

## 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 Opitz-Caltabiano syndrome  
Zespół Opitza i Caltabiano

#### Kod ORPHA

1786

#### Kod OMIM

101805

#### Kod ICD10

Q75.4

#### Kod ICD11

LD25.2

---

#### \*Źródło

orphanet