



## 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

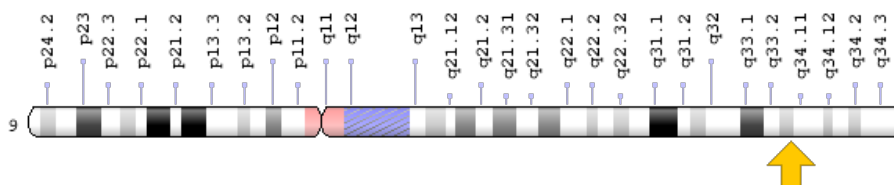
#### Nail-patella syndrome

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 126,614,443 to 126,701,032 on chromosome 9 (Homo sapiens Annotation Release 109, GRCh38.p12) (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

- Developmental Biology (sixth edition, 2000): The Generation of the Dorsal-Ventral Axis  
<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+OR+%28NPS1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

### Catalog of Genes and Diseases from OMIM

- LIM HOMEBOX 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](http://atlasgeneticsoncology.org/Genes/GC_LMX1B.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=LMX1B%5Bgene%5D>
- HGNC Gene Symbol Report  
[https://www.genenames.org/data/gene-symbol-report#!/hgnc\\_id/HGNC:6654](https://www.genenames.org/data/gene-symbol-report#!/hgnc_id/HGNC:6654)
- Monarch Initiative  
<https://monarchinitiative.org/gene/NCBIGene:4010>

- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/4010>
- UniProt  
<https://www.uniprot.org/uniprot/O60663>

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*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/20301311>

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