

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

Kod ORPHA	Kod OMIM	Kod ICD10
3216	221300	Q17.8

Kod ICD11

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*Źródło

orphanet