

Szczelina tęczówki

Kod Orpha: 98944 Kod OMIM: 120200

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

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.