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Case Report

Ollier disease: A case report and literature review ☆,☆☆

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ABSTRACT

Ollier disease is an uncommon disease characterized by several enchondromas and an asymmetric distribution of cartilage lesions, which can vary significantly in size, location, age, and gender. The primary symptom of this condition is a nonossifying chondrocyte mass or hamartomatous chondrocyte growth in the metaphysis of a short or long bone. Specific cases can progress to chondrosarcoma or osteosarcoma. X-ray is the most fundamental diagnostic technique for skeletal illnesses. In this article, we present a case of Ollier disease from Mother and Child Hospital IBN SINA, Rabat, Morocco.

Introduction

Ollier disease is a rare developmental condition characterized by the presence of several asymmetrically distributed enchondromas.

Enchondromas are benign intraosseous cartilaginous lesions that originate from the growth plate cartilage [1].

Diagnosis of Ollier disease is made based on clinical symptoms and imaging findings on conventional radiology examination. Herein we present the case of an 8-year-old patient with Ollier disease followed by a brief review of the literature.

Case report

We present the case of an 8-year-old patient with no past medical history who presents with a painful tumefaction of the right lower limb and tenderness of the right hand. On physical examination no inflammatory changes were found. Plain radiographs of the right hand and right lower limb were performed.

Plain radiograph of the right thigh revealed a well-defined, heterogeneous osteolytic lesion of the distal femur surrounded by a peripheral sclerotic rim. This lesion appeared to be expansile with some cortical discontinuity (Fig. 1).

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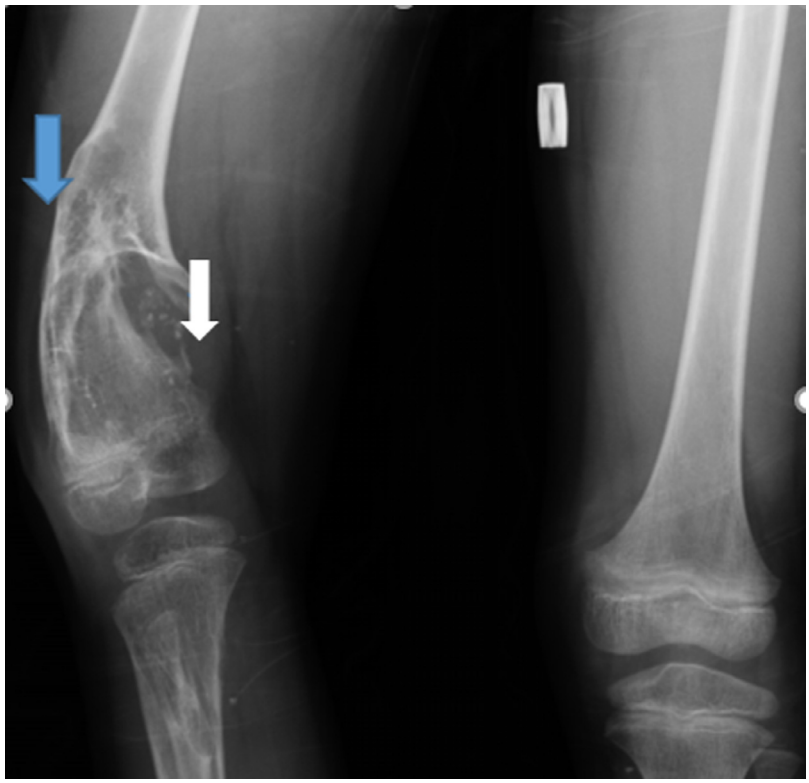

Fig. 1 – Plain radiograph of the right thigh showing a heterogeneous osteolytic lesion surrounded by a sclerotic rim (blue arrow) this expansile lesion is responsible for cortical rupture on the medial aspect of the bone (white arrow).

The hand and wrist X-ray showed a diffuse bone demineralization of the radial and ulnar diaphysis, along with radio lucent lesions located in the phalanges (Fig. 2).

The diagnosis of Ollier disease was made based on clinical presentation, imaging findings, and lesion distribution. Given the cortical involvement, a computed tomography (CT) scan of the right thigh was then recommended, to better analyze the imaging features of this lesion and look for any sign of malignant degeneration.

The CT scan revealed an intramedullary, expansile osteolytic lesion of the distal femur metaphysis, presenting well-defined margins with irregular contours. This lesion was responsible for bone enlargement with thinning of the peripheral cortex which was ruptured on the medial aspect of the bone. No periosteal reaction or soft tissue involvement was found otherwise (Fig. 3). We also discovered other ipsilateral osteolytic lesions of the proximal femoral shaft, iliac bone, spine, and tibial shaft (Fig. 4). We therefore established the presence of many ipsilateral incidental osteolytic lesions with no signs of aggressivity given the absence of bone destruction, periosteal reaction, and soft tissue invasion.

Discussion

Enchondromas are common intraosseous cartilaginous tumors that arise around the growth plate cartilage and are usually benign.

Enchondromatosis occurs when numerous enchondromas coexist, and Ollier disease is the most frequent nonhereditary form of enchondromatosis [1].

In 1889, Ollier first described this condition characterized by multiple, unilaterally distributed endogenous chondromas associated with limb deformities [2]. Maffucci syndrome is a separate entity that has to be differentiated from Ollier disease, in fact it involves the presence of numerous unilateral enchondromas coexisting with cutaneous hemangiomas [3].

The pathogenesis of this disorder is not well established, but many theories regarding somatic mosaic mutations in isocitrate dehydrogenase (IDH)I and IDH2(2) have been associated with both Ollier disease and Maffucci syndrome [4]. Ollier illness is characterized by genomic copy number variations and mutations that influence a variety of important functions [5] several articles have recently proposed heterozygous mutations in the PTHR1, IDH1 (most common), and/or IDH2 genes as genetic aberrations [6].

One of the most accessible and easy to perform diagnostic imaging examination to analyze bone lesions is conventional radiology. Ollier disease lesions typically arise in the shaft and metaphysis of short or long bones, and they are ovale in shape [6]. Although it classically presents as an expansile radiolucent lesion, it typically demonstrates no scalloping or cortical thinning [7].

This condition is well delineated on imaging, demonstrating unilateral bone involvement with numerous intramedullary lytic lesions of long bones located in the hand,

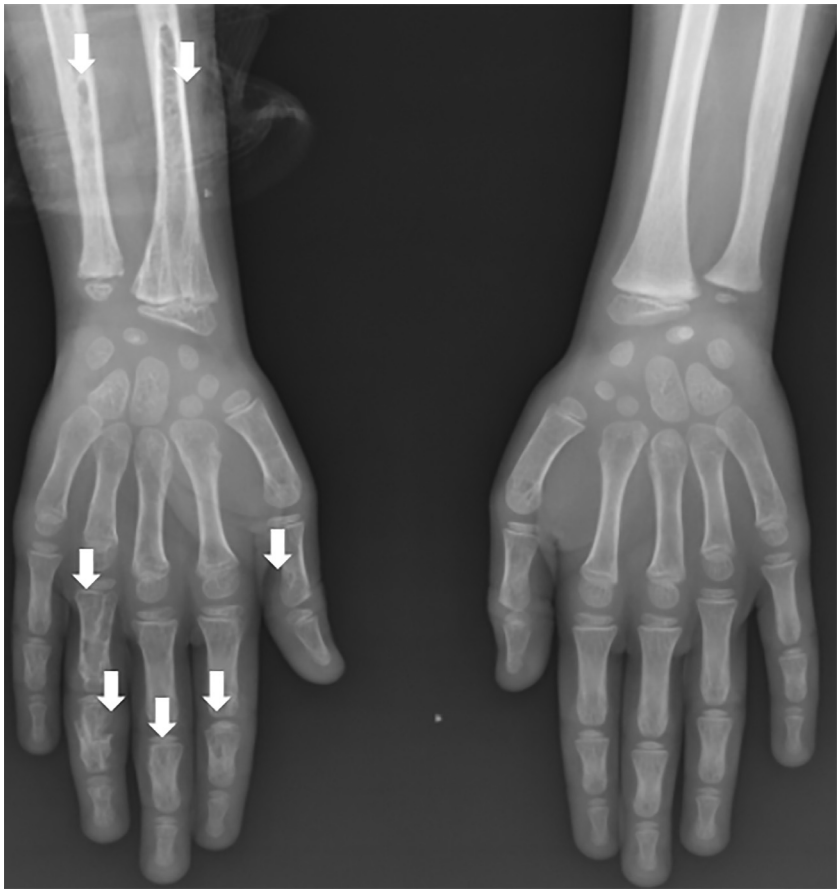

Fig. 2 – X-ray of the hands reveals bone demineralization of the right upper limb's radial and ulnar diaphysis, as well as the presence of osteolytic lesions of the phalanges (white arrow).

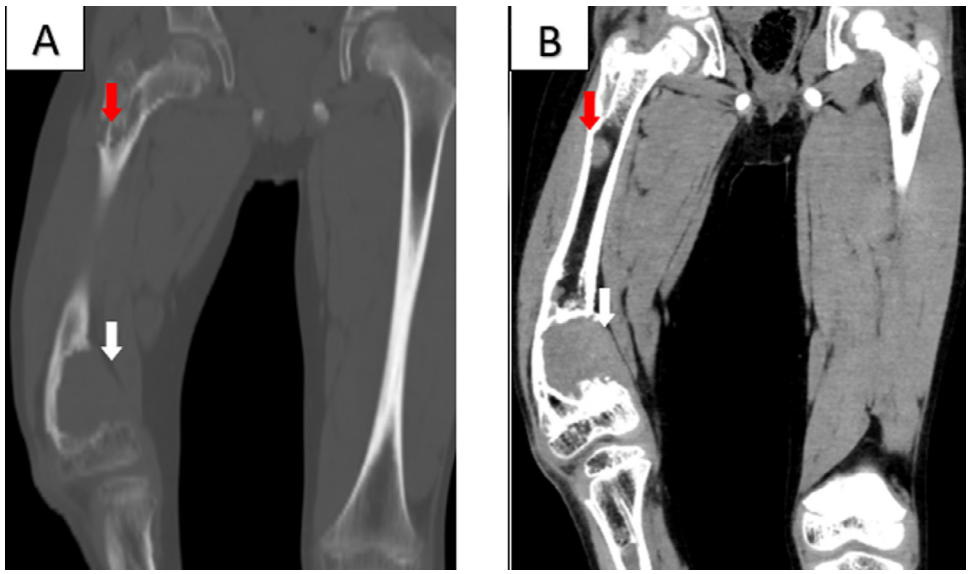

Fig. 3 – Axial/coronal/sagittal CT images demonstrating an osteolytic lesion of the distal metaphysis of the femur that is well-defined with irregular contours and is responsible for a medullary enlargement with a blown aspect of the cortex that is ruptured on the medial aspect (white arrow). No periosteal reaction or soft tissue invasion was found otherwise. Incidental ipsilateral osteolytic lesions are depicted in the upper femoral shaft (red arrow).

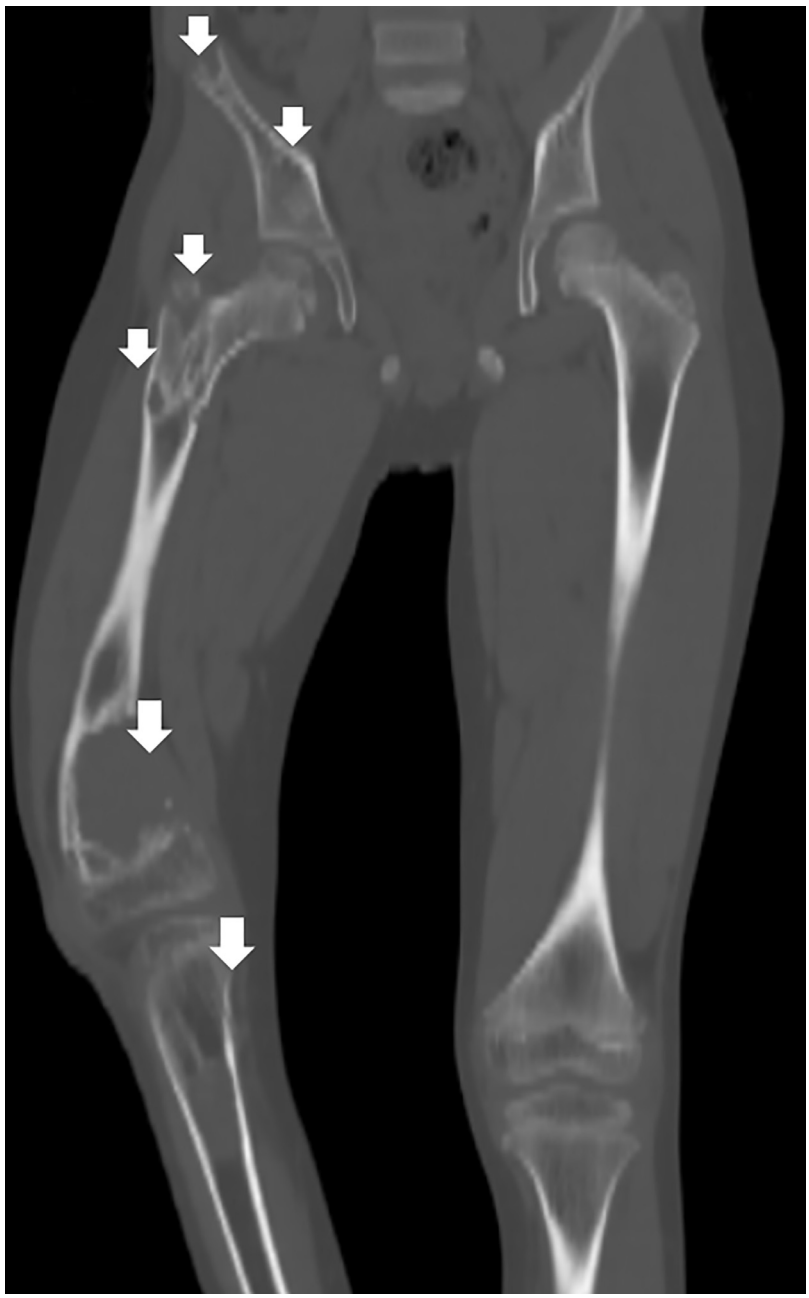

Fig. 4 – Axial/coronal/sagittal CT images showing other ipsilateral osteolytic lesions (white arrow) in the upper femoral shaft, iliac bone, metaphysis, and tibial shaft.

foot, particularly in the metaphyseal regions. Even when there is symmetrical involvement, there is always some unilateral predominance [1].

Computed tomography (CT) is more sensitive than conventional radiology to assess the type of matrix mineralization, calcification pattern, lobulated lesion edges, and the degree and extent of endosteal scalloping [5]. CT is also effective in determining the size of the lesion and existence of any soft tissue component that would support a chondrosarcoma diagnosis [5].

Magnetic resonance imaging (MRI) may be requested in the setting of a pathological fracture or when lesion characterization is required before treatment is initiated [8]. Enchondromas can demonstrate a high uptake on fluorodeoxyglucose-positron emission tomography (FDG-PET) which can be challenging in patients with a known malignancy as it can sometimes mimic a metastatic lesion [5].

The lesions tend to enlarge as the child grows, leading to anomalies such as genu valgum and genu varum, the latter being the most common [1], asymmetrical limb shorten-

ing, and other symptoms. Lesions in short bones can take the shape of scallop-like impressions due to the space limitations; the cortical bone also grows thinner and pathological fractures are more common [6]. The most severe kind of enchondroma development is malignant transformation to secondary chondrosarcomas. According to research, the incidence of secondary cancer in the limb bones ranges from 5% to 50% [4].

Ollier disease and several inherited exostoses must be distinguished. Lesion distribution and location are the most important criterion to differentiate these 2 entities: osteochondromas are eccentric found on the bone surface, whereas enchondromas are found in the middle of bones with intramedullary development [5]. Moreover, based on the radiological appearance, Ollier illness may mimic osteitis fibrosa cystica [9].

There is no specific pharmaceutical treatment for Ollier disease. A long-term surveillance and follow-up can be conducted for patients with Ollier disease who do not have major malformations or functional impairment [6]. Physiotherapy such as ultrasound, cryotherapy, CO_2 laser with stretching, active, mobilization, occupational therapy, and coordination exercises appear to significantly improve the functional abilities [5]. Surgery, on the other hand, is indicated in case of deformity, limb-length disparity, pathological fracture, and malignant transformation [5].

Conclusion

Ollier disease is a rare condition characterized by the development of multiple enchondromas with an asymmetric distribution that vary widely in size, location, age, and gender. The pathogenesis has yet to be fully confirmed. Patient monitoring is necessary since other systemic illnesses and potentially malignant changes may accompany the disease's progression.

Patient consent

Written informed consent for publication was obtained from the father of the child.

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