DISEASE:
Orofaciodigital syndrome type 14

NAME:	Orofaciodigital syndrome type 14
DESCRIPTION:	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.
ORPHACODE:	434179
SYNONYMS:	Microcephaly-cerebral malformation-orofaciodigital syndrome OFD14 Oral-facial-digital syndrome type 14
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	C2CD3
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

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