

## Primary hyperoxaluria

### Primer hiperoksalüri

Elçin Aydın<sup>1</sup> Tülay Öztürk Atasoy<sup>2</sup> Mehmet Argın<sup>3</sup>

<sup>1</sup>Başkent University Faculty of Medicine, Department of Radiology, İzmir, Turkey

<sup>2</sup>Ümraniye Training and Research Hospital, Clinic of Radiology, İstanbul, Turkey

<sup>3</sup>Ege University Faculty of Medicine, Department of Radiology, İzmir, Turkey

#### Abstract

Hyperoxaluria is characterized by nephrolithiasis and nephrocalcinosis caused by supersaturation of calcium oxalate in the urine. Deposits of calcium oxalate can lead to kidney damage, kidney failure, and injury to other organs. Herein, we report a case of primary hyperoxaluria which is a serious though rare condition, can be suspected on the basis of the renal features. Radiological skeletal changes, which are also rather specific, may also share some features with renal osteodystrophy.

**Keywords:** Hyperoxaluria, nephrolithiasis, nephrocalcinosis.

#### Öz

*Hiperoksalüri nefrolitiazis ve nefrokalsinozis ile karakterize, idrardaki kalsiyum oksalatın süpersatürasyonu sonucu oluşur. Kalsiyum oksalat depozitleri böbrek hasarı, böbrek yetmezliği ve diğer organlarda hasarlanmaya neden olabilir. Burada nadir de olsa ciddi bir durum arz eden, renal özellikleri açısından şüphelenilebilen primer hiperoksalüri vakasını sunduk. Radyolojik iskelet değişiklikleri oldukça spesifik olmakla birlikte renal osteodistrofi ile benzer özellikler taşıyabilir.*

**Anahtar Sözcükler:** Hiperoksalüri, nefrolitiazis, nefrokalsinozis.

#### Introduction

Primary hyperoxaluria is a rare condition that causes kidney stones by supersaturation of calcium oxalate in the urine. Deposits of calcium oxalate can lead to kidney damage, kidney failure, and injury to other organs. Primary hyperoxaluria is caused by the deficiency of an enzyme that normally prevents the buildup of oxalate. There are two types of primary hyperoxaluria, distinguished by the enzyme that is deficient. Primary hyperoxaluria is an autosomal recessive disorder and causing urinary precipitation of calcium oxalate, resulting in urolithiasis and often nephrocalcinosis. Progressive renal insufficiency with uremia frequently occurs in childhood or early adulthood. Early diagnosis and conservative treatment can improve long term prognosis of patient (1).

#### Case Report

A 21 year old male patient with a five year history of end stage renal insufficiency was in the kidney transplantation plan. The laboratory tests revealed high liver enzymes. The abdominal ultrasonography was normal except for the small and hyperechoic kidneys. The urine proline, citrulline and oxalic acid levels were high. Plain radiographs of the extremities and axial skeleton were obtained for further evaluation. Plain radiographs revealed diffuse sclerotic densities of the bony structures. There were diffuse multiple lytic lesions with ill-defined borders at the diaphysis of both proximal femurs (Figure 1a-d).

Written informed consent was obtained from the patient for publishing the individual medical records.

#### Discussion

Hyperoxaluria is characterized by nephrolithiasis and nephrocalcinosis caused by supersaturation of calcium oxalate in the urine. Primary hyperoxaluria type 1 and 2 (PH1 and PH 2) are rare autosomal recessive disorders with defective glyoxalate metabolism in the liver resulting in increased oxalate production (1). Secondary hyperoxaluria is due to reduced excretion, excessive

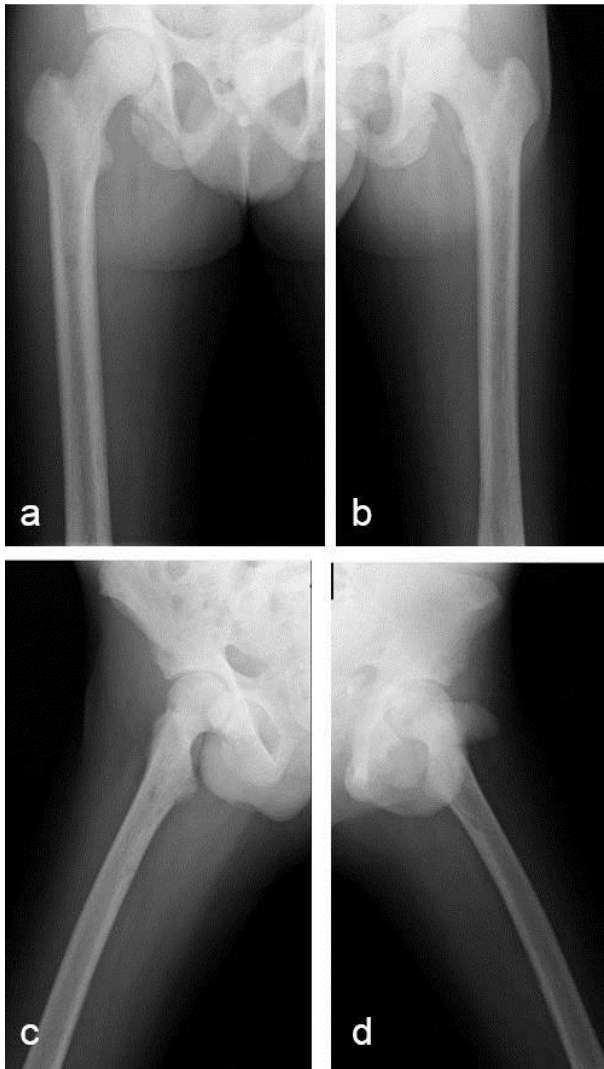
Corresponding Author: Elçin Aydın

Başkent University Faculty of Medicine, Department of Radiology, İzmir, Turkey

Received: 15.07.2016

Accepted: 08.09.2016

dietary intake or increased gut absorption of oxalate (2). Idiopathic hyperoxaluria has no known associated gene defect. In PH1 hepatic deficiency or mistargeting of alanine glyoxylate aminotransferase (AGT) leads to loss of glyoxylate amination to glycine. The accumulated glyoxylate is oxidized to oxalate which cannot be further metabolized and is therefore mainly excreted by the kidney (hyperoxaluria).



**Figure-1. a-d.** Diffuse multiple lytic lesions with an ill-defined border at the both of the proximal femur.

This leads urinary precipitation of calcium oxalate, resulting in urolithiasis and often nephrocalcinosis. Progressive renal insufficiency with uremia frequently occurs in childhood or early adulthood. Although early

diagnosis and conservative treatment can improve long term prognosis of patients (3,4). If end stage renal disease is reached the only curative therapy of PH1 remains combined liver kidney transplantation (5). Preemptive liver transplantation can also cure the metabolic defect in PH1 and may prevent oxalate deposition if performed early enough (6). With declining renal function, high blood levels of oxalate result in deposition of oxalate crystals in other organs, such as bone and bone marrow. Bone tissue is a common site of calcium oxalate crystal deposition but the radiologic appearance of bone involvement has been described in very few reports (7). Moreover the respective role of oxalate crystal deposition and uremic hyperparathyroidism in the genesis of bone lesions is undefined. The earliest specific radiographic skeletal features of oxalosis are fine transverse lines of increased bone density, located symmetrically in the areas of rapid growth, presumably related to crystal precipitation in the site where cartilage normally calcifies (8). These findings are most prominent in areas of hematopoietic rather than fatty marrow and other highly vascularized areas, where blood oxalic acid levels are greater. Increased bone density may also involve the upper and lower end plate of the vertebral bodies. Another manifestation of skeletal oxalate deposition is the radiopaque rim or alternating lucent and dense bands that can be observed in flat bones, mainly of the pelvic girdle, epiphyseal nuclei (target-like epiphyses), tarsal bones, and patella, with the narrow radiolucent rim representing the less involved bone. Precipitation of oxalate crystals in the marrow spaces induces chronic inflammatory granulomatous response, with giant and mononucleated cells around the clusters of crystals, followed by bone resorption. It is difficult to distinguish the changes due to oxalate deposition from that cause by secondary hyperparathyroidism. Diffuse demineralization, "ruger jersey" bone structure, thinned cortices, and subperiosteal bone resorption are reasonably attributed to hyperparathyroidism, but some bone resorption may also be caused by granulomatous reaction around clusters of oxalate crystals, and pathological fractures may occur because of both conditions (9). The diagnosis of PH1 with systemic oxalosis, which is a serious though rare condition, can be suspected on the basis of the renal features. Radiological skeletal changes, which are also rather specific, may also share some features with renal osteodystrophy.

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