Oral Findings of an Infant with Oro-facial-digital Syndrome Type I

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Introduction

Oro-facial-digital syndrome is a group of developmental disorders that represent distinctive genetic anomalies of the oral cavity, face, and digits. There are 14 reported types, with varying inheritance and phenotypes. The most common reported is OFDS type I, which is X-linked dominant and therefore female predominant.

Case Report

This presentation discusses a female infant (34wGA) who presented at the NICU of Children's Wisconsin at four days old with a lobulated tongue and presumed natal teeth in May of 2022. This patient has been followed by the dental team in her first year of development, with persistent lobulated tongue, multiple strong frenula, gingival notching, palatal fissure, and bifid uvula upon clinical exam. She has also been followed by genetics and was diagnosed with OFDS-I, by neurology due to agenesis of corpus callosum, by speech and feeding therapies due to concern of oral dysphagia, by ophthalmology for mild estropia, and by plastic surgery for future surgical needs for tongue and presumed submucosal cleft palate. Physical and occupational therapies have been involved as part of birth to three early intervention.

As she is eating and gaining weight appropriately, no treatment or surgical intervention is indicated at this time. It is important to maintain contact with the medical team in cases such as this about changes in history and upcoming surgeries.

Resources

Franco B, Thauvin-Robinet C. Update on oral-facial-digital syndromes (OFDS). Cilia. 2016 May 2;5:12. doi: 10.1186/s13630-016-0034-4. PMID: 27141300; PMCID: PMC4852435.

Syed S, Sawant PR, Spadigam A, Dhupar A. Oro-facial-digital syndrome type I: a case report with novel features. Autops Case Rep. 2021 Aug 20;11:e2021315. doi: 10.4322/acr.2021.315. PMID: 34458183; PMCID: PMC8387074.

Verma PK, Bhat NK. Oro-facial-digital syndrome: Unspecified type with the spontaneous fusion of cleft palate. Contemp Clin Dent 2021;12:454-8.

Possible Findings of OFDS-1

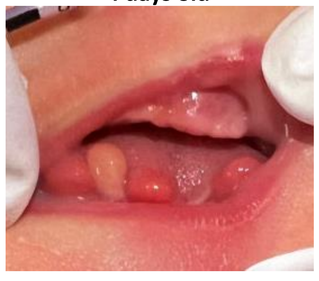
Oral
- **Lobulated tongue**
- **Lingual hamartomas**
- **Ankyloglossia**
- **Cleft palate**
- Median Cleft Lip
- **Gingival frenulae and notching**
- Hyperdontia
- **Hypodontia**
- **Enamel dysplasia**
- Hypoplastic mandible

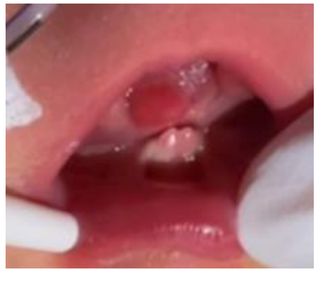
Facial
- Hypertelorism
- Hypoplasia alae of nose
- Hearing defects
- Low set ears
- **Milia**
- Thin hair, alopecia

Digital & Systemic
- Brachydactyly
- Clinodactyly
- Polydactyly
- Syndactyly
- Mental retardation
- **Cerebral and cerebellar abnormalities**
- Kidney defects, polycystic kidney disease
- Liver/pancreas/ovary cysts

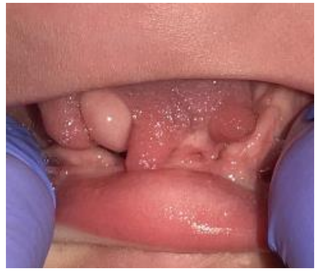
Bolded items denote findings for this patient

4 days old

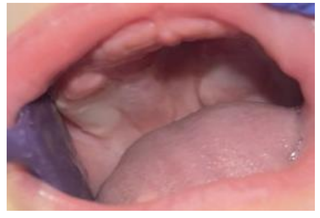


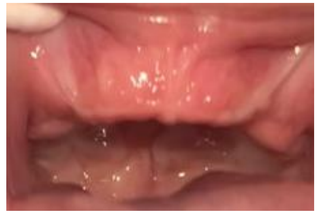


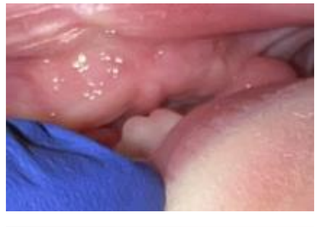
6 months old

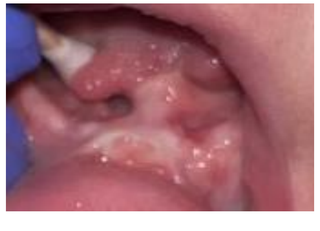


9 months old

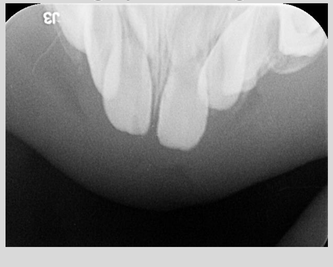


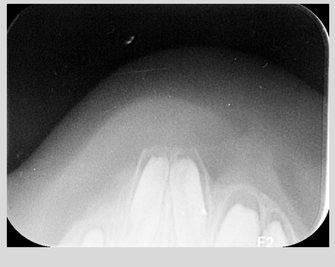






Radiographic findings 9m





Future Treatment Needs

Dental
- Routine examinations, photos and radiographs to monitor changes
- Discuss possible findings and anticipatory guidance with parents
  - Radiographic enamel defect with unerupted #E
  - Palatal folds, possible submucosal cleft palate
  - Hyper/hypodontia
  - High attached frenulae and brushing habits
- May have future orthodontic needs

Plastic Surgery
- Tongue revision surgery
- Lingual or labial frenectomies
- Monitor possible submucosal cleft palate

Continue care with
- Speech Pathology
- Early Intervention
- Neurology
- Audiology
- Ophthalmology
- Dermatology

Follow up for Patients with OFDS-1

It is recommended that patients with OFD-1 are seen at typical recall intervals, based on individual assessment. If the patient presents with oral clinical disturbances, more frequent radiographs (PAs, pano, ceph) may be necessary for comparison as well as consultation with physicians, orthodontists, and surgeons regarding treatment. Early orthodontic intervention may be recommended to treat malocclusions, possible clefting, and changes in arch development.

This multidisciplinary approach may include dentistry, orthodontics, craniofacial plastic surgery, ophthalmology, neurology, speech pathology, dermatology, and ENT, genetics, and more. Follow up and communication with the entire medical team will be important to monitor any changes in health history, as systemic abnormalities (such as kidney disease) may become more prominent later in life.

Conclusion

This patient shows many oral manifestations of Oro-facial-digital Syndrome Type 1. This case report includes clinical and radiographic findings associated with OFDS-I, the importance of interprofessional collaboration, and possible future treatment needs and considerations for this patient and those with OFDS-1.

Comprehensive oral exam of infants with OFDS is crucial to identify structural abnormalities and monitor for future changes in the oral cavity.