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Case Report

Oro-facial-digital syndrome: A report of two cases

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ABSTRACT

The Oro-facial-digital syndrome (OFDS) is a rare developmental anomaly characterized by congenital malformations involving the face, oral cavity, hands and feet. Other organ systems can be involved, defining specific types of OFDS. A recessive mode of inheritance is proposed. To date 13 types have been distinguished based on characteristic clinical manifestations. OFDS has an estimated incidence of 1 in 50,000–250,000 newborns. Two cases of OFDS (15-year-old female and 52-year-old male), who reported to the outpatient clinic of Oral Medicine Department, are presented here. The first case reported with mal-alignment of teeth and the second one with the complaint of bleeding from the palate since 2 weeks. The overlapping features exhibited by different types of OFDS (as seen in these 2 cases) make the diagnosis extremely difficult.

1. Introduction

Oro-facial-digital syndromes (OFDS) are a group of closely related disorders, of which, at least 13 different subtypes have been described [1–3]. Papillon-League–Psaume syndrome, Mohr syndrome, Mohr-Majewski syndrome, Thurston syndrome, Varadi-Papp syndrome and Gabrielli syndrome are different names for different types of OFDS [4–8]. The oro-facial-digital syndrome, also known as oro-digito-facial-dysostosis and dysplasia-linguo-facialis, is a very rare developmental anomaly characterized by congenital malformations involving the face, oral cavity, hands and feet. A recessive mode of inheritance is proposed. Two cases, who reported to the outpatient clinic of Oral Medicine Department, are presented here.

2. Case reports

2.1. Case 1

A 15-year-old girl reported to the department for the correction of mal-aligned teeth.

On general examination, she appeared to be of short stature, moderately nourished and without any systemic disease. Extra orally, the face appeared apparently symmetrical. Depressed nasal bridge, slight flaring of alar regions, mild pitting of lower lip, mild brachycephaly and dryness of facial skin were noted (Fig. 1).

Intra-oral examination revealed a normal mouth opening. All teeth, which were present appeared to have normal morphology, with the exception of a cone shaped maxillary left lateral incisor. Maxillary left canine and lower lateral incisors and canines were found missing and there was no previous history of extraction. Oral hygiene status appeared moderate with no detectable caries lesion. Multiple, thick, abnormal frenula appeared eliminating the labial and buccal sulci in the upper arch. This on extension produced clefts of the alveolar ridge in the upper lateral incisor–canine region bilaterally. Thick fibrous bands in the lower mucobuccal fold appeared eliminating sulcus and clefting the ridge as in the upper arch (Figs. 2 and 3). The tip of the tongue appeared bifid (Fig. 4) and there is infra occlusion.

2.2. Case 2

The second case was a 52-year-old male patient, who reported with complaint of bleeding from the palate since 2 weeks.

On general examination the following features were noted:

(1) Moderately nourished patient with short stature.
(2) Abnormal gait with hypermobility of knee joints bilaterally and polydactyly of upper extremities (Fig. 6).

☆ Asian AOMS: Asian Association of Oral and Maxillofacial Surgeons; ASOMP: Asian Society of Oral and Maxillofacial Pathology; JSOP: Japanese Society of Oral Pathology; JSOMS: Japanese Society of Oral and Maxillofacial Surgeons; JSOM: Japanese Society of Oral Medicine; JAMI: Japanese Academy of Maxillofacial Implants.

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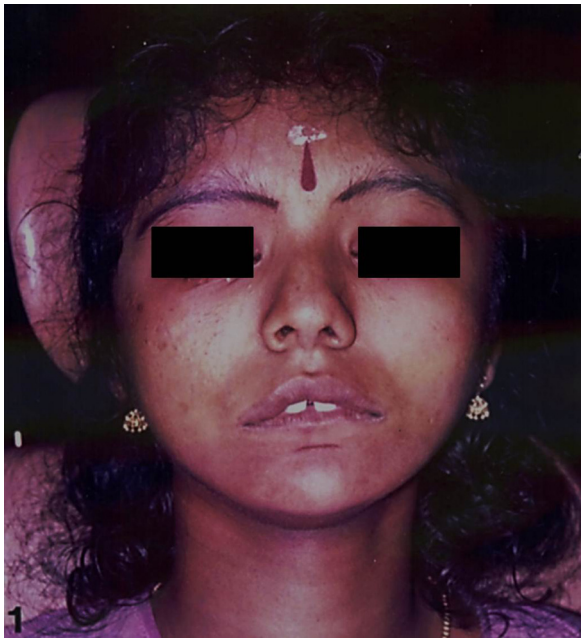


Fig. 1. Facial photograph showing depressed nasal-bridge, slight flaring of alar regions and mild brachycephaly (Case 1).

(3) Apparently symmetrical face with broad and fan-shaped ears, dry-facial skin and patchy alopecia of the scalp.
(4) Depressed nasal bridge, mild flaring of alar regions and mild brachycephaly (Fig. 5).

Intra-orally, his oral hygiene status appeared poor, with no detectable caries lesion.

A normal mouth opening revealed normal morphology for all the present teeth. All the third molars, right upper and left lower first and second molars and left upper first premolars were missing. The patient had a history of extraction of these teeth following caries involvement. Except for increased numbers of frenae, a prominent lingual frenum, and narrow high arched palate with mild grooving in the midline, there were no other features suggestive of this syndrome (Fig. 7). The focus of bleeding appeared to be from a tiny red spot in the posterior aspect of the hard palate, which is clinically suggestive of granulation tissue at the orifice of a minor salivary gland. Multiple prominent minor salivary glands were observed on the palate suggestive of nicotina palatini.

Radiographic evaluation revealed a well-circumscribed area of radiolucency of size 4 cm × 5 cm on the left side of the palate enclosing a well-formed tooth-simulating upper canine (Fig. 8). The maxillary left central incisor which is contacting the lesion showed considerable root resorption. Another well-circumscribed unilocular area of radiolucency was also noted in the periapical region of the right maxillary central and lateral incisors.

Radiographic findings of the first patient were non-contributory except for the missing maxillary left canine and lower lateral incisors.

3. Discussion

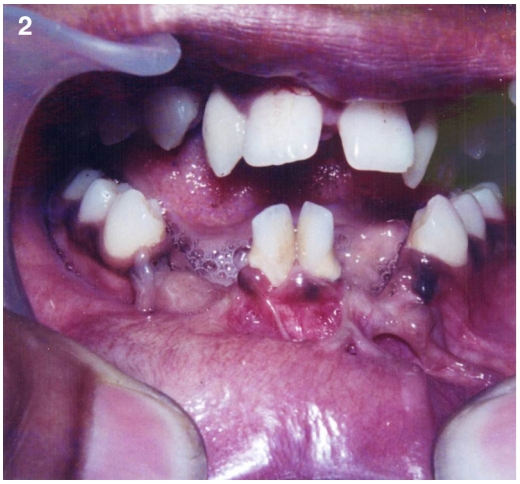
The oro-facial-digital-syndrome, also known as oro-digito-facial-dysostosis and dysplasia-linguo-facialis, is a very rare hereditary developmental anomaly.

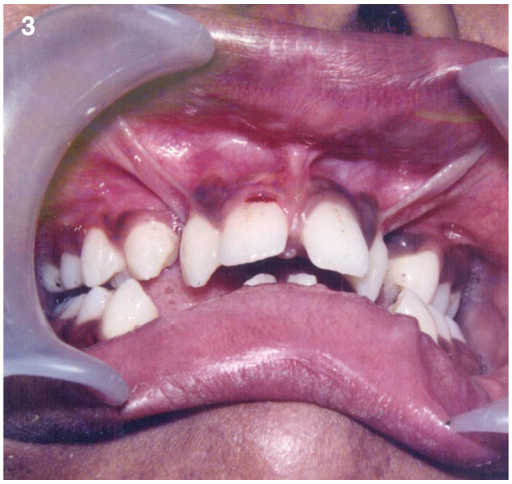
The first description of an oro-facial-digital-syndrome (OFDS) was done by Moher (1941) in a family with oral, facial and digital (OFD) findings, including highly arched palate, lobate tongue with papilliform outgrowths, a broad nasal root, and hypertelorism. Additional findings included syndactyly, brachydactyly, and polydactyly of the hands and feet [1].

A similar OFD phenotype was described by Papillon-Leage and Psaume (1954) in their series of 11 cases, consisting of abnormally developed frenula that, by extension, cleft the jaws and tongue. The clefts were located in lateral-incisor canine region. Malposition of maxillary canine, frequent infra-occlusion, and absence of mandibular laterals were common [9]. Gorlin et al. [11] in 1961 reported a family of four patients having this syndrome in three generations and Schwarz and Fish [12] described its radiographic changes.

For some unknown reason, all subsequent literature reports referred to the X-linked dominant form of Papillon-Leage and Psaume as OFDS type I, even it was described after the recessive form of Moher and Claussen, now referred to as OFDS type II [1].

After the description of OFDS types I and II, the phenotypic spectrum was further expanded with extra-OFD manifestations, with new types, each being characterized either by distinctive clinical findings and/or by a specific mode of inheritance [13]. Subsequently, in a review by Toriello [14], nine different types were listed.





Figs. 2 and 3. Intra-oral photograph showing clefts associated with hyperplasia of frenum-producing cleft in the lateral incisor-canine region bilaterally. Numerous thick fibrous bands appear in mucobuccal fold eliminating sulcus. Missing maxillary left canine, cone shaped upper left lateral incisor (Case 1).