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Case Report

A Long-Standing Fibrous Dysplasia of the Maxilla - Case Report

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Abstract

Fibrous dysplasia is a benign fibro-osseous lesion that is characterized by the replacement of bone with fibrous tissue. It predominantly presents as monostotic form involving one bone or polyostotic variant involving multiple bones. At times, extra skeletal features like endocrine abnormalities and hyperpigmented skin lesions are also observed. The confirmatory diagnosis is made through histopathology and surgery is the treatment of choice, but will depend on the duration, extent and severity of the lesion. Here we are presenting a case report on extensive Fibrous dysplasia of the maxilla of 10 years duration in a 17-year-old boy.

Keywords: Fibrous Dysplasia; Fibro-osseous Lesion; Craniofacial Pathology

Introduction

Fibrous Dysplasia (FD) is a rare benign fibro-osseous lesion that can present as a monostotic form (affecting a single bone) or polyostotic form (affecting multiple bones) [1]. Fibrous dysplasia is non-hereditary and causes the replacement of normal bone tissue by fibrous tissue, within which metaplastic bone formation takes place [2]. FD is attributed to postzygotic somatic mutation in GNAS1 (guanine nucleotide-binding protein, alpha-stimulating activity peptide 1) gene which encodes for G-protein which in turn is involved in the production of cAMP in many tissues including endocrine function, melanocyte activity, and maturation of osteoblasts. The severity of the disease depends on how early the mutations occur; the earlier the mutations, widespread are the manifestations [3]. What makes the diagnosis of this disease particularly challenging is the sporadic nature of the disease, which shows no identifiable clinical pattern [4]. Here we are discussing a case of long-standing Fibrous dysplasia of the maxilla, also involving the palate, alveolar bone, pterygoid plates, floor of the orbit, maxillary, ethmoid and sphenoid sinuses.

Case Report

A 17-year-old male reported with the chief complaint of swelling on the left side of the face for the past 10 years. The swelling was first noticed 10 years back and was diagnosed as fibrous dysplasia. The patient was posted for surgery at that time which was later postponed till he was 18 years old to assess the growth of the lesion. The patient was asymptomatic.

On examination, a swelling on the left side of the face with ill-defined borders, extending on the entire cheek region, involving the left lateral aspect of the nose with obliteration of the nasolabial fold was noted (Figure 1). Intraorally, an extensive swelling was noted on the buccal aspect of the second quadrant, extending from central incisor to the second molar. The involved teeth were displaced inferiorly and prominent spacing could be noted in the anterior region (Figure 2). No palatal changes were elicited. The surface of the swelling appeared smooth. On palpation, the swelling was bony hard and non-tender. The alkaline phosphatase levels were found to be 810 IU/ml.

Radiographically the lesion appeared as an expansile growth with ground glass opacity and few areas of central hypodensity involving left maxillary sinus, ethmoid sinus, sphenoid sinus, floor of the orbit, pterygoid plates, palate and alveolar bone (Figure 3,4).

Figure 1: Extraoral clinical photograph showing the lesion on the left maxilla.

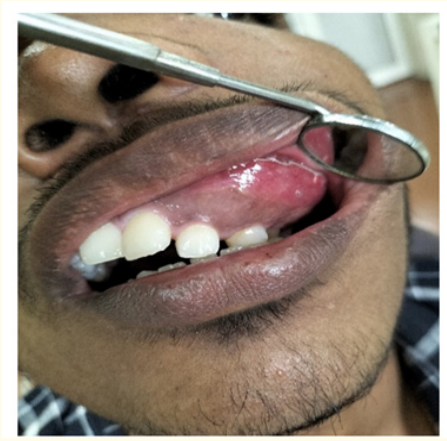

Figure 2: Intraoral clinical photograph showing expansion of the alveolar bone.

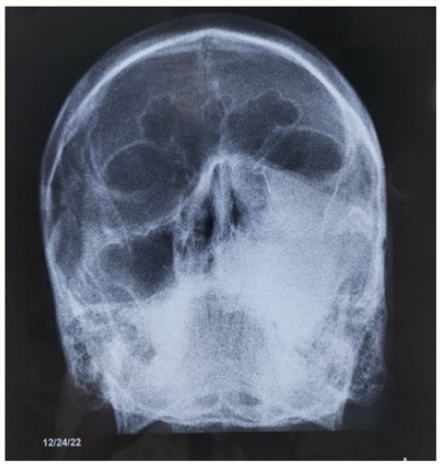

Figure 3: Warers view of the lesion.

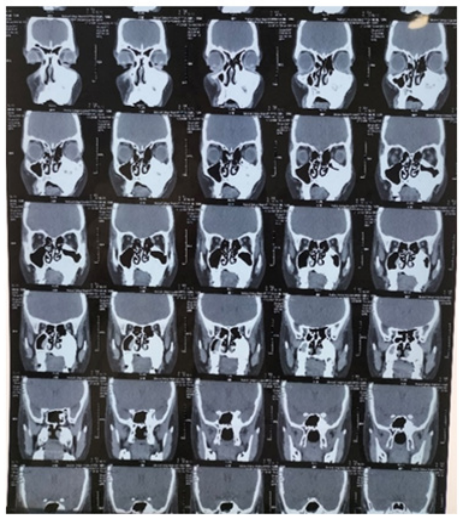

Figure 4: CT image of the lesion.

Histopathology revealed serial sections of H&E stained multiple decalcified hard tissue bits showing a fibrillar connective tissue stroma of moderate cellularity within which numerous trabeculae of mature lamellar bone were seen. Most of the trabeculae were interconnected, whereas few trabeculae were individual. The trabeculae were showing numerous osteocytes and lacked osteoblastic rimming. Peri-trabecular clefting was noted around most of the trabeculae. The inflammatory cell infiltrate was minimal (Figure 5). Correlating the clinical and radiographic findings, histopathological features were suggestive of Fibrous Dysplasia. The patient underwent a recontouring surgery, without complete excision because of the extensive involvement of multiple craniofacial bones. After the surgery, there was significant improvement in the appearance of the patient, but further surgical interventions are warranted considering the sporadic nature of the lesion.

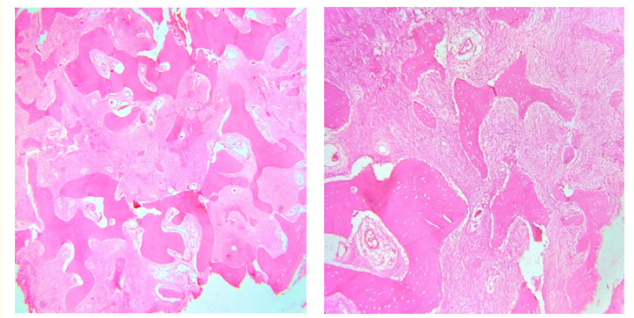

Figure 5: Photomicrographs of the lesion (10x, H&E).

Discussion

Fibrous Dysplasia is a developmental abnormality that is characterized by the replacement of bone by fibrous tissue with secondary metaplastic bone formation. Activating missense mutation of the GNAS gene cause fibrous dysplasia of bone and when more broadly distributed involves, endocrine organs, the melanocyte system, the liver, and likely a broader range of tissues [5]. Occurrence of the lesion in a single bone is termed as Monostotic FD, which accounts for about 80% to 85% of all cases. This form most frequently occurs in the rib (28%), femur (23%), tibia, craniofacial bones (10%–25%) [3]. Even though the mutation can occur from infancy, the disease is diagnosed more commonly in the second decade of life with no apparent gender predilection. The clinical presentation is a painless, slow growing tumour or could show rapid growth at times. Among jaw bones, maxilla is involved more often than the mandible. When involving the maxilla, adjacent bones like spenoid, zygoma, palatal bone are also involved in most of the cases and thus the term craniofacial fibrous dysplasia is more apt rather than monostotic FD [6]. When multiple bones are involved, the term polyostotic FD (20-30% of the cases) is used. Femur, tibia, pelvis, ribs, skull and facial bones, are commonly affected. The symptoms of polyostotic FD is more severe compared to monostotic form, including severe bone pain and spontaneous fractures [3]. Polyostotic fibrous dysplasia is associated with a number of syndromes, the most common being the McCune–Albright syndrome. It involves the triad of skeletal changes, abnormal cutaneous pigmentations called the café-au-lait spots and endocrine disorders [7].

In the radiograph, FD appear as an area of radiolucent ground glass matrix. Cortical thinning may be present due to the expansile lesion but periosteal reaction is not usually seen. The expansile remodeling of a long standing lesion may show thick layer of sclerotic border known as the rind sign [8]. Presence of significant amount of calcifications and osseous tissue may present as sclerotic or mixed lesions [9].

Histopathology will show a moderately cellular fibrous stroma within which irregular, curvilinear trabeculae of individual woven bone are seen, classically called the 'Chinese letter pattern'. Long-standing lesions may show mature interconnected lamellar bone as seen in our case. Osteoblastic rimming is not typically seen surrounding these boney trabeculae. The lesional tissue is seen to merge with the surrounding bone, making it difficult to distinguish from the normal bone radiographically [10]. The stroma appears to consist of multiple cell populations, few being fibroblastic stellate shaped stromal cells, few osteoblasts, osteocytes, osteoclast like giant cells and some immune cells [11].

Differential diagnosis of FD includes ossifying and non-ossifying fibroma, aneurysmal bone cyst, fractured callus, adamantinoma, giant cell tumor, and low-grade osteosarcoma. Histopathology is usually diagnostic [3]. Patients with fibrous dysplasia are usually managed with the 'wait and watch policy' and treatment is deferred till growth completion, as done for our patient. Most lesions are self-limiting and only cosmetic procedures are required such as recontouring, however, the postoperative regrowth of lesions are seen in up to 68% of cases [11]. But the occurrence of complications like optic nerve compression and blindness warrant immediate intervention [10]. Many studies are emerging on the use of Bisphosphonates for the non- surgical management of FD, as it inhibits osteoclastic resorption, reduces pain and strengthens bone [12]. In more severe cases complete excision with graft reconstruction may be possible. Useful biomarkers such as serum alkaline phosphatase and urinary hydroxyproline can be used to monitor response in the nonsurgical treatment of the disease [13]. Management of polyostotic variant of FD is challenging and require a multi- disciplinary approach. Malignant transformation is estimated to be 0.4% in monostotic fibrous dysplasia and 4% in McCune–Albright syndrome [3].

Conclusion

Fibrous dysplasia is a rare but challenging fibro- osseous lesion affecting primarily the skeletal structures. They are of two types; the monostotic FD and polyostotic FD. The craniofacial FD is a term used when multiple craniofacial bones are involved, as seen in this case report. Here we discussed a case of FD affecting the maxilla, maxillary sinus, ethmoid sinus, sphenoid sinus, floor of the orbit, pterygoid plates, palate and alveolar bone. Confirmatory diagnosis was made using histopathology and our patient did not show any endocrinopathies or skin pigmentations. A partial recontouring surgery was done. The patient is being reviewed continuously and will require further cosmetic procedures.

Conflict of Interest

No conflict of interest.

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