Opis choroby *

Definicja

A rare syndromic trigonocephaly characterized by marked malformations of the head and face (essentially acrocephaly), broad depressed nasal bridge, narrow maxillae, abnormalities of the hands and feet (polydactyly, brachydactyly, syndactyly, clinodactyly, camptodactyly, ulnar deviation), obesity and congenital heart disease. This disease is considered a variant of Carpenter syndrome without intellectual disability. There have been no further descriptions in the literature since 1992.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
ACPS4
ACPS4
Akrocefalopolisyndaktylia typu 4
Acrocephalopolysyndactyly type 4

Kod ORPHA
65798

Kod OMIM
201020

Kod ICD10
Q87.0

Kod ICD11
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*Źródło

orphanet