Opis choroby *

Definicja

A rare, severe, multiple congenital anomalies syndrome characterized by severe mandibular hypoplasia, upper limb phocomelia with olygodactyly, absent fibula, and a number of additional skeletal (hypoplastic scapula and ischii, 11 ribs, clubfeet), facial (hypertelorism, hypoplastic supraorbital ridges, wide nasal bridge, microtia with low-set ears) and variable internal organ abnormalities (including arhinencephaly, hypolobulated lungs, and congenital cardiac defects), which usually lead to perinatal death. Surviving patients show features similar to Nagel syndrome.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
1788

Kod OMIM
201170

Kod ICD10
Q75.4

Kod ICD11
LD25.2

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*Źródło

orphanet