

Opis choroby *

Definicja

A rare multiple congenital anomalies/dysmorphic syndrome characterized by auditory canal atresia (resulting in moderate conductive hearing loss) associated with intellectual disability, ventricular septal defect, umbilical hernia, anteriorly displaced anus, various skeletal anomalies (such as mild clubfoot, long fifth fingers, proximally placed thumbs), and craniofacial dysmorphism which includes brachycephaly, prominent forehead, flattened occiput, midface hypoplasia, anteverted nares, and low set, posteriorly rotated ears with overlapping superior helix. There have been no further descriptions in the literature since 1987.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Aural atresia-multiple congenital anomalies-intellectual disability syndrome
	Atrezja ucha - mnogie wady wrodzone - opóźnienie w rozwoju

Kod ORPHA
1488

Kod OMIM
209770

Kod ICD10
Q87.8

Kod ICD11

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*Źródło

orphanet