

Opis choroby *

Definicja

A rare genetic, multiple congenital malformation syndrome characterized by malar and mandibular hypoplasia, microcephaly, ear malformations with associated conductive hearing loss, distinctive facial dysmorphism (with significantly overlap to Treacher Collins syndrome), developmental delay, and intellectual disability.

Dane

Klasyfikacja

Zespół wad wrodzonych MFDM syndrome

Synonimy

MFDM syndrome

Zespół MFDM

Mandibulofacial dysostosis, Guion-Almeida type

Kod ORPHA

79113

Kod OMIM

610536

Kod ICD10

Q87.0

Kod ICD11

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*Źródło

orphanet