Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome without intellectual disability characterized by unilateral or bilateral cleft lip and palate and craniofacial dysmorphism (including frontal bossing, hypertelorism, broad flat nasal bridge, cupped ears/thickened helices, and micrognathia). Additional manifestations are variable congenital cardiac anomalies, pectus excavatum, abnormalities of the hands and feet, ocular abnormalities (myopia, cataract, staphyloma), and conductive or sensorineural hearing loss.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Cleft lip and palate-craniofacial dysmorphism-congenital heart defect-deafness syndrome
Niedobór hialuronidazy 2
Hyaluronidase 2 deficiency

Kod ORPHA
508476

Kod OMIM
-

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet