Opis choroby *

Definicja

Coloboma of choroid and retina is a rare, genetic developmental defect during embryogenesis characterized by the partial absence of retinal pigment epithelium and choroid, most frequently located in the inferonasal quadrant. Patients usually present reduced vision and have an increased risk for retinal detachment. Other ocular anomalies (e.g. coloboma of iris, microcornea, nystagmus, strabismus, microphthalmos) are usually associated, however it may also be isolated.

Dane

Klasyfikacja
Wada morfologiczna

Kod ORPHA
98942

Kod OMIM
120200

Kod ICD10
Q14.8

Kod ICD11
LA13.1

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*Źródło

orphanet