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Case Report

Primary hyperoxaluria: A case report

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ABSTRACT

Renal failure can occasionally be caused by oxalate nephropathy. Glyoxylate metabolism abnormalities and particular hepatic enzyme deficits are the causes of primary hyperoxaluria (PH). Increased intestinal absorption, an excessive diet or an increased intake of oxalate precursors can all result in secondary hyperoxaluria. A 13-month old male child with high blood creatinine, low sodium, low calcium levels, high uric acid and low urine specific gravity is the subject of this research. Medullary papillary calcification was detected using sonography (nephrocalcinosis). Calcium oxalate crystals, sparse lymphocytic infiltration and interstitial fibrosis were seen on a renal biopsy. The patient was put on peritoneal dialysis, progresses to anuria and expired due to renal failure.

1. Introduction

A significant metabolic condition is hyperoxaluria can be primary or secondary. The precise hepatic enzyme deficits that lead to the autosomal recessive hereditary forms of Primary Hyperoxaluria (PH) types I, II, and III result in problems in glyoxylate metabolism. Oxalate crystals build up in the kidneys, causing renal tubular epithelial cells to become damaged and inflamed. This leads to decreased oxalate clearance and the systemic accumulation of calcium oxalate crystals.[1] Most often, secondary hyperoxaluria results from gastrointestinal conditions that enhance intestinal absorption. Other uncommon reasons include chronic pancreatitis, vitamin C, ethylene glycol, consumption of foods rich in oxalate and side effects from a few prescription drugs.[2]

The present case of idiopathic calcium oxalate nephropathy is an infrequent. Awareness of PH and measurement of plasma and urinary oxalate is important. Because early diagnosis, molecular sub typing and early prompt initiation of treatment are important to significantly improve outcome.

The infantile form is more severe and do not provoke kidney stones. “Infantile oxalosis” patient presents with failure to thrive, anaemia, and acidosis.

2. Materials and Methods

Renal biopsy was performed. Routine stains used were hematoxylin and eosin. Polarization microscopy was employed for the detection of crystalline structures and amyloid.

3. Case Presentation

13-months old male child complaint of fever and vomiting for 7 days, generalised edema all over body with breathlessness in the past 3 days. Severe pallor (Haemoglobin-4.5), BP-148/82 mmhg, haematuria.

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Laboratory tests revealed: sodium 122 mEq/L, potassium 4.1 mEq/L, blood urea nitrogen 7 mg/dl, creatinine 8.2 mg/dl, calcium 9 mg/dl, phosphorus 4.9 mg/dl, uric acid 6.7 mg/dl and lactate dehydrogenase 544 IU/ml. ANA, C3, C4, CH50 and anti-ds DNA were within normal limits.

Complete blood count (CBC) showed hemoglobin 4.5 g/dl, haematocrit 13.8%; mean corpuscular volume (MCV): 67.7 fl, Mean Corpuscular Hemoglobin (MCH) 21.9 pg, MCHC 33 g/dl, and platelet 226 $\times$ 10^3 /μL. Urinalysis revealed specific gravity: 1.010, PH: 5, white blood count (WBC): 12 to 16/HPF, red blood count (RBC): 10-15 /HPF, and protein: positive.

Sonography revealed calcification of the medullary papilla along with both kidneys enhanced echo pattern and differentiation (nephrocalcinosis).

The parents declined genetic testing for their child. The patient subsequently progressed to anuria and was put on peritoneal dialysis despite supportive therapy he died of acute renal failure.

Light microscopy: The patient underwent kidney biopsy, which revealed atrophic tubules, and many intra-tubular and interstitial crystals that seemed to be calcium oxalate crystals (Figure 1). The interstitium had moderate fibrosis and scattered lymphocytic infiltration. There is acute tubular injury with sloughing, vacuolization of proximal tubular cells, and flattened, simplified epithelium with frequent intratubular crystals, which are clear on haematoxylin and eosin stain under standard light microscopy and show multi-coloured birefringence under polarized light, with classic fan-shaped morphology (Figure 2).

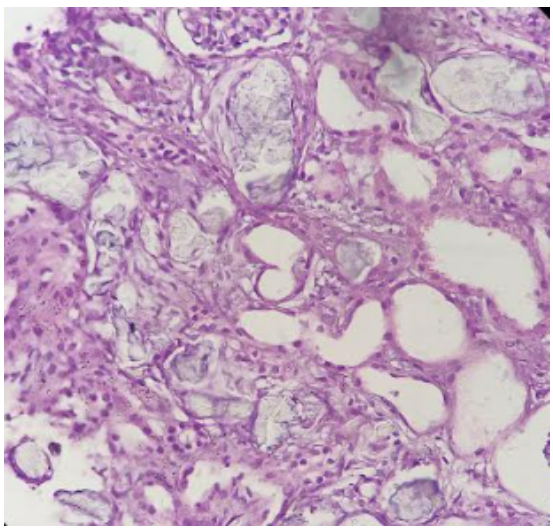

Fig. 1: H & E (40 X) Renal biopsy showing intra-tubular and interstitial crystals

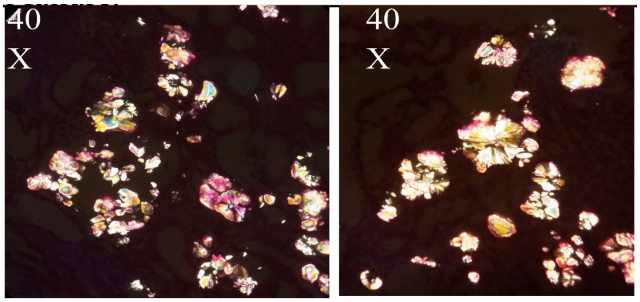

Fig. 2: Under polarised light microscopy, oxalate crystal shows multi-coloured birefringence with classic fan shaped morphology

4. Discussion

Systemic oxalosis, extremely increased urine oxalate, recurrent urolithiasis, and/or progressive nephrocalcinosis are all symptoms of the uncommon autosomal recessive illness known as primary hyperoxaluria. These conditions eventually lead to end-stage renal failure (ESRD).[3]

The most prevalent and severe form of primary hyperoxaluria, type I, which presents in childhood as recurring kidney stones and nephrocalcinosis, is present in 70% to 80% of cases. Compared to Primary Hyperoxaluria-I, Primary Hyperoxaluria-II is less frequent (around 10%), Primary Hyperoxaluria-II has lower urine oxalate levels and has better results. Compared to patients with the other two categories, those with PH-III had a lower risk of developing ESRD.[4]

Calcium phosphate crystals do not polarise but calcium oxalate crystals are gray-white, speculative, and birefringent in polarised light. In healthy or failing kidneys, as well as in transplanted kidneys, small or isolated tubular oxalate crystals are not uncommon; their presence does not indicate renal impairment. Contrarily, significant calcium oxalate deposits in the tubules or interstices are strongly symptomatic of a hyperoxaluric disease.[5,6]

The initial sign or symptom of an oxalate issue in a patient is typically haematuria or dysuria, urolithiasis, or infection. Failure to thrive, anaemia and acidosis are symptoms of renal failure in "infantile oxalosis." Most people begin to have symptoms before the age of 10^1. Until the disease is advanced, the diagnosis of PHs and their many clinical manifestations may be difficult to make. Early intervention can delay ESRD or possibly stop it. The chosen technique of transplantation depends on the type of PH; in PH-I, a combined liver and kidney transplant is favoured, whereas in PH-II, an isolated kidney transplant is the preferred procedure.[7] Childhood or adolescent calcium oxalate stone development, as well as recurring or numerous stones or nephrocalcinosis in individuals with renal failure, are crucial diagnostic indicators that call for metabolic screening and, if necessary, specialised diagnostic tests.[7]

5. Source of Funding

None.

6. Conflict of Interests

The authors had no conflict of interest to disclose.

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