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.

Other Names for This Gene

- BBS10_HUMAN
- C12orf58
- FLJ23560

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of BBS10 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=79738[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?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; BBS10 (https://omim.org/entry/610148)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/79738)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=BBS10[gene])

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Genomic Location

The *BBS10* gene is found on chromosome 12 (https://medlineplus.gov/genetics/chromosome/12/).

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