Opis choroby *

Definicja

An extremely rare form of Oculocutaneous albinism type 1 with minimal pigment present, characterized by blond hair (white at birth), variable iris transillumination (blue irides at birth followed by minimal development of pigment during the first decade of life), visual acuity ranging from 20/80-20/200 and white skin, with or without skin nevi.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
MP OCA type 1
MP OCA type 1
OCA1-MP
OCA1-MP

Kod ORPHA
352734

Kod OMIM
-

Kod ICD10
E70.3

Kod ICD11
EC23.20

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*Źródło

orphanet