Fibrous dysplasia: report of an unusual case

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Abstract: Benign fibro-osseous lesions (BFOLs) of the maxillofacial bones represent a diverse group of pathologic conditions that includes developmental lesions, reactive or dysplastic diseases, and neoplasms. A great deal of controversy surrounding these lesions as to whether they represent distinct entities or a single entity in different stages. It has now been accepted, following genetic studies that these are actually distinct entities having a similar histopathologic appearance. Fibrous Dysplasia is an idiopathic fibro-osseous disease which causes progressive expansion and deformity of bones. Café-au-lait spots have been reported to occur with the polystotic form of the disease but not with the monostotic form. However, it has also been suggested that the abnormal maturation of bones proceeds synchronously in the affected bones of the craniofacial complex, and thus exists in a silent or subclinical stage in apparently normal appearing bones as well. Reported here is a case of Fibrous Dysplasia of the Maxilla in a 12 year old boy with café-au-lait spots over the neck and trunk region. Other bones did not appear to be affected by the disease at this stage. A long term follow-up is required to determine whether other bones will be subsequently affected or not.

Key words: Benign fibro-osseous lesions, Fibrous Dysplasia, monostotic, polystotic

I. Introduction

The Benign Fibro-Osseous Lesions (BFOLs) are a group of clinically and radiologically distinct lesions characterized histologically by the replacement of normal trabecular bone by fibrous connective tissue with an admixture of mineralized product, including osteoid, mature bone and / or cementum like calcification. It is important to subclassify the lesion, as distinct Benign Fibro-Osseous Lesions have different therapeutic protocols.

Fibrous dysplasia is defined as a benign lesion, presumably developmental in nature, characterized by the presence of fibrous connective tissue with a characteristic whorled pattern and containing trabeculae of immature non-lamellar bone. It was first described in 1981 by von Recklinghausen as “Osteitis Fibrosacystica”. After a plethora of nomenclature over the years, the term “Fibrous Dysplasia of Bone” was coined in 1938 by Lichtenstein. In 1942, Jaffe described the clinical manifestations of Fibrous dysplasia. The lesion may be monostotic or polystotic. The craniofacial bones and the proximal femur are commonly involved. The lesion usually arises in the first or second decade, manifesting as a slow growing, painless lesion of the involved bone. Radiographically, the lesion presents as an ill defined radiolucency with poorly discernible borders that tend to blend with the surrounding unaffected bone. The lesion often shows a typical “ground glass” or “orange peel appearance” due the presence of woven bone. Histologically, the lesions are characterized by replacement of normal bone by cellular fibrous connective tissue having a characteristic whorled pattern and trabeculae of immature non-lamellar bone in a “Chinese letter” pattern. The trabeculae of bone typically show an absence of osteoblastic rimming.

The radiographic differential diagnosis includes non-ossifying fibroma, osteofibrous dysplasia, aneurismal bone cyst, giant cell tumor and low grade central osteosarcoma. Histological differential diagnosis includes non-ossifying fibroma, osteofibrous dysplasia, fracture callus, and low grade osteosarcoma. The disease often runs a benign clinical course and becomes dormant by adulthood. However, 1% of cases show malignant transformation and therefore all cases must have an yearly follow up. Computed tomography scans are helpful in recognizing malignant change at an early stage and also give a better visualization of the extent of the lesion.

II. Case Report

A 12 yr old boy visited our private practice with chief complaint of protrusion of upper front teeth. The patient had come for esthetic correction by orthodontic treatment. On examination, the extra oral frontal view revealed an obvious facial asymmetry with the patient’s left side enlarged. A significant amount of the labial vestibule was visible extraorally on the left side, the ala of the nose on the left side was more flattened and the
medial canthus of the left eye more away from the midline (Fig 1). The TMJ examination showed mild hypomobility on the right side on opening of the mouth.

A detailed intra oral examination revealed a solitary unilateral fusiform type of expansion of the buccal cortical plate on the left maxillary posterior teeth region extending from distal of maxillary left first premolar to the distal of maxillary left second molar (Fig 2). The margins of the lesion were blending smoothly with the adjacent normal mucosa. The mucosa covering the expansion was of a normal texture and colour. The area was firm and non tender on palpation. Teeth were placed in normal occlusion with slight rotation of maxillary left second premolar. Teeth in the affected area were not mobile.

Two large irregularly outlined pigmented macules (Cafe au lait spots) were observed on the lower right abdominal region adjacent to the iliac crest (Fig 4). Also, several smaller macules, measuring approx. 1cm or less in diameter were seen over the trunk, lateral and posterior neck region. The patient did not complain of any discomfort associated with the pigmented regions.

Computed Tomographic examination of the maxilla showed a diffuse, mixed radiolucent – radiopaque appearance mimicking a ground glass appearance in the left posterior region (Fig 3). Margins of the mixed radiolucent – radiopaque lesion were not distinct, and the lesion was seen blending imperceptibly with the surrounding bone. The periodontal ligament space of the teeth in the involved area showed narrowing at some places. The normal radiolucent appearance of the left maxillary sinus was not traceable.

Keeping in mind the young age of the patient, the patient was advised regular monitoring and recall. It was decided that surgical intervention would be considered after active growth phase is over. The patient has been on recall for 2 years with no significant increase in the size of the lesion, or presentation of new lesions elsewhere in the body.

III. Discussion

Fibro osseous lesions are a complex and diverse group of lesions characterised by replacement of normal bone by fibrous tissue containing a newly formed mineralised product. Even though earlier workers considered the Benign Fibro-osseous Lesions to represent a spectrum of the same disease entity, there is remarkable evidence available now in support of these lesions being considered as individual entities.

Fibrous dysplasia has a distinct molecular pathogenesis that is postzygomatic activating somatic mutations in GNAS1gene. This gene is located on band 20q13. This gene encodes the α subunit of the G-protein which stimulates the production of cAMP. The cAMP is thought to have an effect on the differentiation of osteoblasts. Two distinct missense mutations in the α subunit of a G-stimulatory protein probably leads to constitutive activation of cyclic adeny l cyclase, resulting in a persistent elevation of cyclic adenosine monophosphate. Cafe-au-lait spots are formed by the overproduction of tyrosinase enzyme, which is the rate limiting step in melanin production. This mutation causes hyperproliferation and incomplete differentiation of marrow stromal cells to abnormal osteoblasts. cAMP also activates Fos, which inhibits osteoblastic-specific genes as well as activates cytokines that promote bone resorption by osteoclasts.

If the mutation occurs in one of the undifferentiated stem cells during early embryonic life, the osteoblasts, melanocytes and endocrine cells that represent the progeny of that mutated cell, all will carry that mutation and express the mutated gene. The clinical presentation of multiple bone lesions, cutaneous pigmentation and endocrine disturbances would result. Skeletal progenitor cells at later stages of embryonic development are assumed to migrate and differentiate as part of the process of the normal skeletal formation. If the mutation occurs during this later period, then the progeny of the mutated cell will disperse and participate in the formation of the skeleton resulting in multiple bone lesions of Fibrous dysplasia. If the mutation occurs during post-natal life, then the progeny of that mutated cell are essentially confined to one site resulting in Fibrous dysplasia affecting a single bone. It is for this reason that polyostotic forms of the disease are commonly associated with cafe-au-lait spots and endocrine disorders, while they are absent in monostotic forms.

Cafe-au-lait spots have not been reported to occur in the craniofacial form of fibrous dysplasia. Presented here is a case report of a young boy showing clinical and radiographic involvement of only a single bone (the maxilla), but also showing distinct cafe-au-lait spots, which are usually reported along with the polyostotic form of the disease. It may, therefore, be hypothesized that a defect in the undifferentiated mesenchymal cells occurred at a stage when endocrinal cells had already differentiated.

References

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References:

Legends For Pictures
Fig 1: Extra oral frontal view revealed a facial asymmetry
Fig 2: Expansion of the buccal cortical plate on the left maxillary posterior teeth region extending from distal of maxillary left first premolar to the distal of maxillary left second molar.
Fig 3: CT view of the maxilla showed a diffuse, mixed radiolucent – radiopaque appearance mimicking a ground glass appearance in the left posterior region.
Fig 4: Two large irregularly outlined pigmented macules lower right abdominal region adjacent to the iliac crest.