

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	Synonimy
Zespół wad wrodzonych ACPS4	ACPS4
	Akrocefalopolisyndaktylia typu 4
	Acrocephalopolysyndactyly type 4

Kod ORPHA	Kod OMIM	Kod ICD10
65798	201020	Q87.0

Kod ICD11

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*Źródło

orphanet