

Title: Bardet-Biedl Syndrome *GeneReview* Table 3

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

Table 3. *BBS2* Benign Allelic Variants

Gene	Sequence Change	Exon	Coding Frequency	References
<i>BBS2</i>	p.I123V	3	Not done	Nishimura et al 2001
<i>BBS2</i>	p.V471V	12	Not done	Nishimura et al 2001

References

Nishimura DY, Searby CC, Carmi R, Elbedour K, Van Maldergem L, Fulton AB, Lam BL, Powell BR, Swiderski RE, Bugge KE, Haider NB, Kwitek-Black AE, Ying L, Duhl DM, Gorman SW, Héon E, Iannaccone A, Bonneau D, Biesecker LG, Jacobson SG, Stone EM, Sheffield VC (2001) Positional cloning of a novel gene on chromosome 16q causing Bardet-Biedl syndrome (*BBS2*). *Hum Mol Genet* 10:865-74