

LHX1 gene

LIM homeobox 1

Normal Function

The *LHX1* gene provides instructions for making a protein that attaches (binds) to specific regions of DNA and regulates the activity of other genes. On the basis of this role, the protein produced from the *LHX1* gene is called a transcription factor. The LHX1 protein is part of a large group of transcription factors called homeodomain proteins. The homeodomain is a region of the protein that allows it to bind to DNA.

The LHX1 protein is found in many of the body's organs and tissues. Studies suggest that it plays particularly important roles in the development of the brain and female reproductive system.

Health Conditions Related to Genetic Changes

17q12 deletion syndrome

17q12 deletion syndrome is a condition that results from the deletion of a small piece of chromosome 17 in each cell. Signs and symptoms of 17q12 deletion syndrome can include abnormalities of the kidneys, urinary tract, and reproductive system; a form of diabetes called maturity-onset diabetes of the young type 5 (MODY5); delayed development; intellectual disability; and behavioral or psychiatric disorders. Some females with this chromosomal change have Mayer-Rokitansky-Küster-Hauser syndrome, which is characterized by underdevelopment or absence of the vagina and uterus. Features associated with 17q12 deletion syndrome vary widely, even among affected members of the same family.

The part of chromosome 17 that is deleted is on the long (q) arm of the chromosome at a position designated q12. This region of the chromosome contains 15 genes, including *LHX1*. A deletion of this region results in a loss of one copy of the *LHX1* gene in each cell, leading to a reduced amount of LHX1 protein. A shortage of this protein likely disrupts the regulation of genes that are necessary for the normal development of several organs, including the brain and female reproductive system. Researchers suspect that a loss of one copy of the *LHX1* gene contributes to intellectual disability, behavioral and psychiatric conditions, and Mayer-Rokitansky-Küster-Hauser syndrome in people with 17q12 deletion syndrome.

Mayer-Rokitansky-Küster-Hauser syndrome

MedlinePlus Genetics provides information about Mayer-Rokitansky-Küster-Hauser syndrome

Other Names for This Gene

- homeobox protein Lim-1
- LIM homeobox protein 1
- LIM-1
- LIM/homeobox protein Lhx1
- LIM1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of LHX1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3975\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3975[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28LHX1%5BTIAB%5D%29+OR+%28LIM+homeobox+1%5BTIAB%5D%29%29+OR+%28%28LIM+homeobox+protein+1%5BTIAB%5D%29+OR+%28LIM-1%5BTIAB%5D%29+OR+%28LIM1%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D%29%29%29>)

Catalog of Genes and Diseases from OMIM

- LIM HOMEBOX GENE 1; LHX1 (<https://omim.org/entry/601999>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/3975>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=LHX1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=LHX1[gene]))

References

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Genomic Location

The *LHX1* gene is found on chromosome 17 (<https://medlineplus.gov/genetics/chromosome/17/>).

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