



## BBS10 gene

Bardet-Biedl syndrome 10

### Normal Function

The *BBS10* gene provides instructions for making a protein that is found in many types of cells. The BBS10 protein is part of a group (complex) of proteins that functions as a chaperonin. Chaperonins help fold other proteins into their correct 3-dimensional shapes so they can perform their usual functions in the body.

Studies suggest that the BBS10 protein helps fold or stabilize certain proteins that are necessary for the normal formation of cilia. Cilia are microscopic, finger-like projections that stick out from the surface of many types of cells. They are involved in cell movement and many different chemical signaling pathways. Cilia are also necessary for the perception of sensory input (such as sight, hearing, and smell).

### Health Conditions Related to Genetic Changes

#### Bardet-Biedl syndrome

More than 35 mutations in the *BBS10* gene have been found to cause Bardet-Biedl syndrome. Mutations in this gene account for about 20 percent of all cases of the disorder.

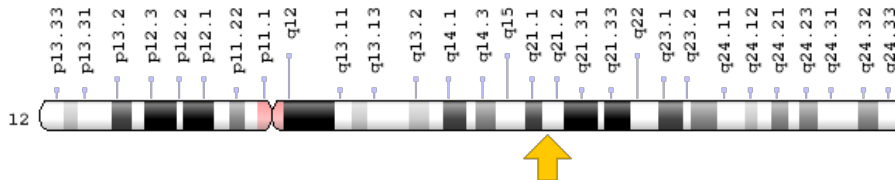
Some *BBS10* gene mutations change single protein building blocks (amino acids) in the BBS10 protein, while other mutations add or delete genetic material in the *BBS10* gene. The most common *BBS10* gene mutation, which is written as C91fsX95, leads to the production of an abnormally short version of the BBS10 protein.

Researchers are studying how mutations in the *BBS10* gene lead to the specific features of Bardet-Biedl syndrome. A malfunctioning BBS10 protein appears to affect the normal formation and function of cilia. Defects in these cell structures probably disrupt important chemical signaling pathways during development and lead to abnormalities of sensory perception. Researchers believe that defective cilia are responsible for most of the features of Bardet-Biedl syndrome, including vision loss, obesity, the presence of extra fingers and/or toes (polydactyly), kidney abnormalities, and intellectual disability.

## Chromosomal Location

Cytogenetic Location: 12q21.2, which is the long (q) arm of chromosome 12 at position 21.2

Molecular Location: base pairs 76,344,474 to 76,348,415 on chromosome 12 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- BBS10\_HUMAN
- C12orf58
- FLJ23560

## Additional Information & Resources

### Educational Resources

- Molecular Cell Biology (first edition, 2000): Cilia and Flagella: Structure and Movement  
<https://www.ncbi.nlm.nih.gov/books/NBK21698/>

### Clinical Information from GeneReviews

- Bardet-Biedl Syndrome  
<https://www.ncbi.nlm.nih.gov/books/NBK1363>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28BBS10%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

### Catalog of Genes and Diseases from OMIM

- BBS10 GENE  
<http://omim.org/entry/610148>

## Research Resources

- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=BBS10%5Bgene%5D>
- HGNC Gene Symbol Report  
[https://www.genenames.org/data/gene-symbol-report/#!/hgnc\\_id/HGNC:26291](https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:26291)
- Monarch Initiative  
<https://monarchinitiative.org/gene/NCBIGene:79738>
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/79738>
- UniProt  
<https://www.uniprot.org/uniprot/Q8TAM1>

## **Sources for This Summary**

- Laurier V, Stoetzel C, Muller J, Thibault C, Corbani S, Jalkh N, Salem N, Chouery E, Poch O, Licaire S, Danse JM, Amati-Bonneau P, Bonneau D, Mégarbané A, Mandel JL, Dollfus H. Pitfalls of homozygosity mapping: an extended consanguineous Bardet-Biedl syndrome family with two mutant genes (BBS2, BBS10), three mutations, but no triallelism. *Eur J Hum Genet.* 2006 Nov; 14(11):1195-203. Epub 2006 Jul 5.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/16823392>
- Marion V, Stoetzel C, Schlicht D, Messaddeq N, Koch M, Flori E, Danse JM, Mandel JL, Dollfus H. Transient ciliogenesis involving Bardet-Biedl syndrome proteins is a fundamental characteristic of adipogenic differentiation. *Proc Natl Acad Sci U S A.* 2009 Feb 10;106(6):1820-5. doi: 10.1073/pnas.0812518106. Epub 2009 Feb 3.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/19190184>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2635307/>
- Muller J, Stoetzel C, Vincent MC, Leitch CC, Laurier V, Danse JM, Hellé S, Marion V, Bennouna-Greene V, Vicaire S, Megarbane A, Kaplan J, Drouin-Garraud V, Hamdani M, Sigaudy S, Francannet C, Roume J, Bitoun P, Goldenberg A, Philip N, Odent S, Green J, Cossée M, Davis EE, Katsanis N, Bonneau D, Verloes A, Poch O, Mandel JL, Dollfus H. Identification of 28 novel mutations in the Bardet-Biedl syndrome genes: the burden of private mutations in an extensively heterogeneous disease. *Hum Genet.* 2010 Mar;127(5):583-93. doi: 10.1007/s00439-010-0804-9. Epub 2010 Feb 23.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/20177705>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3638942/>
- Seo S, Baye LM, Schulz NP, Beck JS, Zhang Q, Slusarski DC, Sheffield VC. BBS6, BBS10, and BBS12 form a complex with CCT/TRiC family chaperonins and mediate BBSome assembly. *Proc Natl Acad Sci U S A.* 2010 Jan 26;107(4):1488-93. doi: 10.1073/pnas.0910268107. Epub 2010 Jan 4.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/20080638>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2824390/>

- Stoetzel C, Laurier V, Davis EE, Muller J, Rix S, Badano JL, Leitch CC, Salem N, Chouery E, Corbani S, Jalk N, Vicaire S, Sarda P, Hamel C, Lacombe D, Holder M, Odent S, Holder S, Brooks AS, Elcioglu NH, Silva ED, Rossillion B, Sigaudy S, de Ravel TJ, Lewis RA, Leheup B, Verloes A, Amati-Bonneau P, Mégarbané A, Poch O, Bonneau D, Beales PL, Mandel JL, Katsanis N, Dollfus H. BBS10 encodes a vertebrate-specific chaperonin-like protein and is a major BBS locus. *Nat Genet.* 2006 May;38(5):521-4. Epub 2006 Apr 2. Erratum in: *Nat Genet.* 2006 Jun;38(6):727. Da Silva, Eduardo [corrected to Silva, Eduardo D].  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/16582908>
  - White DR, Ganesh A, Nishimura D, Rattenberry E, Ahmed S, Smith UM, Pasha S, Raeburn S, Trembath RC, Rajab A, Macdonald F, Banin E, Stone EM, Johnson CA, Sheffield VC, Maher ER. Autozygosity mapping of Bardet-Biedl syndrome to 12q21.2 and confirmation of FLJ23560 as BBS10. *Eur J Hum Genet.* 2007 Feb;15(2):173-8. Epub 2006 Nov 15.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/17106446>
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