

LYST Gene

Subjects: **Genetics & Heredity**

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Lysosomal trafficking regulator

genes

1. Introduction

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.

2. Health Conditions Related to Genetic Changes

2.1. 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 cells 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.

3. Other Names for This Gene

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

References

1. Huizing M, Anikster Y, Gahl WA. Hermansky-Pudlak syndrome and Chediak-Higashi syndrome: disorders of vesicle formation and trafficking. *Thromb Haemost.* 2001Jul;86(1):233-45. Review.
2. Introne W, Boissy RE, Gahl WA. Clinical, molecular, and cell biological aspects of Chediak-Higashi syndrome. *Mol Genet Metab.* 1999 Oct;68(2):283-303. Review.
3. Kaplan J, De Domenico I, Ward DM. Chediak-Higashi syndrome. *Curr Opin Hematol.* 2008 Jan;15(1):22-9. Review.
4. Karim MA, Suzuki K, Fukai K, Oh J, Nagle DL, Moore KJ, Barbosa E, Falik-Borenstein T, Filipovich A, Ishida Y, Kivrikko S, Klein C, Kreuz F, Levin A, Miyajima H, Regueiro JR, Russo C, Uyama E, Vierimaa O, Spritz RA. Apparent genotype-phenotype correlation in childhood, adolescent, and adult Chediak-Higashi syndrome. *Am J Med Genet.* 2002 Feb 15;108(1):16-22.
5. Tchernev VT, Mansfield TA, Giot L, Kumar AM, Nandabalan K, Li Y, Mishra VS, Detter JC, Rothberg JM, Wallace MR, Southwick FS, Kingsmore SF. The Chediak-Higashi protein interacts with SNARE complex and signal transduction proteins. *Mol Med.* 2002 Jan;8(1):56-64.

6. Ward DM, Shiflett SL, Kaplan J. Chediak-Higashi syndrome: a clinical and molecular view of a rare lysosomal storage disorder. *Curr Mol Med*. 2002 Aug;2(5):469-77. Review.
 7. Westbroek W, Adams D, Huizing M, Koshoffer A, Dorward H, Tinloy B, Parkes J, Helip-Wooley A, Kleta R, Tsilou E, Duvernay P, Digre KB, Creel DJ, White JG, Boissy RE, Gahl WA. Cellular defects in Chediak-Higashi syndrome correlate with the molecular genotype and clinical phenotype. *J Invest Dermatol*. 2007 Nov;127(11):2674-7.
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