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Case Report

Ollier disease: multiple enchondromatosis: case report and review of literature

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ABSTRACT

Multiple enchondromatosis is a rare disease in which cartilage tumors appear at the level of the skeleton. The incidence is unknown due to the very few cases reported in world literature. We presented the case of a patient at the plastic surgery department at General hospital Dr. Ruben Leñero, otherwise healthy, referring first clinical manifestations at childhood with an increase in volume and deformity at the second and third fingers of the left hand.

Keywords: Enchondroma, Multiple enchondromatosis, Ollier disease, Cartilage tumor

INTRODUCTION

Enchondromas are boney skeleton tumors, mostly benign, they emerge near the growth plate. When there are multiple cartilaginous tumors, it is called Ollier disease or multiple enchondromatosis according to the WHO nomenclature. Prevalence of Ollier disease is estimated at 1:100,000.[1,4]

Clinical manifestations often appear in the first decade of life, characterized by asymmetrical cartilaginous lesions of unique morphology.[2] It is a known non-hereditary disease with a high risk of malignant transformation into chondrosarcoma. Previous reports revealed that the incidence of malignant transformation is 5-50%.[4,8,10]

Main abnormalities caused by enchondromas include skeletal deformities, extremity length discrepancies and potential malignant degeneration into chondrosarcoma.[3] It is still unclear if this is an isolated genetic defect or a mosaic of mutations. Diagnosis is based on clinical examination and plain X-rays; first described in 1899 as a dyschondroplasia.[13]

CASE REPORT

A 30 year old, healthy male, refers since he was 3 years old, being studied for an increase in the volume of second and third fingers of the left hand, progressively as the patient grew, producing notorious asymmetry, but no limitation of the arch of movement thus not impairing everyday activities. He attended to the plastic surgery emergency department at General hospital, Dr. Ruben Leñero referring pain at rest and difficulty at flexion-extension movements of diseased fingers.

At the clinical examination we found marked asymmetry, mostly at the expense of the medial and distal phalanx of second and third fingers of the left hand, with pain at palpation, but also identifying firm, non-mobile, tumors at the skin surface both at dorsal and volar sides, sensibility and capillary filling remained adequate, rest of the hand presented no alterations (Figure 1 A and B).

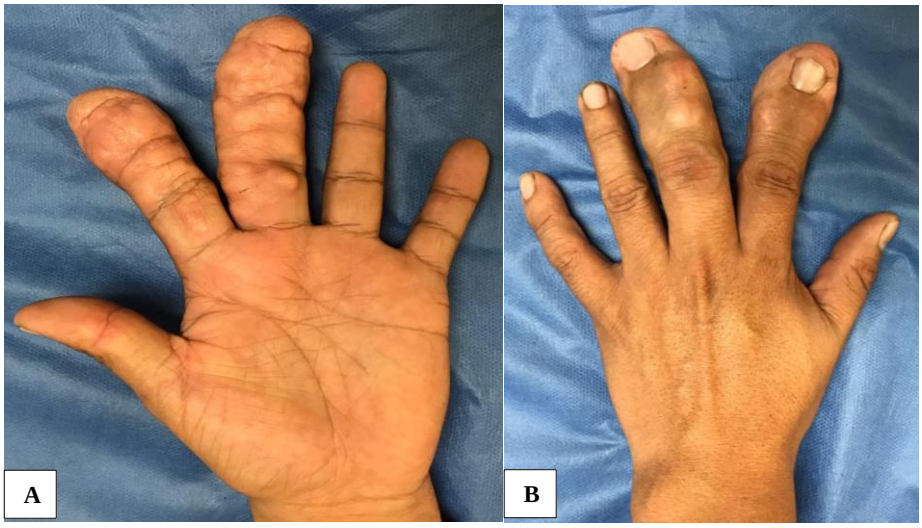

Figure 1 (A and B): Findings.

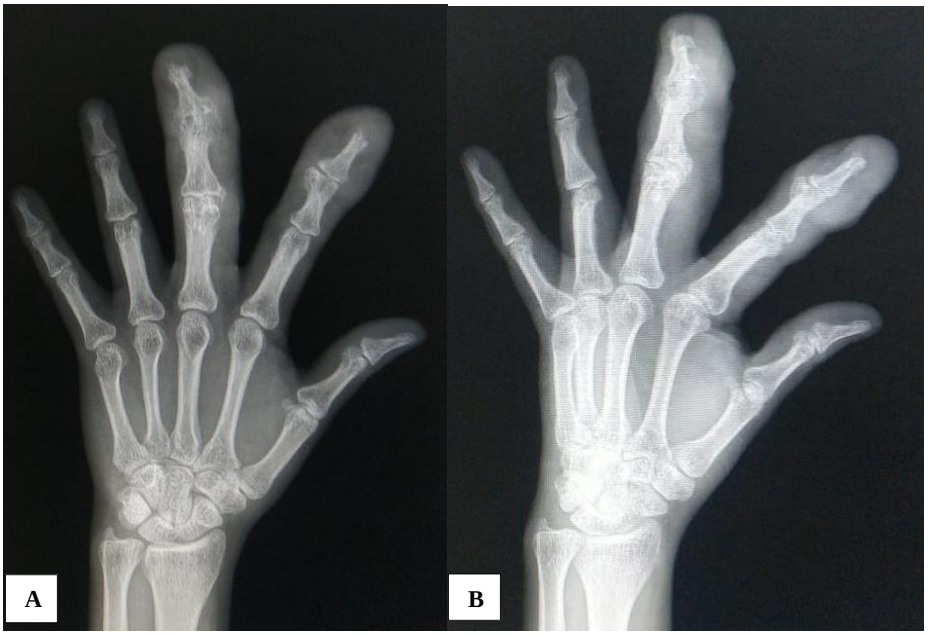

Figure 2 (A and B): X-ray.

Plain X-rays of the hand were taken, finding lytic lesions at the level of the medial and distal phalanx of both fingers, with loss of the proximal and distal interarticular space but no actual evidence of bone deformity (Figure 3 A and B).

The patient referred himself incapable of doing his everyday activities due to pain and decreased mobility, predominantly at the third finger, finding it non-functional. So, an aesthetic-functional amputation was performed (Figure 3 A-D).

The histologic diagnosis was compatible with multiple enchondromatosis, pathology department reported a conglomerate of lesions of 7×0.5 cm all together at the level of the distal phalanx, at the proximal interphalangeal articulation, a 1×q cm single lesion was reported, nodular, solid and firm of irregular borders.

At three months follow up, the patient had adequate evolution, finding improvement of flexion and extension movements, pinch, grip and no pain as referred by the patient. We continued periodical follow up appointments to detect any new tumors that may develop or progression of the disease of the other affected phalanx (Figure 4 A and B).

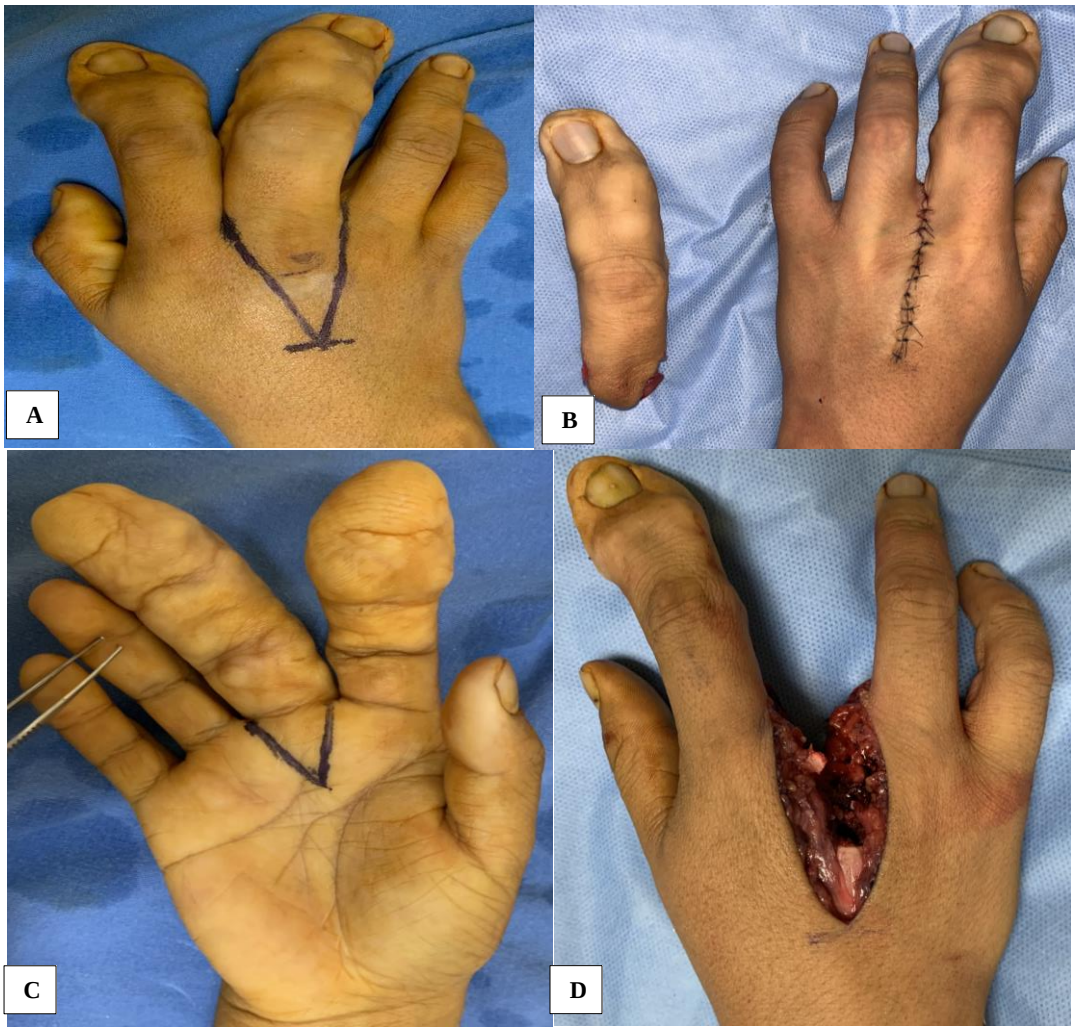

Figure 3 (A-D): Surgical procedure.

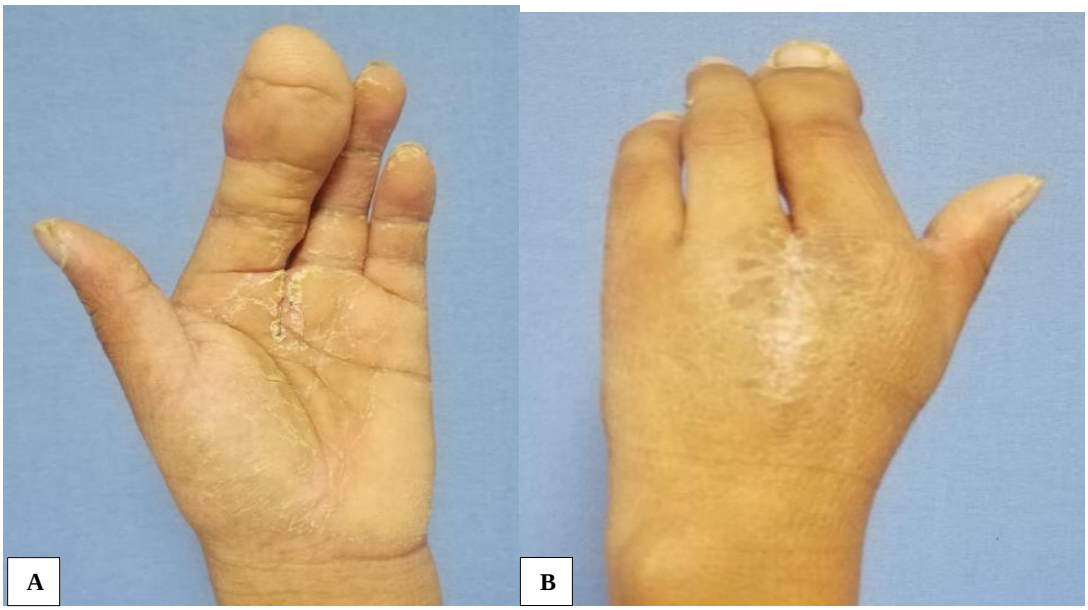

Figure 4 (A and B): 3 months follow up.

DISCUSSION

Enchondromas is a benign cartilage tumor most commonly located in the little finger (65%) and proximal phalange (60%).[8] According to the WHO classification of soft tissue and bone in 2013, multiple enchondromas include Ollier disease and Maffucci syndrome.[9,13]

Three components should be considered in the diagnosis of Ollier disease. They are clinical description, radiographic finding and pathologic imaging. Radiography is highly important in the evaluation of treatments and follow-up prognosis.[8]

Ollier disease involves short tubular bone of the hand mostly, followed by the femur, tibia, fibula, humerus, radius and ulna. Enchondromas can be in various sizes, location, number, age of onset and diagnosis. It is a typical form of chondrosarcoma. Skeletal abnormality such as bending, shortening, pathological fracture and asymmetric deformity can be seen in affected patients.[8]

In 1935 Hunter and Wiles presented s-revision of 40 patients, not being able to determine the risk of malignant degeneration.[6] Jeffe in 1958 estimated risk of more than 50% of malignization in a study based on histological findings that could not differentiate between enchondromas and low-grade chondrosarcomas.[7]

The overall incidence of Ollier disease is low which might be due to infradiagnosis. When encountering with cartilaginous tumor and suspecting enchondroma or chondrosarcoma, one must have Ollier disease as a differential diagnosis.[5] Given the differential diagnosis from Maffucci syndrome, which is another type of multiple enchondromatosis, multiple enchondroma is accompanied with soft tissue hemangiomas and occasionally lymphangiomas.[8,11]

Solitary enchondromas occur in patients aged 10-40 years and 75% of cases happened before age 20 years.[8] Pansuriya et al reported about the genetic evidence of somatic mosaic isocitrate dehydrogenase 1 (IDH1) and isocitrate dehydrogenase 2 (IDH2) mutations in Ollier disease.[12]

CONCLUSION

Multiple enchondromatosis is a rare disease that constitutes a diagnostic challenge with other benign and malignant diseases, that also have a low incidence, thus making it more challenging for a surgeon to diagnose or even think of. Most cases reported of Ollier disease affect long bones metaphyseal region, so it is of relevance to report a case affecting a patient's hand and requiring surgery to improve lifestyle.

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