

Opis choroby *

Definicja

Hypomandibular faciocranial dysostosis is a cranial malformation characterized by facial dysmorphism (proptosis, frontal bossing, midface and zygomatic arches hypoplasia, short nose with anteverted nostrils, microstomia with persistent buccopharyngeal membrane, severe hypoglossia with glossoptosis, severe mandibular hypoplasia, and low set ears) associated with laryngeal hypoplasia and craniosynostosis. Other variable features include cleft palate, optic nerve coloboma and choanal stenosis.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

1790

Kod OMIM

241310

Kod ICD10

Q75.4

Kod ICD11

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*Źródło

orphanet