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

Synonimy
MFDM syndrome
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