

Opis choroby *

Definicja

Dysmorphism-conductive hearing loss-heart defect syndrome is a rare, multiple congenital anomalies syndrome characterized by a distinctive facial appearance (low frontal hairline, bilateral ptosis, prominent eyes, flat midface, broad, ﬂat nares, Cupid's bow upper lip vermilion, and small, low-set, posteriorly rotated ears), in addition to cleft palate, conductive hearing loss, heart defects (atrial or ventricular septal defect) and mild developmental delay/intellectual disability.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

289553

Kod OMIM

615102

Kod ICD10

Q87.8

Kod ICD11

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*Źródło

orphanet