Opis choroby *

Definicja

A rare congenital acrofacial dysostosis characterized by mild intrauterine growth retardation, postnatal short stature, microcephaly, intellectual disability, moderate mandibulofacial dysostosis (including dental anomalies and/or malpositioning, microretrognathia, and malar hypoplasia), and mild pre- and postaxial limb hypoplasia with generalized brachydactyly, mild interdigital webbing, single transverse palmar creases and clinodactyly. Reported facial features include high forehead, widow's peak, downslanted palpebral fissures, sparse lateral eyebrows, and small or dysplastic ears. Variably associated features include frequent caries, preauricular fistulae, inguinal hernia, spina bifida occulta, and cryptorchidism and hypospadias in males.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Opitz-Caltabiano syndrome
Zespół Opitza i Caltabiano

Kod ORPHA
1786

Kod OMIM
101805

Kod ICD10
Q75.4

Kod ICD11
LD25.2

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*Źródło

orphanet