Opis choroby *

Definicja

An extremely rare and fatal association syndrome, characterized by absence of the mandible, cerebral malformations with facial anomalies related to a defect in cleavage in the embryonic brain (e.g. synophthalmia, malformed and low-set ears fused in midline (otocephaly), agenesis of the olfactory bulbs, microstomia, hypoglossia/aglossia) and situs inversus partialis or totalis.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
990

Kod OMIM
202650

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet