RESEARCH PAPER

Primary Oxalosis with Extensive Nephrocalcinosis: A Case Report

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ABSTRACT
Primary hyperoxaluria type 1 (PH1) is a rare autosomal recessive metabolic disorder caused by deficiency of the liver enzyme alanine-glyoxylate aminotransferase, leading to excessive oxalate production, recurrent nephrolithiasis, and progressive nephrocalcinosis. We report a case of a 14-year-old boy with genetically confirmed PH1 (EXON 4 homozygous pathogenic mutation) diagnosed at 3 years of age, who presented with progressively decreasing urine output and abdominal pain. He was undergoing ambulatory peritoneal dialysis for stage V chronic kidney disease. Laboratory evaluation revealed elevated serum oxalate (215 μmol/L) and serum creatinine (5.0 mg/dL post-dialysis). Plain radiographs demonstrated bilateral dense nephrocalcinosis. Ultrasound showed markedly echogenic renal cortices with posterior acoustic shadowing and shrunken kidneys. CT imaging confirmed extensive symmetrical cortical nephrocalcinosis with tram-line calcifications sparing the medulla. The patient had a history of recurrent renal calculi since 2012, requiring pyelolithotomy at age 2 and percutaneous nephrolithotomy with DJ stenting in 2015. Complications included oxalate cardiomyopathy with pulmonary arterial hypertension (>60 mmHg), dilated left atrium, left ventricular hypertrophy, and pulmonary regurgitation. FDG PET-CT revealed mild asymmetrical increased uptake in the left triradiate epiphysis and sacral ala, suggesting early skeletal oxalosis. The patient is currently awaiting combined liver-kidney transplantation. This case highlights the characteristic imaging findings of cortical nephrocalcinosis in PH1 and the importance of early diagnosis and multidisciplinary management to prevent systemic oxalosis...

Keywords: Primary hyperoxaluria type 1, cortical nephrocalcinosis, oxalosis, nephrolithiasis, combined liver-kidney transplantation
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INTRODUCTION

Primary hyperoxaluria type 1 (PH1) is the most common and severe form of the primary hyperoxalurias, a group of autosomal recessive disorders of glyoxylate metabolism [1]. It is caused by a deficiency of the liver-specific peroxisomal enzyme alanine-glyoxylate aminotransferase (AGT), encoded by the *AGXT* gene [2]. This deficiency leads to the accumulation of glyoxylate, which is then converted to oxalate. As oxalate is a metabolic end product with no physiologic role in humans, it must be excreted by the kidneys [1, 2]. The resulting hyperoxaluria leads to the formation of insoluble calcium oxalate crystals, causing recurrent urolithiasis and the progressive deposition of crystals within the renal parenchyma, a process known as nephrocalcinosis [3]. Nephrocalcinosis itself is a significant risk factor for the progression to kidney failure in these patients [3].

The clinical presentation of PH1 is variable, ranging from infantile nephrocalcinosis and early renal failure to a milder, adult-onset form with intermittent stone passage [4]. As renal function declines, the capacity to excrete oxalate diminishes, leading to rising plasma oxalate levels and systemic oxalosis—the widespread deposition of calcium oxalate crystals in extrarenal tissues, including bone, heart, retina, and blood vessels [1, 6]. Radiological evaluation plays a crucial role in diagnosis and monitoring. Cortical nephrocalcinosis, characterized by calcification in the peripheral renal cortex with sparing of the medulla, is a hallmark imaging finding in advanced PH1 [5, 6]. Definitive management for patients with end-stage renal disease (ESRD) involves combined or sequential liver-kidney transplantation, which corrects the underlying hepatic enzyme deficiency while restoring renal function [1]. We present a case of a 14-year-old boy with genetically confirmed PH1 who developed extensive cortical nephrocalcinosis and systemic oxalosis, illustrating the natural history of the disease and its characteristic imaging features.

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CASE PRESENTATION

A 14-year-old boy, the first-born child of non-consanguineous parents, presented to the outpatient department with complaints of progressively decreasing urine output and abdominal pain since 2022. He had been undergoing ambulatory peritoneal dialysis for stage V chronic kidney disease (CKD). His medical history was significant for recurrent renal calculi since 2012, when he first presented at the age of 3 years.

The diagnosis of primary hyperoxaluria type 1 was established at 3 years of age through genetic testing using blood EDTA, which identified an EXON 4 homozygous pathogenic mutation in the AGXT gene, confirming autosomal recessive hyperoxaluria type 1. The patient's early clinical course was characterized by repeated stone-related events. At age 2 years, he underwent pyelolithotomy for renal calculi. Subsequently, at age 5 years (2015), he required percutaneous nephrolithotomy (PCNL) with double-J (DJ) stenting for recurrent stones. Despite these interventions, his renal function progressively declined, and he commenced ambulatory peritoneal dialysis in 2022 (5 days per week).

On presentation, physical examination revealed a chronically ill-appearing adolescent with pallor and mild pedal edema. Blood pressure was elevated at 138/88 mmHg. Cardiovascular examination revealed a systolic murmur, and respiratory examination was unremarkable. Abdominal examination showed no palpable masses or tenderness.

Laboratory investigations revealed markedly elevated serum oxalate at 215 µmol/L (reference range: <46 µmol/L) and post-dialysis serum creatinine of 5.0 mg/dL (reference range: 0.6-1.2 mg/dL). Other laboratory parameters are summarized in Table 1.

Table 1: Laboratory Investigations

Parameter	**Result**	**Reference Range**
Serum oxalate	215 µmol/L	<46 µmol/L
Serum creatinine (post-dialysis)	5.0 mg/dL	0.6-1.2 mg/dL
Serum calcium	8.9 mg/dL	8.5-10.2 mg/dL
Serum phosphorus	5.2 mg/dL	2.5-4.5 mg/dL
Parathyroid hormone (PTH)	215 pg/mL	10-65 pg/mL
25-hydroxy vitamin D	18 ng/mL	30-100 ng/mL
Urinalysis	Calcium oxalate crystals	Negative

Imaging findings:

Plain radiograph of the abdomen (Figure 1) demonstrated bilateral dense nephrocalcinosis, with increased radiodensity in the renal outlines.

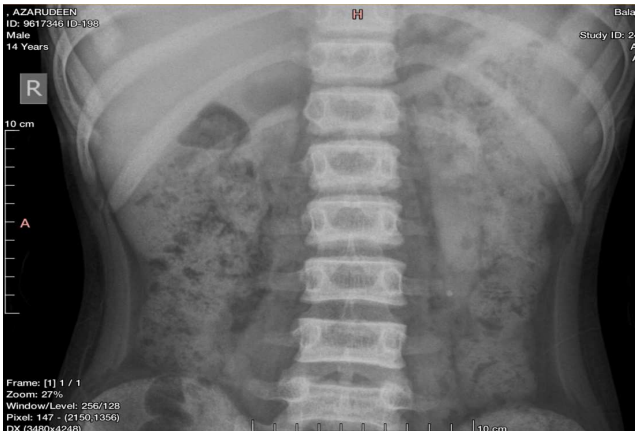

Figure 1: Bilateral increased renal density seen in the radiograph

Ultrasound of the abdomen (Figure 2)

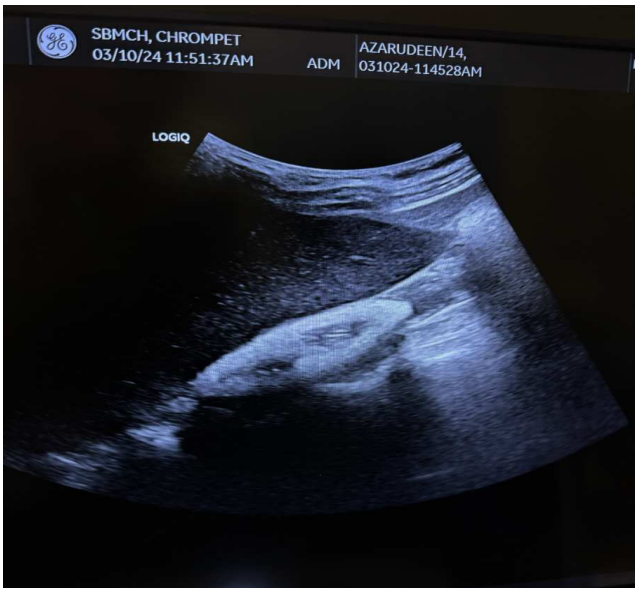

Figure 2: Both the kidneys (figure 2)show marked echogenic cortex with posterior acoustic shadowing denoting diffuse calcifications. -Right kidney appears shrunken in size and measures ~ 5.9 x 1.8 cm. -Left kidney appears shrunken in size and measures ~ 7.8 x 2.1 cm

revealed bilateral shrunken kidneys. The right kidney measured approximately 5.9 × 1.8 cm, and the left kidney measured approximately 7.8 × 2.1 cm. Both kidneys showed markedly echogenic renal cortices with posterior acoustic shadowing, denoting diffuse calcifications. Corticomedullary differentiation was poorly maintained.

Computed Tomography (CT) of the abdomen without contrast (Figures 3)

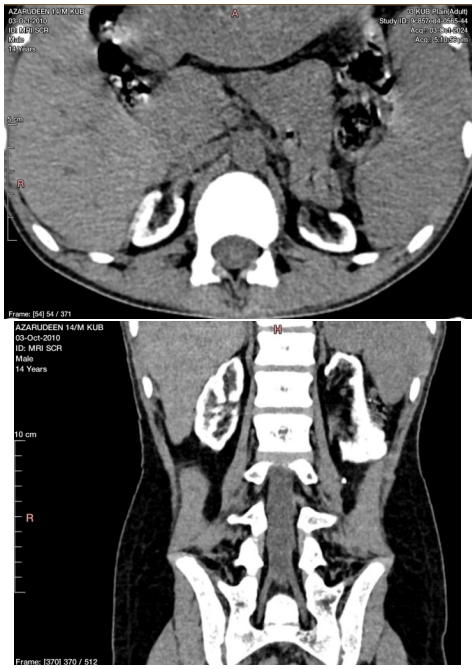

Figure 3: CT scan axial (left) and coronal (right) showing cortical nephrocalcinosis in a 14 year old patient with Primary Hyperoxaluria Type - I

demonstrated symmetrical, dense, uniform calcification confined to the renal cortex bilaterally, with complete sparing of the medullary pyramids. This tram-line pattern of calcification along the peripheral cortex and septa of Bertin is characteristic of cortical nephrocalcinosis in primary hyperoxaluria.

Magnetic Resonance Imaging (MRI) with T1-weighted coronal sequence (Figure 4)

Figure 4: MRI T1 WI Coronal (figure 5) showing cortical nephrocalcinosis in a 14 year old patient with Primary Hyperoxaluria Type – I

showed signal abnormalities corresponding to the calcified renal cortices, though nephrocalcinosis changes can be challenging to identify definitively on MRI due to susceptibility artifacts.

Systemic involvement assessment:

Given the risk of systemic oxalosis, further evaluation was performed:

Echocardiography revealed dilated left atrium, left ventricular hypertrophy, and pulmonary regurgitation, consistent with oxalate cardiomyopathy. Pulmonary arterial pressure was elevated at >60 mmHg, indicating pulmonary arterial hypertension.

FDG PET-CT showed mild asymmetrical increased uptake (SUVmax 9.1) in the triradiate epiphysis of the left hip bone and the left sacral ala of the S1-S2 segment, suggesting early skeletal oxalosis. Mild effusion was noted in both knee joints.

The patient is currently maintained on ambulatory peritoneal dialysis (5 sessions weekly) while awaiting combined liver-kidney transplantation. He has been counseled regarding the importance of medication adherence and avoidance of dehydration. Written informed consent was obtained from the parents for publication of this case report and accompanying images.

DISCUSSION

This case of a 14-year-old boy with genetically confirmed PH1 illustrates the classic and relentless progression of the disease from recurrent nephrolithiasis in early childhood to end-stage renal disease and systemic oxalosis by adolescence, a course that is well-documented in the literature [1, 2]. The underlying pathophysiology is a deficiency of the hepatic enzyme AGT, which disrupts the normal metabolism of glyoxylate and leads to its conversion to oxalate [2]. The excessive oxalate combines with calcium in the urine, forming crystals that aggregate into stones and deposit within the renal tubules and interstitium. This deposition triggers an inflammatory and

fibrotic response, culminating in progressive nephrocalcinosis and renal failure [3]. As highlighted by Tang et al., the presence and extent of nephrocalcinosis are significant risk factors for kidney failure in this patient population [3]. The imaging findings in this case are textbook examples of this process. The plain radiograph demonstrating bilateral renal density, the ultrasound showing densely echogenic cortices with acoustic shadowing, and most definitively, the CT scan revealing tram-line cortical calcification with medullary sparing, are all characteristic features of renal oxalosis [5, 6]. These imaging modalities are not only diagnostic but also serve to monitor disease progression.

Once the glomerular filtration rate falls, the kidneys can no longer adequately excrete the daily oxalate load, leading to systemic oxalosis [1]. Our patient's elevated serum oxalate level of 215 µmol/L is a clear indicator of this systemic overload. The cardiovascular system is particularly vulnerable to oxalate deposition, which can lead to cardiomyopathy, conduction defects, and heart failure [1]. The echocardiographic findings in our patient—dilated left atrium, left ventricular hypertrophy, pulmonary regurgitation, and pulmonary hypertension—are consistent with advanced cardiac oxalosis and represent a poor prognostic sign. Skeletal involvement is another common and debilitating manifestation of systemic oxalosis, as calcium oxalate crystals deposit in the bone marrow, periosteum, and growth plates [6]. The FDG PET-CT findings of increased uptake in the left hip and sacrum likely represent an inflammatory response to these crystal deposits, aligning with the classical skeletal changes described by Chiddarwar et al. [6]. This underscores the importance of whole-body imaging in patients with advanced PH1 to assess the full burden of disease.

The management of PH1 has evolved significantly and must be tailored to the stage of the disease. For patients with preserved renal function, conservative measures such as high fluid intake, pyridoxine (vitamin B6) supplementation (which can reduce oxalate production in a subset of patients with specific *AGXT* mutations), and citrate to inhibit crystallization are essential [1, 4]. However, for patients like ours with ESRD, these measures are insufficient. While intensive hemodialysis can help lower plasma oxalate levels, it is rarely able to keep pace with the continuous endogenous production, allowing systemic deposition to continue [7]. Definitive treatment for PH1 with ESRD is combined or sequential liver-kidney transplantation [8]. Liver transplantation corrects the primary enzymatic defect, as AGT is primarily expressed in hepatocytes, while kidney transplantation restores renal function and oxalate clearance. The goal of this combined approach is to prevent further oxalate production and allow for the slow mobilization and excretion of existing oxalate stores, thereby halting and potentially reversing systemic oxalosis [9,10]. Our patient's placement on the transplant list is the critical next step in his care. This case also highlights the need for early diagnosis and aggressive management of hyperoxaluria from the time of initial presentation to delay the onset of ESRD and prevent the devastating consequences of systemic oxalosis. The characteristic imaging findings should prompt clinicians to consider PH1 early in the differential diagnosis of recurrent stones and nephrocalcinosis.

CONCLUSION

Primary hyperoxaluria type 1 is a rare but devastating metabolic disorder that invariably leads to end-stage renal disease and systemic oxalosis if not adequately managed. The characteristic imaging finding of extensive cortical nephrocalcinosis with medullary sparing on CT is pathognomonic and should prompt immediate diagnostic evaluation. As this case demonstrates, systemic complications, particularly oxalate cardiomyopathy and skeletal involvement, are common once renal failure is established and carry a poor prognosis. Early diagnosis, aggressive supportive care, and timely referral for combined liver-kidney transplantation are paramount to improving outcomes and quality of life for patients with this challenging condition

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