

Opis choroby *

Definicja

Branchiogenic deafness syndrome is a multiple congenital anomalies syndrome, described in one family to date, characterized by branchial cysts or fistulae; ear malformations; congenital hearing loss (conductive, sensorineural, and mixed); internal auditory canal hypoplasia; strabismus; trismus; abnormal fifth fingers; vitiliginous lesions, short stature; and mild learning disability. Renal and uretral abnormalities are absent.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Branchiogenic hearing loss syndrome Zespół Mégarbané i Loiselet Mégarbané-Loiselet syndrome

Kod ORPHA	Kod OMIM	Kod ICD10
50815	609166	Q87.0

Kod ICD11

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*Źródło

orphanet