

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.

### **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.