Case Report

FIBROUS DYSPLASIA OF MAXILLARY BONE: A CASE REPORT

Sonam Bhalla¹, Poonam Goel², Sneha Sethi²

Department of Oral and Maxillofacial Pathology, ¹Bhojia Dental College and Hospital, Bhud, Nalagarh, Distt. Solan, Himachal Pradesh, ²M.M College of Dental Sciences and Research, Mullana, Haryana.

ABSTRACT:
Fibrous dysplasia is a rare benign intramedullary fibro-osseous lesion. It is a genetic noninherited condition caused by mutation in the GNAS1 gene, characterized by abnormal proliferation of fibrous tissue interspersed with normal or immature bone. It occurs in equal proportions in males and females, most often during the first two decades of life. Long bones, skull bones and ribs are the most commonly affected bones. The radiological picture is somewhat variable, including a ground-glass appearance, expansion of the bone and sclerosis surrounding the lesion. Histologically, fibrous dysplasia shows irregularly-shaped trabeculae of immature, woven bone in a background of variably cellular, loosely arranged fibrous stroma. Computed Tomography is the best technique for demonstrating the radiographic characteristics. Here we report a case of fibrous dysplasia in 35 year old male patient.

Key Words: Fibrous dysplasia, maxilla, craniofacial, computed tomography

Corresponding Author: Dr. Sonam Bhalla, Senior lecturer, Department of Oral and Maxillofacial Pathology, Bhojia Dental College and Hospital, Bhud, Nalagarh, Distt. Solan, Himachal Pradesh. Email: sonam.c811@gmail.com


INTRODUCTION
Fibro-osseous lesions of the maxillofacial bones comprise a diverse group of pathologic conditions that include developmental lesions, reactive or dysplastic diseases, and neoplasms, characterized by replacement of normal bone by fibrous tissue containing a newly formed mineralized product. Fibrous dysplasia is a benign intramedullary fibro-osseous lesion originally described by Lichtenstein in 1938¹ and by Lichtenstein and Jaffe in 1942². They are reported to represent approximately 5% to 7% of benign bone tumors.³,⁴ Fibrous dysplasia can present in one bone (monostotic) or multiple bones (polyostotic) and can be associated with other conditions. Craniofacial involvement in fibrous dysplasia is seen in both monostotic and polyostotic forms. Craniofacial involvement occurs in about 30% of monostotic fibrous dysplasia and typically affects the maxilla, mandible and rarely, the calvarium. Polyostotic form of the disease has nearly 100% involvement of the craniofacial bones. The lesions of fibrous dysplasia develop during skeletal formation and growth. Clinical presentation of fibrous dysplasia varies with the primary bone involved and the extent of disease. Fibrous dysplasia has its onset during early life, usually in late childhood or early adolescence. There is an equal distribution in monostotic fibrous dysplasia among the sexes, but the polyostotic form has a clear female predilection. The reason is unknown.

CASE REPORT
A 35 year-old male reported to our department. He developed a swelling on the left side of the face that had persisted for one year. There was no history of pain, trauma, epistaxis, loosening of teeth, trismus, or diminished vision. Intraoral examination of the region revealed a smooth bony hard swelling involving the left maxilla extending from central incisor to first premolar region. Superoinferior dimension at incisor canine region was less than 2 cm and anteroposterior dimension was about 2.5 cm. Buccal cortical plate was expanded. On palpation the consistency was bony hard, non-tender & no local rise of temperature. The skin over the swelling was normal. The swelling was bulging into the gingivo-
buccal sulcus area. Central incisor was extruded and rest of the teeth were normal.

**Figure 1:** Intraoral picture showing swelling in left anterior region of upper jaw

The blood and urine investigations were normal. The computed tomography (CT) scan showed evidence of expansion of left side maxilla involving the alveolus, lesion showed heterogenous density with sclerotic and lytic areas and thin expansile cortex.

**Figure 2, 3 and 4:** Showing expansion of left side of maxilla and involvement of alveolar bone

An incisional biopsy was subsequently performed. Histopathological examination revealed presence of hypercellular connective tissue stroma, composed of irregular outlined bony trabeculae with lacunae containing osteocytes. Few areas shows irregular outlined bony trabeculae rimmed by osteoblast cells. Few giant cells with hyperchromatic nuclei, resembling osteoclast like cells are also seen.

**Figure 5:** Hypercellular connective tissue stroma showing scattered irregular outlined bony trabaculae

**Figure 6:** Bony trabeculae showing lacunae containing osteocytes

Based on the clinical history, radiographic and histologic features of the lesion, a diagnosis of craniofacial fibrous dysplasia was established.

**DISCUSSION**

Fibrous dysplasia is postulated to occur as a result of a developmental failure in the remodeling of primitive bone to mature lamellar bone and a failure of the bone to realign in response to mechanical stress. Failure of maturation leaves a mass of immature isolated trabeculae enmeshed in dysplastic fibrous tissue that are turning over constantly but never (or very slowly) completing the remodeling process and the immature matrix does not mineralize normally.\(^5\) The combination of a lack of stress alignment and insufficient mineralization results in
substantial loss of mechanical strength, leading to the development of pain, deformity and pathologic fractures.

This genetic non-inherited condition is caused by missense mutation in the gene GNAS1 on chromosome 20q13.2-13.3, that encodes the alpha subunit of the stimulatory G protein-coupled receptor, Gsa. The activating mutations occur postzygotically, replacing the arginine residue amino acid with either a cysteine or a histidine amino acid. The mutation selectively inhibits GTPase activity, resulting in constitutive stimulation of AMP-protein kinase A intracellular signal transduction pathways. The systemic manifestations of the mutated Gsa protein-coupled receptor complex include autonomous function in bone through parathyroid hormone receptor; in skin through melanocyte-stimulating hormone receptor; in ovaries through the follicle-stimulating hormone receptor; and in the thyroid and the pituitary gland, through the thyroid and growth hormone receptors respectively. All cells that derive from the mutated cells manifest the dysplastic features.

The clinical presentation varies depending on where in the cell mass the mutation is located and the size of the cell mass during embryogenesis when the mutation occurs. Severe disease may be associated with an earlier mutational event that leads to a larger number or a more widespread distribution of mutant cells. Polyostotic fibrous dysplasia can affect bones derived from mesoderm or neural crest, and is associated with pregastrulation mutation. The sporadic occurrence of these diseases and the characteristic lateralized pattern of skin and bone involvement in the polyostotic forms of fibrous dysplasia suggest this mosaic distribution of abnormal cells. The Gs α mutation was first identified in patients with McCune-Albright syndrome, a rare disorder that combines polyostotic fibrous dysplasia, skin pigmentation, and one of several endocrinopathies. The postnatal manifestation of fibrous dysplasia is seen in both monostotic and polyostotic forms. Monostotic fibrous dysplasia has a different skeletal distribution from polyostotic disease and occurs most commonly in the femur, followed by occurrences in the tibia, craniofacial bones, and the ribs. Craniofacial involvement occurs in about 30% of monostotic fibrous dysplasia and typically affects the maxilla, mandible, and rarely, the calvarium. Polyostotic form of the disease has nearly 100% involvement of the craniofacial bones. Monostotic presentation is more frequent, and lesions enlarge in proportion to skeletal growth. The polyostotic form is less common. By early adolescence, patients with widespread polyostotic fibrous dysplasia may have severe deformities. Polyostotic lesions often continue to enlarge after skeletal maturity, with progressive deformity and an increase in pathologic fractures. Precocious development of secondary sexual characteristics is the most common endocrine presentation in patients with McCune-Albright syndrome. Compared with bone lesions in patients without McCune-Albright syndrome, the skeletal lesions in patients with the syndrome tend to be larger, more persistent, and associated with more complications. Café au lait areas of skin pigmentation frequently are found about the trunk or the proximal parts of the extremities of these patients. These areas have a variegated border that resembles the coast of Maine as opposed to the smooth-bordered (coast-of-California) café au lait areas characteristic of diffuse neurofibromatosis or von Recklinghausen disease. Another rare disorder seen with fibrous dysplasia is Mazabraud syndrome, in which skeletal lesions of fibrous dysplasia are combined with intramuscular myxomas.

CONCLUSION

Fibrous dysplasia is a benign lesion, which rarely affects the head and neck region. CT scan is the investigation of choice. Conventional surgical approach gives better access for complete removal of the lesion.

REFERENCES

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abnormal pigmentation of skin, premature sexual development, hyperthyroidism or still other extraskeletal abnormalities. Arch Pathol. 1942;33:777-816.

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