ORIGINAL RESEARCH PAPER
Volume - 13 | Issue - 08 | August - 2024 | PRINT ISSN No. 2277 - 8179 | DOI : 10.36106/ijsr
INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

CASE SERIES OF FIBROUS DYSPLASIA—A PICTORIAL VIEW

Radiology

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ABSTRACT

Fibrous dysplasia (FD) is a benign condition characterized by the failure to form mature bone tissue, which can affect any bone in the body. It is a genetic noninherited condition caused by mutation 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. It can occur in two forms: monostotic, where it affects only one bone, or polyostotic, where it affects multiple bones.

Variants: Mccune albright syndrome-fibrous dysplasia associated with café au lait spot and precocious puberty. Mazabraud's syndrome- fibrous dysplasia associated with Intramuscular myxoma.

In this study we report seven cases of fibrous dysplasia occurring at various locations with radiological interpretation.

KEYWORDS

FD- Fibrous Dysplasia, CT- Computed Tomogram

CASE REPORT 1:-

A 42-year-old woman presented with persistent headache and pain on the right side of chest over the past four years, accompanied by back pain and difficulty in breathing during physical activity. Upon Physical examination, revealed tenderness over the right chest wall and palpable bony prominence. Multiple radiographs and CT scans were conducted. Chest radiograph and CT scans revealed several abnormalities. The chest radiograph AP view showed a lytic expansile and destructive lesion on the posterior end of the right 6th rib. Skull Radiograph lateral view showed a ground glass matrix, osteolytic lesion on the left side of the skull vault, extending to various cranial structures like sphenoid wing, orbital roof of the frontal bone, zygomatic arch and left superior orbital wall on skull radiography. CT scans of the thorax confirmed an expansile osteolytic lesion on the right 6th rib. Additionally, a CT scan of the skull showed a lytic expansile lesion with ground glass appearance in the left fronto-temporal bone with no accompanying soft tissue component. These radiographic findings collectively indicated a diagnosis of fibrous dysplasia.

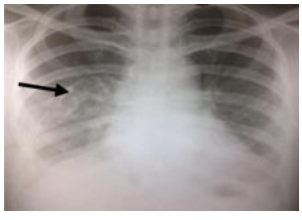

Figure 1: Chest Radiograph PA view showing an expansile destructive lesion of the right 6th rib (black arrow) mainly involving the posterior end.

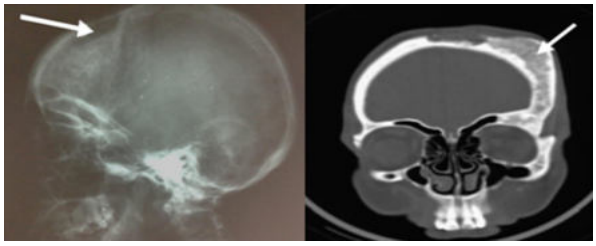

Figure 2 & 3 Skull radiograph lateral view & CT skull-bone window coronal section showing lytic expansile destructive lesion of the left fronto-temporal bones (white arrow) with ground glass matrix. The lesion is extending to orbital roof of the frontal bone, zygomatic and left superior orbital wall along with an expansile cortical thickening extending up to the sphenoid wing, orbital roof of the frontal bone, zygomatic arch and left superior orbital wall.

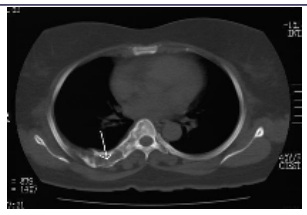

Figure 4: CT thorax Bony window axial cut showing expansile destructive osteolytic lesion of the 6th rib on the right side (white arrow) involving posterior aspect with ground glass matrix.

CASE REPORT 2

A 32-year-old male patient presented with complaints of intermittent chest pain and a noticeable lump over the bilateral chest walls, along with chronic pain and mild swelling in proximal part of left lower limb and left elbow region. Patient had past medical history of a left femur fracture requiring internal fixation. Physical examination revealed tenderness over multiple ribs on both sides and palpable bony prominences. Upon conducting multiple radiographic examinations, an expansile lytic and destructive lesion was observed at the posterior end of the right 7th rib and left 6th rib on PA view chest radiography . Additionally, a distinct and trabeculated cystic lesion with endosteal scalloping and cortical thinning was noted in the meta-diaphyseal region of the left hip joint on radiographs. Similar lesion was also noted in distal part of left humerus. On the basis of clinical symptoms and radiographic findings diagnosis of fibrous dysplasia was made.

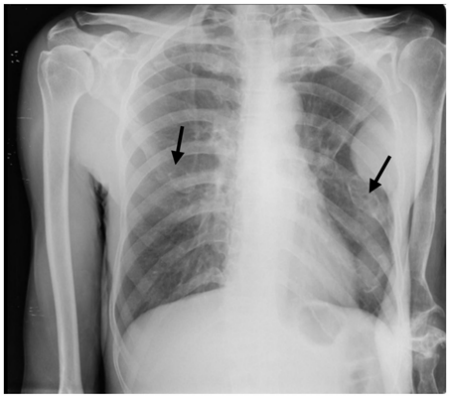

Figure 5: Chest radiograph PA view showing an expansile lytic and destructive lesion of the right 7th rib and left 6th (black arrow), mainly involving the posterior end with ground glass matrix.

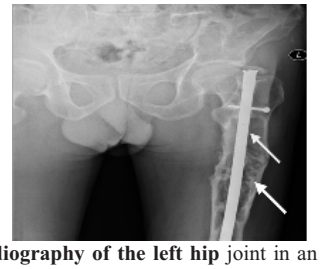

Figure 6:- Radiography of the left hip joint in an anteroposterior (AP) view. Within the left femur, a well-defined and trabeculated lytic expansile and cystic lesion (white arrow) with ground glass matrix is seen in the meta-diaphyseal region. This lesion exhibits characteristics such as endosteal scalloping and cortical thinning. Additionally, the image shows the presence of an intramedullary nail and screws in situ.

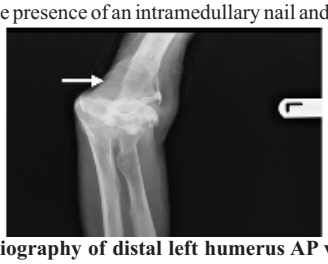

Figure 7: Radiography of distal left humerus AP view showing a well circumscribed lytic expansile and trabeculated cystic lesion (white arrow) noted in the meta-diaphyseal region of distal humerus showing ground glass matrix with endosteal scalloping and thinned cortex.

CASE REPORT 3

A 38-year-old female patient presented with a painless swelling on the right side of mandible and occasional headaches and chest pain persisting for the past three years. Upon Physical examination tenderness over the bilateral chest wall and palpable bony prominences was noticed. Multiple radiographic studies were conducted. Radiographs of the mandible (oblique view) and skull (lateral view) revealed an expansile lytic lesion with a ground glass matrix in mandible and in frontal bone. Additionally, pelvic radiography (AP view) showed bubbly cystic lesions with fusiform enlargement of bones in the ribs on the right side. Similar appearing lesion also noted in left side pubic rami. These collective radiographic findings suggested a diagnosis of fibrous dysplasia.

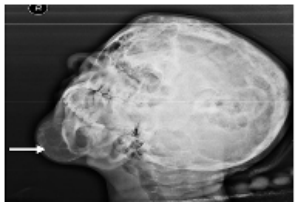

Figure 8: Radiograph Oblique view of Mandible showing an expansile, lytic lesion (cystic) of ground glass haziness (white arrow) with thinned out cortex.

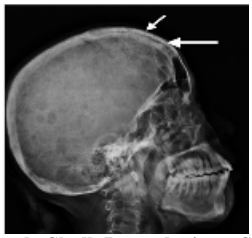

Figure 9: Radiograph Skull Lateral view: Skull vault showing expansile lesion of ground glass matrix (white arrow) in the frontal bone.

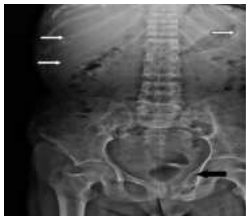

Figure 10:- Radiograph AP view pelvis showing cystic (bubbly) lesions with fusiform enlargement having ground glass matrix involving multiple ribs (white arrows) bilaterally and pubic rami on left side (black arrow).

CASE REPORT 4

A 49-year-old male patient presented with complaints of a deformity of the bilateral lower limbs which had been present for several years and now accompanied by progressive pain and swelling from last 1 year. A series of radiographs were performed to assess the condition which showed a lytic, trabeculated, expansile intramedullary lesion having ground glass matrix and causing endosteal scalloping with thinning of cortex on the left femur radiograph (AP view). Also, on radiographs of the bilateral lower limbs (AP view), a well expansile radiolucent lesion with ground glass matrix noted in the lower shaft of the bilateral tibia and fibula causing endosteal scalloping and deformity with thinned out cortex. The findings on both radiographs suggest a diagnosis of fibrous dysplasia.

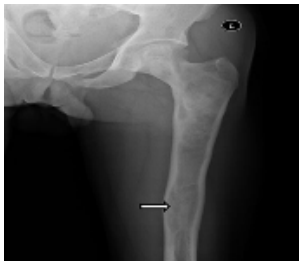

Figure 11: Radiograph AP view left femur showing a lytic, trabeculated, expansile intra- medullary lesion (white arrow) having ground glass matrix and causing endosteal scalloping with thinning of cortex.

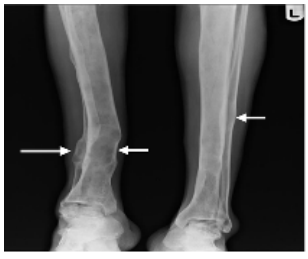

Figure 12:- Radiograph bilateral lower limbs AP view shows a well expansile lytic/lucent lesion in lower shaft of bilateral tibia fibula with deformity (white arrows), having ground glass matrix causing endosteal scalloping with thinned out cortex.

CASE REPORT 5

A 29-year-old male patient presented to the orthopedic outpatient department with a history of bilateral lower limb deformity since childhood, accompanied by progressive pain and swelling in both legs over the past 2 years. On inspection, noticeable bilateral lower limb asymmetry was observed, with the left leg showing more pronounced effects than the right. Limited range of motion was noted, attributed to pain and bony deformities. Radiographs of multiple bilateral lower limb regions were performed, revealing characteristic findings such as a ground-glass appearance, osteolytic lesions, and bone deformities involving multiple bones of bilateral lower limbs.

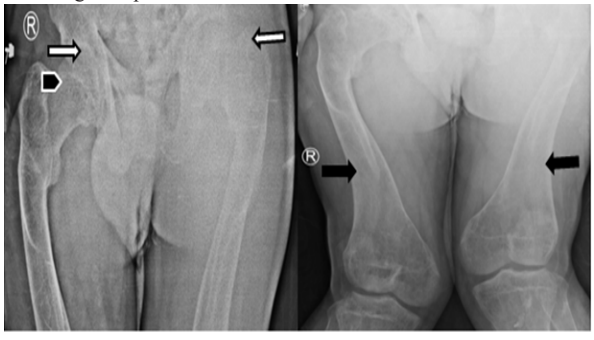

Figure 13:- Radiograph bilateral Hip joint AP view shows shepherd crook deformity in bilateral hip joint (white arrows) as evidenced by a severe coxa varus angulation of the proximal femori with bowing of shaft (black arrows). Underlying lucent expansile lesion with ground glass haziness is seen in upper part of left femur(lack arrow head).

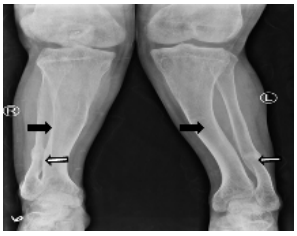

Figure 14:- Radiograph bilateral lower limbs AP view shows shortening of bilateral tibia and fibula with bowing deformity (black arrows). Pathological fractures are also seen in bilateral lower shaft of fibula (white arrows).

CASE REPORT 6

A 45 yr old male patient came up with complaints of pain in both shoulders and feet for the past 4 years. Physical examination revealed palpable bony prominences over the bilateral scapulae and bilateral first metatarsals. Radiograph of the chest (PA view) and bilateral feet (AP view) showed osteolytic, expansile lesions with ground glass matrix in scapulae bilaterally and in both feet, which suggested a diagnosis of fibrous dysplasia.

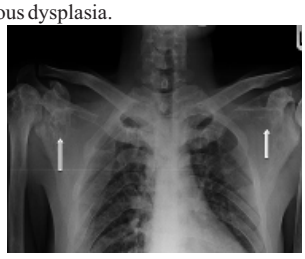

Figure 15:- Radiography of chest X-ray PA view showing an osteolytic expansile lesions (white arrows) with ground glass matrix in scapulae bilaterally.

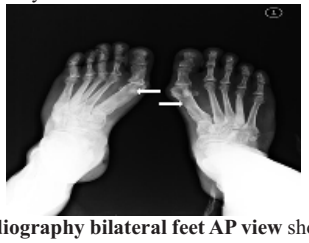

Figure 17:- Radiography bilateral feet AP view showing osteolytic expansile lesions with ground glass matrix (white arrows) in bilateral 1st meta-tarsal.

CASE REPORT 7

A 45-year-old male presented to the orthopedic clinic with complaints of progressive right lower limb pain for the past four years. Physical examination revealed restricted range of motion in the right knee joint with swelling just below the right knee joint. Knee radiographs (AP and lateral view) displayed a well-circumscribed lytic expansile lesion exhibiting ground glass haziness, with endosteal scalloping and thinned cortex in proximal end of tibia.

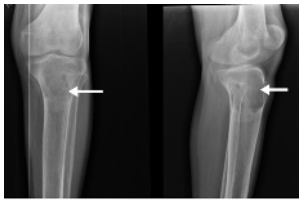

Figure 17:- Radiograph of right Knee AP and lateral view showing a well circumscribed lytic expansile lesion having ground glass matrix (white arrows) causing endosteal scalloping with thinned out cortex in proximal end of the tibia.

INTRODUCTION

Fibrous dysplasia is a developmental anomaly characterized by the replacement of normal bone marrow with fibro-osseous tissue (1, 2). This skeletal disorder affects the bone-forming mesenchyme, leading to a defect in osteoblastic differentiation and maturation (3, 4). As defined by Reed, fibrous dysplasia is an arrest of bone maturation (3).

This is a rare condition and is characterized by deformities of bone, fractures, nerve compressions, and bone pain (5). It can affect a single bone, a small segment, or the entire skeleton diffusely (1, 6).

Fibrous dysplasia is typically seen in young adults (1, 7) and is considered a benign skeletal disorder. It is not a true neoplasm (1), but rather a developmental anomaly that was first described by Lichtenstein and Jaffe and is also known as Lichtenstein-Jaffe disease (1,2,8,9).

Interestingly, fibrous dysplasia has been linked to several endocrinological diseases, including Albright syndrome (7, 8). However, there is no evidence to suggest that the disease has a familial, hereditary, or congenital basis (4, 9).

CLINICAL FEATURES

Fibrous dysplasia is often discovered incidentally, and in most cases, it does not cause symptoms. However, as the condition progresses, some individuals may experience bony expansion and remodeling, or pain. In some cases, complications can arise from pathologic fractures or compression and displacement of adjacent structures, which can lead to serious consequences such as loss of vision or hearing loss in craniofacial fibrous dysplasia.

The most common symptom of fibrous dysplasia is bone pain (5). This condition typically affects young individuals, typically in the first and second decades of life (1, 4, 8).

Other Features Of Fibrous Dysplasia May Include:

Cutaneous lesions in the form of café-au-lait spots, which often have irregular or serrated borders resembling the "Coast of Maine" appearance(1), Facial asymmetry is seen due to hemi-cranial involvement (1). Exacerbation of fibrous dysplasia during pregnancy, which can also lead to the formation of aneurysmal bone cysts (1), sinusitis.

PATHOPHYSIOLOGY

Fibrous dysplasia is thought to occur due to an abnormal activity of mesenchymal cells (4, 9), which is caused by a mutation in the gene that encodes the subunit of a stimulatory G protein (GS alpha) located on chromosome 20 (2, 3, 8, 10). Specifically, a substitution of cysteine with arginine is responsible for the mutation.

The lesions of fibrous dysplasia are characterized by a mixture of fibrous tissue and bone trabeculae (1,10). The fibrous stroma is a myxofibrous tissue with low vascularity, while the bone trabeculae are composed of woven bone (1).

In contrast to normal bone development, fibrous dysplasia does not exhibit osteoblastic activity. However, osteoclasts are typically present, particularly on the concave side of the trabeculae(1,10). The lesions of fibrous dysplasia are characterized by the expansion of cortical bone, which is gradually replaced by firm, rubbery, and gritty fibrous tissue (8).

Fibrous dysplasia is characterized by altered osteogenesis leading to an intramedullary fibro-osseous proliferation with fibrous and osseous tissue components being present in varying degrees (11). It comes in a monostotic or polyostotic form, depending on whether only one single bone or multiple bones are affected. However, there is no progression from the monostotic to the polyostotic form (12).

MICROSCOPIC APPEARANCE

Microscopically, fibrous dysplasia is characterized by the following (11, 13, 14, and 15).

- Varying proportions of fibrous and osseous tissue
- Fibrous tissue is principally made up of bland spindle cells without conspicuous cellular atypia
- Irregular curvilinear branching trabeculae of woven bone with a pattern that has been characterized as “looking like Chinese characters” (13,14).
- Absence of osteoblastic rimming
- Uncommon mitoses unless there is a fracture.

DISCUSSION

Fibrous dysplasia is a developmental disorder that occurs when normal bone marrow is replaced by fibro-osseous tissue (1,2). Fibrous dysplasia is a skeletal developmental disorder that affects the bone-

forming mesenchyme, leading to replacement of normal bone marrow with fibro-osseous tissue (3,4).

It is a rare condition, accounting for 2% of all bone tumors and 7% of all benign bone tumors (2, 9). The most common locations affected are the ribs, femur, tibia, fibula, and facial bones (1, 4,9).

There are two types of fibrous dysplasia:-

Monostotic fibrous dysplasia—

It is the most common form of fibrous dysplasia, affecting a single bone (1, 2, 4, 7) in approximately 70%, (3,8) of cases.

Polyostotic fibrous dysplasia—

It involves multiple bones and is more common in females (3). In some cases, polyostotic fibrous dysplasia can be associated with soft tissue myxoma (1), typically occurring in multiple sites within the muscles.

The polyostotic form of fibrous dysplasia can be further divided into three subtypes (2).

Cranio-facial type— - only the cranio-facial complex is involved
Lichtenstein Jaffe type

Albright syndrome:- Albright syndrome is characterized by a triad of polyostotic fibrous dysplasia, usually affecting one side (uniateral) of the body (1).

IMAGING FEATURES OF FIBROUS DYSPLASIA

X RAY:-

Fibrous dysplasia typically displays three main imaging features (12) :-

- Cystic/lucent
- Sclerotic
- Mixed

Beyond that, the appearance of fibrous dysplasia is typically smooth and homogeneous, with the following features:

- Endosteal scalloping: the bone surface is curved inward, forming a scalloped shape.
- Cortical thinning: the outer layer of the bone is thinned, but not broken (12).
- Additionally, the borders of the lesion are well-defined, and the surrounding bone is intact, but thinned due to the expansive nature of the fibrous dysplasia (12).
- The fibrous dysplastic lesion is characterized by its eccentric and medullary location (1), situated within the spongy tissue of the bone. The lesion's presence alters and remodels the normal architecture of the bone, resulting in a distinctive appearance marked by delicate woven bone spicules that give a "ground glass" appearance (1).
- Shepherd's crook deformity
- The Shepherd's crook deformity is a characteristic feature of fibrous dysplasia, which is characterized by a marked varus deformity of the affected femur. This deformity occurs as a result of abnormal modeling of the bone, which is caused by alterations in the normal biomechanical properties of the bone. In other words, the bone's natural structure and function are disrupted, leading to a curvature of the femur that resembles a shepherd's crook.

Computed Tomography

Computed Tomography (CT) is considered the modality of choice for evaluating fibrous dysplasia (12) particularly in craniofacial lesions. CT imaging features include:

- Ground-glass opacities: present in 56% of cases (16)
- Homogeneously sclerotic: seen in 23% of cases
- Cystic: present in 21% of cases
- Well-defined borders: characteristic of fibrous dysplasia
- Expansion of the bone: with the surrounding bone intact.
- Endosteal scalloping: may be visible on CT imaging.

Additionally, the attenuation of lesions typically ranges from 60-140 HU (Hounsfield units) and may enhance after the application of contrast media (12). Areas of low enhancement and cyst formation aid in differentiating the lesion from malignancy. (7)

MRI

On MRI, the majority of fibrous dysplasia lesions typically appear hypointense on T1-weighted images and exhibit variable intensity on T2-weighted images (1,6). This variable intensity on T2WI can be mistaken for a soft tissue tumor (6). Furthermore, the lesion's fibrous tissue component enhances intensely after contrast administration (6), which can lead to a mistaken diagnosis of a tumor. Additionally, areas of high intensity on T2WI are thought to correspond to non-mineralized regions and cystic change that may be visible on CT scans (6).

COMPLICATIONS

Fibrous dysplasia is typically a monostotic and asymptomatic condition, but in severe cases, it can lead to bone deformity and subsequent musculoskeletal dysfunction or osteoarthritis (17). Lesions in the spine can increase the risk of developing scoliosis, which can result in functional limitations (18). In rare cases, fibrous dysplasia can affect the craniofacial region, causing associated cranial nerve deficits, such as vision and hearing loss. While malignant transformation to sarcoma is rare, (19) it can occur when radiation therapy has been previously used to treat the condition (20).

TREATMENT

Fibrous dysplasia is usually self-limiting except in syndromic cases which require surgery (10).

- The bone and soft tissue tumor program at CHOP offers a team approach to treating fibrous dysplasia, with a multidisciplinary team of specialists. Treatment options include:
- Non-surgical treatment: bisphosphonates to prevent bone loss and reduce pain, administered by endocrinologists
- Surgical treatment: Surgical correction is an effective treatment (2,3,8,9) and is done depending on the severity of the condition, including:
- Fracture repair: metal plates and screws on the outside of the bone or a metal rod placed inside the bone
- Deformity correction: to address progressive deformity and associated pain and functional impairment.

DIFFERENTIAL DIAGNOSIS

In patients with polyostotic McCune-Albright syndrome, it is crucial to accurately assess the cafe au lait skin lesions, as they can have serrated borders, which distinguishes them from neurofibromatosis (21). When diagnosing fibrous dysplasia, radiologists must be aware that the imaging presentation can vary and may resemble other bone diseases. In fact, monostotic fibrous dysplasia can mimic several conditions, including simple bone cyst, giant cell tumor, fibroxanthoma, osteoblastoma, hemangioma, osteofibrous dysplasia, and Paget disease. Polyostotic fibrous dysplasia can also resemble neurofibromatosis, hyperparathyroidism, enchondromatosis, and eosinophilic granuloma (22). To favor a diagnosis of fibrous dysplasia, clinicians should consider factors such as patient age, bone location (long bone diaphysis), presence of a ground glass matrix, and non-aggressive appearance (23). In cases where malignancy cannot be ruled out after clinical and imaging workup, a bone biopsy may be necessary to confirm the diagnosis (24).

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