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

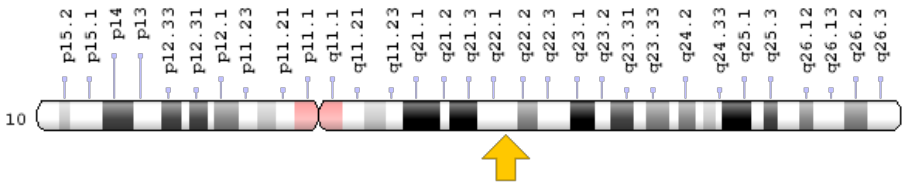
CHST3-related skeletal dysplasia

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 71,964,395 to 72,013,562 on chromosome 10 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (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

- Developmental Biology (sixth edition, 2000): Osteogenesis: The Development of Bones
  https://www.ncbi.nlm.nih.gov/books/NBK10056/
- Essentials of Glycobiology (second edition, 2009): Proteoglycans and Sulfated Glycosaminoglycans
  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%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%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 Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:1971
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:9469
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/9469
- UniProt
  https://www.uniprot.org/uniprot/Q7LGC8

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