

## BBS1 gene

Bardet-Biedl syndrome 1

### Normal Function

The *BBS1* gene provides instructions for making a protein found in cells throughout the body. The BBS1 protein is part of a group (complex) of proteins that plays a critical role in the formation of cell structures called 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 30 mutations in the *BBS1* gene have been identified in people with Bardet-Biedl syndrome. Mutations in this gene are the most common cause of Bardet-Biedl syndrome, accounting for about one-quarter of all cases.

Most *BBS1* gene mutations change single protein building blocks (amino acids) in the BBS1 protein or lead to the production of an abnormally short version of the protein. The most common mutation replaces the amino acid methionine with the amino acid arginine at protein position 390 (written as Met390Arg or M390R).

Mutations in the *BBS1* gene likely 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.

### Other Names for This Gene

- BBS1\_HUMAN
- BBS2L2
- FLJ23590

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of BBS1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=582\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=582[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28BBS1%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

- BBS1 GENE; BBS1 (<https://omim.org/entry/209901>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/582>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=BBS1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=BBS1[gene]))

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## Genomic Location

The *BBS1* gene is found on chromosome 11 (<https://medlineplus.gov/genetics/chromosome/11/>).

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