LYST gene
lysosomal trafficking regulator

Normal Function

The LYST gene (also known as CHS1) provides instructions for making a protein known as the lysosomal trafficking regulator. Researchers believe that this protein plays a role in the transport (trafficking) of materials into structures called lysosomes. Lysosomes act as recycling centers within cells. They use digestive enzymes to break down toxic substances, digest bacteria that invade the cell, and recycle worn-out cell components. Although the lysosomal trafficking regulator protein is involved in the normal function of lysosomes, its exact role is unknown. Studies suggest that this protein may help determine the size of lysosomes and regulate their movement within cells.

Health Conditions Related to Genetic Changes

Chediak-Higashi syndrome

At least 30 mutations in the LYST gene have been identified in people with Chediak-Higashi syndrome. These mutations impair the normal function of the lysosomal trafficking regulator protein, which disrupts the size, structure, and function of lysosomes and related structures within cells.

LYST mutations that cause the severe, childhood form of Chediak-Higashi syndrome typically result in the production of an abnormally short, nonfunctional version of the lysosomal trafficking regulator protein. The mutations responsible for the milder, adult version of this disease usually change a single protein building block (amino acid) in the protein. In these cases, the altered protein may retain some function.

People with LYST mutations have abnormally large lysosomes and related structures in cells throughout the body. These enlarged structures interfere with normal cell functions. For example, enlarged lysosomes in certain immune system cells prevent these cells from responding appropriately to bacteria and other foreign invaders. As a result, the malfunctioning immune system cannot protect the body from severe, recurrent infections.

In pigment cells called melanocytes, cellular structures called melanosomes (which are related to lysosomes) are abnormally large. These structures produce and distribute a pigment called melanin, which is the substance that gives skin, hair, and eyes their color. People with Chediak-Higashi syndrome have oculocutaneous albinism because melanin is trapped within the giant melanosomes and is unable to contribute to skin, hair, and eye pigmentation.
Researchers believe that abnormal lysosome-like structures inside blood cell fragments called platelets underlie the abnormal bruising and bleeding seen in people with Chediak-Higashi syndrome. Similarly, abnormal lysosomes in nerve cells probably cause the neurological problems associated with this disease.

**Chromosomal Location**

Cytogenetic Location: 1q42.3, which is the long (q) arm of chromosome 1 at position 42.3

Molecular Location: base pairs 235,661,031 to 235,883,708 on chromosome 1 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)

**Other Names for This Gene**

- Beige homolog
- Chediak-Higashi syndrome 1
- CHS
- CHS1
- LYST_HUMAN

**Additional Information & Resources**

**Educational Resources**

- Eurekah Bioscience Collection: Defects in Trafficking
  https://www.ncbi.nlm.nih.gov/books/NBK6177/#A53468

  https://www.ncbi.nlm.nih.gov/books/NBK21743/#A1185

**Clinical Information from GeneReviews**

- Chediak-Higashi Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK5188
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28LYST%5BTIAB%5D%29+OR+%28lysosomal+trafficking+regulator%5BTIAB%5D%29+OR+%28%28Chediak-Higashi+syndrome%29+OR+%28CHS1+AND+Chediak-Higashi%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- LYSOSOMAL TRAFFICKING REGULATOR
  http://omim.org/entry/606897

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_LYST.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=LYST%5Bgene%5D
- HGNC Gene Family: BEACH domain containing
  https://www.genenames.org/cgi-bin/genefamilies/set/1230
- HGNC Gene Family: WD repeat domain containing
  https://www.genenames.org/cgi-bin/genefamilies/set/362
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:1130
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/Q99698

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11487012
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10527680
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18043242
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11857544

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11984006 
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2039936/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12125812

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17554367

Reprinted from Genetics Home Reference:

Reviewed: February 2008
Published: October 2, 2018

Lister Hill National Center for Biomedical Communications
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