Reumatol Clin. 2013;9(3):181–185

Reumatología Clínica

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Case Report

Large Joint Destructive Arthropathy and Tumoral Calcinosis Associated to Primary Oxalosis: Case Report and Literature Review☆

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ARTICLE INFO

Article history:
Received 21 February 2012
Accepted 19 September 2012

Keywords:
Tumoral calcinosis
Arthropathy
Primary hyperoxaluria
Bone oxalosis

ABSTRACT

A case of destructive arthropathy of hips and shoulders with tumoral calcinosis associated with calcium oxalate deposits in a patient with primary oxalosis and end stage renal disease on hemodialysis.

Artropatía destructiva de grandes articulaciones y calcinosis tumoral asociada a oxalosis primaria: reporte de un caso y revisión de la literatura

Palabras clave:
Calcinosis tumoral
Artropatía
Hiperoxaluria primaria
Oxalosis ósea

RESUMEN

Se presenta el caso de artropatía destructiva de caderas y hombros con calcinosis tumoral asociada a depósitos de oxalato de calcio en un paciente con oxalosis primaria e insuficiencia renal terminal en hemodiálisis.

Introduction

Oxalate crystal deposition disease is a rare condition seen primarily in patients with primary hyperoxaluria (PHO) and oxalate nephropathy and forms of secondary oxalosis in patients with ESRD on chronic dialysis, oxalate supplementation, thiamine deficiency and pyridoxine or oxalate formation due to Aspergillus niger. Musculoskeletal manifestations of calcium oxalate deposition disease are similar to those presented in calcium pyrophosphate crystals arthropathies. We report the case of a patient with arthropathy and tumoral calcinosis and associated deposits of calcium oxalate.

Case Description

The case is a 22-year-old Latin American Mestizo O positive male, with no toxic habits. The patient had no family history of kidney stones. He was diagnosed with short stature at age 6. At age 8 he presented repeated episodes of kidney stones and at 10 was diagnosed with primary hyperoxaluria, with progression to ESRD. At 12 he underwent renal replacement therapy with peritoneal dialysis and at 13 underwent living donor transplantation with graft loss after 5 months, so hemodialysis was started at a rate of 3 sessions a week and has remained so since then. 2 years after starting hemodialysis he presented bone pain and carpal, metacarpophalangeal and bilateral knee symmetrical and additive arthritis. At 16 he had a pathological fracture of the right femur, which required open reduction and internal fixation. The bone pain and polyarthritis followed a progressive course, with no response to treatment with non steroidal anti-inflammatory drugs (NSAID), and the patient noted the development of tumors located in soft tissue, which prevented gait at age 19. The symptoms persisted

☆ Please cite this article as: Horta-Baas G, et al. Artropatía destructiva de grandes articulaciones y calcinosis tumoral asociada a oxalosis primaria: reporte de un caso y revisión de la literatura. Reumatol Clin. 2013;9:181–5.

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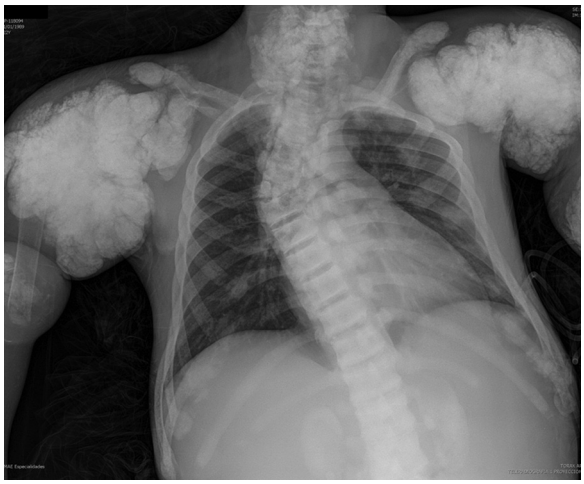

Fig. 1. Nodular calcifications of the shoulders. Superior and inferior vertebral plate osteosclerosis (rugby jersey spine) and vertebral collapse of dorsal vertebrae 5 and 6. Bulbous growth on the ends of the ribs and clavicles. Osteosclerosis of clavicles and rib fracture sequelae. Fracture of the right humerus.

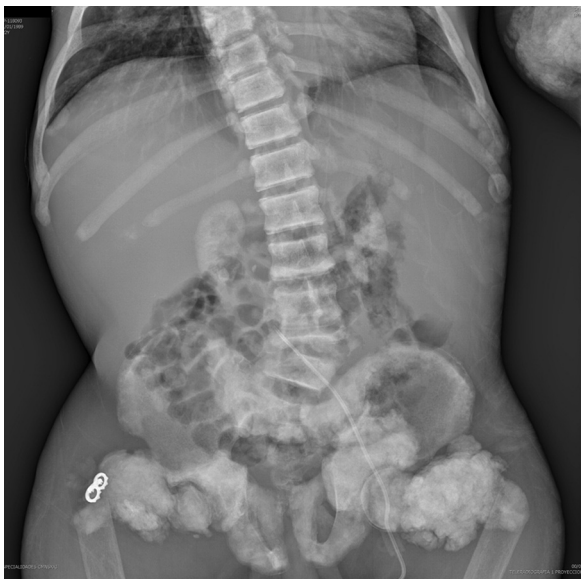

Fig. 2. Bilateral nephrocalcinosis, vertebral osteoscleorosis (rugby jersey spine), osteosclerosis in Paget-like pelvic bones. Osteomalacia. Nodular calcifications. Bilateral subtrochanteric fracture. Surgical material at the proximal end of the right femur.

despite infiltration with glucocorticoids, low-dose oral steroids, opioid analgesics and NSAID. During the patients latest assessment we found that the patient had low height, pectus carinatum, short limbs, wrist and ankle subluxation and periarticular tumors located on shoulders, hips and knees, at the expense of soft tissue.

Laboratory studies showed: Vitamin A 7.1 ng/ml (low <10), ferritin 1342 ng/ml (30–400), cortisol 7.87 mg/dl (5–25) parathyroid hormone (PTH) 4.08 pg/ml (10–65), iron saturation percentage of 101% (15–55), iron binding capacity without saturating 63 g/dl (250–450), iron 64 mg/dl (50–170 mg/dl), creatinine 3.4 mg/dl (0.4–1.2), BUN 36.45 mg/dl (5–23), glucose 83 mg/dl, calcium 9.7 mg/dl (8.4–10.2), phosphorous 5.1 mg/dl (2.7–4.5), magnesium 2.4 mg/dl (1.6–2.6), alkaline phosphatase, 296 U/l (40–129) LDH 507 U/l (240–480), gamma glutamyl transferase 640 U/l (10–71), alanine aminotransferase 23 U/l (2–41), aspartate transaminase 50 U/l (2–38), albumin 2.3 g/dl (3.4–4.8) and uric acid 6.5 mg/dl (2.4–7).

An abdominal ultrasound showed both kidneys to be hypoplastic with increased echogenicity and renal calcifications, with hepatic and spleen enlargement and a normal pancreas. An abdominopelvic computed tomography showed nephrocalcinosis, hepatomegaly, splenomegaly, arteriosclerosis and, osteosclerosis of vertebral and pelvic bones.

A transthoracic echocardiogram showed a systolic pressure of 61 mmHg in the pulmonary artery, normal left ventricular diameter, a thickened wall, normal mobility, mild dilated right chambers, valvular sclerosis, mild mitral regurgitation and moderate tricuspid regurgitation.

The chest X-ray (Fig. 1) demonstrated nodular periarticular calcification in the shoulders, dorsal vertebral osteosclerosis and vertebral collapse of thoracic vertebrae 5 and 6, with sequelae of rib fractures and a fracture of the right humerus. An abdominal X-ray (Fig. 2) showed bilateral nephrocalcinosis, vertebral osteosclerosis which also affected pelvic bones, bilateral nodular calcifications and a subtrochanteric fracture.

Discussion

PHO is an autosomal recessive inborn error of metabolism leading to an enzyme deficiency of alanine-glyoxylate aminotransferase in hepatic peroxisomes. The enzyme deficiency causes an overproduction of oxalate which is eliminated by the kidneys and precipitates forming crystals that are deposited in various tissues. PHO diagnosis is performed before the age of 5 in 65% of cases. The main cause of death is uremia, which in 80% of cases occurs before age 20.[1]

Since oxalate is eliminated through the kidney, this is the first and primary target organ, leading to the appearance of repeated stone formation in the first decades of life, nephrocalcinosis and early renal failure.[2] When terminal renal failure occurs and oxalic acid cannot be excreted, rapidly evolving tissue deposits develop particularly in the kidneys and skeleton.[2,3] Bones are one of the main affected organs, with unusually serious lesions having been described, especially in patients with chronic renal failure on dialysis.[4] Oxalate deposits and the surrounding granulomatous reaction induce lesions similar to secondary hiperparatiroidism which are particularly serious.[4]

The pattern of joint involvement is more commonly acute or chronic symmetrical polyarthritis or oligoarthritis, with involvement of the metacarpophalangeal and proximal interphalangeal joints, with or without tenosynovitis, along with miliary or cottony skin and finger arterial calcifications[5,6] (Fig. 3), but can occur in other joints, such as knees, elbows, ankles and the first metatarsophalangeal joint. In autopsy studies, calcium oxalate deposits in joint tissue and bone oxalosis occur in approximately 90% of patients with renal insufficiency undergoing chronic hemodialysis.[6]

El Hage et al. conducted a review of 12 consecutive patients with type 1 PHO, all with renal involvement, 4 with ESRD undergoing dyalisis.[7] The main symptom was bone pain and was present in only 4 of the severely involved patients and appeared in the second year of dialysis. The 2 most severely affected patients had evidence of pathologic fractures.[7]

Kidney damage is usually the result of a combination of nephrolithiasis, secondary nephrocalcinosis and interstitial fibrosis. Renal failure is associated with the rapid deposit of the crystals in the kidney, myocardium, skin, blood vessels and bones; when the glomerular filtration rate decreases below 30–40 ml/min/1.73 m^2, oxalate cannot be efficiently excreted by the kidneys and reaches saturation levels.[8] Oxalate saturation depends on serum levels and these are inversely related to the glomerular filtration rate.[9] This saturation, which occurs almost universally in the serum of patients with terminal uremic PHO, causes the systemic oxalosis affecting