CLINICAL CASE - TEST YOURSELF | Musculoskeletal Imaging

A 55-Year-Old Female with Chronic Discomfort and Pain in the left Knee

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PART A

A 55-year-old female presented for a scheduled MRI examination presenting with persistent pain and swelling in her left knee. She had a known medical condition from early childhood, age 12. Her condition had progressively worsened, with the pain becoming more pronounced during physical activity and causing significant discomfort when walking. The patient also reported intermittent stiffness and a limited range of motion in the affected knee, which was worse in the morning. The patient denied any history of trauma, fever, or other systemic symptoms. There was no significant family history of bone diseases or similar conditions.

On physical examination, there was visible swelling around the left knee joint. The overlying skin appeared normal with no discoloration. The range of motion was reduced.

KEY WORDS | Ollier Disease, Enchondromatosis, Multiple Enchondromas, Knee, Orthopedics, Radiology, MRI

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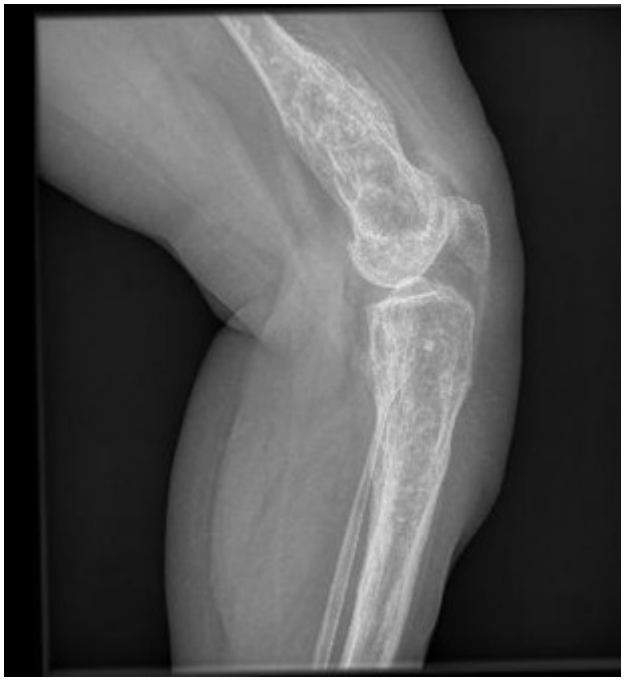

Figure 1A *Left Knee Lateral X-Ray*

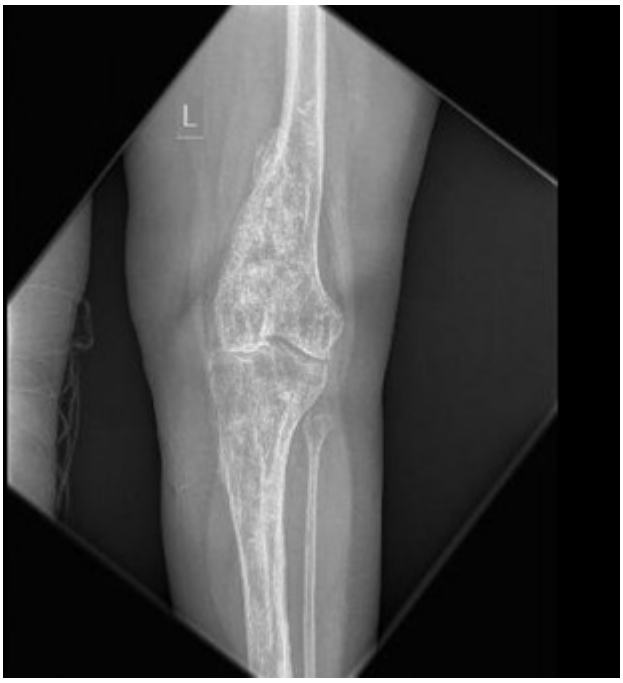

Figure 1B *Left Knee Frontal AP X-Ray*

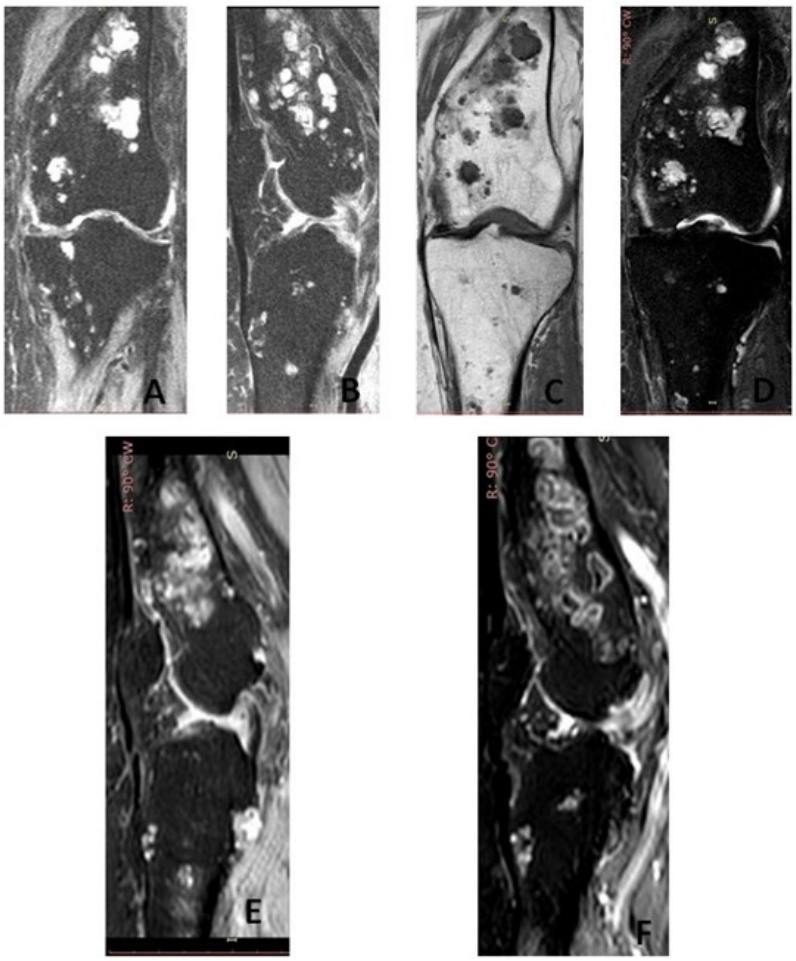

Figure 2A *PDW FAT SAT COR,* ***Figure 2B*** *PDW FAT SAT SAG,* ***Figure 2C*** *T1W - COR,* ***Figure 2D*** *STIR COR,* ***Figure 2E*** *T1W FAT SAT SAG,* ***Figure 2F*** *T1W – FAT SAT SAG + GD*

PART B

Diagnosis: The clinical presentation, combined with the radiographic findings (figure 1,2), led to a diagnosis of Ollier Disease - characterized by multiple enchondromas, affecting the left femoral bone and tibia.

Enchondromas are common benign, usually asymptomatic, cartilaginous tumors that develop in the metaphyses, often in close proximity to the growth plate, and can extend into the diaphyses of long tubular bones [1]. Enchondromatosis, also known as Ollier disease, is defined by the presence of multiple enchondromas, which consist of asymmetric cartilaginous lesions with remarkable variability in terms of size, location, number, progression, the age of onset, and surgical requirements [2,3].

Ollier disease is characterized by the development of hypertrophic cartilage, which, arising from the epiphyseal plate, extends into the metaphysis and, with continued skeletal growth, into the diaphysis. The long bones and the short tubular bones of the hands and feet are affected. This process leads to severe deformity of the affected bones, resulting in abnormal structure and premature fusion of the growth plates. Enchondromas also cause skeletal abnormalities, limb length discrepancies, and pain, and they have the potential to transform into chondrosarcomas (in 20-50% of cases)[4,5,6,7].

Unlike multiple hereditary osteochondromatosis, Ollier disease is not inherited. The coexistence of multiple enchondromatosis with hemangiomas of the soft tissues is characterized as Maffucci syndrome. In Maffucci syndrome, phleboliths are usually present, which aid in diagnosis and in distinguishing it from Ollier disease. The combination of enchondromatosis with multiple exostoses is characterized as metachondromatosis.

Incidence. The incidence of Ollier disease is estimated to be 1 in 50,000 to 100,000[1,2,3].

Frequency: The exact frequency of Ollier disease is unknown. In the United States, enchondromas constitute 12-14% of benign bone tumors and 3-10% of all bone tumors [1,2,3].

Gender: Enchondromas occur equally in males and females. Ollier disease is slightly more common in females, with a female-to-male ratio of 1.5:1. [1,2,3]

Race: Ollier disease does not show any racial predilection. It has been described in various ethnic groups. [1,2,3]

Age: Solitary enchondromas are usually observed in individuals aged 20-40 years. Ollier disease is most commonly found in individuals aged 0-10 years. The association of enchondromatosis with intracranial malignant conditions is similar in both children and adults, although Ollier disease appears to be more frequent in children [1,2,3].

Patients typically present with pain, swelling, and potential deformities in the affected limb, depending on the number and size of the enchondromas.

While the lesions are benign, there is a risk of malignant transformation into a chondrosarcoma, particularly in adulthood, necessitating careful monitoring.

Enchondromas are radiologically characterized by multiple radiolucent, homogeneous lesions with an ovoid or elongated shape and well-defined, slightly thickened bony margins [1,3]. These lesions typically align parallel to the axis of the long bones. When located at the bone margins, they produce a characteristic appearance similar to a bone cyst.

Metaphyseal Expansion: Enchondromas can cause expansion of the metaphyses.

Delayed Bone Growth: A delay in bone development may be observed (Loder RT et al., 2004).

Calcification of Lesions: Over time, the lesions usually calcify and exhibit diffuse stippling. The disappearance of calcifications is a sign of malignant transformation, with the underlying enchondroma being replaced by sarcomatous tissue.

Calcified enchondromas need to be differentiated from: **Bone infarcts, Chondrosarcoma**

If the lesion is purely lytic, differential diagnosis should include [4,5,6]:

- **Benign Lytic Lesions** (such as non-ossifying fibroma)
- **Simple Bone Cyst**
- **Fibrous Dysplasia**
- **Eosinophilic Granuloma**
- **Chondrosarcoma (particularly clear cell**

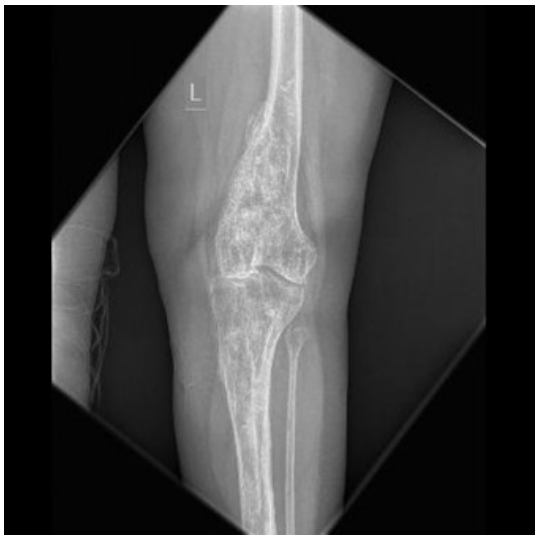

Figure 1A *Left Knee Lateral X-Ray*
Figure 1B *Left KneeFrontal AP X-Ray*
Figure 1A,B*: The X-ray revealed multiple radiolucent lesions with well-defined margins and scattered calcifications within the distal femur and proximal tibia (figure 1A, 1B). The lesions were suggestive of enchondromas, with mild cortical thinning and evidence of periosteal reaction on the posterior surface of the tibia.*

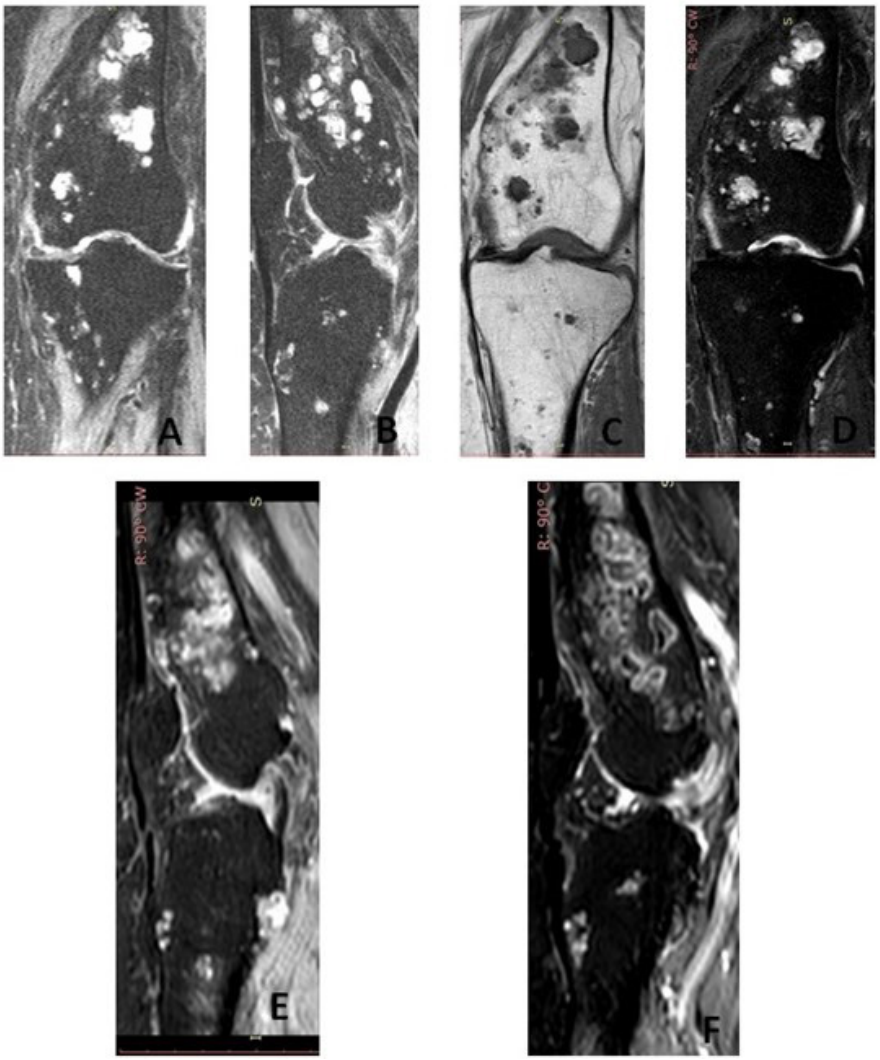

Figure 2A-F: *numerous lobulated focal lesions, some of which converge in specific areas and exhibit internal septations. The majority of these lesions are confined within the bone marrow. These lesions display low signal intensity on T1-weighted sequences (Figure 2C, 2E), high signal intensity on T2 – PD weighted sequences (Figure 2A, 2B, 2D), and demonstrate ring-like and septal enhancement following the intravenous administration of a paramagnetic contrast agent (Figure 2F). There were no signs of malignant transformation, such as soft tissue extension or cortical breach.*

chondrosarcoma), which tends to affect the ends of bones, especially the proximal femur.

- Enchondromas appear as well-circumscribed, radiolucent lesions in most cases with internal calcifications revealed on X-rays.
- CT scans evaluate cortical involvement and planning potential surgical interventions.
- MRI is crucial for assessing the extent of the lesions and distinguishing benign enchondromas from malignant transformations.

In MRI imaging, the T1-weighted sequence often shows an intermediate to low-signal intensity in cartilage-containing lesions. A distinctive feature of chondroid matrix structures, known as 'rings and arcs,' appears as internal foci of low signal intensity. This characteristic pattern helps identify the presence of cartilage within the lesion, as seen in enchondromas and other cartilage-based growths[3,4].

On T2-weighted imaging, cartilage typically appears predominantly hyperintense and well-defined due to its hydrophilic nature and high water content. Similar to T1, the 'rings and arcs' pattern is also present on T2, highlighting the unique appearance of a chondroid matrix. Importantly, there is an absence of bone marrow or soft tissue edema in these lesions, which further supports a diagnosis involving cartilaginous material without significant surrounding inflammatory response[3,4].

In post-contrast imaging with gadolinium (T1 C+ (Gd)), there is enhancement along the scalloped margins of the lesion, along with curvilinear translesional septa, which accentuates the 'rings and arcs' pattern. This enhancement pattern can occasionally mimic that seen in chondrosarcomas, although the latter are generally more aggressive and may present with additional features such as surrounding soft tissue invasion or bone marrow involvement [3,4].

This case underscores the importance of early and accurate diagnosis of Ollier Disease to manage symptoms effectively and monitor for potential complications, including malignancy. Regular follow-up and a multidisciplinary approach involving radiology, orthopedics and oncology are crucial for optimizing patient outcomes. **R**

Conflict of Interest:
The authors declared no conflicts of interest.
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