Opis choroby *

Definicja

Orofaciodigital syndrome type 14 is a rare subtype of orofaciodigital syndrome, with autosomal recessive inheritance and C2CD3 mutations, characterized by severe microcephaly, trigonocephaly, severe intellectual disability and micropenis, in addition to oral, facial and digital malformations (gingival frenulae, lingual hamartomas, cleft/lobulated tongue, cleft palate, telecanthus, up-slanting palpebral fissures, microretrognathia, postaxial polydactyly of hands and duplication of hallux). Corpus callosum agenesis and vermis hypoplasia with molar tooth sign, on brain imaging, are also associated.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Microcephaly-cerebral malformation-orofaciodigital syndrome
OFD14
Zespół ustno-twarzowo-palcowy z małogłowiem i malformacjami mózgu
OFD14
Oral-facial-digital syndrome type 14

Kod ORPHA
434179

Kod OMIM
615948

Kod ICD10
Q87.0

Kod ICD11
LD25.00

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*Źródło

orphanet