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 disabaility. There have been no further descriptions in the literature since 1992.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
3210

Kod OMIM
272350

Kod ICD10
Q82.0

Kod ICD11
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*Źródło

orphanet