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.

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

Tests Listed in the Genetic Testing Registry

- Tests of LMX1B (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4010[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?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 HOMEOBOX TRANSCRIPTION FACTOR 1, BETA; LMX1B (https://omim.org/entry/602575)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/4010)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=LMX1B[gene])

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Genomic Location

The *LMX1B* gene is found on chromosome 9 (https://medlineplus.gov/genetics/chromosome/9/).

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