Warpe B et al. GAIMS J Med Sci 2024;4(2) (Jul-Dec):157-162
Online ISSN: 2583-1763

Case Report

An Unusual Case of Osteofibrous Dysplasia of Tibia in a Female Child

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ABSTRACT

Osteofibrous dysplasia (OFD) is an uncommon bone tumor characterized by a non-cancerous, developmental skeletal disorder. It comprises a fibrous matrix with immature bone tissue surrounded by osteoblasts, typically located in the cortex of long bones. Distinguishing between osteofibrous dysplasia, fibrous dysplasia, classical adamantinoma, and osteofibrous dysplasia-like adamantinoma can pose a diagnostic challenge. OFD is challenging to diagnose on histopathology alone without radiological and clinical correlation and we highlight the importance of the same. Therefore, initial clinico-radiological correlation must be followed by histopathological examination in guiding prompt inception of timely treatment. Here, we present a unique, uncommon case of OFD in a seven-year-old girl, wherein pathological fractures occurred due to an underlying bone lesion incidentally detected on X-ray.

Keywords: Non-neoplastic, Osteofibrous dysplasia, Tibia

INTRODUCTION

Osteofibrous dysplasia (OFD), also known as ossifying fibroma of the long bones, is a benign fibro-osseous developmental skeletal anomaly. It constitutes only 0.2% of all primary bone tumors.[1] The term "osteofibrous dysplasia" was coined in 1981 by Capanacci and Laus due to its histological similarity to fibrous dysplasia.[2,3] Osteofibrous dysplasia tends to affect males slightly more than females, with children under the age of ten years being the most commonly affected demographic population.[4] It is a rare bone tumor that originates exclusively from the midshaft of the tibia.[5] Osteofibrous dysplasia typically manifests as a painless lesion, often accompanied by bowing of the tibia or pathological fractures, although lesions may also be incidentally discovered during imaging studies.[6] Here, we present a similarly rare case with a correlation between clinical-pathological and clinico-radiological findings.

CASE REPORT

A seven-year-old girl was brought to the orthopedic department by her parents, complaining of pain in the lower right leg region persisting for five years.

Her medical history revealed a playful fall in 2018, during which a cystic bony lesion was incidentally discovered in the mid and lower shaft of her right tibia. At that time, she underwent conservative management with pain relief medication and splints. In 2020, she experienced another fall from a height of three feet, resulting in a fracture of the mid-shaft of her right tibia, which was treated with Ender's nail insertion. The implant was removed six months after surgery. Subsequently, in March 2021, she underwent surgery for the insertion of a telescopic rod to prevent pathological fractures due to the underlying cystic lesion. Following this procedure, the patient regained the ability to walk but continued to experience tenderness at the surgical site. The tenderness

worsened with exertion but improved with rest and medication.

Upon physical examination at our facility, mild swelling was observed on the medial aspect of the right lower leg, along with healed surgical scars on both sides of the right proximal tibia and in the midline. Laboratory investigations revealed a hemoglobin level of 11.3 g%, a white blood cell count of 10,220, relative neutrophilia on peripheral blood smear, normal platelet count, and normal renal and liver function tests except for elevated serum alkaline phosphatase levels. C-reactive protein levels were within normal limits.

X-ray imaging showed a lesion along the long axis of the right tibia (Figure 1), appearing radiolucent with sharp margins and a narrow transition zone defined by well-circumscribed borders. Focal resorption of the inner layer of the cortex, indicative of endosteal scalloping, was also observed, along with a fibrous component within the lesion. There was no evidence of calcification or involvement of adjacent soft tissue. Given the patient's young age, the anatomical location, and the appearance of the lesion, senior radiologists suggested a differential diagnosis including benign conditions such as osteofibrous dysplasia, adamantinoma, giant-cell tumor, fibrous dysplasia, and osteoid osteoma.

In order to arrive at a definitive diagnosis, we needed to correlate the radiological findings with histopathological evidence. Therefore, in mid-September of 2023, a percutaneous biopsy was performed by an orthopedic surgeon using an 11-G Jamshidi biopsy needle to confirm the diagnosis and guide further management. Tissue samples were obtained from the distal tibia on the right side, specifically from the bony lesion. These samples, appearing as multiple fragmented tissue bits, gray-white in color, measured a total of 1.5x0.7x0.3cm and were received for histopathological examination in a single block.

Upon microscopic examination, the biopsy sections revealed two distinct components: bony trabeculae and fibrous strands. The predominant fibrous stromal component consisted of spindle to stellate cells arranged in a storiform pattern and in short fascicles within a myxoid hemorrhagic background. Some areas of the stroma exhibited hyalinization. Bony trabeculae and fragments were interspersed within the fibrous stroma, exhibiting characteristics of immature woven bone as well as peripheral mature lamellar bone. Osteoblasts were observed rimming the woven and lamellar bony trabeculae. Additionally, numerous scattered multinucleated osteoclastic giant cells and hemosiderin pigmentation were noted in the background (Figure 2 to 6).

IHC was not done in our case due to cost restraints. The clinico-radiological findings of the case typically coincide with histopathology findings of OFD. The epithelial cell clusters are absent in OFD, like in our case on multiple tissue sections studied.

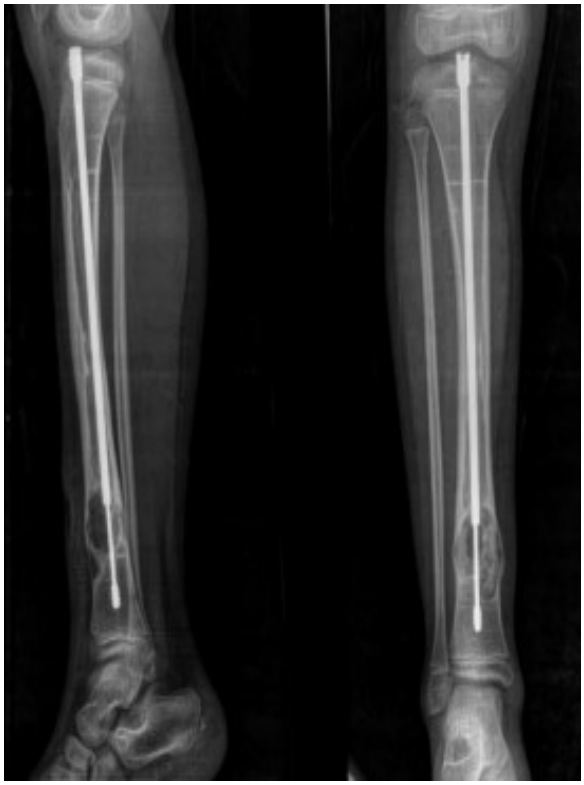

Figure 1: Radiological X-ray image - Lateral and anterior X-ray of right tibia showing old united fracture of tibia with telescopic nail in-situ and cystic expansile lesion with well defined margin at the lower end.

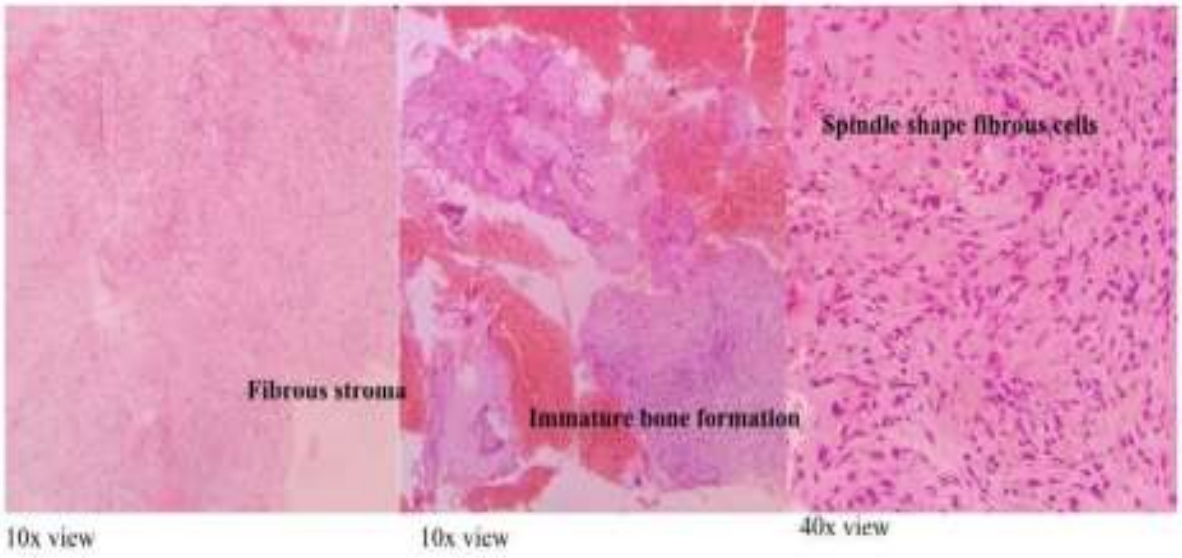

Figure 2: Microphotograph of tibial mass depicting benign fibrous stroma in short interlacing fascicles (H&E, X100).

Figure 3: Microphotograph of tibial mass depicting immature woven bone. Background shows marked hemorrhage with fragments of dead mature lamellar bone at the left of image (H&E, X100).

Figure 4: Microphotograph of tibial mass depicting benign fibrous spindle stromal cells arranged in short interlacing fascicles (H&E, X400).

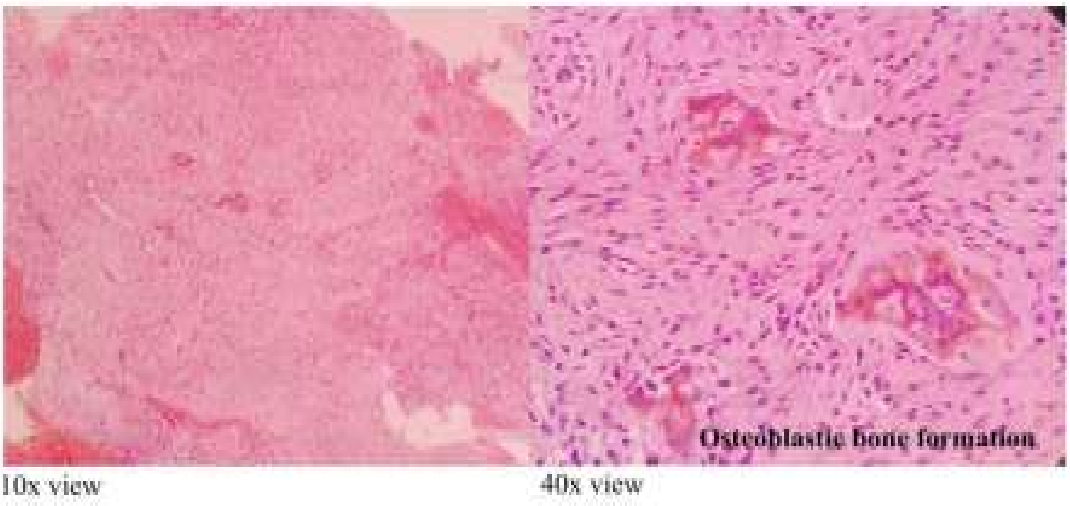

Figure 5: Microphotograph of tibial mass depicting benign fibrous stroma in short interlacing fascicles with intervening areas of immature woven bone (H&E, X100).

Figure 6: Microphotograph of tibial mass depicting immature woven one with rimmed osteoblasts (H&E, X400).

DISCUSSION

OFD manifests as a benign fibroosseous lesion predominantly found within the cortical bone. Histologically, it is characterized by a fibrous stroma intertwined with irregularly shaped, immature trabeculae of woven bone, typically encircled by active osteoblasts. OFD exhibits a predilection for the tibia, as observed in our case, often manifesting unilaterally, most frequently in the anterior mid-diaphysis region. It rarely extends into the metaphysis. While the tibia is the primary site of involvement, isolated occurrences affecting other bones such as the fibula, radius, ulna, humerus, or clavicle have been sparsely reported. Clinically, OFD presents as a painless fusiform swelling along the tibial diaphysis, often accompanied by local tenderness. It is frequently detected incidentally during X-ray examinations conducted for unrelated reasons. The dysplastic nature of the affected bone renders it fragile, predisposing individuals to pathological fractures or progressive bone deformities, such as tibial bowing.[7]

X-ray serves as the primary diagnostic tool for evaluating OFD. In radiological examination, distinct eccentric intracortically located osteolytic lesions are typically observed, characterized by well-defined margins that are sclerotic and circumscribed, with an inner ground glass density. Additional findings may include cortical expansion and anterior bowing, as well as occasional thinning and disruption of the cortex. The lesion itself may present with a lobular or bubbly appearance, often displaying multiple lytic areas. Notably, there is an absence of periosteal reaction, and the overall appearance is non-aggressive.[7]

Diagnosis holds significant importance in guiding therapeutic strategies. Treatment predominantly leans towards conservative measures, as the progression of the lesion tends to be slow and halts with skeletal maturity. However, in cases where surgery is necessary due to severe morbidity, excision surgery is preferred over curettage to minimize the risk of recurrence, particularly in younger patients.[8,9] Surgical interventions are generally deferred until skeletal maturity, considering OFD's self-limiting nature. However, in instances where surgical intervention is warranted; for instance, in cases of bowing deformity, pathological fracture, or the presence of a sizable mass; it is recommended.

As part of our patient's treatment plan, it was recommended to remove the telescopic rod and proceed with curettage, bone grafting, and fixation using plates. Regrettably, the patient failed to attend scheduled follow-up appointments.

Osteofibrous dysplasia (OFD), osteofibrous dysplasia-like adamantinoma (OFD-LA), and classic adamantinoma are closely related lesions typically located in the mid tibial diaphysis. Distinguishing between them involves various factors such as age, lesion characteristics, location, radiological and histological appearance, as well as clinical course.[8] However; the definitive diagnosis heavily relies on histopathological reports, which are considered the gold standard.[9] Nevertheless, it's essential to conduct a thorough clinicoradiological assessment before histopathology in cases of skeletal lesions.

Classic adamantinoma, a variant of adamantinoma, is a rare malignant primary fibro-osseous bone tumor primarily occurring in the tibial diaphysis of young adults, with an age range of 4 to 75 years and a median age of 30.8 years. Unlike OFD, it typically manifests in mature skeletons. Classic adamantinoma exhibits an indolent course but has a high recurrence rate, especially with incomplete excision. Its histogenesis remains uncertain, but the presence of obvious basaloid epithelial elements within the fibro-osseous stroma is characteristic. In this instance, the stromal component took a backseat to the epithelial component, with the epithelial characteristics exhibiting more prominence. The epithelial features displayed enhanced development, characterized by cells with round nuclei and a greater abundance of eosinophilic or amphophilic cytoplasm arranged in basaloid or tubular patterns.These epithelial cell clusters are absent in OFD. Radiologically, classic adamantinoma demonstrates complete involvement of the medullary cavity, extension into adjacent soft tissues, skip lesions in the ipsilateral fibula, and

moth-eaten bone appearance. It exhibits local aggressiveness, with potential for distant metastasis and poor prognosis, necessitating extensive surgical intervention. While OFD may share some radiological features, its medullary involvement is typically partial.[2,7]

Osteofibrous dysplasia-like (differentiated) adamantinoma (OFD-LA) represents a less aggressive variant compared to classic adamantinoma, manifesting as strictly intracortical with a slow progressive nature. OFD-LA exhibits superior clinical outcomes when compared to classic adamantinoma. OFD-LA occurs in the age range of the pediatric population with an average age of 14 years. It involves the shaft of the tibia anteriorly. It is a locally aggressive, intermediate WHO grade bone tumor with both malignant and benign features. This variant is distinguished by inconspicuous clusters of epithelial cells (keratin-positive) nestled within the fibro-osseous stroma, serving as its defining characteristic. The stromal spindle component holds greater prevalence. However, the bony aspect differs from that observed in OFD. The trabeculae display less branching and anastomosing, with less prominent osteoblasts, and absence of clustered osteoclasts if present.[2]

Fibrous dysplasia (FD) primarily affects children and young adults, representing a developmental bone abnormality that can occur in any bone. In its monostotic form, which involves a single bone, common sites include the ribs, craniofacial bones, and femur. FD typically manifests with an intramedullary location and without cortical destruction, presenting as a benign fibro-osseous lesion. Radiologically, thinning of the cortex may be observed, although deformity is rarely evident.[7] Histologically, FD exhibits characteristics such as the absence of zonation pattern, lack of maturation to lamellar bone, infrequent osteoblastic rimming of bony trabeculae, and absence of keratin-positive cells in the stroma. The histopathological features indicate a failure in bone maturation, with arrested development resulting in immature woven bone formation.[7] Previously, FD was referred to as osteitis fibrosa or generalized fibrocystic disease of bone. Surgery is generally not indicated, as this lesion tends to be self-limiting and responsive to hormonal changes during puberty.

Osteoid osteoma is a benign bone tumor that predominantly affects children, characterized by a nidus smaller than 2 cm in diameter on plain radiographs. Unlike OFD, it may exhibit a solid periosteal reaction with cortical thickening. Patients may experience nocturnal pain relieved by NSAIDs. Histologically, osteoid osteoma shows osteoid deposition at the center and haphazardly arranged woven bone trabeculae with prominent osteoblastic rimming. Zonation phenomena are absent. Fifty percent of cases occur in the long bones of the lower limbs, with common sites including the femoral neck and the end of the diaphysis of long bones. This tumor typically has an excellent prognosis, with rare or absent recurrence. Cortical involvement occurs in 75% of cases, while intramedullary or subperiosteal involvement is rare.

OFD can sometimes resemble giant cell tumors of bones (GCT) in radiographic appearance.[7] Unlike OFD, GCT typically occurs in mature skeletons. GCT presents with well-defined, non-sclerotic margins. Histologically, it exhibits a characteristic morphology characterized by numerous osteoclast-like giant cells embedded within mononuclear neoplastic cells in the stroma. Tumor cells do not deposit osteoid. GCT primarily affects the epiphysis and infrequently the metaphysis of long bones, with common sites including the distal femur and proximal tibia.[7]

Malignant bone tumors, such as Ewing's sarcoma (ES), are aggressive and demonstrate periosteal reaction, resulting in an "onion skin" radiological appearance, unlike OFD.[7] ES ranks as the second most common malignant bone tumor, predominantly affecting children and young adults, with rarity above the age of 30. It often occurs in the bony diaphysis, particularly in the long bones of the lower limbs. Common clinical presentations include a painful enlarging bony mass. Treatment typically involves neoadjuvant chemotherapy followed by surgery. Histopathologically, ES is characterized by small round tumor cells arranged in clusters and nests.

CONCLUSION

Osteofibrous dysplasia (OFD) represents a developmental bone irregularity that primarily impacts long bones. Typically, conservative measures suffice for treatment, unless there are pathological fractures or significant bone deformities. Prior to any biopsy procedure, it's crucial to establish a correlation between clinical and radiological findings. Histopathology serves as the cornerstone for definitive diagnosis, distinguishing OFD from other potential conditions. It's worth noting that OFD is seldom documented in medical literature due to rare conditions and lack of adequate case reporting.

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Source of support: Nil

Conflict of interest: None declared

How to cite: Warpe B, Joshi S, Patel K, Solanki R. An Unusual Case of Osteofibrous Dysplasia of Tibia in a Female Child. GAIMS J Med Sci 2024;4(2):157-162.

https://doi.org/10.5281/zenodo.13353655