

Opis choroby *

Definicja

A rare syndrome described in three members of a family (a boy, his father, and his paternal grandmother) that is characterized by the association of aniridia with patella aplasia or hypoplasia. The grandmother also had bilateral cataracts and glaucoma. There have been no further descriptions in the literature since 1975.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

1069

Kod OMIM

106220

Kod ICD10

Q87.8

Kod ICD11

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*Źródło

orphanet