Primary Hyperoxaluria-Imaging of Renal Oxalosis

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Primary Hyperoxaluria is a rare autosomal recessive hereditary disorder due to deficient alanine-glyoxylate aminotransferase enzyme with defective glyoxylate metabolism leading to excessive oxalate production and deposition into the tissues (oxalosis). Deposition of excessive calcium oxalates in nephrons leads to crystallization (nephrocalcinosis) which increases risk for end-stage renal disease. We are presenting a case of primary hyperoxaluria type I confirmed with genetic studies. UROLOGY 134: e3–e4, 2019. © 2019 Elsevier Inc.

A 9-month-old male infant with poor weight gain was brought with end-stage renal disease (ESRD). An uncle and 2 nephews died of the same disease. Serum creatinine (640 μmol/L) and plasma oxalate were high (66.0 μmol/L). Pathogenic mutation of alanine-glyoxylate aminotransferase enzyme gene confirmed the diagnosis of primary hyperoxaluria type I (PH1). Ultrasound showed bilateral enlarged kidneys (~6.8 cm length) with increased parenchymal echogenicity and loss of corticomedullary differentiation (Fig. 1). Plain radiograph and noncontrast CT showed bilateral dense nephrocalcinosis (Figs. 2,3). While on hemodialysis (4 times/wk), he underwent liver transplantation and is awaiting renal transplantation.

PH1 is the most common and serious type of PH, a rare autosomal recessive hereditary disorder (prevalence of 1-3/million population).[1] Deficiency of alanine-glyoxylate aminotransferase causes defective glyoxylate metabolism, excessive oxalate production, and deposition (oxalosis).[2] Supersaturation of calcium oxalates in nephrons leads to crystallization and renal parenchymal deposition (nephrocalcinosis). Nephrocalcinosis increases risk for ESRD whereas nephrolithiasis is not significantly associated with ESRD.[3] Oxalosis occurs whenever serum oxalate is >30.0 μmol/L. Urine oxalate >45 mg/day is classical of PH. Imaging should include ultrasound, radiograph and CT.[4] Prenatal screening is essential for suspected PH. Treatment of choice in PH1 with ESRD is liver and kidney transplantation (combined or sequential).[5]

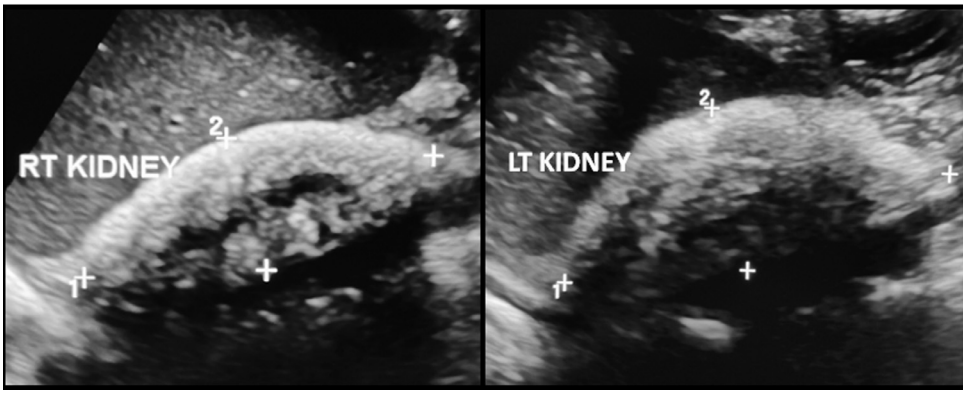

Figure 1. USG shows bilateral enlarged kidneys with increased parenchymal echogenicity and calcification with loss of corticomedullary differentiation. USG, ultrasound.

Compliance with ethical standards.

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Ethical approval (animals): *This article does not contain any studies with animals performed by any of the author(s).*

Ethical approval: *All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki Declaration and its later amendments or comparable ethical standards.*

Informed consent: *Informed consent was obtained from individual participant included in the study.*

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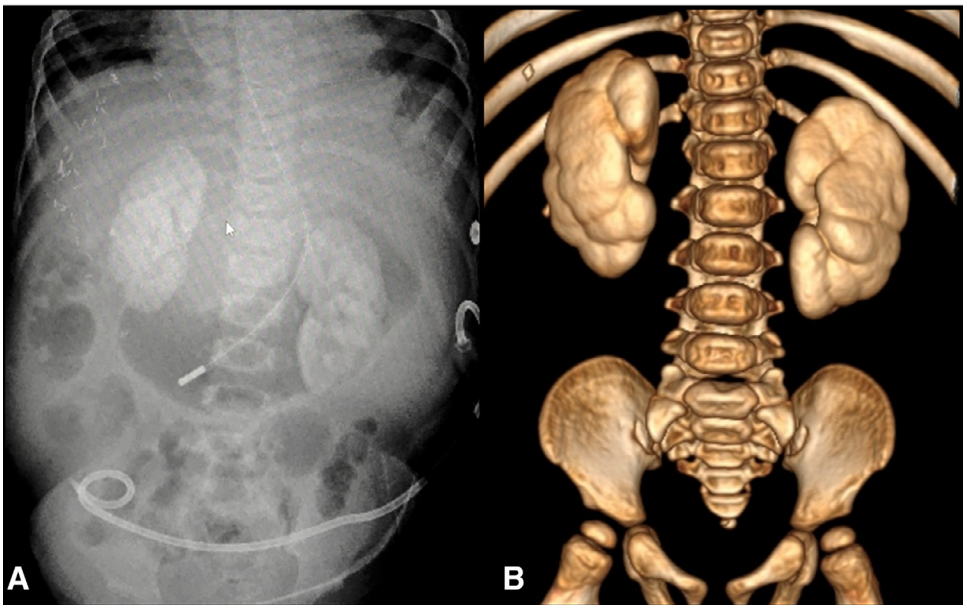

Figure 2. (**A**) Plain radiograph shows bilateral nephrocalcinosis; (**B**) Reconstructed coronal CT image of bilateral nephrocalcinosis.

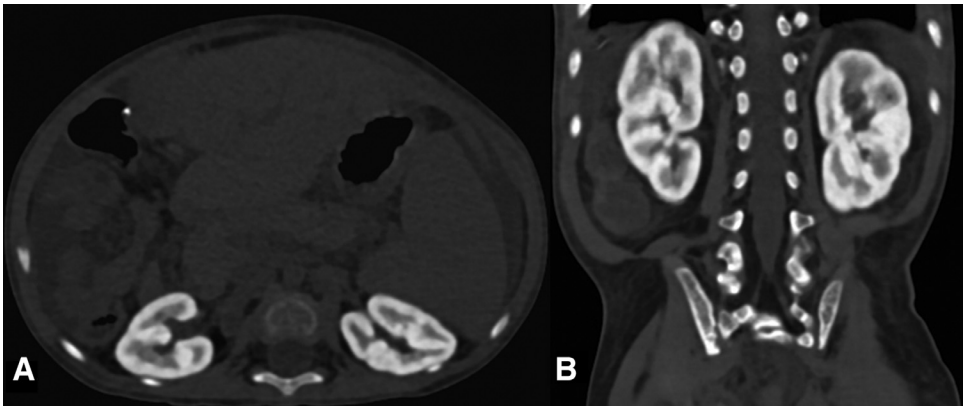

Figure 3. (**A**, **B**): Axial section and coronal reformat of precontrast CT abdomen shows bilateral extensive nephrocalcinosis.

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