

Opis choroby *

Definicja

A rare congenital malformation syndrome, most commonly presenting with hemifacial microsomia associated with ear and/or eye malformations and vertebral anomalies of variable severity. Additional malformations involving the heart, kidneys, central nervous, digestive and skeletal systems may also be associated.

Dane

Klasyfikacja

Zespół wad wrodzonych OAV spectrum

Synonimy

Jednostronna lub obustronna i asymetryczna dysplazja uszno-żuchwowa
Spektrum OAV
Oculoauriculovertebral spectrum

Kod ORPHA

141132

Kod OMIM

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Kod ICD10

Q87.0

Kod ICD11

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*Źródło

orphanet