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Diagnostic Dilemma in a Distal Femoral Intramedullary Lesion: Enchondroma Mimicking Low-Grade Chondrosarcoma with Discordant Biopsy Findings: A Case Report

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Learning Point of the Article:

Discordant imaging and biopsy findings in cartilaginous bone tumours should prompt repeat biopsy and multidisciplinary evaluation, as accurate clinico-radiological correlation is essential to distinguish enchondroma from low-grade chondrosarcoma and guide appropriate treatment.

Abstract

Introduction: Differentiating enchondroma from low-grade chondrosarcoma remains challenging due to significant overlap in clinical presentation, imaging characteristics, and histopathological appearance. Accurate diagnosis is crucial in guiding appropriate treatment and preventing unnecessary aggressive management.

Case Report: A 25-year-old female presented with progressive right knee pain for 20 days. Te X-ray was inconclusive. Magnetic resonance imaging demonstrated a well-defined intramedullary lesion within the distal femur measuring approximately 7.6 × 2.6 × 3 cm. Initial computed tomography (CT)-guided biopsy results were inconclusive. Repeat imaging, including magnetic resonance imaging and positron emission tomography-CT, suggested a non-aggressive lesion without metastatic disease. After multidisciplinary tumor board discussion, a repeat biopsy demonstrated a cartilaginous neoplasm compatible with enchondroma. The patient subsequently underwent intralesional curettage with antibiotic-loaded bone cement filling. Final histopathological analysis confirmed a benign enchondroma.

Conclusion: This report highlights the importance of clinicoradiological correlation and repeat biopsy when initial investigations provide conflicting information in suspected cartilage tumors.

Keywords: Enchondroma, distal femur tumor, intramedullary cartilage tumor, low-grade chondrosarcoma, bone tumor biopsy, diagnostic dilemma, curettage and cementation, musculoskeletal oncology.

Introduction

Cartilaginous tumors of bone represent a spectrum of lesions ranging from benign enchondromas to malignant chondrosarcomas. Accurate differentiation between these entities remains a well-known challenge in orthopedic oncology because they share overlapping clinical, radiological, and histopathological characteristics [1,2,3]. Although enchondromas are benign lesions, low-grade chondrosarcomas may present with similar imaging findings.

Enchondromas are intramedullary tumors composed of hyaline cartilage arising from residual cartilage rests in the bone marrow cavity. These lesions most frequently occur in the metaphyseal or diaphyseal regions of long bones and are often discovered incidentally during imaging studies performed for unrelated

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Author's Photo Gallery


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Figure 1: Initial plain radiograph of the right knee joint (anteroposterior and lateral views) showing no definite abnormality in the distal femur. The lesion was not clearly appreciable on radiographs.

conditions [1]. While many enchondromas remain asymptomatic, some patients may present with pain, swelling, or a pathological fracture.

The presence of persistent pain in a cartilage tumor located in a weight-bearing bone, such as the femur, raises concern for malignancy. Low-grade chondrosarcoma represents a slow-growing malignant cartilage tumor that may closely resemble an enchondroma on both radiological and histological examination [2]. Radiological signs suggesting malignancy include endosteal scalloping involving more than two-thirds of cortical thickness, cortical disruption, periosteal reaction, and soft-tissue extension [4,5].

Histopathological diagnosis can also be challenging because cartilage tumors frequently demonstrate variable cellularity and architecture. Sampling errors or inadequate biopsy specimens may lead to inconclusive or misleading results. Consequently, management decisions should rely on a combination of clinical presentation, imaging findings, histopathology, and multidisciplinary discussion.

This report describes a case of distal femoral enchondroma presenting as a diagnostic dilemma due to discordant biopsy findings, emphasizing the role of repeat biopsy and comprehensive evaluation in establishing the correct diagnosis.

Case Report

A 25-year-old female presented to the orthopedic outpatient department complaining of pain in the right knee for approximately 20 days. The pain had developed gradually without any identifiable precipitating event. It was described as a dull aching discomfort localized around the knee joint without radiation. The symptoms progressively increased over time and were not associated with any specific aggravating or relieving factors. There was no history of trauma, fever, weight loss, night pain, or systemic symptoms. The patient denied any previous similar complaints or underlying medical conditions. Her past medical and family history was unremarkable.

Clinical examination

Physical examination revealed no visible deformity or swelling around the knee. There were no scars, sinus tracts, or dilated

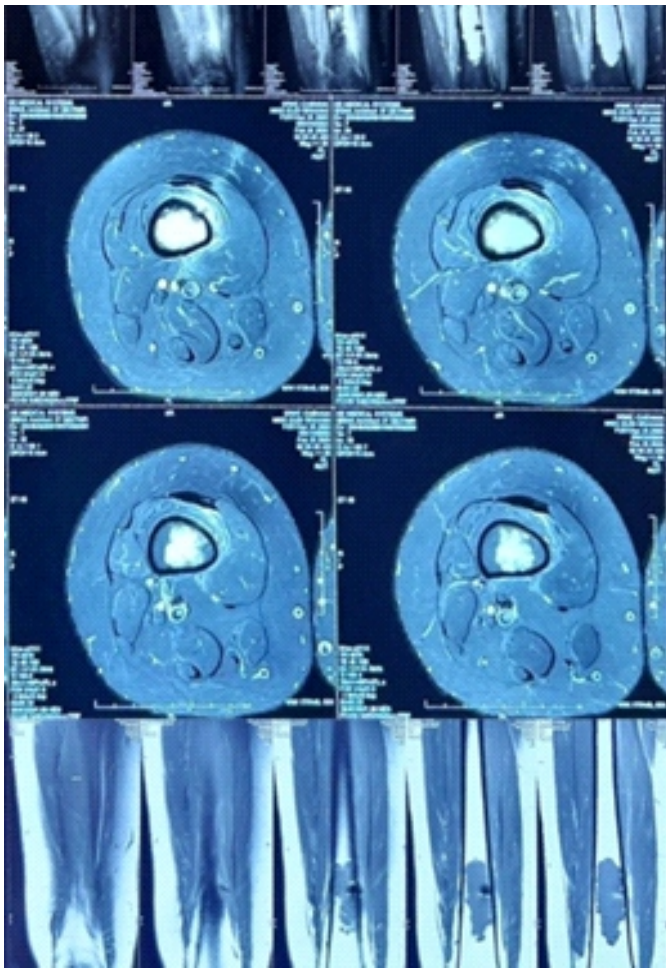

Figure 2: Repeat magnetic resonance imaging of the right femur with knee screening performed at our institution, demonstrating a well-defined lobulated intramedullary lesion involving the mid and distal femoral diaphysis. The lesion appears hypointense on T1-weighted sequences and hyperintense on T2/short tau inversion recovery images without significant cortical destruction, periosteal reaction, or soft-tissue extension, features suggestive of a benign cartilaginous lesion, likely an enchondroma.

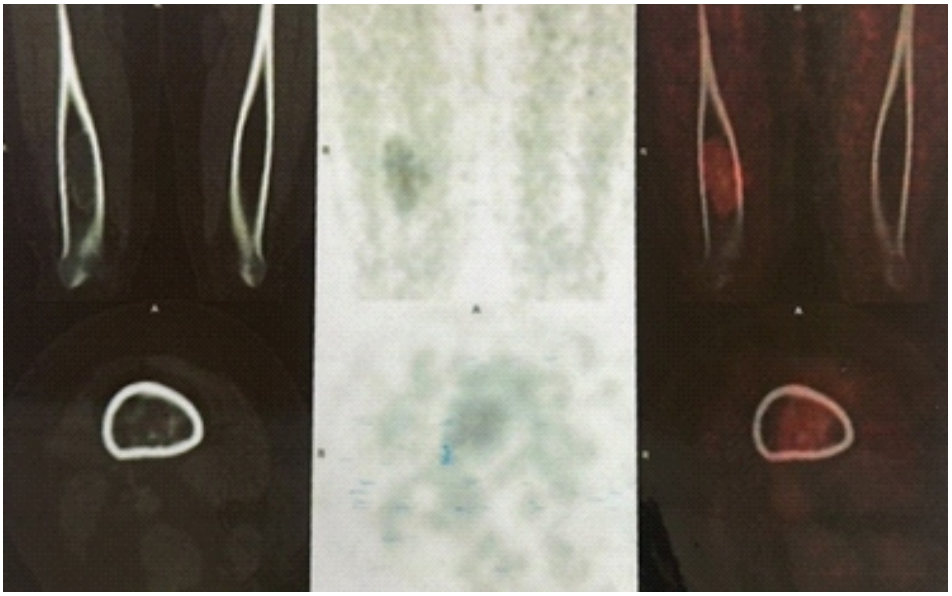

Figure 3: 18F-fluorodeoxyglucose positron emission tomography-computed tomography scan demonstrating focal low-grade metabolic uptake within the intramedullary lesion of the distal right femur (standardized uptake value max ~2.6). No additional metabolically active lesions or distant metastases are identified elsewhere in the body, supporting the likelihood of a benign cartilaginous lesion.

superficial veins. Palpation demonstrated localized tenderness over the anterior aspect of the distal femur. No local warmth was detected. The range of motion of the knee joint was from full extension to approximately 100° of flexion, with mild discomfort during deeper flexion. Sensory examination of the limb was normal, and distal pulses were palpable. Based on the clinical findings, a benign bone lesion such as fibrous dysplasia was initially suspected.

Initial imaging and biopsy

The initial X-ray taken was found to be inconclusive (Fig. 1). Magnetic resonance imaging of the right knee performed at an outside facility demonstrated a well-circumscribed, lobulated intramedullary lesion located in the distal femoral metaphyseal-diaphyseal region. The lesion measured approximately 7.6 cm in craniocaudal length, 2.6 cm transversely, and 3 cm in the anteroposterior dimension. On magnetic resonance imaging (MRI) sequences, the lesion appeared hypointense on T1-weighted images and hyperintense on short tau inversion recovery sequences. No cortical destruction, periosteal reaction, or surrounding soft-tissue mass was identified. The radiological differential diagnosis included enchondroma, low-grade chondrosarcoma, and simple bone cyst [6]. A computed tomography (CT)-guided biopsy was performed right after MRI. Histopathological analysis revealed necrotic osteoid tissue without identifiable tumor cells. As the findings were inconsistent with imaging characteristics, the biopsy slides were reviewed at another laboratory for a second opinion. The second report described fibrous tissue with areas of calcification and hyalinization, along with scattered normal bony trabeculae. No malignant cells or granulomatous inflammation were identified. These findings remained inconclusive.

Further evaluation

The patient subsequently presented to our institution after 1 week for further management. Repeat MRI performed demonstrated a well-defined non-aggressive intramedullary lesion involving the middle and distal femoral diaphysis (Fig. 2). The imaging characteristics were consistent with a benign cartilage tumor, most likely an enchondroma. To further evaluate metabolic activity, a positron emission tomography-CT (PET-CT) scan was obtained the next day (Fig. 3). The study demonstrated low-grade fluorodeoxyglucose uptake with

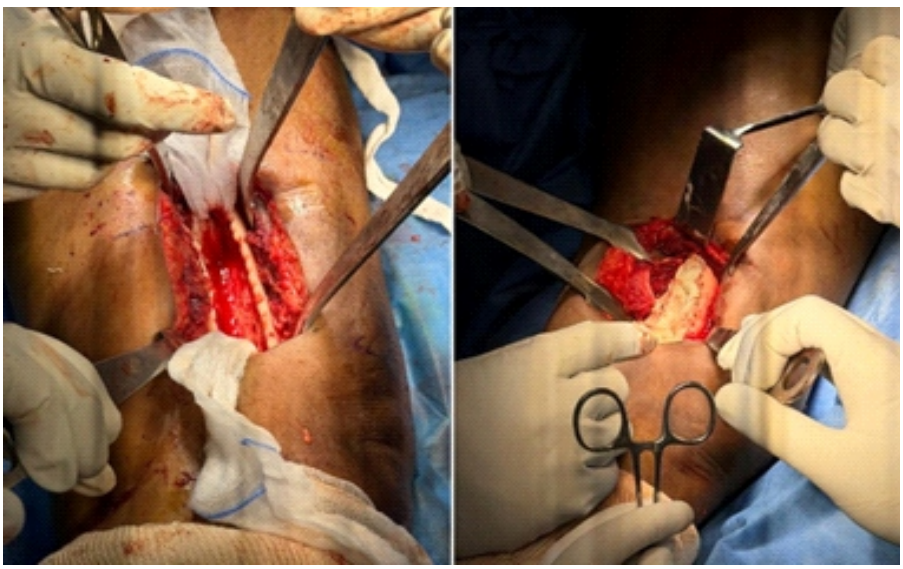

Figure 4: Intraoperative clinical photographs demonstrating exposure of the distal femur lesion through a cortical window with intralesional curettage of the tumor cavity, followed by filling of the defect with antibiotic-loaded polymethylmethacrylate bone cement.

Figure 5: Post-operative plain radiograph of the right femur (anteroposterior and lateral views) demonstrating the distal femoral cavity following intralesional curettage, filled with antibiotic-loaded polymethylmethacrylate bone cement. The cement occupies the tumor cavity with maintained cortical integrity and satisfactory post-operative alignment.

a maximum standardized uptake value (SUV) of approximately 2.6 within the lesion. No additional metabolically active lesions or distant metastases were detected. Low metabolic activity on PET-CT has been associated with benign cartilage tumors, further supporting the diagnosis of enchondroma [7].

Given the discrepancy between imaging findings and biopsy results, the case was discussed in a multidisciplinary tumor board meeting, including orthopedic oncologists, radiologists, medical oncologists, and radiation oncologists. Considering the persistent symptoms, inconclusive histopathology, and imaging findings suggestive of a benign lesion, the tumor board recommended performing a repeat CT-guided biopsy to obtain a more representative tissue sample.

Repeat CT-guided biopsy was planned and performed after 1 week. Histological examination demonstrated cartilage fragments composed of chondrocytes embedded within a hyaline matrix without features of malignancy. These findings were consistent with a cartilaginous neoplasm suggestive of enchondroma.

Based on the biopsy findings and tumor board recommendation, surgical treatment was planned. After 5 days with all the required fitness for surgery, the patient underwent tumor excision with intralesional curettage of the distal femoral lesion (Fig. 4). The resulting cavity was filled with antibiotic-loaded polymethylmethacrylate bone cement. Intralesional curettage combined with bone cement filling is widely used in the management of symptomatic enchondromas. Cement provides immediate mechanical stability, facilitates early mobilization, and allows easier radiographic detection of recurrence during follow-up (Fig. 5) [8,9,10,11].

The excised tissue specimen was sent for definitive histopathological examination (Fig. 6). Microscopic evaluation revealed lobules of mature hyaline cartilage containing chondrocytes located within lacunae. The cells exhibited bland nuclear morphology without atypia, mitotic activity, or necrosis. These findings confirmed the diagnosis of a benign cartilaginous tumor consistent with enchondroma.

Discussion

Distinguishing enchondroma from low-grade chondrosarcoma remains a difficult diagnostic problem in musculoskeletal oncology [12]. Both lesions share similar clinical manifestations and imaging appearances, which often complicates clinical decision-making.

Pain has historically been considered a feature suggestive of malignancy in cartilage tumors. However, several studies have reported that benign enchondromas may also produce pain due to mechanical stress, cortical thinning, or associated bone marrow edema [2]. Therefore, clinical symptoms alone cannot reliably distinguish between benign and malignant lesions.

Radiological evaluation remains an essential component of the diagnostic process. Enchondromas typically appear as well-defined intramedullary lesions with chondroid matrix calcifications and minimal cortical involvement. In contrast, low-grade chondrosarcomas may demonstrate cortical destruction, extensive endosteal scalloping, periosteal reaction, and soft-tissue extension [4,5].

In the present case, MRI consistently demonstrated a well-defined intramedullary lesion without aggressive radiological features. These findings strongly favored a benign cartilage tumor.

Histopathological examination is traditionally considered the gold standard for diagnosis. Nevertheless, biopsy interpretation may be complicated by sampling error, tumor heterogeneity, or insufficient tissue. Small biopsy specimens may not adequately represent the entire lesion, potentially resulting in inconclusive findings.

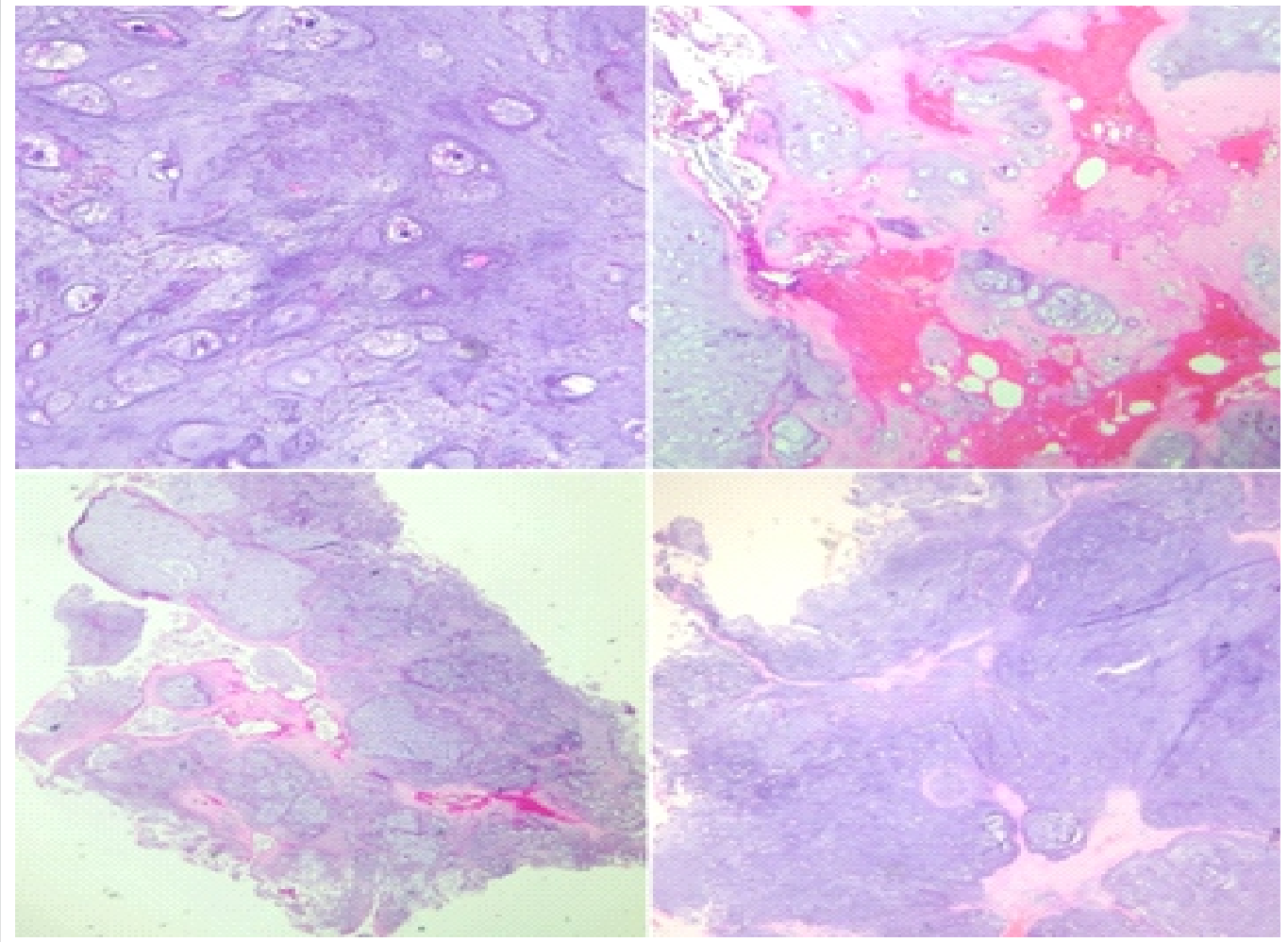

Figure 6: Histopathological photomicrographs (hematoxylin and eosin staining) demonstrating lobules of mature hyaline cartilage separated by fibrous septae. The chondrocytes are seen within well-formed lacunae exhibiting bland cytological features with relatively small, uniform nuclei and inconspicuous nucleoli. Occasional binucleated chondrocytes are present without significant nuclear atypia, increased mitotic activity, or necrosis. These findings are consistent with a benign cartilaginous lesion suggestive of enchondroma.

This challenge was illustrated in our case, where the initial biopsy failed to demonstrate tumor tissue. Only after a repeat biopsy was the cartilaginous nature of the lesion identified.

PET-CT has recently been explored as an adjunct tool for differentiating benign from malignant cartilage tumors. Previous studies have suggested that lesions demonstrating low SUVs are more likely to represent benign pathology [7]. In our patient, the SUV value of approximately 2.6 further supported a benign diagnosis.

The standard treatment for symptomatic enchondromas involves intralesional curettage followed by bone grafting or cementation. Bone cement provides immediate structural support and allows early weight-bearing. In addition, recurrence can be easily detected due to the contrast between cement and surrounding bone on radiographs.

This case emphasizes the importance of integrating clinical evaluation, imaging findings, histopathological analysis, and multidisciplinary discussion in the management of cartilage tumors [8,9,10,11,13].

Conclusion

Cartilage tumors of long bones may present significant diagnostic challenges due to overlapping features between benign enchondromas and low-grade chondrosarcomas. When imaging findings and biopsy results are inconsistent, repeat biopsy and multidisciplinary evaluation are essential to establish an accurate diagnosis. Appropriate clinicoradiological correlation helps guide optimal treatment and prevents unnecessary aggressive interventions.

Clinical Message

Discordant imaging and biopsy findings in suspected cartilage tumors should prompt repeat biopsy and multidisciplinary evaluation to avoid misdiagnosis and inappropriate treatment.

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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