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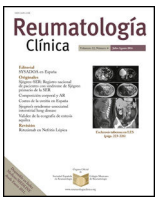

Case Report

Radiological features of crystal-induced arthropathy associated with hereditary hemochromatosis with homozygous C282Y mutation☆

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ABSTRACT

I present a clinical case of a 64-year-old male patient with hemochromatosis (homozygous C282Y) and crystal induced arthropathy showing the most common radiological features found in this metabolic disorder and the differences that may exist when compared to other primary degenerative processes or other inflammatory pathologies.

Características radiológicas de la artropatía microcristalina asociada a hemocromatosis hereditaria con mutación homocigota C282Y

Palabras clave:
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RESUMEN

Se expone el caso clínico de un paciente varón de 64 años con hemocromatosis (homocigoto C282Y) y artropatía microcristalina mostrando las características radiológicas más comunes que se encuentran en este trastorno metabólico y las diferencias que pueden existir al compararla con otros procesos degenerativos primarios u otras patologías inflamatorias.

Introduction

Haemochromatosis is a metabolic disorder characterised by elevated serum iron levels in various organs; the homozygous C282Y mutation is the most frequently detected. Joint involvement is common and forms part of the differential diagnosis of crystal-induced arthritis.

Clinical case

A 64-year-old patient with a personal history of haemochromatosis and homozygous C282Y genetic testing (since 2010), total thyroidectomy (1993), as complications he had undergone periodic phlebotomy (last in 2012), left hepatectomy (2014) due to a hepatocarcinoma of 1.8 cm in segment III with no significant portal hypertension, currently in complete remission and Child–Pugh A cirrhosis in the functional stage. Referred for rheumatology consultation to study migratory inflammatory arthralgia in the small joints of the hands, knees and ankles. Physical examination revealed painful limitation on flexo-extension of the metacarpophalangeal joints, and proximal interphalangeal joints of 2nd, 3rd and 4th finger bilaterally. Non-painful limitation on flexo-extension of the left ankle with no associated tenosynovitis, and limitation in the last degrees of flexion in both knees with bilateral patellar rubbing, with no frank synovitis, therefore it was

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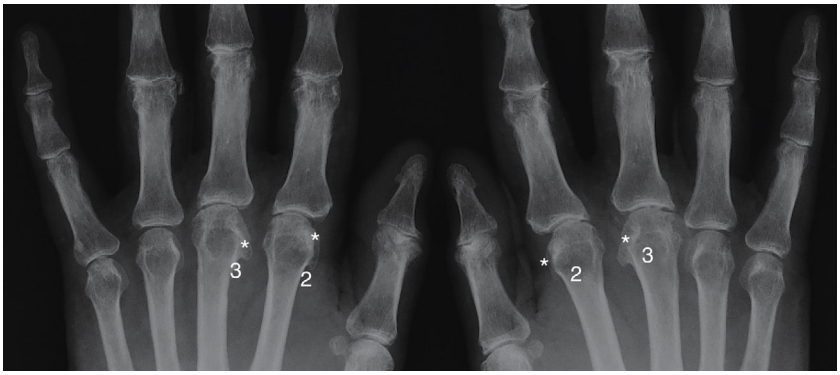

Fig. 1. AP X-ray of the hands showing severe degenerative changes in a patient with hereditary haemochromatosis. It can be observed that the changes are more aggressive in the 2nd and 3rd metacarpophalangeal joints bilaterally with reduced joint space, subchondral sclerosis, and hook-like osteophytes in the radial area (*).

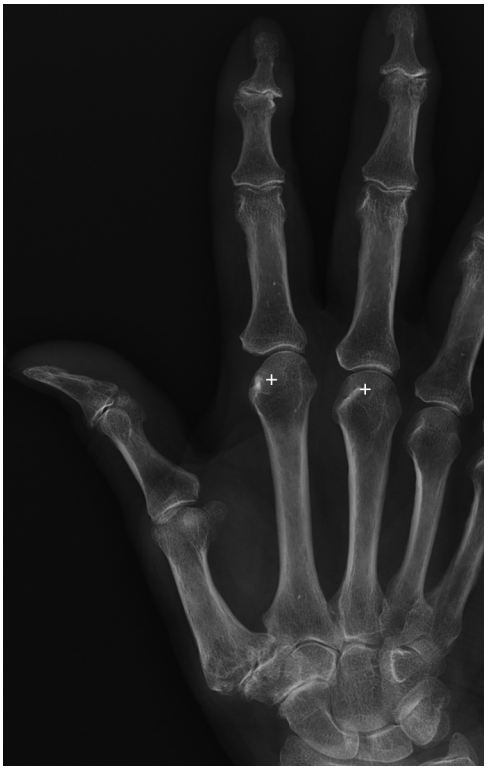

Fig. 2. AP X-ray of the hand of a patient with osteoarthritis, without haemochromatosis. As an example for comparison, neither involvement of the metacarpophalangeal joints nor the formation of hook-like osteophytes (+) can be observed.

not possible to test the synovial fluid. Worthy of note from the blood tests were Hb 14.8 g/dl, total leukocytes 6310, platelets 128 000/mm^3, prothrombin time 58%, ESR 30 mm, CRP <.3, glucose 99 mg/dl, GOT/GPT 11/25 U/L, GGT/FA 17/60 U/L, iron 69 μg/dl, ferritin 108 ng/ml, magnesium 2 mg/dl. PTH, vitamin D 25 EtOH within normal limits. Autoimmune study: ANA absent, rheumatoid factor negative, ACPA negative.

The radiological study showed more severe degenerative changes in the 2nd and 3rd metacarpophalangeal joints (with reduction of the joint space, subchondral cysts, and characteristically the formation of hook-like osteophytes appearing on radial face of these joints (Fig. 1). A radiological comparison of the metacarpal-phalangeal joints of a patient diagnosed with primary osteoarthritis is shown (Fig. 2). Degenerative changes can be seen in the knees (reduced joint space predominantly in the inner compartment, subchondral sclerosis, marginal osteophytes and chondrocalcinosis) (Fig. 3), and more severe changes in the left tibiotalar joint (Fig. 4).

The clinical diagnosis was crystal-induced arthropathy associated with haemochromatosis and osteoarthritis. The flare-ups of joint inflammation were treated with low doses of colchicine (.5 mg)/24 h and methylprednisolone, since non-steroidal anti-inflammatory drugs were contraindicated for this patient. After the liver surgery he had no further symptoms of inflammation although the mechanical arthralgia persisted in the hands and ankles.

Discussion

There are various studies that attempt to demonstrate radiological and clinical differences in these patients compared to primary osteoarthritis.[1–3] Conventional radiology is the standard

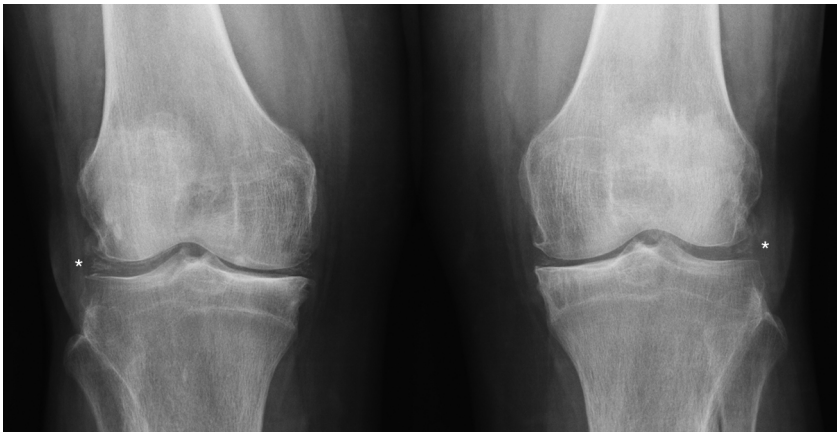

Fig. 3. AP X-ray of the knees. Showing radiological chondrocalcinosis (*), reduced joint space, subchondral sclerosis and marginal osteophytes.

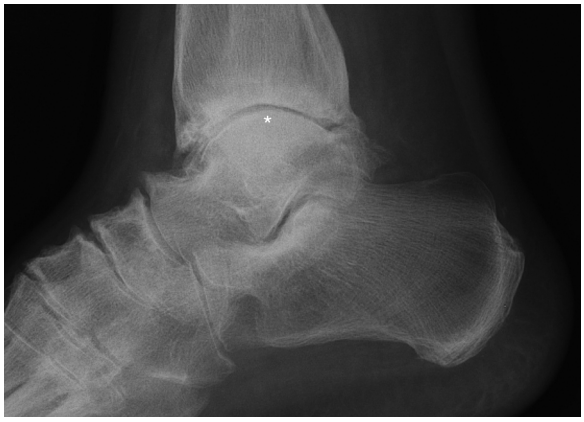

Fig. 4. Lateral X-ray of left ankle. Severe joint impingement and marginal osteophytes (*).

technique,[4] a radiographic scoring system specific to haemochromatosis has also been proposed.[5] The radiological features show greater involvement of the 2nd and 3rd metacarpophalangeal joints, and hook-like osteophytes in the radial area of these joints are usually observed.[1,2,4,5] They have a greater tendency to symmetrical presentation in the hands (which distinguishes the condition from other degenerative processes), and marginal erosion can appear. Chondrocalcinosis presents in 20%–50% of the cases, and can be observed more frequently in the wrist, knee and ankle. The degenerative radiological changes are more severe, the tibiotalar joint is one of the most affected,[4] in fact it has been confirmed that patients with C282Y homozygous haemochromatosis are more likely to require joint replacement.[2] With regard to treatment, colchicine has been demonstrated as effective for patients with radiological chondrocalcinosis, and recurrent joint inflammation.[4]

Conflict of interests

The author has no conflict of interests to declare.

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