

Opis choroby *

Definicja

A form of oculocutaneous albinism characterized by light hair at birth that darkens with age, white skin, transparent irides, photophobia, nystagmus, foveal hypoplasia and reduced visual acuity.

Dane

Klasyfikacja

Choroba

Synonimy

OCA6

OCA6

Kod ORPHA

370097

Kod OMIM

113750

Kod ICD10

E70.3

Kod ICD11

EC23.20

*Źródło

orphanet