Opis choroby *

Definicja

A rare genetic eye disease characterized by microcornea, coloboma of the iris and the optic disc, axial enlargement of the globe, staphyloma, and severe myopia. Additional manifestations are mild cornea plana, iridocorneal angle abnormalities with elevation of intraocular pressure, and shallow anterior chamber depth. Variable expressivity of the phenotype has been described, including unilateral or bilateral involvement, or variable extent of coloboma, among other features.

Dane

Klasyfikacja
Choroba

Synonimy
MACOM syndrome
Zespół MACOM

Kod ORPHA
468672

Kod OMIM
602499

Kod ICD10
Q15.8

Kod ICD11
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*Źródło

orphanet