

Title: Bardet-Biedl Syndrome *GeneReview* Table 7

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Note: The following information is provided by the authors listed above and has not been reviewed by *GeneReviews* staff.

Table 7. *MKKS/BBS6* Benign Allelic Variants

Gene	Sequence Change	Exon	Coding Frequency	References
<i>BBS6/MKKS</i>	c.-675_674ins [GTGGCGGCCT]	1A	2.25%	Slavotinek et al 2002
<i>BBS6/MKKS</i>	c.-74G>A	3	0/94	Slavotinek et al 2002
<i>BBS6/MKKS</i>	c.117C>T; nil	3	Not done	Slavotinek et al 2002
<i>BBS6</i>	p.G49V	3	8.3%	Stone et al 2000
<i>BBS6</i>	c.534C>T; nil	3	Not done	Slavotinek et al 2002
<i>BBS6</i>	p.R517C	6	6.3%	Stone et al 2000
<i>BBS6</i>	p.G532V	6	6.4%	Stone et al 2000

References

Slavotinek AM, Searby C, Al-Gazali L, Hennekam RC, Schrandt-Stumpel C, Orcana-Losa M, Pardo-Reoyo S, Cantani A, Kumar D, Capellini Q, Neri G, Zackai E, Biesecker LG (2002) Mutation analysis of the *MKKS* gene in McKusick-Kaufman syndrome and selected Bardet-Biedl syndrome patients. *Hum Genet* 110:561-7

Stone DL, Slavotinek A, Bouffard GG, Banerjee-Basu S, Baxevanis AD, Barr M, Biesecker LG (2000) Mutation of a gene encoding a putative chaperonin causes McKusick-Kaufman syndrome. *Nat Genet* 25:79-82