P3H1 gene
prolyl 3-hydroxylase 1

Normal Function

The *P3H1* gene provides instructions for making an enzyme called prolyl-3 hydroxylase 1 (sometimes known as leprechac). This enzyme works with two other proteins, cartilage associated protein and cyclophilin B, as part of a complex that helps process certain forms of collagen. Collagens are proteins that provide strength, support, and the ability to stretch (elasticity) to many body tissues.

The complex modifies a protein building block (amino acid) called proline in collagen molecules. This modification, which is known as proline 3-hydroxylation, appears to be critical for the normal folding and assembly of collagen. It also may be important for releasing collagen molecules into the spaces around cells (the extracellular matrix). The secretion of collagen from cells is necessary for the proper formation of connective tissues, such as bones, tendons, and cartilage, that form the body's supportive framework.

Studies suggest that prolyl-3 hydroxylase 1 has several additional functions. For example, this enzyme may play a role in interactions between certain types of cells and the extracellular matrix that surrounds them. Other research indicates that prolyl-3 hydroxylase 1 may act as a tumor suppressor, preventing cells from growing and dividing too fast or in an uncontrolled way.

Health Conditions Related to Genetic Changes

**Osteogenesis imperfecta**

At least four mutations in the *P3H1* gene have been identified in people with a rare, severe form of osteogenesis imperfecta classified as type VIII. These mutations prevent cells from producing any functional prolyl-3 hydroxylase 1. Without this enzyme, certain forms of collagen are not modified through proline 3-hydroxylation. The altered collagen molecules are incorrectly folded, and some abnormal collagen is secreted from cells more slowly than usual. These collagen defects weaken connective tissues, resulting in extremely slow growth and thin, brittle bones that may fracture before birth.
**Chromosomal Location**

Cytogenetic Location: 1p34.2, which is the short (p) arm of chromosome 1 at position 34.2

Molecular Location: base pairs 42,746,335 to 42,767,084 on chromosome 1 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)

![Chromosomal Location Diagram](image)

Credit: Genome Decoration Page/NCBI

**Other Names for This Gene**

- GROS1
- growth suppressor 1
- Leucine- and proline-enriched proteoglycan 1
- MGC117314
- P3H1_HUMAN

**Additional Information & Resources**

**Educational Resources**

  https://www.ncbi.nlm.nih.gov/books/NBK26810/#A3551

  https://www.ncbi.nlm.nih.gov/books/NBK21582/

- National Institute of Child Health and Human Development: Second Gene Discovered for Recessive Form of Brittle Bone Disease (February 8, 2007)

  https://www.ncbi.nlm.nih.gov/books/NBK9874/?rendertype=figure&id=A2050
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28LEPRE1%5BTIAB%5D%29+OR+%28leprecan%5BTIAB%5D%29%29+OR+%28%28GROS1%5BALL%5D+OR+%28P3H1%5BALL%5D%29%29+AND+%22last+3600+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- PROLYL 3-HYDROXYLASE 1
  http://omim.org/entry/610339

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_P3H1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=P3H1%5Bgene%5D
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:64175
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/Q32P28

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15044469


Reprinted from Genetics Home Reference:
  https://ghr.nlm.nih.gov/gene/P3H1

Reviewed: November 2007
Published: March 5, 2019

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services