



[Home](#) → [Medical Encyclopedia](#) → Chediak-Higashi syndrome

URL of this page: [//medlineplus.gov/ency/article/001312.htm](https://medlineplus.gov/ency/article/001312.htm)

Chediak-Higashi syndrome

Chediak-Higashi syndrome is a rare disease of the immune and nervous systems. It involves pale-colored hair, eyes, and skin.

Causes

Chediak-Higashi syndrome is passed down through families (inherited). It is an autosomal recessive disease. This means that both parents are carriers of a non-working copy of the gene. Each parent must pass their non-working gene to the child for them to show symptoms of the disease.

Defects have been found in the *LYST* (also called *CHS1*) gene. The primary defect in this disease is found in certain substances normally present in skin cells and certain white blood cells.

Symptoms

Children with this condition may have:

- Silver hair, light-colored eyes (albinism)
- Increased infections in the lungs, skin, and mucous membranes
- Jerky eye movements (nystagmus)

Infection of affected children with certain viruses, such as Epstein-Barr virus (EBV), can cause a life-threatening illness resembling the blood cancer lymphoma called hemophagocytic lymphohistiocytosis.

Other symptoms may include:

- Decreased vision
- Intellectual disability
- Muscle weakness
- Nerve problems in the limbs (peripheral neuropathy)
- Nosebleeds or easy bruising
- Numbness
- Tremor
- Seizures

- Sensitivity to bright light (photophobia)
- Unsteady walking (ataxia)

Exams and Tests

Your health care provider will perform a physical exam. This may show signs of a swollen spleen or liver or jaundice.

Tests that may be done include:

- Complete blood count, including white blood cell count
- Blood platelet count
- Blood culture and smear
- Brain MRI or CT
- EEG
- EMG
- Nerve conduction tests

Treatment

There is no specific treatment for Chediak-Higashi syndrome. High doses of vitamin C may help some children whose condition is in the stable phase. Hematopoietic stem cells appear to have been successful in several patients.

Antibiotics are used to treat and, sometimes, prevent infections. Antiviral drugs, such as acyclovir, and chemotherapy drugs are often used in the accelerated phase of the disease. Blood and platelet transfusions are given as needed. Surgery may be needed to drain abscesses in some cases.

Support Groups

More information and support for people with Chediak-Higashi syndrome and their families can be found at:

- National Organization for Rare Disorders (NORD) -- rarediseases.org/rare-diseases/chediak-higashi-syndrome/ [<https://rarediseases.org/rare-diseases/chediak-higashi-syndrome/>]

Outlook (Prognosis)

Death often occurs in the first 10 years of life, from long-term (chronic) infections or accelerated disease that results in the lymphoma-like illness (hemophagocytic lymphohistiocytosis). However, some affected children have survived longer.

Possible Complications

Complications may include:

- Frequent infections involving certain types of bacteria
- Lymphoma-like cancer triggered by viral infections such as EBV
- Early death

When to Contact a Medical Professional

Contact your provider if you have a family history of this disorder and you are planning to have children.

Talk to your provider if your child shows symptoms of Chediak-Higashi syndrome.

Prevention

Genetic counseling is recommended before becoming pregnant if you have a family history of Chediak-Higashi syndrome.

References

Coates TD. Disorders of phagocyte function. In: Kliegman RM, St. Geme JW, Blum NJ, Shah SS, Tasker RC, Wilson KM, eds. *Nelson Textbook of Pediatrics*. 21st ed. Philadelphia, PA: Elsevier; 2020:chap 156.

Dinauer MC, Coates TD. Disorders of phagocyte function. In: Hoffman R, Benz EJ, Silberstein LE, et al, eds. *Hematology: Basic Principles and Practice*. 8th ed. Philadelphia, PA: Elsevier; 2023:chap 51.

Holland SM, Gallin JI. Evaluation of the patient with suspected immunodeficiency. In: Bennett JE, Dolin R, Blaser MJ, eds. *Mandell, Douglas, and Bennett's Principles and Practice of Infectious Diseases*. 9th ed. Philadelphia, PA: Elsevier; 2020:chap 12.

Review Date 9/18/2023

Updated by: Anna C. Edens Hurst, MD, MS, Associate Professor in Medical Genetics, The University of Alabama at Birmingham, Birmingham, AL. Review provided by VeriMed Healthcare Network. Also reviewed by David C. Dugdale, MD, Medical Director, Brenda Conaway, Editorial Director, and the A.D.A.M. Editorial team.

Learn how to cite this page



A.D.A.M., Inc. is accredited by [URAC](http://www.urac.org), for Health Content Provider (www.urac.org). URAC's [accreditation program](#) is an independent audit to verify that A.D.A.M. follows rigorous standards of quality and accountability. A.D.A.M. is among the first to achieve this important distinction for online health information and services. Learn more about A.D.A.M.'s [editorial policy](#), [editorial process](#), and [privacy policy](#).

The information provided herein should not be used during any medical emergency or for the diagnosis or treatment of any medical condition. A licensed medical professional should be consulted for diagnosis and treatment of any and all medical conditions. Links to other sites are provided for information only – they do not constitute endorsements of those other sites. No warranty of any kind, either expressed or implied, is made as to the accuracy, reliability, timeliness, or correctness of any translations made by a third-party service of the information provided herein into any other language. © 1997-2025 A.D.A.M., a business unit of Ebix, Inc. Any duplication or distribution of the information contained herein is strictly prohibited.

