LMX1B gene
LIM homeobox transcription factor 1 beta

Normal Function

The LMX1B gene provides instructions for producing a protein that attaches (binds) to specific regions of DNA and regulates the activity of other genes. On the basis of this role, the LMX1B protein is called a transcription factor. The LMX1B protein appears to be particularly important during early embryonic development of the limbs, kidneys, and eyes.

Health Conditions Related to Genetic Changes

Nail-patella syndrome

At least 145 mutations in the LMX1B gene have been found to cause nail-patella syndrome. Most mutations result in the production of an abnormally short, nonfunctional version of the LMX1B protein or change a single protein building block (amino acid). Mutations that substitute one amino acid for another amino acid reduce or eliminate the protein's ability to bind to DNA, disrupting the regulation of other genes during early development. Deletions of the entire LMX1B gene or large portions of the gene have also been shown to cause nail patella syndrome. It is unclear exactly how mutations in the LMX1B gene lead to the signs and symptoms of nail-patella syndrome.

Chromosomal Location

Cytogenetic Location: 9q33.3, which is the long (q) arm of chromosome 9 at position 33.3

Molecular Location: base pairs 126,613,928 to 126,701,032 on chromosome 9 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI
Other Names for This Gene

- LIM homeo box transcription factor 1, beta
- LIM homeobox transcription factor 1, beta
- LMX1.2
- LMX1B_HUMAN
- MGC138325
- MGC142051
- NPS1

Additional Information & Resources

Educational Resources

  https://www.ncbi.nlm.nih.gov/books/NBK10053/

Clinical Information from GeneReviews

- Nail-Patella Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1132

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28LMX1B%5BTIAB%5D%29%29+OR+%28NPS1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- LIM HOMEOBOX TRANSCRIPTION FACTOR 1, BETA
  http://omim.org/entry/602575

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_LMX1B.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=LMX1B%5Bgene%5D
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:4010
Sources for This Summary


Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/gene/LMX1B

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