

Opis choroby *

Definicja

A rare, severe, multiple congenital anomalies syndrome characterized by severe mandibular hypoplasia, upper limb phocomelia with oligodactyly, 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, hyplobulated 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

*Źródło

orphanet