Opis choroby *

Definicja

Orofaciodigital syndrome type 12 is a rare subtype of orofaciodigital 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

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Moran-Barroso syndrome
OFD12
OFD12
Oral-facial-digital syndrome type 12

Kod ORPHA
141327

Kod OMIM
-

Kod ICD10
Q87.0

Kod ICD11
LD25.00

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*Źródło

orphanet