ISSN- O: 2458 - 868X, ISSN–P: 2458 – 8687
Index Copernicus Value: 68 . 16
PubMed - National Library of Medicine - ID: 101731606

International Journal of Medical Science and Innovative Research (IJMSIR)
IJMSIR : A Medical Publication Hub
Available Online at: www.ijmsir.com
Volume – 5, Issue –2, April - 2020, Page No. : 24 - 30

A Rare Craniofacial Fibrous Dysplasia – Case Report and Review of Literature.

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Citation this Article: Dr. Sonal Madan, Dr. Deval Kale, Dr. Setu P. Shah, "A Rare Craniofacial Fibrous Dysplasia – Case Report and Review of Literature", IJMSIR- April - 2020, Vol – 5, Issue -2, P. No. 24 – 30.

Type of Publication: Case Report

Conflicts of Interest: Nil

Abstract

Craniofacial fibrous dysplasia has been regarded as a developmental disorder characterized by replacement of normal bone with benign cellular fibrous connective tissue. It is a bone tumor that, although benign, has the potential to cause significant cosmetic and functional disturbance, particularly in the craniofacial skeleton. Most patients have chief complain of painless swelling. Craniofacial fibrous dysplasia should be treated as conservatively as possible. Bisphosphonate, vit. D3 and calcium can be used to fill the lytic lesion and for the thickening of the cortices. Facial disfigurement is the main reason for the surgical intervention. Functional disturbance or involvement of optic foramen or foramen magnum can be the other reason for the surgery. Here we report complete case of 17 year old male with craniofacial fibrous dysplasia with medical and surgical management.

Keywords: Fibrous dysplasia, craniofacial, bisphosphonate, alkaline phosphatase.

Introduction

Fibrous dysplasia is a non-malignant condition in which normal bone and marrow are replaced by fibrous tissue and haphazardly distributed woven bone. Fibrous dysplasia 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). Fibrous dysplasia has four varieties - monostotic, polyostotic, craniofacial and cherubism. Craniofacial form of the disease occurs in 10-25% of patients with monostotic form & in 50% with polystotic 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.[1] 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- dihydroxy vitamin D levels in most cases are normal. Malignant transformation is rare and usually precipitated by radiation therapy. Treatment of this disease is non-specific and multidisciplinary approach is necessary.[2] Here we report the case of craniofacial fibrous dysplasia with its clinical and radiographic findings alongwith its medical and surgical management.

Case report

In 2012, 17 year old patient reported to department of oral and maxillofacial surgery with the chief complain of swelling on left side of the face in the lower jaw region since last 7 years. Patient was relatively asymptomatic before 7 years. He noticed a bony hard, painless swelling over the angle of mandible on left side of face which has gradually increased in size. He had reported to dental surgeon at Bhavnagar in 2005 and being under age, was kept under observation. In June 2012 he had reported to oral and maxillofacial surgeon in Rajkot. Surgeon had removed bone from the inferior border & from the facial surface of mandible on left side under general anesthesia by extra-oral approach. While operating he had encountered severe bleeding and soft mass inside the bone so he stopped his procedure & closed the operated area. By histopathological examination of the partially removed lesion, it was diagnosed as fibrous dysplasia. Patient was referred to College of dental science and research centre, Ahmedabad.

Patient was not having past medical history. On general examination, patient was moderately built, well conscious, well oriented to time, place and person. No signs of pallor, icterus, clubbing, cyanosis or lymphadenopathy.

Extra-oral examination revealed two swellings with gross facial asymmetry. The 15 x 10 cm 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. Overlying skin appeared stretched but normal. Initial evaluation of his ears, eyes, mouth, and nose were unremarkable. The swelling was involving mandibular body, antegonial notch, angle & ramus region. It was bony hard, non-tender, 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. No other swelling on contra lateral side or in any other part of body reported. 5 cm linear operative scar was noted in submandibular region. There was no paraesthesia on left side of lower lip.(Fig. 1)

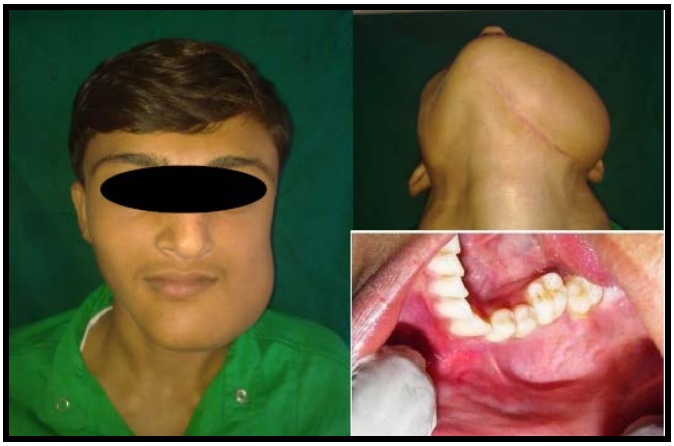

Fig. 1: Extraoral and intraoral clinical view.

On Intraoral examination, swelling was present at lower left buccal vestibule to ascending ramus region, without any overlying mucosal changes. 36, 37 were lingually tilted. There was obliteration of lower buccal vestibule on left side. 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 and behind the molars area, too. Clinically all tooth were present except all third molars. Mandibular left 1st molar was tilted lingually.(Fig. 1)

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. Panoramic view showed near complete opacification of left mandibular body, angle, ramus & bony prominence below inferior border of mandible extending from parasymphyseal region to neck of condyle. There was superior displacement of inferior alveolar canal on left side, a characteristic feature for fibrous dysplasia. Lingual tilting of the mandibular left first molar was also seen.(Fig. 2)

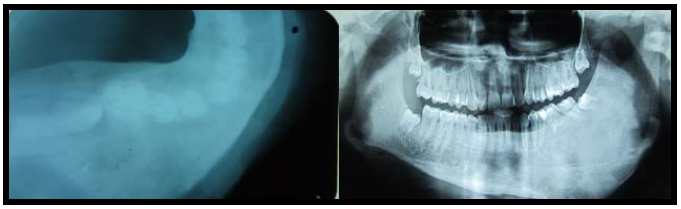

Fig. 2: Occlusal and panoramic radiograph

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. There was deficiency of the buccal cortex along the inferolateral aspect of the left body of mandible 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. The left wing of sphenoid, roof of sphenoid sinus, and both left pterygoid plates were also involved. Orbit revealed involvement of roof and lateral wall causing mild anteromedial displacement of intraorbital contents and encroachment of the orbital fissure. 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. Left anteroinferior aspect of the occipital bone, posterior and inferior part of the left mastoid process were also abnormal. Bilateral maxilla, hard palate and other bones on contralateral side were not involved.(Fig. 3)

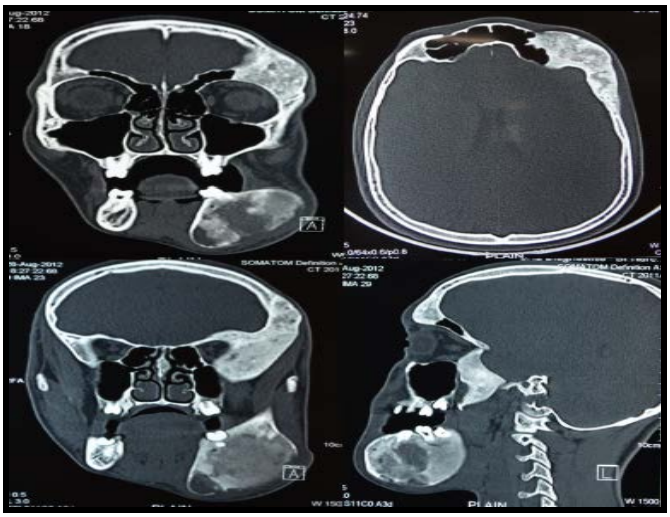

Fig. 3: Preoperative C.T. scan showing involvement of cranial bones.

All pre-operative haematological investigations were advised. Additional investigations including serum calcium, serum phosphorous, parathyroid hormone level (PTH) and serum alkaline phosphatase (Serum ALP) level were advised due to previously diagnosed fibro-osseous lesion. All reports were within normal limits expect serum ALP level which was 580 IU/L. (Normal range 20-140 IU/L) We referred the patient to endocrinologist. As the patient was below maturation age with higher serum ALP level and with pre-diagnosed fibrous dysplasia case, we decided to start the medical management first. And to also monitor

serum ALP level for next one year till the maturation age.

No.	Drug name	Group	Dosage
1.	Risedronate – 35 mg	3rd gen. Bisphosphonate	1 tab/week
2.	Calcitriol – 0.25 mcg	Bioactive form of Vit. D3	2 cap/week
3.	Chelocalciferol (1000 IU) + Calcium carbonate (500 mg)	Vit D3	1 tab everyday at bedtime
4.	Cholecalciferol (60,000 IU)	Vit D3	1 tab/15 days

Month and year	Level of serum ALP
August 2012	580 UI/L
November 2012	249 UI/L
January 2013	229 UI/L
March 2013	212 UI/L
August 2013	221 UI/L
October 2013	190 UI/L

Medical management was continued for 1 year. During that period serum ALP level was continuously monitored to see the bone activity. In october 2013, when lower serum ALP level was achieved and patient was of 18 years at that time. We again advise for the C.T. scan. On comparison of the both C.T. scan significant calcification was observed over the 1 year period.(Fig. 4) So we decided to go for the surgical management alongwith the continuous medical management with the consent from the endocrinologist.

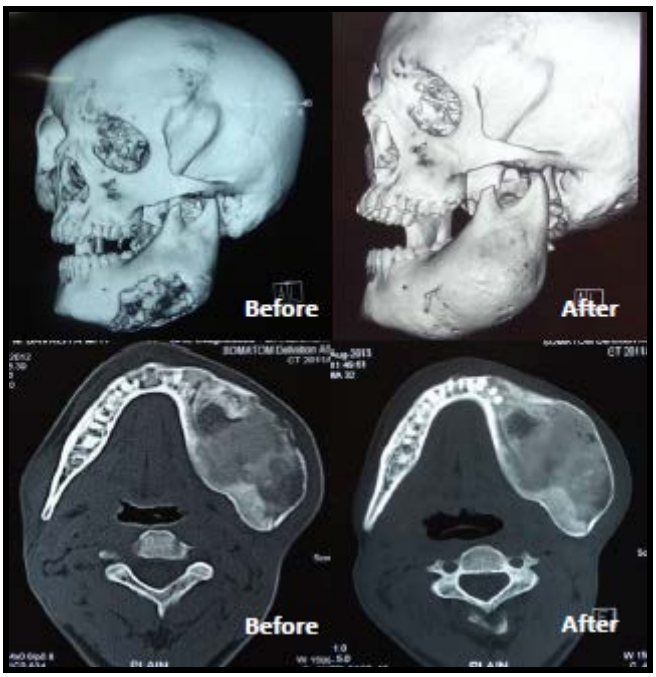

Fig. 4: Comparison of C.T. scans before and after medical management

As patient complain was only facial asymmetry, we opted upon to do the surgical procedure only on mandible. For the rest of the bone we decided to observe for the further progress or any further complain if any. Extra-oral submandibular approach was preferred, as to hide the future scar line in the skin crease below the mandible and to revise the dirty scar of the previous surgery. After reflection of the flap, bone shaving was done in piecemeal manner to prevent fracture of the mandible. By checking facia symmetry required bone was removed from the facial surface and from the inferior border of the mandible. As due to 1 year medical management, less and controllable bleeding was observed as compared to severe and uncontrolled bleeding for which surgical procedure was terminated. Layerwise suturing was done. Skin sutures were removed after 10 days.

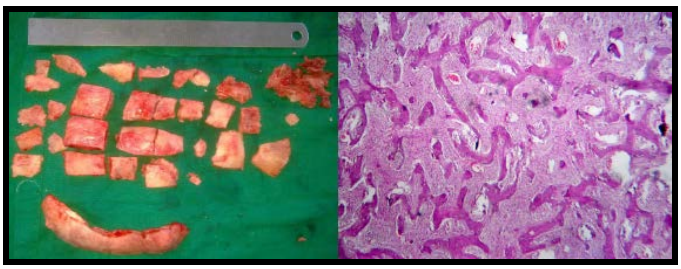

Fig. 5: Specimen and histopathological examination. (4x magnification showing Chinese letter appearance)

By histopathological examination of the specimen after surgery, diagnosis of craniofacial fibrous dysplasia was confimed. Sebsequent every year serum ALP & serum calcium levels were observed, which were normal since last 7 years. No subsequent complain regarding the fibrous dysplasia of the other cranial bones.(Fig. 5)

Discussion

Fibrous dysplasia was first reliably reported one century ago by von Recklinghausen, where he had described patients with a pathologic condition of the bone characterized by deformity and fibrotic changes that he termed as osteitis fibrosa generalisata. In 1938, Lichtenstein and Jaffe first introduced the term fibrous dysplasia. They had also noted that fibrous dysplasia of the bone is a condition that can affect single as well as multiple bone. The relationship between lesions and endocrinopathy as recognised by McCune and Albright et al. during the same period and triad of polyostotic fibrous dysplasia, precocious puberty and area of cutaneous pigmentation (café-eu-lait spots) is now known as McCune Albright syndrome. Since then, large number of cases have been reported and considerable advances have been made in the understanding and treatment of the disease.[3]

Fibrous dysplasia is often a deforming and devastating condition that begins in childhood. It is as uncommon, nonfamillial congenital disorder of bone that is found equally in both sexes. It accounts for about 2.5% of all bone tumors and 7.5% of all benign neoplasm.[3]

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. 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. 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. 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.[4]

DA Lisle et al. had reported 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. 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.[4]

There is some role for biochemical markers in management of craniofacial fibrous dysplasia. Serum ALP and urinary hydroxyproline are examples of useful

markers and are used to monitor response in the non-surgical management of the disease rather than diagnosis. They are also indicative of active stage of the disease.[5] In our case we continuously monitoring serum ALP level throughout medical management. The one of the reason to perform the surgery after control on the level of serum ALP was, to avoid surgery in active phase of the disease.

There are no long term studies of the natural history of craniofacial fibrous dysplasia but as can be seen in the appendicular skeleton, progression/expansion of craniofacial disease also has a tendency to slow or stop after skeletal maturation. Growth hormone excess is the single aspect of the disease that is associated with the craniofacial morbidities like vision and hearing loss.[6] This was the other reason to perform the surgery after completion of the maturation age.

As suggested, if the disease involves mandible with gross facial asymmetry, than the treatment should be conservative excision. For the other cranial bones the treatment should be medical management and observation. No surgery should be considered for the other cranial bones, unless symptomatic.[3] In our case we have followed the same principle.

Bisphosphonates (previously referred as bisphosphonates or diphosphonates) are a group of agents which are analogues to pyrophosphates. They are absorbed onto hydroxyapatite crystals in bone mineral and because their structure renders them resistant to enzyme degradation, they act principally by inhibiting bone resorption with some effect on bone formation. Alongwith calcium administration, net effect is to promote bone mineral accretion while the same time reducing bone turnover. Ultimately this increases bone density & bone mineralisation occurs more rapidly. Bisphosphonates are well tolerated over the longer period.[7] The above mechanism in our case was observed by the comparison of the C.T. scan before and after the medical therapy. Also increased density and calcification helped us with drastic reduction in the blood loss compared to previously terminated procedure.

The risk of developing sarcoma is 400 times higher in patients who have been treated previously with radiation tan in non-radiated patients. Radiation therapy should not be used to treat fibrous dysplasia.[8]

Conclusion

Craniofacial fibrous dysplasia is a benign disease that has the potential to cause significant cosmetic and functional disturbance. Lesional cells are committed osteogenic precursor cells with impaired capacity to differentiate into normal functioning osteoblast. The defects in osteoblast differentiation is associated with Gsα mutation of both neural crest and mesoderm derived osteogenic cells and may thus affect any part of the osteogenic compartment. It is a disease that functionally and aesthetically cripples the affected person. Much progress has been made over the past decade especially for the identification of genetic mutation linked to the etiology of the disease. With proper understanding, diagnosis and management, however good outcome can often be achieved.

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