Opis choroby *

Definicja

A group of rare genetic hypopigmentation disorders characterized by a generalized reduction in pigmentation of hair, skin and eyes and variable ocular findings including nystagmus, reduced visual acuity and photophobia. Variants include OCA1A (the most severe form), OCA1B, OCA1-minimal pigment (OCA1-MP), OCA1-temperature sensitive (OCA1-TS), OCA2, OCA3, OCA4, OCA5, OCA6, OCA7 and OCA8.

Dane

Klasyfikacja
Grupa fenomenów

Synonimy
OCA
OCA

Kod ORPHA
55

Kod OMIM
-

Kod ICD10
E70.3

Kod ICD11
EC23.20

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*Źródło

orphanet