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.

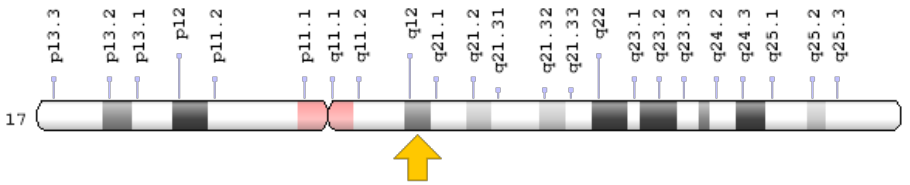
The part of chromosome 17 that is deleted is on the long (q) arm of the chromosome at a position designated q12. This region of the chromosome contains 15 genes, including *LHX1*. A deletion of this region results in a loss of one copy of the *LHX1* gene in each cell, leading to a reduced amount of LHX1 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

Chromosomal Location

Cytogenetic Location: 17q12, which is the long (q) arm of chromosome 17 at position 12

Molecular Location: base pairs 36,937,475 to 36,944,612 on chromosome 17 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

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

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience Database: LIM Domain and Its Binding to Target Proteins
  https://www.ncbi.nlm.nih.gov/books/NBK6372/
- Molecular Biology of the Cell (fourth edition, 2002): Homeodomain Proteins Constitute a Special Class of Helix-Turn-Helix Proteins
  https://www.ncbi.nlm.nih.gov/books/NBK26806/#A1240

Clinical Information from GeneReviews

- 17q12 Recurrent Deletion Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK401562

Scientific Articles on PubMed

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

Catalog of Genes and Diseases from OMIM

- LIM HOMEOBOX GENE 1
  http://omim.org/entry/601999

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_LHX1.html
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:6593
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:3975
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/3975
- UniProt
  https://www.uniprot.org/uniprot/P48742

Sources for This Summary

- Hobert O, Westphal H. Functions of LIM-homeobox genes. Trends Genet. 2000 Feb;16(2):75-83. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10652534
- OMIM: LIM HOMEOBOX GENE 1
  http://omim.org/entry/601999
- Mefford HC, Clauin S, Sharp AJ, Moller RS, Ullmann R, Kapur R, Pinkel D, Cooper GM, Ventura M, Ropers HH, Tommerup N, Eichler EE, Bellanne-Chantelot C. Recurrent reciprocal genomic rearrangements of 17q12 are associated with renal disease, diabetes, and epilepsy. Am J Hum Genet. 2007 Nov;81(5):1057-69.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17924346
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2265663/

- Moreno-De-Luca D; SGENE Consortium., Mulle JG; Simons Simplex Collection Genetics Consortium., Kaminsky EB, Sanders SJ; GeneSTAR., Myers SM, Adam MP, Pakula AT, Eisenhauer NJ, Uhas K, Weik L, Guy L, Care ME, Morel CF, Boni C, Salbert BA, Chandrareddy A, Demmer LA, Chow EW, Surti U, Aradhya S, Pickering DL, Golden DM, Sanger WG, Aston E, Brothman AR, Gliem TJ, Thorland EC, Ackley T, Iyer R, Huang S, Barber JC, Crolla JA, Warren ST, Martin CL, Ledbetter DH. Deletion 17q12 is a recurrent copy number variant that confers high risk of autism and schizophrenia. Am J Hum Genet. 2010 Nov 12;87(5):618-30. doi: 10.1016/j.ajhg.2010.10.004. Erratum in: Am J Hum Genet. 2011 Jan 7;88(1):121.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/21055719
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2978962/
- Nagamani SC, Erez A, Shen J, Li C, Roeder E, Cox S, Karaviti L, Pearson M, Kang SH, Sahoo T, Lalani SR, Stankiewicz P, Sutton VR, Cheung SW. Clinical spectrum associated with recurrent genomic rearrangements in chromosome 17q12. Eur J Hum Genet. 2010 Mar;18(3):278-84. doi: 10.1038/ejhg.2009.174.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19844256
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2987224/

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