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