Oral-Facial-Digital Syndrome Type 1: Oral Findings in a 6-Year-Old Girl

Zuhal Kirzioglu¹ Esra Oz¹

¹Department of Paediatric Dentistry, The University of Süleyman Demirel, Isparta, Turkey

Address for correspondence Esra Oz, PhD, Department of Paediatric Dentistry, The University of Süleyman Demirel, Isparta, Turkey (e-mail: esrakaraagac@sdu.edu.tr).

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Abstract

Keywords

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Oral-facial-digital syndrome (OFDS) is a group of congenital anomalies with 13 different forms. OFDS type 1 (OFDS1) is a developmental genetic anomaly related to the X chromosome, that is often seen in girls, and affects the face, oral cavity, and extremities. In this study, we discuss the oral findings of a 6-year-old girl with OFDS1 and her situation after 2.5 years.

Introduction

Oral-facial-digital syndrome type 1 (OFDS1) is a developmental genetic disease that was first reported by Papillon–League and Psaume in 1954 and described by Gorlin and Psaume 8 years later. OFDS1 is characterized by malformation of the oral cavity, face, and fingers and an X-dominant inheritance in girls.¹ It has an incidence of 1/50,000 to 1/250,000 live births.²

Facial deformities can include frontal bossing, broad nasal root, hypoplasia of the mandibular ramus, alar cartilage aplasia, pseudocleft of the upper lip, hypertelorism, and strabismus. Digital abnormalities affect mainly the hands (50–70%) and less so the feet (25%) and include brachydactyly, syndactyly, clinodactyly, and, rarely, polydactyly.^{2,3} Brain anomalies, developmental deficits, and mental disability due to central nervous system involvement have been reported in patients with OFDS1.⁴ Polycystic kidney disease (PKD) distinguishes OFDS1 from the other forms of OFDS and is seen in approximately 50% of patients.²

The oral/maxillofacial disorders include micrognathia, high-arched cleft palate, thickened alveolar ridges, ankyloglossia, lobulated tongue with hamartomas between the lobes, thick hyperplastic frenula in the upper and lower mucobuccal areas, cleft near canine separated by the frenulum, tooth malformation, congenitally missing and supernumerary teeth, and enamel hypoplasia.^{2,3,5}

Case Report

A 6-year-old girl was brought to our clinic for an evaluation of dental problems by her parents. The patient was the daughter of a 20-year-old mother born at the end of a marriage to a relative. The patient's medical history revealed the diagnosis of OFDS1.

Corpus callosum agenesis, a cerebral cystic structure, and hydrocephaly were observed on a cranial magnetic resonance image. She manifested severe, early developmental delay. PKD was also diagnosed in both the patient and the mother. The patient had a hernia operation at 1 month, a cleft palate operation at 9 months, and tongue nodule operation. The mother reported that the patient had received special education.

The physical examination revealed brachydactyly, micrognathia, a thin upper lip, a broad nasal root, and strabismus (►Fig. 1). The maxillary segment formed a pseudo-cleft when the patient smiled.

An intraoral examination revealed that the patient had a deep V-shaped palate distinctly separate from the anterior segment, ankyloglossia, irregular morphology of the primary teeth, and tooth rotations (►Fig. 2). A thin upper lip, lobulated tongue, numerous hyperplastic frenula and a cleft separated by the frenulum in the mandible, congenitally missing teeth, tooth caries, developmental enamel defects were seen in the patient and the patient's mother (►Fig. 3).

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Fig. 1 Facial appearance of the patient and her fingers.

Fig. 2 The oral findings of patient. (a) Lobulated tongue. (b) Cleft and hyperplastic oral frenulas. (c) Deep V-shaped palate. (d) Ankyloglossia and tooth rotations.

A panoramic radiograph taken of the patient revealed supernumerary teeth (right and left permanent maxillary canine, right permanent mandibular central), congenitally missing teeth (right and left permanent mandibular canine), inactive caries of 55, 54, 53, 65, 73, 74 and 75, narrowed pulp chambers and canals in the primary teeth, right permanent mandibular first molar with three roots, extracted right and left maxillary primary first molars, only roots of the left and right mandibular primary first and second molars, space loss

Fig. 3 The oral findings of mother. (a) Lobulated tongue. (b) Hyperplastic oral frenula, a cleft near canine, congenitally missing teeth, enamel defects.

due to teeth extracted early (► **Fig. 4**). The mother's panoramic radiograph revealed that the left permanent mandibular lateral and right mandibular permanent central were congenitally missing, the right primary mandibular central was retained, caries were common in the teeth, narrowed pulp chambers and canals were seen, and numerous permanent teeth had been missing. Diastemata were also present between the teeth due to a unilateral cleft (► **Fig. 5**).

The patient's cephalometric film revealed that both the upper jaw (SellaNasion-A = SNA = 81.6 degrees) and lower jaw (SellaNasion-B = SNB = 78.5 degrees) were in a normal position and the patient was skeletal class I (SNA-SNB = ANB = 3.1 degrees). The patient's vertical direction of development was normal (GonionGnathion-SellaNasion = GoGn-SN = 31.6 degrees, ► **Fig. 6**). Procedures and space maintainers were programmed for the patient, and appointments were scheduled for treatment.

The patient was lost to follow-up for 2.5 years. At that age, the patient was on medication with Enalapril Sandoz 10 mg 3 months for the PKD and had a few teeth restorations and extractions.

The patient's oral examination and panoramic radiograph revealed that the bilateral deep clefts extending from the buccal frenulas divided the palate clearly in two lateral processes and the anterior segment (containing central and canine teeth). The first molar teeth had enclosed roots. The permanent teeth had normal morphology but pulp volumes had decreased. There were diastemata and rotations of the teeth due to lower frenula, deterioration of the relationship between the jaws more clearly on the right side due to the bilateral clefts on upper jaw, and open bite and increased space loss due to failure of the space retainers (► **Figs. 7 and 8**).

Protective dental treatment was applied to the patient. It was explained to the patient and her parents that regular oral care and appointments are important so that oral problems do not occur. The treatments needed by the patient were planned according to the analysis, and the parents were informed.

We consulted with other departments (orthodontics, periodontics, etc.) related to the current situation of the patient. In the follow-up programs, in a multidisciplinary approach, frenectomy of labial frenulas followed by fixed

Fig. 4 The panoramic radiograph of patient (January 16, 2015).

Fig. 5 The panoramic radiograph of mother (January 16, 2015).

orthodontic treatment is a treatment choice for the patient. If the patient cannot be treated with any orthodontic treatment, prosthetic procedures are planned to provide aesthetic and function rehabilitation. It was decided to follow up the growth and development of the face and jaw at regular

intervals along with regular follow-up. The patient was also followed by professionals in plastic surgery and nephrology.

Discussion

Our patient had OFDS1, which is the most frequent type of OFDS of the 13 different forms, and had typical clinical and radiological findings of the disease.

PKD, which is frequently seen in patients with OFDS1, was present in our patient and her mother. As kidney pathologies develop at later ages, the renal function follow-up is of great importance.⁶ Angiotensin converting enzyme inhibitor drugs, such as Enalapril Sandoz, taken for PKD can produce oral complications, such as dry mouth, loss of taste, gingival hyperplasia, and ulcerations.⁷ Although the mother did not have any mental problems, her oral problems indicate that such oral problems may also occur in a child with mental retardation in the future.

Our patient with OFDS1 had oral symptoms similar to those described in the literature.⁸ About 33% of patients with OFDS1 have a middle lip/upper lip pseudocleft,⁹ but this finding was not present in our patient.

Fig. 6 The cephalometric film of patient (January 16, 2015).

Fig. 7 The panoramic radiograph of patient after 2.5 years (May 23, 2017).

Fig. 8 The oral findings of patient after 2.5 years. (a) Diastemata due to lower frenulas, tooth rotations. (b) Bilateral deep clefts extending from the buccal frenulas. (c) Numerous hyperplastic oral frenulas.

OFDS1 is a syndrome that includes missing and supernumerary teeth.⁹ Only cases with supernumerary canines have been reported in the literature^{10,11}; a case with an excessive number of both primary and permanent incisors has also been reported.⁵ In our case, the upper bilateral supernumerary canines and lower, supernumerary canines were present. Also, the patient reported central and bilateral congenitally missing permanent mandibular canines, which has not been described in the literature previously.

OFDS2, Joubert, and Opitz trigonocephaly syndromes were present with similar findings to OFDS1. Autosomal recessive inheritance, but no hyperplastic oral frenula, is seen in patients with OFDS2. Similar findings are observed in patients with Opitz trigonocephaly syndrome, in addition to short stature and skin laxity.¹²

Although there are many articles in the literature that show the oral findings of OFDS1,^{5,8,10,11,13,14} the articles reporting changes in oral findings as a result of long follow-

up of patients are limited.^{15,16} Also, the oral and facial changes have been reported after orthodontic treatment of patients 21 and 14 years of age with OFDS1.^{17,18}

Although it is not specific, treatment of children with this syndrome requires a multidisciplinary approach in the medical and dental fields. The efficacy of dental specialist in this group is important to prepare and monitor a preventive program in children who are drug dependent due to PKD.

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