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A Rare Case Of Monostotic Craniofacial Fibrous Dysplasia In A 8-Year-Old Female- Case Report

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Abstract:
Craniofacial fibrous dysplasia is an uncommon skeletal disorder characterized by the replacement of normal bone with fibrous tissue. This report presents a rare case of monostotic craniofacial fibrous dysplasia in an 8-year-old female patient. The child exhibited symptoms including facial asymmetry and swelling, which prompted further investigation. Diagnostic imaging, including X-rays and CT scans, revealed the extent of the osseous involvement, confirming the diagnosis. Surgical intervention was performed to alleviate symptoms and prevent further complications. This case highlights the importance of early diagnosis and management of craniofacial fibrous dysplasia, especially in pediatric patients, to ensure optimal outcomes.

Keywords: Craniofacial fibrous dysplasia, skeletal disorder, fibrous tissue.

Introduction:

Fibrous dysplasia is an uncommon bone disorder that is not inherited, where normal bone tissue is replaced by fibrous tissue, resulting in bones that are weak and prone to deformity [1]. The condition arises from a postzygotic mutation in the GNAS gene, which causes continuous activation of the Gs alpha protein and disrupts normal bone development through altered signaling pathways [2]. Fibrous dysplasia can manifest as monostotic, involving only one bone, or polyostotic, affecting multiple bones, and may be linked to other syndromes like McCune-Albright syndrome[3]. Symptoms can vary widely, from asymptomatic lesions to pain, fractures, deformities, and impaired function, depending on the disease's location and severity [4]. Diagnosis is usually made through radiographic imaging, which typically reveals a "ground glass" appearance, and is confirmed by histopathological analysis [5]. Treatment primarily aims to manage symptoms, including pain relief, fracture prevention, and surgical correction of severe deformities, as no cure currently exists [6]. Ongoing research is focused on developing targeted therapies that address the disease's molecular basis to improve patient outcomes.

CASE REPORT:

A 8 year old Female patient reported to the department of Cranio- Facial Surgery with the complaint of swelling over the right cheek region for 8 months. Upon eliciting the history patient gives a history of swelling which started eight months back and currently reached to this size. No history of pain and discomfort, but aesthetics was her primary concern. Patient was non diabetic, non hypertensive with no underlying comorbidities and not using any medications. Upon clinical extraoral examination the swelling was about 5 x 4 cm in size oval in shape located 2 cm from the corner of the mouth, 1.5cms below the lateral canthus of the eye and 1cm in front to the tragus of the ear (Figure 1 and 2). No visible pulsation was noted. Asymmetry of the face was clearly noted over the right side of the face. Upon palpation the swelling was hard in consistency the skin over the swelling was normal and pinchable. Apart from the local examination the whole body was examined and looked for cafe au lait spots which were absent.

Intraoral examination revealed swelling was present over the upper right labial and buccal vestibular region extending from 51 tooth to distal aspect of 16 tooth region. No mucosal changes were noted and no pus discharge was observed. The Electrical pulp test was normal and all the teeth were vital.

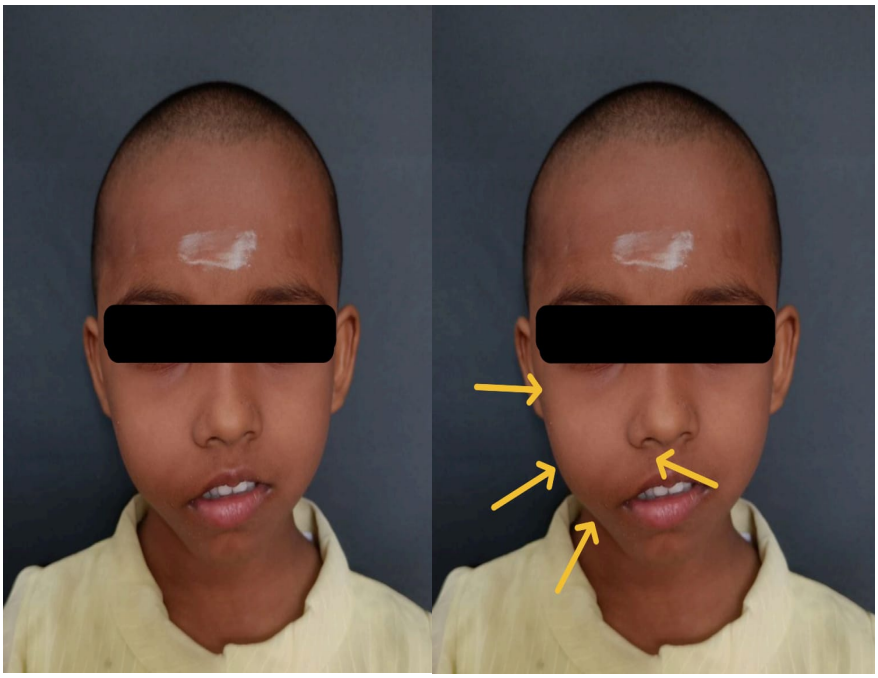

Figure 1 : Depicting the Extraoral picture of the patient - Frontal view.

As it was, the hard tissue lesion patient was advised for full skull Computed Tomography (Figure 2) . Impression was given as Pterygoid process, Greater and lesser wing of sphenoid, Frontal process of maxilla, Temporal, Zygomatic process and orbital surface of the zygomatic bone and alveolar process (along molar teeth) appears expanded with intact cortex with loss of cortico medullary differentiation and being replaced classically by homogeneous ground glass opacities with mass effect on the right maxillary sinus. Features likely suggestive of Fibrous Dysplasia.
Apart from that OPG (Figure 4) was taken which shows the characteristic ground glass appearance.

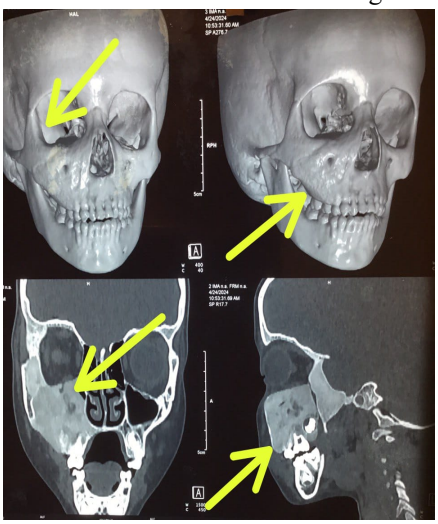

Figure 3: Depicting 3D Reconstructed models as well as coronal and sagittal section views.

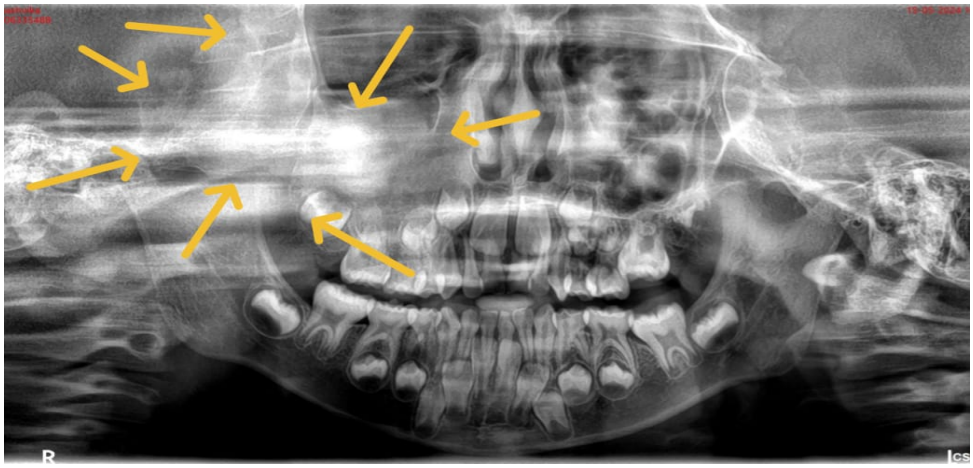

Figure 3 : Depicts characteristic Ground Glass Appearance.

Upon clinical and radiographic findings it was provisionally diagnosed as Fibrous dysplasia. To confirm the diagnosis Incisional biopsy was sent for histopathology.

Impression of the histopathology report was given as the Multiple H and E section shows irregular shaped curvilinear trabeculae of lamellar and woven bone with few showing osteoblastic rimming in a fibrocellular loosely arranged connective tissue with numerous spindle fibroblasts. Few trabeculae also show resting/reversal lines (Figure 4).

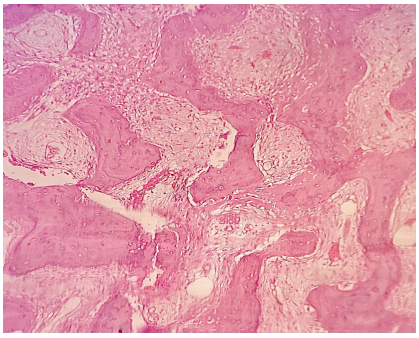

Figure 4 : Depicting the histopathological slide image (100 X Magnification).

Apart from all the above mentioned investigations, Lab investigations like Thyroid Function test, Luteinizing hormone, Follicular stimulating hormone, Estradiol, Parathormone levels were evaluated and they were within normal limits. As all the hormone levels were within normal range mazabraud syndrome was ruled out.

Serum phosphorus, serum calcium, Alkaline phosphatase was evaluated.

Serum phosphorus and calcium levels were within normal limits whereas Alkaline phosphatase levels were raised (i.e 278 IU/L).

After all the investigations the case was diagnosed as Craniofacial Fibrous Dysplasia which is usually a monostotic variant.

Under standard aseptic conditions under general anesthesia left nasotracheal intubation was done. Incision marking was done i.e crevicular incision extended from 51 region to 16 tooth region with two vertical releasing incisions. Administration of 2% local anesthesia with 1:80000 adrenaline

Local infiltration was given. Crevicular incision was given and full thickness mucoperiosteal flap was elevated. Exposure was done superiorly till the infraorbital rim region and posteriorly till the zygomatic arch as well as till the tuberosity of maxilla. Osseous recontouring was done using the vulcanite bur. Hemostasis achieved and closure done using 4-0 Vicryl suture material (Figure 6). Post operative events were uneventful.

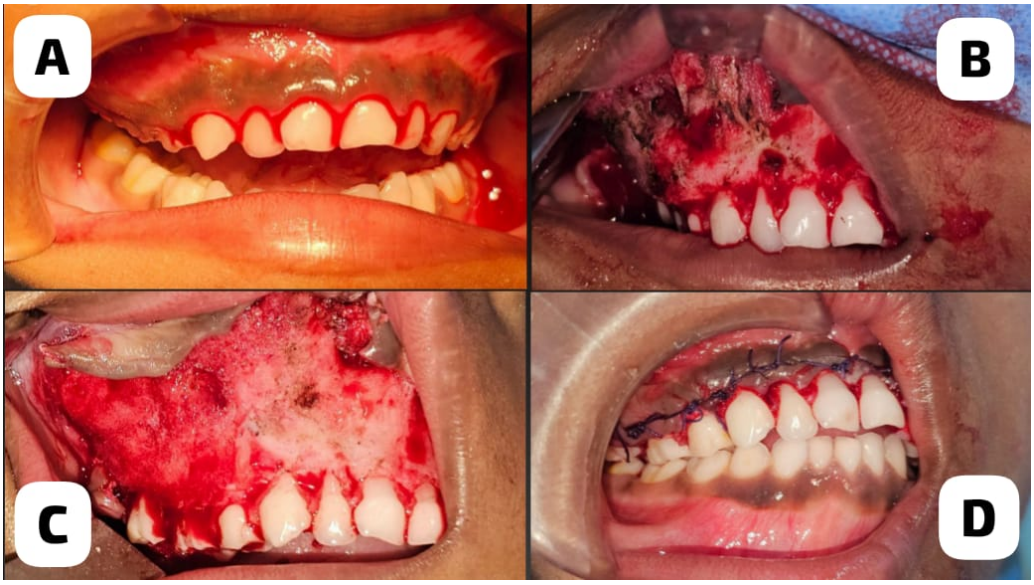

Figure 5: Depicting the surgical procedure.
A.Incision B. Exposure of Osseous lesion C. Osseous Recontouring D. Closure
Post operative healing was satisfactory. Patient was followed up after 3 months. .

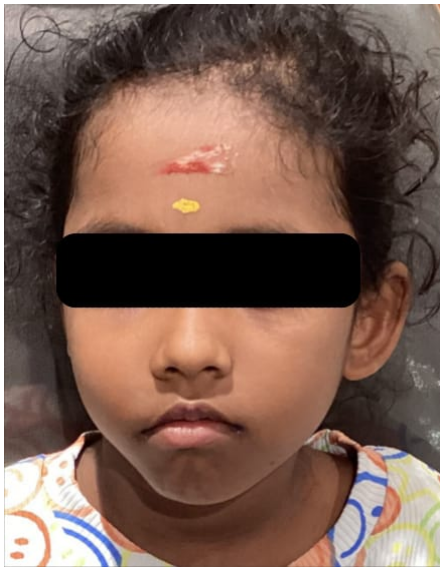

Figure 6 : Depicting the Postoperative photo after 3 months of follow up.

DISCUSSION:

Fibrous dysplasia (FD) is a rare, non-inherited skeletal disorder characterized by the replacement of normal bone with abnormal fibrous tissue. This process leads to the formation of structurally weak bones that are prone to deformities and fractures. The condition can present in one of two forms: monostotic, where only a single bone is affected, or polyostotic, where multiple bones are involved [7]. FD most commonly affects the bones of the skull, face, ribs, femur, and tibia, although any bone can be involved .

The condition is caused by a postzygotic mutation in the GNAS gene, which encodes the Gs alpha subunit of a guanine nucleotide-binding protein. This mutation results in the constitutive activation of the Gs alpha protein, leading to aberrant signaling through the cyclic AMP (cAMP) pathway. The downstream effects of this mutation disrupt normal bone formation, leading to the replacement of normal bone with fibrous tissue and immature, woven bone .Continuous stimulation of adenylate cyclase and an increase in intracellular cAMP levels. The elevated cAMP levels result in abnormal cell signaling, particularly affecting osteoblasts—the cells responsible for bone formation.

The dysregulated osteoblast activity in FD leads to the replacement of normal lamellar bone with fibrous tissue and immature woven bone [8]. This abnormal tissue lacks the structural integrity of normal bone, making the affected bones weaker and more susceptible to deformity and fracture. The histological appearance of fibrous dysplasia is characterized

by irregular trabeculae of woven bone within a fibrous stroma. The trabeculae are typically shaped like "Chinese characters" and lack the organized structure of normal bone [9].

The clinical presentation of fibrous dysplasia varies widely depending on the location and extent of the affected bone. In monostotic fibrous dysplasia, where only one bone is involved, patients may be asymptomatic or present with localized pain, swelling, or a visible deformity . The disease most commonly affects the craniofacial bones, femur, tibia, ribs, and humerus. In cases involving the craniofacial bones, patients may present with facial asymmetry, sinus obstruction, or visual disturbances if the orbit is involved [10]. In polyostotic fibrous dysplasia, where multiple bones are affected, the clinical presentation is more severe and may include bone pain, multiple fractures, and significant deformities. Polyostotic fibrous dysplasia is often associated with McCune-Albright syndrome, a condition characterized by the triad of polyostotic fibrous dysplasia, café-au-lait skin pigmentation, and endocrine abnormalities such as precocious puberty [11].

Patients with fibrous dysplasia are at risk for a variety of complications, depending on the bones involved. These complications can include fractures, deformities, nerve compression, and functional impairment. For example, involvement of the cranial bones can lead to vision or hearing loss [12]. while involvement of the femur can lead to bowing of the bone (often referred to as "shepherd's crook" deformity) and increased risk of fractures [13].

The diagnosis of fibrous dysplasia is typically established through a combination of clinical evaluation, radiographic imaging, and histopathological examination. The clinical history and physical examination may reveal signs suggestive of FD, such as localized bone pain, swelling, or deformity. However, imaging studies are essential for confirming the diagnosis and assessing the extent of the disease.

Radiographs of affected bones typically reveal a characteristic "ground glass" appearance, caused by the presence of fibrous tissue within the bone [14]. This radiographic finding is highly suggestive of FD, although it is not specific to the condition. In cases of craniofacial involvement, computed tomography (CT) scans are often used to provide detailed images of the affected bones and to assess the extent of bone involvement [15].

Magnetic resonance imaging (MRI) can also be useful in evaluating FD, particularly for assessing the soft tissue components of the lesion and for identifying any associated complications, such as nerve compression [16].

Histopathological examination of a biopsy specimen is the gold standard for diagnosing fibrous dysplasia. The biopsy typically shows irregular trabeculae of woven bone within a fibrous stroma. These trabeculae are often described as having a "Chinese character" appearance due to their irregular shape. Immunohistochemical staining can be used to demonstrate the presence of the mutated Gs alpha protein, further confirming the diagnosis.

There is currently no cure for fibrous dysplasia, and treatment is primarily focused on managing symptoms and preventing complications. The management of FD is typically multidisciplinary, involving orthopedic surgeons, endocrinologists, and other specialists like Cranio Maxillofacial Surgeons as needed.

In asymptomatic patients or those with mild symptoms, conservative management may be appropriate. This approach includes regular monitoring of the affected bones through imaging studies, as well as symptomatic treatment with analgesics for pain control. Bisphosphonates, a class of drugs that inhibit bone resorption, have been used to manage bone pain in FD patients, although their efficacy in altering the course of the disease remains uncertain [17].

Surgical intervention may be required in cases where the disease causes significant deformity, functional impairment, or is associated with a high risk of fractures. Surgical options include corrective osteotomy to realign deformed bones, bone grafting to provide structural support, and internal fixation to stabilize fractures or prevent future fractures. In cases of craniofacial involvement, surgery may be necessary to relieve nerve compression, correct facial asymmetry, or address other functional or cosmetic concerns [18].

In patients with McCune-Albright syndrome or those with isolated endocrine abnormalities, hormonal management may be necessary. This may include the use of medications to manage precocious puberty, thyroid dysfunction, or other endocrine issues associated with FD.

Although rare, there is a small risk of malignant transformation of fibrous dysplasia into osteosarcoma or other bone malignancies. This risk is higher in patients who have received radiation therapy, highlighting the importance of avoiding unnecessary radiation exposure in these patients [19].

The prognosis for patients with fibrous dysplasia varies widely depending on the extent of the disease and the presence of associated complications. In many cases, the disease remains stable over time, with patients experiencing only mild symptoms or no symptoms at all. However, in some cases, the disease can progress, leading to significant morbidity due to fractures, deformities, and functional impairment.

Research into targeted therapies for fibrous dysplasia is ongoing, with the aim of developing treatments that address the underlying molecular mechanisms of the disease. Potential therapeutic targets include the Gs alpha protein, cAMP signaling pathways, and the abnormal osteoblast activity that characterizes FD. While these therapies are still in the experimental stages, they hold promise for improving outcomes in patients with this challenging condition.

Other research is exploring the role of bone remodeling pathways, such as the RANK/RANKL/OPG pathway, in the pathogenesis of FD. Understanding these pathways could lead to the development of new treatments that help to normalize bone remodeling and prevent the formation of fibrous tissue .

In addition to molecular therapies, advances in surgical techniques and imaging technologies are improving the management of FD. Minimally invasive surgical techniques, such as endoscopic approaches for craniofacial FD, are reducing the morbidity associated with surgery and improving patient outcomes.
Advanced imaging techniques, such as three-dimensional CT and MRI, are providing more detailed assessments of the disease and helping to guide surgical planning [20].

CONCLUSION:

In conclusion, this case report highlights the clinical and diagnostic challenges associated with monostotic craniofacial fibrous dysplasia in an 8-year-old female. The presentation of facial asymmetry and swelling underscored the importance of early recognition and thorough diagnostic evaluation, including imaging studies and histopathological confirmation. Surgical intervention was crucial in managing the patient's symptoms and preventing potential complications. This case emphasizes the need for a multidisciplinary approach in the treatment of craniofacial fibrous dysplasia, particularly in pediatric patients, to ensure optimal functional and cosmetic outcomes. Early diagnosis and appropriate management are essential in minimizing the impact of this rare disorder on the patient's quality of life.

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