Opis choroby *

Definicja

A very rare, syndromic genetic deafness characterized by mild to moderate conductive hearing loss, dysmorphic pinnae and lip pits or dimples. The pinnae are usually small, cup-shaped, with helix folded forward, and hearing loss is associated with malformed ossicles and displacement of the external auditory canal.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Conductive hearing loss-malformed external ear syndrome
Zespół Mengela i Konigsmarka
Mengel-Konigsmark syndrome

Kod ORPHA
3216

Kod OMIM
221300

Kod ICD10
Q17.8

Kod ICD11
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*Źródło

orphanet