

Title: Bardet-Biedl Syndrome *GeneReview* Table 5

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

Table 5. *BBS4* Benign Allelic Variants

Gene	Sequence Change	Exon	Coding Frequency	References
<i>BBS4</i>	c.-17C/A	1	Not done	Katsanis et al 2002
<i>BBS4</i>	c.42G/A; nil	1	A allele present in one individual and in 0/84 controls	Katsanis et al 2002
<i>BBS4</i>	c.-6A/G	3	G allele present in one individual and in 0/81 controls	Katsanis et al 2002
<i>BBS4</i>	c.91G/A; nil	4	Not done	Katsanis et al 2002
<i>BBS4</i>	c.8A/C; nil	5	C allele present in one individual and in 0/93 controls	Katsanis et al 2002
<i>BBS4</i>	c.28insA	5		Katsanis et al 2002
<i>BBS4</i>	c.17C/T	6		Katsanis et al 2002
<i>BBS4</i>	c.1061C/T	13		Katsanis et al 2002
<i>BBS4</i>	c.20C/T	13		Katsanis et al 2002
<i>BBS4</i>	c.18C/T	15	C allele present in one individual and in 0/93 controls	Katsanis et al 2002
<i>BBS4</i>	c.-45C/T	16		Katsanis et al 2002
<i>BBS4</i>	c.1561G/C	16	C allele present in one individual and in 0/87 controls	Katsanis et al 2002

References

Katsanis N, Eichers ER, Ansley SJ, Lewis RA, Kayserili H, Hoskins BE, Scambler PJ, Beales PL, Lupski JR (2002) *BBS4* is a minor contributor to Bardet-Biedl syndrome and may also participate in triallelic inheritance. *Am J Hum Genet* 71:22-9