Chediak-Higashi Syndrome

National Cancer Institute

Source

National Cancer Institute. Chediak-Higashi Syndrome. NCI Thesaurus. Code C2941.

A rare autosomal recessive immunodeficiency disorder characterized by abnormal intracellular protein transport. Chediak-Higashi syndrome (CHS) is characterized by immune deficiency; partial oculocutaneous albinism; a bleeding disorder due to deficient platelet dense bodies; neutropenia; neutrophils with impaired chemotaxis and bactericidal activity; recurrent infection; and abnormal natural killer (NK) cell function. CHS may be associated with hepatosplenomegaly, lymphadenopathy, anemia, thrombocytopenia, roentgenologic changes in bones, lungs and heart, and skin and psychomotor abnormalities; it is often fatal in childhood as a result of infection or an accelerated lymphoma-like phase. CHS occurs in mink, cattle, and mice, as well as man.