Opis choroby *

Definicja

A rare subtype of Griscelli syndrome characterized by pigmentary dilution in skin and hair with irregular clumps of pigment in hair shafts resulting in silvery hair, in association with increased susceptibility to recurrent infections and immunological abnormalities, in particular impairment of T-cell and natural killer cytotoxic activity eventually leading to hemophagocytic lymphohistiocytosis. Patients may present neurological manifestations related to infiltration of the central nervous system in the context of the hemophagocytic syndrome. The disease is mostly fatal in the first decade of life.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
Griscelli-Pruniéras syndrome type 2
Hipopigmentacja - Niedobór odporności z lub bez upośledzenia neurologicznego
Zespół Griscelliego i Pruniérasa typu 2
Hypopigmentation-immunodeficiency with or without neurologic impairment syndrome

Kod ORPHA
79477

Kod OMIM
607624

Kod ICD10
E70.3

Kod ICD11
4A01.23

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*Źródło

orphanet