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Case report

Craniofacial and monostotic variants of fibrous dysplasia affecting the maxillofacial region

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ABSTRACT

Fibrous dysplasia, a benign fibro-osseous lesion, is a skeletal developmental anomaly of the bone forming mesenchyme that manifests as bony expansion or enlargement due to an abnormal osteoblastic and fibrous connective tissue proliferation and defective osteoblastic differentiation and maturation. Craniofacial fibrous dysplasia is a form of fibrous dysplasia affecting the cranial base, vault and the maxillofacial region to varying extents. Either of the jaws may be involved by the monostotic variety of fibrous dysplasia too. Histopathological examination of a biopsy specimen alone is inadequate to make a definitive diagnosis, but rather, correlation with the clinical features, biological behavior, radiographic appearance and intra-operative findings of these lesions is imperative, so that they can be distinguished from other fibro-osseous lesions and an appropriate treatment modality can be instituted.

1. Introduction

The term "fibrous dysplasia" was first used in 1938 by an American pathologist, Louis Lichtenstein [1]. Fibrous dysplasia is a benign hamartomatous developmental dysplastic disorder involving a process of altered osteogenesis affecting bones of the skeleton, characterized by replacement of the normal bone substance of the interior of bone by fibro-osseous tissue exhibiting varying degrees of osseous metaplasia. It is primarily a developmental abnormality of the bone forming mesenchyme in which fibrous tissue gradually expands and replaces the bone [2]. Patients with fibrous dysplasia exhibit slow, asymptomatic enlargement of one or multiple bones, the pathogenesis of which is a replacement of medullary bone with a fibrous connective tissue proliferation containing variable amounts of osseous matrix that do not mature to lamellar bone. In other words, there occurs an abnormal cellular proliferation of the bone mesenchymal cells, but with their inability to fully differentiate, manifesting as bony expansion comprising of immature woven bone which fails to mature into organized lamellar bone.

Fibrous dysplasia is a congenital anomaly resulting from a post-zygotic mutation of the G protein gene located on the long arm of chromosome 20, the GNAS 1 gene in osteoblastic progenitor cells, leading to increased proliferation and abnormal differentiation of bone. The mutation occurs in somatic cells after conception (post zygotic), either during embryonic development or after birth. The extent and form of Fibrous dysplasia depends on the stage of development and location where the mutation occurs. The earlier during embryogenesis the mutation, the more generalized and extensive the disease [3].

The classic forms of fibrous dysplasia are the monostotic, polyostotic and the craniofacial varieties [4]. Eighty to 85% of patients with fibrous dysplasia have the monostotic form. Monostotic fibrous dysplasia affects only one bone, most commonly the rib, proximal femur etc. The polyostotic variety comprises around 15–25% of cases of fibrous dysplasia and may affect up to 75% of the skeleton. It can further be divided into three types [2]:

(i) Jaffe-Lichtenstein type, in which multiple bones are involved, sometimes accompanied by "café au lait" (coffee with milk) pigmentation of the skin.
(ii) McCune–Albright's syndrome, characterized by an extensive skeletal involvement, café au lait skin pigmentation and multiple endocrinopathies, including gonadal hyperfunction leading to sexual precocity (especially in females), hyperthyroidism, hyperparathyroidism, Cushing's disorder, acromegaly, pituitary adenomas and so on.
(iii) Mazabraud syndrome, characterized primarily by polyostotic fibrous dysplasia and intra muscular myxomas [4].

☆ AsianAOMS: Asian Association of Oral and Maxillofacial Surgeons; ASOMP: Asian Society of Oral and Maxillofacial Pathology; JSOP: Japanese Society of Oral Pathology; JSOMS: Japanese Society of Oral and Maxillofacial Surgeons; JSOM: Japanese Society of Oral Medicine; JAMI: Japanese Academy of Maxillofacial Implants.

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The craniofacial variety of fibrous dysplasia is a form of the disease that affects the maxillofacial region. It is not a restricted to a single bone, but may be confined to a single anatomical site. These lesions primarily affect the maxilla, but may also cross sutures and involve the contiguous or adjacent bones like the sphenoid, zygoma, temporal bone, frontonasal bones and base of the skull. It is almost always unilateral. Craniofacial fibrous dysplasia can cause severe facial deformity and asymmetry, and most devastating of all, blindness by involving the orbital bones and compressing orbital canal contents.

Presentation of fibrous dysplasia is most often during the second and third decades of life with most cases diagnosed before the age of 30. They most often manifest clinically as a slow-growing, painless expansion of the involved bone/bones. Both sexes are equally affected in the monostotic form, but a female predilection has been reported in the polyostotic form [5]. Majority of cases of fibrous dysplasia become quiescent and "burn out" in early adulthood (late teenage years or early twenties) when skeletal maturity has been reached [6]. Some even regress with age. Other cases of FD are either reactivated or first activated by pregnancy, suggesting that sex hormones could influence some of them [7]. Some cases present for the first time even in adulthood and some have even been reported in the 7th and 8th decades [8].

Four cases of fibrous dysplasia affecting the maxillofacial region, seen in patients of three different age groups are presented. Two were classical cases of craniofacial FD, one seen in a young 20-year-old adult male and the other in a 68-year-old elderly female. The other two were cases of monostotic FD affecting the mandible seen in female patients in their mid thirties. The wide age range among these patients may merely reflect the ages the lesions have been first detected, diagnosed and recorded, rather than a later age of commencement of growth, as a late reporting by the patient and hence a late diagnosis is entirely possible as most of the cases of FD are insidious in onset and are painless and asymptomatic throughout their course.

2. Case reports

2.1. Case 1

A 20-year-old male patient reported with the chief complaint of an unsatisfactory facial appearance (Fig. 1A) caused by a swelling in the region of his right cheekbone, which often drew comments from his friends and colleagues. History revealed that he had first noticed the swelling when he was 15 years old, which had slowly but steadily increased in size during his late teenage years, reaching its present size about a year ago, after which it had persisted as such without any further increase in size. There was no history of any trauma to the region and no pain or paresthesia in the affected area. There was also no history of manifestations of partial nasal passage obstruction, such as altered resonance of speech, nasal stuffiness, mouth breathing or sleep apnea. There was no history of epiphora indicating an absence of obstruction of the nasolacrymal apparatus.

On examination, the patient exhibited an obvious facial asymmetry caused by a diffuse unilateral bony expansion of the right zygomatico-maxillary complex (Fig. 1A). There was obliteration of the right nasolabial fold and the right lower eyelid appeared to be elevated as compared to its left counterpart. The exaggerated right malar prominence was even more obvious from the bird's eye view position. There was an outward expansion of the right zygomatic arch as well, which was visible on inspection as well as palpable. There was no evidence of pain or paresthesia in the entire region of the bony expansion and in the area supplied by the right infraorbital nerve. The bony expansion was smooth and diffuse, with normal overlying skin. The inter incisal mouth opening, temporomandibular movements and occlusion were all normal and there was no displacement of the maxillary teeth seen. Differential diagnosis included a benign fibro-osseous lesion, osteoma, etc.

Radiographs (paranasal sinus and submentovertex views) and CT scans (coronal sections) revealed a diffuse expansion of the right maxilla and zygomatic complex with increase in bulk and contour of the right zygomatic arch, body of zygoma and thickening of the zygomatic buttress region (Fig. 1C). The involved bone appeared to have a granular, "ground-glass" like texture and indistinct margins which blended with the adjacent normal appearing bone. There was partial obliteration and a diffuse opacification of the right maxillary antrum. The nasal septum appeared to deviate to the left. The frontal sinus and nasoethmoid regions appeared unaffected. Axial sections of CT scans (Fig. 1D and E) revealed a typical "ground glass" opacification of the base of the skull in the region of the middle cranial fossa. There was contiguous involvement of the basisphenoid, greater and lesser wings of sphenoid including the sella turcica, the zygomatic body and arch, and maxilla on the right side. The lesional bone had ill-defined borders and merged with the adjacent normal bone.

Laboratory investigations like serum calcium, phosphorus and alkaline phosphatase were all within normal limits. $T_{3,T4}$ and TSH levels were checked to rule out endocrinopathies, a possible feature of the McCune-Albright's syndrome. A full body scintigraphy was done using Technetium (^{99m}Tc-MBP) whole body bone scan and a SPECT study of skull bones was carried out, which revealed hot spots indicative of a polyostotic involvement of multiple skull bones on the right side (Fig. 1F). There was no involvement of the rest of the skeleton. A provisional diagnosis of craniofacial fibrous dysplasia was made correlating the clinical presentation with the radiographic and CT, SPECT and Technetium scan findings.

Treatment planned for the patient was a contour excision of the right zygomatico-maxillary region to restore the facial symmetry and esthetics.

An Al Quayat–Bramley approach (Fig. 1G) was employed to expose the zygomatic arch and body, which was reduced in bulk and thickness using vulcanite trimmers under copious saline irrigation. The anterolateral wall of the maxilla and the zygomatic buttress were exposed using an intraoral buccal sulcus approach (Fig. 1H). A segment of lesional bone was excised for histopathological examination (Fig. 1I). The infraorbital nerve was identified and preserved during the shaving and contouring of the expanded bone. The consistency of the bone was softer than normal and could be trimmed and shaved quite easily. The surgical sites were thoroughly irrigated before layer-wise closure. Post operative healing was smooth and uneventful and excellent cosmetic results were achieved (Fig. 1B), with no evidence of recurrence or regrowth even after one year of follow up.

Histopathological examination of the excised bony specimen revealed replacement of normal bone architecture by a richly cellular, densely collagenous moderately vascular fibrocellular connective tissue stroma which was richly populated with numerous uniform-appearing, spindle shaped fibroblasts (Fig. 2A and B). Within the stroma, there were numerous long, fine, slender, delicate, often branching, curvilinear trabeculae of immature woven bone. The bony trabeculae had numerous osteoblasts and osteocytes within them, and also exhibited a distinct osteoblastic rimming. A few basophilic reversal lines were seen within some of the bony trabeculae. There was also seen artifactual separation of many of the trabeculae from the surrounding stroma.

2.2. Case 2

A 72-year-old female patient reported the complaint of a long-standing swelling above her right eye, which had lately become