Opis choroby *

Definicja

A rare, genetic, developmental defect of the eye characterized by a uni- or bilateral abnormal lens shape (contraction of the lens with a notch) due to segmentally defective, or absent, development of the zonule and flattening of the equator in the region of the zonular defect, typically manifesting with reduced visual acuity. Other ocular anomalies, such as iris, choroid or optic disc colobomas, as well as cataracts and retinal detachment, may be associated.

Dane

Klasyfikacja
Wada morfologiczna

Kod ORPHA
98943

Kod OMIM
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Kod ICD10
Q12.2

Kod ICD11
LA12.0

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*Źródło

orphanet