Eur J Pediatr (1998) 157: 255–256

YOUR DIAGNOSIS?

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An 8-year-old boy with renal failure, nephrolithiasis and bone pain

Received: 30 January 1997
Accepted: 5 July 1997

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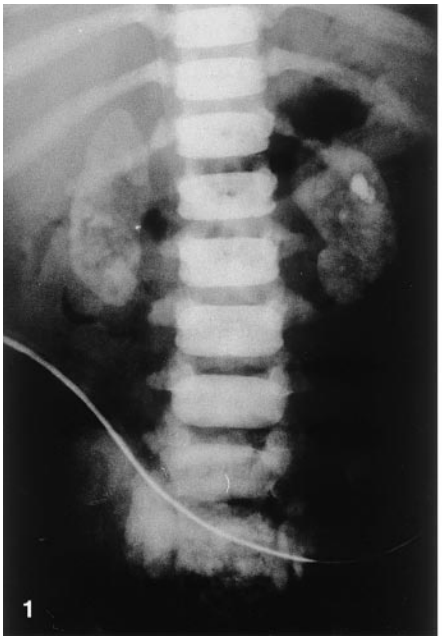
Information

An 8-year-old boy with renal failure and nephrolithiasis was referred from a local hospital for the evaluation of bone pain. He had undergone repeated surgical procedures for the removal of renal and ureteric stones since 3 years of age but his renal function progressively declined and he developed irreversible renal failure. He was maintained on haemodialysis in the local hospital for 2 years during which time his clinical condition progressively deteriorated. Severe bone pain set in soon after and he experienced difficulty in walking. Because of social problems, dialysis therapy was discontinued 10 months ago. Since then, he is at home without any therapy and is immobilized. He is the fourth child of first degree cousins and his 4-year-old brother had surgery for a bladder stone 2 years ago. On admission, he had the physical features of uraemia with oedema, foul breath and pallor. His height and weight were below the 3rd percentile. Height standard deviation score was −5.4. Laboratory tests revealed anaemia and renal failure. Haemoglobin concentration was 5.6 g/dl. BUN and creatinine values were elevated to 148 mg/dl and 6.9 mg/dl, respectively. Creatinine clearance was 7.5 ml/1.73 m^2 per minute Serum calcium was 9.4 mg/dl, phosphorus 8.4 mg/dl, alkaline phosphatase 114 IU/l (normal range 98–278 IU/l), parathyroid hormone 39.9 pg/ml (12–72 pg/ml).

Abdominal radiography revealed bilateral nephrocalcinosis and multiple renal stones in the left kidney. Increased radiodensity in the vertebral bodies was also noted (Fig. 1). The capital femoral epiphyses were displaced and increased density was found at the proximal and distal ends of the long bones with radiolucent areas in the metaphysis (Fig. 2).



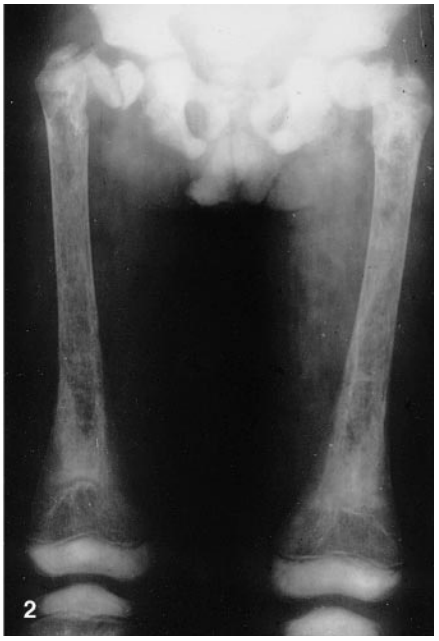


Fig. 1 Abdominal X-ray. Nephrocalcinosis, nephrolithiasis (left kidney) and increased radiodensity in the vertebral bodies

Fig. 2 Increased density at the proximal and distal ends of the long bones with radiolucent areas in the metaphyses and displacement of the capital femoral epiphyses

Diagnosis: Systemic oxalosis induced bone disease

Iliac crest bone biopsy demonstrated refractile crystals surrounded by a marked giant cell reaction. These crystals were birefringent under polarized light, typical for calcium oxalate (Fig. 3).

Discussion

The association of nephrocalcinosis, recurrent urolithiasis and renal failure in children strongly suggests primary hyperoxaluria type 1. The disease is a rare autosomal recessive disorder caused by deficiency of the liver specific peroxisomal enzyme alanine-glyoxylate amino transferase encoded on the long arm of chromosome 2 [4]. Increased synthesis of oxalate and glycollate in the liver leads to the biochemical hallmarks of primary hyperoxaluria type 1. Renal features are generally the first and the main manifestation of the disease. Deteriorating renal function diminishes the excretion of oxalate which leads to increased oxalate crystal deposition in extrarenal sites including bone, bone marrow, blood vessels, central nervous system, peripheral nerves, heart and retina, i.e. systemic oxalosis [2]. Nature and extent of the bone disease are related to the severity and duration of renal disease. Bone pain is an important clinical feature. The earliest radiographic sign of bone oxalosis is a fine radiodense transverse line at the zone of cartilage calcification caused by oxalate precipitation where cartilage normally calcifies [5]. Over a period of time more oxalate accumulates and this line becomes a broad band. Subperiostal cortical defects and large lucent areas different from renal osteodystrophy are important radiological manifestations of the disease [1, 5]. These lucencies and subperiostal cortical defects were reported as areas of oxalate deposition with giant cell destruction of bone [1].

Skeletal problems become more prominent with dialysis which does not prevent crystal accumulation [1, 5]. Combined liver and kidney transplantation should be considered before extrarenal complications occur [3].

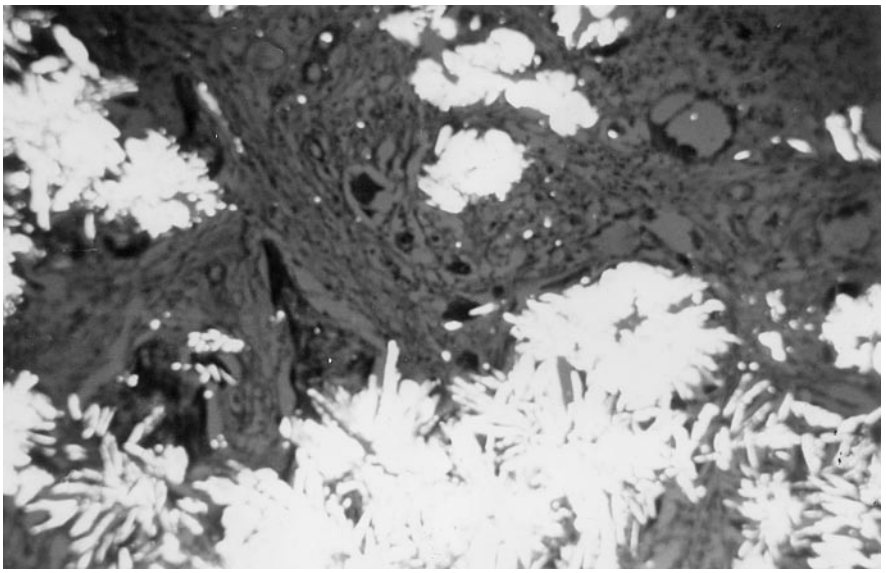


Fig. 3 A high-power photomicrograph of birefringent oxalate crystals in bone under polarized light

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