

Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by growth retardation, short stature, feeding difficulty and failure to thrive, cardiac anomalies (septal defects and/or valve dysplasia), joint laxity, short extremities, brachydactyly, carpal and tarsal fusion, cervical vertebral fusion, inner ear malformation with bilateral conductive hearing loss, and dysmorphic facial features (such as hypertelorism, upslanting palpebral fissures, posteriorly rotated ears, anteverted nares, and long philtrum). Additional variable manifestations include gastroesophageal reflux and genitourinary anomalies, among others.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Forney syndrome

Niedomykalność zastawki mitralnej - głuchota -
wady szkieletu

Zespół Forney'a

Zespół Forney'a, Robinsona i Pascoe

Forney-Robinson-Pascoe syndrome

Mitral regurgitation-deafness-skeletal anomalies
syndrome

Mitral regurgitation-hearing loss-skeletal
anomalies syndrome

Kod ORPHA

3238

Kod OMIM

157800

Kod ICD10

Q87.8

Kod ICD11

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*Źródło

orphanet