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

Synonimy
MFDA
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