

Polisyndaktylia - wada serca

Kod Orpha: 2934 Kod OMIM: 263630

Opis choroby *

Definicja

A rare, life-threatening developmental defect during embryogenesis characterized by polysyndactyly of fingers and toes as well as complex congenital heart defects (e.g. atrioventricular septal defects, aortic dextroposition, single ventricle, hypo- or hypertrophy of one side of the heart). Additional features may include dysmorphic traits (large fontanel, high forehead, ptosis, hypertelorism, epicanthus, low-set malformed ears, prominent root of the nose, bulbous nose, anteverted nares, long and smooth philtrum, thin upper lip, micrognathism, hirsutism, single transverse crease) nail hypoplasia, phalange agenesis/hypoplasia, flexion contractures, polysplenia, multiple hepatic/renal cysts, atrophic biliary vesicle, ductal plate malformation and genital anomalies (e.g. micropenis, undescended testes, hypoplastic scrotum). The syndrome is usually fatal in utero or in infancy, but survival cases have been reported.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Bonneau syndrome
Zespół Bonneau

Kod ORPHA

2934

Kod OMIM

263630

Kod ICD10

Q87.8

Kod ICD11

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[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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