

Opis choroby *

Definicja

A severe form of oculocutaneous albinism type 1 (OCA1) characterized by complete absence of melanin and manifesting as white hair and skin, blue, fully translucent irises, nystagmus and misrouting of the optic nerves.

Dane

Klasyfikacja

Podtyp kliniczny

Synonimy

OCA1A

Albinizm oczno-skórny tyrozynazo-ujemny

OCA1A

Tyrosinase-negative oculocutaneous albinism

Kod ORPHA

79431

Kod OMIM

203100

Kod ICD10

E70.3

Kod ICD11

EC23.20

*Źródło

orphanet