Opis choroby *

Definicja

A rare, syndromic, inherited retinal disorder characterized by cone-rod type congenital amaurosis, severe retinal dystrophy leading to visual impairment and profound photophobia (without night blindness), and trichomegaly (bushy eyebrows with synophrys, excessive facial and body hair (including marked circumaleolar hypertrichosis). There have been no further descriptions in the literature since 1989.

Dane

Klasyfikacja
Choroba

Kod ORPHA
1021

Kod OMIM
204110

Kod ICD10
H35.5

Kod ICD11
9B70

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*Źródło

orphanet