CRANIOFACIAL POLYOSTOTIC FIBROUS DYSPLASIA : REPORT OF A CASE WITH REVIEW OF LITERATURE

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Fibrous dysplasia is a nonspecific hamartomatous skeletal developmental anomaly, characterized by slow progressive replacement of a localized area of bone by an abnormal proliferation of isomorphic fibrous tissue. The exact etiology of it is in ambiguity. The recent concept with etiopathogenesis proposes that post-zygotic mutation in the guanine nucleotide binding protein á–stimulating activity polypeptide-1 gene. The Fibrous dysplasia can be of monostotic type which is common and affects maxilla, and polyostotic type, a rare variety. The incidence of cranial fibrous dysplasia is even rare amongst the primary bone lesions, representing less than one percent. There is no specific treatment for fibrous dysplasia. Untreated fibrous dysplasias are associated with complications like hearing loss, trismus, and facial disfigurement for which cosmetic surgical treatment is rendered.

Keywords: Fibro-osseous, Metaplasia, Hamartomatous, Taurodontism.

The Fibrous dysplasia is a benign bone lesion characterized by replacement of the normal substance of the interior of the bone by fibro-osseous connective tissue histologically exhibiting varying degrees of osseous metaplasia. It is considered to be a non-neoplastic hamartomatous developmental lesion of unknown origin [1]. A unique case of craniofacial polyostotic fibrous dysplasia which has involved maxilla, mandible and frontal bone causing severe facial disfigurement and that has been successfully managed surgically is being reported.

Case Report

An 18-year-old female reported to the Department of Oral Medicine & Radiology with a gradually increasing painless swelling in the right middle third of the face since 9 years. History of past illness revealed that patient had undergone surgical intervention 3 years ago. Patient also gives a history of similar swellings in the forehead, right maxillary region and right body of mandible. Extra oral examination showed gross facial asymmetry with diffuse, hard, nontender swellings in the right maxillary region, right forehead, and in the right body of mandible (Fig.1), with no local rise in temperature. Intraoral examination showed diffuse swelling in the right maxillary region with bicortical expansion and displacement of the teeth (Fig.2). On palpation it was hard to firm in consistency with no tenderness.



Fig. 1: Clinical photograph showing diffuse swelling in the right middle third of face and right frontal region.

On radiographic examination, orthopanta-mograph showed a mixed radiopaque and radiolucent lesion in the right body of mandible, opacification and obliteration of right maxillary sinus (Fig.3). Para nasal sinus view, Posterior- Anterior skull view, Lateral cephalogram showed opacification in the same region. Occlusal radiographs showed diffuse radiopacity in the right maxillary posterior region (Fig.4) and widened cortices in the right madibular body

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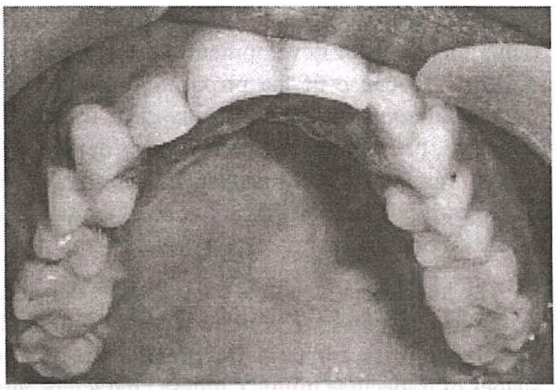


Fig.2: Clinical photograph showing diffuse swelling with bicortical expansion in the right posterior maxillary region and rotation of the 15, 16, 17.



Fig.3: Panoramic radiograph showing mixed radiopaque and radiolucent lesion in the right body of mandible and opacification, obliteration of right maxillary sinus.

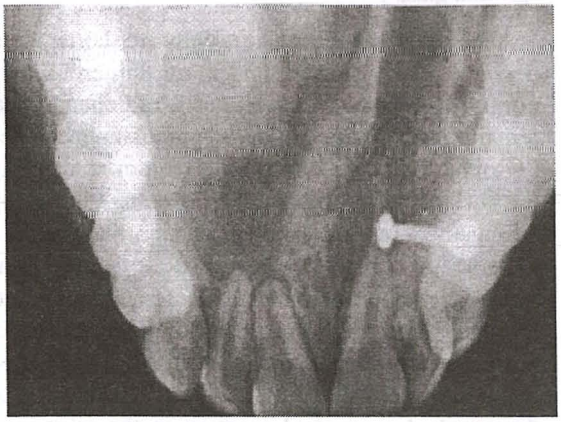


Fig.4: Occlusal radiograph showing diffuse radiopacity in the right maxillary posterior region.



Fig.5: Madibular occlusal radiograph showing widened cortices in the right madibular body.



Fig.6: Intra oral periapical radiograph showing ground glass appearance and loss of lamina dura in relation the teeth.

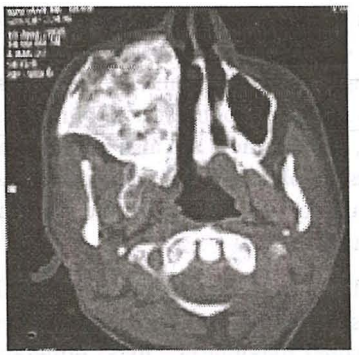


Fig.7: Computed tomography showing multiple lytic lesions of ground glass opacity in the bilateral frontal and multiple right sided faciomaxillary bones with few specks of calcifications.

(Fig.5). Intra oral periapical radiograph showed ground glass appearance and loss of lamina dura in relation the teeth (Fig.6). Computed tomography revealed ground glass opacity in multiple right sided faciomaxillary bones and the mandible with few specks of calcifications within lytic lesions (Fig.7). The biochemical marker, serum alkaline phosphatase was significantly elevated; however serum calcium and phosphorus levels were normal. The histopathological report revealed this lesion as mature fibrous dysplasia. The lesion in the right maxilla and right body of the mandible was surgical recontoured successfully and improvement in the facial appearance of the patient was noted post- operatively.

Discussion

The etiology of fibrous dysplasia is unknown. Currently it is considered to be caused by mutations occurring post-zygotically in the gene coding for the á-subunit of the stimulatory G-protein, in the guanine nucleotide binding alpha stimulating (GNAS) complex locus in chromosome 20q13 [5].

Fibrous dysplasia involves the maxilla almost twice as often as the mandible and occurs more frequently in the posterior aspect, before the age of 30 years. The lesions are painless, firm, expansile, slowly enlarging bony swellings usually occurring unilaterally producing facial asymmetry. Craniofacial fibrous dysplasia is most common in women than in men, in the ratio of 3:1, involving the bones of the face or the base of the skull and the most commonly affected bone is the sphenoid .Leontiasis ossea is a form of facial swelling caused by maxillary expansion with loss of the nasomaxillary angle causing a feline facial appearance. Symptoms such as exophthalmos, blindness, headache, ptosis, and deafness are also noted. Cranial nerve palsies involving glossopharyngeal and vagus nerves due to narrowing of the jugular foramen can occur [3, 7, 9].

Fibrous dysplasia is frequently associated with dental anomalies including rotation, oligodontia, displacement, enamel hypoplasia, hypo mineralization and taurodontism possibly due to activating mutations in tooth development either directly or indirectly because of the proximity of abnormal bone . Unerupted teeth are rarely seen in fibrous dysplasia, probably because most cases develop after tooth eruption. Fibrous dysplasia is often associated with cafe´-au-lait pigmentation of the skin and rarely involving oral mucosa [8].

The histological features composed of curvilinear trabeculae of woven bone surrounded by a moderately cellular fibroblastic proliferation. The shapes of trabeculae mimic Chinese characters, and the bone lacks osteoblastic rimming. The histological features of FD are site-specific; in the cranial bones, they are pagetoid in nature, whereas in the gnathic bones, they are hyper cellular, and in the axial/appendicular skeletons, they have a Chinese-character pattern [11, 12].

Three radiographic patterns of craniofacial fibrous dysplasia as described by Fries (1957) are of a mixed lucent and sclerotic pattern, known as 'pagetoid' seen in 56% of cases , homogeneous sclerotic in 21% and a predominantly cystic appearance with a round or ovoid lucency surrounded by a dense sclerotic boundary in 21% [6]. The characteristic radiographic feature that differentiates fibrous dysplasia from other pathologies is areas of homogeneous grey hazy density, usually described as 'ground glass'.

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), 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. Radiographic margin of FD usually blends with the adjacent unaffected tissue making the outline of the lesion difficult to demarcate.A periosteal reaction in the jaws may strongly suggest sarcomatous transformation [10].

The Computed tomography shows an expansile lesion with ground-glass density in the medullary cavity of the affected bone. Three common patterns are observed on CT: a mixed pattern with lytic and sclerotic regions 55%, homogeneous sclerotic 34%, and predominantly cystic or lytic 11%. The three-dimensional bone reconstruction with the helical CT shows optimal visualization of the extent of FD involving the skull base. Magnetic resonance imaging generally shows an intermediate signal on T1-weighted images and a hypo intense signal on T2-weighted images. The hypo intense signal intensity on T2-weighted images is caused by numerous bony trabeculae. In the early stages of FD, there may be areas of hyper intensity on T2-weighted images [1, 2].

The GNAS mutation can be detected in circulating mononuclear cells, bone cells, melanocytes and endocrine cells. The probability of detection is proportional to the number of mutated cells and the severity of the disease. Looking for the mutation may be useful to establish the diagnosis while avoiding a bone biopsy, or to confirm a pathological result because of the pathological examination is sometimes challenging. Biochemical markers of bone turnover can be used to assess the activity of FD and the response to therapy. The serum osteocalcin, total and bone specific alkaline phosphatase, and C-terminal type I collagen crosslink's (CTX) have been shown to be elevated in active disease and to diminish in response to antiresorptive treatment. [5].

Malignant Transformation

The malignant or sarcomatous transformation of fibrous dysplasia is a rare complication, occurring in 0.4–4% of cases. The craniofacial region is the most common site affected. The commonest types of sarcoma complicating fibrous dysplasia are osteosarcoma, fibro sarcoma and chondrosarcoma[5].

Treatment

Luck (1950) said that "no effective treatment is known" for fibrous dysplasia. The active phase of fibrous dysplasia usually starts in early childhood and gradually becomes quiescent and finally, with termination of skeletal growth, the process becomes inactive and then it ceases to grow. The surgical intervention is generally directed at the cosmetic facial deformities and cranial nerve compression. Surgery is indicated at any age, if important function is threatened, deformity becomes substantial or complications develop. Medical treatment has a role in the management of craniofacial FD. Some authors have reported the use of steroids, mainly in the treatment of visual symptoms from optic nerve compression.

Malocclusion and caries index scores were higher in patients with maxillomandibular FD and the orthodontic tooth movement presented a challenge, so these patients will benefit from an early orthodontic evaluation and regular, judicious dental care to maintain good oral health [4, 7, 8].

Conclusion

FD is a benign progressive disease that has the potential to cause significant cosmetic and functional disturbance, especially visual impairment. Diagnosis of FD must be established based on expert assessment of clinical, radiographic, and histopathological findings. Since lesions in the late stage of FD in the skull base can cause lifethreatening problems, surgery can be done before significant clinical symptoms occur. Periodic CT can be used to follow the progression of the disease and assess the need for any surgical intervention.

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Continued from P. 91

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