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
Zespół wad wrodzonych

Synonimy
Branchiogenic hearing loss syndrome
Zespół Mégarbané i Loiselet
Mégarbané-Loiselet syndrome

Kod ORPHA
50815

Kod OMIM
609166

Kod ICD10
Q87.0

Kod ICD11
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*Źródło

orphanet