Opis choroby *

Definicja

A rare developmental defect during embryogenesis characterized by macrostomia or abnormal mouth contour, preauricular tags or pits, and uni- or bilateral ptosis due to external ophthalmoplegia. This syndrome belongs to the oculoauriculovertebral spectrum, a developmental disorder affecting the structures derived from the first and second branchial arches.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
83619

Kod OMIM
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Kod ICD10
Q87.0

Kod ICD11
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*Źródło

orphanet