

Opis choroby *

Definicja

Coloboma of macula is a rare, non-syndromic developmental defect of the eye characterized by well-circumscribed, oval or rounded, usually unilateral, atrophic lesions of varying size presenting rudimentary or absent retina, choroid and sclera located at the macula leading to decreased vision and, on occasion, other symptoms (e.g. strabismus). It is usually isolated, but may also be associated with Down syndrome, skeletal or renal disorders.

Dane

Klasyfikacja

Wada morfologiczna

Kod ORPHA

98945

Kod OMIM

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Kod ICD10

Q14.8

Kod ICD11

LA13.2

*Źródło

orphanet