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A case of isolated craniofacial fibrous dysplasia – Radiologist's perspective



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ABSTRACT

Fibrous dysplasia is a disease characterized by replacement of bone by fibro osseous tissue. FD has four different disease patterns. They are monostotic, polyostotic and McCune–Albright syndrome. Craniofacial pattern of disease occurs in 10–25% of patients with monostotic form and in 50% with polyostotic form. It also occurs as isolated craniofacial form. In isolated variety no extracranial lesions are present. Radiographic diagnosis plays an important role in diagnosis, classification and assessing prognosis of fibrous dysplasia. In this paper we report a case of isolated craniofacial type of fibrous dysplasia in a young female patient involving the maxilla and skull bones with a complete radiographic CT assessment of the extent of the lesion. Temporal bone involvement and bilateral lesion in certain cranial bones are the rare findings noted in this case.

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1. Introduction

Fibrous dysplasia is a skeletal developmental anomaly of the bone forming mesenchyme that manifests as a defect in osteoblastic differentiation and maturation. FD is caused due to the activating mutations in the GNAS gene which encodes for the alpha subunit of the signalling G protein, Gs alpha.[1] Following the suggestion of McCune and Bruch (1937), that this disorder is to be considered as a distinct clinical entity, Lichtenstein introduced the term Fibrous dysplasia.[2,3] Fibrous dysplasia is described in terms of three major types, monostotic involving a single bone, polyostotic involving multiple bones and McCune–Albright syndrome, a polyostotic form of fibrous dysplasia that also involves endocrine abnormalities.[4] Craniofacial pattern of disease occurs in 10–25% of patients with monostotic form and in 50% with polyostotic form. It also occurs as isolated craniofacial form. In isolated variety no extracranial lesions are present.[5] Facial region may be affected by a form of fibrous dysplasia that is not strictly monostotic, but may be confined to a single anatomical region. These lesions affect primarily the maxilla and simultaneously cross sutures and enter into the adjacent facial bones. This type of fibrous dysplasia does not meet the precise criteria for the monostotic or polyostotic forms and has been termed craniofacial fibrous dysplasia.[6,7] Though conventional radiographs are needed to evaluate the trabecular pattern, lamina dura and effect on adjacent structures, advanced imaging modalities like CT is essential for assessing the exact extension of the disease. In this paper we report a case of isolated craniofacial type of fibrous dysplasia involving the right maxilla and adjacent cranial bones in a young female patient with a complete radiographic assessment of the extent of the disease. As in most cases of fibrous dysplasia,

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clinical and radiographic features are sufficient to make a diagnosis, a radiologist is to be consulted to give a more informative three dimensional representation of the extent of the lesion. Also radiographs are used as reference tools for comparison in future.

2. Case report

A female patient of age 15 years came with a complaint of asymmetry of face due to a slow growing painless swelling in the right side of her upper jaw for the past one year. Extra oral examination revealed a diffuse swelling in the right side cheek region (Fig. 1). The swelling was bony hard in consistency and nontender. Intra oral examination revealed a swelling of size 3 × 2 cm present obliterating the right buccal sulcus and palatal surface extending from the mesial aspect of 12 to distal aspect of 17 (Fig. 2). The swelling was bony hard in consistency and nontender. A provisional diagnosis of fibro osseous lesion was given. Intra oral periapical radiograph of right anterior and posterior maxillary regions revealed ground glass appearance of trabeculae, with ill defined periphery, blending into the surrounding bone and disappearance of distinct lamina dura (Figs. 3 and 4). Maxillary occlusal view revealed ground glass appearance of trabeculae and expansion with a thinned out buccal cortex (Fig. 5). Panoramic radiograph revealed fine granular opacity involving the right maxillary antrum (Fig. 6). CT axial view (Fig. 7) revealed a homogenously dense lesion involving the right maxilla, obliterating the right maxillary sinus, with parallel thickening of the outer cortical border and a residual air space that has maintained the normal anatomic shape, characteristic feature found in fibrous dysplasia. Involvement of pterygoid plates is also seen.



Fig. 2 – Preoperative intra oral view.

Further axial sections (Figs. 8–10) and coronal sections (Figs. 11 and 12) revealed obliteration of the sphenoid sinus, squamous portion of right temporal bone, posterior ethmoidal air cells and frontal bone. Complete skeletal analysis was done to rule out extracranial involvement. Biochemical analysis revealed an elevated serum alkaline phosphatase level (987 U/L – ref value (60–306 U/L)) and normal serum calcium and phosphorous levels. A working diagnosis of Fibrous dysplasia – craniofacial type was given as the lesion involved multiple bones of the craniofacial region only with no extracranial involvement. Histopathology revealed irregularly shaped bony trabeculae of varied sizes in a fibrocellular stroma (Fig. 13) which correlated with the clinical diagnosis. As the



Fig. 1 – Preoperative extra oral view.



Fig. 3 – IOPA – ground glass appearance.

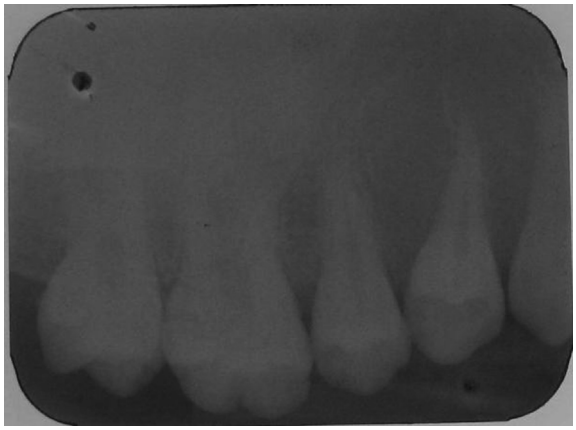


Fig. 4 – IOPA – ground glass appearance.

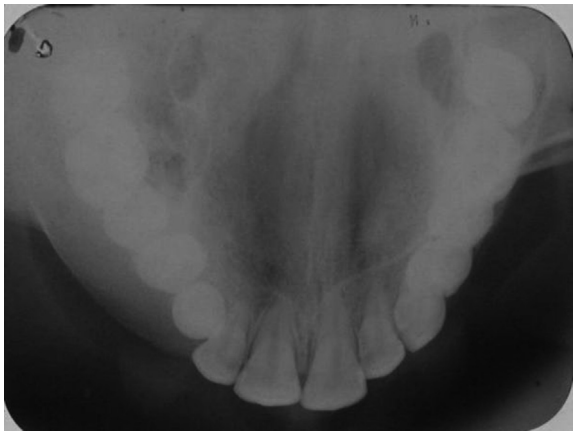


Fig. 5 – Maxillary occlusal view – expansion of cortex.

patient was asymptomatic except for the facial asymmetry, a conservative surgical recontouring was done with improved facial symmetry (Figs. 14 and 15). The patient is kept under observation.



Fig. 7 – CT axial – right maxilla and pterygoid plates involved.

3. Discussion

Reed's definition states that fibrous dysplasia is an arrest of bone maturation, woven bone with ossification resulting from metaplasia of a nonspecific fibro osseous type.[4] Fibrous dysplasia is described in terms of three major types, monostotic involving a single bone, polyostotic involving multiple bones and McCune–Albright syndrome, a polyostotic form of fibrous dysplasia that also involves endocrine abnormalities.[4] Initial manifestation is mostly found in 3–15 years of age. Two thirds with polyostotic type are asymptomatic till 10 years of age and monostotic types are asymptomatic as old as 20–30 years of age. Craniofacial pattern of disease occurs in 10–25% of patients with monostotic form and in 50% with polyostotic form. It also occurs as isolated craniofacial form. In isolated variety no extracranial lesions are present. Sites of involvement most commonly include the frontal, sphenoid, maxillary and ethmoid bones. Occipital and temporal bones are less commonly affected. In the present case, involvement of



Fig. 6 – OPG – right maxillary sinus involvement.



Fig. 8 – CT axial – bilateral sphenoid bone involvement.



Fig. 10 – CT axial – frontal bone involvement.

temporal bone is also seen. Lesions more commonly are unilateral except for very rare extensive lesions of the maxillofacial region that are bilateral.[8] The present case shows bilateral involvement of cranial bones. Hypertelorism, cranial asymmetry, facial deformity, visual impairment, exophthalmos and blindness may occur because of involvement of orbital or periorbital bones. Involvement of sphenoid wing and temporal bones may result in vestibular dysfunction, tinnitus and hearing loss. When the cribriform plate is involved, hyposmia or anosmia may result.[5] In the present case no neurologic deficit was found. The average age of patients with craniofacial fibrous dysplasia is 20–30 years.[9] The incidence is equal in males and female.[1] Radiograph usually reveals a lytic, ground glass lesion consistent with FD.[10] The density and trabecular pattern of fibrous dysplasia lesions vary considerably. The variation is more pronounced in the mandible and more homogeneous in the maxilla. The internal aspect of bone may be more radiolucent, more radiopaque, or a mixture of these two variations compared with normal bone. Early lesions may be more radiolucent than mature lesions and in rare cases may appear to have granular internal septa, giving the internal aspect a multilocular appearance. The abnormal trabeculae usually are shorter, thinner, irregularly shaped, and more numerous than normal trabeculae. This creates a radiopaque pattern that can vary; it may have a granular appearance ("ground-glass" appearance, resembling the small fragments of a shattered windshield), a pattern resembling the surface of an orange (peau d'orange), a wispy arrangement (cotton wool), or an amorphous, dense pattern. A distinctive characteristic is the organization of the abnormal trabeculae into a swirling pattern similar to a fingerprint. Occasionally radiolucent regions resembling cysts may occur in mature lesions of fibrous dysplasia. These are



Fig. 9 – CT axial – posterior ethmoidal and right temporal bone.



Fig. 11 – CT coronal – involvement of right maxilla.

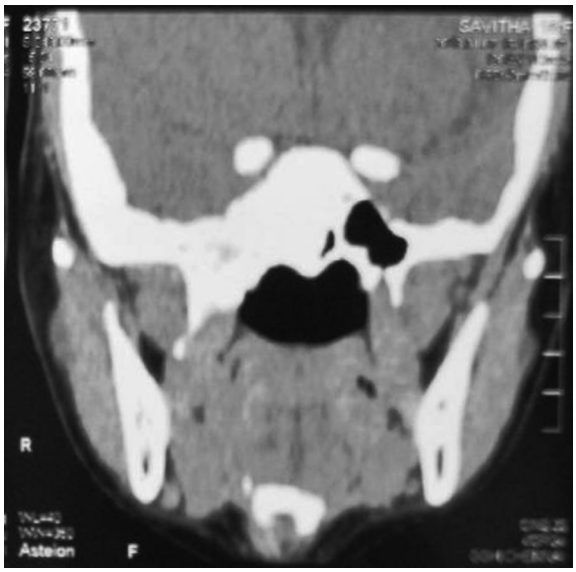


Fig. 12 – CT coronal – involvement of sphenoid bone.

bone cavities that are analogous to simple bone cysts.[8]in the present case a ground glass appearance of trabeculae is observed in intra oral periapical radiograph. FD has characteristic appearance on CT and consists of three varieties, ground glass pattern (56%), homogenously dense pattern (23%), and cystic variety (21%).[11] In this case homogenously dense pattern is seen. Malignant degeneration is reported in less than 1% of cases of fibrous dysplasia, monostotic or craniofacial lesions with the greatest potential.[12] Radiographic features suggestive of malignant degeneration include rapid increase in the size of the lesion, change from a previously mineralized bony lesion to a lytic lesion along with clinical findings of increasing pain and enlarging soft tissue mass.[13,14] Also, a dramatic elevation of serum alkaline phosphatase may herald malignant transformation.[13]

FD is histologically characterized by a benign – appearing spindle cell fibrous stroma containing scattered, irregularly shaped trabeculae of immature (woven) bone, lacking osteoblast, that evolve directly from the stroma.[15]

Conservative management may be indicated if predicated on sound knowledge of a patient's clinical status and

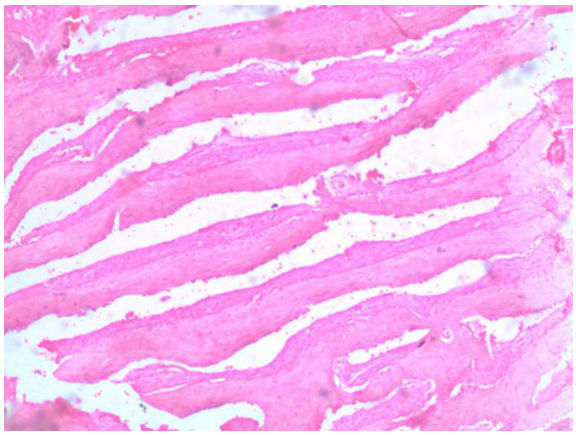


Fig. 13 – Photomicrograph – trabeculae of woven bone.



Fig. 14 – Postoperative extra oral view.

radiological findings. A patient with preserved vision and acceptable or minimal cosmetic deformity may be a candidate for serial clinical and radiological observation. Acute changes heralded by the sudden onset of diplopia, pain, or changes in visual function must be seriously considered and expedient surgical intervention may be warranted.[16]

Generally, a conservative surgical approach including facial osseous debulking and contouring has been recommended for stable disease with minor symptoms. In progressive and more symptomatic cases with functional impairment and cosmetic deformity, radical resection of diseased tissue, optic nerve decompression, and



Fig. 15 – Postoperative intra oral view.

reconstruction of the facial areas with autogenous bone grafts may be needed.[17]

Different studies have reported clinical improvement in patients with fibrous dysplasia after bisphosphonate therapy,[18–20] although there is no histological evidence of abnormal osteoclastic activity in fibrous dysplasia of bone. Medical management of fibrous dysplasia remains poorly characterized. At present, no medical therapy has been clearly proven to cure or impede the progression of fibrous dysplasia. Medications, such as bisphosphonates, calcitonin, and mithramycin, targeted at reducing bone resorption/osteoclastic activity have been prescribed. Preliminary data appear promising although inconclusive.[21,22] Corticosteroid agents have also been used in the care of patients with fibrous dysplasia. Systemic corticosteroids have been used successfully as a temporizing measure in patients with acute visual loss.[23]

In most cases the radiographic characteristics of fibrous dysplasia and the clinical informations are sufficient to allow the practitioner to make a diagnosis without a biopsy. There are reports of exaggerated growth from stimulation of a lesion during surgical intervention in young patients. The prognosis generally is good, although poor outcomes are more frequent in younger patients and in those with polyostotic forms of the disease. The risk of malignant transformation is low.[24]

4. Conclusion

CT can give a more accurate three dimensional representation of the extent of the lesion and can serve as a precise base line study for future comparisions.[8] In female patients, hormonal changes from pregnancy or the use of oral contraceptives may stimulate growth or result in the development of lesions within the area of fibrous dysplasia, such as aneurismal bone cysts or giant cell granulomas.[9] Patient is kept under monitoring and is asked to report any changes. The prognosis is expected to be good as most lesions stop growing after skeletal maturation.

Conflicts of interest

All authors have none to declare.

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