

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

Kod ORPHA	Kod OMIM	Kod ICD10
508501	617927	Q87.0

Kod ICD11

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*Źródło

orphanet