Chediak-Higashi Syndrome

Chediak-Higashi syndrome is an autosomal recessive disorder caused by a defect in microtubule polymerization. This defect leads to a decrease in phagocytosis and impaired lysosome degranulation within phagosomes which rely on microtubular polymerization. This defect leads to recurrent pyogenic infections, often caused by Staph aureus and Streptococci species. Individuals with Chediak-Higashi syndrome also commonly have partial albinism, and are described to have light skin and silvery hair due to defective melanization of melanosomes. In the melanocytes, which are the cells responsible for pigmentation, autophagocytosis of melanosomes occurs due to the defective microtubule polymerization. Neurologic involvement is variable but often includes peripheral neuropathy. The mechanism of peripheral neuropathy has not been elucidated but axonal and demyelinating peripheral neuropathy have been noted. The infections in Chediak-Higashi syndrome tend to be serious and even life-threatening with very few patients with this condition living to adulthood.



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Pathophysiology

Autosomal Recessive
Recessive-chocolate
This syndrome is inherited in an autosomal recessive fashion.

Defective Lysosomal Trafficking Regulator Gene (LYST)
Broken LYST-Lights for traffic
Mutations in the LYST gene lead to impaired function of lysosomes throughout the body, and gives rise to abnormal lysosomal size and structure. As normal cell function is dysfunctional, immune cells are unable to appropriately respond to bacteria and other foreign invaders.

Defect In Microtubular Function
Broken Microtubule-truck
Chediak-Higashi syndrome is caused by a defect in microtubule polymerization, leading to decreased phagocytosis and impaired lysosome degranulation within phagosomes and defective melanocytes.

Decrease In Phagocytosis
Down-arrows on Mac-man
Defective microtubular function leads to a decrease in phagocytosis and impaired lysosome degranulation within phagosomes which rely on microtubular polymerization. This defect leads to recurrent pyogenic infections, often caused by Staph aureus and Streptococci species.

Signs And Symptoms

Recurrent Pyogenic Infections
Pus-pie
Defective phagocytosis and impaired lysosome degranulation within the phagocytes leads to recurrent pyogenic infections that are often severe and can be life threatening.

Staph Aureus And Streptococci
Staff-of-Oreos and Stripper-with-cock-eyes
Individuals with Chediak-Higashi suffer from recurrent pyogenic infections, often caused by Staph aureus and Streptococci species.

Partial Albinism
Albino owl
Individuals with Chediak-Higashi syndrome commonly have partial albinism and are described to have light skin and silvery hair due to defective melanization of melanosomes. In the melanocytes, which are the cells responsible for pigmentation, autophagocytosis of melanosomes occurs.

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Peripheral Neuropathy

Purple-wavy Neuron-extremities

Peripheral neuropathy is a general term that describes damage to the nerves of the peripheral nervous system. Symptoms can include abnormal sensory function and motor symptoms. The mechanism of peripheral neuropathy in Chediak-Higashi syndrome has not been elucidated but axonal and demyelinating peripheral neuropathy have been noted.

Pancytopenia

Pan-side-toe-peanut

Typically patients suffer from multiple infections before they enter an accelerated phase, characterized by fever, lymphadenopathy, jaundice, and widespread lymphohistiocytic organ infiltrates. During this phase, patients also develop pancytopenia in the form of anemia, neutropenia and thrombocytopenia.

Diagnosis

Giant Granules In Granulocytes And Platelets

Granules with Granny and Plates

Diagnosis of Chediak-Higashi syndrome is made through the presence of eosinophilic peroxidase-positive, abnormally large "giant" granules in granulocytes and platelets.

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