Opis choroby *

Definicja

A rare ciliopathy characterized by oral anomalies (multiple oral frenula, missing incisors), facial dysmorphism (such as square face with small forehead, upslanting palpebral fissures, and cleft lip, among other features), digital anomalies (brachydactyly, brachymesophalangy, polydactyly), and short stature. Additional reported manifestations include short femoral neck, bilateral cervical ribs, abnormal vertebral bodies, and gracile long bones.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
OFD18
OFD18
Oralfacialdigital syndrome typu 18
Zespół ustno-twarzowo-palcowy typu 18
Oral-facial-digital syndrome type 18
Orofaciodigital syndrome type 18

Kod ORPHA
508501

Kod OMIM
617927

Kod ICD10
Q87.0

Kod ICD11
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*Źródło

orphanet