CASE REPORT

FIBROUS DYSPLASIA: BRIEF REVIEW IN LIGHT OF A CASE REPORT

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ABSTRACT

Fibrous dysplasia (FD) of monostotic type is the most common type, usually asymptomatic and becomes inactive at puberty. We present a case of 40 year old lady presented with pain over right hip region for few years. X-ray, bone scan and MRI revealed a solitary lesion over right femoral neck suggestive of FD. She underwent surgical curettage and internal fixation and histopathology turned out to be FD. In view of this case report we present a pictorial essay encompassing various clinical aspects of FD.

Key words: Fibrous dysplasia; monostotic; polyostotic; McCune-Albright syndrome.

Introduction

Fibrous dysplasia (FD) is a developmental disorder in which normal bone marrow and cancellous bone is replaced by immature bone and fibrous tissue. FD represents approximately 2.5% of all bone lesions, as well as 7% of all benign bone tumors.[1] The distribution shows minimal preference towards the female sex (M: F = 1:1.2).[2]

Historical Facts

The first description of FD was presented by Weil in 1922.[3] Later on in 1937, Albright et al[4] reported a syndrome characterized by precocious puberty, areas of skin pigmentation, endocrine abnormalities, and fibro-osseous lesions of bone. Same year, McCune and Bruch[5] reported a child with similar clinical findings (McCune-Albright Syndrome). However, the term FD was presented by Lichtenstein[6] in 1938 while in 1942, Jaffe[7] separated the disease into its principal clinical forms - polyostotic and monostotic.

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Etiology

Mutation in the gene that encodes the subunit of a stimulatory G protein (Gsá) located on chromosome 20 is considered the basic reason.[8] This mutation results in a substitution of the cysteine or the histidine-amino acids of the genomic DNA in the osteoblastic cells-by another amino acid, arginine.[9] These osteoblasts elaborate a fibrous tissue in the bone marrow instead of normal bone. Cancellous bone maintenance is perturbed, and bone undergoing physiologic remodeling is replaced by an abnormal proliferation of fibrous tissue. The extent and pattern of disease depend on the stage of development and the location at which the mutation occurs. All the bones can be affected.

Clinical Presentation

The monostotic form of FD comprises approximately 80% of all cases and is seen in patients between 10 and 70 years old. The most common sites of involvement include femur, rib, tibia, mandible, skull, and humerus. Uncomplicated monostotic lesions are generally asymptomatic and usually do not cause

significant deformity. As a rule, monostotic FD does not convert to the polyostotic form, lesions do not increase in size over time, and the disease becomes inactive at puberty.[10] The polyostotic form of FD comprises 20% of all cases and involves many or few bones, most commonly the skull and facial bones, pelvis, spine and shoulder. It is often unilateral and tends to involve larger segments of bone and is frequently associated with fractures and severe deformities. The shepherd's crook deformity is a common bowing deformity with varus angulation of the proximal femur. Although it generally becomes quiescent at puberty, but existing deformities may progress. The term "leontiasis ossea" describes a rare form of polyostotic disease that involves the frontal and facial bones and results in marked deformities resembling a lion's face.[11]

McCune-Albright syndrome is an endocrinopathy occurring mainly in girls, consisting of the triad of precocious puberty, polyostotic FD (more disabling than pure polyostotic disease), and characteristic cutaneous pigmentation ("café au lait").[12] Mazabraud syndrome is the rare combination of FD and soft-tissue myxomas with a higher incidence of transformation to osteosarcoma.[13]

Imaging of Fibrous Dysplasia

Conventional X-rays

Plain radiography is the first line investigation and most FD appears as a well circumscribed bony lesion with a ground glass or hazy appearance of the matrix (Fig. 1). The degree of haziness directly correlates with its underlying histopathology. More radiolucent lesions are composed of predominantly fibrous elements, whereas more radiopaque lesions contain a greater proportion of woven bone. A cystic appearance seen in some lesions corresponds with areas of necrosis.[12] There is a narrow zone of transition and no periosteal reaction or soft tissue mass. In long bones the lesions are usually located in the metaphysis or diaphysis. There is sometimes focal thinning of the overlying cortex, called "scalloping from within". Repeated fractures through lesions in the proximal femur can result in the formation of a so-called shepherd's crook deformity.

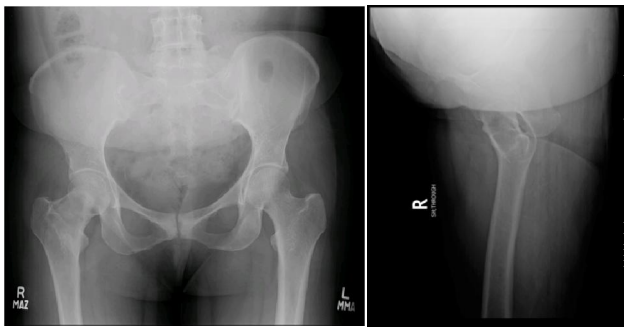
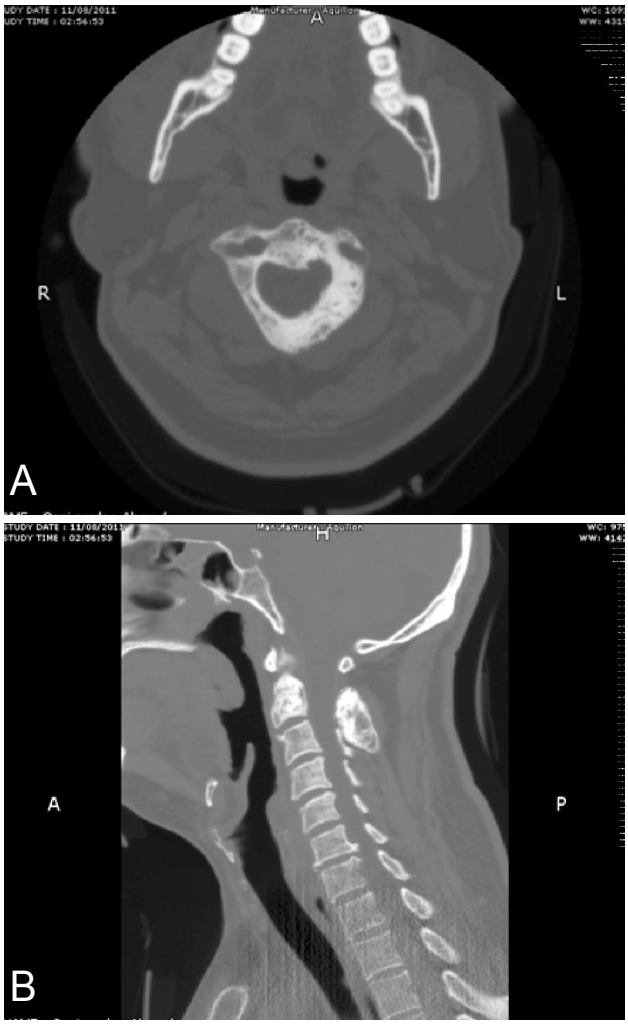


Figure 1: X-ray pelvis: There is a well-defined lucent lesion in the right femoral neck having sclerotic margins but no significant bone expansion. No fracture or periosteal reaction is seen.

Computerized Tomography (CT)

The diagnosis of FD on CT is usually straightforward and the most common appearance is an expanded bone showing a ground-glass appearance (56%) (Fig. 2), followed by the homogeneously dense pattern (23%) and the cystic variety (21%).[14]



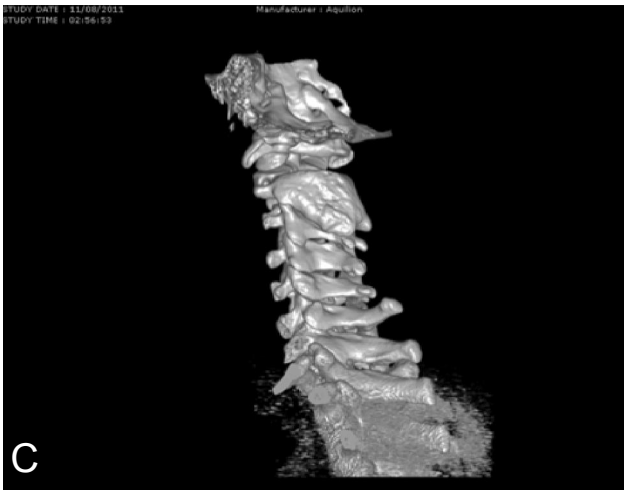
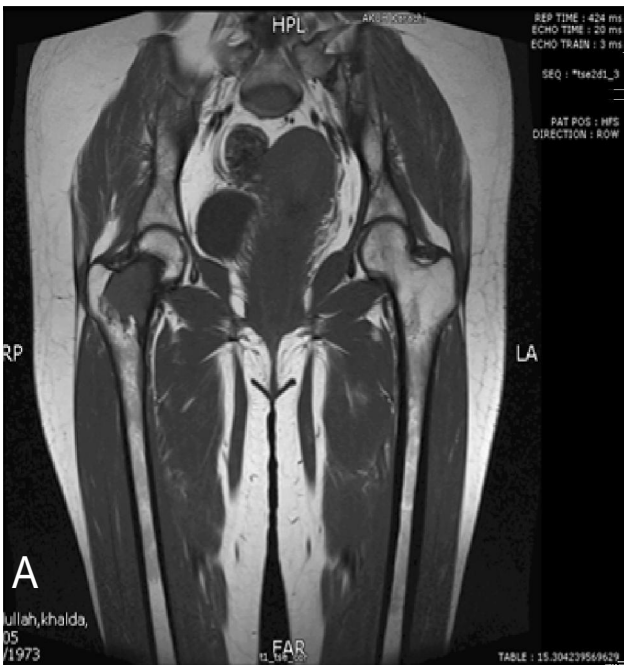


Figure 2: CT scan (A: axial, B: sagittal, C: 3 D, of a different patient) showing sclerosis and coarsening of the trabecular pattern along with expansion is identified involving the body, lamina, left pedicle as well as spinous process of the C2 vertebra (predominantly the left side).

Magnetic Resonance Imaging (MRI)

The MRI characteristics of FD are variable, typically showing signal intensity that is intermediate to low on T1-weighted images, intermediate to high on T2-weighted images, and heterogeneous enhancement after administration of gadolinium (Fig. 3) and may mimic with a tumor.[15]



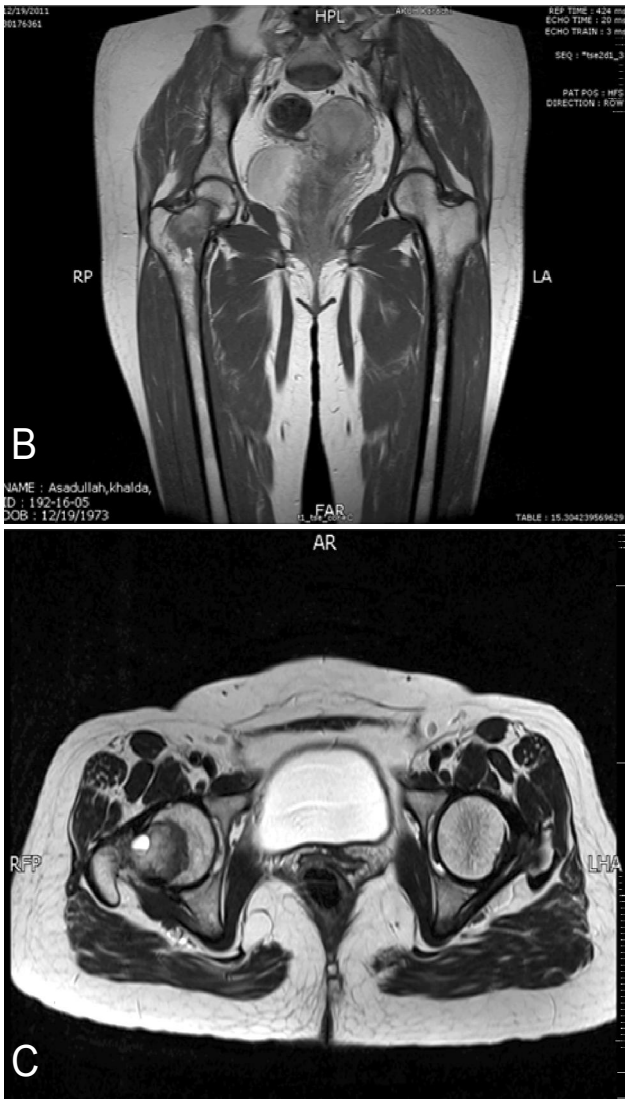


Figure 3: MRI of case patient showing T1 weighted pre contrast (A) and post contrast (B) images revealing enhancing lesion over right femoral neck and axial T2 (C) image showing cyst with fluid level.

Radionuclide Bone Scan

Being a sensitive imaging modality, the majority of lesions in FD are tracer avid on Tc-99m MDP bone scans but intensity of tracer uptake varies with predominant histological behavior of lesions (Fig. 4). Machida et al[16] analyzed 59 lesions in 26 patients with fibrous dysplasia. Four (14%) of 29 cystic lesion and two (7%) of 30 "ground glass" lesions had radiotracer uptake equivalent to normal bone. The remainder showed supra-normal tracer uptake. Bone scanning is helpful in conjunction with radiography to detect polyostotic involvement (Fig. 5).

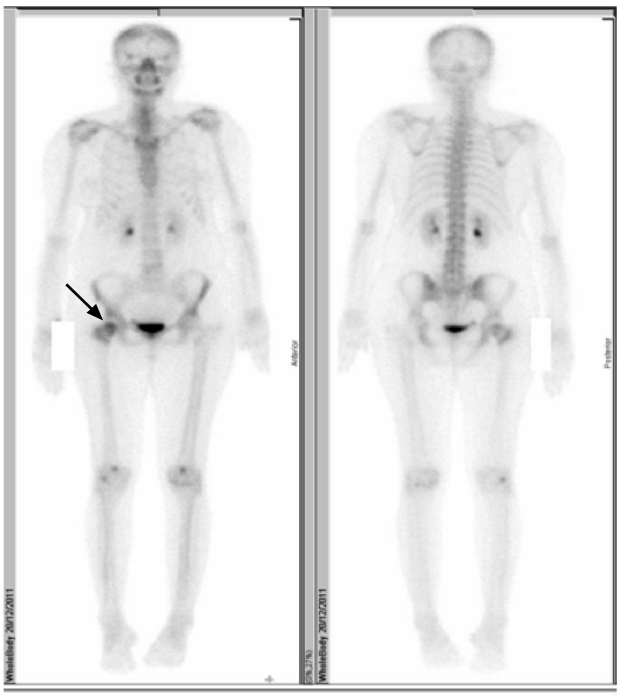


Figure 4: Whole body bone scan of the patient showing a solitary area of increased tracer deposition over right femoral neck and trochanteric region (arrow) (monostotic FD).

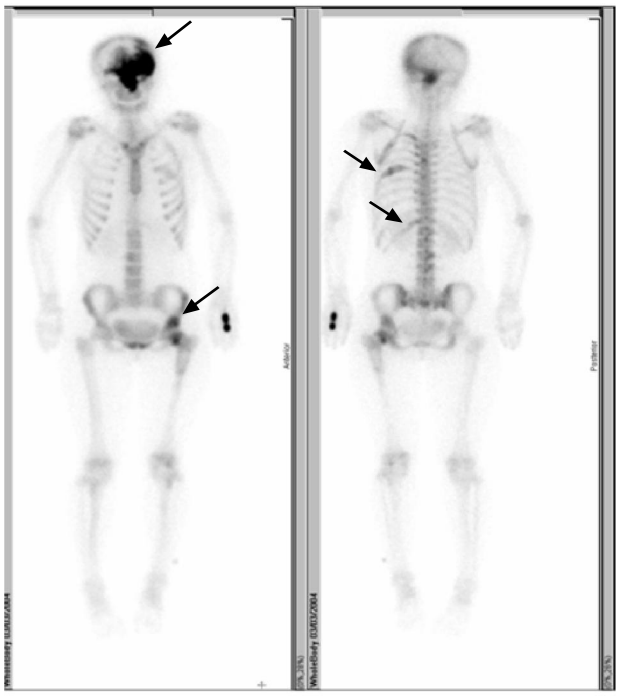


Figure 5: Whole body bone scan of a different patient with multiple areas of increased tracer deposition over facial bones, left 12th and 7th ribs, left femoral neck and proximal part of left acetabulum (arrows) (polystotic FD).

Complications

Pain is the most common presentation of FD and usually related to pathologic fracture and its risk is increased with a coexisting aneurysmal bone cyst[17] and needs surgical intervention (Fig. 6). Malignant degeneration of fibrous dysplasia complicates less than 1% of all cases, presenting clinically as pain and swelling with cortical destruction and associated soft-tissue masses on radiograph.[18]

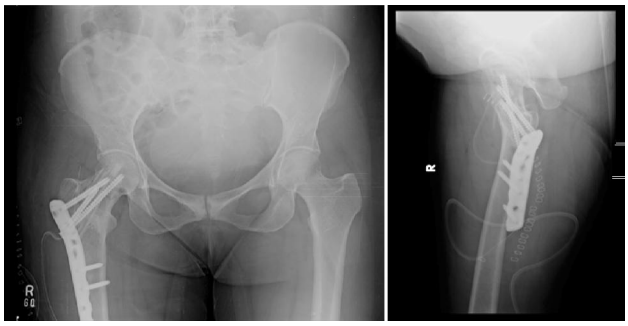


Figure 6: Plain X-rays of case patient after surgical curettage and internal fixation.

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