Opis choroby *

Definicja

A rare multiple congenital anomalies syndrome characterized by a spectrum of developmental anomalies including cleft lip and/or palate, craniosynostosis, intellectual disability and/or learning disability, radioulnar synostosis, genital and vesicorenal anomalies. Observed facial dysmorphism includes hypertelorism, blepharophimosis, blepharoptosis, high arched eyebrows. Less common features reported include anterior chamber defects, cardiac anomalies (e.g. ventricular septal defect; see this term), caudal appendage, umbilical hernia/omphalocele and diastasis recti.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Craniofacial-ulnar-renal syndrome
Zespół czaszkowo-twarzowo-łokciowo-nerkowy
Zespół Malpuecha, Michelsa, Mingarelli i Carnevale'a
Malpuech-Michels-Mingarelli-Carnevale syndrome

Kod ORPHA
293843

Kod OMIM
265050

Kod ICD10
Q87.8

Kod ICD11
LD2F.1Y

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*Źródło

orphanet