Opis choroby *

Definicja

A rare, genetic, multiple congenital anomalies/dysmorphic syndrome characterized by uveal coloboma (typically bilateral) variably associated with cleft lip, palate and/or uvula, hearing impairment, and intellectual disability. The spectrum of eye involvement is also variable and includes iris coloboma extending to the choroid, disc, and/or macula, microphthalmia, cataract, and extraocular movement impairment.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
1473

Kod OMIM
120433

Kod ICD10
Q13.8

Kod ICD11
LD2F.1Y

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*Źródło

orphanet