Case Report

A rare case report of craniofacial fibrous dysplasia

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Abstract

Fibrous dysplasia (FD) is a fibro-osseous lesion of the osseous structures of the body. The exact cause is unknown; however, recently, the cause has been reported to be postzygomatic somatic mutation in guanine nucleotide-binding protein, alpha stimulating 1 gene located at chromosome 20q13.2. The three subtypes of FD are monostotic, polyostotic and craniofacial. The term craniofacial FD (CFD) is used to describe FD where the lesions are confined to contiguous bones of the craniofacial skeleton. This report describes the case of CFD of a 20-year-old male patient who had unusual presentation involving right maxilla and frontal bone of the left side of the face. The clinical features, radiological findings and treatment have been discussed.

Keywords: Craniofacial fibrous dysplasia, frontal, maxilla

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INTRODUCTION

Fibrous dysplasia (FD) is an uncommon nonhereditary, skeletal developmental anomaly where normal bone is replaced by an excessive proliferation of cellular fibrous connective tissue intermixed with irregular bony trabeculae. It may arise as a single lesion referred to as monostotic or can occur with multiple lesions that affect many bones also known as polyostotic. A small set of polyostotic FD can also occur as a component of a multisystem developmental disorder known as McCune-Albright syndrome that is also associated with endocrine hyperfunction and cafe au lait cutaneous macules.[1] With an incidence of 1:4000–1:10,000, it seems to be a rare disease.[2] Craniofacial FD (CFD) affects the bones of the craniofacial complex, including the mandible and maxilla, cranial base and vault. It is one of the three types of FD.[3] FD of bone evolve from activating missense mutations in Gs alpha gene in pluripotent embryonic stem cells. The inheritance of these mutations remains in a population of postnatal skeletal stem cells or mesenchymal stem cells which direct the formation of atypical bone in FD.[4] These conditions have a slight female predilection.[2] The bones commonly involved are maxilla (12%) and mandible (12%), involvement of the ethmoid, sphenoid, frontal and temporal bones are infrequent. The affected bones show expansion, thickening and sclerosis. Depending on the involved bone patients may have visual abnormalities, hearing disturbances, facial asymmetry and tooth displacement.[5] We report a case of CFD in a 20-year-old male patient.

CASE REPORT

A 20-year-old male patient presented with a painless swelling of the right maxilla and frontal bone of the left side of the face for the past 2 years [Figure 1]. Clinically,

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the patient presented with facial asymmetry. There was no history of trauma, trismus, diminished vision, loosening of teeth or epistaxis. Extraoral examination revealed well-defined bony hard swelling in the right maxillary region. There was involvement of the left frontal bone which showed the prominence in that region. Intraorally, there was enlargement of the right side of palate and expansion of alveolar buccal plate was present extending from 11 to 17 [Figure 2]. There was no swelling present elsewhere in the body, and café-au-lait spots were absent. Routine investigations such as hemogram, serum calcium and serum alkaline phosphatase (ALP) were performed. All parameters were within normal limits except ALP. ALP was raised to 300 U/L. The computed tomography (CT) scan showed a radiodense mass with ground-glass appearance involving left frontal and right maxillary bone and its expansion causing facial asymmetry [Figure 3]. CT scan showed thickening of the frontal region with loss of anterior wall of frontal sinus and dense ossifications of the maxilla and its antrum [Figure 4]. An incisional biopsy was obtained from the lesion in the maxilla and histopathological analysis was done. Macroscopy showed two pieces of bony hard-tissue greyish white in color measuring 1 cm × 0.5 cm and 0.5 cm × 0.5 cm approximately [Figure 5]. Microscopic examination showed irregular bony trabeculae in Chinese script pattern scattered within fibrous stroma [Figure 6]. Bony trabeculae showed no osteoblastic rimming [Figure 7]. Poorly formed metaplastic bone was separated by cellular fibrous connective tissue stroma. The bone appeared woven rather than lamellar. Based on the clinical history, radiographic assessment and histological features of the lesion, a diagnosis of craniofacial FD was deduced. Surgical recontouring was restricted only to maxilla. Contour correction of the alveolar bone was performed using stainless steel drill and soft-tissue reduction was also performed [Figure 8]. No treatment of frontal bone was done because there were no ocular symptoms and patient was unwilling too.

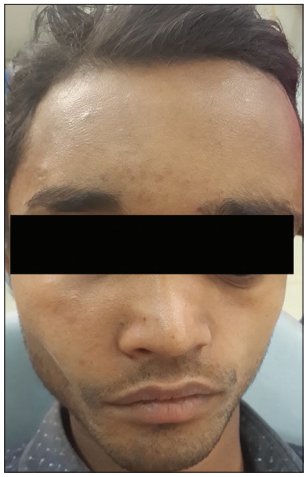

Figure 1: Clinical appearance of the patient

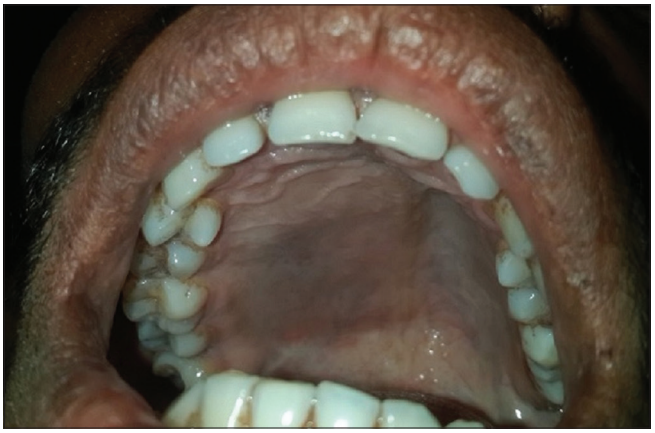

Figure 2: Enlargement of the right side of the palate and expanded alveolar plate extending from 11 to 17

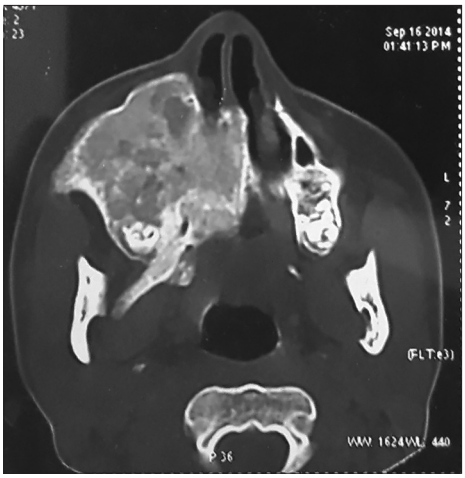

Figure 3: Axial computed tomography image shows increased dimensions of the zygoma and obliteration of the right maxillary sinus

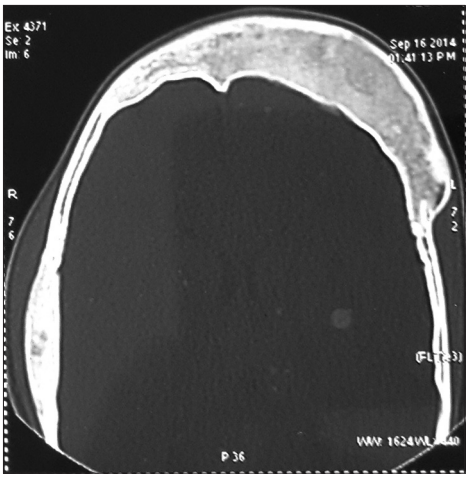

Figure 4: Axial computed tomography image shows expansion of left frontal bone

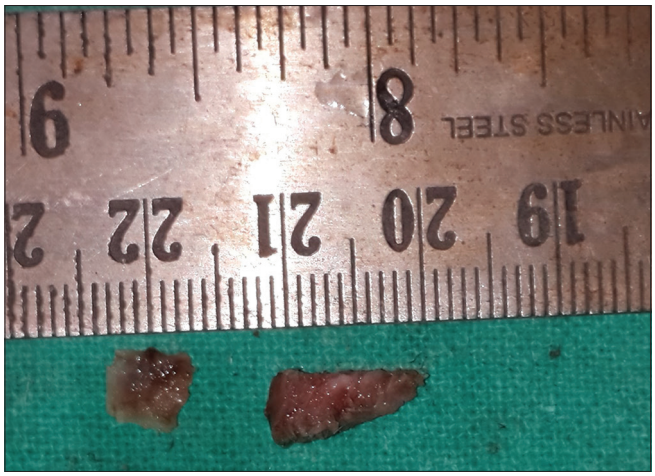

Figure 5: Incisional biopsy specimen showing two bits of hard tissue

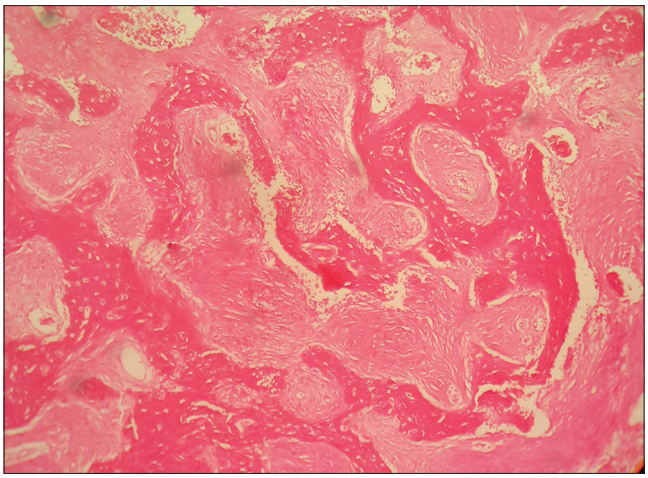

Figure 6: Decalcified section showing C-shaped bony trabeculae (H&E, ×100)

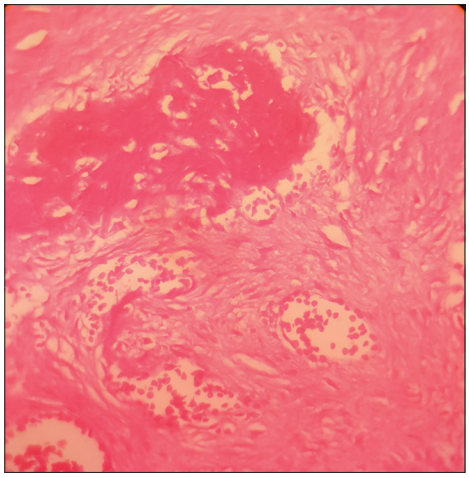

Figure 7: Decalcified section showing no osteoblastic rimming (H&E, ×400)

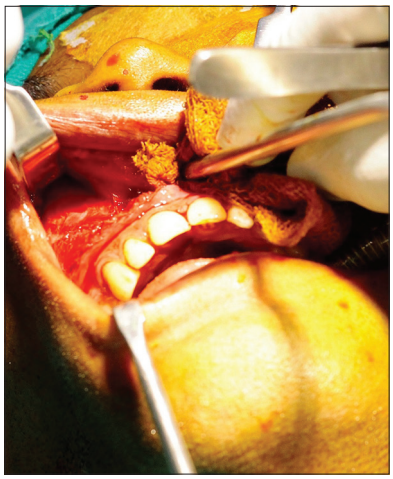

Figure 8: Surgical recontouring of the maxilla

DISCUSSION

FD is a fibro-osseous lesion characterized by developmental hamartoma by blending of fibrous and osseous tissue, with resultant secondary bony metaplasia, without osteoblast maturation producing immature, newly formed and weakly calcified bone. It can be monostotic or polyostotic. The craniofacial bones are affected in 10%–25% of cases in monostotic forms and in 50% of cases in polyostotic forms. FD essentially affects children and young adult[6] such as in the present case, a 20-year-old male. CFD is a benign, slowly progressive bone disorder in which normal craniofacial bones are replaced by fibrous tissue in which secondary metaplastic bone formation occurs.[7] FD is relatively rare in the craniofacial region, (only 20% of all locations).[8] Involvement of frontal, sphenoid, nasoethmoid and maxillary bones may result in nasal obstruction, sinus obliteration mainly frontal and maxillary sinus and subsequent sinusitis. Other features associated with CFD are dystopia, dysesthesias in the distribution of the trigeminal nerve, epiphora and also headaches.[9] Latest researchers suggest that the activated G-protein Wnt/B-catenin signaling pathway is involved in modulation of bone formation. Patients with activating guanine nucleotide-binding protein, alpha stimulating mutations specifically showed activated Wnt/B-catenin signaling. The typical radiographic feature of FD is a radiolucent, hazy or ground-glass, pattern. The patterns are due to defective mineralization of immature abnormal bone and it is usually different from the radiographic appearance of normal bone.[1] There are three types of computed tomography (CT images described as ground glass (56%), homogeneous dense (sclerotic) (23%) and radiolucent (cystic) (21%). These findings are characteristic of FD.[10] Magnetic resonance imaging (MRI) also may help in assessing cranial nerve involvement and soft-tissue structures adjacent to the lesion. Bone scintigraphy is usually recommended to rule out the polyostotic variant of FD.[11] On MRI, FD

shows homogenous, moderately low signal intensity on T1-weighted images. On T2-weighted images, the tissue usually exhibits very high signal intensity. After intravenous Gd-diethylenetriaminepentaacetic acid, lesions display moderate to significant central contrast enhancement with some rim enhancement. The degree of contrast enhancement on T1-weighted images depends on amount and degree of bone trabeculae and collagen present. Both CT and MRI are excellent imaging modalities in defining the compressive effect of CFD on the orbit, optic canals and adjacent paranasal sinuses.[5] Serum ALP is significantly high in FD. Elevated Serum ALP is usually a reliable marker for predicting the prognosis of patients with FD.[12] There is no specific treatment exists for FD. Radical resection is the only technique to obtain complete resolution of FD. Wait-and-see is indicated in cases of stable lesions and ceases to grow once the patient reaches puberty. Reconstructive techniques allow obtaining adequate aesthetical and functional results. Aggressive lesions are treated by radical resection, except in pediatric patients with residual large defects in which it can be acceptable to try to resolve symptoms through bone shaving, reserving more aggressive treatments in cases of relapse or after skeletal maturity.[13] Bisphosphonates are often used as medical treatment as they may reduce the increased bone resorption. They may alleviate bone pain in FD; however, the effect of medical therapy on skeletal destruction is not clear.[14] To conclude, most of the cases of FD can be treated by conservative recontouring. The surgical procedure is indicated only after active growth phase of bones and a follow-up should be done to check for flare-ups.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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