



A Rare Case of Craniofacial Fibrous Dysplasia

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Abstract

Craniofacial fibrous dysplasia is rare form of fibrous dysplasia. It can have devastating complications depending on which ostia are involved. Here we present the rare case of craniofacial fibrous dysplasia and highlighting clinical and radiological picture including CT scan images showing exclusive involvement of craniofacial bones. There is no universal treatment option for such disease rather than wait & watch. Presented case has been managed by riseronic acid & bisphosphonate with no surgical intervention as patient's growth was not completed. Significant reduction in serum alkaline phosphatase level & reformation of buccal cortical plate defect reported with said medical management.

Keywords: *Fibrous dysplasia, monostotic, polyostotic, craniofacial.*

Introduction

Fibrous dysplasia (FD) is a non-malignant condition in which normal bone and marrow are replaced by fibrous tissue and haphazardly distributed woven bone. FD is caused by somatic activating mutations in the α subunit of the stimulatory G protein encoded by the gene guanine nucleotide-binding protein, α -stimulating activity polypeptide (GNAS 1, 20q13.2).^[1] Fibrous dysplasia (FD) has three varieties- monostotic, polyostotic, and craniofacial. Cranio-facial form of the disease occurs in 10-25 % of patients with monostotic

form & in 50% with polyostotic form. It also occurs in an isolated craniofacial form. In the isolated variety, no extracranial lesions are present. Sites of involvement most commonly include the frontal, sphenoid, maxillary, and ethmoidal bones. The occipital & temporal bones are less commonly affected.^[2] Patient reports with either great facial deformity or hampered vital function. Laboratory findings include elevated serum alkaline phosphatase but calcium, parathyroid hormone, 25 hydroxy vitamin D, 1,25- dihydroxyvitamin D levels in most cases

are normal. Malignant transformation is rare and usually precipitated by radiation therapy.^[3,4]

Treatment of this disease is non-specific and multidisciplinary approach is necessary.

The present paper reports a case of craniofacial fibrous dysplasia with its characteristic radiographic findings & wider range of cranial bones involvement.

Case Report

A 17 year old male student complained of facial asymmetry due to swelling on lower aspect of left cheek since 7 years. Swelling was almond shaped in the beginning, not associated with pain & noticeably increased in this interval. No history of trauma or infection to this area has been reported. Patient approached regional doctor 7 years back who advised him for radiological investigation like panoramic projection, lateral cephalogram and CT scan examination after which he informed about the increasing tendency of the lesion & advised to wait for surgery until 18 years. Meanwhile swelling was gradually increasing in size reached up to the size of approximately tennis ball and caused severe facial disfigurement and asymmetry, raising esthetic appeal for the patient to undergo surgical intervention for which he consulted the same doctor before 1 year. The surgical note of the doctor available explains that sub-mandibular bony mass was removed & due to major blood loss, complete removal of the lesion was not done & patient was referred patient to college of dental science and research centre, bopal, Ahmadabad for the expert opinion & needful management. During anamnesis patient denied headaches, pain to the enlarged area, fevers/night sweats, or congestion/sinus pressure, difficulty in mastication, speech or vision or any other associated complain. His voice was of normal quality, and he had full range of motion in his jaw. The patient's neck did not demonstrate thyromegaly or lymphadenopathy. He did not have any skin lesions. The patient had no other complaints, had no other significant medical history and was otherwise in good health.

On Extra Oral examination, two swellings with facial asymmetry were noted. The dome shaped swelling was extending from symphyseal area to left tragus of ear anteroposteriorly, superoinferiorly it was extended from the smile line superiorly to 2 cm below the inferior border of mandible up to the midline of neck inferiorly [figure 1]. Overlying skin appeared stretched but normal. Initial evaluation of his ears, eyes, mouth, and nose were unremarkable. This prominence involved mandibular body, antegonial notch, angle & ramus region. It was non-tender, bony hard, non-mobile & of normal body temperature. Swelling did not bleed on palpation. Another bony hard swelling also palpated from lateral side of left frontal bone to squamous part of left temporal bone involving roof of orbit, lateral wall of orbit, and lateral canthus of eye region [figure 1]. No other swelling on contra lateral side or in any other part of body reported.

On Intraoral examination, swelling was present at lower left buccal vestibule to ascending ramus region, without any overlying mucosal changes. 36, 37 were linguallly tilted. There was obliteration of lower buccal vestibule on left side [figure 2]. Swelling was bony hard in consistency. Lingual bony bulge was palpable on the medial part of body and ramus of mandible below the molars teeth from attached gingiva of molars to deep in the lingual vestibule & behind the molars area, too.

On Radiographic investigation, mandibular left lateral topographic occlusal view showed cortical expansion from left canine region to left external oblique ridge region on both buccal and lingual side [figure 3].

Panoramic view showed near complete opacification of left mandibular body, angle, ramus & bony prominence below inferior border of mandible extending from para-symphyseal region to neck of condyle. There was superior displacement of inferior alveolar canal on left side, a characteristic feature for fibrous dysplasia. [figure 4]

CT scan was performed for better characterization of the area. Mandible revealed severe expansion on both the sides with heterogenous areas of lysis & sclerosis diffusely involving the entire left side including the coronoid & condylar processes, symphysis and also extends to adjacent right anterior mandible for a distance of 1.5 cm crossing the midline. [figure 5]. There was deficiency of the buccal cortex along the infero-lateral aspect of the left body of mandible [figure 5] probably related to past surgical attempt.

Similar bony expansion with spotty sclerosis was also noted in left frontal bone, involving a narrow region anterior to the coronal suture and the entire lower half [figure 6]. The left wing of sphenoid, roof of sphenoid sinus, and both left pterygoid plates were also involved [figure 7]. Orbit revealed involvement of roof and lateral wall causing mild antero-medial displacement of intra-orbital contents [figure 8] and encroachment of the orbital fissure [figure 6]. This was a radiographic finding. Patient did not reveal any clinically observable finding related to it. Left squamous temporal bone revealed similar expansion and sclerosis [figure 9]. Left antero-inferior aspect of the occipital bone, posterior & inferior part of the left mastoid process were also abnormal [figure 10]. Bilateral maxilla, hard palate, and other bones on contralateral side were not involved.

Hematological investigations revealed serum calcium, phosphorus level and serum parathormone levels within normal range but highly elevated serum alkaline phosphatase level i.e. 525 UI/L. (reference value for adults: 20-140 UI/L, for <17 years: 0-390 UI/L)

Bone biopsy showed numerous short, irregularly shaped trabeculae of woven bone & confirmed the diagnosis of fibrous dysplasia [figure 11].

Due to highly elevated serum alkaline phosphatase level patient was referred to endocrinologist for expert opinion and to rule out any other endocrine abnormality. As patient was still in growing age, endocrinologist advised to wait for surgical intervention & gave

bisphosphonate, risedronic acid & vitamin D3 drug therapy. Patient is taking the same drugs since 1 year. Regular follow-up is going on for drug therapy efficiency on the lesion. Serum alkaline phosphatase level has significantly reduced to half after 2 months of drug therapy, and reached to 190 UI/L after 12 months of therapy. No major side effects of the drugs were reported till date.

We have obtained CT scan of existing lesion of the patient after 12 months of drug therapy and it showed reformation of buccal cortical plate defect on left side and no further increase in size or involvement of any other bone.[figure 12]

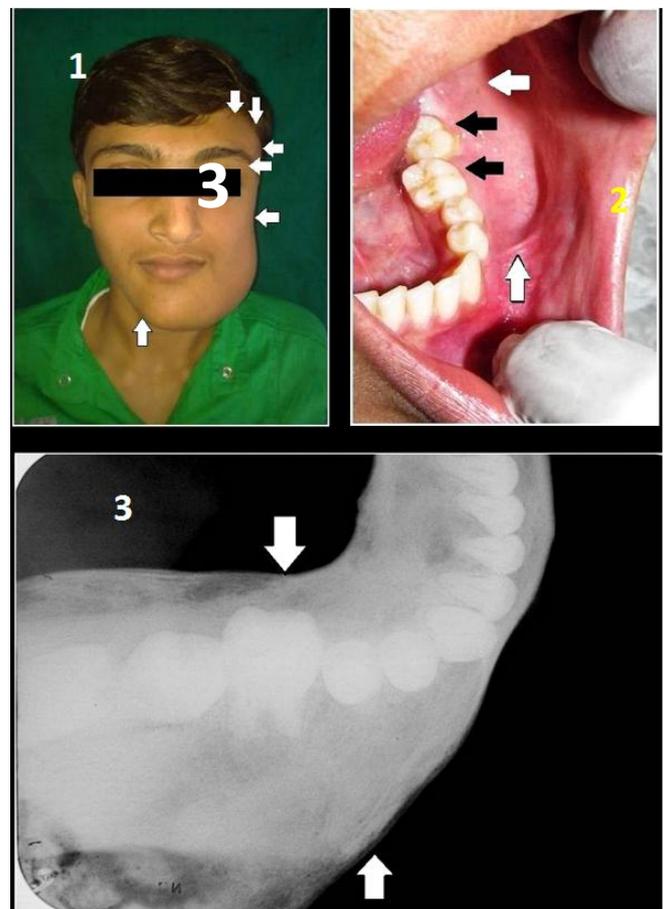


Figure 1- Extraoral extension, **Figure 2-** Intraoral extension, **Figure 3-** Bicortical expansion

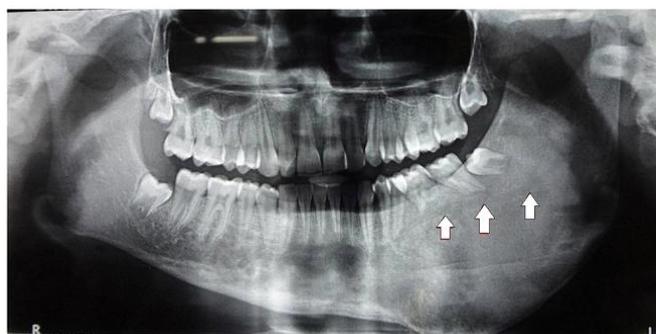


Figure 4- Superior displacement of inferior alveolar canal

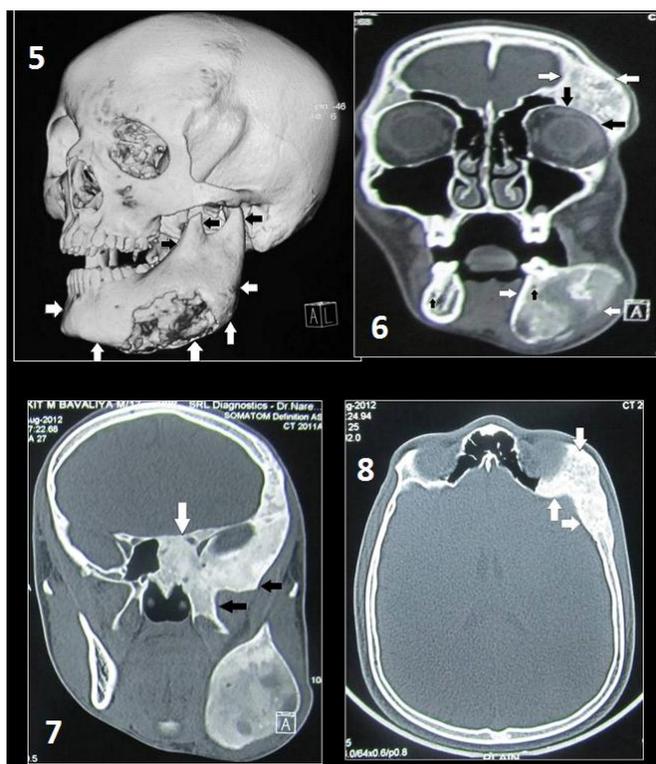


Figure 5- Buccal cortical plate defect (related to previous surgery), **Figure 6-** Involvement of orbit (black arrows) and frontal bone(white arrows), **Figure 7-** Involvement of the left wing of sphenoid, left pterygoid plates(black arrows) & roof of sphenoid sinus (white arrows), **Figure 8-** Mild antero-medial displacement of intra-orbital contents

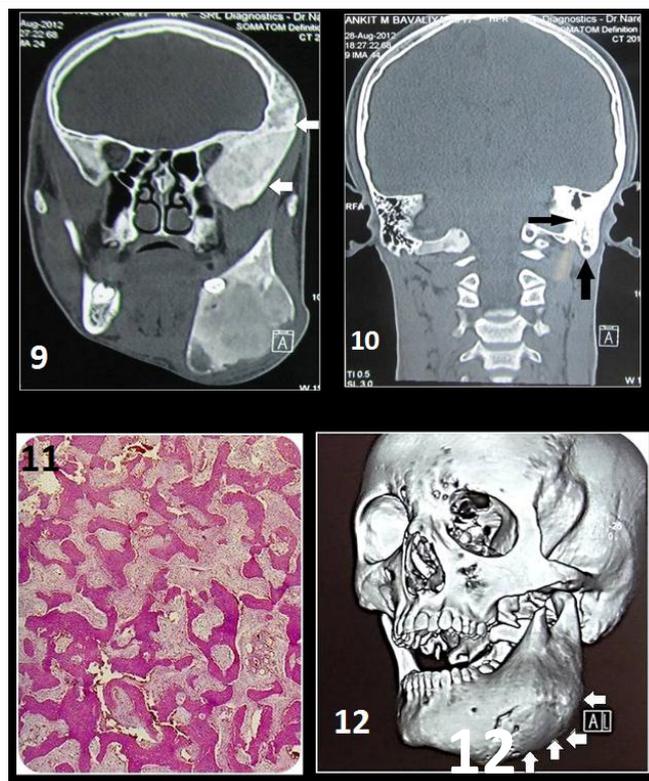


Figure 9- Involvement of left squamous temporal bone, **Figure 10-** involvement of antero-inferior aspect of occipital bone, posterior & inferior part of the mastoid, **Figure 11-** Numerous short, irregularly shaped trabeculae of woven bone, **Figure 12-**Reformation of buccal cortical plate defect.

Discussion

Fibrous Dysplasia is a disturbance of bone metabolism that is classified as a benign fibro-osseous lesion. The fibrous connective tissue containing abnormal bone replaces normal bone. [3, 5, 6]

Isolated variety of craniofacial fibrous dysplasia typically presents at around 10 years of age and then progresses throughout adolescence life with no extracranial lesions & slight female predilection.^[2,7] Identical disease course has been reported in present case except patient was male.

The craniofacial form of fibrous dysplasia can be diffuse and may involve multiple skull bones.^[8] Fibrous dysplasia of the skull most frequently presents as painless bony enlargement of part of the jaw or face in patients in the first decade of life.^[7] The clinical presentation depends on the site, duration, extent and nature of the lesion. It

ranges from a mild local swelling with little or no pain to a gross deformity with complications such as visual disturbance and sensorineural hearing loss etc. Our patient reported with complain of facial asymmetry without any other associated complain.

DA Lisle *et al* (2008) reported in his case study of 25 patients that the disease found 46% in maxilla, 34% in mandible and 20% in other cranial bones. However, in our case left mandible was completely involved and the maxilla was completely spared. He also reported that craniofacial fibrous dysplasia most commonly involves the bones of the face or the skull base, with involvement of parietal or occipital bones being relatively rare. Skull base lesions usually involve the sphenoid or temporal bones while lesions confined to the clivus are extremely rare.^[9] In the case presented here there were involvement of bones of face and skull base with involvement of anteroinferior aspect of occipital bone and clivus which are extremely rare.

In the other study McDonald-Jankowski (1999) reported the most common radiographic presentation of fibrous dysplasia as a poorly defined, ovoid (fusiform) area of dysplastic bone exhibiting a ground glass appearance.^[10] This finding was well appreciated in our case [figure 5]. The trabecular pattern appeared irregular and thickened with no discernible pattern of orientation.

Steven R. Singer *et al* (2004) reported in their case report about diffuse enlargement of the left mandible, extending from left canine area to the condyle and encompassing the inferior border, the ramus and the coronoid process.^[8] Justin Clark *et al* (2010) reported thinning of buccal cortex in the mandible.^[7] These findings coequal with present case.

Petrikowski *et al* (1995) suggested that upward displacement of the mandibular canal may be unique to fibrous dysplasia and could be pathognomonic.^[11] In the case presented here the superior displacement of mandibular canal was seen & considered pathognomonic for diagnosis.

Steven R. Singer *et al* (2004) also reported about displacement of the teeth on affected side with loss of lamina dura and narrowing of periodontal ligament space.^[8] This findings were seen in our case.

Justin Clark *et al* (2010) also reported involvement of facial and skull bones with difficulty in vision, hearing and hyperesthesia on the affected area and elevated serum alkaline phosphatase level.^[7] All the findings recorded by them match with our case except absence of any neural complain surprisingly till date inspite of involvement of many cranial bones.

Elevated serum alkaline phosphatase and urine hydroxyproline are usually noted in the active stage of the disease. These markers can also be used to track treatment progress.^[8] In our case highly elevated serum alkaline phosphatase level was noted and with the treatment of bisphosphonates, risedronic acid and vitamin D3, it has been significantly reduced to reference level. Plotkin *et al* examined 18 children and adolescents with fibrous dysplasia treated with bisphosphonates therapy and found that serum alkaline phosphatase level decreased with no serious side effects, however they noted no radiographic change or improvement but in our case cessation of progress of lesion, absence of involvement of any other new bone and reformation of existing buccal cortical plate defect has been noted.

The case described here is unique because the patient reported that the lesion had been present for more than 7 years & within this time the lesion had involved the craniofacial bones in addition to mandible.

The mnemonic FEGNOMASHIC can be helpful when considering the differential diagnosis.^[7] The present case is reported in its mature/radiopaque stage may help to exclude many of the lesions from the mnemonic, mainly describing lytic lesions.

Treatment with bisphosphonates to inhibit osteoclastic and osteoblastic activity is a medical option than surgery but it is difficult to measure

success-other than with symptoms. We reported significant reduction in serum alkaline phosphatase level after 12 months of drug therapy along with stilled progress of lesion and reformation of buccal cortical defect. Non surgical management does not carry very promising outcome though now when the genetic basis for the disease is known, more specific therapy can be targeted to achieve complete successful remission.^[7]

Conclusion

Fibrous dysplasia is significant for the dentist because it may affect the facial, cranial and jaw bones leading to many deformities and dysfunctions of orofacial area. This is a case of craniofacial FD having extensive lesions in craniomaxillofacial region. Craniofacial Fibrous Dysplasia is important to ensure the accurate diagnosis and appropriate management especially in growing age where surgery needs to be deferred.^[6] We have treated this case with risedronic acid, vitamin D3 and bisphosphonates therapy and reported significant reduction in serum alkaline phosphatase level, cessation of progress of lesion and reformation of existing buccal cortical plate defect.

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