Opis choroby *

Definicja

A form of oculocutaneous albinism (OCA) characterized by a spectrum of hypopigmentation of skin hair and eyes, ranging from little or no pigmentation to localized pigementation. Nystagmus, photophobia and reduced visual acuity are frequently present. The subtypes include OCA1A, OCA1B, type 1 minimal pigment oculocutaneous albinism (OCA1-MP) and type 1 temperature sensitive oculocutaneous albinism (OCA1-TS).

Dane

Klasyfikacja
Choroba

Synonimy
OCA1
OCA1

Kod ORPHA
352731

Kod OMIM
606952

Kod ICD10
E70.3

Kod ICD11
EC23.20

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*Źródło

orphanet