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