Primary hyperoxaluria type I is a rare autosomal recessive metabolic disorder characterized by absence of liver specific peroxisomal alanine glyoxylate aminotransferase (AGT) leading to elevated urinary excretion of glycolate and oxalate [1]. Calcium oxalate has a very low solubility leading to nephrocalcinosis, urolithiasis and progressive renal insufficiency [2]. Due to the rarity of the disease, these specific diagnostic tests are not performed routinely unless the diagnosis is strongly suspected. We report a case of primary hyperoxaluria in which the diagnosis was not entertained because there was no past history of urinary calculi and no evidence of nephrocalcinosis on plain X-ray and ultrasonography. The disease was detected and diagnosed only after kidney transplantation.

A 19-year-old female presented in September 1993 with uraemic symptoms, severe anaemia and azotaemia (serum creatinine, 13.6 mg/dl). Routine urinalysis was unremarkable. Ultrasound of kidneys showed right kidney of 7.1 × 3.1 cm size and left kidney of 10.7 × 5.5 cm. Both kidneys showed increased echogenicity. The pelvicocalyceal system of left kidney appeared mildly dilated and a plain abdominal X-ray (Figure 1) revealed a radio-opaque density at upper pole of the left kidney. A micturating cystourethrogram did not show reflux. Cystoscopy with retrograde urogram showed no evidence of obstruction. A diagnosis of end-stage renal disease (ESRD) of unknown aetiology was made. Live related donor kidney transplant was performed. The graft function was excellent and creatinine reached 0.9 mg/dl on third post-transplant day. From the tenth day, there was gradual increase in serum creatinine. When creatinine rose to 1.4 mg/dl on 25th day, a kidney biopsy was performed which showed tubules filled with oxalate crystals (Figure 2). Over the next few days there was progressive decline in graft function due to recurrence of primary disease. A repeat abdominal X-ray (Figure 3) after the patient restarted dialysis, revealed dense nephrocalcinosis in the native kidneys. If this nephrocalcinosis had been obvious at the initial diagnosis of ESRD, renal transplantation would have been avoided.

This case highlights the fact that in young patients with ESRD of unknown origin with any evidence of renal calculi, oxalosis must be excluded.

References

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Fig. 1. Plain abdominal X-ray taken at the time of diagnosis of ESRD.

Fig. 2. Brightly refractile oxalate crystals in the tubular lumina under polarized light.

Fig. 3. Abdominal X-ray 3 months after graft failure, demonstrating nephrocalcinosis.