

Opis choroby *

Definicja

Orofaciodigital syndrome type 14 is a rare subtype of orofacioidigital 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 Microcephaly-cerebral malformation-orofacioidigital syndrome

Synonimy

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

*Źródło

orphanet