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 4. *BBS2* Pathogenic Allelic Variants

Gene	Mutation	Exon	Reference
BBS2	p.I314fsX324 homozygote	8	Nishimura et al 2001
BBS2	p.V75G homozygote	2	Nishimura et al 2001
BBS2	p.Y24X homozygote	1	Katsanis et al 2001
BBS2	p.Y24X heterozygote	1	Katsanis et al 2001
BBS2	p.Q59X heterozygote	2	Katsanis et al 2001
BBS2	p.Q59X heterozygote	2	Katsanis et al 2001
BBS2	p.Y24X heterozygote	1	Katsanis et al 2001
BBS2	p.R275X homozygote	8	Katsanis et al 2001
BBS2	p.R315W homozygote	9	Katsanis et al 2001
BBS2	p.D170fsX171 homozygote	4	Katsanis et al 2001
BBS2	p.C210fsX246 homozygote	6	Katsanis et al 2001
BBS2	p.D104A heterozygote	2	Katsanis et al 2001
BBS2	p.R634P heterozygote	15	Katsanis et al 2001
BBS2	p.D104A heterozygote	2	Katsanis et al 2001
BBS2	IVS1-1G>C heterozygote		Katsanis et al 2001
BBS2	p.V158fsX200 heterozygote	4	Katsanis et al 2001
BBS2	p.N70S heterozygote	2	Katsanis et al 2001
BBS2	p.L168fsX170 heterozygote	4	Katsanis et al 2001
BBS2	p.R216X heterozygote	6	Katsanis et al 2001
BBS2	p.T558I homozygote	14	Katsanis et al 2002
BBS2	p.N70S heterozygote	3	Katsanis et al 2000, Katsanis et al 2001
BBS2	p.T558I homozygote	14	Katsanis et al 2001

.0001 BBS2 I314fsX324. All affected members of a consanguineous Bedouin family were found to carry a homozygous deletion of a single nucleotide in exon 8 [Nishimura et al 2001].

.0002 BBS2, V75G. All affected members of a large consanguineous Bedouin BBS kindred were found to carry a non-conservative valine to glycine substitution

in exon 2 of BBS2 [Nishimura et al 2001]. The valine at this position is conserved in human, bovine, rabbit, rat, mouse and zebrafish BBS2 orthologues and this variant was postulated to be the disease-causing mutation, despite there being a second variant carried on the same chromosome (I123V).

.0003 BBS2, Y24X. This mutation was identified in the homozygous form in two unrelated individuals with BBS [Katsanis et al 2001]. One of those individuals also carried a heterozygous A242S mutation in BBS6 (see tri-allelic inheritance, section X). Y24X was also identified in compound heterozygosity with the Q59X mutation, in an individual who additionally had a heterozygous Q147X mutation within *BBS6* ([Katsanis et al 2001]; see triallelic inheritance).

.0004 BBS2, Q59X. One affected individual identified was a compound heterozygote for Q59X and Y24X in *BBS2* [Katsanis et al 2001]. A further mutation was identified in this individual: Q147X in BBS6.

.0009 BBS2, R275X. A homozygous arginine to termination mutation at codon 275 was identified in an individual with BBS [Katsanis et al 2001].

.0006 BBS2, R315W. A homozygous arginine to tryptophan mutation at codon 315 was identified in an individual with BBS [Katsanis et al 2001].

.0007 BBS2, D170fsX171. A homozygous frameshift mutation at codon 170 resulting in a termination codon at residue 171 was identified in an individual with BBS [Katsanis et al 2001].

.0008 BBS2, C210fsX246. A homozygous frameshift mutation at codon 210 resulting in a termination codon at residue 246 was identified in an individual with BBS [Katsanis et al 2001].

.0009 BBS2, D104A. One individual with BBS was identified who was compound heterozygous for an aspartic acid to alanine substitution at codon 104 of BBS2, and an arginine to proline substitution at codon 634 [Katsanis et al 2001]. This mutation was also identified in heterozygous form in an individual with BBS who was linked to the BBS1 locus.

.0010 BBS2, R634P. One individual with BBS was identified who was compound heterozygous for an arginine to proline substitution at codon 634 and an aspartic acid to alanine substitution at codon 104 of BBS2 [Katsanis et al 2001].

.0011 BBS2, IVS-1G>C. An affected individual identified was heterozygous for a G to C substitution at the -1 position of the intron 1 splice acceptor site [Katsanis et al 2001].

.0012 BBS2, V158fsX200. An affected individual linked to the BBS1 locus was found to carry a heterozygous frameshift mutation at codon 158, resulting in a premature stop codon at residue 200 [Katsanis et al 2001].

.0013 BBS2, N70S. An individual with BBS who was homozygous for a missense mutation in *BBS6* (Y37C) was found to additionally carry a heterozygous asparagines to serine substitution in *BBS2* [Katsanis et al 2001].

.0014 BBS2, L168fsX200. An individual with BBS was found to be a compound heterozygote for two mutations within *BBS2*: a frameshift mutation at codon 168 resulting in a stop codon at residue 170; and a nonsense mutation at codon 216 resulting in the introduction of a premature termination codon [Katsanis et al

2001]. A heterozygous C499S mutation in *BBS6* was identified in the same individual.

.0015 BBS2 R216X. An individual with BBS was found to be a compound heterozygote for two mutations within *BBS2*: the first a nonsense mutation at codon 216 resulting in the introduction of a premature termination codon; the second a frameshifting mutation at codon 168 resulting in a stop codon at residue 170 [Katsanis et al 2001]. A heterozygous C499S mutation in *BBS6* was identified in the same individual.

.0016 BBS2 T558I. A homozygous threonine to isoleucine mutation at codon 558 of *BBS2* was identified in an individual with BBS [Katsanis et al 2001]. The same individual was later also identified to have a homozygous A364E mutation in *BBS4* [Katsanis et al 2002].

References

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