## Opis choroby *

Definicja

A rare, multiple congenital anomalies syndrome with intellectual disability commonly characterized by facial dysmorphism (e.g. sagittal craniosynostosis, hypertelorism, strabismus, low-set dysplastic ears, retrognathia or micrognathia, mandibular ankyloses, cleft palate, aplasia uvulae), congenital heart defects (e.g. atrioventricular septal defect, anomalous venous return), genital anomalies (e.g. cryptorchidism, microphallus), as well as growth delay and intellectual disability. In some cases, tracheobronchial anomalies, large joint contractures, syndactyly, rib anomalies and hypoplastic kidneys are reported. Rarely, no cardiac anomaly may be reported.

Dane

**Klasyfikacja**
Zespół wad wrodzonych

Synonimy
Craniosynostosis-congenital heart disease-intellectual disability syndrome
Kraniostenoza strzrałkowa z wrodzoną wadą serca, upośledzenie umysłowe i ankyloza żuchwy
Kraniosynostoza - wrodzona choroba serca - niepełnosprawność intelektualna
Zespół Pfeiffera, Singera i Zschieschego
Pfeiffer-Singer-Zschiesche syndrome

**Kod ORPHA**
2872

**Kod OMIM**
218450

**Kod ICD10**
Q87.8

**Kod ICD11**
LD2F.1Y

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*Źródło

orphanet