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