

Opis choroby *

Definicja

An extremely rare form of oculocutaneous albinism type 1 characterized by temperature sensitive hair pigmentation leading to dark hair on the hands, feet, legs, arms and chest (cooler body areas) and white or pale yellow hair on the scalp, axilla and pubic area (warmer body areas). Nystagmus and reduced visual acuity are also noted.

Dane

Klasyfikacja

Podtyp kliniczny

Synonimy

OCA1-TS

OCA1-TS

TS OCA type 1

TS OCA type 1

Kod ORPHA

352737

Kod OMIM

606952

Kod ICD10

E70.3

Kod ICD11

EC23.20

*Źródło

orphanet