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

Kod ORPHA

Synonimy

OCA5

Kod OMIM

Kod ICD10

370091

615312

E70.3

Kod ICD11 EC23.20

<u>*Źródło</u>

orphanet