Title: Bardet-Biedl Syndrome *GeneReview* Table 9
Authors: Forsythe E, Beales PL
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Note: The following information is provided by the authors listed above and has not been reviewed by *GeneReviews* staff.

Table 9. *BBS7* Pathogenic Allelic Variants

Gene	Mutation	Exon	Reference
BBS7	p.H323R homozygous	10	Badano et al 2003
BBS7	p.H323R homozygous	10	Badano et al 2003
BBS7	p.T2111 homozygous	6	Badano et al 2003
BBS7	K237fsX296 homozygous	7	Badano et al 2003

.0001 *BBS7* H323R. This amino acid substitution was identified in the homozygous state in all affected individuals from two unrelated BBS pedigrees [Badano et al 2003].

.0002 *BBS7* T211I. This amino acid substitution was identified in the homozygous state in all individuals in a consanguineous BBS pedigree [Badano et al 2003]. All affected family members additionally carried a E234K heterozygous change in *BBS1*, raising the possibility of complex inheritance between BBS7 and BBS1 [Badano et al 2003].

.0003 *BBS7* K237fsX296. A four-base pair deletion within exon 7of *BBS7*, resulting in the introduction in a premature stop codon within exon 9, was identified in the homozygous state in the only affected individual in a BBS family from Saudi Arabia.

References

Badano JL, Ansley SJ, Leitch CC, Lewis RA, Lupski JR, Katsanis N (2003) Identification of a novel Bardet-Biedl syndrome protein, BBS7, that shares structural features with BBS1 and BBS2. Am J Hum Genet 72:650-8