Fibrous dysplasia of maxilla: A case report
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Abstract
Fibrous dysplasia (FD) is classified as a fibro-osseous lesion, benign in nature which exhibits distinctive features like exchange of normal bone by fibro-osseous connective tissue with different degrees of osseous metaplasia. The craniofacial type of FD which can affect jaw bones is a rare entity. It may cause pain, deformities, and pathological fracture of bone. FD constitutes approximately 2-5% of all bone tumors and 7% of all benign tumors. Here, we are reporting a case of FD affecting maxilla. Diagnosis was based on clinical, radiographical, and histological findings. Differential diagnosis poses a great value in case of FD.

Keywords: Craniofacial, fibro-osseous lesion, fibrous dysplasia, monostotic, polyostotic

Introduction
The fibrous dysplasia (FD) is a type of fibro-osseous lesion with the disturbance of the bone metabolism. Fibrous content of connective tissue replaces normal bone by abnormal bone.[1-3] There is somatic activating mutations of the stimulatory G protein (α subunit) encoded by the gene guanine nucleotide binding protein, a stimulating activity polypeptide (GNAS).[2] However, craniofacial dysplasia (CFD) is a rare variant of FD. Involvement of craniofacial structures in CFD may have symptoms such as vestibular dysfunction, tinnitus, hearing loss, hypertelorism, visual impairment, exophthalmos, and blindness.[3] The uneven shaped trabeculae in radiographic help in the differential diagnosis. Most commonly seen in the second decade of life.[4]

Case Report
A male patient aged 28 years reported to our institution with the chief complaint of painless swelling over the left part of the face since birth. The patient had history of painless swelling which is growing proportionally as patient grows. There was no relevant medical history. On extra-oral examination, facial asymmetry with swelling on the left maxillary region of the face was noted. The swelling was extending from the right infraorbital margin till upper alveolar region superior-inferiorly and from ala of the nose to 1 cm anterior to the pinna of the ear anteroposteriorly. On inspection, there was diffuse swelling, roughly ovoid in shape, extending from the ala of the nose to the tragus of the ear on the left side of the face [Figure 1]. The overlying skin is normal in appearance. On palpation, the swelling measures 7 cm × 5 cm extending from 22 to 28 margins are ill-defined, hard in consistency and vestibular obliteration on the left maxillary buccal vestibule. The vestibular obliteration was present extending from the left maxillary lateral incisor to the left maxillary third molar [Figure 2]. It was provisionally diagnosed as FD of maxilla. Cone beam computed tomography (CT) showed mixed radiolucent and radiopaque features with cortical bone expansion [Figure 3]. Incisional biopsy was taken from maxillary vestibule, the lesional area.

On histopathological examination, the H and E stained decalcified hard tissue section showed low to moderate cellular fibrous stroma with spindle cells surrounding irregular trabeculae of woven bone. Osteocytes exhibited Chinese letter pattern in some areas without osteoblastic rimming [Figure 4]. Some areas showed osteoblastic rimming. Few areas of blood filled cavity demarcated by connective tissue septa resembling aneurysmal bone cyst-like features are seen along with dilated blood vessels [Figure 5].

Discussion
The FD is nonhereditary disorder with skeletal developmental anomaly of the bone-forming mesenchyme that manifests as
Fibrous dysplasia of maxilla

Nonitha, et al.

an osteoblastic differentiation and maturational defect.\cite{4,5} Abnormal bone comprising fibrous component in connective tissue which substitute the normal bone.\cite{6,7} FD is caused by a mutation in GNAS1 gene (20q13.2), the gene that encodes G protein that is responsible for the production of cyclic adenosine monophosphate (cAMP) in affected tissue, resulting in: (a) Increased activity of the affected endocrine organs, causing precocious puberty, increased growth hormone, hyperthyroidism, and increased cortisol production; (b) Increase in proliferative activity of melanocytes results in abnormal patches of light brown with irregular margins (cafe-au-lait spots); (c) cAMP alter differentiation of osteoblast leading to FD; (d) fibroblast growth factor 23 -mediated phosphate wasting with or without hypophosphatemia in relation with FD.\cite{4}

FD may involve any bone consisting 7% of all benign osseous tumors. FD may affect children and young adults. The monostotic form dysplasia has equal incidence in both genders and is 6 times more frequent than the polyostotic variant. Recent research has shown a slight female preponderance.\cite{1} On contrary, we reported a case of male patient. In the monostotic form, maxilla is affected more commonly than mandible.\cite{4,6} FD is classified into:\cite{1,4}

**Monostotic form**

It represents 70-80% of FD.\cite{2} Affected parts are ribs, femur, tibia, craniofacial bones, and humerus 28%, 23%, and 10-25%, respectively. Pain and pathological fracture are the features most commonly observed from the first decade to seventh decade. There is a less deformity as compared to polyostotic form.\cite{9}
Polyostotic form

It accounts 20-30% of FD. It involves the skull and facial bones, clavicle, shoulder girdle, pelvis, spine, femur, and tibia. Either it is unilateral or bilateral. Several bones are affected involving single limb/both limbs with or without axial involvement. Before the first decade about two-third of the patients are symptomatic. Pain and pathological fracture are common features. Bowing of weight bearing bones with curved femoral neck and proximal shaft of femur increase - Shepherd’s crook are pathognomonic features. FD with pigmented lesion; cafe-au-lait (Jaffe’s type) Albright’s syndrome constitutes of pigmented lesion, FD, endocrine disturbance.\(^6\)

Craniofacial form

It accounts 10-25% of monostotic form and 50% polyostotic. Frequently involved sites are frontal, sphenoid, maxillary, and ethmoid bones. The associated features are cranial asymmetry, hypertelorism, facial deformity, visual dysfunction, exophthalmos, and orbital bone involvement may cause blindness. Tinnitus, vestibular dysfunction, and deafness may occur with the involvement of sphenoid wing and temporal bone. It may affect cribiform plate causing hyposmia and anosmia.\(^4\)

Radiographic appearance

- Type I - Unilocular or multilocular radiolucency with a well-circumscribed border of fine bony trabecular network.
- Type II - Similar to Type I with increased trabeculation. The appearance is more opaque and mottled.
- Type III - Ground glass or Peau d’ orange appearance seen due to numerous delicate trabeculae and thus the lesion appears opaque blend with the normal bone.\(^6\)

The reported case has Type III radiographic presentation.

Further any sarcomatous alteration in the lesion can be identified by magnetic resonance imaging/CT scan.\(^6\)

Histologic appearance

Lesion contains fibrillar connective tissue with trabeculae of woven immature bone, irregular in shape. Large osteocytes and collagen fibers of trabeculae were extending into fibrous tissue. The trabeculae may show Chinese letter pattern.\(^6\) The absence of osteoblastic rimming. Bone formation by stellate osteoblast was noted. Osteoclastic activity may be seen.\(^{1,6}\) In our case, osteoblastic rimming was absent.

Differential diagnosis of FD can be made on clinical, radiographic, laboratory, and histological findings. The pathological conditions which mimics FD can be classified as other fibro-osseous lesions (cemento-osseous dysplasia, and ossifying fibroma), bone cysts, cementoma, Paget’s disease, cherubism, hyperparathyroidism, chronic sclerosing osteomyelitis, and osteogenic sarcoma.\(^9\)

Treatment and prognosis

Bisphosphonates can be used in inoperable cases.\(^{5,10}\) Surgical curettage and bone replacement with graft. The frequency of malignant transformation is 0.4-1%.\(^{4,10}\) The radiotherapy increased malignant transformations more than 400 times.\(^{11}\)

Conclusion

FD may affect craniofacial and jaw bones and lead to deformities. The mutation of protein can have several effects on hormone receptors resulting in endocrinopathies. The monostotic FD is a rare entity and here we reported FD of maxilla in a male patient with no endocrinopathy.

Clinical significance

Various fibro-osseous lesions may also show similar features like FD, so clinician should have knowledge about differentiating features. Although FD is a benign osseous tumor, it could show the malignant transformation. Considering this fact appropriate diagnosis and follow-up is necessary.

References