

## Opis choroby \*

### Definicja

Orofaciodigital syndrome type 12 is a rare subtype of orofacioidigital syndrome, with sporadic occurrence, characterized by cardiac (septum hypertrophy) and central nervous system abnormalities (myelomeningocele, Sylvius aqueduct stenosis, corpus callosum agenesis, vermis hypoplasia), in addition to oral, facial and digital malformations (gingival frenulae, bifid tongue, supernumerary teeth, macrocephaly, hypertelorism, pre- and post-axial polydactyly in hands, preaxial polydactyly in feet and club feet). Skeletal anomalies, such as short tibiae and central, Y-shaped metacarpals, are also associated.

### Dane

<b>Klasyfikacja</b>	<b>Synonimy</b>
Zespół wad wrodzonych	Moran-Barroso syndrome
	OFD12
	OFD12
	Oral-facial-digital syndrome type 12

<b>Kod ORPHA</b>	<b>Kod OMIM</b>	<b>Kod ICD10</b>
141327	-	Q87.0

**Kod ICD11**  
LD25.00

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### \*Źródło

orphanet