Opis choroby *

Definicja

A rare, genetic, developmental defect of the eye characterized by a uni- or bilateral notch, gap, hole or fissure, typically located in the inferonasal quadrant of the eye, involving only the pigment epithelium or the iris stroma (incomplete) or involving both (complete), manifesting with iris shape anomalies (e.g. 'keyhole' or oval pupil) and/or photophobia. Association with colobomata in other parts of the eye (incl. ciliary body, zonule, choroid, retina, optic nerve) and complex malformation syndromes (such as CHARGE syndrome) may be observed.

Dane

Klasyfikacja
Wada morfologiczna

Kod ORPHA
98944

Kod OMIM
120200

Kod ICD10
Q13.0

Kod ICD11
LA11.4

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*Źródło

orphanet