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Osteofibrous Dysplasia in a 4-Month-Old: A Rare Presentation in Early Infancy

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Learning Point of the Article:

Osteofibrous dysplasia presents as a lytic, tender swelling in infants, which is confirmed with early imaging and biopsy, and then treated conservatively with close monitoring in uncomplicated cases.

Abstract

Introduction: Osteofibrous dysplasia (OFD) is a non-malignant lesion of cortical bone, where fibrous tissue replaces normal bone. Its presentation is rare in infants and may clinically and radiologically mimic other malignant lesions such as adamantinoma, making diagnosis challenging. Biopsy and imaging play a key role in diagnosis.

Case Report: The patient, a 4-month-old infant of Indian origin, presented with a tender swelling over the proximal aspect of the right tibia. Radiographic evaluation revealed a lytic lesion. Further magnetic resonance imaging and histopathological evaluation confirmed the diagnosis of OFD. We planned to treat it conservatively and monitor the lesion size regularly, given the patient's age and absence of complications. Surgical intervention was deferred unless there is progression or functional compromise.

Conclusion: This case highlights the diagnostic challenges of OFD in infants and its differentiation from malignant lesions. Through this report, we aim to broaden clinical awareness regarding the occurrence of OFD in young infants and emphasize the role of imaging and biopsy in establishing a definitive diagnosis.

Keywords: Osteofibrous dysplasia, pediatric orthopedics, tibial swelling, infant bone lesions, fibro-osseous tumor.

Introduction

Osteofibrous dysplasia (OFD) is a benign fibro-osseous condition of bone that typically affects the cortical bone of the anterior mid-shaft of the tibia in children [1]. The term OFD was first devised by Dahlin and Johnson in 1972 [2]. The usual age of presentation of OFD ranges from 7 days to 22 years [3]. Skeletal neoplasms of the newborn and neonates are rarely encountered, and only a few cases have been reported. It is frequently misdiagnosed owing to a lack of distinctive radiographic findings and inadequate sampling materials [4]. OFD typically presents as a monostotic lesion, affecting a single bone in about 85% of cases. However, 5% of cases show polyostotic involvement (multiple bones affected) [5]. This report describes a rare case of OFD in a 4-month-old infant.

Case Report

A 4-month-old male infant was brought to D. Y. Patil Hospital by his mother with a chief complaints of swelling in the right lower limb for the last 10 days. There was no history of trauma, fall, or injury. The child was irritable and crying, but was feeding well.

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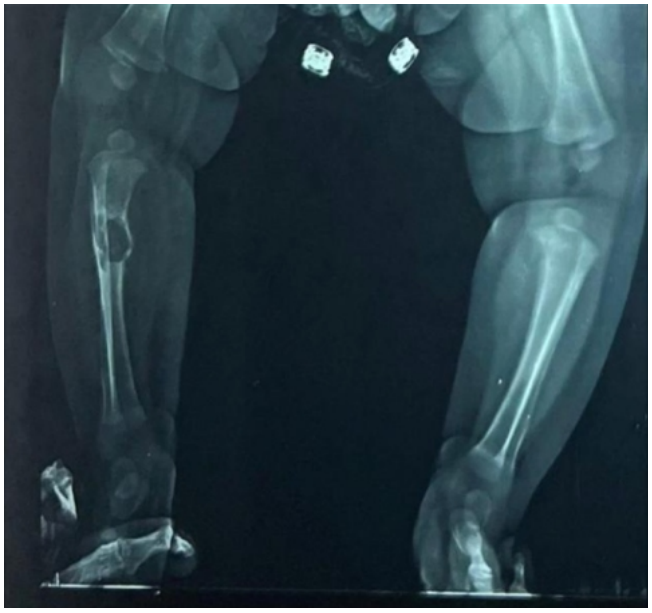

Figure 1: Anteroposterior X-rays.

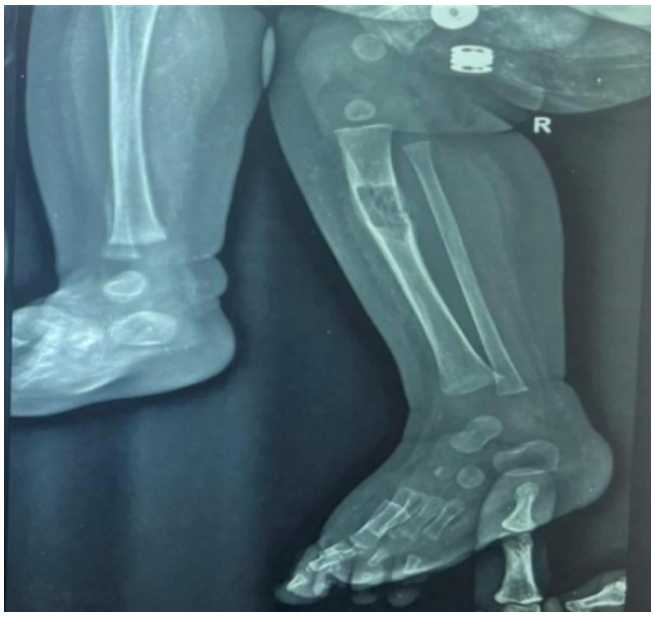

Figure 2: Lateral X-rays.

The swelling was bony, hard, and non-progressive over the proximal tibia. Tenderness was noted over the swelling, indicated by the infant's crying during palpation. The swelling was firmly fixed, without any discharging sinuses. He had no history of fever and chills. No associated history of any trauma, ENT or ocular bleeding, loss of consciousness, or vomiting, and was then admitted for further evaluation.

The patient had no significant medical history. He had a normal full-term delivery with no significant birth complications.

Developmental milestones were age-appropriate for 4 months. He was fully immunized for his age. Family history was notable for a non-consanguineous marriage. No known family history

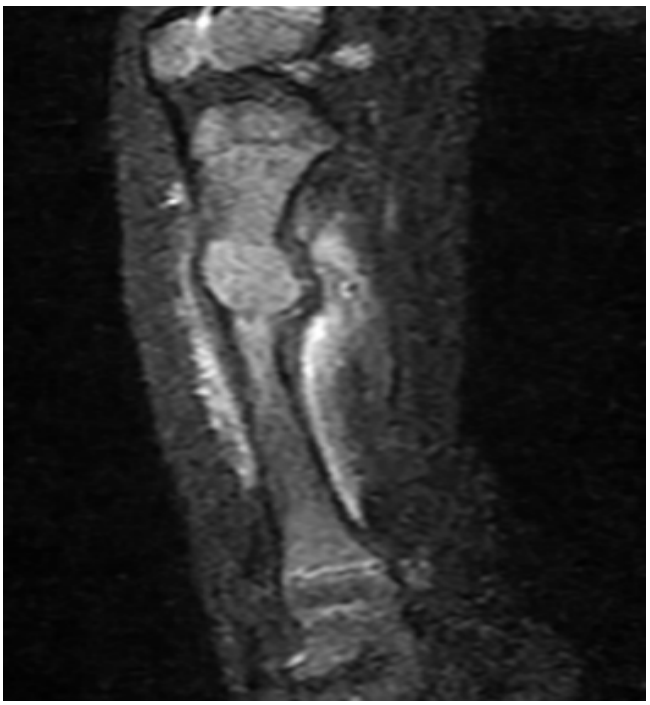

Figure 3: Sagittal T1-weighted magnetic resonance imaging shows an expansile mass replacing the medullary space, showing intermediate intensity signals.

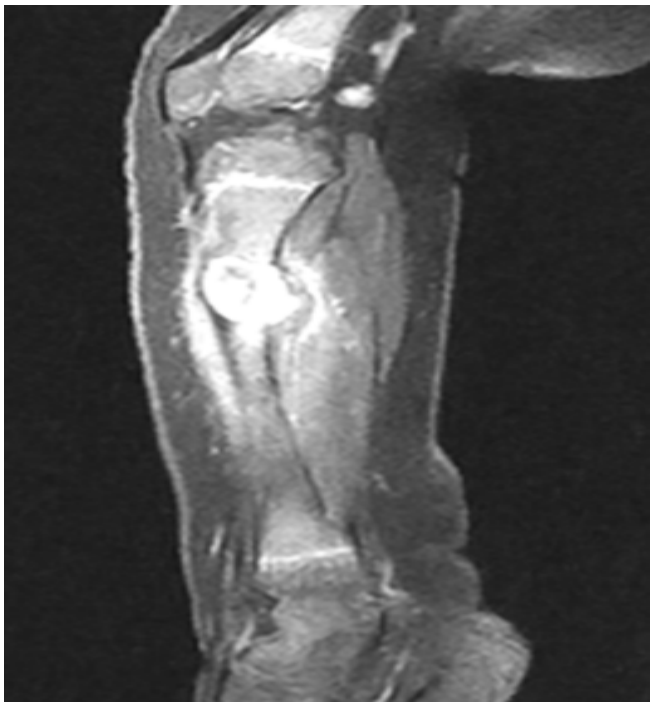

Figure 4: Sagittal T2-weighted magnetic resonance imaging reveals heterogeneous hyperintense signal intensity.

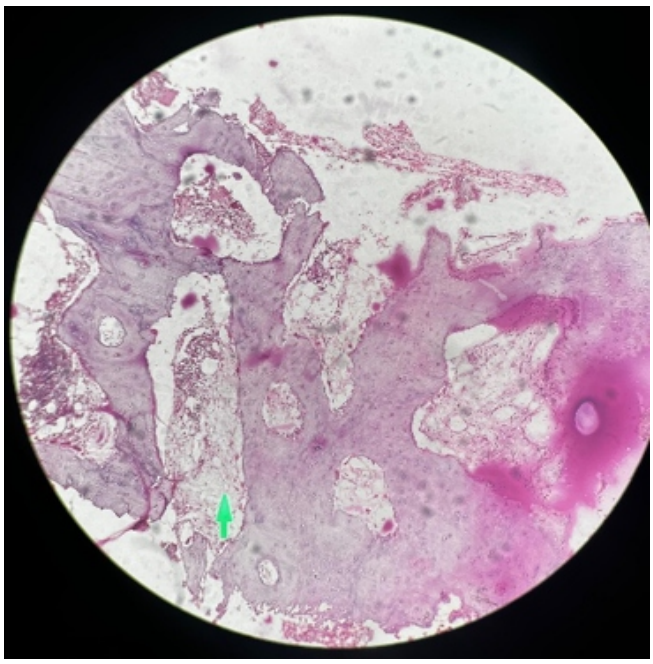

Figure 5: Woven bone trabeculae rimmed by osteoblasts.

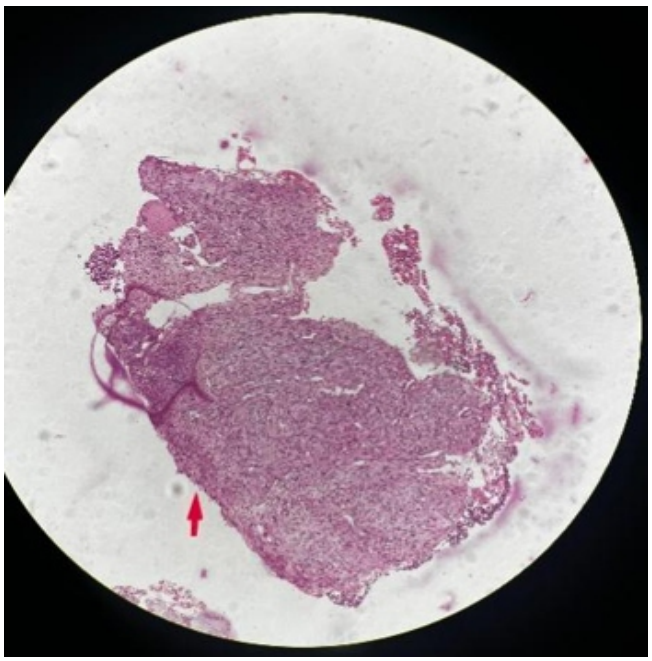

Figure 6: Bland fibroblastic stroma.

of genetic disorders or similar illnesses. The patient was breastfed from birth. On general examination, the patient was conscious and alert, responding to auditory and visual stimuli, recognized his mother, and had an appropriate cry for his age. He was afebrile, with a pulse rate of 120/min, and SPO2 of 100% on room air. Pallor, icterus, cyanosis, clubbing, lymphadenopathy, and edema were all negative.

On inspection of the right lower limb, a diffuse swelling measuring 1.7×1 cm was noted over the proximal tibia. No scars, erythema, dilated veins, tattoo marks, discolouration, fissure, sinus, or ecchymoses were present. There was no local rise in temperature. Toe movement and sensation were present. Distal pulses were palpable.

X-ray reports revealed an ill-defined expansile lytic lesion

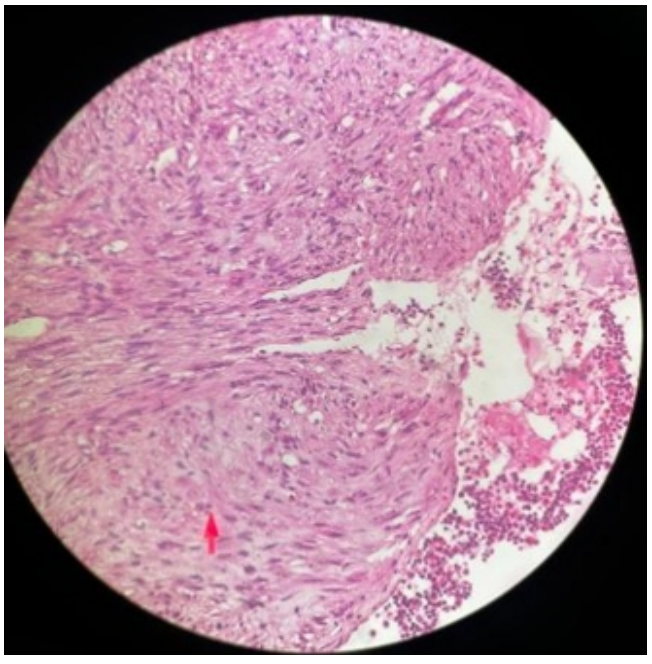

Figure 7: Fibrous stroma with spindle to stellate-shaped cells.

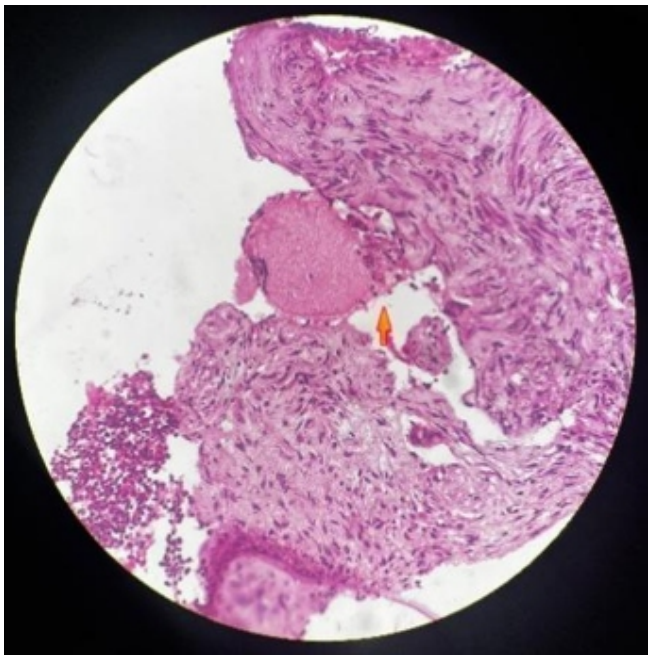

Figure 8: Multinucleated giant cell in the fibrous stroma.

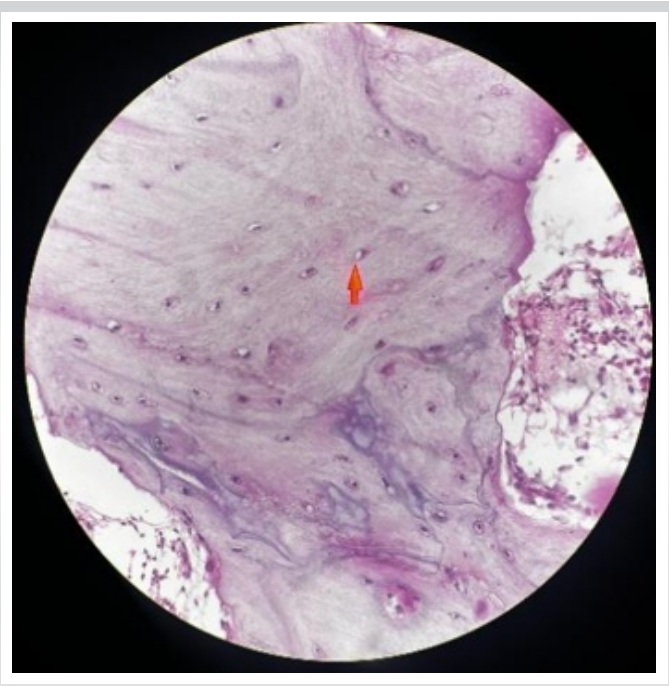

Figure 9: Woven bone with osteocytes in the lacunae.

involving the diaphysis of the proximal right tibia.

Therefore, we then performed a J-needle biopsy with above knee cast application. The sample was sent for histopathology examination, which showed a lesion composed of fibrous stroma and bony trabeculae, which was lined by osteoblasts.

Radiographs

Anteroposterior (Fig. 1) and lateral (Fig. 2) X-rays demonstrate an expansile osteolytic lesion involving the medullary cavity of the proximal diaphysis of the right tibia. The margins were well-defined, multilobulated and irregular.

MRI

Proton density fat saturated images demonstrated hyperintense lesions with few hypointense areas. (Fig 3 and 4).

Histopathology (Fig. 5, 6, 7, 8, 9)

Histopathology revealed bony trabeculae lined by osteoblasts within a fibrous stroma

Discussion

OFD is a rare, benign, fibro-osseous cortical bone lesion most commonly found in children, typically between 1 and 10 years of age, and predominantly affecting the anterior tibial cortex [1]. Neonatal or infantile presentation is extremely rare, with very few cases reported worldwide. The earliest documented case was in a 3-day-old neonate, described by Castaldo et al., who presented with a tibial swelling later confirmed as OFD on biopsy [6]. Another case by Hindman et al. reported a pathological fracture of the tibia at birth due to congenital OFD [7].

Our case of a 4-month-old infant thus represents a rare early-age manifestation, expanding the known age spectrum of OFD and highlighting the importance of including it in the differential diagnosis of tibial lesions, even in infancy.

Clinical and radiological features

The child presented with a firm, non-mobile, tender swelling over the proximal tibia without systemic signs such as fever or trauma. These features are consistent with typical OFD presentations [1, 8].

Radiographs showed a well-defined, eccentric, expansile lytic lesion with multilobulated contours, involving the diaphyseal cortex of the proximal tibia – hallmark radiological features of OFD [1,5]. MRI confirmed intermediate T1 and heterogeneously hyperintense T2 signal changes within the lesion, with no soft-tissue involvement or periosteal reaction – features supportive of a benign fibro-osseous process [6, 9].

Radiographic surveillance is recommended every 6–12 months until skeletal maturity, particularly for:

- Large lesions
- Unusual sites (e.g., fibula, radius)
- Patients <5 years at diagnosis
- Suspected recurrence or progression.

MRI should be repeated in case of symptom changes or rapid lesion growth, to exclude soft-tissue invasion or malignant transformation [8, 9].

Histopathology and differential diagnosis

Histopathology remains the gold standard for OFD diagnosis. The biopsy in our case revealed woven bone trabeculae lined by osteoblasts within a fibrous stroma, a confirmatory feature of OFD [1,10].

Differentiation from other fibro-osseous lesions is essential:

- Fibrous dysplasia lacks osteoblastic rimming and tends to be more medullary rather than cortical [10].
- Classic adamantinoma includes nests of epithelial cells that stain positive for cytokeratin, indicating malignancy [10].
- OFD-like adamantinoma, an intermediate lesion, shows sparse epithelial elements but can behave more aggressively [9, 10].

The exact etiology of OFD remains uncertain. It is considered a

developmental lesion, not a true neoplasm. Some studies suggest a potential link between OFD and adamantinoma due to histological and radiological similarities.

Molecular studies have identified cytokeratin expression in stromal cells of some OFD cases, blurring the lines between OFD and OFD-like adamantinoma [9,10]. While adamantinoma exhibits epithelial gene expression (e.g., keratin-14 and -19), OFD does not consistently show this. However, rare progression of OFD to adamantinoma has been described, making long-term vigilance essential, even in histologically benign lesions [9].

Moreover, GNAS mutations, which are commonly found in fibrous dysplasia, are typically absent in OFD, helping distinguish the two entities at the molecular level [10].

Some experts view OFD and OFD-like adamantinoma as part of a spectrum, where OFD may be a precursor lesion to adamantinoma. OFD-like adamantinoma contains sparse epithelial cells and has intermediate behavior. However, no clear progression pathway has been proven definitively [9].

This overlap has led to ongoing controversy in the classification and nomenclature of these lesions. Immunohistochemistry (cytokeratin positivity) and close clinical monitoring are crucial to distinguish benign from low-grade malignant lesions, especially in recurrent or aggressive cases.

Thus, histology and immunohistochemistry play a crucial role in distinguishing these entities.

Management

OFD is considered self-limiting, often regressing spontaneously after skeletal maturity. Studies show that conservative management (biopsy and observation) is usually sufficient, particularly when deformity and fracture risk are low [1,8].

Dala-Ali et al. reported in a multicenter review that angular deformity <10° at presentation predicted good outcomes without surgery [11]. Recurrence rates post-curettage are high (~70%), but malignant transformation is exceedingly rare [1,11].

In our case, a diagnostic biopsy and protective above-knee cast were sufficient to manage the child, given the absence of deformity or fracture. Surgical intervention, such as curettage or resection, should be reserved for progressive deformity, functional impairment, or ambiguous diagnosis [8,11].

Children diagnosed with OFD require regular follow-up with radiographs, especially during growth spurts, to monitor lesion size, cortical expansion, and tibial alignment [8,11]. Although most regress or remain stable, rare progression or deformity (especially anterior bowing) may develop and should be addressed promptly.

Conclusion

This case highlights an unusual presentation of OFD in early infancy and contributes to the limited existing literature on its occurrence at such a young age. Reporting rare infantile presentations broadens the recognized clinical spectrum of the disease and improves understanding of its natural history. Increased awareness of OFD in infants may support more informed clinical decision-making and help optimize long-term outcomes through age-appropriate management strategies.

Clinical Message

Although OFD is uncommon in infancy, it should be included in the differential diagnosis of lytic tibial lesions in the pediatric population to avoid misdiagnosis. Awareness of this entity allows for appropriate evaluation and timely diagnosis, thereby preventing unnecessary aggressive treatment. Conservative management with regular radiological follow-up is a safe and effective approach in the absence of progression or functional impairment.

Declaration of patient consent: The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given the consent for his/ her images and other clinical information to be reported in the journal. The patient understands that his/ her names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Conflict of interest: Nil **Source of support:** None

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