

## 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

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#### Kod ICD10

Q87.8

#### Kod ICD11

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#### \*Źródło

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