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Case Report

CASE REPORT

Ollier disease: the first report in Syria

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Abstract

Ollier disease is a rare nonhereditary skeletal disorder, characterized by multiple enchondromas, which are noncancerous growth of cartilage. In this report, we present a case of Ollier disease in a 10-year-old Syrian boy. The patient presented with multiple boney masses on hands; he had a history of pathological fractures when he was 5, which caused crippling. We analyzed the clinical, radiographical and pathological characteristics of our patient, which helped us to reach the final diagnosis. Ollier disease is a benign bone tumor, but it has a risk of malignant transformation into chondrosarcoma. The aim of this report is to document the presence of Ollier disease in Syria to help other Syrian physicians considering this disease in the differential diagnosis if they face similar presentations.

INTRODUCTION

Ollier disease was first described in 1898; it is a skeletal disorder characterized by multiple enchondromas, which are cartilage nodules that develop within the bones. The prevalence of Ollier disease is 1 in 100 000 and the pathogenesis of the disease is unknown.

The most common complaint is cosmetic deformity due to multiple swellings on hands and feet, and most patients present with limping caused by asymmetric shortening of an extremity [1, 2]. Diagnosis of Ollier disease depends on clinical, radiographic and pathologic findings. Enchondroma in Ollier disease has a potential risk of malignant transformation into chondrosarcoma. Treatment options have limited effect, but when complications exist, surgery is recommended [3].

CASE REPORT

A 10-year-old Syrian boy presented to pediatric hospital complaining of limping with multiple painless asymmetric nodular swelling of proximal phalanges bones (Fig. 1A). He had a history of multiple fractures in long bones characterized by abnormal healing started when he was 5. Physical examination revealed that intelligence was unaffected, vital signs were normal and the patient also developed progressive skeletal distortions in proximal and distal phalanx of hands with a slight functional limitation, enlargement in distal end of both right forearm and right leg compared with left ones. We noticed shortness of right lower limb, which caused crippling (Fig. 1B). Examination of his parents was normal. The patient status was stable at his last review and he is being followed up annually.

We collected blood cell data during his first visit to the hospital. All laboratory test results are demonstrated in Table 1.

A plain radiograph was performed, showing multiple bone lesions in extremities, whereas skull and chest were normal (Fig. 2).

We performed a biopsy from nodules in the right hand to exclude malignancy (Fig. 3).

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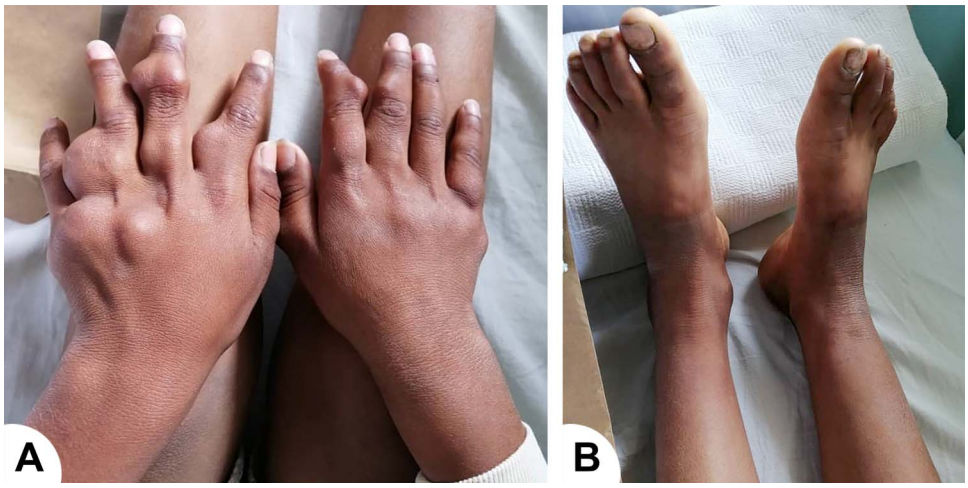

Figure 1: Clinical photographs of both hands showing multiple swellings (**A**) and shortness of right lower limb (**B**).

Table 1: Laboratory tests on admission.

Variable	Result	Normal range
WBC	9600/µl	4400–11 000/µl
Neutrophils	76%	40–70%
Lymphocytes	17%	20–40%
HB	13 g/dl	13–16 g/dl
RBCs MCV	79 fl	82–96 fl
PLT	293 µl	150–450 × 103/µl
Cl	104 mmol/l	95–105 mmol/l
Na	138 mmol/l	135–148 mmol/l
K	3.83 mmol/l	3.5–5.0 mmol/l
Ca	10.2 mg/dl	8.5–10.5 mg/dl
P	4.44 mg/dl	2.5–4.5 mg/dl
Uric acid	3.48 mg/dl	3.4–7.0 mg/dl
ALP (Alkaline phosphatase)	297 IU/l	44–147 IU/l
Urea	25 mg/dl	10–50 mg/dl
Creatinine	0.67 mg/dl	0.7–1.36 mg/dl
ALT	13 IU/l	8–20 IU/l
AST	24 IU/l	8–20 IU/l
ESR	24 mm/h	0–20 mm/h
TP (total protein)	7.61 g/dl	6–8 g/dl
CRP	0.31 mg/l	<2 mg/l

DISCUSSION

Enchondromas are neoplasms of hyaline cartilage, which are a type of benign bone tumor family. The most affected bones are the short tubular bones of the hands and feet, but it may present in long bones such as the femur, humerus and tibia. Ribs and flat bones are rarely affected. If there are multiple enchondromas, we call this condition enchondromatosis, also known as Ollier disease [1, 4].

Ollier disease was first described at the end of the 19th century, and in 1978, Spranger et al. put a classification of enchondromatosis based on radiographic appearance, anatomic site and mode of inheritance. The result was six subtypes of enchondromatosis: Ollier disease is classified as Spranger type I and Spranger type II is Maffucci syndrome, which is the same as Ollier disease but with hemangiomas [5].

Ollier disease is usually non-familial disorder, and therefore, it appears to occur spontaneously. To date, no evidence suggests that it is inherited. The atypical distribution of lesions in the disease suggests that postzygotic somatic mosaic mutations are the cause of the malformation of endochondral bone, such as heterozygous mutations in the isocitrate dehydrogenase [6]. Also heterozygous mutations and missense mutations in PTH/PTHrP type I receptor (PTHR1) or dysregulation in the Indian hedgehog signaling pathway (IHH) may cause growth of enchondroma in Ollier disease patients, as these two play a role in regulating chondrocyte differentiation, proliferation and maturation especially during endochondral ossification [7].

The diagnosis of Ollier disease is based on clinical manifestations and specific radiographic findings, but histopathologic examinations have limited influence, unless malignancy is suspected [8].

The lesions may affect multiple bones and generally occur bilaterally, with unilateral predominance leading to asymmetric distribution, which cause leg discrepancy and pathologic fractures. Patients usually present with palpable painless bony masses, which can be in various size, location, number and age of onset [1].

Plain radiography is highly important in the evaluation of the disease and usually provides adequate information. Radiographic findings include multiple, radiolucent, homogeneous lesions with a well-defined margin. When the lesions increase in size and cause pain, ultrasound, magnetic resonance imaging and scintigraphy are recommended [2, 3].

The enchondroma is composed of benign hyaline cartilage nodules with well-defined margins. Histopathologic examination differentiates between benign enchondromas and malignant chondrosarcomas [9].

The differential diagnosis may include hereditary multiple exostosis. The osteochondromas seen in hereditary multiple exostosis are located at the bone surface, whereas enchondromas are located in the center of bones [1].

Treatment of Ollier disease is usually conservative. Surgical intervention is performed in cases of limb-length discrepancy, pathological fracture and malignant transformation [10].

The prognosis of Ollier disease is difficult to assess as there can be a variation in the size, number and location. There is a risk of malignant transformation, in these case a careful lifelong follow up is necessary [1].

This is the first case reported of Ollier disease in Syria. This case raises the awareness between Syrian physicians

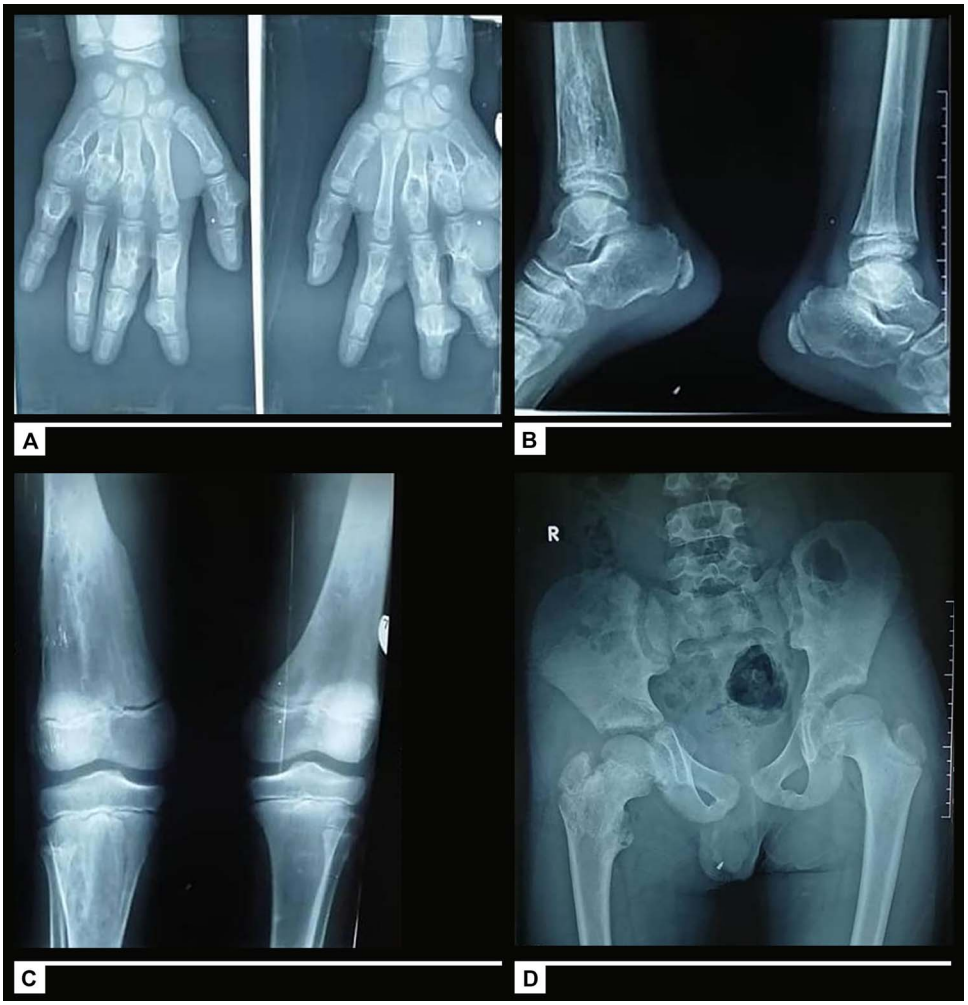

Figure 2: Multiple radiolucent, well-defined, lytic lesions are seen in metacarpal and phalanges bones (**A**), distal end of right leg and right femur (**B** and **C**) and proximal end of right femur (**D**), in keeping with multiple enchondromas.

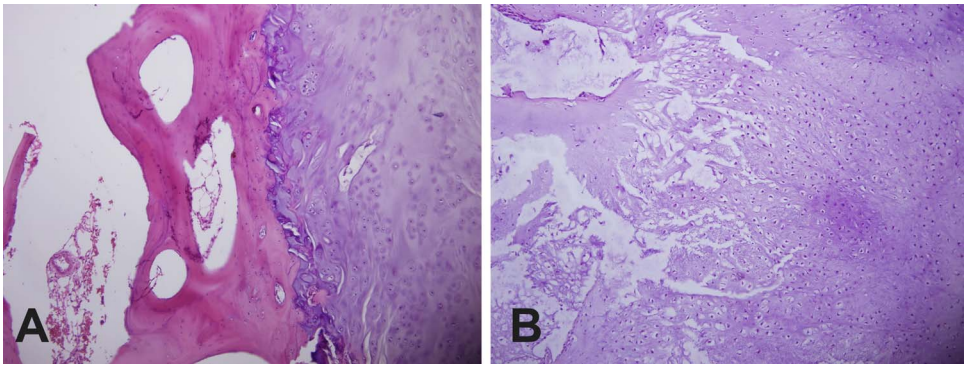

Figure 3: Microscopic examination of enchondromas showed hyaline cartilage, osseous tissue and trabeculae (**A**) and chondrocytes with cytological atypia (**B**).

to discover more cases of Ollier disease, which in turn help patients to obtain pre-diagnosis and better therapy outcome.

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CONFLICT OF INTEREST STATEMENT

None declared.

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ETHICAL APPROVAL

No approval was required.

PATIENT CONSENT

Written informed consent was obtained from the patient for publication of this article and the figures related to it.

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