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(CASE REPORT)

CORTICAL-PREDOMINANT NEPHROCALCINOSIS AS AN EARLY RADIOLOGIC MARKER OF FULMINANT INFANTILE PRIMARY HYPEROXALURIA TYPE 1: A CASE REPORT

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ABSTRACT

Background: Primary hyperoxaluria type 1 (PH1) is a rare autosomal recessive disorder of glyoxylate metabolism leading to hepatic oxalate overproduction, progressive nephrocalcinosis, and early end-stage renal disease (ESRD), particularly in its severe infantile form. Early radiological recognition is critical for prompt metabolic and genetic evaluation.

Case Presentation: A 3-month-old infant born to consanguineous parents presented with status epilepticus secondary to severe acute kidney injury (AKI), anuria, profound electrolyte disturbances (hyponatremia 113–122 mmol/L; hyperkalemia 6.8 mmol/L), metabolic acidosis (pH 7.00), and severe anemia (Hb 4.3 g/dL). Renal ultrasound demonstrated bilaterally normal-sized kidneys with diffuse, intense hyperechogenicity involving both cortex and medulla, complete loss of corticomedullary differentiation, and no obstruction. The pattern appeared cortical-predominant. Follow-up ultrasounds confirmed persistent symmetric "bright kidney" appearance. Non-contrast CT revealed dense symmetric cortico-medullary nephrocalcinosis (>100–200 HU) without obstructing calculi or hydronephrosis. Renal biopsy showed abundant birefringent calcium oxalate crystals, highly suggestive of oxalate nephropathy in the context of infantile fulminant disease.

Conclusion: Diffuse cortical-predominant nephrocalcinosis on ultrasound in infants with severe unexplained AKI should prompt urgent suspicion of PH1. Early radiologic recognition accelerates metabolic evaluation, renal biopsy confirmation, and multidisciplinary management, including potential RNAi therapy.

Keywords: Primary hyperoxaluria type 1; Infantile oxalosis; Cortical nephrocalcinosis; Renal ultrasound; Bright kidneys; Oxalate nephropathy.

1 INTRODUCTION

Primary hyperoxaluria type 1 (PH1) results from deficiency of hepatic alanine-glyoxylate aminotransferase (AGT), leading to excessive oxalate production and systemic oxalosis [1]. The infantile form (<6 months) is particularly aggressive and often presents with fulminant renal failure and massive nephrocalcinosis [1,2].

Ultrasound is the first-line imaging modality and plays a pivotal diagnostic role. Two sonographic patterns have been described in pediatric PH1:

- A medullary pattern (more frequent, less specific)
- A cortical or cortico-medullary pattern, associated with rapid progression to ESRD [3]
- We report a case of severe infantile PH1 with early cortical-predominant nephrocalcinosis, emphasizing the diagnostic value of imaging.

2 CASE PRESENTATION

A 3-month-old infant born to first-degree consanguineous parents was admitted for generalized tonic–clonic seizures. Examination revealed hypotonia, pallor, lethargy (Glasgow 12–13), and oliguria progressing to anuria.

Laboratory Findings

Creatinine: 145 µmol/L Urea: 2.3 g/L

Hyponatremia: 113 mmol/L Hyperkalemia: 6.8 mmol/L

Metabolic acidosis: pH 7.00, HCO_3^- 7 mmol/L Hemoglobin: 4.3 g/dL

Imaging Findings

Initial Ultrasound

Both kidneys were normal in size for age. There was marked diffuse hyperechogenicity of the entire parenchyma with complete loss of corticomedullary differentiation. The hyperechogenicity was cortical-predominant with posterior acoustic shadowing. No hydronephrosis or calculi were identified.

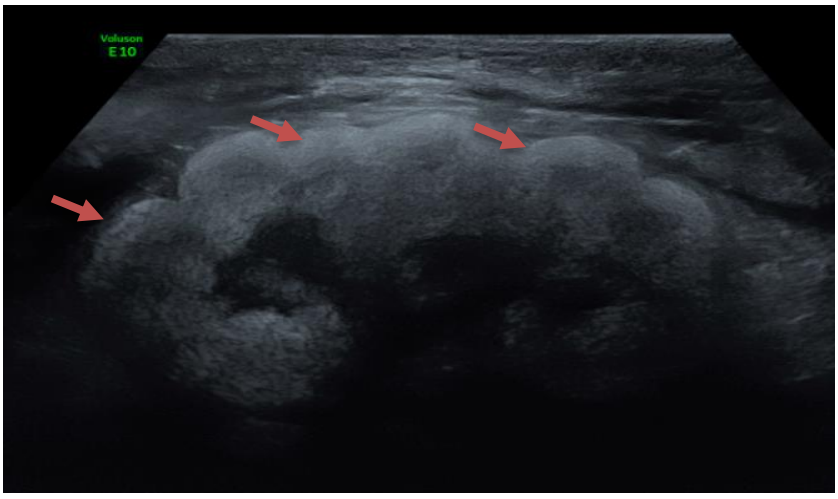

Figure 1: Initial renal ultrasound: marked diffuse hyperechogenicity predominantly involving the cortex (Orange arrow), with complete loss of corticomedullary differentiation and posterior acoustic shadowing. Typical "bright kidneys" appearance in fulminant infantile primary hyperoxaluria type 1

2.1 Follow-Up Ultrasound

Persistent bilateral symmetric diffuse hyperechogenicity was observed over weeks 1–4, consistent with progressive oxalate nephrocalcinosis.

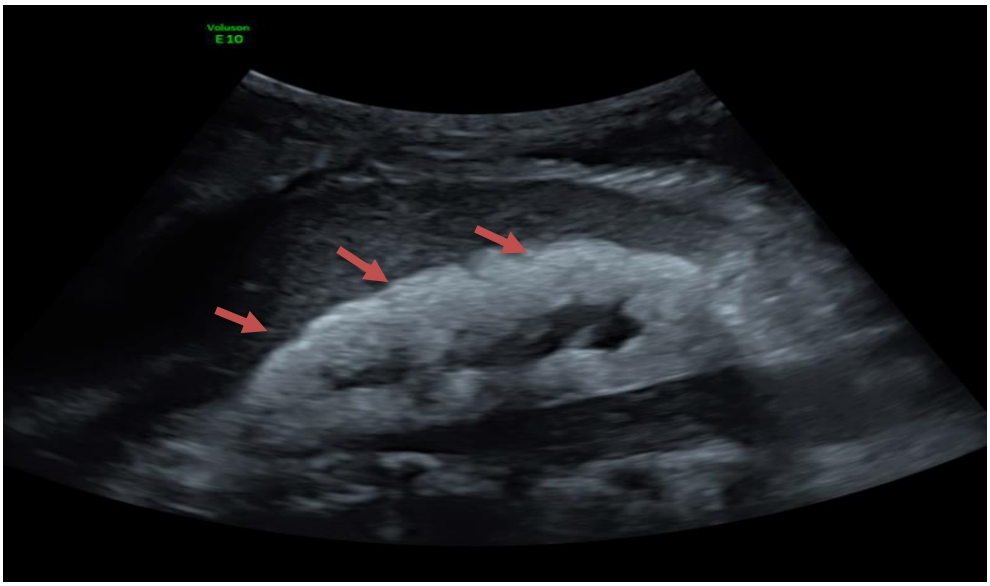

Figure 2: Follow-up ultrasound (weeks 1–4): persistent bilateral symmetric diffuse hyperechogenicity (Orange arrow), consistent with progressive oxalate nephrocalcinosis.

2.2 Non-Contrast CT

CT confirmed symmetric dense cortico-medullary nephrocalcinosis with high-attenuation deposits (>100–200 HU). No discrete obstructing stones were seen.

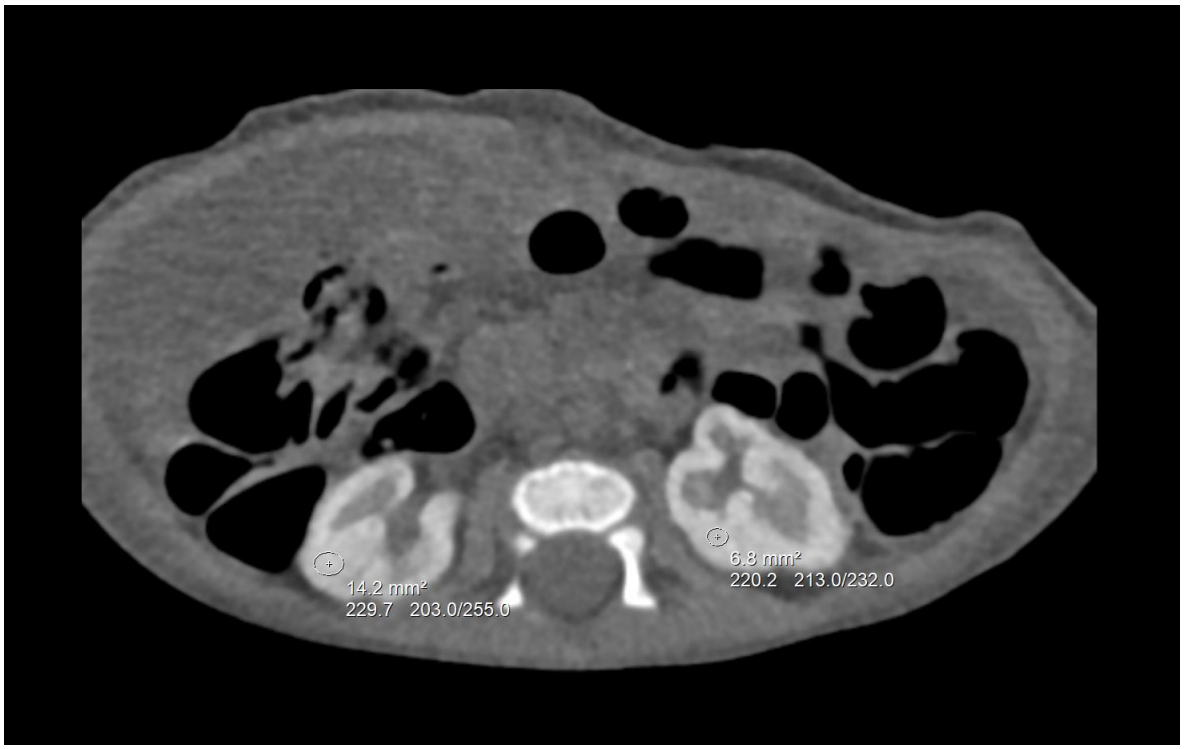

Figure 3: Non-contrast CT: dense symmetric bilateral cortico-medullary nephrocalcinosis with high-attenuation deposits (>200 HU), without obstructing calculi or hydronephrosis.

2.3 Management and Outcome

Urgent peritoneal dialysis was initiated. Electrolyte disturbances were corrected. The patient required transfusions, erythropoietin therapy, and antihypertensive management. Persistent anuria necessitated chronic dialysis.

Renal biopsy revealed acute tubular necrosis and extensive intratubular/interstitial calcium oxalate crystals (birefringent under polarized light).

3 DISCUSSION

PH1 is the most severe form of primary hyperoxaluria and accounts for the majority of infantile oxalosis cases [1,2]. Oxalate overproduction leads to early intrarenal crystal deposition, causing tubular injury, interstitial inflammation, and rapid decline to ESRD.

Ultrasound is highly sensitive for nephrocalcinosis detection. Diallo et al. described two distinct sonographic patterns in pediatric PH1: medullary and cortical [3]. The cortical pattern, as observed in our case, correlates with more aggressive disease and faster progression to ESRD.

Strauss et al. further emphasized the broad imaging spectrum of PH1 and the value of early radiologic suspicion [4]. In our patient, cortical-predominant hyperechogenicity without obstruction was a key diagnostic clue.

Non-contrast CT provides superior density characterization and confirms extensive oxalate deposition while excluding obstructive uropathy [4,5]. Although radiation exposure should be minimized in infants, CT can support diagnosis when ultrasound findings are atypical or severe.

Recent expert consensus from ERKNet and OxalEurope underscores the importance of early imaging-based suspicion to accelerate genetic testing and initiate therapy [2]. The advent of RNA interference therapy, particularly lumasiran, has significantly changed the therapeutic landscape by reducing hepatic oxalate production [6]. Early diagnosis may therefore influence long-term outcomes.

Differential diagnoses include distal renal tubular acidosis and Bartter syndrome; however, these typically demonstrate medullary-limited nephrocalcinosis and lack the profound metabolic derangements and early renal failure characteristic of infantile PH1.

4 CONCLUSION

Cortical-predominant diffuse nephrocalcinosis on ultrasound in an infant with severe unexplained AKI should immediately raise suspicion for PH1. Early radiological recognition enables prompt histopathological confirmation (renal biopsy demonstrating calcium oxalate crystals) and therapeutic intervention.

STATEMENTS ON COMPLIANCE WITH ETHICAL STANDARDS

Disclosure of conflict of interest

The authors declare no conflicts of interest regarding this manuscript.

Statement of informed consent

Informed consent was obtained from the legal guardians of the patient included in this study.

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