

Opis choroby *

Definicja

A rare mandibulofacial dysostosis characterized by the association with scalp alopecia and sparse eyebrows and eyelashes. Craniofacial dysmorphic features include zygomatic and mandibular dysplasia or hypoplasia, cleft palate, micrognathia, dental anomalies, auricular dysmorphism, and eyelid anomalies, among others. Patients may experience limited jaw mobility, glossoptosis, upper airway obstruction, and conductive hearing loss.

Dane

Klasyfikacja

Zespół wad wrodzonych MFDA

Synonimy

MFDA

Obustronna i symetryczna dysplazja uszno-
żuchwowa z łysieniem

Kod ORPHA

443995

Kod OMIM

616367

Kod ICD10

Q75.4

Kod ICD11

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*Źródło

orphanet