

Opis choroby *

Definicja

A rare congenital optic disc excavation characterized by deep fundus excavation of chorioretinal atrophy surrounding a relatively normal appearing optic disc. Retinal vasculature is normal, and retinochoroidal coloboma and glial anomalies are absent. Patients present with mostly unilateral markedly reduced visual acuity. Association with other congenital defects or systemic diseases is uncommon.

Dane

Klasyfikacja

Wada morfologiczna

Kod ORPHA

519400

Kod OMIM

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Kod ICD10

Q14.2

Kod ICD11

LA13.7Y

*Źródło

orphanet