

Opis choroby *

Definicja

A rare genetic eye disease characterized by optic disc anomalies (bilateral colobomatous optic discs, retinal vessels arising from the peripheral optic disc) and macular atrophy. Peripapillary chorioretinal atrophy and chorioretinal and iris coloboma have also been described. Patients present with horizontal nystagmus and poor visual acuity.

Dane

Klasyfikacja

Choroba

Kod ORPHA

435930

Kod OMIM

212550

Kod ICD10

Q14.8

Kod ICD11

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*Źródło

orphanet