CASE REPORT

Ollier's disease: a case report and review of literature

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ABSTRACT

Background: Ollier's disease is a rare nonfamilial skeletal disorder characterized by the occurrence of multiple enchondromas with asymmetrical involvement of the extremities either exclusively or predominantly. The etiology and underlying factors contributing to this are under investigation. The diagnosis is based on clinical and radiographic features with the complementary role of radionuclide bone scanning to assess radiologically depicted lesions and to ascertain the complete extent of skeletal involvement.

Case Presentation: The present study describes the case of a 55-year-old-male patient who presented with multiple marked swellings from fingers of the right forehand and forearm having suppurative discharge and gross deformity that developed when he was eight years of age. On examination, gross enlargement of the right thumb and fifth digit was seen with hard nodules along with redness, increased vascularity, and areas of necrosis. X-ray showed multiple expansile lytic lesions involving metaphysis and diaphysis with ring and arc calcifications involving distal radius and ulna and almost all metacarpal and phalanges. Bone scan showed asymmetrical unilateral involvement of tubular limb bones sparing the axial skeleton.

Conclusion: Ollier's disease is a benign, rare nonhereditary skeletal disorder with its malignant transformation being a serious form of complication. Diagnosis rests on the clinical and radiological features.

Keywords: Ollier's disease, multiple enchondromas, malignant transformation.

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Introduction

Also termed Multiple enchondromatosis, Dyschondroplasia, and Multiple cartilaginous enchondroses, the disorder was first described by the French surgeon Louis Léopold Ollier's in 1899 [1,2]. The disorder is characterized by the presence of at least three enchondromas (i.e., benign intraosseous cartilaginous lesions) and asymmetrical distribution of the bones [3,4]. Ollier's disease primarily affects the long bones and cartilage of the joints of the arms and legs, specifically the metaphyses. The pelvis is often involved; and even more rarely, the ribs, breastbone (sternum), and/or skull may also be affected [5]. The disease often manifests in infancy and early childhood, usually starting with the appearance of palpable and painless bony masses on the involved limbs. At present, the pathogenesis of Ollier's disease is not decisively determined [6]. It is proposed that it may be related to somatic mosaic mutations of isocitrate dehydrogenase (IDH)1 and IDH2 [7] resulting in abnormalities in signaling pathways controlling the proliferation and differentiation of chondrocytes, leading to the development of intraosseous cartilaginous foci [7,8]. It has been accentuated that Ollier's disease usually halts spontaneously with skeletal maturity; therefore, any lesion displaying activity or increased uptake after the termination of the growth period requires exhaustive examination. Scintigraphy has been recommended as worthwhile in observing the lesions and of the development of any malignant transformation [9]. The most severe complication is the malignant transformation of enchondromas toward secondary chondrosarcomas [1,10].

Case Report

A 55-year-old-male patient presented to us with multiple marked swellings having suppurative discharge and gross deformity of the right hand. These swellings appeared when he was 8 years of age. The swellings were soft,

asymptomatic, and developed from the fingers of the right hand and distal forearm. The size of the swellings increased with age causing gross deformity of the hand; however, there was no appearance of such swellings in any other part of the body. There were no associated symptomatic complaints. The patient had a history of falls 1 year back with no significant trauma. The patient had no family history of similar pathology. For the last 3 months, swellings on the thumb and fifth digit became infected. Patient was referred for metastatic workup via scintigraphy.

Physical examination

On examination, gross enlargement of the right thumb and fifth digit was seen (Figure 1a and b). Multiple hard nodules were noted along with redness, increased vascularity, and multiple areas of necrosis. Small swellings with minimal deformity on second and fourth digits were noted with sparing of third digit. Deformity of the right forearm (more marked on the ulnar side) was also seen. Systemic examination was unremarkable.

X-ray findings

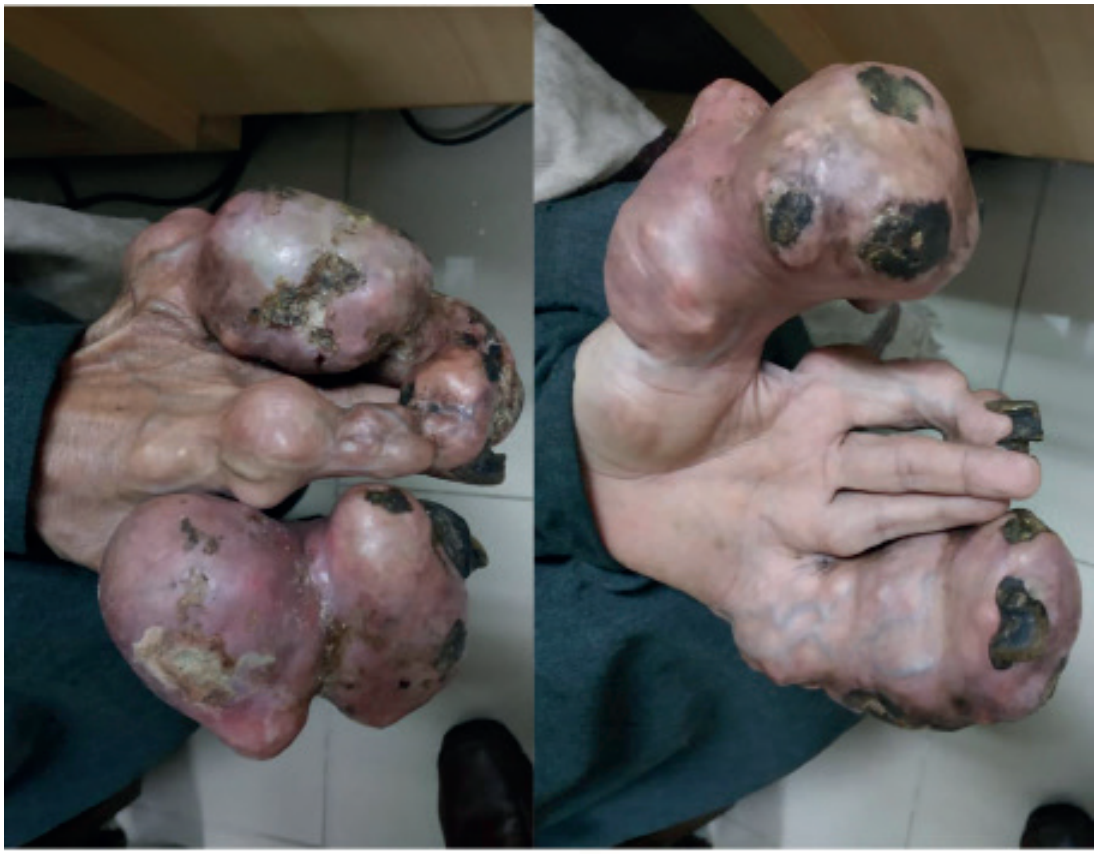
X-ray left forearm and hand show multiple expansile lytic lesions involving metaphysis and diaphysis with ring and arc calcifications involving distal radius and ulna and almost all metacarpal and phalanges sparing only third digit causing gross deformity of the hand and distal forearm (Figure 2a). On X-ray of left arm, a large oval shape expansile lytic area involving diaphysis of proximal left humerus was found (Figure 2b).

Scintigraphic findings

Bone scan showed grossly expansile metacarpals and phalangeal regions of medial and lateral ends of left hand showing moderate to intense radiotracer uptake (Figure 3a). Distal radius and ulna also show expansile patchy uptake. A focal area of increased uptake was noted in the proximal and middle parts of the shaft of the left humerus. Mild increased uptake was also seen in left-sided upper ribs, acromion of the left scapula, and lateral border of the left clavicle (Figure 3b). Bone scan appearance of asymmetrical unilateral involvement of tubular limb bones sparing the axial skeleton was reported to be consistent with multiple enchondroma seen in Ollier's disease.

Discsussion

Enchondromas are benign chondrogenic tumors composed of hyaline cartilage that typically occur in the



a b

Figure 1. *(a) and (b) Multiple enchondromas with gross enlargement of the right thumb and fifth digit. Multiple hard nodules along with redness, increased vascularity, and multiple areas of necrosis. Small swellings with minimal deformity on second and fourth digits with sparing of third digit.*

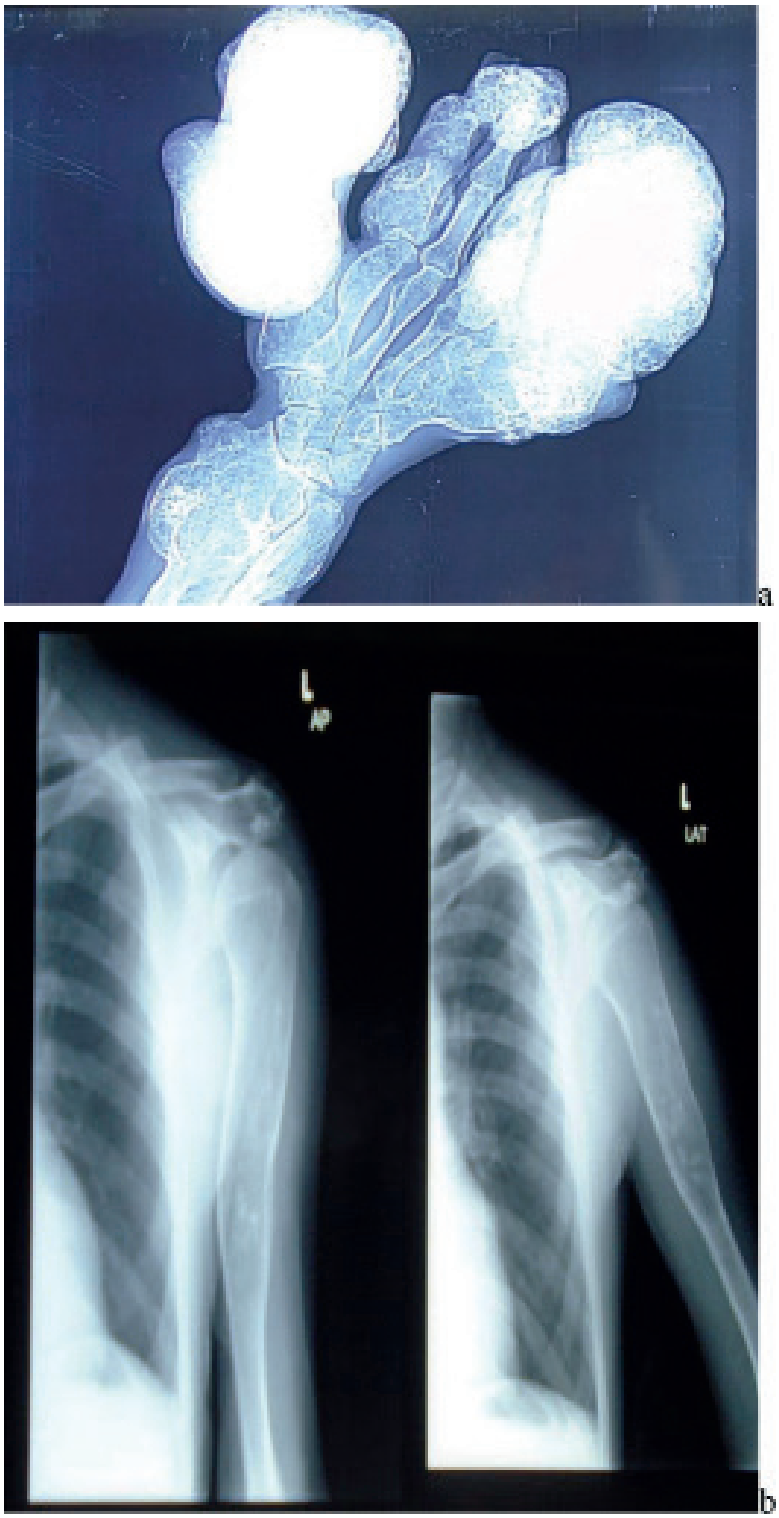


Figure 2. *(a) Multiple expansile lytic lesions involving metaphysis and diaphysis with ring and arc calcifications involving distal radius and ulna and almost all metacarpal and phalanges sparing only third digit causing gross deformity of the hand and distal forearm. (b) A large oval shape expansile lytic area involving a diaphysis of the proximal left humerus.*

medullary cavity of the diaphysis or metaphysis. Lesions affecting the proximal bones are more severe and the region of the knee joint and the lower end of the radius and ulna are particularly common sites and are more prone to pathological fractures [1,10]. The estimated prevalence of Ollier's disease is approximately 1 in 10^5. Due to its rarity, literature focusing on it is limited [11]. The characteristic X-ray finding includes multiple, radiolucent, homogenous oval, or elongated lesions in the metaphyses of tubular bones with extension into the diaphyseal and epiphyseal regions [4,12]. Some of these enchondromas may show expansile behavior called enchondromas protuberance

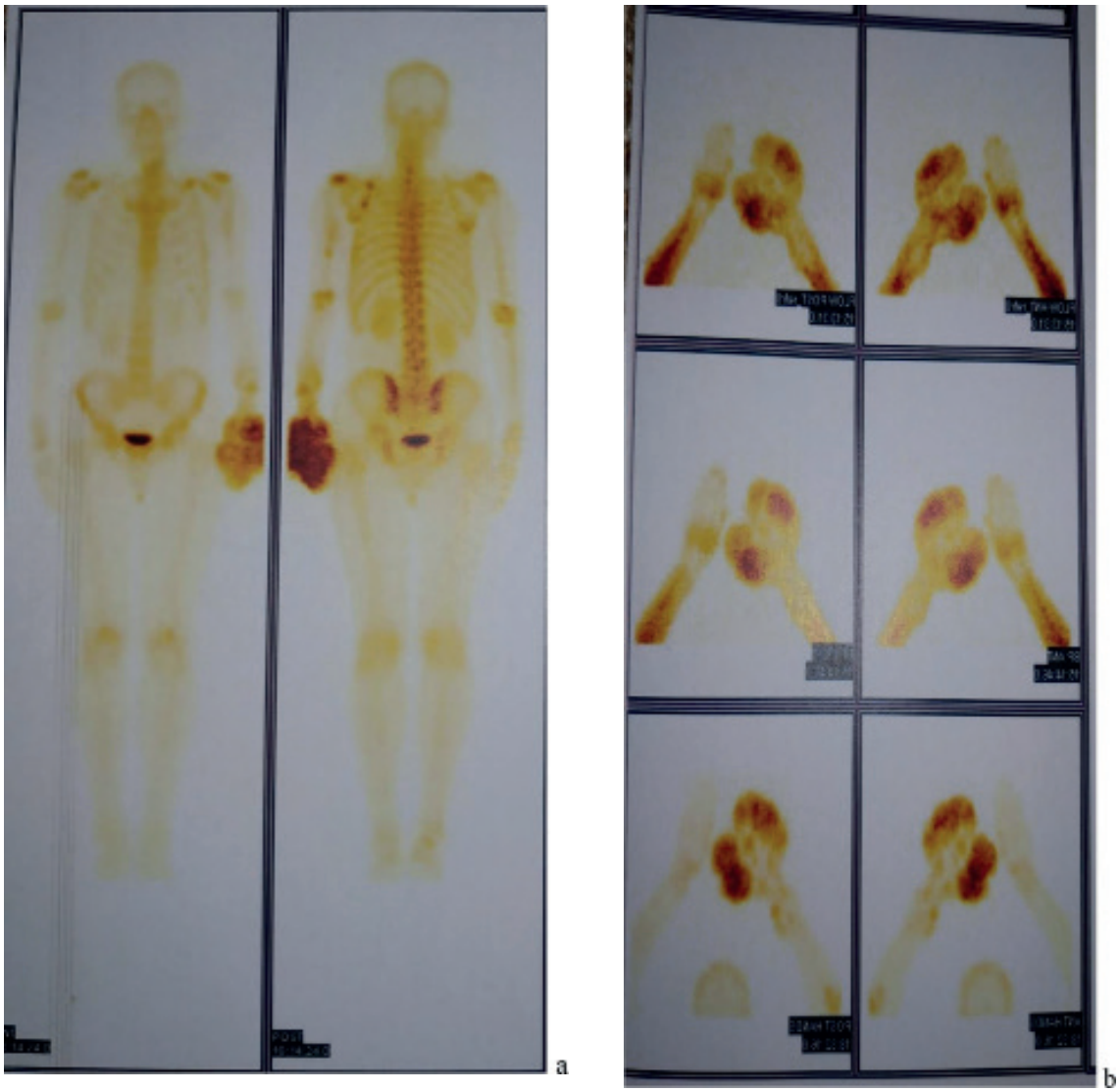


Figure 3. *(a) and (b) Grossly expansile metacarpals and phalangeal regions of medial and lateral ends of the left hand with moderate to intense radiotracer uptake. Distal radius and ulna also show expansile patchy uptake. Focal area of increased uptake is seen in the proximal and middle parts of the shaft of the left humerus, left-sided upper ribs, acromion of the left scapula, and lateral border of the left clavicle.*

[1,8]. The radiographic findings are typically more severe than the clinical examination would suggest [4]. Histologic analysis has a limited role and is mainly used if malignancy is suspected [9]. Technetium-99m methylene diphosphonate (MDP) whole body bone scan is a sensitive investigation to ascertain the extent of skeletal involvement particularly in the asymptomatic sites and is recommended as useful in the monitoring of lesions and the development of any malignant transformation [2,5]. Enchondroma can have high MDP uptake on scintigraphic imaging and occasionally may mimic metastatic lesions in patients being screened for metastatic disease [6]. Most frequently, malignant transformation occurs in long bones and flat bones while this is less common in the hands and feet [1]. The reported incidence of malignant transformation is highly variable in the range of 5%-50% in the literature [1] There is no medical treatment for Ollier's disease [4] Asymptomatic patients of multiple enchondromatosis do not need medical or surgical treatments [8] Lifelong monitoring is required in Ollier patients given the risk of malignant transformation [1]. Surgical intervention is necessary in case of pathological fractures, growth defect, and malignant transformation [1]. The prognosis for Ollier's disease is difficult to assess [4]. Patients with malignant transformation from enchondroma toward grade I, II, and III chondrosarcoma have a 5-year survival rate of 90%, 81%, and 43%, respectively, and 10-year survival rate of 83%, 64%, and 29%, respectively [1].

Conclusion

In brief, Ollier's disease is rare disease characterized by multiple enchondromas with predominately unilateral limb involvement. Advancements in research methods are leading to a more clear understanding of underlined pathogenesis. We presented the clinical, radiological, and scintigraphic findings in a systematical style through the

exploration of a typical case and a literature review, in order to effectively guide clinical practice.

Conflict of interest

The authors declare that there is no conflict of interest regarding the publication of this article.

Funding

None.

Consent to participate

Written and informed consent was taken from patient to publish this case report.

Ethical approval

Ethical approval is not required at our institution to publish an anonymous case report.

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