Case 10766

Eurorad

Cortical nephrocalcinosis in a case of type-1 primary oxaluria

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Section: Paediatric radiology
Area of Interest: Kidney
Procedure: Screening
Imaging Technique: Ultrasound-Colour Doppler
Special Focus: Transplantation Case Type: Clinical Cases
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Patient: 15 years, male

Clinical History:

Bladder calculi at the age of 3, removed by cystolithotomy. Vomiting and pedal oedema for the last nine months, and has been having dialysis for the last last three months. Serum uric acid and phosphorus-normal, mild hypocalcaemia. Plasma oxalate elevated, plasma oxalate to creatinine ratio elevated.
Imaging Findings:

Abdominal ultrasound showed bilateral renal cortical hyperechogenicity with relative medullary sparing, significant posterior acoustic shadowing with loss of cortico-medullary differentiation and poor paranchymal vascularity (Fig. 1, 2), suggestive of cortical nephrocalcinosis in a background of end-stage renal disease.
Discussion:

Background:

Type 1 primary hyperoxaluria is a rare autosomal recessive genetic disease caused by a deficient liver enzyme; alanine–glyoxylate aminotransferase. This disorder is characterised by excessive synthesis of oxalic acid (oxalate) and urinary excretion of both oxalate and glycolate. An abnormal deposition of calcium oxalate can occur in bones, joints, nerves, heart, vessels, skin, retinae and kidneys [1]. In the urinary tract, oxalate is a metabolite excreted by the kidney that induces the death of renal epithelial cells when it is present at excessive concentrations [2–5].

Clinical perspective:

Patient usually presents with recurrent renal or bladder calculi from a young age or with medical renal disease.

Imaging perspective:

The sonographic pattern of cortical nephrocalcinosis is quite specific, with diffuse markedly hyperechoic peripheral renal cortex with possible global acoustic shadowing and a lack of corticomedullary differentiation.

However, the final diagnosis of type 1 primary hyperoxaluria can be approached by analysis of plasma oxalate, urinary glycolate, and oxalate levels. An enzymatic study obtained from liver biopsy can quantify alanine–glyoxylate aminotransferase activity. An antenatal diagnosis is possible because of the molecular analysis of the different

mutations [6, 7]. In type 1 primary hyperoxaluria, marked hyperechogenicity is directly related to the amount of oxalate of calcium deposits.

Outcome:

Progressive deterioration in renal function resulting in end-stage renal disease (ESRD) would warrant dialysis and liver–kidney transplantation [8].

Differential Diagnosis List: Cortical nephrocalcinosis in a case of Type 1 primary hyperoxaluria, Cortical necrosis, Congenital nephritic syndrome, Renal toxicity (due to medication and infection), Haemolytic uraemic syndrome

Final Diagnosis: Cortical nephrocalcinosis in a case of Type 1 primary hyperoxaluria

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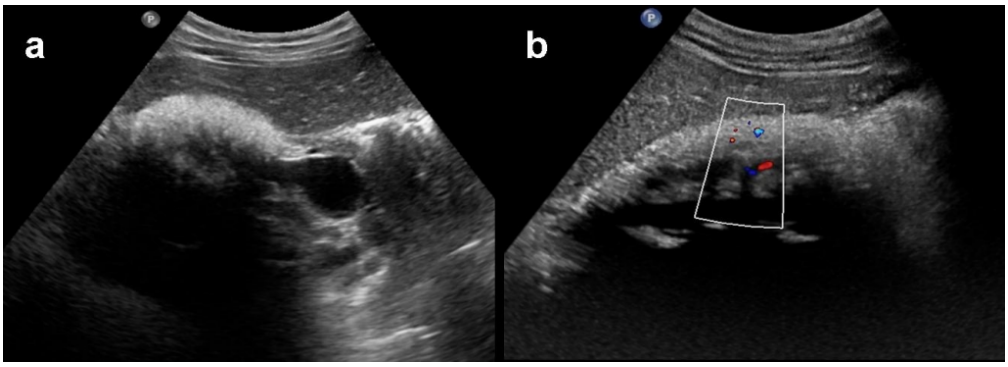
Figure 1

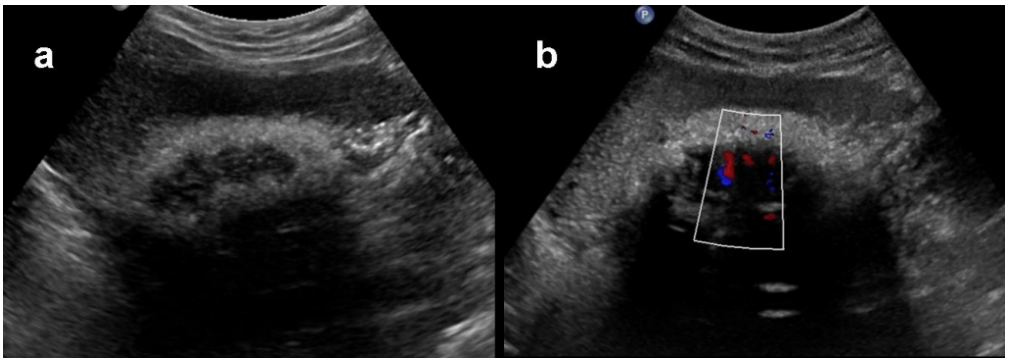
a



Description: (a) Axial sonogram of right kidney showing raised renal cortical echogenicity with relative medullary sparing and posterior acoustic shadowing; (b) Sagittal colour Doppler images with poor paranchymal colour uptake. **Origin:** Department of Radiology, Amrita Institute of Medical Science, Kochi, Kerala, India.

Figure 2

a



Description: (a) Sagittal sonogram of left kidney showing raised renal cortical echogenicity with relative medullary sparing, loss of cortico–medullary differentiation and posterior acoustic shadowing; (b) colour Doppler images with poor colour uptake. **Origin:** Department of Radiology, Amrita Institute of Medical Science, Kochi, Kerala, India.