CHST3 gene
carbohydrate sulfotransferase 3

Normal Function

The CHST3 gene provides instructions for making an enzyme called chondroitin 6-O-sulfotransferase 1 or C6ST-1. This enzyme has an important role in the development and maintenance of the skeleton. In particular, it is essential for the normal development of cartilage, which is a tough, flexible tissue that makes up much of the skeleton during early development. Most cartilage is later converted to bone, except for the cartilage that continues to cover and protect the ends of bones and is present in the nose and external ears.

The C6ST-1 enzyme modifies molecules called chondroitin sulfate proteoglycans, which are abundant in cartilage and give this tissue its rubbery, gel-like consistency. The C6ST-1 enzyme carries out a process known as sulfation, in which a chemical group called a sulfate is transferred from one chemical compound to another. Specifically, the enzyme takes sulfate from a molecule called 3'-phosphoadenyl-5'-phosphosulfate (PAPS) and adds it to a specific location on chondroitin sulfate proteoglycans. Sulfation of these molecules is a critical step in cartilage formation.

Health Conditions Related to Genetic Changes

CHST3-related skeletal dysplasia

At least 24 mutations in the CHST3 gene have been found to cause CHST3-related skeletal dysplasia, a condition characterized by progressive bone and joint abnormalities. Most of the mutations change single protein building blocks (amino acids) in the C6ST-1 enzyme. Other mutations result in the production of an abnormally short version of the enzyme. Each of these genetic changes reduces or eliminates the activity of C6ST-1, preventing it from transferring sulfate groups to chondroitin sulfate proteoglycans. Defective sulfation of these molecules disrupts the normal development of cartilage and bone, resulting in short stature, joint dislocations, and the other features of CHST3-related skeletal dysplasia.
**Chromosomal Location**

Cytogenetic Location: 10q22.1, which is the long (q) arm of chromosome 10 at position 22.1

Molecular Location: base pairs 71,964,362 to 72,013,564 on chromosome 10 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)

Credit: Genome Decoration Page/NCBI

**Other Names for This Gene**

- C6ST
- C6ST-1
- C6ST1
- carbohydrate (chondroitin 6) sulfotransferase 3
- chondroitin 6-O-sulfotransferase 1
- CHST3_HUMAN
- galactose/N-acetylglucosamine/N-acetylglucosamine 6-O-sulfotransferase 0
- GST-0
- HSD

**Additional Information & Resources**

**Educational Resources**

  https://www.ncbi.nlm.nih.gov/books/NBK10056/

  https://www.ncbi.nlm.nih.gov/books/NBK1900/

**Clinical Information from GeneReviews**

- CHST3-Related Skeletal Dysplasia
  https://www.ncbi.nlm.nih.gov/books/NBK62112
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28CHST3%5BTIAB%5D%29+OR+%28%28carbohydrate+sulfotransferase+3%5BTIAB%5D%29+OR+%28C6ST%5BTIAB%5D%29+OR+%28C6ST1%5BTIAB%5D%29+OR+%28C6ST-1%5BTIAB%5D%29+OR+%28chondroitin+6-O-sulfotransferase+1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D+OR+Genetic+Phenomena%5BMH%5D%29+AND+english%5BLa%5D+AND+human%5Bmh%5D

Catalog of Genes and Diseases from OMIM

- CARBOHYDRATE SULFOTRANSFERASE 3
  http://omim.org/entry/603799

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=CHST3%5Bgene%5D

- HGNC Gene Family: Sulfotransferases, membrane bound
  https://www.genenames.org/cgi-bin/genefamilies/set/763

- HGNC Gene Symbol Report

- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:9469

- NCBI Gene

- UniProt
  https://www.uniprot.org/uniprot/Q7LGC8

Sources for This Summary


  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/9883891

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19320654

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20830804

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18698629

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