Title: Oral-Facial-Digital Syndrome Type 1 *GeneReview* – Table 2
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Note: The following information is provided by the authors and has not been reviewed by *GeneReviews* staff.

Table 2. Clinical Features and Causal Genes in OFD Syndromes

	Oral Features	**Facial Features**	**Hand Anomalies**	**Foot Anomalies**	**Skin/Hair Features**	**Renal Features**	**Cardiac Features**	**Cerebral Features**	**Skeletal Features**	**Other Abnormalities**	**Inheritance**	**Gene**	**Main References**
OFD I	Gingival frenulae Lingual hamartomas Cleft/lobulated tongue Cleft palate	Hypertelorism Cleft lip **Pseudocleft of the upper lip**	**Brachydactyly** Clinodactyly Polydactyly	Preaxial polydactyly	**Milia** **Alopecia**	**Polycystic kidney disease**	-	**Corpus callosum agenesis** **Cerebellar hypoplasia**	-	Moderate intellectual disability (50%)	X-linked dominant (lethal in males)	*OFD1*	Thauvin-Robinet at al [2006] Prattichizzo et al [2008]
OFD II	Gingival frenulae Lingual hamartomas Cleft/lobulated tongue Cleft palate	-	Brachydactyly Clinodactyly **Polydactyly**	**Broad hallux** Pre/postaxial polydactyly	Thick hair	-	Rare	Porencephaly Hydrocephaly	Median Y-shaped metacarpal	-	Autosomal recessive	-	Toriello et al [1993] Prpic et al [1995]
OFD III	Bifid uvula Lingual hamartomas Lobulated tongue Tooth hypoplasia	Hypertelorism Bulbous nose Low-set ears	Postaxial polydactyly	Postaxial polydactyly	-	**End-stage renal failure at 13 and 24 years old**	-	**Cerebellar vermis hypoplasia** **DW malformation** with cystic dilation of the fourth ventricle Myoclonia / **eye movement**	-	Pectus excavatum Severe intellectual disability	Autosomal recessive	*TMEM231*	Sugarman et al [1971] Smith & Gardner-Medwin [1993] Roberson et al [2015]
OFD IV	Gingival frenulae Lingual hamartomas Lobulated tongue Cleft palate	Epicanthus **Micrognathia** Low-set ears	Brachydactyly Clinodactyly Pre/postaxial polydactyly	Pre/postaxial polydactyly	-	Renal cysts	-	Porencephaly Occipital encephalocele Agenesis of corpus callosum Cerebellar vermis hypoplasia with MTS	Pectus excavatum **Tibial abnormalities**	Short stature Variable intellectual disability	Autosomal recessive	*TCNT3*	Toriello et al [1997] Thomas et al [2012]
OFD V	Gingival frenulae (rare)	**Midline cleft lip**	Postaxial polydactyly	Postaxial polydactyly	-	-	-	-	-	**India origin**	Autosomal recessive	-	Valiathan et al [2006]
OFD VI	Gingival frenulae Lingual hamartomas Lobulated tongue Cleft palate	Hypertelorism Cleft lip	Brachydactyly Clinodactyly Syndactyly **Median/ postaxial polydactyly**	**Broad hallux** **Preaxial polydactyly**	-	Renal agenesis Renal dysplasia	Rare	**Cerebellar vermis hypoplasia with MTS**	**Median Y-shaped metacarpal**	Variable intellectual disability Possible retinopathy	Autosomal recessive	*OFD1* *TMEM216* *C5orf42* *TMEM138* *TMEM107* KIAAO753	Poretti et al [2012] Darmency-Stamboul et al [2013] Lopez et al [2014] Li et al [2016] Lambacher et al [2016] Chevrier et al

	Oral Features	Facial Features	Hand Anomalies	Foot Anomalies	Skin/Hair Features	Renal Features	Cardiac Features	Cerebral Features	Skeletal Features	Other Abnormalities	Inheritance	Gene	Main References
													[2016] Author, personal data
OFD VII	Gingival frenulae Lingual hamartomas Cleft palate	Hypertelorism **Cleft lip** Asymmetry	Clinodactyly	-	-	**Polycystic kidney disease**	-	-	-	Moderate intellectual disability	X-linked dominant	*OFD1 ?*	Whelan et al [1975] Nowaczyk et al [2003]
OFD VIII	Gingival frenulae Lingual hamartomas Lobulated tongue **Epiglottis hypoplasia**	**Midline cleft lip** Telecanthus Large nose	**Bifid thumb** Postaxial polydactyly	Preaxial polydactyly	-	-	-	-	**Tibia and radius hypoplasia**	Psychomotor delay **Precocious lethality**	X-linked recessive	-	Edwards et al [1988]
OFD IX	Gingival frenulae Lingual hamartomas Lobulated tongue Cleft palate	Midline cleft lip Synophrys	Brachydactyly Clinodactyly Polydactyly	Bifid toes	-	-	SD	-	-	Short stature **Microphthalmia** **Coloboma**	Autosomal recessive	*SCLT1* *TBC1D32/ C6orf170*	Gurrieri et al [1992] Erickson & Bodensteiner [2007]
OFD X	Gingival frenulae Cleft palate	Telecanthus Flat nasal root Retrognathia	**Oligodactyly** Preaxial polydactyly	-	-	-	-	-	**Shortened 4 limbs** **Bilateral short radii** **Fibular agenesis**	-	Sporadic	-	Figuera et al [1993]
OFD XI	Gingival frenulae Cleft palate	Hypertelorism Auricular pits Blepharo-phimosis	Postaxial polydactyly	Postaxial polydactyly	-	-	-	**Ventricular dilatation**	**Odontoid hypoplasia** **Vertebral abnormalities**	**Deafness** **Severe intellectual disability** **Behavioral abnormalities**	Sporadic	-	Gabrielli et al [1994] Obregon & Barriero [2003]
OFD XII	Gingival frenulae Bifid tongue Supernumerary teeth	Macrocephaly Hypertelorism	Pre/postaxial polydactyly	Preaxial polydactyly Club feet	-	-	Septum hypertrophy	**Aqueductal stenosis** **Corpus callosum agenesis** **Cerebellar vermis hypoplasia** **Myelomeningocele**	**Short tibiae** **Central Y-shaped metacarpal**	-	Sporadic	-	Moran-Barroso et al [1998]
OFD XIII	Lingual hamartomas	Cleft lip	Brachydactyly Clinodactyly Syndactyly	Brachydactyly Clinodactyly Syndactyly	-	-	Mitral and tricuspid valves dysplasia	**Leukoaraiosis**	-	**Neuropsychiatric disease** **Epilepsy**	Sporadic	-	Degner et al [1999]
OFD XIV	Gingival frenulae Lingual hamartomas Cleft/lobulated tongue Cleft palate	Telecanthus	**Postaxial polydactyly**	**Duplication of hallux**	-	-	-	Corpus callosum agenesis **Cerebellar vermis hypoplasia with MTS**	-	**Severe microcephaly** Micropenis	Autosomal recessive	*C2CD3*	Thauvin-Robinet al [2014]

	Oral Features	Facial Features	Hand Anomalies	Foot Anomalies	Skin/Hair Features	Renal Features	Cardiac Features	Cerebral Features	Skeletal Features	Other Abnormalities	Inheritance	Gene	Main References
Unclassified OFD	Lobulated tongue Cleft palate	Median cleft lip	Postaxial polydactyly	**NA**	Thick hair	Fused kidneys	**TOF** **VSD**	Corpus callosum agenesis		Possible moderate intellectual disability Hirschsprung disease	Autosomal recessive	*DDX59*	Shamseldin et al [2013]
Unclassified OFD	Lingual hamartomas	Frontal bossing Hypertelorism Micro-retrognathia	Postaxial polydactyly	Duplication of hallux Syndactyly	-	-	**Coarctation of the aorta**	-	5th Y-shaped metacarpal	+/- intellectual disability	Autosomal recessive	*WDPCP*	Saari et al [2015] Toriyama et al [2016] Author, personal data
Unclassified OFD	Lingual hamartoma	Frontal bossing Cleft lip Low-set ears Micro-retrognathia	Preaxial polydactyly	Polydactyly	-	-	**TOF**	NA	**Y-shaped metatarsal**	Intellectual disability, micropenis	Autosomal recessive	*INTU*	Toriyama et al [2016]
Unclassified OFD	Cleft palate	Cleft lip	Postaxial polydactyly	Postaxial polydactyly	-	-	**-**	Cerebellar vermis hypoplasia, DW malformation	12th rib hypoplasia	-	Autosomal recessive	*TMEM231*	Li et al [2016]
Unclassified OFD	Gingival frenulae	Cleft lip	Brachydactyly Preaxial polydactyly	Postaxial polydactyly	-	-	**-**	-	Short stature Short mesoaxial phalanges	-	Autosomal recessive	*IFT57*	Thevenon et al [2016]

DW = Dandy-Walker; MTS = molar tooth sign; SD = septal defects; TOF = tetralogy of Fallot; VSD = ventricular septal defect

Characteristic features for each subtype are in **bold**.

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