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Craniofacial Polyostotic Fibrous Dysplasia With Shepherd's Crook Deformity of The Proximal Part of The Femur: A Case Report

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ABSTRACT

Craniofacial fibrous dysplasia is one of the benign fibro-osseous lesions which affect the bones of the craniofacial complex, involving the maxilla and mandible. Fibrous dysplasia is a skeletal developmental disorder of the bone-forming mesenchyme that causes a defect in osteoblastic differentiation and maturation. Fibrous dysplasia shows predilection of 5% of all bone tumors and around 7% of all benign tumours. This article presents a unique case of unilateral fibrous dysplasia of the upper and lower jaws, along with the skeletal deformity having shepherd's crook appearance. The clinical features including imaging studies and advanced radiography are described for obtaining a definitive diagnosis.

KEYWORDS : Fibrous Dysplasia, Lichtenstein-Jaffe's disease, Maxillo-mandibular, Monostotic form, Polyostotic form, Craniofacial form.

INTRODUCTION

Lichtenstein[1] in 1938 and Jaffe in 1942[2] first described Fibrous dysplasia as a benign intramedullary fibro-osseous lesion. McCune and Bruch in 1937 first suggested that this disorder should have its own place as a distinct clinical entity. In the successive years, Lichtenstein described this disorder as "fibrous dysplasia". It is a skeletal developmental anomaly of the bone-forming mesenchyme that manifests as a defect in osteoblastic differentiation and maturation. The characteristic feature of this lesion is the proliferation of fibrous tissue inside the medullary bone secondary to bony metaplasia, immature, weakly calcified bone, without osteoblast maturation appearing radiolucent on radiographs, appearing typically as "ground-glass"[3]. Fibrous dysplasia can involve one bone known as monostotic form or multiple bones are known as polyostotic form and are also associated with other conditions. This fibro-osseous lesion develops during skeletal bone formation and growth and has different appearances. The exact etiological cause is unknown. But it is usually caused by the mutation in GNAS1 (Guanine Nucleotide-Binding Protein) gene (20q13.2). As a result, there is a substitution of the histidine-amino acids of DNA in the osteoblastic cells-by arginine which is another amino acid causing osteoblastic cells replacement by fibrous tissue[4]. In fibrous dysplasia, craniofacial involvement is approximately 50% of polyostotic form and 10-25% of monostotic form. The commonly involved bone in the facial skeleton are mandible (12%) and maxilla (12%) showing the involvement of the sphenoid, ethmoid, frontal, and temporal bones[5]. Clinically it shows expansion, thickening, and sclerosis of the bones involved and may result in hearing disturbances, visual complications, tooth displacement and facial asymmetry. Laboratory reports provide a confirmatory diagnosis for polyostotic fibrous dysplasia when alkaline phosphatase are elevated up to 30% and the further rise in level may show malignant degeneration. Malignant degeneration is rare and occurs in less than 1% of cases of fibrous dysplasia. Craniofacial lesions and monostotic form have the highest potential for malignant degeneration[6].

This article presents a case of craniofacial polyostotic fibrous dysplasia in a 28-year-old male patient.

CASE REPORT

28-year-old male patient reported with the chief complains of swelling on the left side of the face since 8years. There was no relevant medical and family history with similar findings. The general physical examination showed a moderately built patient with trendelenburg gait and satisfactory vital signs. Extraoral examination revealed a smooth bony-hard swelling involving left zygoma region with normal overlying skin. Intraoral examination revealed the presence of bilateral expansion of cortical plate in the distal part of the alveolar ridge of lower jaw. The surrounding mucosa was normal in color, without any signs of inflammation or ulceration. There was a multiple number of missing teeth with poor oral hygiene (Figure1 and 2). On palpation affected areas were painless, with hard consistence and smooth surface. The provisional diagnosis based on the clinical features was fibrous dysplasia affecting lower jaw prominently. Further Imaging Studies with extraoral, intraoral radiographs, computed tomography and advanced imaging was made for the accurate definition of the bone density and for obtaining the final diagnosis. The lesion was confined to the bone only without soft-tissue involvement which was helpful in distinguishing it from a malignancy. Periapical radiograph of the lower right first premolar and molar was observed with mixed radio-opaque and radiolucent pattern. The occlusal radiograph further revealed the expansion of the cortical plate with a greyish "ground-glass" pattern that was similar to the density of cancellous bone with no visible trabecular pattern. The radiolucent region was composed of fibro-osseous tissues with a fluid-filled cavity (Figure 2 and 3). Orthopantomogram revealed Mandibular involvement having multilocular radiolucency with "ground-glass" appearance (Figure 4). The CT scan showed a radio-dense mass involving right frontal, temporal, zygomatic regions in the maxilla, and mandible causing facial asymmetry with ground-glass appearance and expansion of involved bones(Figure 5).The x-ray of Pelvic bone (antero-posterior view) revealed radiographically lateral bowing of the proximal part of the thigh and shortening of the limb (Figure 6). Blood samples were taken to measure the levels of serum alkaline phosphatase and serum calcium. Laboratory reports further confirmed the diagnosis with the increased levels of the same. From the involved area, soft tissue and bone were taken for histological examination. The histological findings showed fibrous changes of the bone without any soft tissue involvement. Areas filled with cellular connective tissue stroma consisting of fibroblasts arranged in a whorled pattern were observed. Trabeculae lined by osteoblasts were noticed showing a transition into woven bone surrounded by osteoclasts. Clumps of multinucleated giant cells were also noted near the areas of cystic changes. This

led to the final diagnosis of Fibrous dysplasia. The patient was advised to visit his dentist regularly for re-examination for any change in growth formation and pain. In the re-examine period the level of alkaline phosphatase was also measured.

Discussion

Fibrous dysplasia is a non-hereditary developmental or growth disorder where normal bone is replaced by abnormal fibrous tissue with small, abnormally arranged bone trabeculae. It is considered by some authors to be a hamartomatous malformation that results from an idiopathic arrest in maturation at the woven bone stage[5]. Craniofacial involvement in fibrous dysplasia is seen in monostotic as well as polyostotic forms. Monostotic fibrous dysplasia differs in skeletal distribution from polyostotic disease and occurs most commonly in the femur followed by tibia, craniofacial bones, and ribs[7]. Craniofacial involvement occurs in about 30% of monostotic fibrous dysplasia affecting the maxilla, mandible, and rarely the calvarium. The polyostotic form of the disease shows 100% involvement of the craniofacial bones and also affects the multitude of skeletal bones unilaterally[8].

In this case, the patient was showing an involvement of craniofacial bones along with the pelvic bone. Fibrous dysplasia has its onset during early life, usually in late childhood or early adolescence. Patients with the polyostotic form of the disease are common in younger age group. In the present case, the patient age was 26 years. There is an equal gender distribution in monostotic fibrous dysplasia but the polyostotic form has a clear female predilection[5].But in the present case, the patient was male which is a rare entity. Radiographic features of fibrous dysplasia vary depending upon the amount of bony and fibrous matrix within the lesion and are sub-classified into three different patterns: pagetoid type 56%, sclerotic type 23%, and the radiolucent type 21%[8]. In this case, the radiograph revealed a sclerotic type of lesions; which is seen commonly in the facial bones. The lytic and pagetoid types usually show calvarial bones involvement. Radionuclide scanning in fibrous dysplasia shows areas of intensely increased uptake due to diffuse microscopic ossification. Scintigraphy is helpful in determining the activity and multicentricity of the lesion; it is helpful in diagnosing when plain radiographs are equivocal[9]. Computerized Tomography establishes the extent of bone involvement. Involvement of optic canals, orbital fissures, ostiomeatal complex and frontonasal ducts can be best evaluated by CT scanning. CT characteristics of fibrous dysplasia include expansion of the involved bone with heterogeneous pattern associated with scattered or confluent islands of bone formation. On magnetic resonance imaging, fibrous dysplasia exhibits homogenous, moderately low signal intensity on T1 weighted images. On T2 weighted images, the tissue usually exhibits high signal intensity. Advance imaging modalities such as CT and MRI are excellent imaging in defining the constrictive effect of craniofacial fibrous dysplasia involving the orbit, optic canals, and paranasal sinuses. Surgical excision and recontouring of the affected bone tissue is usually a successful way of treatment. However, it leads to a huge functional and aesthetic deficit, as well as long-term postoperative complications. The conservative therapeutically approach with limited reduction in the size of these lesions is effective in managing the symptoms. Because patients suffering from FD may be at high risk of malignant transformation, periodic follow-up is mandatory to detect such transformation[6]. The surgical approach aims for stable occlusion, facial aesthetics, and evasion of post-operative relapse. Biphosphonates are used few cases when an intervention is necessary but the surgery cannot be performed. Some authors suggest applying calcitonin in combination with surgical treatment. Usually, the prognosis is good although the unacceptable results occur more frequently among young patients or patients with polyostotic forms of the disorder[10].

CONCLUSIONS

Fibrous dysplasia in the maxillo-mandibular region is rare and can be difficult to differentiate from other benign and malignant bone disorders. The general dental practitioner can be the first to detect such conditions mostly when the only affected areas are in the maxillo-mandibular region so sufficient knowledge on this condition is important for the proper diagnosis, treatment, and prevention of further complications. It is mandatory to carry out imaging studies, histological and laboratory tests for obtaining the definite diagnosis, treatment, and management of fibrous dysplasia.

PHOTOGRAPHS ALONG WITH LIGANDS

Figure 1: Extraoral (A) and Intraoral (B) view of the patient

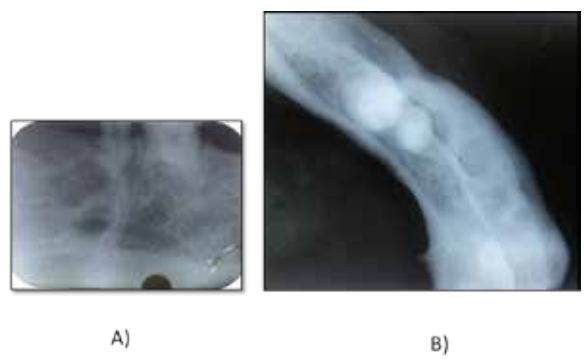

Figure 2:Periapical (A) and occlusal (B) radiograph showing multilocular radiopaque and radiolucent appearence

Figure 3: PA view (A) and Jug-Handle view (B) showing diffuse radiopacity with irregular central opacities and prominent left zygomatic bone

Figure 4: Orthopantomogram revealing Mandibular involvement having multilocular radiolucency with "ground-glass" appearance.

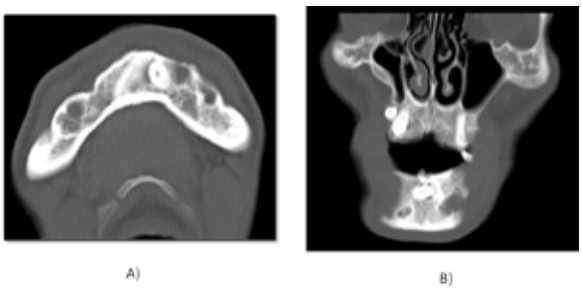

Figure 5: Axial CT (A) image showing expansion of the mandible with radiopaque and radiolucent changes and the coronal CT (B) image showing enlargement of left zygomatic bone

Figure 6: Anteroposterior radiograph of the pelvis with polyostotic fibrous dysplasia and classic shepherd's crook deformity on the left side.

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