Griscelli Syndrome Type 2

National Cancer Institute

Source

National Cancer Institute. Griscelli Syndrome Type 2. NCI Thesaurus. Code C111814.

A rare, autosomal recessive genetic syndrome caused by mutations in the RAB27A gene. It is characterized by hypopigmentation of the skin, hair and eyes, recurrent infections, neutropenia, and immune system abnormalities. Patients are prone to develop hemophagocytic lymphohistiocytosis.