Opis choroby *

Definicja

A very rare acrofacialdyosotosis characterized by short stature, acrocephaly, ocular hypertelorism, ptosis of eyelids, ocular proptosis, downslanting palpebral fissures, high nasal bridge, anteverted nostrils, short philtrum, cleft palate, micrognathia, abnormal external ears, preauricular pits, mixed hearing loss, bulbous digits, metatarsus varus, pectus excavatum and various radiological abnormalities. Features of this syndrome were reported to overlap with otopalatodigital syndrome types 1 and 2. There have been no further descriptions in the literature since 1988.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Kaplan-Plauchu-Fitch syndrome
Zespół Kaplana, plauchu i Fitcha

Kod ORPHA
949

Kod OMIM
201050

Kod ICD10
Q87.0

Kod ICD11
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*Źródło

orphanet