

MKKS gene

MKKS centrosomal shuttling protein

Normal Function

The *MKKS* gene (also called the *BBS6* gene) provides instructions for making a protein that plays an important role in early development, specifically in the formation of the limbs, heart, and reproductive system. This protein's structure suggests that it may belong to a family of proteins called chaperonins. Proteins must be folded into the correct shape to function properly, and chaperonins help them do that.

The MKKS protein combines with other proteins to form a structure known as the chaperonin complex. The chaperonin complex serves as a scaffold for the assembly of another molecule called the BBSome. The BBSome helps transport materials that support the function of cilia, the microscopic, finger-like projections on the surface of cells. Cilia help transmit information.

Researchers speculate that the MKKS protein may also be directly involved in transporting important molecules to different locations within the cell. Specifically, the MKKS protein may help transport SMARCC1 protein from the cytoplasm into the nucleus of the cell. SMARCC1 helps regulate the activity of certain genes.

Health Conditions Related to Genetic Changes

McKusick-Kaufman syndrome

A few variants (also called mutations) in the *MKKS* gene have been found to cause McKusick-Kaufman syndrome. McKusick-Kaufman syndrome is a condition that affects the development of the hands, feet, heart, and reproductive system. Two particular variants have been found to cause McKusick-Kaufman syndrome in the Old Order Amish population. Each of these variants changes a single protein building block (amino acid) in the MKKS protein. One variant replaces the amino acid histidine with the amino acid tyrosine at protein position 84 (written as p.His84Tyr or p.H84Y). The other variant replaces the amino acid alanine with the amino acid serine at protein position 242 (written as p.Ala242Ser or p.A242S). Those with McKusick-Kaufman syndrome in the Old Order Amish population typically have both of these variants in each copy of the *MKKS* gene. This combination of variants is rarely found in individuals with McKusick-

Kaufman syndrome outside of the Old Order Amish population.

Some variants in the *MKKS* gene can also cause Bardet-Biedl syndrome, a condition that is related to McKusick-Kaufman syndrome.

Though it is not clear exactly how variants in the *MKKS* gene lead to the specific signs and symptoms of McKusick-Kaufman syndrome, the p.His84Tyr and p.Ala242Ser variants may impair the protein's ability to deliver SMARCC1 into the nucleus of the cell. This change likely affects the activity of certain genes that are critical during early development.

Bardet-Biedl syndrome

MedlinePlus Genetics provides information about Bardet-Biedl syndrome

Other Names for This Gene

- BBS6
- HMCS
- KMS
- MKS

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of *MKKS* ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8195\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8195[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28MKKS%5BTIAB%5D%29+OR+%28BBS6%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D%29>)

Catalog of Genes and Diseases from OMIM

- *MKKS* CENTROSOMAL SHUTTILING PROTEIN; *MKKS* (<https://omim.org/entry/604896>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/8195>)

- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=MKKS\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=MKKS[gene]))

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Genomic Location

The *MKKS* gene is found on chromosome 20 (<https://medlineplus.gov/genetics/chromosome/20/>).

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