

Oculofaciocardiodental Syndrome: A Rare Disorder with Canine Radiculomegaly

AISHWARYA RATHOD¹, PRIYANKA JAISWAL², PANKAJ BANODE³, GAJENDRA MANAKSHE⁴, DEEPIKA MASURKAR⁵

(CC) BY-NC-ND

ABSTRACT

The ocular, face, dental, and cardiac systems are all impacted by the uncommon condition known as Oculofaciocardiodental (OFCD) syndrome. OFCD is only present in females due to its X-linked genetic inheritance and male embryonic lethality. The most common ailment in these patients is canine radiculomegaly. This particular feature highlights the role of dentists in detecting this syndrome. Accompanied with a report on a female patient with unique facial traits, we provide a comprehensive description of OFCD. In this case we have discussed with the typical features of OFCD. A 20-year-old female patient with OFCD syndrome presented with unique characteristics affecting ocular, cardiac, facial, and dental systems. Patient had congenital cataract, elongated roots of canine and incisors, elongated face with convex profile, cardiac abnormalities like atrial septal defect and ventricular septal defect. On the basis of clinical and radiographic findings and necessary medical diagnostic procedures, patient was diagnosed with OFCD syndrome. But due to lack of keen observation, the syndrome was undiagnosed and treatment was delayed. It is critical for every clinician to find out reasons and answers of abnormal finding in patient.

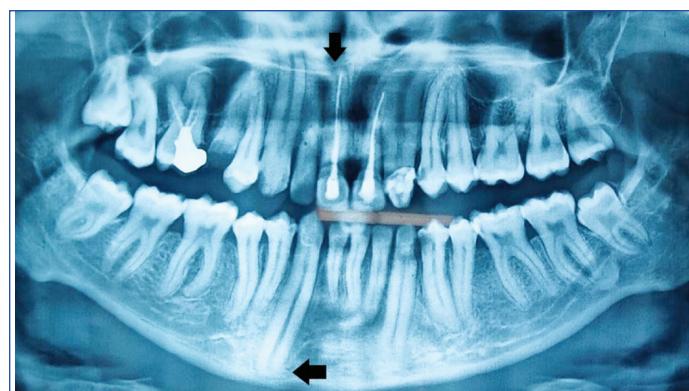
Keywords: Electrocardiography, Mutation, Orthopantomogram, Ultrasonography

CASE REPORT

A 20-year-old female patient came to department of periodontics with the chief complaint of deposits over teeth since two months. She had completed her orthodontic treatment almost a year back. Earlier she had multiple over retained and malaligned teeth. The root canal treatment was done for central incisors previously. Height of patient was 152 cm. The weight of the patient was 55 kg. Intraoral findings included anterior central incisors with porcelain fused metal crown, teeth deposits, halitosis, congenitally missing right lower lateral incisor, proximal caries in relation to the left lower canine and first premolar. At four months of age, the patient underwent surgery for a congenital cataract in her right eye, performed by an ophthalmologist. Patient had congenital cataract and glaucoma as seen in B scan ultrasonography. A comprehensive dental and medical history was recorded and patient had no any significant family history. She was born after normal pregnancy with weight 2500 g. Patient was having normal intelligence. For diagnosis and treatment plan, patient was advised Orthopantomogram radiograph. After radiographic evaluation it was seen that patient had elongated roots of canines (maxillary and mandibular) and incisors (maxillary) [Table/Fig-1].

Extraoral examination revealed patient was tall with elongated face and convex profile. The eyebrows were broad and laterally curved, as shown in [Table/Fig-2].

Second toe of the right leg had hammer toe as shown in [Table/Fig-3].



[Table/Fig-1]: Orthopantomogram showing elongated upper and lower canines and upper central incisors.



[Table/Fig-2]: Extraoral features- Elongated face with convex profile.



[Table/Fig-3]: Hammer toe.

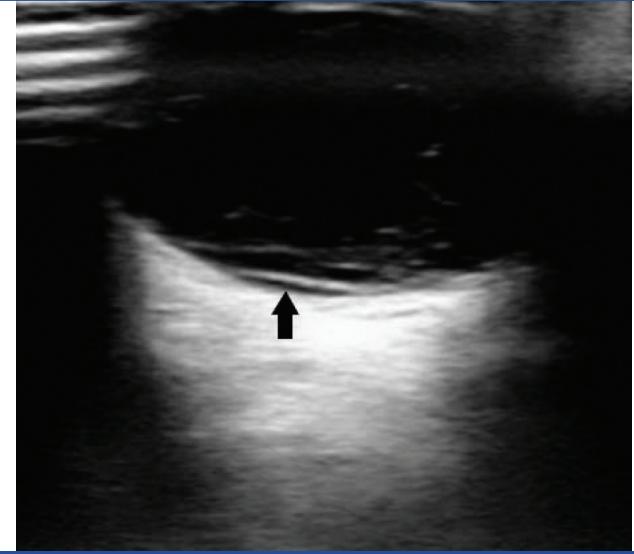
The differential diagnosis for this case was Oculofaciocardiodental (OFCD) syndrome and Marfan syndrome. The provisional and final diagnosis was OFCD syndrome. Patient consent was taken. Full mouth scaling was done and good dental hygiene was advised. For further investigation, the patient was sent to cardiologist and ophthalmologist. The changes observed in electrocardiography and B scan ultrasonography have been illustrated in [Table/Fig-4-6].



[Table/Fig-4]: White arrow shows defect between right and left atrium (Atrial septal defect).



[Table/Fig-5]: Patent Ductus Arteriosus (PDA).



[Table/Fig-6]: Retinal detachment.

DISCUSSION

Hayward originally described the OFCD condition in 1980 [1]. Gorlin RJ et al., later identified it as a rare hereditary syndrome with multisystemic involvement [2]. There have been no more than 100 OFCD cases documented worldwide to date [3]. OFCD syndrome is an X-linked disorder that only affects females and manifests as distinctive ocular, facial, cardiac, and dental symptoms whose prevalence is less than one in a million and which may

be fatal in males [4]. Congenital cataracts, secondary glaucoma, microphthalmia or microcornea are examples of eye abnormalities. The face is long and narrow, with a cleft palate, large philtrum, high nasal bridge, broad nasal tip with laterally curved, thick eyebrows. Atrial Septal Defects (ASDs) and ventricular septal defect, as well as mitral valve prolapse, are cardiac disorders. Canine radiculomegaly is the most frequent dental anomaly. It occurs when the roots of the maxillary incisors and canines grow until they come into contact with the orbital cortical plate or the lower boundary of the jaw [5]. Oligodontia, retained primary teeth, delayed tooth eruption and development and variable root length are further observations.

There has been a normal distribution of intelligence. The effect on an OFCD patient's quality of life varies on which organs are affected. The expressiveness of this illness is incredibly variable. While eye problems might result in impaired vision or blindness and heart defects can have potentially fatal implications, root gigantism may make orthodontic treatment more difficult [6].

OFCD is challenging for medical specialists to identify and frequently goes undetected, despite patients seeking care in various disciplines at various periods depending on priority. The characteristics of this medical condition exhibit a wide range of symptoms, including difficult-to-identify dental defects at birth. Many dysmorphic syndromes include dental deformities as a key component, which frequently serves as a key clinical indicator of the underlying disease. An uncommon dominant X-linked disease known as OFCD syndrome has recognisable ocular, facial, cardiac, and dental characteristics. The X-chromosome, one of the two sex chromosomes, contains the gene linked to this condition. OFCD syndrome is brought on by Back Central Optic Radius (BCOR) gene mutations. In individuals with OFCD syndrome, the BCOR gene has undergone a number of mutations. The eyes' usual growth and many additional tissues and organs before birth are disrupted by these mutations because they stop the altered gene from producing any functional proteins [5].

The following conditions may occur in this syndrome: The most significant characteristics for diagnosing OFCD are those that are associated with facial, ocular, and dental traits. These characteristics are given in [Table/Fig-7]. Additional observations include oligodontia, retained primary teeth, delayed tooth development and eruption, and varying root length. There have been not more than 100 cases of the extremely rare condition known as the OFCD syndrome reported [7]. Many patients with OFCD syndrome have received incorrect diagnoses of rubella embryopathy due to congenital cataracts and heart abnormalities. The characteristics of this syndrome exhibit a wide range of symptoms, which is difficult to identify dental defects at birth. It is crucial to differentiate between Marfan syndrome and OFCD when making a diagnosis. While both Marfan and OFCD syndromes are genetic illnesses, Marfan syndrome is autosomal dominant and can affect both sexes, whereas OFCD is mostly X-linked and affects females. Marfan syndrome affects the skeletal, ocular, cardiovascular, and other systems, whereas OFCD mainly affects the ocular, facial, cardiac, and dental systems [8]. In addition to OFCD defects, Marfan syndrome also affects the musculoskeletal, central nervous, pulmonary, and integumentary systems [9].

Feature	Findings
Dental	Oligodontia
	Radiculomegaly
	Late dentition
	Root dilacerations
	Incorrect positions of teeth
Ocular	Exotropia
	Congenital cataract
	Microphthalmia
	Ptosis

Facial	Wide tip of the nose
	Narrow face
	Elevated nasal bridge
	Eyebrows with a curve
	Clefts of the orofacial region
Cardiac	ASD- Atrial septal defect
	VSD- Ventricular septal defect
	MVP- Mitral valve prolapse
Other	Normal intelligence
	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 [4]

Table/Fig-7: Characteristics occurring in OFCD syndrome.

Nguyen TT et al., in their case mentioned that a female patient with long root had found abnormalities in the teeth, ocular, facial, and cardiac features, with radiculomegaly of the canines being a specific feature of OFCDs. The patient's genetic analysis shown a pathogenic heterozygous deletion at intron 11 of the BCOR gene [10]. Martinho J et al., reported case in which a multidisciplinary strategy was used to treat a female patient with OFCD syndrome who was referred for orthodontic intervention and occlusal rehabilitation [11]. Zhou Y et al., reported patient with bilateral congenital cataracts and systemic findings include ASD, patent ductus arteriosus, congenital clubfoot, and syndactyly. Also, patient was noted to have narrow palpebral fissure, almond-shaped eyes, flat nasal bridge, and broad nasal tip. Authors concluded that patients with multiple medical issues which may fit in to diagnosis of OFCD should be diagnosed earlier [12]. Davoody A et al., in their case presented a female patient with facial features and an overview of OFCD [6].

In the present case, the dental abnormality present in the patient was radiculomegaly of the canine and maxillary incisors. The length of maxillary canine increases until they reach the orbit or inferior border of the mandible, the canines is still growing. Patient had reported that she had misaligned teeth and her dental age was not coinciding with the chronological age. She had completed orthodontic treatment one year back.

Cardiac abnormalities may include atrial septal defect, ventricular septal defect, mitral valve defects, moderate cardiomegaly, ventricular and atrial hypertrophy, and mitral valve prolapse are possible cardiac malformations. OFCD syndrome frequently included reports of benign peripheral pulmonic stenosis [13]. In this case, patient reported with ASD and PDA. Additionally documented conditions included congenital cataracts, microphthalmia, subsequent glaucoma, strabismus, ptosis, regressive vision impairment, and hypertelorism.

Exotropia occurs frequently. In this case, patient had congenital cataract and glaucoma as seen in B scan ultrasonography. Among the described facial traits was elongated narrow face, elevated nasal bridge with separated nasal cartilages, eyebrows with a curve and thick, a long philtrum, and clefts of the orofacial region. Except for a cleft of the hard/soft palate, the present case shows patient's facial features displaying all of the OFCD syndrome's hallmarks. Patient was with normal intelligence and having hammer toe (second toe).

CONCLUSION(S)

The present case report aims to provide analysis of a patient with a congenital cataract, cardiac defect, affecting facial features, and teeth anomalies. It emphasises the diagnostic consideration, especially for dentist. With the evaluation of the patient's clinical presentation and radiographic findings, the present case report seeks to enhance our understanding of the complexities of the OFCD syndrome. Despite having easily diagnosable distinctive findings in the medical and dental anomalies, many medical and dental practitioners frequently ignore them.

REFERENCES

- [1] Hayward JR. Cuspid gigantism. *Oral Surg Oral Med Oral Pathol.* 1980;49:500-01.
- [2] Gorlin RJ, Marashi AH, Obwegeser HL. Oculo-facio-cardiodental (OFCD) syndrome. *Am J Med Genet.* 1996;63:290-92.
- [3] Ng D, Thakker N, Corcoran CM, Donnai D, Perveen R, Schneider A, et al. Oculo-facio-cardiodental and Lenz microphthalmia syndromes result from distinct classes of mutations in BCOR. *Nat Genet.* 2004;36:411-16.
- [4] Schulze BR, Horn D, Kobelt A, Tariveridian 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.
- [5] Wilkie GJ, Chambers IG. A very large maxillary cuspid. *Oral Surg Oral Med Oral Pathol.* 1990;70:159-60.
- [6] Davoody A, Chen IP, Nanda R, Uribe F, Reichenberger EJ. Oculo-facio-cardiodental syndrome: A rare case and review of the literature. *Cleft Palate Craniofac J.* 2012;49(5):e55-e60.
- [7] Feberwee HE, Feenstra I, Oberoi S, Sama IE, Ockeloen CW, Clum F, et al. Novel BCOR mutations in patients with oculo-facio-cardiodental (OFCD) syndrome. *Clin Genet.* 2014;85:194-97.
- [8] Pyeritz RE. The Marfan syndrome. *Ann Rev Med.* 2000;51(1):481-510.
- [9] Laganà G, Venza N, Paoloni V, Bertoldo F, Ruvolo G, Cozza P. A 3D geometric morphometric analysis of the palatal morphology in Marfan's syndrome: A preliminary study. *J Clin Diagn Res.* 2018;12:14-17.
- [10] Nguyen TT, Truong ATH, Hoang VA, Van Huynh D, Van Nguyen T, Le CT, et al. Oculo-facio-cardio-dental (OFCD) syndrome: A case report. *J Med Case Reports.* 2024;18(1):18. Available from: <https://doi.org/10.1186/s13256-023-04244-x>.
- [11] Martinho J, Ferreira H, Paulo S, Paula A, Marto CM, Carrilho E, et al. Oculo-facio-cardio-dental syndrome: A case report about a rare pathological condition. *Int J Environ Res Public Health.* 2019;16(6):928. Doi: 10.3390/ijerph16060928.
- [12] Zhou Y, Wojcik A, Sanders VR, Rahmani B, Kurup SP. Ocular findings in a patient with oculo-facio-cardiodental (OFCD) syndrome and a novel BCOR pathogenic variant. *Int Ophthalmol.* 2018;38(6):2677-82. Doi: 10.1007/s10792-017-0754-5.
- [13] Verma G, Singh GK, Tandon P, Verma SL. A rare syndrome with unusual dental findings: Oculo-facio-cardio-dental syndrome. *J Oral Maxillofac Pathol.* 2014;18(2):331. Doi: 10.4103/0973-029X.140928.

PARTICULARS OF CONTRIBUTORS:

1. Senior Lecturer, Department of Periodontics, Vidarbha Youth Welfare Society Dental College and Hospital, Amravati, Maharashtra, India.
2. Professor, Department of Periodontics, Datta Meghe Institute of Higher Education and Research, Sharad Pawar Dental College, Sawangi, Wardha, Maharashtra, India.
3. Professor and Head, Department of Interventional Radiology, Datta Meghe Institute of Higher Education and Research, Jawaharlal Nehru Medical College, Sawangi, Wardha, Maharashtra, India.
4. Professor and Head, Department of Cardiology, Datta Meghe Institute of Higher Education and Research, Jawaharlal Nehru Medical College, Sawangi, Wardha, Maharashtra, India.
5. Senior Lecturer, Department of Periodontics, Yogita Dental College Hospital, Khed, Maharashtra, India.

NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:

Dr. Aishwarya Rathod,
Senior Lecturer, Department of Periodontics, Vidarbha Youth Welfare Society
Dental College and Hospital, Amravati-444602, Maharashtra, India.
E-mail: aishwaryarathod55@gmail.com

AUTHOR DECLARATION:

- Financial or Other Competing Interests: None
- Was informed consent obtained from the subjects involved in the study? Yes
- For any images presented appropriate consent has been obtained from the subjects. Yes

PLAGIARISM CHECKING METHODS:

- Plagiarism X-checker: Oct 18, 2024
- Manual Googling: Jan 24, 2025
- iThenticate Software: Apr 15, 2025 (12%)

ETYMOLOGY:

Author Origin

EMENDATIONS:

9

Date of Submission: **Oct 17, 2024**

Date of Peer Review: **Nov 15, 2024**

Date of Acceptance: **Apr 17, 2025**

Date of Publishing: **Aug 01, 2025**