231Arg	AD	VMD	[1, 9, 10] Check kinnick
Ser231Thr	AD	VMD	[1, 5, 22]
Ile232Asn	AD	VMD	[1, 15]
Pro233Ala	AR	VMD	[23]
Pro233Leu	AD	VMD	[1, 24]
Pro233Gln	AD	VMD	[1, 25]
Ala234Val	AD	VMD	[26]
Val235Leu	AD	VMD	[1, 25]
Val235Met	AD	VMD	[1, 10, 13]
Thr237Arg	AD	VMD	[1, 9, 10, 15]
Thr237Ser	AD	VMD	[1, 15]
Gln238Leu	AR	ARB	[27]
Val239X	AR	ARB	[28]
Thr241Asn	AD	VMD	[1, 6, 19, 29]
Val242Met	AD	VMD	[1, 4, 30]
Ala243Glu	AR	ARB	[31]
Ala243Thr	AD	VMD	[1, 7, 12]
Ala243Val	AD	VMD	[1, 6, 9-11, 18, 21, 29]
Cys251Tyr	AR	ARB	[32]
Arg255Gln	AR	ARB	[33, 34]
Arg255Trp	AR	VMD and ARB	[8, 28, 33-37]
Arg255Trp	AD	VMD	[1, 24, 38]
Arg255Arg	AR	ARB	[36]
Arg255X	AR	ARB	[39]

Asn259X	AR	VMD and ARB	[6]
Pro260X	AR	ARB	[40]
Pro260X	AD	VMD	[29]
Val273Met	AR	ARB	[34]
Val273del	AR	ARB	This study
Pro274Arg	AR	VMD and ARB	[1]
Val275Ile	AR	VMD	[1]
Phe276Leu	AD	VMD	[1, 7]
Thr277Met	AR	ARB	[33, 34, 36, 41], This study
Phe281Ser	AR	ARB	[33], Luo *et al.* Novel Best1 Mutations in chinese patients. *Arvo Abstract. 2017*
Phe281X	AD and AR	VMD	[1, 7]
Phe283X	AR	ARB	[31]
Tyr284Cys	AD	VMD	[1, 2]
Trp287X	AD and AR	VMD	[1, 37, 42]
Trp287X	AR	ARB	[36, 43]
Ala291Val	AD	VMD	[37]
Glu292Lys	AD	VMD	[1, 22, 44, 45]
Gln293His	AD	VMD	[1, 5, 37]
Gln293Lys	AD	VMD	[1, 10, 16]
Gln293X	AR	ARB	[46]
Leu294Phe	AD	VMD	[12]
Leu294Phe	AR	ARB	[40]
Leu294Val	AD	VMD	[1, 5, 29, 47]
Leu294X	AD	VMD	[1, 6, 19]
Ile295Thr	AD	VMD	[29, 47, 48]
Ile295Val	AD	VMD	[1, 2]
Ile295Leu	AR	ARB	[6]
Ile295X	AD	VMD	[9, 10, 13, 15, 26, 29, 49]
Asn296Asp	AD	VMD	[1, 42]
Asn296His	AD	VMD	[1, 7]
Asn296Lys	AD	VMD	[1, 50]
Asn296Ser	AD	VMD	[1, 2, 12, 25, 51]
Asn296Ser	AR	ARB	[36]
Pro297Ala	AD	VMD	[1, 7, 10, 13]
Pro297Ser	AD	VMD	[1, 10, 52, 53]
Pro297Thr	AD	VMD	[1, 38]
Phe298Cys	AD	VMD	[47]
Phe298Ser	AD	VMD	[1, 26, 29, 50]

Phe298Val	AD	VMD	[22]
Gly299Glu	AD	VMD	[1, 10, 16, 17]
Gly299Ala	AD	VMD	[1, 18]
Gly299Arg	AD	VMD	[1, 5]
Glu300Asp	AD	VMD	[1, 6, 7, 10, 52-54]
Glu300Lys	AD	VMD	[1, 6, 7, 9, 10, 19, 44, 48]
Asp301Glu	AD	VMD	[1, 8-12, 29, 54, 55]
Asp301Gly	AD	VMD	[1, 37, 38]
Asp301Asn	AD	VMD	[9, 10]
Asp301X	AD	VMD	[1, 7, 19]
Asp302Ala	AD	VMD	[1, 6, 19, 47, 51]
Asp302Gly	AD	VMD	[1, 7]
Asp302His	AD	VMD	[1, 56]
Asp302Asn	AD	VMD	[51, 57]
Asp302Val	AD	VMD	[1, 7]
Asp302X	AD	VMD	[18, 50]
Asp303Glu	AD	VMD	[1, 56]
Asp303Gly	Semi-dominant	ARB	[35]
Asp303Gly	AD	VMD	[1]
Asp303Asn	AD	VMD	[26]
Asp304Gly	AD	VMD	[1, 26]
Asp304Asn	AD	VMD	[1, 12]
Asp304Val	AD	VMD	[58]
Asp304X	AD	VMD	[4, 20, 24]
Phe305Ser	AD	VMD	[1, 10, 13]
Phe305Tyr	AD	VMD	[1]
Glu306Asp	AD	VMD	[1, 4]
Glu306Gly	AD	VMD	[1, 5, 7]
Glu306Asn	AD	VMD	[7]
Thr307Ala	AD	VMD	[1, 7]
Thr307Ile	AD	VMD	[1, 7, 10, 19, 54, 59]
Thr307Asn	AD	VMD	[44, 48]
Asn308Ser	AD	VMD	[1, 56]
Trp309Arg	AD	VMD	[1, 12]
Ile310Thr	AD	VMD	[1, 2, 9, 10, 12]
Val311Gly	AD	VMD	[1, 9, 10]
Asp312Glu	AD, AR, Semi-dominant	VMD	[1, 57, 60, 61]
Asp312Asn	AR	ARB	[11, 26, 39, 62]
Asp312Asn	AD and AR	VMD	[1, 11]
Gln316His	AR	VMD	[1]
Gln316Pro	AD	VMD	[1, 19]

Val317Met	AR	VMD and ARB	[1, 36, 62]
Ser318X	AR	ARB	[34]
Leu319Pro	AR	ARB	[11]
Met325Thr	AR	ARB	[62-64]
Gln327X	AR	ARB	[65]
Pro346His	AD	VMD	[4]
Tyr347X	AR	ARB	[66]
Arg355His	AR	ARB	[36]
Arg356X	AR	ARB	[33, 34, 66, 67]
Ala357Val	AR	ARB	[33]
Ile366X	AR	VMD and ARB	[1, 68]
Met373X	AD and AR	VMD	[1, 6]
Glu374X	AR	ARB	[40]
Glu374X	AD	VMD	[49]
Leu472X	AR	VMD	[69]
His490X	AD and AR	VMD	[1]
His490X	AR	ARB	[70]
Val492Ile	AD	VMD	[1]
Ser517X	AR	ARB	[33]
Thr536Thr	AD	VMD	[55]
Glu557Lys	AD	VMD	[1]
Glu557X	AR	VMD	[71]

Supplemental Table 2. List of ARB and VMD mutations and accompanying inheritance patterns for BEST1 residues 221-585.

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