Title: Bardet-Biedl Syndrome *GeneReview* Table 6

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Date: February 2014

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

Table 6. *BBS4* Pathogenic Allelic Variants

Gene	Mutation	Exon	Reference
BBS4	p.A364E homozygote	13	Katsanis et al 2002
BBS4	c.77-220del homozygote	Exons 3-4	Mykytyn et al 2001
BBS4	c.77-220del homozygote	Exons 3-4	Mykytyn et al 2001
BBS4	p.R295P homozygote	12	Mykytyn et al 2001
BBS4	IVS4+1G>C homozygote	4	Mykytyn et al 2001
BBS4	p.V195fsX209 heterozygote	8	Mykytyn et al 2001
BBS4	IVS6-2A>C homozygote	7	Mykytyn et al 2001
BBS4	IVS3-2A>C homozygote	4	Katsanis et al 2001
BBS4	p.A364E homozygote	13	Katsanis et al 2001
BBS4	p.L327p heterozygote	12	Katsanis et al 2001
BBS4	p.N165H heterozygote	8	Katsanis et al 2002
BBS4	p.S457I heterozygote	15	Katsanis et al 2002

.0001 BBS4 77-220del. A homozygous 6-kb deletion resulting in the removal of the whole of exons 3 and 4 was identified in all individuals affected with BBS from two families: an Italian and an Israeli Arab family [Mykytyn et al 2001]. The deletion breakpoints occurred within Alu elements in introns 2 and 4 and haplotype analysis suggested that the mutation arose independently in the two families.

.0002 BBS4 R295P. All affected individuals of a large consanguineous Bedouin kindred were found to carry a homozygous arginine to proline missense mutation within exon 12 of BBS4 [Mykytyn et al 2001].

.0003 BBS4 IVS4+1G>C. A homozygous G to C substitution at the -2 position of the splice donor site of intron 4 was identified in the two affected siblings from a European BBS family [Mykytyn et al 2001].

.0004 BBS4 V195fsX209. A heterozygous two-base pair insertion in exon 8 of *BBS4* was identified in a small non-consanguineous BBS family which is predicted to result in a premature stop codon at residue 209 [Mykytyn et al 2001]. Sequence analysis of the coding region did not reveal a second mutation within *BBS4* in this family.

.0005 BBS4 IVS6-2A>C. A homozygous A to C substitution at the -2 position of the splice acceptor site of intron 6 was identified in the two affected siblings from a European BBS family [Mykytyn et al 2001].

.0006 BBS4 IVS3-2A>G. In a consanguineous family from Saudi Arabia the affected individual was found to carry a homozygous A to G substitution at the -2 position of the splice acceptor site of intron 3 [Katsanis et al 2002].

.0007 BBS4 A364E. A homozygous missense mutation resulting in an alanine to glutamine substitution was detected in an affected individual from a consanguineous Kurdish family [Katsanis et al 2002]. Furthermore, the affected individual had previously been shown to be homozygous for a T558I mutant *BBS2* allele [Katsanis et al 2001], which may suggest that the disease is inherited in a tetra-allelic pattern in this family.

.0008 BBS4 L327P. A non-conservative leucine to proline substitution was identified in heterozygous form in affected members from a BBS family [Katsanis et al 2002]. With the absence of functional data on the BBS4 protein, it is unclear whether this change is a pathogenic alteration.

.0009 BBS4 N165H. A non-conservative asparagine to histidine substitution was identified in heterozygous form in affected members from a BBS family [Katsanis et al 2002]. With the absence of functional data on the BBS4 protein, it is unclear whether this change is a pathogenic alteration.

.0010 BBS4 S457I. A non-conservative serine to isoleucine substitution was identified in heterozygous form in affected members from a BBS family [Katsanis et al 2002]. With the absence of functional data on the BBS4 protein, it is unclear whether this change is a pathogenic alteration.

References

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