KAT6B gene
lysine acetyltransferase 6B

Normal Function

The *KAT6B* gene provides instructions for making a type of enzyme called a histone acetyltransferase. These enzymes modify histones, which are structural proteins that attach (bind) to DNA and give chromosomes their shape. By adding a small molecule called an acetyl group to particular locations on histones, histone acetyltransferases control the activity of certain genes.

Little is known about the function of the histone acetyltransferase produced from the *KAT6B* gene. It is active in cells and tissues throughout the body, where it interacts with many other proteins. It appears to regulate genes that are important for early development, including development of the skeleton and nervous system.

Health Conditions Related to Genetic Changes

**Genitopatellar syndrome**

At least eight mutations in the *KAT6B* gene have been identified in people with genitopatellar syndrome, a rare condition characterized by genital abnormalities, missing or underdeveloped kneecaps (patellae), intellectual disability, and abnormalities affecting other parts of the body. The mutations that cause genitopatellar syndrome occur near the end of the *KAT6B* gene in a region known as exon 18. These mutations lead to the production of a shortened histone acetyltransferase enzyme. Researchers suspect that the shortened enzyme may function differently than the full-length version, altering the regulation of various genes during early development. Because the altered enzyme takes on a different function, these mutations are described as "gain-of-function." However, it is unclear how these changes lead to the specific features of genitopatellar syndrome.

**Ohdo syndrome, Say-Barber-Biesecker-Young-Simpson variant**

More than 10 mutations in the *KAT6B* gene have been found to cause the Say-Barber-Biesecker-Young-Simpson (SBBYS) variant of Ohdo syndrome. This condition has signs and symptoms that overlap with those of genitopatellar syndrome (described above), although some of the specific developmental abnormalities differ between the two conditions. Mutations that cause the SBBYS variant of Ohdo syndrome have been identified throughout the *KAT6B* gene, although many of them occur in exon 18. Studies suggest that these mutations likely prevent the production of functional histone acetyltransferase from one copy of the *KAT6B* gene in each cell. A shortage of this enzyme impairs the regulation of various genes during early development. Because these mutations lead to a reduction in the enzyme, they are
described as "loss-of-function." However, it is unclear how these changes lead to the specific features of the condition.

Coloboma

Cancers

Genetic changes involving the KAT6B gene have been associated with certain types of cancer. These mutations are somatic, which means they are acquired during a person’s lifetime and are present only in certain cells. The genetic changes are chromosomal rearrangements (translocations) that disrupt the region of chromosome 10 containing the KAT6B gene. Researchers have found a translocation that attaches this region of chromosome 10 to part of chromosome 16 in some people with a cancer of blood-forming cells called acute myeloid leukemia (AML). This translocation has also been identified in some people with therapy-related myelodysplastic syndrome, a blood disorder that can occur after a person has undergone chemotherapy for another form of cancer.

It is unclear how translocations involving the KAT6B gene are related to the development of cancer. These changes likely alter histone modification, which could prevent normal regulation of gene activity. Impaired gene regulation may contribute to the growth of cancers by allowing abnormal cells to grow and divide uncontrollably.

Tumors

Somatic changes involving the KAT6B gene have also been identified in some people with uterine leiomyomas, which are noncancerous growths in the uterus that are also known as uterine fibroids. Uterine leiomyomas are common in adult women. These growths can cause pelvic pain and abnormal bleeding, and, in some cases, lead to an inability to have biological children (infertility). The genetic change associated with uterine leiomyomas is a translocation between the region of chromosome 10 containing the KAT6B gene and a particular region of chromosome 17. It is unclear how this translocation is related to tumor development. Changes in histone modification that impair normal gene regulation may allow certain cells to divide in an uncontrolled way, leading to the growth of a tumor.
Chromosomal Location

Cytogenetic Location: 10q22.2, which is the long (q) arm of chromosome 10 at position 22.2

Molecular Location: base pairs 74,824,927 to 75,032,624 on chromosome 10 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- GTPTS
- histone acetyltransferase KAT6B
- histone acetyltransferase MORF
- histone acetyltransferase MOZ2
- histone acetyltransferase MYST4
- K(lysine) acetyltransferase 6B
- KAT6B_HUMAN
- monocytic leukemia zinc finger protein-related factor
- MORF
- MOZ-related factor
- MOZ2
- MYST-4
- MYST histone acetyltransferase (monocytic leukemia) 4
- MYST4
- qkf
- querkopf
- ZC2HC6B
Additional Information & Resources

Educational Resources

- Epigenomics Help (2010): Histone Modification "Writers"  

Clinical Information from GeneReviews

- KAT6B Disorders  
  https://www.ncbi.nlm.nih.gov/books/NBK114806

Scientific Articles on PubMed

- PubMed  
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28KAT6B%5BTIAB%5D%29+OR+%28MORF%5BTIAB%5D%29+OR+%28MOZ2%5BTIAB%5D%29+OR+%28MYST4%5BTIAB%5D%29+OR+%28querkopf%5BTIAB%5D%29+AND+%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D

Catalog of Genes and Diseases from OMIM

- LEIOMYOMA, UTERINE  
  http://omim.org/entry/150699

- LEUKEMIA, ACUTE MYELOID  
  http://omim.org/entry/601626

- LYSINE ACETYLTRANSFERASE 6B  
  http://omim.org/entry/605880

- MYELODYSPLASTIC SYNDROME  
  http://omim.org/entry/614286

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
  http://atlasgeneticsoncology.org/Genes/MYST4ID41488ch10q22.html

- ClinVar  

- HGNC Gene Symbol Report  

- Monarch Initiative  
  https://monarchinitiative.org/gene/NCBIGene:23522
• NCBI Gene
• UniProt
  https://www.uniprot.org/uniprot/Q8WYB5

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/22265014
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3276659/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/22715153
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3696352/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10497217

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/22077973
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3213399/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12542485

• Moore SD, Herrick SR, Ince TA, Kleinman MS, Dal Cin P, Morton CC, Quade BJ. Uterine leiomyomata with t(10;17) disrupt the histone acetyltransferase MORF. Cancer Res. 2004 Aug 15;64(16):5570-7.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15313893

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11157802
Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/22265017
Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3276665/

Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17694082

Reviewed: February 2013
Published: June 23, 2020

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services