CASE REPORT

A rare syndrome with unusual dental findings: Oculo-facio-cardio-dental syndrome

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ABSTRACT
Radiculomegaly of a tooth is a rare condition. When it is associated with other dental abnormalities, facial dysmorphism and congenital cataracts, the condition is named as oculofaciocardiodental (OFCD) syndrome. This is an X-linked dominant trait, reported only in women, suggesting that it is lethal in males. The most consistent and pathognomic dental finding of OFCD is radiculomegaly (extremely long roots), particularly of the canines and occasionally of other teeth including premolars and incisors, which can easily be diagnosed on dental panoramic radiographs by a dentist or an orthodontist. Till date, 21 cases have been reported worldwide. The aim of this report is to present a new case of a 24 year old female affected by this syndrome and to evaluate it from a dental perspective to call the attention of dentists to this rare anomaly.

Key words: Congenital cataract, facial dimorphism, radiculomegaly, X-linked dominant inheritance

INTRODUCTION

Oculo facio cardio dental (OFCD) syndrome was first reported by Hayward[¹] in 1980 and named by Obwegeser and Gorlin,[²] as a rare genetic syndrome with multisystemic involvement. Rarestness can be assumed that till date, as per published literature, only 21 cases have been reported worldwide.[³]

This is a X-linked rare congenital anomaly, with incidence of less than 1 in 1 million people and it might be lethal in males.[²⁴] It is characterized by heterogeneous clinical features such as dental radiculomegaly, congenital cataract, facial dymorphism and congenital heart disease.[⁵]

Radiculomegaly is characteristic of this syndrome and was first reported by Weine,[⁶] Wilkie and Chambers.[⁷] Gorlin et al.[²] reported that, in addition to the radiculomegaly, the apex of the canines do not close until adulthood and the roots continue to grow until the orbit and lower border of the mandible are reached.[⁸⁹]

CASE REPORT

A 24 year old female reported to the Department of Orthodontics and Dentofacial Orthopaedics for the management of malaligned teeth in upper and lower jaw. She had normal intelligence and was tall. Family history revealed that her father had similar Class III malocclusion, but no other symptoms were observed. 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 2500 g. She was having episodes of cyanosis while crying i.e. a bluish tint of skin, lips and finger nails; and was diagnosed to have ventricular septal defect (VSD) by echo-cardiography. A VSD is a defect in the septum between the heart’s two lower chambers, the ventricles. This defect allows oxygen-rich blood from left ventricle to mix with oxygen-poor blood from right ventricle. The problem persisted until the defect was repaired. VSD was repaired with open-heart surgery at the age of two years.

Patient had also reported an history of umbilical hernia at birth. A hernia at birth will push the belly button out. It showed more when she used to cry because the pressure from crying makes the hernia bulge out more. To handle the situation, the parents used to apply pressure by adhesive tapes over the belly button as prescribed by the paediatrician.
The umbilical hernia shrunk and closed by itself in around 22 months of age.

The patient had congenital cataract and was operated for both eyes at four and at six years of age by an ophthalmologist [Figure 1a]. There was no family history of cataract, birth defects, genetic disease or cardiac anomalies. She also had syndactyly of second and third toes. The fingers were normal. [Figure 1b and c].

Parents mentioned delayed eruption of the primary dentition, initiated at the age of 2 year, 6 months. Severe dental and skeletal abnormalities were found on detailed clinical examination.

Extra-oral examination presented a long and narrow face with concave profile. The eyebrows were laterally curved and thick [Figure 2]. Intraoral examination revealed a Class III malocclusion with a negative over jet and deep overbite.

Dental and chronological age of the patient was not coinciding as multiple retained deciduous teeth, namely maxillary right lateral incisor, first and second molars; left lateral incisor and first molar along with root stumps of both right and left canine were retained. In the mandibular arch, right central incisor, lateral incisor, canine and left canine and second molar teeth were retained. Both the maxillary right premolars and mandibular left second premolar were absent on clinical examination [Figure 3].

Severe dental and skeletal abnormalities were found on detailed radiographic examination. Lateral cephalometric radiograph showed skeletal Class III and high angle pattern [Figure 4a] Posterio-anterior radiograph revealed a skeletal midline shift to the right [Figure 4b]. The panoramic radiograph showed permanent teeth with extremely long roots and open apices. The roots of maxillary canines were in relation with the inferior border of the orbits and the lower

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**Figure 1:** (a) Clinical features of patient at the age of 22 years demonstrated deeply set eyes and a broad nasal tip. As a consequence of glaucoma, the globe of the left eye is larger than right eye. (b) Syndactyly of 2nd and 3rd toes in both the foot. (c) Fingers in both the hands were normal

**Figure 2:** (a) Frontal Photograph showing clinical features of patient as right-sided microphthalmia with upward slanting palpebral fissures, enlargement of the left eye secondary to glaucoma, laterally curved eyebrows, long narrow face and a wide nasal tip. (b) Photograph showing midline diastema. (c) Profile photograph showing deficient mid face, large mandible and concave facial profile

**Figure 3:** Intra oral photographs of patient showing class III malocclusion, reverse over jet, multiple retained deciduous teeth and malformation of permanent teeth
canine roots almost reached the lower border of the mandible. Permanent maxillary right first premolar was missing while second premolar was impacted with distal aggregation. The maxillary left central incisor had dilacerated root; all four third molars were congenitally missing [Figure 4c]. All the main findings were recognized in this patient, therefore the case was diagnosed as OFCD syndrome. An integrated orthodontic, endodontic and prosthodontic treatment was planned.

**DISCUSSION**

Although patients seek treatment in different specialties at different times depending on priority, the diagnosis of oculofaciocardiodental syndrome is difficult for medical specialists and the syndrome is often undiagnosed. Dental anomalies form an integral aspect of many dysmorphic syndromes, often representing important clinical clues to the true underlying diagnosis.

OFCD syndrome is a rare X-linked dominant condition associated with characteristic ocular, facial, cardiac and dental features. The gene associated with this condition is located on the X-chromosome, which is one of the two sex chromosomes. Mutations in the BCOR gene cause OFCD syndrome. Several mutations in the BCOR gene have been found in people with OFCD syndrome. These mutations prevent the production of any functional protein from the altered gene, which disrupts the normal development of the eyes and several other organs and tissues before birth.

Patients generally suffer from severely malaligned teeth and seek orthodontic treatment. Because panoramic radiographs are obtained routinely before orthodontic treatment, an orthodontist has a chance to diagnose the syndrome early. The most consistent dental finding of this syndrome is radiculomegaly of canine and occasionally of other teeth including premolars and incisors. Other dental abnormalities include oligodontia, fused teeth, supernumerary teeth, malformed permanent teeth, malocclusion, root dilacerations, macroodontia and enamel defects. Dental eruption in both deciduous and the permanent dentition is consistently slow and delayed. Extreme overbite, and constricted maxilla may be associated features.

Because of the congenital cataracts and cardiac anomalies, many patients with OFCD syndrome have been misdiagnosed as having rubella embryopathy. The eye abnormalities (microphthalmia, cataract and glaucoma) can affect one or both eyes leading to possible loss of vision or blindness. Facial features include a long, narrow face with deep-set eyes and a broad nasal tip that may be divided by a cleft.

Congenital cataracts, microphthalmia, secondary glaucoma, strabismus, ptosis, regressive vision impairment and hypertelorism were also reported. Exotropia is very common. Babies with this condition may be born with an atrial or ventricular septal heart defect or mitral valve prolapse. Ocular and cardiac findings, in isolation, may be seen in a variety of other conditions.

Cardiac anomalies may include atrial septal defect (ASD) and VSD; mitral valve defects; mild cardiomegaly; ventricular and atrial hypertrophy and mitral valve prolapse. Benign peripheral pulmonary stenosis was commonly reported in OFCD syndrome. This patient also had ASD and VSD. All these defects can be easily diagnosed by echocardiography. Cardiac catheterization can also be done to help the cardiologist to decipher whether blood is mixing between the two chambers of the heart. These defects are repaired with open heart surgery. Repairing the defects can greatly improve a child’s health and quality of life.

Many children have umbilical hernia at birth. The hernia usually is not painful or dangerous, and it often closes on its own without treatment before 12 months of age. If the hernia has not closed by 5 years of age, the child may require surgery to close it. Syndactyly, wherein two or more digits are fused together, is a very frequent finding. The patient showed complete, complex Type I syndactyly of 2nd and 3rd toes. [Figure 5a and b].

Facial anomalies include long narrow face, high nasal bridge, broad nasal tip with separated nasal cartilages, bifid nose, laterally curved and thick eyebrows, ear deformity, long philtrum, cleft palate and/or submucous cleft palate.

Others associated features include reduced birth weight; septate vagina; anteriorly placed anus; hammer-type flexion of the second and fourth toes; radio-ulnar synostosis; hypoplastic thumbs; vertebral and rib anomalies; cerebral
and cranial defects; bifid uvula; intestinal malrotation; hearing impairment; mental and psychomotor retardation.

MANAGEMENT

Management of OFCD syndrome requires appropriate cardiac, ophthalmic and dental care. The present patient came to the Dental Department after management of cardiac, ocular and other problems. For dental management, an integrated orthodontics, endodontics and prosthodontics treatment was planned and started based on the investigation.

CONCLUSION

Orthodontists, along with all dentists, might be in the best position to diagnose OFCD syndrome because they work with dental radiographs. They can therefore compare the eruption, root formation and apical closure of teeth throughout the orthodontic treatment period. The diagnosis of this syndrome is vital so as provide better prospects of future life.

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