

Albinizm oczno-skórny typu 5

Kod Orpha: 370091 Kod OMIM: 615312

Opis choroby *

Definicja

A form of oculocutaneous albinism characterized by white skin, golden hair, photophobia, nystagmus, foveal hypoplasia and impaired visual acuity, that affects males and females equally. Patients have been reported only in a consanguineous Pakistani family. The responsible gene has not yet been detected.

Dane

Klasyfikacja

Choroba

Synonimy

OCA5

OCA5

Kod ORPHA

370091

Kod OMIM

615312

Kod ICD10

E70.3

Kod ICD11

EC23.20

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.