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