GALNS gene
galactosamine (N-acetyl)-6-sulfatase

Normal Function
The GALNS gene provides instructions for producing an enzyme called N-acetylgalactosamine 6-sulfatase. This enzyme is located in lysosomes, which are compartments within cells that break down and recycle different types of molecules. N-acetylgalactosamine 6-sulfatase is involved in the breakdown of large sugar molecules called glycosaminoglycans (GAGs) or mucopolysaccharides. Specifically, this enzyme removes a chemical group known as a sulfate from a GAG called keratan sulfate. Keratan sulfate is particularly abundant in cartilage and the clear covering of the eye (cornea).

Health Conditions Related to Genetic Changes
Mucopolysaccharidosis type IV
More than 148 mutations in the GALNS gene have been found to cause mucopolysaccharidosis type IV (MPS IV). Most of these mutations change single DNA building blocks (nucleotides) in the gene. All of the mutations that cause MPS IV reduce or eliminate the function of N-acetylgalactosamine 6-sulfatase.

The lack of N-acetylgalactosamine 6-sulfatase activity leads to the accumulation of keratan sulfate within lysosomes. Because keratan sulfate is predominantly found in cartilage and the cornea, the buildup of this substance causes skeletal abnormalities and cloudy corneas. Researchers believe that a buildup of GAGs may also cause the features of MPS IV by interfering with the functions of other proteins inside lysosomes and disrupting the movement of molecules inside the cell.
Chromosomal Location

Cytogenetic Location: 16q24.3, which is the long (q) arm of chromosome 16 at position 24.3

Molecular Location: base pairs 88,813,734 to 88,856,966 on chromosome 16 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- chondroitinase
- chondroitinsulfatase
- FLJ17434
- FLJ42844
- FLJ98217
- galactosamine (N-acetyl)-6-sulfate sulfatase
- galactose-6-sulfate sulfatase
- GALNAC6S
- galNAc6S sulfatase
- GALNS_HUMAN
- GAS
- MPS4A
- N-acetylgalactosamine-6-sulfatase
- N-acetylgalactosamine-6-sulfatase precursor
- N-acetylgalactosamine-6-sulfate sulfatase
Additional Information & Resources

Educational Resources

  https://www.ncbi.nlm.nih.gov/books/NBK1934/?rendertype=figure&id=ch41.f5

- Madam Curie Bioscience Database: Defects in Glycosaminoglycan Degradation (Mucopolysaccharidoses)
  https://www.ncbi.nlm.nih.gov/books/NBK6177/#A53462

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28GALNS%5BTIAB%5D%29+OR+%28galactosamine++6-sulfate+sulfatase%5BTIAB%5D%29+OR+%28%28MPS4A%5BTIAB%5D%29+OR+%28N-acetylglactosamine-6-sulfatase%5BTIAB%5D%29+OR+%28chondroitinsulfatase%5BTIAB%5D%29+OR+%28GALNS%5BTIAB%5D%29+OR+%28%28galactose-6-sulfate+sulfatase%5BTIAB%5D%29+OR+%28N-acetylglactosamine-6-sulfate+sulfatase%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

- GALACTOSAMINE-6-SULFATE SULFATASE
  http://omim.org/entry/612222

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=GALNS%5Bgene%5D

- HGNC Gene Symbol Report

- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:2588

- NCBI Gene

- UniProt
  https://www.uniprot.org/uniprot/P34059
Sources for This Summary

- OMIM: GALACTOSAMINE-6-SULFATE SULFATASE
  http://omim.org/entry/612222

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17876718

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16837223

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16287098

Reprinted from Genetics Home Reference:

Reviewed: July 2010
Published: June 9, 2020

Lister Hill National Center for Biomedical Communications
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