Opis choroby *

Definicja

A rare, genetic, multiple congenital malformation syndrome, characterized by cleidocranial dysplasia (wide fontanelles, calvaria dysostosis, absent or hypoplastic clavicles), absent thumbs and halluces, hypoplastic distal and medial phalanges of fingers, pelvic dysplasia with hip dislocations. Dysmorphic features include sparse scalp hair, protruding eyes, low-set ears, anteverted nares, midfacial hypoplasia, tented upper lip, high arched palate, and micrognathia. Brain malformations are frequently associated. From birth, affected individuals tend to be significantly hypotonic and present with global developmental delay, and respiratory, feeding and swallowing difficulties.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Cleidocranial dysplasia-micrognathia-absent thumbs syndrome
Dysplazja obojczykowo-czaszkowa - małożuchwie - brak kciuków

Kod ORPHA
3472

Kod OMIM
216340

Kod ICD10
Q87.8

Kod ICD11
LD24.23

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*Źródło

orphanet