A rare case of Enchondromatosis of the knees and hands with involvement of Hoffa's fat pad and peri-articular soft-tissues

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ABSTRACT

We report a case of a 56-year old man with chronic pain in both knees for several years. This patient had already undergone surgery on his left knee in 2002 after an x-ray showed multiple lytic and well margined lesions in the distal femur and proximal tibia with ground-glass matrix, involving Hoffa's fat pad and the patellar ligament. Histology was consistent with an enchondroma. The most recent MRI examination showed enchondromatosis involving both knees with bilateral extension into Hoffa's fat pad and the patellar ligament. Subsequently, we performed an additional radiographic examination of the hands and feet, as well as an MRI of both hands to identify other possible enchondromas in the most common sites for this disease. Enchondromatosis with soft tissue involvement is extremely rare, and involvement of Hoffa's fat pad has not been reported in the scientific literature. The clinical presentation of this case and the general aspects of Enchondromatosis are discussed.

CASE REPORT

CASE REPORT

We present a case of a 56 year-old man who came to our institution for an MRI of both knees, because he had experienced several years of knee pain associated with functional limitation and soft tissue swelling in the left knee and both hands [Fig. 1-3].

The patient conveyed that he already knew he had an enchondroma in his left knee, because in 2002 he had an x-ray of the left knee. This radiograph showed multiple lytic, well margined lesions of the distal femur and proximal tibia, with a ground-glass matrix involving Hoffa's fat pad and the patellar ligament. After this x-ray he had surgery to remove the lesion. Histology was consistent with an enchondroma, and in 2003 and 2006 he underwent two subsequent arthroscopies for joint curettage and a partial medial meniscectomy. At our institution the patient underwent MRI examination of both knees which revealed the presence of multiple bone lesions in the proximal metaphysis of the tibia and distal metaphysis of the femur. Subsequently, we performed radiographic examinations of both knees, hands and feet, as well as an MRI of both hands.

DISCUSSION

Enchondroma is a rare, benign tumor of chondroid nature, that usually develops in the metaphyses and diaphyses of the long bones and the cartilage of the joints of the arms and legs. When multiple enchondromas are present, the condition is called enchondromatosis, also known as Ollier disease (WHO terminology) [1,2,3].

Ollier disease, first described in 1889, has an estimated prevalence of 1/100,000 [4].

Enchondromas in Ollier disease can be extremely variable in terms of size, number, location, evolution, and age at diagnosis [5]. The distribution of these may be unilateral or bilateral, and when bilateral, they are always asymmetrical [6].

Spranger et al. attempted to classify Enchondromatosis into 6 types, of which Ollier disease (type I) and Maffucci syndrome (type II) are the most common, while the other 4 subtypes (metachondromatosis, spondyloenchondrodysplasia, enchondromatosis with irregular vertebral lesions, and generalized enchondromatosis), are extremely rare [5].

For a long time, enchondroma has been considered a developmental disorder caused bythe failure of normal endochondral bone formation. With the identification of genetic abnormalities in enchondroma, however, it is currently thought of as a neoplasm. The gene(s) causing the different enchondromatosis syndromes are largely unknown. Active hedgehog signaling is reported to be important for enchondroma development and PTH1R mutations have been identified in ~10% of those with Ollier disease [7].

Usually, enchondroma manifests itself early in childhood without any significant gender bias, as a single lesion (solitary enchondroma) which is most often found incidentally when radiographic studies are performed for other reasons, or with the appearance of palpable bony masses on a finger or a toe [2].

Enchondromas frequently affect the long tubular bones, particularly the tibia and the femur; flat bones, especially the pelvis, can also be affected [2]. The skull and vertebral bodies are very rarely involved, and soft tissue involvement has never been reported in scientific literature [1,7,8].

The complications caused by enchondromas include: skeletal deformities, limb-length discrepancy and the potential risk for malignant change to chondrosarcoma (estimated around 25%), although controversy exists in the literature about the development of secondary chondrosarcoma from enchondroma [9,10,11].

The diagnosis of Ollier disease is based on clinical features and diagnostic imaging, while histological analysis has a limited role, and it is mainly used only if malignancy is histological analysis has a limited role, and it is mainly used only if malignancy is suspected [3].

Radiography typically shows multiple, radiolucent, homogenous lesions with an oval or elongated shape, endosteal erosion and ground glass appearance of the matrix; dystrophic calcification within the matrix of small cartilage nodules or fragments of lamellar bone are often described [2,10].

Computed tomography (CT) is superior to radiography in detecting matrix mineralization as well as in the evaluation of the calcification pattern, the margins and the degree and extent of endosteal scalloping. Moreover, CT is particularly useful in the characterization of lesions that occur in the pelvis, or other areas with complex anatomy that can be difficult to evaluate on radiographs [8].

On magnetic resonance (MR) imaging, the non-mineralised component of enchondromas appear as low to intermediate signal intensity lesions on T1-weighted sequences, and intermediate to high signal intensity lesions on T2-weighted sequences. Low signal intensity septa on T2-weighted MR images may also be evident, corresponding pathologically to enchondral ossification or fibrous septations [8].

Most of the typical radiographic features of Enchondromatosis can be appreciated on the radiographs acquired on our patient. Looking closely at the radiographs of both knees, [Fig.4-5] one can appreciate soft tissue swelling of Hoffa's fat pads associated with chondroid matrix lesions of the tibial tuberosities bilaterally, and another similar lesion in the distal metaphysis of the left femur; these lesions are related to Ollier disease.

The case presented here has all of the typical features of the Ollier disease, but also peculiar involvement of the soft tissues, that are, in our specific case, Hoffa's fat pad and the patellar ligament.

In the Pubmed database (keywords: Ollier, Enchondromatosis, Hoffa) there is no evidence of a similar situation (involvement of Hoffa's fat pad and the patellar ligament in Ollier disease), so we will discuss the general aspects of Ollier disease with particular attention to the soft tissue involvement, as in our case.

Clinical observation of the right hand [Fig.6] demonstrated palpable swelling of the proximal interphalangeal joint of the 3rd finger, which when combined with the radiographic features [Fig.7], is compatible with enchondromas.

MR imaging of both knees showed the non-mineralised component of the enchondromas as low to intermediate signal intensity on T1-weighted sequences, intermediate to high signal intensity on T2-weighted sequences and high signal with Short Tau Inversion Recovery (STIR) sequences [Fig.8-10]. Similar findings can be appreciated in the distal metaphysis of the femur, in the posterior region of both tibial plates (not visible on radiographs) and in the anterior tibial tuberosity. The enchondroma in this last site expands into Hoffa's fat pad and into the patellar ligament which appears thickened and enveloped as "a sleeve," because of a coexisting chronic reactive tendinopathy [Fig.11-12].

MR imaging of the right hand shows an ovoid lesion (about 20mm in diameter) in the distal diaphysis and middle third of the proximal phalanx of the third finger, with sharp margins, low signal intensity on T1, intermediate signal intensity on T2 and high signal intensity on STIR, compatible with an enchondroma. This lesion is associated with soft tissue intra-articular involvement [Fig.13]. On the lateral (radial) side of the II metacarpophalangeal joint of the left hand there is swelling of the peri-articular soft tissues, with a maximum

diameter of 20 mm, with low signal intensity on T1, low-intermediate signal on T2 and intermediate-high signal in STIR, indicating a proliferative synovitis, which is associated with reactive edema and erosions of the second metacarpal head [Fig.14-15].

Possible differential diagnoses for this imaging appearance include bone infarct, chondrosarcoma, epidermoid inclusion cyst, unicameral bone cyst, giant cell tumor and fibrous dysplasia [1].

Bone infarct appears on radiographs as a central lesion in metaphysis or diaphysis with well defined serpiginous borders and on MRI it has low signal intensity in both T1-w and T2-w sequences.

Chondrosarcoma appears on radiographs as a mixed lytic-sclerotic expansile lesion with irregular borders, and the sclerotic component represented by typical chondroid ring-and-arc calcifications; on MRI, it presents with low signal intensity in T1-w and high signal in T2-w sequences.

An epidermoid inclusion cyst appears on radiographs as an expansile radiolucent bone lesion with sclerotic remodeling, and on MRI it has low-intermediate signal intensity in T1-w and high signal in T2-w sequences.

A unicameral bone cyst appears on radiographs as a solitary, lytic, usually metaphyseal lesion with well defined sclerotic margins, and on MRI it presents with low signal in T1-w and high signal in T2-w sequences.

A giant-cell tumor often appears on radiographs as a lytic lesion, with sharp, well defined margins and extensive subchondral extension. On MRI it has low signal intensity in T1-w and heterogeneous high signal in T2-w sequences.

Fibrous dysplasia appears on radiographs often as a purely lytic lesion, but may have a ground-glass appearing matrix, and calcifications can be present to a variable degree. On MRI, in T1-w sequences it has intermediate signal intensity with more low signal area in the distal portion, and in T2-w sequences it is tipically high signal, with low, intermediate or markedly high signal areas within.

There is no medical treatment for enchondromatosis. Surgery is indicated only in the case of complications, such as pathological fractures, growth disturbance or malignant transformation [6]. For this reason, follow-up has a very important role in this disease.

We strongly recommend multidisciplinary assessment of difficult cases in order to avoid mistakes in decision making which could lead to detrimental treatment. Biopsy should be performed only if a lesion is larger than 7 cm in diameter and/or presents significant changes in follow-up imaging [13].

The prognosis for Ollier disease is difficult to assess [7]. As generally is the case, forms with an early onset appear more severe [4].

TEACHING POINT

Enchondromas are cartilaginous tumors, usually benign, usually intraosseous, which develop near the growth plate, causing bone deformities, especially when they develop during growth. These lesions may expand, with variability, into adjacent soft tissues(tendons, ligaments, fat pads), altering the biomechanics of joints leading to onset of clinical manifestations related to the functions performed by the anatomical structures involved.

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FIGURES

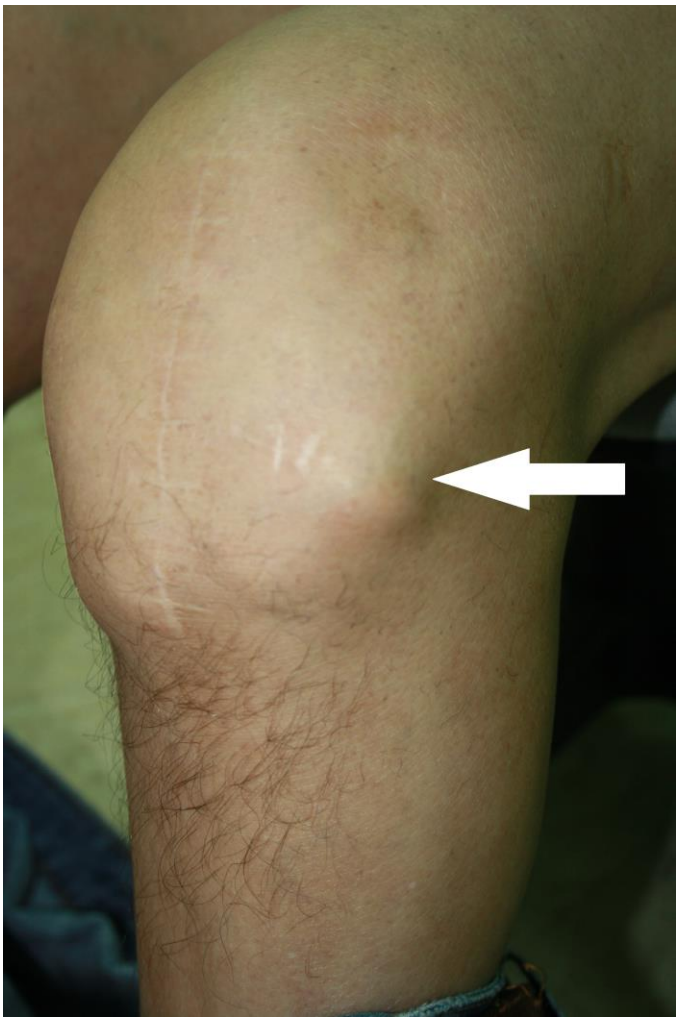

Figure 1. 56 year-old man with Enchondromatosis. Picture of the left knee which shows a scar from previous surgery and a mass laterally (arrow)

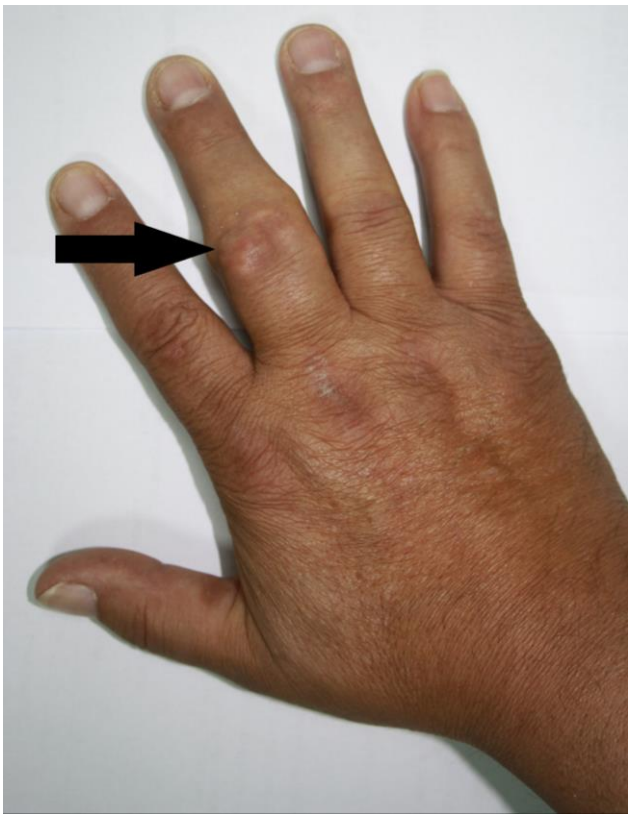

Figure 2. 56 year-old man with Enchondromatosis. Picture of the right hand which shows swelling of the proximal inter-phalangeal joint of third finger (arrow).

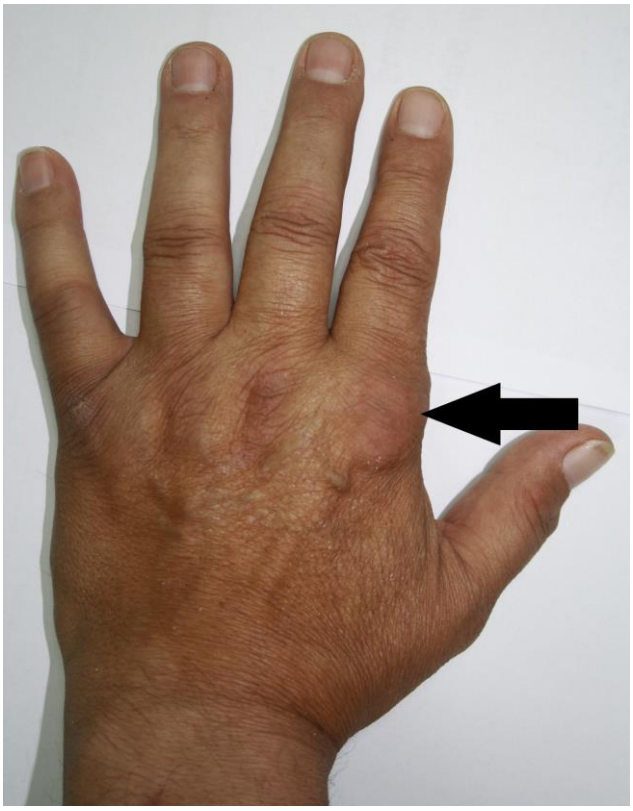

Figure 3. 56 year-old man with Enchondromatosis. Picture of the left hand which shows swelling medially to the metacarpo-phalangeal joint of the second finger (arrow).

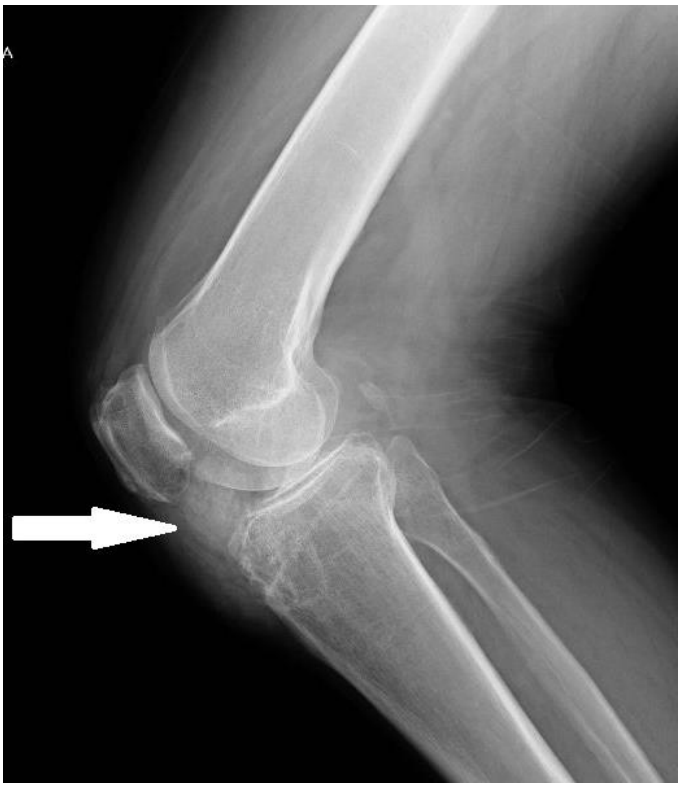

Figure 4. 56 year-old man with Enchondromatosis. CR image of the right knee (lateral view) which shows a radiopacity extending into Hoffa's fat pad (arrow).

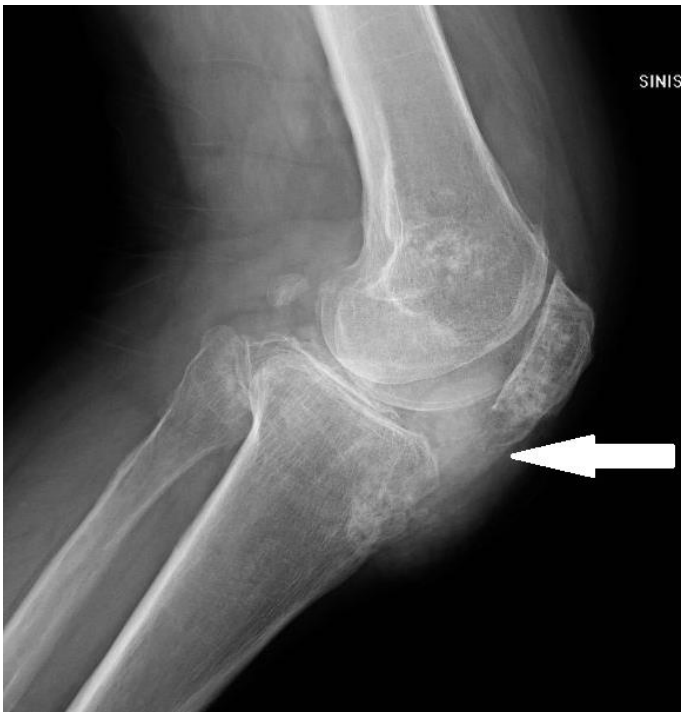

Figure 5. 56 year-old man with Enchondromatosis. CR image of the left knee (lateral view) which shows a radiopacity extending into Hoffa's fat pad (arrow).

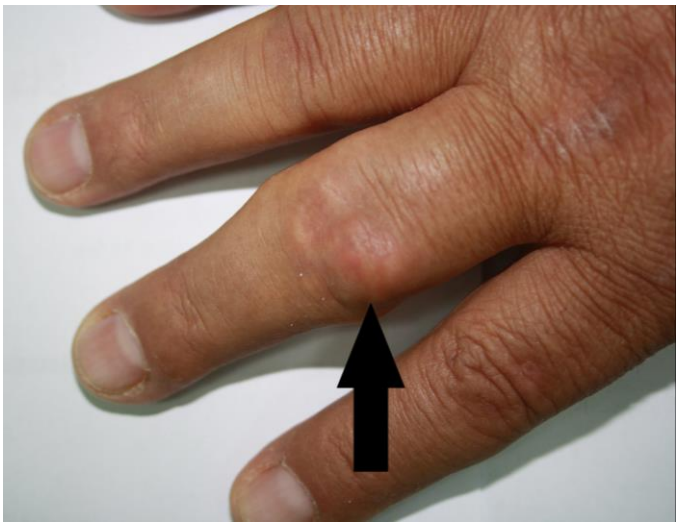

Figure 6. 56 year-old man with Enchondromatosis. Magnified picture of the third finger of the right hand which shows swelling of the proximal inter-phalangeal joint.

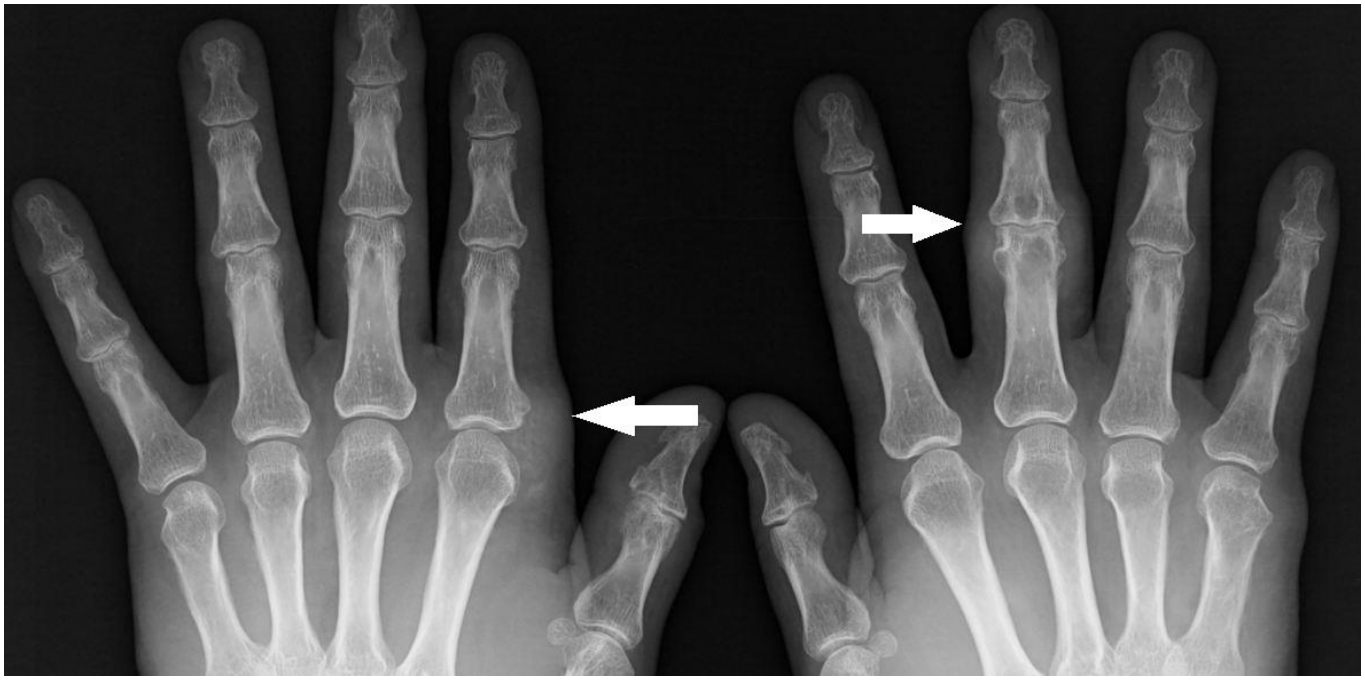

Figure 7. 56 year-old man with Enchondromatosis. CR image of both hands (AP view) which shows swelling of the metacarpo-phalangeal joint of left second finger and of the proximal inter-phalangeal joint of right third finger (arrows).

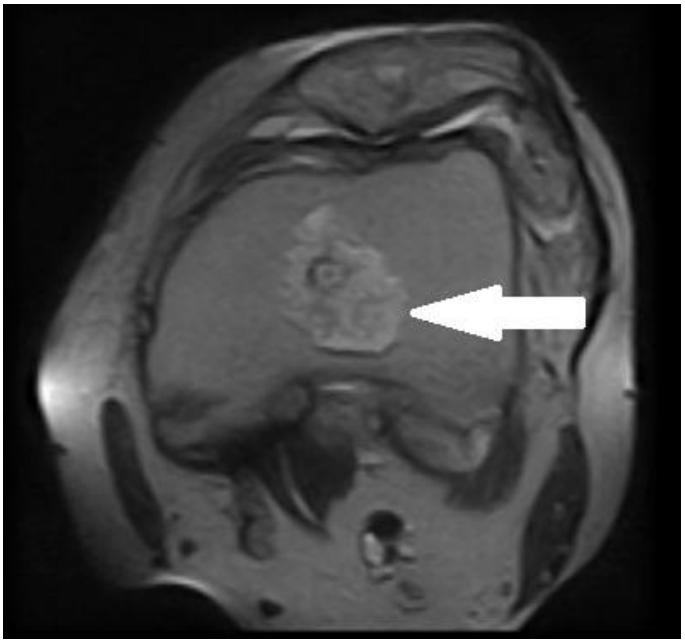

Figure 8. 56 year-old man with Enchondromatosis. Image acquired on a G-Scan 0,25 Tesla MR scanner. T2-weighted TSE (TR: 3800; TE: 28; TI: 0; FA: 90) axial image of left knee through the patello-femoral joint which shows an enchondroma centered in the distal femur (arrow).

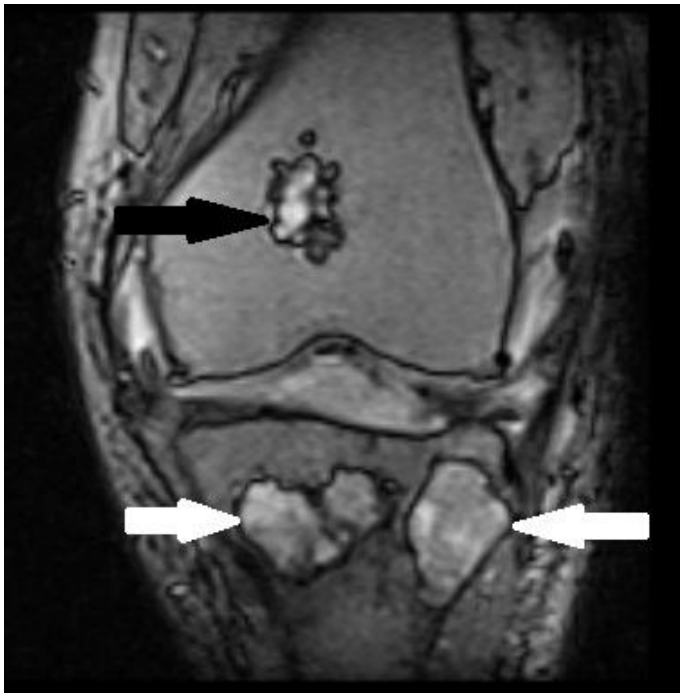

Figure 9. 56 year-old man with Enchondromatosis. Image acquired on a G-Scan 0,25 Tesla MR scanner. GE-T1-weighted (TR: 1200 TE: 22 TI: 0 FA: 35) coronal image of left knee which shows multiple enchondromas in the distal femur (black arrow) and proximal tibia (white arrows).

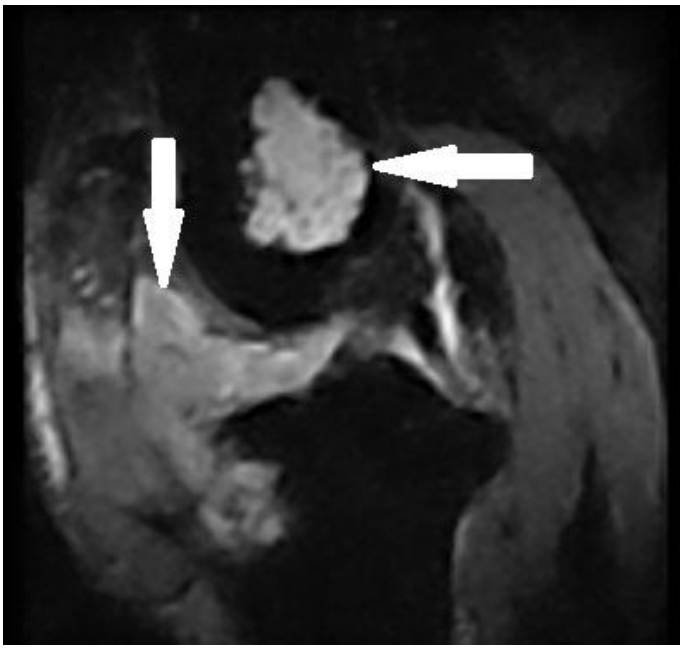

Figure 10. 56 year-old man with Enchondromatosis. Image acquired on a G-Scan 0,25 Tesla MR scanner. Sagittal STIR (TR: 2380 TE: 25 TI: 80 FA: 90) image of the left knee which shows a large enchondroma in the distal femur (horizontal arrow), and another in the anterior tibial tuberosity with involvement of Hoffa's fat pad and the patellar tendon (vertical arrow).

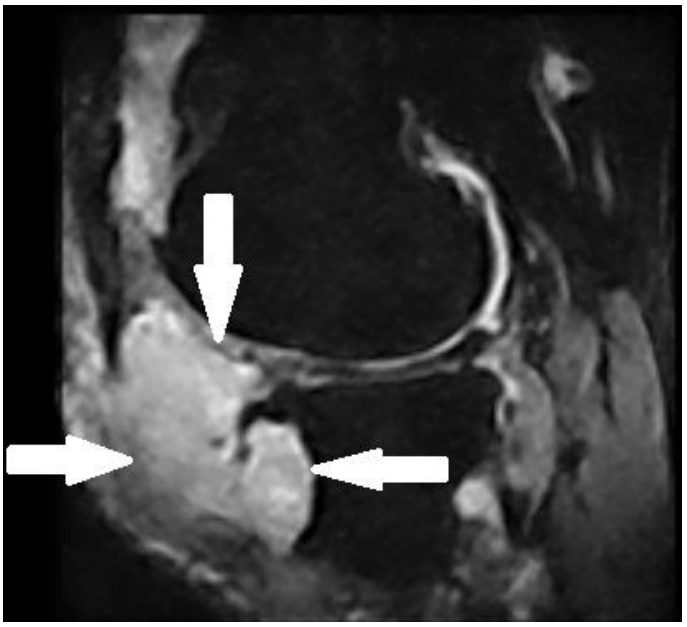

Figure 11. 56 year-old man with Enchondromatosis. Image acquired on a G-Scan 0,25 Tesla MR scanner. Sagittal STIR (TR: 2180 TE: 25 TI: 80 FA: 90) image of the right knee which shows an enchondroma in the anterior tibial tuberosity with extensive involvement of Hoffa's fat pad and the patellar tendon (arrows).

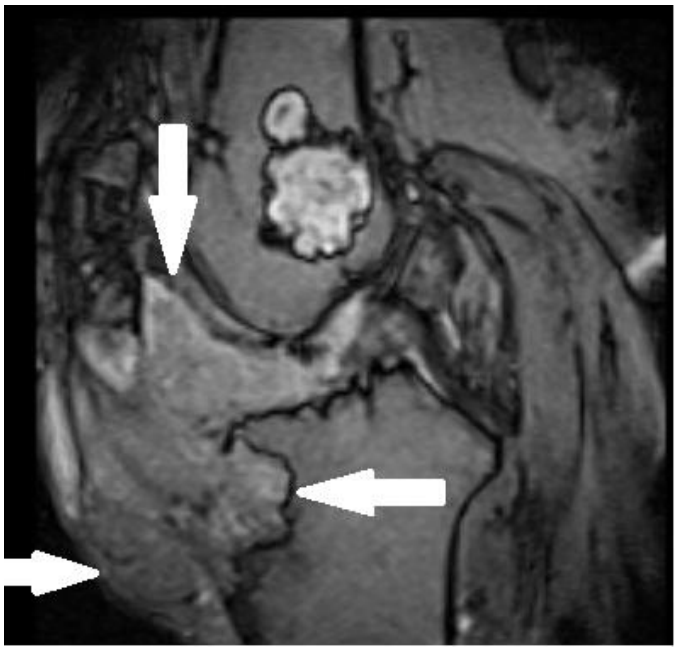

Figure 12. 56 year-old man with Enchondromatosis Image acquired on a G-Scan 0,25 Tesla MR scanner. GE-T1-weighted (TR: 1200 TE: 22 TI: 0 FA: 35) sagittal image of the left knee which shows extensive involvement of Hoffa's fat pad and the patellar tendon by an enchondroma localized in the anterior tibial tuberosity (arrow).

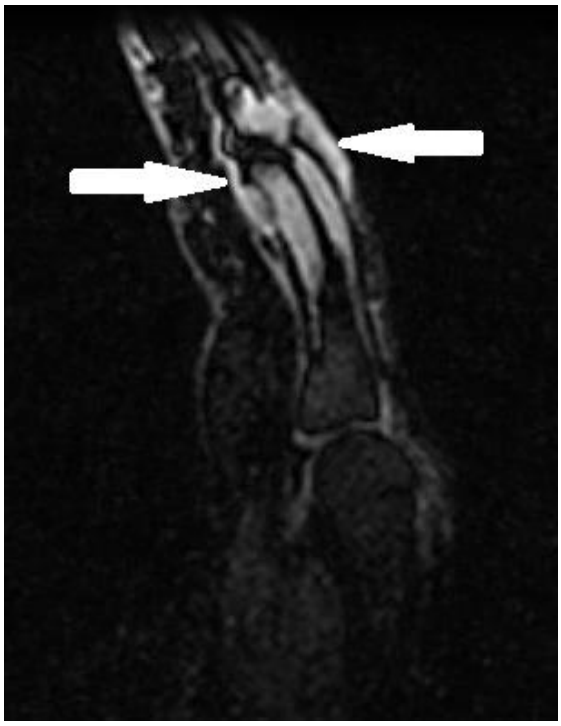

Figure 13. 56 year-old man with Enchondromatosis. Image acquired on a G-Scan 0,25 Tesla MR scanner. Sagittal STIR (TR: 2180 TE: 25 TI: 80 FA: 90) image of the right hand which shows, in the distal and middle third of the diaphysis of the proximal phalanx of third finger, the presence of an ovoid lesion with sharp margins, compatible with a chondroma; this lesion is associated with intra-articular soft-tissue involvement (arrows).

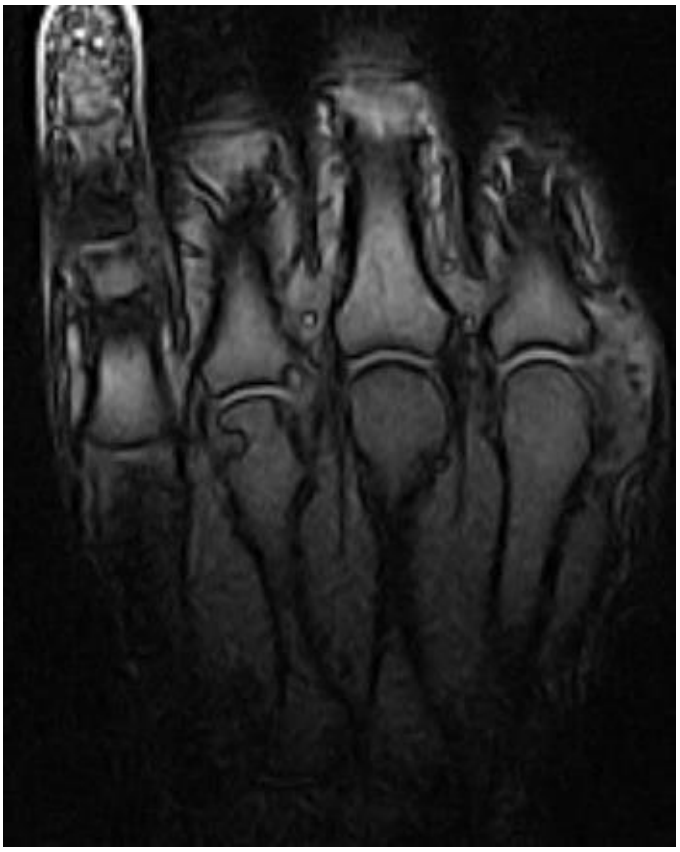

Figure 14. 56 year-old man with Enchondromatosis. Image acquired on a G-Scan 0,25 Tesla MR scanner. Coronal X-Bone (TR: 1300 TE: 20 TI: 0 FA: 35) image of the left hand which shows swelling of the peri-articular soft tissues on the lateral (radial) side of II metacarpophalangeal joint, consistent with a proliferative synovitis (arrow).

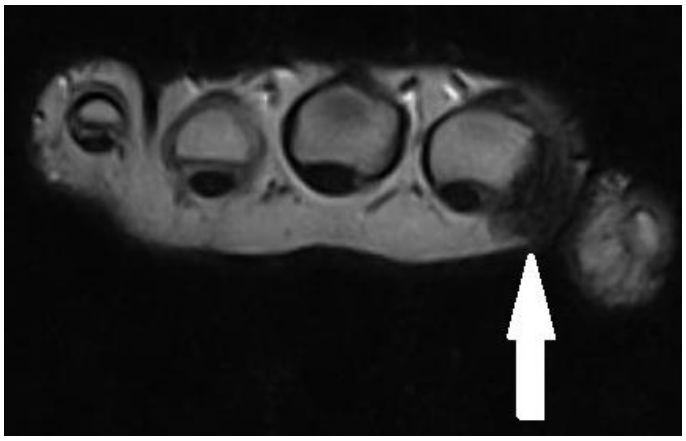

Figure 15. 56 year-old man with Enchondromatosis. Image acquired on a G-Scan 0,25 Tesla MR scanner. T2-weighted TSE (TR: 3500; TE: 28; TI: 0; FA: 90) axial image of the left hand which shows swelling of the peri-articular soft tissues on the lateral (radial) side of the II metacarpophalangeal joint, consistent with a proliferative synovitis (arrow).

Etiology	Probable correlation with genetic abnormalities.
Incidence	Not reported.
Prevalence	1/100,000
Gender Ratio	Not significant gender bias.
Age predilection	Childhood.
Risk factors	PTH1R gene mutation have been identified in ~10% of cases.
Prognosis	Prognosis in Ollier disease is difficult to assess. Generally, forms with an early onset appear more severe.
Treatment	There is no medical treatment for enchondromatosis. Surgery is indicated only in case of complications.
Findings on imaging	• X-Ray: multiple, radiolucent homogenous lesions with an ovoid or elongated shape, endosteal erosion, ground glass appearance and dystrophic calcifications. • MRI: o T1W: low-intermediate signal o T2W: intermediate-high signal o STIR: high signal

Table 1: Summary table for enchondromatosis

	X-Ray	MRI
Enchondromatosis	Radiolucent, homogenous lesions with an ovoid or elongated shape, endosteal erosion and ground glass appearance of the matrix.	T1-w: low-intermediate signal. T2-w: intermediate-high signal. STIR: high signal.
Bone Infarct	Central lesion in metaphysis or diaphysis with well defined serpiginous borders.	Irregular serpiginous margins. T1-w: low signal. T2-w: low signal.
Chondrosarcoma	Mixed lytic-sclerotic expansile lesion with irregular borders. The sclerotic component is represented by typical chondroid ring-and-arc	T1-w: low-intermediate signal. T2-w: very high signal in non mineralized-calcified portions.
Epidermoid Inclusion Cyst	Expansile radiolucent bone lesion with sclerotic remodeling.	T1-w: low-intermediate signal. T2-w: high signal.
Unicameral Inclusion Cyst	Solitary, lytic, usually metaphyseal lesion with well defined sclerotic margins.	T1-w: low signal. T2-w: low signal.
Giant-cell Tumor	Lytic lesion, with sharp, well defined margins and extensive subchondral lesion.	T1-w: low signal. T2-w: heterogeneous high signal.
Fibrous dysplasia	It is often a purely lytic lesion, but may have a ground-glass appearance matrix. Calcifications can be present to a variable degree.	T1-w: intermediate signal with a more low signal area in the distal portion. T2-w: typically high signal, with low, intermediate or markedly high signal areas within.

Table 2: Differential diagnosis table of fistulation in soft-tissue sarcoma

ABBREVIATIONS

CT = computed tomography
MRI = magnetic resonance imaging
PTH1R = parathyroid hormone 1 receptor
STIR = Short Tau Inversion Recovery
WHO = World health association

KEYWORDS

enchondromatosis; Ollier disease; benign hyaline tumour; chondroma; enchondroma; Hoffa's fat pad

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