Opis choroby *

Definicja

A very rare acrofacial dysostosis characterized by normal intelligence, shortness of stature, and mild acrofacial dysostosis (malar hypoplasia, micrognathia and webbing of digits with shortening of the fourth metacarpals) associated with oligodontia, normal or high arched palate, aplasia cutis verticis with pili torti, mild cutaneous syndactyly of digits 2-5, webbing of digits and shortening of the fourth metacarpals, and unilateral cleft lip. Features are similar to those seen in Zlotogora-Ogur syndrome, although the latter shows no sign of acrofacial dysostosis. There have been no further reports in the literature since 1997.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
1787

Kod OMIM
601829

Kod ICD10
Q75.4

Kod ICD11
LD25.2

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*Źródło

orphanet