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Ollier Disease: A Case Report and Literature Review

O. Saidi[1*], K. Mouaadine[1], B. Chkirate[1]

[1]Pediatrics Department IV, Child Hospital, CHU IBN SINA, Rabat, Morocco

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***Corresponding author:** Saidi Oumayma
Pediatrics Department IV, Child Hospital, CHU IBN SINA, Rabat, Morocco

Abstract **Case Report**

Ollier's disease is a non-hereditary disease whose manifestations most often begin in the first decade of life. It is characterized by the growth of benign cartilaginous tumors called enchondromas and is associated with bone deformities and shortenings. We report the case of a 3-year-old patient with Ollier disease, suspected on clinical findings and confirmed by standard radiographs and biopsy. Diagnosis of Ollier disease is based on specific clinical manifestations and radiographic findings. Histological analysis has a limited role and is mainly used in cases of suspected sarcomatous transformation. Treatment of Ollier disease is generally conservative, except in the case of complications. No drug treatment is currently available. Surgery is performed in cases of deformity, pathological fracture, deviation, limb inequality and malignant sarcomatous transformation. The prognosis and course of the disease are influenced by orthopedic complications and the high risk of sarcomatous degeneration.

Keywords: Ollier disease, enchondromatosis, osteolytic lesion.

INTRODUCTION

Ollier disease is a non-hereditary disorder characterized by the growth of benign cartilage tumors, known as enchondromas, within the bony metaphyses, close to the conjugation cartilages. Cartilage lesions are scattered throughout the skeleton, with a unilateral predominance. They occur electively in the limbs, particularly the extremities, and are associated with bone deformities and shortening, leading to rheumatological and orthopedic problems and slowed growth [1]. Diagnosis of Ollier disease is based on specific clinical manifestations and radiographic findings, and treatment is generally conservative, surgery is performed in cases of limb length inequality, pathological fracture and malignant transformation [2].

CASE REPORT

we present the case of 3 years old patient, with no notable pathological history He had presented, since the age of 6 months, painless bone swellings that did not limit daily activity (on the right and left ankles, and on the proximal and distal interphalangeal joints of the left hand), in a context of apyrexia and preservation of general condition. Admitted for recent onset of further swellings in the right and left shoulder, rib cage, lower end of left femur and left ankle, clinical examination reveals saturoweight retardation (-3DS), diffuse hard and painless bony masses, unequal length of lower limbs (Fig 1), costal chaplet (Fig 3), and valgum knee.

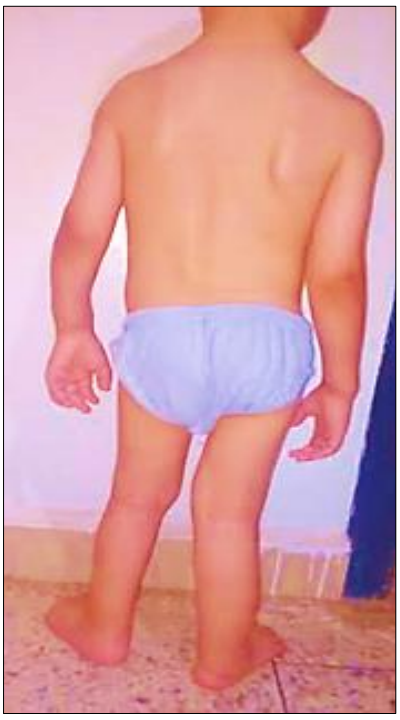

Fig. 1: Photograph showing inequality in lower limb length, with the right lower limb longer than the left.

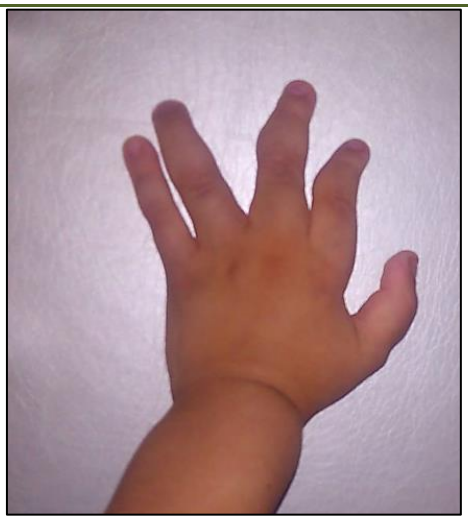

Fig. 2: Photograph of the left hand showing bony masses on the proximal and distal interphalangeal joints.

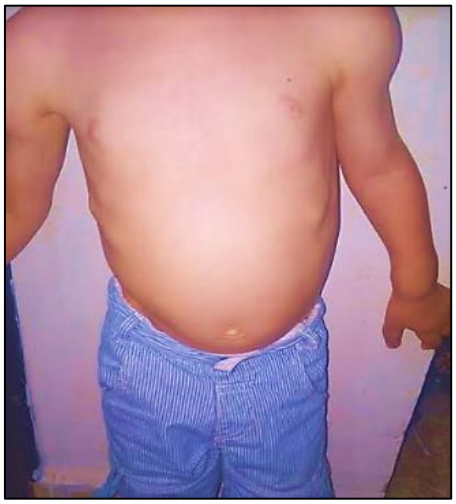

Fig. 1: Photograph showing a costal rosary and bone swellings on the distal end of the left forearm and humerus.

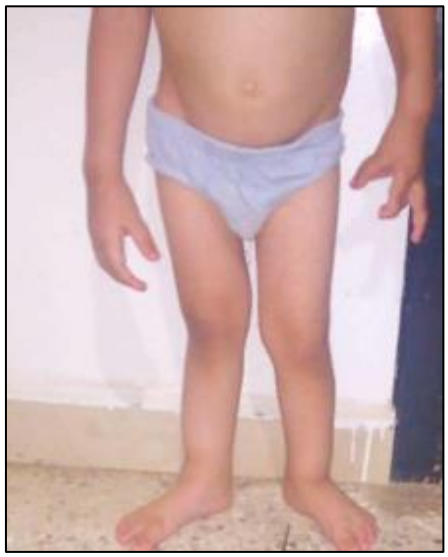

Fig 4: Photograph showing a bone mass at the lower end of the left femur.

Biological assessment was in favor of a mild inflammatory syndrome: erythrocyte sedimentation rate:18mm at 1st hour and CRP = 12.68 mg/l, a strictly normal ionogram and phosphocalcic balance.

Standard radiographs showed multiple osteolytic bone lesions: on the acromion and coronoid process on both sides, the metaphyses of the proximal and distal ends of the 2 humeri, the metaphyses of the 2 bones of the left forearm, the base of the 1st phalanx of the thumb, the left index, middle and ring fingers and the metacarpals opposite, the left side of the pelvis, the epiphyses and metaphyses of the 2 femurs (Fig 5), the metaphyses of the 2 tibias and left fibula and the talus on both sides.

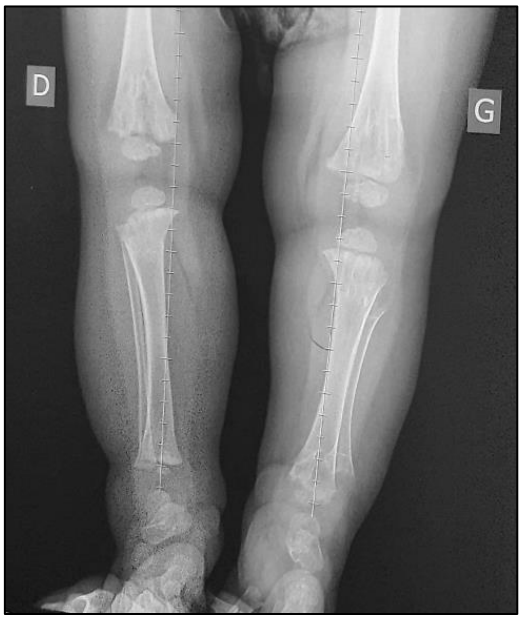

Fig. 5: Radiographs of both legs revealed multiple metaphyseal-epiphyseal osteolytic lesions on the metaphyses of the 2 tibiae and left fibula, as well as the talus on both sides.

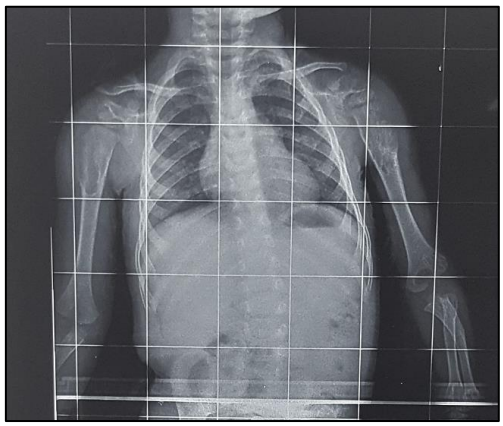

Fig. 6: Radiographs of the thorax and 2 upper limbs showing multiple osteolytic lesions on the proximal and distal ends of the 2 humeri, the metaphyses of the 2 bones of the left forearm and the acromion and coronoid process on both sides.

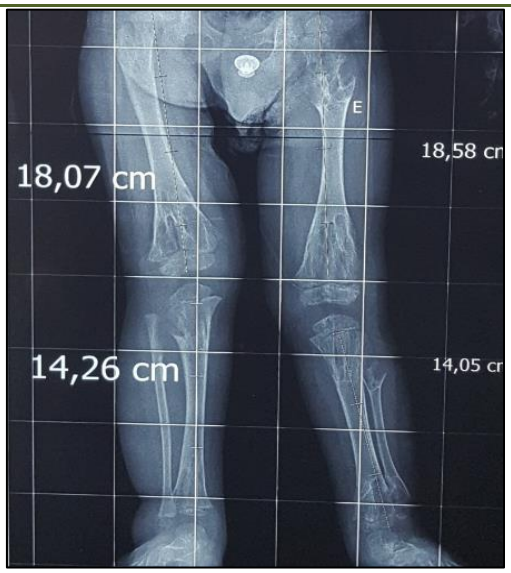

Figure 7: A standard X-ray of the lower limbs showed unequal length of the lower limbs, with the right lower limb being longer than the left.

Bone biopsy revealed a morphological aspect compatible with a chondroma, with no histological signs of malignancy. The diagnosis of Ollier disease was based on the clinical presentation and radiographic findings. Bone biopsy ruled out sarcomatous transformation of the bone lesions. The patient was put on analgesic therapy with regular rheumatological and orthopedic follow-up.

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 [3].

Described in 1899 by Louis Léopold Ollier [4], its prevalence is estimated at 1/100,000, but may be higher due to under-detection of mild forms without skeletal deformities. This condition is characterized by multiple, unilaterally distributed endogenous chondromas associated with limb deformities [5]. 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.

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 [6]. Ollier illness is characterized by genomic copy number variations and mutations that inflence a variety of important functions [7], several articles have recently proposed heterozygous mutations in the PTHR1, IDH1 (most common), and/or IDH2 genes as genetic aberrations [8].

The usual clinical manifestation of Ollier disease includes palpable, asymptomatic, painless bony masses on the fingers, toes and metacarpals, with a unilateral predominance [9]. Although lesions are usually bilateral, with unilateral predominance leading to asymmetric distribution. The masses increase in size as the child grows, resulting in asymmetrical limb shortening and deformities such as valgum and varum knee, the latter being the most common [9].

Enchondromas are most commonly seen in the phalanges and metacarpals, and are rarely seen in the carpal bones [7]. Ollier disease is also frequently observed in long bones such as the femur and tibia. The trochanters of the femur are frequently affected, while the neck of the femur is relatively spared [10].

An international multicenter retrospective study conducted between 2007 and 2011 found that 59% of patients had enchondromas in the femur, 47% in the tibia, 32% in the humerus, 27% in the fibula, 25% in the pelvis, 45% in the small tubular bones of the hands and 21% in the small tubular bones of the feet [11].

Correlation between clinical presentation and radiological findings is necessary to establish the definitive diagnosis [12]. Imaging plays a very important role in the diagnosis of Ollier disease, helping to confirm the diagnosis, pinpoint its anatomical location, measure the size of enchondromas, and detect signs of sarcomatous transformation [13].

On standard radiography Ollier disease has specific radiographic features, with multiple osteolytic lesions in the center of the tubular bones of the hand, foot and long bones, particularly in the metaphyseal regions [7]. Symmetrical involvement is sometimes present, but is always predominantly unilateral [14].

Enchondromas appear as radiolucent images, located on the metaphysis of long bones or on the bones of the hands and feet, with diaphyseal extension. These radiolucent areas have a homogeneous appearance and are surrounded by a fine border of osteosclerosis.

During the course of the disease, pathological fractures are observed in some patients due to cortical thinning [14]. According to reviews, pathological fractures are observed in 40-60% of patients at the initial presentation of the disease [15].

Signs of malignant transformation should be sought, including extension into the soft tissue, cortical erosion, enchondromal irregularities and non-uniform mineralization [1-17]. Computed tomography (CT) is more sensitive than conventional radiology to Asses the type of matrix mineralization, calcification pattern, lobulated lesion edges, and the degree and extent of endosteal scalloping [7]. CT is also effective in determining the size of the lesion and existence of any

soft tissue component that would support a chondrosarcoma diagnosis [7].

MRI is particularly useful for assessing intraosseous and soft tissue involvement [18]. It may be requested in cases of pathological fracture, when characterization of the lesions is necessary prior to treatment [16]. Enchondromas can demonstrate a high uptake on florodeoxyglucosepositron emission tomography (FDG-PET) which can be challenging in patients with a known malignancy as it can sometimes mimic a metastatic lesion [7].

Histological analysis has a limited role and is mainly used in cases of suspected sarcomatous transformation [1]. The anatomopathologist must specify the histological and cytological caract features of the chondrocytes, which are of capital importance in the diagnosis of the type of cartilaginous tumor [19]. Histological analysis of chondrocytes must also be carried out in viable, non-remaned and non-necrotic territories [19].

Diagnosis is therefore based on a combination of radiographic (cortical destruction, soft tissue extension), clinical and histological criteria. 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 [8].

Physiotherapy such as ultrasound, cryotherapy, CO2 laser with stretching, active, mobilization, occupational therapy, and coordination exercises appear to signifiantly improve the functional abilities [5]. Surgery, on the other hand, is indicated in case of deformity, limb-length disparity, pathological fracture, and malignant transformation [7].

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