Dental Management of a Patient With Oculo-facio-cardio-dental Syndrome

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ABSTRACT

Oculo-facio-cardio-dental (**OFCD**) syndrome is an X-linked dominant syndrome characterized by congenital cataract, microphtalmia, secondary glaucoma, long narrow face, high nasal bridge, pointed nose with cartilages separated at the tip, cleft palate, atrial septal defect (ASD), ventricular septal defect (VSD), radiculomegaly, delayed eruption, oligodontia, and persistence of primary teeth. The aim of this report is to indicate the importance of early diagnosis and management of oculo-facio-cardio-dental syndrome. (**J Dent Child 2008;75:306-8**)

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KEYWORDS: OCULO-FACIO-CARDIO-DENTAL SYNDROME, DENTAL MANAGEMENT, RADICULOMEGALY

Oculo-facio-cardio-dental (**OFCD**) syndrome is an X-linked condition with characteristic ocular, facial, cardiac, and dental findings in affected females and presumed lethality in affected males.[1] The combination of teeth with large roots and congenital cataracts were first described by Hayward in 1980.[2] Wilkie et al in 1993 described a mother and daughter with ocular, facial, cardiac, and dental features, and proposed that they represented a distinct syndrome with autosomal dominant inheritance.[3] This unique combination of ocular, facial, cardiac, and dental findings was gathered under the name of oculo-facio-cardio-dental syndrome in 1996 by Gorlin et al.[4]

The characteristic findings of OFCD syndrome are microphtalmia, congenital cataract, glaucoma, long narrow face, high nasal bridge, bifid nasal tip, ventricular septal defect, atrial septal defect and mitral valve abnormalities.[1,5]

The most common dental findings are radiculomegaly, and delayed eruption of both the primary and permanent dentition. Primary teeth are retained, and permanent teeth are missing, malpositioned, and malformed.[5,6] Some other common dental findings, such as root dilaceration, microdontia, and enamel defects, have been reported in the dental literature.[1-3,4,5,7-9]

The gene for the condition has been identified, with mutations having been established in affected individuals in the BCOR gene on chromosome Xp11.4. The exact function of the gene is not known, and the etiopathogenic relationship between intragenic termination mutations and eventual radiculomegaly and other dental anomalies described in OFCD syndrome is also unclear.[9]

The aim of the present report is to emphasize the importance of early diagnosis and management of oculo-facio-cardio-dental syndrome.

CASE DESCRIPTION

A 12-year-old female patient was referred to our pediatric dentistry clinic for a routine dental examination. She was 8 years old when first diagnosed with OFCD syndrome in another medical center. She was the only child of nonconsanguineous parents, and was born at term. Her birth height, weight, and head circumference were unavailable. Her motor and mental development was normal. She was noted to have bilateral congenital cataracts, microphtalmia in the right eye, and left exotropia at birth. A mild cardiomegaly was also detected. There was no family history of cataracts, birth defects, or genetically transmitted diseases. The patient had surgery to remove her bilateral cataracts at 4 weeks of age.

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Physical examination showed a bulbous, bifid nose. Her oral findings were delayed eruption of the primary and permanent dentition, oligodontia, retained primary teeth, and radiculomegaly in her permanent maxillary incisors (Figure 1a-c). The panoramic radiograph revealed maxillary right and left permanent central incisors and maxillary right lateral incisor with extremely long roots and open apices (Figure 2). Eighteen of her permanent teeth (maxillary right permanent canine, first and second premolars, second molar; maxillary left permanent lateral incisor, canine, first and second premolars, second molar; mandibular left permanent lateral incisor, canine, second premolar, second molar; mandibular right permanent lateral incisor, canine, first and second premolars, second molar) were congenitally missing. Two of her primary teeth (maxillary right primary first molar and maxillary left primary second molar) were restored with composite restorations. Stainless steel crowns were prepared to restore 2 profound cavities (mandibular left primary second molar and mandibular right primary second molar). Teeth with a poor prognosis (maxillary right primary second molar and mandibular left primary first molar) were extracted, and space maintainers were placed for the maxilla and mandible (Figure 3a-c). Topical fluoride was applied every 6 months, and the patient was scheduled for regular control for the space maintainers every 3 months.

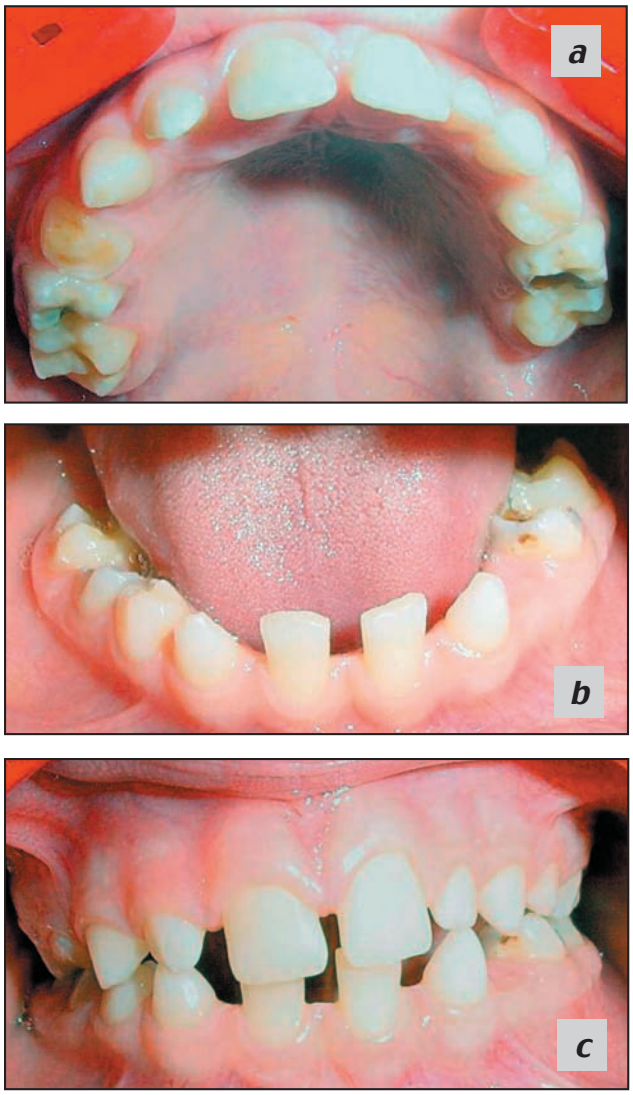

Figure 1a–c: Intraoral view of the patient (after extraction of mandibular left first primary molar tooth).

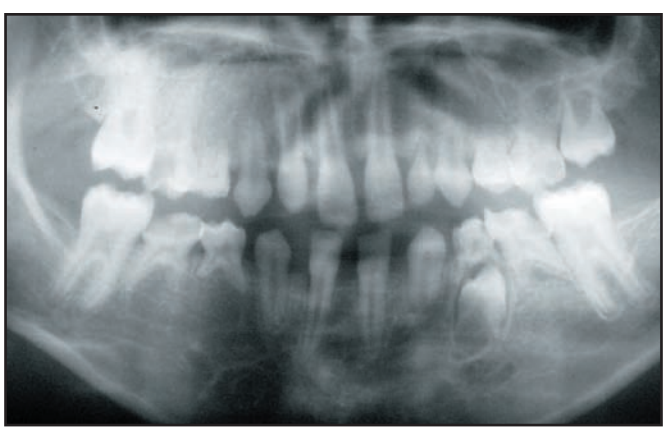

Figure 2: Panoramic radiograph of the patient.

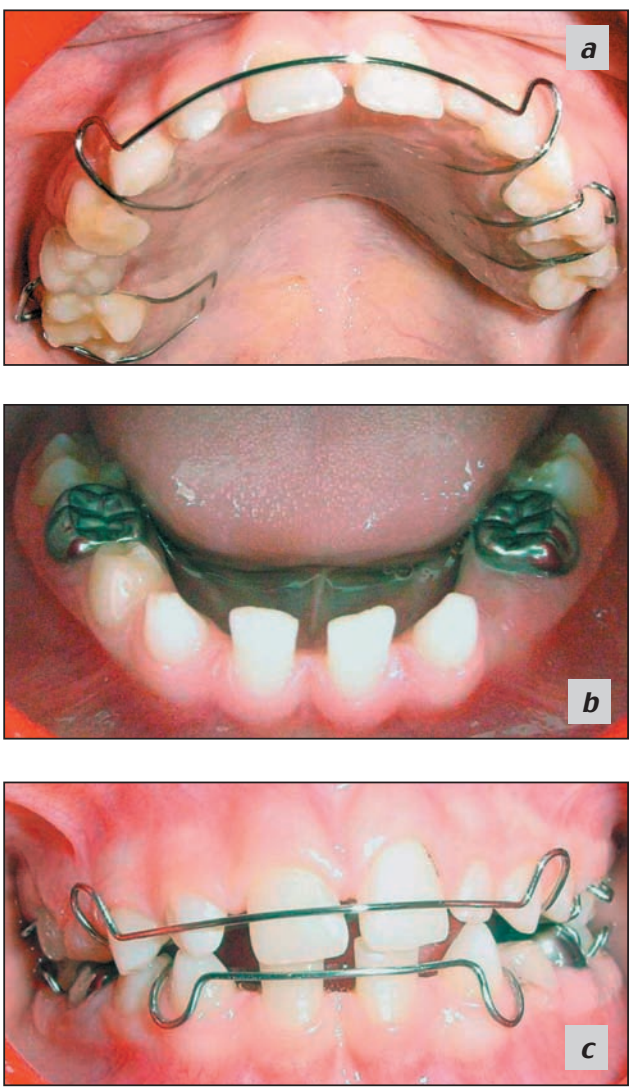

Figure 3a–c: Intraoral view of the patient after treatment.

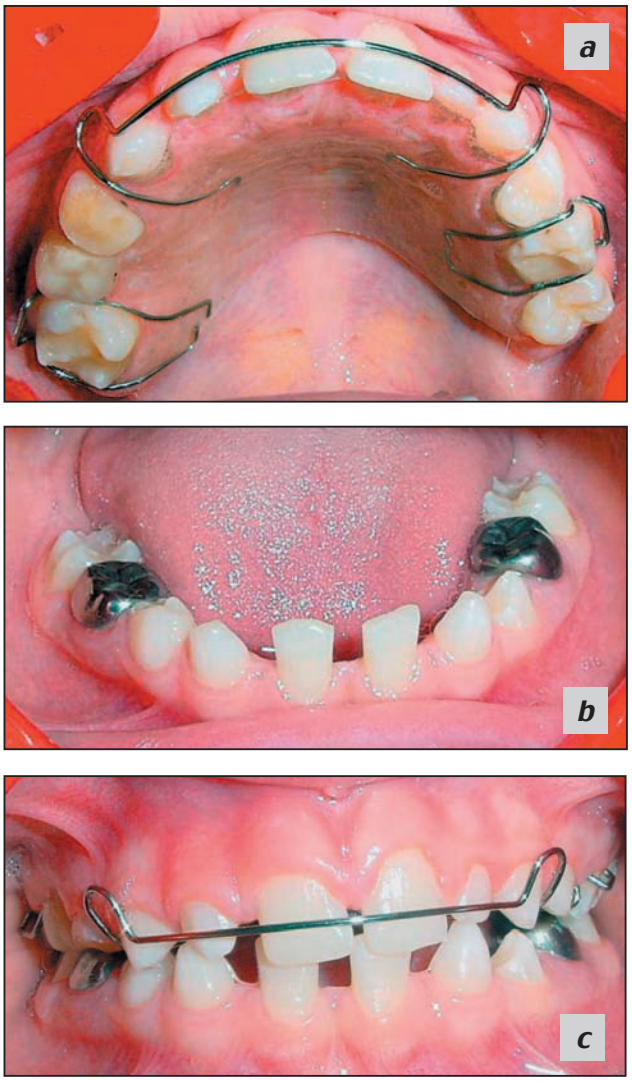

Figure 4a–c: Intraoral view of the patient 1 year after treatment.

One-year follow-up of the patient revealed a good space maintenance (Figure 4a-c).

DISCUSSION

OFCD syndrome is a rare condition with ocular, facial, cardiac, and dental abnormalities.[1,5] It is characterized by congenital cataract, secondary glaucoma, microphtalmia, narrow face, high nasal bridge, broadening of the nasal tip with separated cartilages, cleft palate, ventricular septal defect, atrial septal defect, and mitral valve abnormalities, canine radiculomegaly, oligodontia, delayed eruption of dentition, retained primary teeth, malocclusion, root dilaceration, variable root length, microdontia, and enamel defects.[5,9] This syndrome is extremely rare—so far, only 18 known cases have been documented.

OFCD syndrome has been associated with radiculomegaly—particularly of the canines and occasionally of other teeth, including premolars and incisors.[2,4] In the present case, radiculomegaly was detected only in her permanent maxillary incisors. Although there is no radiculomegaly detected in the both maxillary and mandibular primary canines, the other findings confirmed the syndrome. The absence of radiculomegaly in the canines in the present case could be due to the fact that the subject was congenitally missing all her permanent canines.

Hypodontia has been reported in many OFCD syndrome cases.[3,4,7] In the present case, 18 permanent teeth (excluding maxillary and mandibular permanent third molars) were congenitally missing. This can cause physical and psychological problems in the patient. Therefore, the aim of the treatment planning was to preserve the primary teeth as long as possible.

Early diagnosis and treatment of OFCD patients is very important to continue the development of the teeth and the jaws.

REFERENCES

1. Oberoi S, Winder AE, Johnston J, Vargervik K, Slavotinek AM. Case reports of oculofaciocardiodental syndrome with unusual dental findings. Am J Med Genet 2005;136:275-7.
2. Hayward JR. Cuspid gigantism. Oral Surg Oral Med Oral Pathol 1980;49:500-1.
3. Wilkie AO, Taylor D, Scambler PJ, Baraitser M. Congenital cataract, microphtalmia, and septal heart defect in two generations: A new syndrome? Clin Dysmorphol 1993;2:114-9.
4. Gorlin RJ, Marashi AH, Obwegeser HL. Oculo-facio-cardio-dental (OFCD) syndrome. Am J Med Genet 1996;63:290-2.
5. Schulze BRB, Horn D, Kobelt A, Tariverdian G, Stellzig A. Rare dental abnormalities seen in oculo-facio-cardio-dental (OFCD) syndrome: Three new cases and review of nine patients. Am J Med Genet 1999;82:429-35.
6. Opitz C, Horn D, Lehmann R, Dimitrova T, Fasmers-Henke K. Oculo-facio-cardio-dental (OFCD) syndrome. J Orofac Orthop 1998;59:178.
7. Hedera P, Gorski JL. Oculo-facio-cardio-dental syndrome: Skewed x chromosome inactivation in mother and daughter suggest x-linked dominant inheritance. Am J Med Genet 2003;123:261-6.
8. Marashi AH, Gorlin RJ. Radiculomegaly of canines and congenital cataracts—a syndrome? Oral Surg Oral Med Oral Pathol 1990;70:802-3.
9. McGovern E, Al-Mudaffer M, McMahon C, Brosnahan D, Fleming P, Reardon W. Oculo-facio-cardio-dental syndrome in a mother and daughter. Int J Oral Maxillofac Surg 2006;35:1060-2.