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Ollier´s disease: Multiple enchondromatosis

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Abstract
Ollier disease is a rare condition involving the development of intraosseous benign tumors known as enchondromas. These are usually formed during early stages of growth when the cartilage or growth plate is still biologically active. Once the patient matures, these tumors cause dysmorphic deformities and fragile bone fractures in adulthood. It is important to search for cutaneous vascular tumors in order to differentiate with Maffuci syndrome. Malignization of the enchondromas is also a severe complication. Regular control of the patients is key to detect malignant transformations and stablish an early treatment. Malignant transformation involves the development of increased vascular structures around the tumor, thus gammagraphic imaging is key to control the disease. Literature describing this disease is rare and treatment options are limited. Surgical treatment is often required when the osteochondromas transform or become very symptomatic; causing pain and articular malfunction. Our report describes a case of Ollier disease diagnosed in our center and discusses the core concepts of the disease and therapeutic management.

Keywords: Ollier disease, multiple enchondromatosis, enchondromas

Introduction

Case Report

A 38-year-old patient presented to our hospital trauma care with intense shoulder pain and considerable swelling. The patient referred the pain was triggered by a mild traumatism in the shoulder a few days ago.

The physical examination of the patient showed a shortened shoulder and a left superior limb partially atrophied. Sensitive and vascular peripheral systems on the superior limb were conserved, however motor function was compromised. Fully abduction and external rotation of the shoulder was not possible.

X-ray images were taken of the shoulder (Figure 1). An image compatible with multiple enchondromas was seen. Further anamnesis of the patients proved more symptoms in other skeletal areas which were not related to the initial trauma. These regions included hands and both knees. The patient described difficulties to write and increased finger pain with intense usage of both hands. Knee symptoms were also prominent: the patient referred long term limitations in walking and climbing up stairs. The physical examination of the knee proposed palpable osseous appendices in both internal and external compartments of both knees. Further images were taken, including CT scans (Figure 2 and 3).

With a thorough anamnesis and the clinical manifestations of the patient and the help of complimentary tests, the patient was diagnosed with Ollier disease.

Further studies including MRI and gammagraphy of the patient's shoulder (Figure 4) were performed due to an increment of the swelling and continuous pain, suspecting a malignant transformation of enchondromas into chondrosarcomas.

The shoulder MRI with contrast showed one of the enchondromas invading part of the axillary vascular bundle. However, the gammagraphy showed no malignant development.

The patient was treated initially using low doses of corticoids and oral pain killers. During the follow up of the patient, posterior gammagraphic controls were done every 6 months. There was no malignant development in a 2 year follow up.

However, the intense pain in the shoulder made us consider a surgical solution: part of the large osteocondroma found in the shoulder was removed and an iliac crest autograft with hydroxyapatite additive compound was added in order to fill up the anatomical defect.

Currently, the patient presents no symptoms and continues a regular follow up.

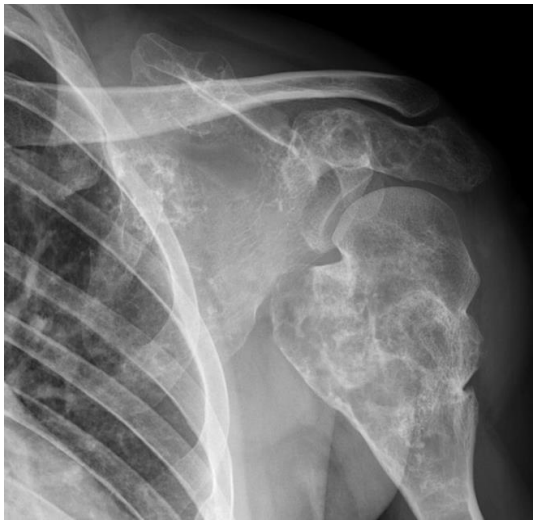

Fig 1: Sholder X-Ray image showing extense osteochondroma in the proximal humerus and scapula.

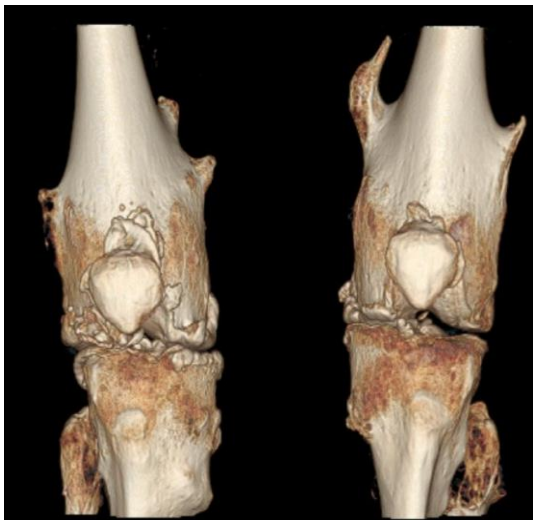

Fig 2: 3D reconstruction CT scan of both knees. Intense affection of the articular surface.

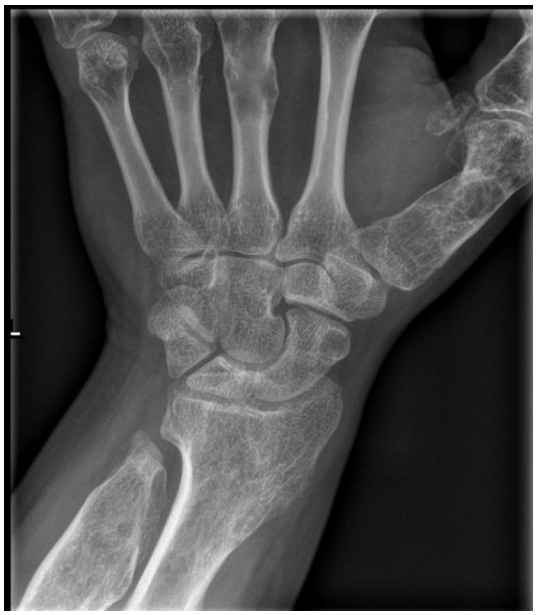

Fig 3: X- Ray image of the left hand. Osteochondromas were found affecting the forearm, wrist and fingers.

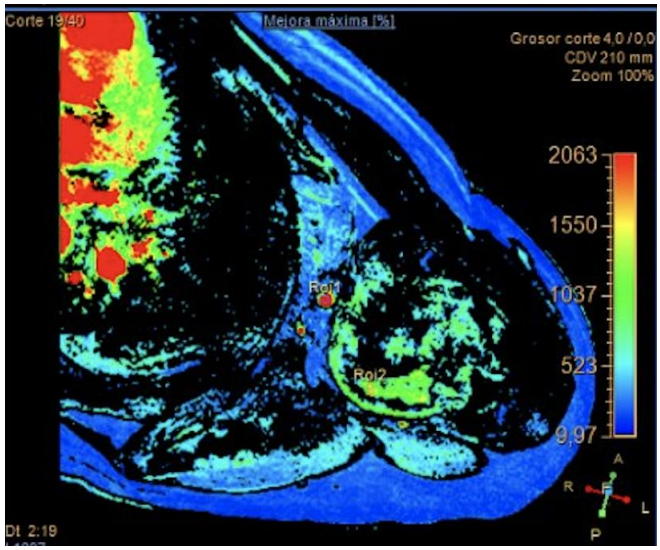

Fig 4: Gamagraphic study showing heterogeneous capitation typical of osteochondroma. No hyperemia or intense vascular invasion is seen.

Discussion

Ollier disease or multiple enchondromatosis is a rare pathology with a prevalence of 1 of 100 [1]. The disease is not heritable disease, thus there are some genes identified that could trigger the onset of the disease in patients [2].

Multiple enchondromatosis is usually asymptomatic during the first years of life. During childhood, there is usually a quick onset of clinical manifestations such as bone pain, unspecific articular malfunction and leading to the presence to palpable tumors in bone surfaces. The first osseous areas to affect are the hand and fingers followed by tibia and femur[3]

The most severe complication is the malignant transformation forming chondrosarcomas [4]. There are some risk factors that could favour this malignancy that include: central enchondromas such as pelvic and spine locations, age above 40 years old and bigger tumors [5].

There should be a gammagraphic control of the disease as patients in their third and fourth decade of life are more prone to develop malignant sarcomas.

Ollier disease is also associated to hepatic carcinoma, pancreatic and ovarian cancer.

Patients with 3 or more enchondromas should be screened in order to detect Ollier disease. An early diagnosis of the disease and the control of the osteochondromas increase patient's survival, malignancies can be detected and treated before systemic affection occurs.

Moreover, differential diagnosis with Maffucci syndrome should be done. In this entity, the enchondromas are also associated to vascular deformities in the skin.

The diagnosis of Ollier disease is mainly done by means of clinical and imaging data. Malignancy is often presented as acute increases in pain in locations of the osteochondromas. Biopsy and pathological samples are not always recommended in order to determine the malignancy of the tumors as it is difficult to differentiate an osteochondroma from a low-grade osteosarcoma. For this purpose, gammagraphical studies are best to accurately determine dysplasia transformation [7].

Treatment of asymptomatic benign lesions includes pain killers and gammagraphic control. When the chondromas become symptomatic or there are signs of malignant transformation, then surgery may be proposed. Surgical procedures include curettage resection of the lesion and usage

of tissue grafts in order to compensate anatomic defects. Radio or chemotherapy are not efficient with these tumors as many malignancies involve low grade and well differentiated cell lines.

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