REVIEW ARTICLE

Fibrous dysplasia of the craniofacial region - A brief understanding of the disease

Amara Deepthi[1], B. S. Jayanth[1], Fouziya Begum[2], O. Shreyas[3], Akshatha Rao[4]

[1]Department of Oral and Maxillofacial Surgery, Coorg Institute of Dental Sciences, Virajpet, Karnataka, India, [2]Department of Prosthodontics, Coorg Institute of Dental Sciences, Virajpet, Karnataka, India, [3]Department of Cleft and Craniofacial Surgery, DCKH, St. Joseph's Hospital, Mysore, Karnataka, India, [4]Department of Oral and Maxillofacial Surgery, Century International Institute of Dental Sciences and Research Centre, Poinachi, Karnataka, India

Keywords
Congenital, fibrous dysplasia, monostotic, polyostotic fibrous dysplasia

Correspondence
Dr. Amara Deepthi, Department of Oral and Maxillofacial Surgery, Coorg Institute of Dental Sciences, Virajpet - 571 218, Karnataka, India.
Phone: +91-9481951298/+91-9030931298.
E-mail: deepumedico@gmail.com

Received 02 August 2016;
Accepted 20 September 2016

doi: 10.15713/ins.jcri.139

Abstract

Face is the perception of a person's mind and is an important channel of a person's identity. Any abnormality in the face leads to decreased self-esteem of a person. Many syndromes and congenital disorders affect this region of the human body. One such condition which greatly affects the appearance of the face and also sometimes causing functional disturbances is fibrous dysplasia of the craniofacial region. A proper understanding of the disease is required for its appropriate management. This article helps to understand the etiology, pathogenesis, clinical, histologic, radiologic features, and various treatment options available for its management.

Introduction

Fibrous dysplasia (FD) is a non-heritable, genetic disorder in which normal bone is replaced by immature, haphazardly distributed bony and fibrous tissues. The cause of this disorder is a gene mutation that prevents differentiation of cells within the osteoblastic lineage.[1] FD when confined to the bones within the craniofacial skeleton, it is known as craniofacial FD" (CFD).[2] It is a lesion that grows slowly and is often identified incidentally with routine imaging, particularly for monostotic disease, or when gradual swelling and facial asymmetry become noticeable.

Von Recklinghausen coined the term "osteitis fibrosa generalisata" in 1891 to describe this condition in a patient with skeletal deformities. Lichtenstein renamed it as "FD" in 1938. Perhaps the best terminology to describe FD is "fibro-osseous dysplasia" or "fibrous osteodysplasia."[3]

Incidence and Distribution

It is a rare condition known to have an incidence of 1:4000-1:10,000 and represent 7% of all benign bone tumors and has a slight female predilection. It usually presents in the first three decades of life and usually stabilizes after reaching skeletal maturity. In few individuals, it persists even after three decades owing to the nature of the disease. Maxilla is involved almost twice frequently as the mandible. Usually, unilateral side is involved and more commonly in the posterior region.[3]

Types of FD

Monostotic FD

This type is the most common and is seen in 70-80% of cases. A monostotic variant involves only single bone. It is usually less severe form, and in the craniofacial area, it involves the mandible (12%), maxilla (12%) and other long bones like the femur (17%), ribs (24%) or tibia (13%).[4,5] In skull, it commonly involves the ethmoid, sphenoid, frontal, and the temporal bones.[6] These locally extend to the maxillary sinus, orbital floor, zygomatic process, and skull base involvement which results in malocclusion and significant facial asymmetry. They are not well circumscribed. Thus, they are not true monostotic lesions and can be best described as "CFD." It usually has a

delayed presentation compared to polyostotic variant, if it is not associated with endocrinopathies.[7]

Polyostotic FD

It is reported in almost 20-30% of cases and involves more than one bone. In most of these cases, head and neck are involved.

Types of polyostotic FD:

a. Jaffe-Lichtenstein syndrome (Jaffe type): In this type, FD involves a variable number of bones and is also accompanied by pigmented skin lesions or café-au-lait spots.
b. McCune-Albright syndrome (MAS): In this type, almost all skeletal bones are involved. They also present with pigmented skin or oral mucosal lesions and various endocrine disturbances. It is the most severe form of FD. It is seen in almost 3% of cases.[4,7]
c. Mazabraud syndrome: In this type of FD, the disease is associated with intramuscular myxomas.

Pathogenesis

The pathologic basis for FD as mentioned by Ricalde *et al.* is a post-zygotic mutation in the GNAS-1 gene located on chromosome 20, which results in the formation of excess 30, 50-cyclic adenosine monophosphate (cAMP) in mutated cells. The increase in cAMP might prevent the differentiation of cells within the osteoblastic lineage. The mutation occurs in the 201 position, which is usually occupied by an arginine (R201), but in FD, is replaced by either a cysteine (R201C), a histidine (R201H), or a guanine (R201G). GNAS-1 mutation and subsequent cAMP excess impairs the ability of cells in the osteoblastic lineage to fully differentiate. The condition of cAMP excess contributes to the mutated osteoblastic cells overexpressing interleukin (IL)-6, which in turn activates surrounding osteoclasts, and thereby expands the bone lesions. The accepted explanation for the variability in the severity and extent of the disease is related to the stage at which the post-zygotic mutation occurred.[1]

Clinical Features

The majority of the cases involving the jaws first present around the age of 25 years and overall at 15 years age. The most common presenting symptom in the craniofacial region is a gradual, painless enlargement of the involved bones evident as facial asymmetry. Swelling is seen mostly in cases involving the jaws.[8] If there is obliteration of bony cavities or constriction of foramina: Proptosis, diplopia, orbital dystopia, blindness, epiphora, strabismus, tinnitus, hearing loss, nasal obstruction, facial paralysis, etc., may be evident.[3] Pain is not an initial presenting symptom but might occur later due to the disease obstructing the sinus or compressing the nerves passing through various foramina.[4] Resorption of roots is rarely observed. Loss of lamina dura is seen in the radiographs of the affected region. Although the possibility of pathological fracture is highest in polyostotic cases, especially in MAS, de Mattos *et al.* reported that even 50% of monostotic cases fracture. The most cases involving the jaws do not become "quiescent during puberty," typical of extragnathic cases of FD.[8]

Histologic Features

The lesion is mainly fibrous, consisting of proliferating fibroblasts arranged in a compact stroma of interlacing collagen fibers with irregular trabeculae of bone scattered throughout the lesion without a definite pattern or arrangement. The main features are delicate trabeculae of immature bone without any osteoblastic rimming enmeshed in a fibrous stroma depicting a "Chinese letter" pattern. Coarse woven bone replaces the well-organized lamellar bone.[4,9]

Radiological Features

Plain radiography, computed tomography (CT)-scans or magnetic resonance imaging of the lesion, shows a poorly defined lesion which merges with the adjacent bone. Lesions at early stage appear radiolucent. They later become more radiopaque and present with "ground glass appearance" or diffuse radiopacity.[9] The "ground glass" appearance was apparent only in approximately 38% of cases on plain radiographs, in contrast to 100% on CT. Nevertheless, CT readily displays the full extent of the lesion, especially anatomically complex areas. The maxillary sinus is involved in almost all cases. Usually, fusiform expansion is seen in the mandible and enlargement of the normal contour in the maxilla. All cases reported buccolingual expansion and displacement and/or thinning of the lower border of the mandible. Teeth are displaced in approximately 35% cases.[8]

Three distinct patterns were described by Panda *et al.*[3,10] The pagetoid type (on CT imaging) characterized by bone expansion and scattered islands of bone formation in a low-attenuation field. The sclerotic type with homogeneous ground-glass appearance. The cystic type is appearing as a well-defined low-attenuation lesion with a sclerotic margin.

Diagnosis

Diagnosis cannot be relied on the histologic appearance alone. A proper diagnosis is based on the history, clinical features, radiographic, and operative findings along with the histologic appearance.[9]

FD needs to be distinguished from other benign fibro-osseous lesions like ossifying fibroma.[3]

Treatment Options

Treatment is centered mainly to address cosmetic or functional reasons. The surgical approaches vary from careful monitoring and waiting for cessation of growth of the lesion after puberty

to conservative or radical resection.[10] The surgical treatment is mainly targeted to correct the facial deformity with the restoration of the obliterated foramina (in cases where they cause problems such as visual disturbances, orbital dystopia, proptosis and nasal malfunction).

Follow-up is required for lesions arising secondarily within the FD. The two most important are aneurysmal bone cysts and osteosarcomas.[3,8]

The treatment options may be basically divided into four categories:

1. Observation
2. Medical therapy
3. Surgical remodeling
4. Radical excision and reconstruction.[3]

Small asymptomatic lesions which are cosmetically acceptable could be monitored with observation alone.

Patients with large symptomatic lesions may be managed using long-term bisphosphonates. They have been proven effective to reduce symptoms and also to increase the cortical bone thickness. They retard bone erosion by inhibiting the action of osteoclasts. The reason is that the drug stabilizes the lesion and thereby decreases the patient's pain.[3] Large, symptomatic or critical lesions may be managed using intravenous bisphosphonates like pamidronate or zoledronic acid. Medical therapy has not occupied a prominent role for the management of these lesions till to date. Steroids are currently used as supportive therapy in few cases.[11]

Conservative surgery is done as a remodeling procedure to address the disfiguring deformity and thereby to achieve acceptable aesthetics.[8] Long-term follow-up of the patient is important after the conservative surgery. Stabilization of the lesion usually occurs once the bone maturation completes.

Surgery is generally indicated if there is a threat to vision or if the disease involves the optic canal.[8] Nowadays, most authors prefer radical surgical therapy, which permits complete removal of the lesion followed by immediate reconstruction.[11]

Due to the recurrence associated with this disease, opinions vary among the surgeons treating FD. Serum alkaline phosphatase levels (ALP) are important markers for detecting the recurrence of this disease.[12] If the post-operative serum ALP level does not increase following the complete resection of the lesion, then the chance of recurrence is very less.[3]

Partial resection could be considered in patients, whose condition does not allow complete resection or if the patient is older than 17 years. Here, regrowth is not much probable and surgery is done only to address the aesthetic issues.[3]

Chen and Noordhoff proposed a treatment algorithm incorporating aggressive and radical surgery for the management of CFD.

They divided the head and face into four zones based on the aesthetic and functional consequences of the disease along with consideration of anatomic factors for surgery in each area.[3,11,13]

Zone 1 includes the fronto-orbito-malar regions of the face which are esthetically very important. They can be adequately reconstructed with simple bone grafting techniques after excision. For this region, they recommended radical excision and reconstruction.

Zone 2 includes the region of the hair-bearing scalp. Since it is not an area of primary aesthetic concern, as such, intervention is optional for the patient.

Zone 3 includes the central skull base with the pterygoid, sphenoid, mastoid, and petrous temporal bone. Only observation is recommended here, due to the difficulty associated to obtain access to these areas during the surgery.

Zone 4 includes tooth bearing regions of the skull, maxilla, and the mandible. Conservative management should be considered, due to the difficulty in reconstructing the defects in this region.

Reconstruction (using iliac crest, rib, and calvaria grafts or revascularized free flaps) is generally performed immediately to avoid functional disturbances and also restore the facial form.[11,14,15] Some authors remove thin bone, remodel, and re-implant these dysplastic bone grafts for reconstruction of the surgical defects, as they offer an excellent platform for the migration of osteoblasts from the surrounding normal bone.[11,16]

Conclusion

CFD is one of the distinctive entities in the region of the head and neck. Proper treatment planning and long-term follow-up play a key role. Although many treatment options are available for the management of these lesions, complete success could not be achieved with any procedure. There is still, a need to study this disease further for the better understanding and management of this disease.

References

1. Ricalde P, Magliocca KR, Lee JS. Craniofacial fibrous dysplasia. Oral Maxillofac Surg Clin North Am 2012;24:427-41.
2. Dicaprio MR, Enneking WF. Current concepts review Fibrous dysplasia. J Bone Joint Surg Am 2005;87:1848-64.
3. Menon S, Venkatswamy S, Ramu V, Banu K, Ehtaih S, Kashyap VM. Craniofacial fibrous dysplasia: Surgery and literature review. Ann Maxillofac Surg 2013;3:66-71.
4. Bhattacharya S, Mishra RK. Fibrous dysplasia and cherubism. Indian J Plast Surg 2015;48:236-48.
5. Eversole LR. Craniofacial fibrous dysplasia and ossifying fibroma. Oral Maxillofac Surg Clin North Am 1997;9:625-42.
6. Mohammadi-Araghi H, Haery C. Fibro-osseous lesions of craniofacial bones. The role of imaging. Radiol Clin North Am 1993;31:121-34.
7. Bhadada SK, Bhansali A, Das S, Singh R, Sen R, Agarwal A, *et al.* Fibrous dysplasia & McCune-Albright syndrome: An experience from a tertiary care centre in North India. Indian J Med Res 2011;133:504-9.
8. MacDonald DS. Maxillofacial fibro-osseous lesions. Clin Radiol 2015;70:25-36.
9. Prabhu S, Sharanya S, Naik PM, Reddy A, Patil V, Pandey S, *et al.* Fibro-osseous lesions of the oral and maxillo-facial region: Retrospective analysis for 20 years. J Oral Maxillofac Pathol 2013;17:36-40.
10. Panda NK, Parida PK, Sharma R, Jain A, Bapuraj JR.

A clinicoradiologic analysis of symptomatic craniofacial fibro-osseous lesions. Otolaryngol Head Neck Surg 2007;136:928-33.
11. Valentini V, Cassoni A, Marianetti TM, Terenzi V, Fadda MT, Iannetti G. Craniomaxillofacial fibrous dysplasia: Conservative treatment or radical surgery? A retrospective study on 68 patients. Plast Reconstr Surg 2009;123:653-60.
12. Park BY, Cheon YW, Kim YO, Pae NS, Lee WJ. Prognosis for craniofacial fibrous dysplasia after incomplete resection: Age and serum alkaline phosphatase. Int J Oral Maxillofac Surg 2010;39:221-6.
13. Chen YR, Noordhoff MS. Treatment of craniomaxillofacial fibrous dysplasia: How early and how extensive? Plast Reconstr Surg 1990;86:835-42.
14. Munro IR, Chen YR. Radical treatment for fronto-orbital fibrous dysplasia: The chain-link fence. Plast Reconstr Surg 1981;67:719-30.
15. Edgerton ME, Persing AJ, Jane JA. The surgical treatment of fibrous dysplasia. Ann Surg 1985;202:459-79.
16. Ricalde P, Horswell BB. Craniofacial fibrous dysplasia of the fronto-orbital region: A case series and literature review. J Oral Maxillofac Surg 2001;59:157-67.

How to cite this article: Deepthi A, Jayanth BS, Begum F, Shreyas O, Rao A. Fibrous dysplasia of the craniofacial region - A brief understanding of the disease. J Adv Clin Res Insights 2016;3:205-208.