Opis choroby *

Definicja

A rare multiple congenital anomalies/dysmorphic syndrome characterized by intellectual disability, severe visual impairment due to ocular malformations (microphthalmos and microcornea with sclerocornea), short stature, hypotrichosis, dental anomalies, and dysmorphic facial features (such as a narrow nasal bridge with marked distal flaring and low-set, protruding ears). There have been no further descriptions in the literature since 1992.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
1806

Kod OMIM
268320

Kod ICD10
Q87.8

Kod ICD11
LD27.0Y

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*Źródło

orphanet