



MKKS gene

McKusick-Kaufman syndrome

Normal Function

The *MKKS* gene provides instructions for making a protein that plays an important role in the formation of the limbs, heart, and reproductive system. The structure of this protein suggests that it may act as a chaperonin, which is a protein that helps fold other proteins. Proteins must be folded into the correct 3-dimensional shape to perform their usual functions in the body. Abnormally folded proteins can also interfere with the functions of normal proteins.

Although the structure of the MKKS protein is similar to that of a chaperonin, some studies have suggested that protein folding may not be this protein's primary function. Within cells, the MKKS protein is associated with structures called centrosomes. Centrosomes play a role in cell division and the assembly of microtubules, which are proteins that transport materials in cells and help the cell maintain its shape. Researchers speculate that the MKKS protein may be involved in transporting other proteins within the cell.

Health Conditions Related to Genetic Changes

McKusick-Kaufman syndrome

Two mutations in the *MKKS* gene have been identified in people with McKusick-Kaufman syndrome in the Old Order Amish population. Each of these mutations changes a single protein building block (amino acid) in the MKKS protein. One mutation replaces the amino acid histidine with the amino acid tyrosine at protein position 84 (written as His84Tyr or H84Y). The other mutation replaces the amino acid alanine with the amino acid serine at protein position 242 (written as Ala242Ser or A242S). Affected Amish people have these two mutations in both copies of the *MKKS* gene.

The mutations that underlie McKusick-Kaufman syndrome alter the structure of the MKKS protein. Although the altered protein disrupts the development of several parts of the body before birth, it is unclear how *MKKS* mutations lead to the specific features of this disorder.

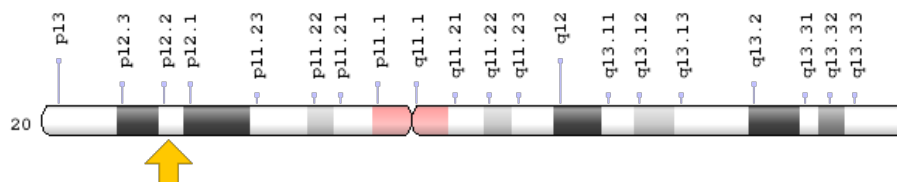
The signs and symptoms of McKusick-Kaufman syndrome overlap significantly with those of another condition called Bardet-Biedl syndrome, which can make the two conditions difficult to tell apart in infancy and early childhood. Although both syndromes can be caused by changes in the *MKKS* gene, it remains unclear why some mutations cause McKusick-Kaufman syndrome and others cause Bardet-Biedl syndrome.

Bardet-Biedl syndrome

Chromosomal Location

Cytogenetic Location: 20p12.2, which is the short (p) arm of chromosome 20 at position 12.2

Molecular Location: base pairs 10,401,009 to 10,434,239 on chromosome 20 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- Bardet-Biedl syndrome 6 protein
- BBS6
- HMCS
- KMS
- MKKS_HUMAN
- MKS

Additional Information & Resources

Educational Resources

- Howard Hughes Medical Institute: First Bardet-Biedl Syndrome Gene Identified (August 28, 2000)
<https://www.hhmi.org/news/first-bardet-biedl-syndrome-gene-identified>
- Molecular Cell Biology (fourth edition, 2000): Folding of Proteins in Vivo Is Promoted by Chaperones
<https://www.ncbi.nlm.nih.gov/books/NBK21750/#A553>
- The Cell: A Molecular Approach (second edition, 2000): Chaperones and Protein Folding
<https://www.ncbi.nlm.nih.gov/books/NBK9843/#A1200>

Clinical Information from GeneReviews

- Bardet-Biedl Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1363>
- McKusick-Kaufman Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1502>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?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>

Catalog of Genes and Diseases from OMIM

- MKKS GENE
<http://omim.org/entry/604896>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_MKKS.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=MKKS%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:7108
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:8195>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/8195>
- UniProt
<https://www.uniprot.org/uniprot/Q9NPJ1>

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