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