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Case Report

Craniofacial fibrous dyplasia: A case report with emphasis on radiographic findings

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ABSTRACT

Fibrous dysplasia (FD) is an idiopathic skeletal disorder where normal bone gets replaced by poorly organized fibrous connective tissue. The lesion is classified into two forms: monostotic and polyostotic. This disorder arises from sporadic mutation of the α-subunit of the Gs stimulatory protein. Although histopathology is a gold standard in the diagnosis of any pathology, radiology remains an important investigation. In radiology, fibrous dysplasia is very often associated with the term ground glass matrix. The knowledge of its unique pathogenesis and course are crucial to understand imaging findings and potential complications. Here, we are presenting case report of fibrous dysplasia involving the craniofacial bones.

1. Introduction

Fibro-osseous disorders are the group of disorders in which normal bone is replaced by fibrous tissue consisting of new mineralized tissue. FD is one of the fibro-osseous disorders. FD is a developmental condition characterized by replacement of normal bone by proliferative cellular fibrous connective tissue along with irregularly placed bony trabeculae.[1] FD results from a mutation in GNAS 1 (Guanine nucleotide binding protein alpha stimulating activity polypeptide 1) gene. The severity of condition depends on the time of occurrence of mutation during fetal or postnatal life.[2] When the disease is localized to a single bone called monostotic FD and affecting multiple bones is polyostotic FD. The McCune-Albright syndrome, Mazabraud syndrome and Jaffe–Lichtenstein syndrome are seen associated with FD.[3] Here, the patient showed typical ground glass appearance in most of the skull and facial bones. Also, the FD commonly affects maxilla but here primary involvement of jaw is in mandible. Hence, the case puts immense value in the literature.

2. Case Report

A 23 years old male patient reported to the Department of oral Medicine and Radiology with the complaint of missing teeth in mandibular anterior teeth region of the jaw since 15 years. The patient gave history of traumatic exfoliation of deciduous teeth in road traffic accident when he was 8 years old and then there was no eruption of permanent teeth at that place. For this same complaint he visited many dentists and on radiological examination they suggested him orthodontic treatment as they observed that 33 and 34 were impacted.

On intra-oral examination missing 33 and 34 were observed. All other teeth were present in normal position.

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Also, diffuse swelling was seen extending from mesial aspect of 32 to distal aspect of 35 mesiodistally and from crest of edentulous alveolar ridge to mandibular vestibule superoinferiorly. The approximate size of the swelling was 3 X 2 cm. The overlying mucosa appears normal. (Figure 1) The swelling was hard, non-tender, non-reducible and non-compressible. Teeth in vicinity to the swelling were vital. The provisional diagnosis of fibro-osseous lesion was made. The differential diagnosis for this case was fibrous dysplasia. Complete hemogram showed all parameters within normal range.

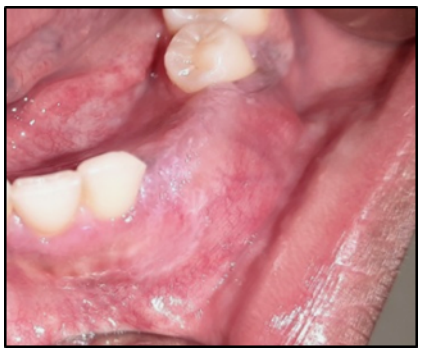

Fig. 1: Intraoral presentation

On radiographic examination of OPG mixed radiolucent radiopaque lesion having ground glass appearance was seen extending from apex of 45 to distal region of 38 mesiodistally and from alveolar crest in edentulous area to approximately 10 mm away from inferior border of mandible superoinferiorly.(Figure 2) On CBCT the lesions having ground glass appearance were seen in the mandible (Figure 3 A & B), zygomaticomaxillary complex on left side and frontal process of zygomatic arch on left side (Figure 4), petrous part of temporal bone and mastoid air cells on left side (Figure 5), pterygoid plates on left side (Figure 6), anteroinferior part of sella tursica (Figure 7), involvement of lateral wall of orbit on left side and lateral wall of maxillary sinus on left side (Figure 4). Widening of skull base was also present (Figure 5). The inferior alveolar nerve on left side displaced superiorly (Figure 3 B) hence radiographically diagnosed as craniofacial fibrous dysplasia.

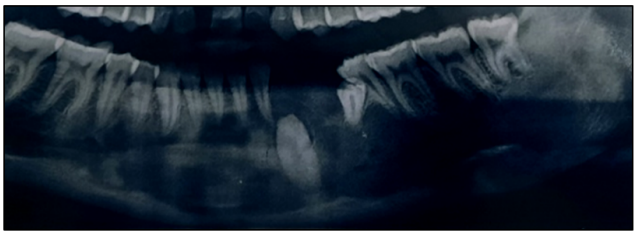

Fig. 2: Cropped image of OPG

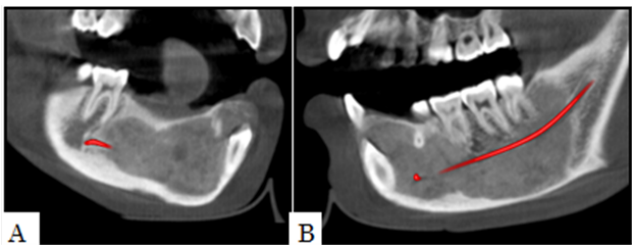

Fig. 3: A & B: Lesion in mandible

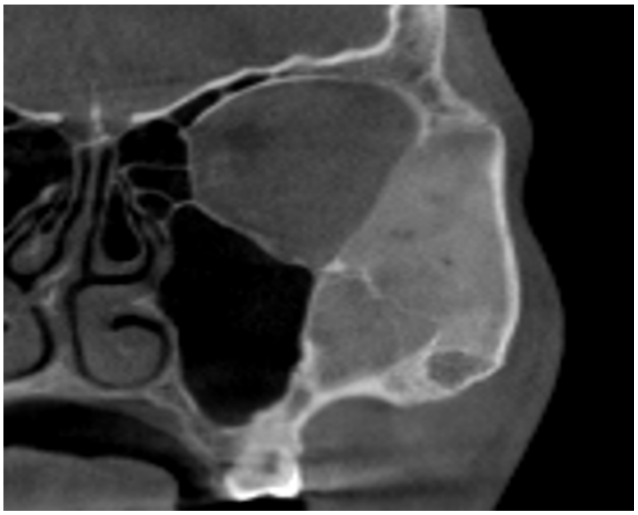

Fig. 4: Zygomaticomaxillary complex

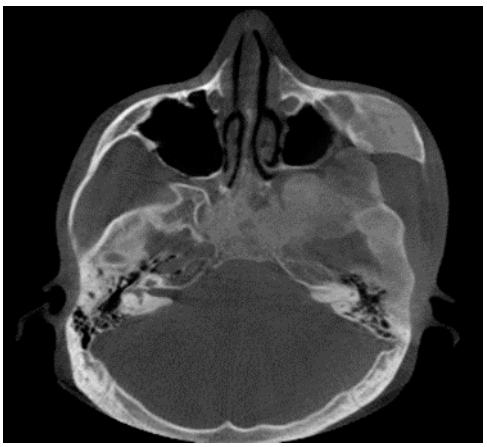

Fig. 5: Petrous part of temporal bone and mastoid air cells

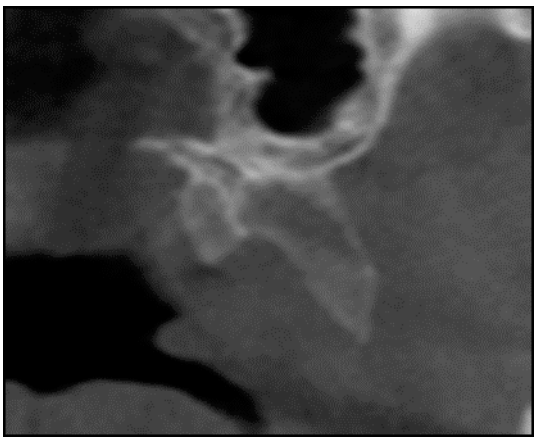

Fig. 6: Pterygoid plate

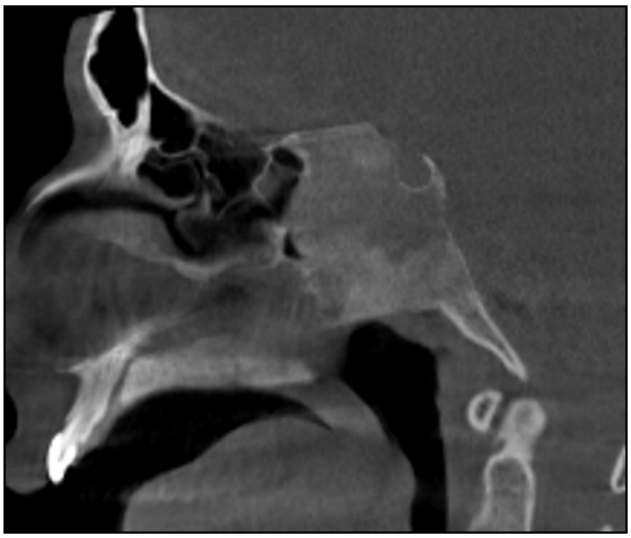

Fig. 7: Anteroinferior part of sella tursica

Though the diagnosis of fibrous dysplasia can be done based on clinical and radiographic findings we decided to for incisional biopsy for confirmation. Initially aspiration was done, which was negative. The incisional biopsy was done at 33, 34 region and the fibrous tissue was sent for histopathologic examination.

On histopathologic examination multiple decalcified tissue sections were seen only in connective tissue stroma. The connective tissue stroma was fibrocellular with haphazardly arranged bundles of collagen fibers. Also numerous plum fibroblasts were seen. At some places cementum like globular calcification was seen and at other places calcifications showed retraction from adjacent connective tissue stroma. Numerous variable sized blood capillaries seen at some places. Areas of extravasation and degeneration were seen at some places. Hence, it was diagnosed as fibrous dysplasia with increased vascularity (Figure 8).

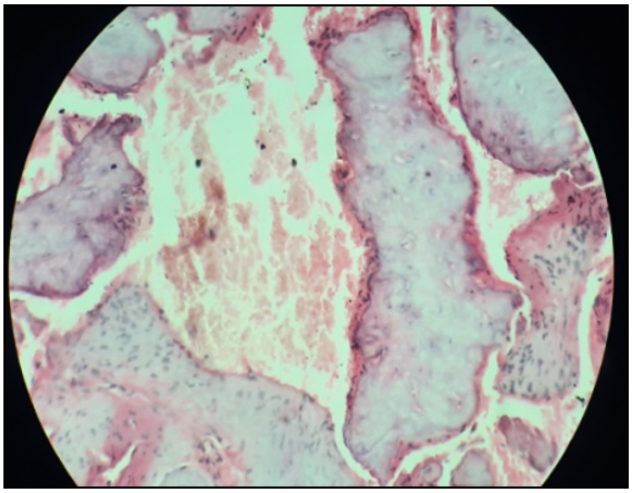

Fig. 8: Histopathologic slide

3. Discussion

FD is a most common fibro-osseous disorder arising due to mutation of α-subunit of the Gs stimulatory protein. Here the bone is distorted and replaced by fibrous tissue which is poorly organized and unsound in structure.[3] This condition is believed to be hamartomatous in origin.[4] It is considered to be a disease of young population having incidence of 1:4000 to 1:10,000.[5] Here the bones of face and skull are involved which results in asymmetry of face and spontaneous fractures.[6]

According to Reed's definition, FD is an arrest of bone maturation and presence of woven bone with ossification resulting from metaplasia of a nonspecific fibro-osseous type.[7]

3.1. Pathogenesis

The mutations in the alpha subunit of a G stimulatory protein causes activation of adenylyl cyclase, which results in elevation of cyclic adenosine monophosphate (cAMP). This causes stimulation of endocrine receptors. The increase in cAMP has many downstream effects.[8] It puts abnormal expression in several target genes like c-fos (a proto-oncogen which is a human homolog of retroviral oncogen v-fos), c-jun (a proto-oncogen which is cellular homolog of viral oncoprotein v-jun), interleukin-6 (IL-6) leading to osteoblastic recruitment and functional disturbance in dysplastic bone.[9] The increased number of osteoclasts and bony resorption demonstrated in FD is related to IL-6. In a study done by Candeliere et al. showed FD-affected bone marrow spaces contain high levels of c-fos and bones of healthy subjects or uninvolved bones of FD patients had no c-fos expression.[9] Increase in intracellular cAMP of FD-affected bones causes cell proliferation with defect in differentiation. The important feature of FD

is bone expansion, explained by cellular proliferation and the immature woven bone manifesting inappropriate differentiation due to the mutated gene.

The mutation occurring in early embryonic life manifests the mutated genes in various forms like multiple bone lesions, cutaneous pigmentation and endocrine disturbances leading to McCune Albright syndrome. When mutation occurs in later stage the progenies of mutated cell disperse and take part in the formation of skeleton resulting in multiple bone lesions that is polyostotic FD. The mutation occurring in postnatal phase, the progenies of mutated cell are manifested in one site only leading to monostotic FD.[2]

Types: According to Eversol[2]

1. Monostotic
2. Polyostotic
3. Polyostotic with endocrinopathy (McCuneAlbright)
4. Osteofibrous dysplasia

The craniofacial structures are affected in 10% of monostotic FD, 50% of mild cases of polyostotic FD, and 100% of severe cases of polyostotic FD. Maxilla and mandible are commonly affected and involvement of temporal bone seen in 18% cases.[6]

3.2. Clinical features

Monostotic FD is more common and seen in about 80% patients of FD. It involves people at the age range of 20–30 years. The common locations in decreasing order of frequency are the rib, femur, tibia, craniofacial bones, and humerus. The monostotic FD commonly present pain or a pathologic fracture.[10]

Approximately 20-30% FD are polyostotic. This more frequently involves the skull and facial bones, pelvis, spine, and shoulder girdle. The common sites of involvement in decreasing order are the femur, tibia, pelvis, ribs, skull and facial bones, upper extremities, lumbar spine, clavicle, and cervical spine. It may manifest unilaterally or bilaterally, and may affect various bones of a single limb or both limbs. It may or may not affect axial skeleton. It initially shows pain in the involved limb with a limp, spontaneous fracture, or both.[10]

The monostotic FD is often discovered incidentally, while the polyostotic FD diagnosed during first few years of life. The majority of bony lesions arise and become clinically significant by 10 years of age and almost no new bony lesions seen after the age of 15 years.[3] The lesions are active in early childhood and expand during process of linear growth then become static after puberty, the activity decreases throughout adulthood.[3] The females are affected more, almost twice commonly than males, according to some authors; however, the others say that incidence in males and females is equally.[1] FD arises as an asymptomatic painless swelling which later causes facial asymmetry. The maxilla affected commonly than mandible. Occasionally, there are reports of a painful and suppurative swelling with ulceration. Other symptoms include nasal obstruction due to involvement of paranasal sinuses, visual disturbances because of involvement of orbit, and the lesion in temporal may cause hearing loss. The patient may complain of facial pain, headaches and facial numbness.[1]

The craniofacial FD occurs in 10– 25% of patients having monostotic FD and in 50% having polyostotic FD. It may occur as craniofacial FD isolated form without extracranial. The most common sites in craniofacial FD are frontal, sphenoid, maxillary, and ethmoidal bones, occipital and temporal bones involved less commonly. Hyper telorism, cranial asymmetry, facial deformity, visual impairment, exophthalmos, and blindness may be seen after involvement of orbital and periorbital bones. Involvement of the sphenoid wing and temporal bone result in vestibular dysfunction, tinnitus, and hearing loss. Involvement of cribriform plate may lead to hyposmia or anosmia.[10]

3.3. Radiographic features

Fibrous dysplasia affects the maxilla twice often than mandible and more frequently the posterior regions of the jaw. The periphery of FD lesions is poorly defined, with a gradual and broad transition between the dysplastic and normal bone. This may be referred as blending of the fibrous dysplastic bone into the normal trabecular pattern. The abnormal trabeculae are short and thin than normal bone, and more irregular in shape. They are more numerous in number than the normal trabeculae. Due to these changes a series of classical radiologic patterns can be seen in FD.[11]

Fries gave three radiographic presentations of craniofacial FD.[12,13]

1. Pagetoid or ground glass appearance.
2. Sclerotic pattern.
3. Cyst-like pattern.

The pagetoid or ground glass pattern shows bony expansion along with alternate areas of radiopacity and radiolucency. The sclerotic pattern shows bony expansion with homogenous radiopacity. The cyst-like pattern seen as a round or oval lesion having sclerotic border.

As per Shafer's Textbook of Oral pathology[10]

1. Small unilocular radiolucencent lesion or a larger multilocular radiolucency, with well circumscribed border.
2. Similar to the first, but has increase in trabeculation giving mottled appearance.
3. Ground glass or peau d'orange appearance which is more radiopaque than the former having numerous delicate trabeculae.

Obisesan et al. have classified FD of craniofacial bones into six types[14]

1. Peau d'orange/Orange peel type
2. Whorled plaque like type
3. Diffuse sclerotic type
4. Cyst like type
5. Pagetoid type
6. Chalky type

Table 1:

Pattern	Description
Ground glass appearance	Sand-blasted or etched glass
Sclerotic appearance	Homogenous increase in radiodensity with gradual merge with surrounding normal bone
Cyst-like (simple bone cyst)	Radiolucent regions may occur in mature lesions of FD
Orange peel appearance (Peau d'orange)	Alternate areas of radiolucency and opacity with granular appearance
Fingerprint bone pattern (swirling pattern)	Well circumscribed lesion containing plaques of amorphous materials having intermediate radiodensity
Chalky type	Well circumscribed dense amorphous radiopaque lesion

Displacement of teeth and root resorption seen in rare cases. The periodontal ligament (PDL) space may show narrowing. The increased density of bone may make PDL space to appear widened. Lamina dura is ill-defined. FD may expand into the antrum and maintains the original outer contour, whereas it is more convex extension in the neoplasm. The lateral wall is involved first and last wall involved is the most posterosuperior portion. FD behaves more characteristically in the mandible as it displaces the inferior alveolar canal in superior position.[1]

Skeletal radiographic features include Leg-length discrepancy seen in about 70% of patients showing limb involvement. The structural integrity of the bone is weakened, so the weight-bearing bones become bowed. There is increase in curvature of the femoral neck and proximal shaft leading to a Shepherd's crook deformity, which is a characteristic sign.[10]

Nisha Dua et al in 2015 reported two cases of FD each involving swelling in the maxilla and showing ground glass appearance, loss of lamina dura, involvement of maxillary sinus and expansion of buccal cortical plate.[6] Elizabeth H. Theng et al in 2021 reported a case series of craniofacial FD with inflammation of periorbital region.[15]

3.4. Syndrome association

1. Jaffe–Lichtenstein syndrome - Multiple bone involvement with café-au-lait pigmentation.
2. McCune Albright syndrome – polyostotic FD associated with café-au-lait pigmentation and endocrinopathies including hyperthyroidism, sexual dysfunction.
3. Mazabraud's syndrome - FD along with multiple soft tissue myxomas.

3.5. Differential diagnosis

The metabolic bone diseases like hyperparathyroidism, has same radiographic presentation, but it is more generalized and no bony expansion seen. Paget's disease may show similar radiographic pattern and bony expansion but involvement of the complete bone is seen, and is seen in elderly individuals. The osteomyelitis presenting as the usual enlargement of jaws may show similar presentation however in cases of osteomyelitis reactive bone formation is seen at the periosteum. Osteosarcoma may show more aggressive radiographic findings like a more bizarre internal structure.[11]

3.6. Investigations

Routine investigations include complete hemogram. Serum ALP is elevated occasionally. Serum calcium, parathyroid hormone, 25-hydroxyvitamin D, and 1,25-dihydroxyvitamin D levels are normal in most cases. Cases of aggressive polyostotic FD may show hypophosphatemia, hyperphosphaturia, and osteomalacia.[6]

3.7. Management and prognosis

The management of FD uses bisphosphonates and results have not been that good. But many authors reported pain and inflammation were improved, bone destruction was reduced, increased osseous density, rarefaction of osteolytic lesions, also the radiologic aspects and osseous metabolism showed improvement.[1] The primary treatment of choiceis surgery, which includes conservative recontouring of jaw. But surgical approach of cranio-maxillo-facial lesions is controversial because excision of all affected bone is usually of no use as it is impossible to find the boundaries of the disease. It is indicated in cases if an important function is affected or develops complications.[6]

Clinical findings like increase in pain and an enlargement of soft tissue suggest malignant transformation. Osteosarcoma is the most common which often affects the craniofacial bones.[16] The fibrosarcoma, chondrosarcoma, and Ewing's sarcoma may be seen.[17]

4. Source of Funding

None.

5. Conflict of Interest

None.

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