Mutation	Inheritance	Phenotype	Citation
Thr2Ala	AD	VMD	[1, 2]
Thr2Ile	AD	VMD	[2]
Thr2Asn	AD	VMD	[2, 3]
Thr2Ser	AD	VMD	[4]
Ile3Asn	AD	VMD	[5]
Ile3Thr	AD	VMD	[6]
Thr4Ala	AD	VMD	[2, 7]
Thr4Ile	AD	VMD	[2, 8]
Tyr5His	AD	VMD	[2]
Tyr5X	AR	VMD	[9]
Thr6Ala	AD	VMD	[2, 10]
Thr6Ile	AD	VMD	[11]
Thr6Lys	AD	VMD	[2]
Thr6Pro	AD	VMD	[2, 12-15]
Thr6Arg	AD	VMD	[2, 16, 17]
Ser7Asn	AD	VMD	[1, 17]
Val9Ala	AD	VMD	[2, 12, 14, 15, 18, 19]
Val9Leu	AD	VMD	[2, 20]
Val9Met	AD	VMD	[2, 11, 15, 21, 22]
Ala10Thr	AD	VMD	[2, 15, 22-24]
Ala10Val	AD	VMD	[12, 15]
Asn11Ile	AD	VMD	[2, 25]
Arg13Cys	AD	VMD	[2, 11]
Arg13His	AR	VMD and ARB	[8, 17, 26, 27]
Arg13His	AD	VMD	[2, 15, 28, 29]
Arg13Pro	AD	VMD	[2]
Gly15Asp	AD	VMD	[2, 18]
Gly15Arg	AD	VMD	[30]
Ser16Phe	AD	VMD	[2, 17, 23, 31-33]
Ser16Tyr	AD	VMD	[2, 13]
Phe17Cys	AD	VMD	[2, 16, 33]
Phe17Ser	AD	VMD	[2]
Arg19Leu	AD	VMD	[2]
Leu20Val	AD	VMD	[4, 34]
Leu21Arg	AD	VMD	[11]
Leu21Val	AD	VMD	[2, 15]
Trp24Cys	AD	VMD	[15, 16, 23, 24]
Arg25Asn	AD	VMD	[2]
Arg25Gln	AR	ARB	[35]
Arg25Gln	AD	VMD	[15, 24]

Arg25Trp	AR	ARB	[17, 36]
Arg25Trp	AD	VMD	[1, 2, 15, 16, 18, 22, 37, 38]
Gly26Arg	AD	VMD	[2, 25]
Ser27Arg	AD	VMD	[2, 15, 22]
Tyr29Cys	AD	VMD	[2, 39]
Tyr29His	AD	VMD	[2, 25]
Tyr29X	AR	VMD	[2, 19]
Lys30Glu	AD	VMD	[26]
Lys30Asn	AD	VMD	[2]
Lys30Arg	AD	VMD	[2, 16, 40, 41]
Tyr33His	AR	VMD	[8]
Gly34Gly	AR	ARB	[29, 36, 41, 42]
Glu35Lys	AR	ARB	[17, 41]
Ile38er	AD	VMD	[43]
Leu40Pro	AR	ARB	[17, 44]
Leu41Pro	AR	ARB	[45, 46]
Leu41Pro	AD	VMD	[2, 25]
Tyr44His	AR	ARB	[17]
Arg47Cys	AR	VMD	[2, 38]
Arg47His	AR	ARB	[17, 29, 47]
Arg47His	AD	VMD	[2, 32]
Leu52Pro	AR	ARB	[29]
Gln58Leu	AD	VMD	[2, 15, 22]
Gln58X	AR	ARB	[26, 48]
Gln59X	AR	ARB	[35]
Tyr72Asp	AD	VMD	[2]
Ile73Phe	AD	VMD	[20]
Ile73Met	AD	VMD	[17]
Ile73Asn	AD	VMD	[2, 33]
Ile73Val	AD	VMD	[2, 14]
Gln74X	AR	ARB	[17]
Leu75Phe	AD	VMD	[3]
Leu75Pro	AD	VMD	[2, 4]
Ile76Met	AD	VMD	[17]
Ile76Asn	AD	VMD	[2]
Ile76Thr	AD	VMD	[17]
Ile76Val	AD	VMD	[2, 21]
Ser79Tyr	AR	VMD	[4]
Phe80Cys	AD	VMD	[38]
Phe80Leu	AD	VMD	[1, 2, 16, 23]
Phe80Val	AD	VMD	[20]
Val81Met	AD	VMD	[1, 2, 17, 21]
Val81Leu	AR	VMD	[4]

Leu82Val	AD	VMD	[2, 6, 12, 15, 19, 34]
Gly83Leu	AD	VMD	[23]
Phe84Val	AD	VMD	[2, 23]
Tyr85His	AD	VMD	[2, 12, 14, 15, 19, 20, 34]
Val86Met	AD	VMD	[2]
Leu88X	AR	ARB	[49]
Val89Ala	AD	VMD	[2, 19, 50]
Thr91Ile	AD	VMD	[2, 16, 18, 47]
Arg92Cys	AD and AR	VMD	[2, 12, 15, 18, 29]
Arg92Gly	AD	VMD	[2, 18]
Arg92His	AD	VMD	[2, 19, 33, 34, 51]
ARg92Ser	AD	VMD	[2, 15, 22]
Trp93Cys	AD	VMD	[2, 12, 14, 15, 23, 28, 52]
Trp93Pro	AR	ARB	[53]
Trp93Arg	AD	VMD	[2, 23]
Trp93Ser	AD	VMD	[17]
Trp94Leu	AD	VMD	[17]
Gln96Glu	AD	VMD	[2, 41]
Gln94His	AD	VMD	[2, 12, 15]
Gln96Arg	AD	VMD	[2, 34, 54, 55]
Trp97His	AD	VMD	[17]
Glu98Lys	AD	VMD	[56]
Asn99Asp	AR	ARB	[17]
Asn99His	AD	VMD	[34]
Asn99Lys	AD	VMD	[2, 15, 22, 57]
Asn99Tyr	AD	VMD	[2, 21]
Leu100Arg	AD	VMD	[2, 11, 15, 22, 25]
Pro101Leu	AR	ARB	[58]
Pro101Leu	AD	VMD	[2, 41]
Pro101Thr	AR	ARB	[2, 59]
Pro101Thr	AD	VMD	[16, 20]
Trp102Arg	AD	VMD	[2, 25]
Asp104Glu	AD	VMD	[2, 12, 14, 15, 19]
Asp104His	AD	VMD	[2, 25]
Arg105Gly	AD	VMD	[17, 30]
Leu109Tyr	AR	ARB	[17]
Phe113Leu	AD	VMD	[2]
Glu115X	AD	VMD	[4]
Asp118Ala	AR	ARB	This Study
Glu119Gln	AD	VMD	[20]
Leu123X	AR	ARB	[17]

Thr124Met	AR	ARB	[26]
Thr127Mer	AR	ARB	[26]
Arg130Leu	AR	ARb	[8, 27]
Arg130Ser	AD and AR	VMD	[20, 60]
Tyr131Cys	AR	ARB	[47]
Asn133Asp	AR	ARB	[26]
Asn133Lys	AD	VMD	[2, 16, 41, 61]
Leu134Val	AR	ARB	[62]
Leu134Val	AD	VMD	[2, 63]
Gly135Ser	AD	VMD	[12, 15, 16, 21]
Leu140Val	AR	ARB, RP	This study, [64]
Leu140Arg	AD	VMD	[2, 16]
Arg141His	AR	VMD and ARB	[2, 11, 19, 35, 45, 48, 53, 58, 65-68]
Arg141His	AD	VMD	[15]
Arg141Ser	AR	VMD	[2, 67]
Ser142Gly	AD	VMD	[29]
Val143Ala	AD	VMD	[11]
Val143Phe	AD	VMD	[2, 29]
Ser144Gly	AD and AR	VMD	[9]
Ser144Asn	AD	VMD	[2, 32]
Ala146Lys	AD	VMD	[2, 69]
Ala146Ser	AD	VMD	[70]
Ala146Thr	AR	ARB	[29]
Pro148Ser	AR	ARB	[48]
Arg150Pro	AR	ARB	[47]
Pro152Ala	AR	VMD and ARB	[2, 45]
Ser157X	AD	VMD	Luo *et al.* Novel Best1 Mutations in chinese patients. *Arvo Abstract. 2017*
Gln159X	AR	ARB	[48]
Ala160Pro	AR	ARB	[36]
Met163Arg	AR	ARB	[8, 17, 29]
Glu167Gly	AR	ARB	[26, 71]
Tyr167Cys*	AD	VMD	[71]
Lys173X	AR	VMD	[8]
Leu174X	AR	ARB	[72]
His178X	AD	VMD	[2, 21]
Asn179X	AR	VMD	[20, 38]
Phe188Ser	AR	ARB	[62]
Leu191Pro	AR	ARB	[53]
Leu191Pro	AD	VMD	[2, 39]
Lys194X	AD	VMD	[2, 13]

Ala195Val	AR	VMD and ARB	[2, 8, 11, 17, 26]
Ala195Val	AD	VMD	[1, 2, 13, 16, 25]
Arg200X	AR	ARB	[26, 45, 73]
Ile201Thr	AD and AR	VMD	[2, 16, 21]
Ile201X	AR	VMD	[2]
Arg202Trp	AR	ARB	[74]
Ile205Tyr	AD	ARB	[75]
Ile205X	AR	VMD	[2]
Leu207Ile	AD	VMD	[16]
Ser209Asn	AD	VMD	[2, 15, 22]
Ile211Thr	AD	VMD	[16]
Glu213Lys	AD	VMD	[2]
Glu213Lys	AR	ARB	[76]
Val216Ile	AR	ARB	[47]
Leu217Phe	AD	VMD	[2, 39]
Arg218Cys	AR	ARB	[26]
Arg218Cys	AD	VMD	[2, 3, 4, 8, 11, 12, 15-17, 23, 28, 33, 38, 54, 57, 77]
Arg218His	AR	ARB	[78]
Arg218Gly	AD	VMD	[2, 38]
Arg218His	AD	VMD	[1, 2, 16, 20, 21, 23, 29, 33, 40, 41, 63]
Arg218Asn	AD	VMD	[2]
Arg218Gln	AD	VMD	[23]
Arg218Ser	AD	VMD	[2, 12, 15, 22, 70]
Gln220Pro	AD	VMD	[2]
Gln220X	AR	ARB	[79]

Supplemental Table 1. List of ARB and VMD mutations and accompanying inheritance patterns for BEST1 residues 1-220. *may be incorrect, but is recorded as listed in article.

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