JOURNAL OF CLINICAL AND BIOMEDICAL SCIENCES

CASE REPORT

OPEN ACCESS

Received: 07.10.2022
Accepted: 30.10.2022
Published: 22.12.2022

Citation: Revanth RB, Deepti N, Anil KS, A, Yashas U, Guru YM. A Case Report of Polyostotic form of Fibrous Dysplasia: Imaging Features in Radiograph, Computed Tomography and Magnetic Resonance Imaging. J Clin Biomed Sci 2022; 12(4): 154-156. https://doi.org/10.58739/jcbs/v12i4.121

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Funding: None

Competing Interests: None

Published By Sri Devaraj Urs Academy of Higher Education, Kolar, Karnataka

ISSN
Print: 2231-4180
Electronic: 2319-2453

A Case Report of Polyostotic form of Fibrous Dysplasia: Imaging Features in Radiograph, Computed Tomography and Magnetic Resonance Imaging

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Abstract

Fibrous dysplasia is a disease that causes aberrant development of osteoblasts, which results in fibrous stroma replacing normal bone. Lichtenstein first described it in 1938. It is a sporadic disorder caused due to a postzygotic mutation in the GNAS1 gene. It can affect a single bone (monostotic) or several bones (polyostotic). The most common locations are the skull & ribs. It is typically an incidental finding and asymptomatic. When a pathologic fracture or malignant alteration complicates it, symptoms could develop. As polyostotic FD is a rare disorder that only occasionally occurs in 20–25% of patients, therefore the aim of this article is to report such a rare case of polyostotic FD in a 30-year-old female patient showing the characteristic changes.

Keywords: Polyostotic fibrous dysplasia; GNAS1 gene mutation; Sporadic disorder

Introduction

A developing benign medullary fibro-osseous condition known as fibrous dysplasia (FD) is characterised by the cessation of mature lamellar bone formation and the formation of woven bone, which may be multifocal.[1] It can happen in either a monostotic or polyostotic form, affecting any bone. According to WHO's 5th edition categorization of soft tissue & bone cancers, it has been referred to as a benign bony neoplasm.[2] It is a rare condition that can affect people of all ages, including children and adults. Typically, young people and children receive the initial diagnoses. Although the actual incidence is unknown, it is assumed to account for around 5% of benign bone lesions. There is no bias based on gender.

The primary criterias for diagnosing fibrous dysplasia consists of both radiological and clinical features and if the imaging characteristics are recognisable, the lesion doesn't need to be histologically examined.[3]

Case History

A thirty-year old female patient presented with right hip discomfort and weight bearing difficulty after an episode of fall since the past 1 month. On local examination, right limb was externally rotated, swelling and tenderness was present in the middle 1/3rd of the thigh. Restricted range of movements of the hip joint was noted and active movements of ankle and toes were present. General physical examination was unremarkable. Vital signs were normal.

Imaging features

1. **On plain radiograph:** Few expansile lytic lesions with thinning of adjacent cortex involving the epi metaphyseal regions of bilateral femur was noted with narrow zone of transition and mild lateral bowing/angulation of bilateral femur. Pathological subtrochanteric fracture was also noted. There is no calcification or periosteal reaction or soft tissue component within the expansile lytic lesions.
2. **On computed tomography:** CT scan coronal reformatted image showed few well-defined expansile lucent lesion in the epimetaphyseal region of bilateral femora with interspersed areas of ground glass density. There is no calcification or periosteal reaction or soft tissue component. Pathological subtrochanteric fracture was also noted.
3. **On Magnetic resonance imaging:** Multiple ill-defined T1 and T2 heterogenous mixed signal intensity lesions with no restriction of diffusion on DWI are noted extensively involving bilateral head, neck & shaft of femur and pelvic bones. On post contrast study these lesions showed heterogenous enhancement. Also, noted subtrochanteric fracture of right femur and diffuse hyperintensities in muscles of the right gluteal and right thigh region, Secondary to fracture/denervation.

Discussion

The first descriptions of the benign disorder fibrous dysplasia were made by Lichtenstein in 1938 and Jaffe in 1942.4 With 0.8% primary and 7% benign bone lesions, it is a rather rare disorder. In 70–80% of cases, it is monostotic, while in 20–30% of cases, it is polyostotic.

Diagnostic criteria according to WHO classification of soft tissue and bone tumors (5th edition):

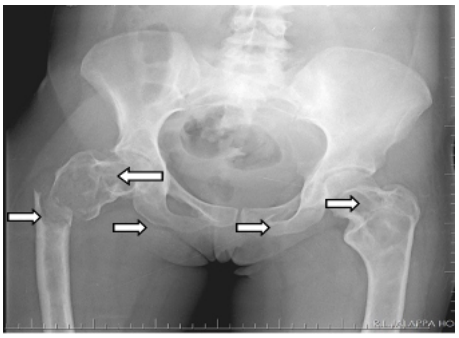

Fig 1. Radiograph AP view of pelvis showing Few expansile lucent lesions (White arrows) with thinning of adjacent cortex involving the epi-metaphyseal regions of bilateral femur was noted with narrow zone of transition and mild lateral bowing/angulation of bilateral femur

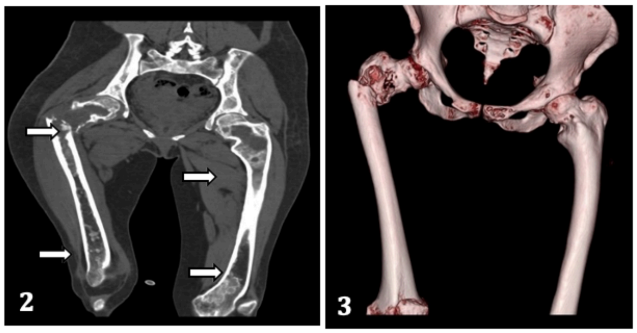

Fig 2. 2 & 3: Computed tomography scan coronal reformatted image & 3D surface shaded display image showing few well-defined expansile lucent lesion in the epi-metaphyseal region of bilateral femora with interspersed areas of ground glass density. There is no calcification or soft tissue component or periosteal reaction. Pathological subtrochanteric fracture was also noted

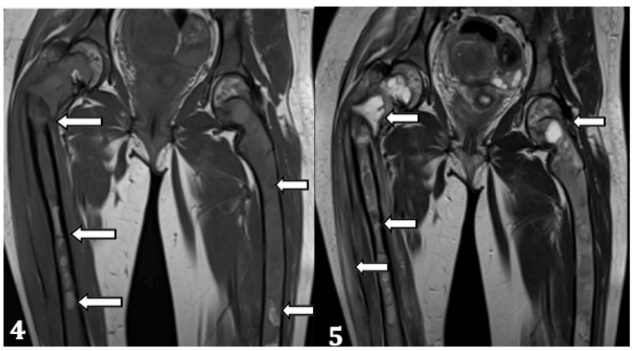

Fig 3. T1 & T2 weighted coronal MRI shows Multiple ill-defined T1 and T2 heterogenous mixed signal intensity lesions extensively involving bilateral head, neck & shaft of femur and pelvic bone Also, noted subtrochanteric fracture of right femur. Diffuse hyperintensities in muscles of the right gluteal and right thigh region were also noted (Orange arrow), Secondary to fracture / denervation

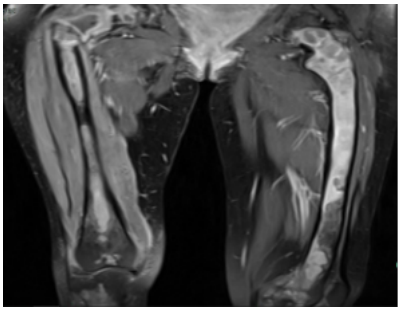

Fig 4. T1 weighted post-contrast coronal MRI shows heterogenous enhancement of the lesions involving bilateral head, neck & shaft of femur and pelvic bone

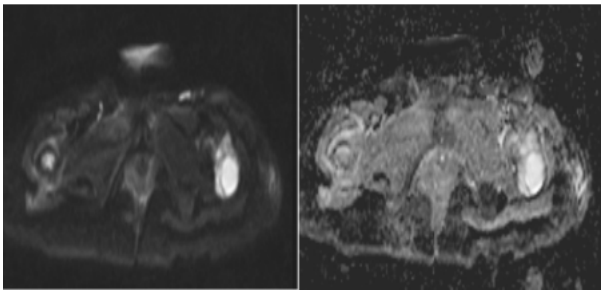

Fig 5. 7 & 8: Diffusion weighted imaging & apparent diffusion coefficient image

Essential features include

- A bone lesion with compatible imaging characteristics.
- Osseous part consisting of irregular curvilinear branching trabeculae of woven bone without apparent osteoblastic rimming.
- Fibrous part consisting of bland fibroblasts.

The following additional criterion is desirable

- Evidence of GNAS activating missense mutations Imaging features include:

Plain radiography imaging

The appearance of fibrous dysplasia is usually smooth and homogeneous with endosteal scalloping and cortical thinning. There are clearly marked borders and the cortex is generally intact but thinned due to the expansive nature of the lesion. Other features include: ground-glass matrix, may be completely lucent (cystic) or sclerotic, well-circumscribed lesion with no periosteal reaction and in some cases rind sign, i.e. when a lesion is surrounded by a layer of thick, sclerotic reactive bone. In extremities, bowing deformities and shepherd crook deformity of the femoral neck might be seen.

Computed Tomography

Furthermore the above mentioned radiography findings, other findings like ground-glass opacities, homogeneously sclerotic lesion, cystic lesion, with well-defined borders can be seen.

Magnetic resonance imaging

Due to the numerous variations in how bone lesions look and the fact that they frequently resemble tumours or other more aggressive diseases. The ability of MRI to distinguish fibrous dysplasia from other conditions is not particularly useful. However, heterogenous signal intensity is seen on T1 and T2 weighted imaging with heterogenous post-contrast enhancement.

Through dietary changes and exercise, management attempts to gauge the severity of the condition and preserve bone quality. The prognosis is favourable and typically no more therapy is needed.

However, leg length disparities, deformities, and impingement or nerve compression disorders can all result from monostotic fibrous dysplasia.[4] Surgical excision may be considered if a bulk effect is significant.

Differential diagnosis of Paget's disease and osteofibrous dysplasia should be ruled out.[5]

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