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Rare case of polyostotic fibrous dysplasia of bilateral tibia with pathological fracture and associated endocrinopathy managed conservatively

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Abstract
Introduction: Fibrous dysplasia represents a non-malignant lesion that arises from a developmental anomaly during the process of woven bone remodeling. Typically, these lesions are monostotic and asymptomatic, they may present as polyostotic, predominantly affecting the long bones, ribs, and craniofacial skeletal structures.
Case presentation: We present a rare case of polyostotic fibrous dysplasia with bilateral tibial involvement and associated endocrinopathy of hyperthyroidism and hyperparathyroidism. There are very few reported cases in the literature. A 22-year-old male presented with bilateral tibial pathological fracture after trivial trauma, with radiographs showing lytic lesion in bilateral tibial shaft. The patient was managed conservatively due to the associated medical comorbidities, hyperthyroidism, and thyroid storm.
Conclusion: This case presents an unusual presentation of polyostotic fibrous dysplasia with bilateral tibia lesions and pathological fracture with associated endocrinopathy. It was managed conservatively, and the fracture united at a faster rate than what was expected from literature, suggesting that these can be treated conservatively without the need for internal fixation with autogenous or allogenous bone grafts.

Keywords: Polyostotic fibrous dysplasia, bilateral tibia, conservative, endocrinopathy, hyperthyroidism

Introduction

Fibrous dysplasia is a non-heritable, benign, fibro-osseous lesion that is medullary mono-ostotic in 80% of cases and polyostotic in 20%. It is considered a developmental failure during the remodeling of woven bone when bones fail to remodel, such that mature lamellar bone fails to form. Owing to loss of maturation, the unorganized mass of the trabecular bone is left enmeshed in the dysplastic fibrous tissue. This condition, combined with insufficient mineralization, causes significant loss of mechanical strength, leading to deformity, pathological fracture, and bone pain. Generally, four forms of this disease have been described: 1. Mono-ostotic, involving a single bone, 2. Polyostotic form involving multiple bones 3. Craniofacial form 4. Cherubism: inherited form involving maxilla and mandible.

2-3% of polyostotic FD cases have café au lait skin lesions and precocious puberty: McCune –Albright syndrome

Some cases of polyostotic FD have isolated endocrinopathies such as Hyperthyroidism, Hyperparathyroidism, diabetes mellitus, and Cushing's syndrome without full McCune Albright.

Fibrous dysplasia is an incidental finding in most mono-ostotic cases. Other patients presented with bone pain, pathological fractures, and deformities. Bones plastically deform under mechanical pressure from body weight, and the type of deformity depends on the location of the lesion. The proximal femur lesion has a characteristic Shepherd's hook deformity, tibial bow (Saber tibia), protrusio acetabulae, and scoliosis. Deformity in monostotic lesions halts after skeletal maturity, whereas polyostotic lesions continue to progress.

Fibrous dysplasia can affect multiple sites in the axial and appendicular skeletons with craniofacial abnormalities. Radiographs typically show expansile lesions with endosteal scalloping and thinning of the cortex, demonstrating a classic "ground glass" appearance,

with no periosteal reaction. The most commonly involved areas were the proximal femur and skull base.

For incidental lesions, especially in the mono-ostotic form, observation is performed because there is minimal risk of pathological fracture or deformity. Various treatment methods for polyostotic lesions include injectable bisphosphonates, pamidronate, and oral risedronate, which inhibit osteoclastic bone resorption, clinically reduce pain, and improve bone strength.

Surgical management is directed for the treatment or prevention of pathological fractures. Simple curettage and bone grafting have high failure rates, with local recurrence and reappearance of polyostotic lesions.

Previous studies have not reported indicators of FD, although there are several treatment methods, including conservative treatment (medications and braces), surgical procedures (curettage, curettage and graft, and internal fixation). Generally, curettage of the lesion leaves a cavity that predisposes the bone to destabilization or even pathological fracture. Therefore, external or internal fixation was performed for stabilization.

Case report

A 23-year-old male with complaints of bilateral leg pain and inability to walk after a trivial fall presented to KEM Hospital on 27th November, 2021. He also had a history of right humerus fracture, which was managed conservatively four years previously.

On physical examination, tenderness was present bilaterally on the mid part of the tibia. On radiographic investigation, X-ray of bilateral tibia (AP and lateral view). The radiographic examination revealed lytic lesions in the diaphyseal regions of both the tibias. [figure 1] To find out the cause of the lytic lesions and to rule out other causes, the patient was sent for a skeletal survey, which included X-ray of the spine, pelvis, skull, and hands. X-ray of the right humerus was done to look for the previous injury.

X-rays revealed no lesions except the humerus X-ray which showed similar lytic lesions as those seen in the tibias. [figure 2]

Bilateral tibia X-ray showing a lytic lesion in the mid-shaft diaphyseal region, which has a typical ground-glass appearance. Association of pathological fractures in both tibia.

Laboratory investigation: serum parathyroid 123 pg/ml
Serum calcium, 9 mg/dl, phosphorus- 3.1mg/dl,
Alkaline phosphatase 282 U/L
Free T3, 8.91; Free T4, 4.40; TSH, 0.05
Primary hyperparathyroidism and primary hyperthyroidism
As the lesions were located in the diaphyseal region with typical lytic lesions with ground-glass appearance, we diagnosed the lesions as polyostotic fibrous dysplasia.

Management: The patient was managed conservatively on the bilateral above knee slab for 1 and half months due to the associated medical comorbidities of hyperthyroidism thyroid storm. Both fractures united with callus formation seen in both tibia. [Figure 5 and 6]

Discussion

Fibrous dysplasia, a non-malignant intramedullary fibro-osseous condition, was initially documented by Lichtenstein in 1938 and further described by Lichtenstein and Jaffe in 1942 [1]. The etiology of this condition has been associated with a Gsα gene mutation occurring in somatic cells post-fertilization, specifically located on chromosome 20q13.2-13.3 [2, 3].

Various management strategies exist, including observation, pharmacological interventions, and surgical procedures. Numerous cases are incidentally discovered through radiographic imaging when patients are asymptomatic. If the radiographic findings are characteristic of fibrous dysplasia, a biopsy is generally unwarranted. These lesions typically do not pose a risk of pathologic fracture or deformity, and clinical observation is often sufficient [4].

While open biopsies are infrequently required, they may be necessary to confirm the diagnosis in patients presenting with atypical symptoms.

Surgical interventions may be indicated for addressing deformities, persistent pain, lesion progression, or bone involvement that increases the risk of pathological fracture. Surgery may also be performed to prevent or treat pathological fractures [5-9].

Liens *et al.* [10] documented the immediate impacts of pamidronate on nine individuals, comprising eight with polyostotic and one with monostotic disease. Each patient received intravenous pamidronate infusions over a three-day period, with a total dosage of 180 mg (60 mg daily), administered every six months. This treatment was complemented with calcium (500 to 1500 mg daily) and vitamin D (800 to 1200 IU daily).

Basic curettage carries a significant risk of recurrence, while curettage combined with an autogenous cancellous bone graft demonstrates varying outcomes.

Autogenous cancellous grafts are absorbed more rapidly and replaced by dysplastic fibrous tissue. In contrast, cortical autogenous grafts used to fill curetted cavities remain intact for a longer duration than cancellous grafts. Cortical allogeneic grafts exhibit the least and slowest internal replacement by host bone, resulting in most grafts persisting for an extended period. This characteristic renders fibrous dysplasia one of the rare conditions where allogeneic grafts are biologically more advantageous than autogenous grafts [4].

O'Sullivan and Zacharin *et al.* observed favorable outcomes utilizing intramedullary interlock nailing and bisphosphonates for femur and tibia fibrous dysplasia lesions in 10 patients diagnosed with McCune-Albright syndrome. The mean follow-up period was 18 months. All patients experienced improved quality of life, characterized by reduced pain scores, decreased fracture rates, and enhanced ambulatory ability [11].

A retrospective analysis by Zhang *et al.* examined 39 FD patients who underwent intramedullary nailing. The procedure encompassed curettage, grafting, and intramedullary nailing. At final follow-up, 33 patients reported no pain, while seven experienced occasional mild discomfort. The affected areas included the femur (31 cases), coxa vara (8 cases), tibia (14 cases), fibula (1 case), spine (2 cases), bilateral lower limbs (1 patient), ipsilateral femur and tibia (1 patient), and ipsilateral femur and fibula (1 patient) [12].

Guille *et al.* proposed that for lesions covering extensive areas, refracture risk increases after plate and screw removal. Consequently, intramedullary nailing is recommended for various anatomical locations, excluding proximal femoral lesions in the lower limbs. This method

results in fewer complications at the lesion or fracture site compared to plate and screw fixation, while providing adequate stability by anchoring in healthy distal bones. This approach is particularly beneficial for expansive lesions [13].

Polyostotic fibrous dysplasia with pathological fracture of both tibias is rarely reported. We describe an uncommon case involving a 21-year-old male with concurrent endocrine disorders.

Unlike previous studies, this case presented an atypical manifestation of fibrous dysplasia featuring bilateral tibial lesions and pathological fracture, along with a rare combination of endocrine abnormalities (Hyperthyroidism and hyperparathyroidism) without the classic Mccune Albright Syndrome. Due to hyperthyroidism and thyroid storm, the patient was deemed unfit for surgery and was instead treated conservatively with above-knee plaster for 6 weeks. Although literature suggests that pathological fractures in fibrous dysplasia typically require extended healing periods, in this instance, the fracture site healed within 6 weeks.

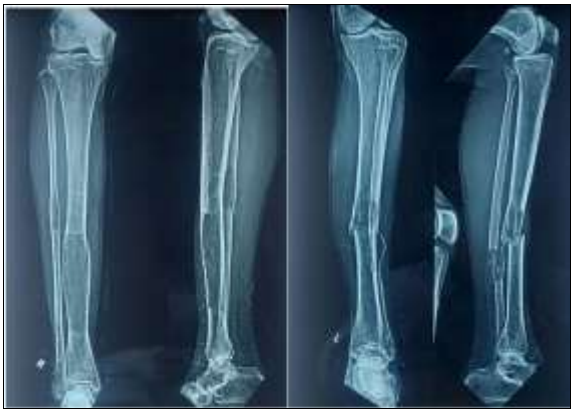

Fig 1: Radiographs Anteroposterior and lateral view of bilateral tibia.

Bilateral tibia X-ray showing a lytic lesion in the mid-shaft diaphyseal region, which has a typical ground-glass appearance. There is associated pathological fracture in both thighs

Fig 2: Anteroposterior radiograph of the humerus

The X-ray of the humerus shows a lytic lesion and ground-glass appearance.

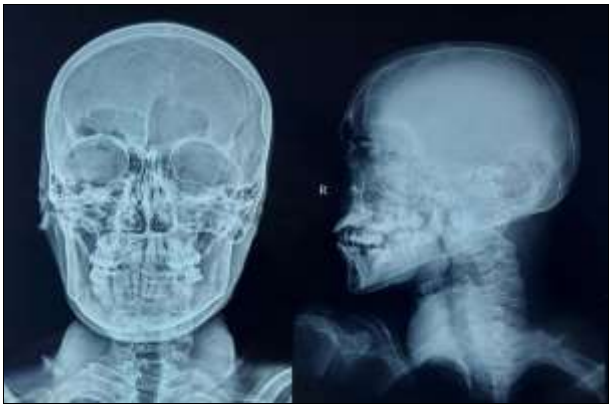

Fig 3: Anteroposterior and lateral X-ray of skull

No lesions were seen on the skull.

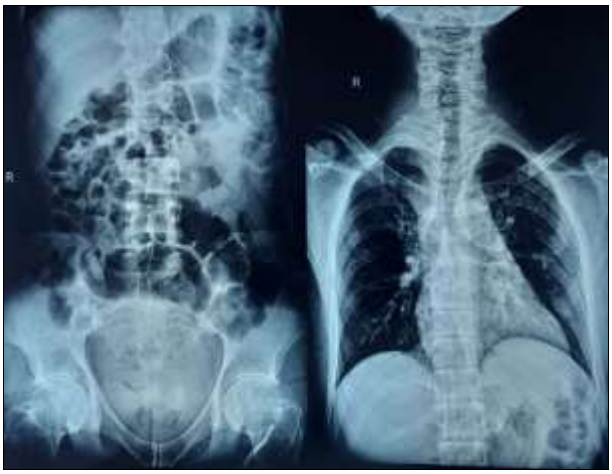

Fig 4: X-ray of whole spine (Antero-posterior view)

No lesions were seen on the X-ray

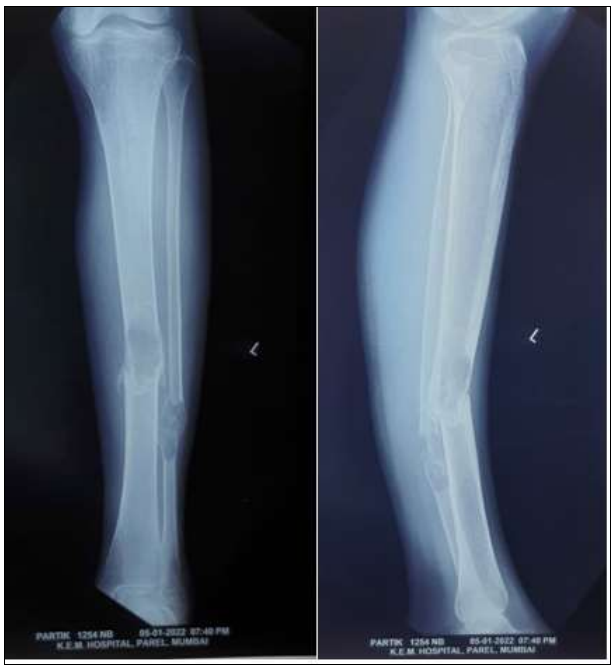

Fig 5: 6-week post trauma radiograph anteroposterior and lateral views of the left tibia

Pathological fracture showing callus formation and healing with recurvatum deformity within 6 weeks.

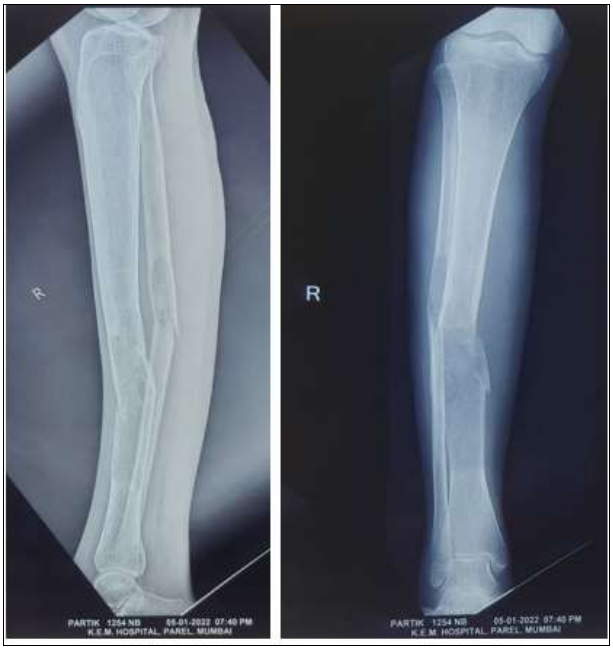

Fig 6: 6-week post-trauma radiograph of the anteroposterior and lateral views of the right tibia. Pathological fracture showed callus formation and healed with recurvatum deformity within 6 weeks

Conclusion

A 22-year-old man presented with an unusual presentation of polyostotic fibrous dysplasia of bilateral tibia and pathological fracture. He was treated with conservative management due to associated hyperthyroidism and thyroid storm, showing unexceptionally faster healing.

Clinical message

A rare case of polyostotic fibrous dysplasia with endocrinopathy showing a faster healing rate can be treated conservatively without the need for internal fixation with autogenous or allogenous bone grafts.

Acknowledgments

Dr. SS bava examined the patient with respect to pathological fracture. Dr. Sujata helped with radiological examination. Dr. Rajendra phunde was involved in the literature review. Dr. Prince Uchadiya was involved in patient management and was a major contributor to the manuscript. All authors have read and approved the manuscript. There is no conflict of interest.

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