

Say-Barber-Biesecker Syndrome

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KAT6B gene

lysine acetyltransferase 6B

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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.

More About This Health Condition

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.

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Coloboma

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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.

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 histone acetyltransferase (monocytic leukemia) 4 MYST-4 MYST4 qkf querkopf ZC2HC6B

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

Tests of KAT6B

Scientific Articles on PubMed

PubMed

Catalog of Genes and Diseases from OMIM

LEIOMYOMA, UTERINE; UL

LEUKEMIA, ACUTE MYELOID; AML

LYSINE ACETYLTRANSFERASE 6B; KAT6B

MYELODYSPLASTIC SYNDROME; MDS

Gene and Variant Databases

NCBI Gene

ClinVar

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Genomic LocationThe KAT6B gene is found on chromosome 10.

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