Case Report

Oculo-facio-cardio-dental Syndrome: *Report of a Rare Case*

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Abstract: Oculo-facio-cardio-dental (OFCD) syndrome is an extremely rare condition with ocular, facial, cardiac, and dental abnormalities. It is often unrecognized by many medical and dental professionals. Only 17 cases have been documented to date. Because all reported patients have been female, it has been suggested that OFCD syndrome is an X-linked dominant trait. Isolation and diagnosis of this syndrome is hard for the medical specialists. Only unique and specific findings are observed in dental and skeletal structures, which can easily be diagnosed by an orthodontist or general dentist. Therefore, it was the aim of this study to present a new case of this syndrome and to evaluate it from an orthodontic perspective in order to call the attention of orthodontists to this rare anomaly. (*Angle Orthod* 2006;76:184–186.)

Key Words: Oculo-facio-cardio-dental syndrome; OFCD; Congenital cataracts; ASD; VSD; Canine radiculomegaly

INTRODUCTION

Oculo-facio-cardio-dental (OFCD) syndrome is an extremely rare condition with ocular, facial, cardiac, and dental abnormalities. It is often unrecognized by many medical and dental professionals, and only 17 cases have been documented to date.[1–10]

Hayward[1] was the first to describe an association between radiculomegaly of the canines and congenital cataracts. Gorlin et al[11] was the first to name the condition as OFCD syndrome. Because all reported patients have been female, it has been suggested that OFCD syndrome is an X-linked dominant trait.[4,9] Isolation and diagnosis of this syndrome is hard for the medical specialists.

Only unique and specific findings are observed in dental and skeletal structures, which can easily be diagnosed by an orthodontist or a general dentist. Therefore, the aim of this report is to present a new case of this syndrome, to evaluate it from an orthodontic perspective, and to bring this rare anomaly to the attention of the orthodontic community.

CASE REPORT

Patient ZÖ was a 15-year-old female when first referred to the Orthodontic Department of Süleyman Demirel University, Faculty of Dentistry. She had normal intelligence and a tall stature. Her chief complaints were malalignment of her anterior teeth and speech problems. A detailed medical and dental history was obtained from the patient and from her parents. After a normal pregnancy, she was born with a birth weight of 3800 g. Her early psychomotor development was mildly retarded. Parents also mentioned a delayed eruption of the primary dentition. On a past medical history, she was diagnosed as having ostium secundum type atrial septal defect (ASD) and membranous ventricular septal defect (VSD). Both these defects were repaired when she was two years old.

The patient had congenital cataracts and was operated for both eyes at four and at six years of age. There was no family history of cataracts, birth defects, genetic disease, or cardiac anomalies.

Severe dental and skeletal abnormalities were found in a detailed clinical examination. A long and narrow face, high nasal bridge, broad nasal tip with separated cartilages, and a long philtrum were noteworthy in extraoral examination (Figure 1). The eyebrows were laterally curved and thick. The patient had a straight profile. An intraoral examination revealed a Class II malocclusion with an extremely deep overbite (Figure 2).

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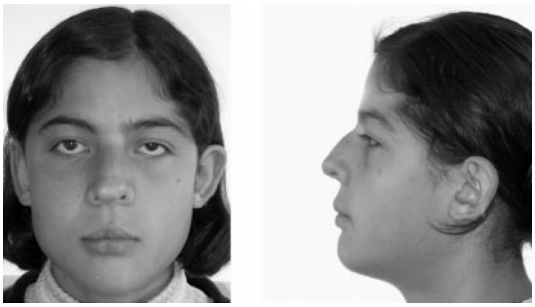

FIGURE 1. Extraoral photographs.

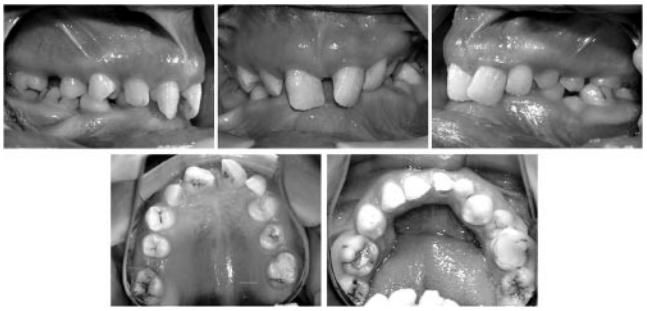

FIGURE 2. Intraoral photographs.

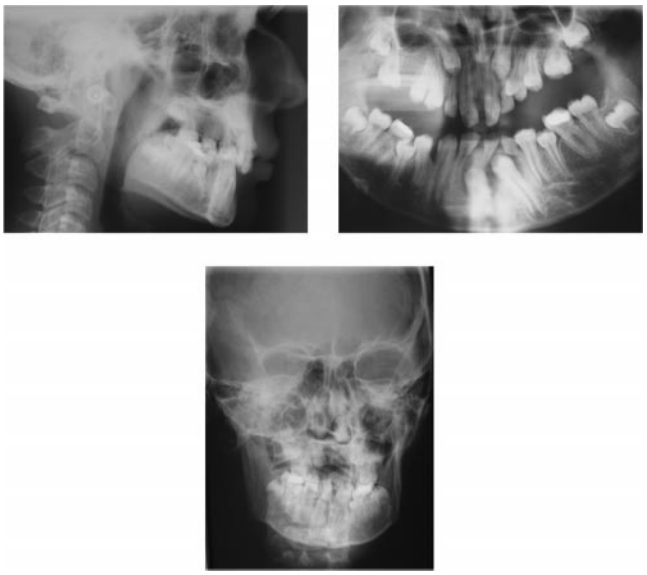

FIGURE 3. Cephalometric, panoramic, and posteroanterior radiographs.

A delayed eruption of the permanent teeth was observed. A skeletal Class II and high angle pattern is evident in the lateral cephalometric radiograph (Figure 3). The panoramic radiograph revealed teeth with extremely long roots and open apices (Figure 3). The upper canine roots were in relation with the inferior border of the orbits, and the lower canine roots almost reached the lower border of the mandible. She had her upper first molars extracted. The lower left lateral incisor was congenitally absent. The maxillary canines, the mandibular left canine, mandibular second bicuspids, and mandibular right central incisor were impacted. The maxillary left bicuspids had dilacerated roots. Enamel defects were observed in the mandibular left central incisor. All third molar germs were present. Posteroanterior radiograph revealed a skeletal midline shift to the right (Figure 3).

The patient was examined by a cardiologist and an ophthalmologist. On physical examination, her blood pressure was 110/70 mm Hg and pulse rate was 80 bpm, with a regular rhythm. Cardiac auscultation revealed a systolic murmur over the apex.

Echocardiography revealed a mild mitral and moderate tricuspid insufficiency and mild pulmonary hypertension (40 mm Hg). No residual ASD or VSD was detected.

The ophthalmological examination revealed amblyopia and exophoria on the right eye and regressive vision impairment.

Her intelligence was not tested but may reach average level. Because all main findings were recognized in this patient, the case was diagnosed as OFCD syndrome. An integrated orthodontic, endodontic, and prosthodontic treatment was planned and started.

DISCUSSION

OFCD is a very rare syndrome. To date, only 17 cases have been reported.[1–10] The syndrome has been reported only in women, suggesting that the syndrome is lethal in males. An X-linked dominant inheritance seems to be likely because Wilkie et al[4] and Hedera and Gorski[9] reported transmission of the syndrome from mother to daughter.

Because of the congenital cataracts and cardiac anomalies, many patients with OFCD syndrome have been misdiagnosed as having rubella embryopathy.[12] Orthodontists can play a major role in the differential diagnosis of this rare syndrome. First, dental findings of this syndrome are characteristic and unique. The diagnosis is only confirmed when the radiculomegaly becomes evident.[8] Ocular and cardiac findings, in isolation, may be seen in a variety of other conditions. Dental specialists often establish the association between these findings. Second, patients with OFCD generally suffer from severe malalignment of their teeth and seek orthodontic treatment. Because the panoramic radiographs are obtained routinely before orthodontic treatment, an orthodontist has a chance to diagnose the syndrome early, as in this case.

In the literature, similar findings were reported in patients with OFCD syndrome. These findings can be categorized as follows.

Dental findings

The most remarkable dental finding is the radiculomegaly of the teeth, especially the canines.[1–10] The lengths of the canines are far beyond the average length of a normal maxillary canine (27 mm). In this case, the length of lower left canine was 46 mm, and it continues to elongate. The canines continue to grow until the orbit or inferior border of the mandible is reached.[8] The timing of root closure is obscure and especially the canine teeth have an open apex.[1–10] The dental eruption in both the deciduous and the permanent dentition is consistently slow and delayed.[1–10] Missing, malpositioned, malformed permanent teeth and oligodontia are also reported.[1,2,4–8] Wilkie et al[4] reported unusually large crowns in several incisor and canine teeth. In this case, maxillary central incisors and mandibular right lateral incisor had large crowns. We supposed that gigantism of the premaxilla is correlated with continuous elongation of the upper anterior teeth.

Ophthalmologic findings

Bilateral congenital cataracts,[1,2,4–10] microphthalmia,[2,4,5,7] regressive vision impairment,[1,2,4–7] and ptosis[4,5,7] were reported in patients with OFCD syndrome. Although secondary glaucoma is common in these patients,[1,4,5,7] no evidence for it was observed in this case.

Cardiac findings

ASD,[4,5,7,9] VSD,[4,5,7,10] mild cardiomegaly,[8] ventricular and atrial hypertrophy,[8] benign peripheral pulmonic stenosis,[8] and mitral valve prolapse[2] were commonly reported in OFCD syndrome. Similarly, our patient also suffered from ASD and VSD.

Facial findings

A long, narrow face,[2,4–9] high nasal bridge,[2,5,7,9] broad nasal tip with separated nasal cartilages,[2,5–7,9] laterally curved and thick eyebrows,[4,5,7] long philtrum,[5,7] and clefts of the hard/soft palate[2,5–7,9] were reported. All facial findings of the OFCD syndrome were obvious in our patient except a cleft of the hard/soft palate.

Other findings

Hearing impairment,[5] mental[6,7] and psychomotor retardation,[4,9] and syndactyly of toes two and three were also reported.[2,4–7,9] However, none of these findings were present in our patient.

CONCLUSIONS

- OFCD syndrome is an extremely rare condition with ocular, facial, cardiac, and dental abnormalities.
- Although characteristic findings are observed in dental and skeletal structures, which can easily be diagnosed, it is often overlooked by many medical and dental professionals.

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