Opis choroby *

Definicja

A rare, genetic, non-syndromic developmental defect of the eye disorder, with high clinical and genetic heterogeneity, most frequently characterized by bilateral, symmetrical, non-progressive cataracts which present at birth or in early-childhood. Additional ocular manifestations (e.g. anterior segment dysgenesis, colobomas, nystagmus, microcornea, microphthalmia, myopia) may be associated, however other organs/systems are usually not affected.

Dane

Klasyfikacja
Choroba

Kod ORPHA
91492

Kod OMIM
601885

Kod ICD10
Q12.0

Kod ICD11
LA12.1

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*Źródło

orphanet