Ossifiyng Fibroma in a Pre-Existing Treacher Collins Sydrome Case

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ABSTRACT

Treacher Collins syndrome (TCS), or mandibulofacial dysostosis, is a congenital disorder of craniofacial development having an incidence of about 1 in 50,000 live births. The essential components of the syndrome were described in 1990 by E Treacher Collins hence the syndrome was named after him. It mostly affects the structures derived from first and second brachial arches with variable expressivity. Characterized by hypoplasia of facial bones (mandible, maxilla and cheek bone), antimongoloid slant of palpebral fissures, coloboma of the lower lid and various ear anomalies. Ossifying fibroma is a rare, destructive, deforming, slow growing, benign fibro-osseous tumor. It is frequently seen in the craniofacial bones, with the mandible being the most common site of involvement.

Keywords: Treacher Collins Syndrome (TCS), TCOF1 Gene, Mandibulofacial Dysostosis, Ossifying Fibroma.

INTRODUCTION

Treacher Collins syndrome (TCS) or Franceschetti syndrome is an autosomal dominant disorder affecting craniofacial development. The syndrome is said to involve the first and second brachial arches during the embryonic development. Early descriptions were given by Berry (1889), Treacher Collins (1900), and Franceschetti and Klein (1949), and hence the names Berry's syndrome and FranceschettiZwahlen-Klein syndrome.[1] In 1949 Franceschetti and Klein did and extensive research on the syndrome and coined the term mandibular facial dysostosis to describe the characteristic clinical features which are restricted to head and face. Some of the common features are slanting of antimongoloid palpebral features, malar and mandibular hypoplasia, colobama of lower eyelid, malformation of auricles and cleft palate.

According to the 1992 World Health Organization (WHO) classification, ossifying fibroma is defined as demarcated or rarely encapsulated neoplasm consisting of fibrous tissue containing varying amounts of mineralized material resembling bone and/or cementum. It is a benign fibro-osseous lesion of the jaw characterized by replacement of normal bone by fibrous tissue containing a newly formed mineralized product.[2]

CASE REPORT

An 18 year old male patient came to the department of oral medicine and radiology with the chief complains of discomfort and growth in upper left front teeth region of jaw since 1 month. History of growth which was initially smaller in size and gradually attained the present size, not associated with pain. Family history was not significant, patient had a set of extra oral features like accessory ear on right side of face since birth, and patient's mother revealed that patient has satisfactory intelligence level. Previous medical history disclosed surgical treatment done for accessory auricle present in front of right side of tragus and commissuoroplasty was undertaken for the closure of comissures on both right and left side which were extended to both cheek regions.

On general examination patient had normal gait, stature and was moderately built. Extra oral examination findings were recorded like antimongloid slanting of the palpebral fissures with sparse eyelashes, colobama of the lower eyelid, hypoplasia of the malar prominence,micrognathia and the maxilla appeared to be prognathic (fig1-3). Intraorally all the teeth were fully erupted except 18 28 38 48 and 33, prognathic maxilla with proclination of upper anteriors and hypoplastic mandible.

On examination of specific growth, extra oral diffuse solitary swelling was seen on left side of face which was round in shape, gradually increased in size (approximately 4×3cm) extending supero-inferiorly from the lower left eyelid to 2cm above the lower border of mandible and antero-posteriorly from left ala of the nose to 3 cm ahead of left tragus area, with well defined borders, left ala of the nose was raised, surface texture and color of overlying skin was normal,. On palpation all the inspectory findings were confirmed about size, shape, color, texture and borders, non tender and hard in consistency. Intraoral examination revealed normal mouth opening, swelling was seen extending in upper left labial vestibule from 21 to 23 region, palatally extending from gingival margin of 21 posteriorly 1cm away from junction of hard and soft palate, mesiodistally 5 mm below the gingival margin of 24 crossing the midline towards the right side of

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Figure-1-3: Extraoral features of the patient

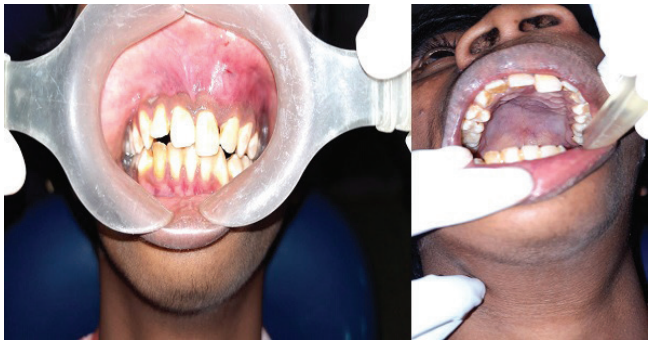

Figure-4 & 5: Intraoral features

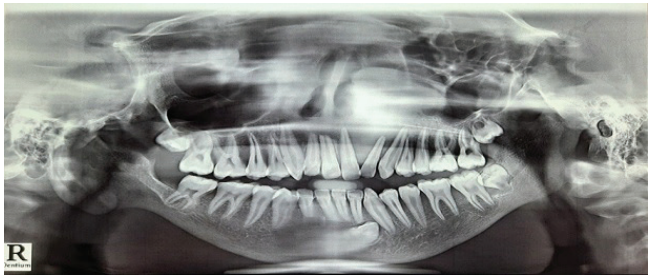

Figure-6: Panaromic radiograph

the palate approximately of size 3*2 cm. Overlying mucosa and surrounding structures were normal with well defined margins, accompanied by extrusion of 21 and displacement of 22 (fig4,5). On palpation all the inspectory findings about shape and size and borders were confirmed, non tender, hard in consistency and grade 1 mobility of 21, 22, and 23.

Investigation

Panoramic radiograph shows prominent antigonial notch & short ramus on right side of mandible, hypoplasia of the mandible, impacted 18 28 38 with only crown formation, impacted 33 and completely missing 48 were the other features seen. A well-defined homogenous radiopacity was seen extending from mesial surface of 11 extending towards the left side and involving 21 22 23 24 upto distal surface of 25approximately of size 4*3cm involving the left maxillary sinus region. Displacement and root resorption of 11 21 22 and deviation of nasal septum towards right side.

CT report's showed large ill defined cystic mass arising from maxilla involving left maxillary sinus and left nasal cavity with scalloped borders and destruction of cortex in many areas noticed.(fig-7)

Incisional biopsy of the lesion was done under local anesthesia in labial vestibular region in 22 and 23 region and sent for histopathological examination which revealed the growth as ossifying fibroma.

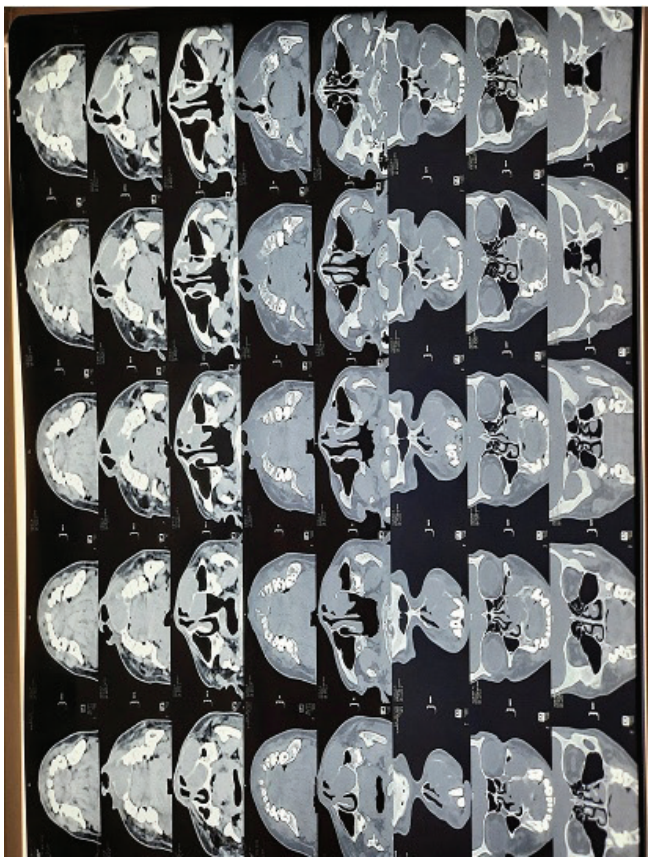

Figure-7: Axial and coronal section of CT scan

DISCUSSION

Treacher Collins Syndrome (TCS) or Mandibulofacial Dysostosis is a hereditary disorder firstly described by THOMSON in 1846; its incidence is around 1:40,000 to 1:70,000 cases per live-born children, having no difference between genders or among races. Its transmission occurs by a dominant autosomal transmission with variable expressivity is suspected. There are a 50% of chances of inheriting such condition when one of its parents presents such syndrome.[3]

The gene responsible for causing altered changes in TCS is seen in the chromosome 5 long arm (5q31-33), named as TCOF1 , which is responsible for formation of a low-complexity protein, called treacle.[4] It is suspected that the function of this protein is to act on the transportation of other proteins from the cytoplasm to the nucleus. The mutations are caused by alterations in the sequence of the gene which affects the protein which is originated by such gene, and the deletion of the exon 24 of the gene TCOF1 is the most frequent one.

The structures affected in TCS are derived from the first and second pharyngeal arch. The typical characteristics of the syndrome are 1) Antimongoloid palpebral fissures with either a notch or coloboma of the outer third of the lower lid, and occasional absence or paucity of the lashes of the lower lid. 2) Malformation of the external ear, and occasionally of the middle and inner ear, with low implantation of the auricle. 3) Hypoplasia of the facial bones, especially the malar bones and mandible 4) Macrostomia, high palate, malocclusion and abnormal position of the teeth. 5) Atypical

hair growth in the form of tongue-shaped processes of the hair-line extending.[4]

Ossifying fibroma is one of the most common inflammatory hyperplasia affecting oral cavity. In 1872, MENZEL first described ossifying fibroma, but only in 1927 MONTGOMERY assigned its terminology.[5] Ossifying fibromas are benign asymptomatic neoplasms often considered to be fibro-osseous lesions that generally have slow growth and present proliferation of fibrous cell tissue, with a varying quantity of bone products that include bone, cement or a combination of these. Ossifying fibroma having female predilection generally manifests in the third or fourth decades of life. Most common site is mandibular premolar-molar region, and about 30% of cases occur in maxilla. The tumor occurring in children has been named as juvenile aggressive ossifying fibroma, which presents at an earlier age and is more aggressive clinically and more vascular on pathologic exam.[6]

Ossifying fibromas are formed from pluripotent mesenchymal cells that originate from the periodontal ligament. These cells are capable of forming bone tissue and cementum.[7] In most cases, the lesions are radiolucent with radiopaque foci, depending on the quantity of tissue calcification, which gives rise to varying degrees of radiopacity. Bone swelling or expansion at the buccal and/or lingual cortical plates is the most frequent clinical sign of ossifying fibroma. Large size of the maxillary tumor is one of the remarkable finding at the time of diagnosis, probably attributable to the large amount of available space in the maxillary sinus into which they could expand similar to our case. Though the growth was sufficiently large, patient did not present with significant clinical symptoms as it was extending over the sinus region. The radiological appearance depends upon its maturity.[8] the most important radiographical feature of this lesion is well-circumscribed and sharply defined border. Loss of lamina dura and root resorption and/or divergence of associated teeth may be noted similar to the ones seen in our case.[9]

CONCLUSION

Early diagnosis of TCS allows prompt and appropriate treatment of aesthetic and functional deficiencies in these patients. Individuals with severe form of TCS usually undergo, over a period of time, multiple major reconstructive surgeries that are rarely fully corrective. The ossifying fibroma of the maxilla is an uncommon benign tumor leading to Cosmetic and dental occlusal problems which are often the first manifestations of these lesions as they are clinically asymptomatic. Complete surgical excision is needed to reduce the reoccurrence of such lesion.

Authors' Contributions

All authors read and approved the final manuscript.

Consent

All authors declare that written informed consent was obtained from the patients(or other approved parties) for publication of this case report and accompanying images.

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