

Opis choroby *

Definicja

A rare, isolated, congenital, head and neck morphological anomaly characterized by the unilateral hypoplasia/agenesis of the depressor anguli oris muscle, resulting in an asymmetric crying facies in neonatal period/infancy (drooping of one corner of the mouth during crying) while eye closure, nasolabial fold and forehead wrinkling are symmetric. Although isolated in the majority of cases, newborns presenting with this morphological anomaly should be referred for further screening for 22q11.2 deletion syndrome and/or other coexisting cardiovascular, musculoskeletal, cervicofacial, respiratory, genitourinary and endocrine anomalies.

Dane

Klasyfikacja

Wada morfologiczna

Synonimy

Isolated asymmetric crying facies

Izolowana asymetryczna płacząca twarz

Kod ORPHA

1166

Kod OMIM

-

Kod ICD10

Q87.0

Kod ICD11

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*Źródło

orphanet