Opis choroby *

Definicja

A rare developmental defect of the eye characterized by usually bilateral absence of the normal protrusion of the cornea from the sclera, the corneal curvature being the same as that of the adjacent sclera. Most patients develop hyperopia, hazy corneal limbus, and arcus lipoides at an early age. The condition may present as an autosomal dominant or an autosomal recessive form, with the latter showing more severe signs and symptoms (such as a round and opaque thickening located centrally in the cornea) and more frequent association with other ocular anomalies.

Dane

Klasyfikacja
Wada morfologiczna

Kod ORPHA
53691

Kod OMIM
217300

Kod ICD10
Q13.4

Kod ICD11
LA11.1

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*Źródło

orphanet