

Opis choroby *

Definicja

A form of oculocutaneous albinism (OCA) characterized by skin and hair hypopigmentation (light blond to dark brown), nystagmus, iris transillumination, visual acuity ranging from 6/9 to 3/60 and hypopigmentation of the peripheral ocular fundus. Photophobia is not a major feature.

Dane

Klasyfikacja

Choroba

Synonimy

OCA7

OCA7

Kod ORPHA

352745

Kod OMIM

615179

Kod ICD10

E70.3

Kod ICD11

EC23.20

*Źródło

orphanet