Radiology Case Reports 15 (2020) 1806–1808

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Case Report

A case of failure to thrive secondary to primary hyperoxaluria type 1

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ARTICLE INFO

Article history:
Received 21 May 2020
Revised 4 July 2020
Accepted 6 July 2020

Keywords:
Primary Hyperoxaluria
Failure to thrive
Kidney stone
Ultrasound

ABSTRACT

Primary hyperoxaluria type 1 is a rare genetic condition characterized by oxalate deposition in the kidneys. We report findings of an 8-month old female presenting with failure to thrive, poor oral intake, and kidney stones resulting in the diagnosis of primary hyperoxaluria type 1. The patient exhibits a unique presentation without renal failure at the time of diagnosis suggesting a previously unreported comorbidity in early stages of disease.

Introduction

Primary hyperoxaluria type 1 (PH1) is a rare genetic autosomal recessive condition (1-3 per million) characterized by oxalate deposition in the kidneys as calcium oxalate stones due to a deficiency in the liver enzyme alanine glyoxylate aminotransferase. This results in nephrocalcinosis and eventually end stage renal disease (ESRD) [1–3]. Definitive diagnosis is obtained through genetic testing. Initial management includes fluid hydration and crystallization inhibitors. As the disease progresses to ESRD, patients will eventually require liver and kidney transplantation [3]. In infants, PH1 frequently presents with failure to thrive (FTT) in the setting of ESRD [1,2]. We describe the initial presentation of hyperoxaluria without renal compromise, complicated by severe FTT that did not respond to standard interventions.

Case Report

An 8-month-old full term Yemeni female born to consanguineous parents who are first cousins presented to the hospital with white stones in the diaper and increased irritability. Family history was negative for renal disease. The patient had a 4-year-old sibling with FTT of unknown etiology after negative work-up. Review of the patient's growth chart showed severe FTT (<0th %) with no weight gain for 2 months (Fig. 1). BUN/creatinine, bicarbonate, and electrolytes were normal.

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https://doi.org/10.1016/j.radcr.2020.07.019

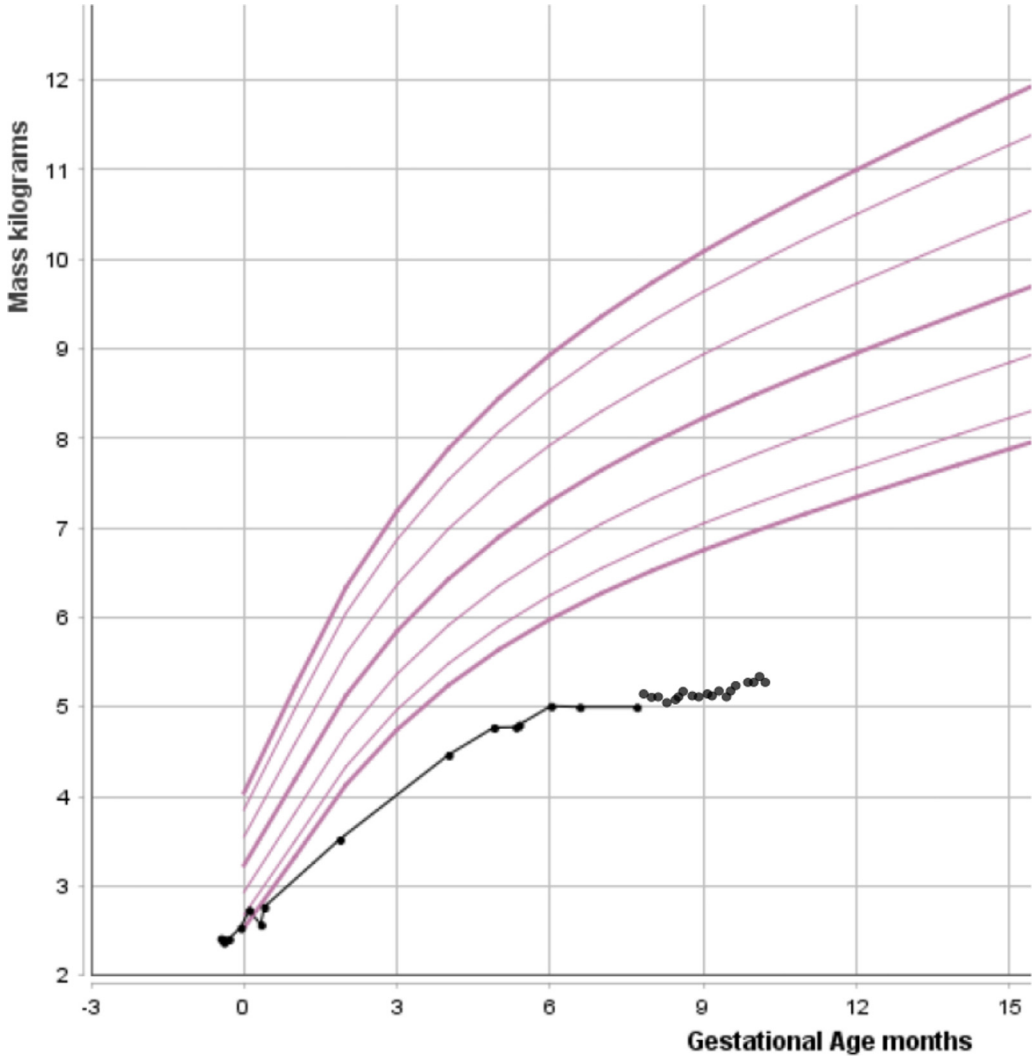

Figure 1 – Growth chart.
Severe failure to thrive (< 0th %) with no weight gain from 6 to 8 months. Persistent poor weight gain throughout admission despite interventions.

Ultrasound showed bilateral medullary nephrocalcinosis and bilateral nephrolithiasis up to 8 mm (Fig. 2). Subsequent stone analysis showed calcium oxalate composition and urine tests showed high levels of glycolic and oxalic acids. Genetic testing confirmed the diagnosis of PH1 with pathogenic variant of gene AGXT (c584T>G, p.Met195Arg). The patient was started on pyridoxine, potassium citrate, and potassium phosphate to decrease stone formation. While hospitalized the patient continued to pass kidney stones. She also had poor weight gain, likely due to periods of food refusal, irritability, and emesis. An extensive workup for other causes of FTT (renal tubular acidosis, malabsorption, metabolic/hormonal disorders) was negative. After 1 month in the hospital with no weight gain despite assistance from speech therapy, an NG tube was placed to meet fluid (150 ml/kg/day) and caloric goals (100 kcal/kg/day); she then gained 15 g/day (Fig. 1). After 62 days in the hospital and 470 g weight gain, she was discharged to a rehabilitation center to receive continued feeding therapy. Ultrasound prior to discharge showed unchanged nephrocalcinosis and nephrolithiasis.

Discussion

There are no previous case reports describing patients with PH1 and FTT that could not be attributed to the patient's declining renal function [4]. In fact, there are few descriptions in the literature of infantile hyperoxaluria without accompanying ESRD at time of diagnosis [4,5]. According to the rare disease registry, 60% of patients with PH1 present with abdominal pain [6]. We theorize that her food aversion, which persisted despite behavioral therapies, is related to colic from her nephrolithiasis.

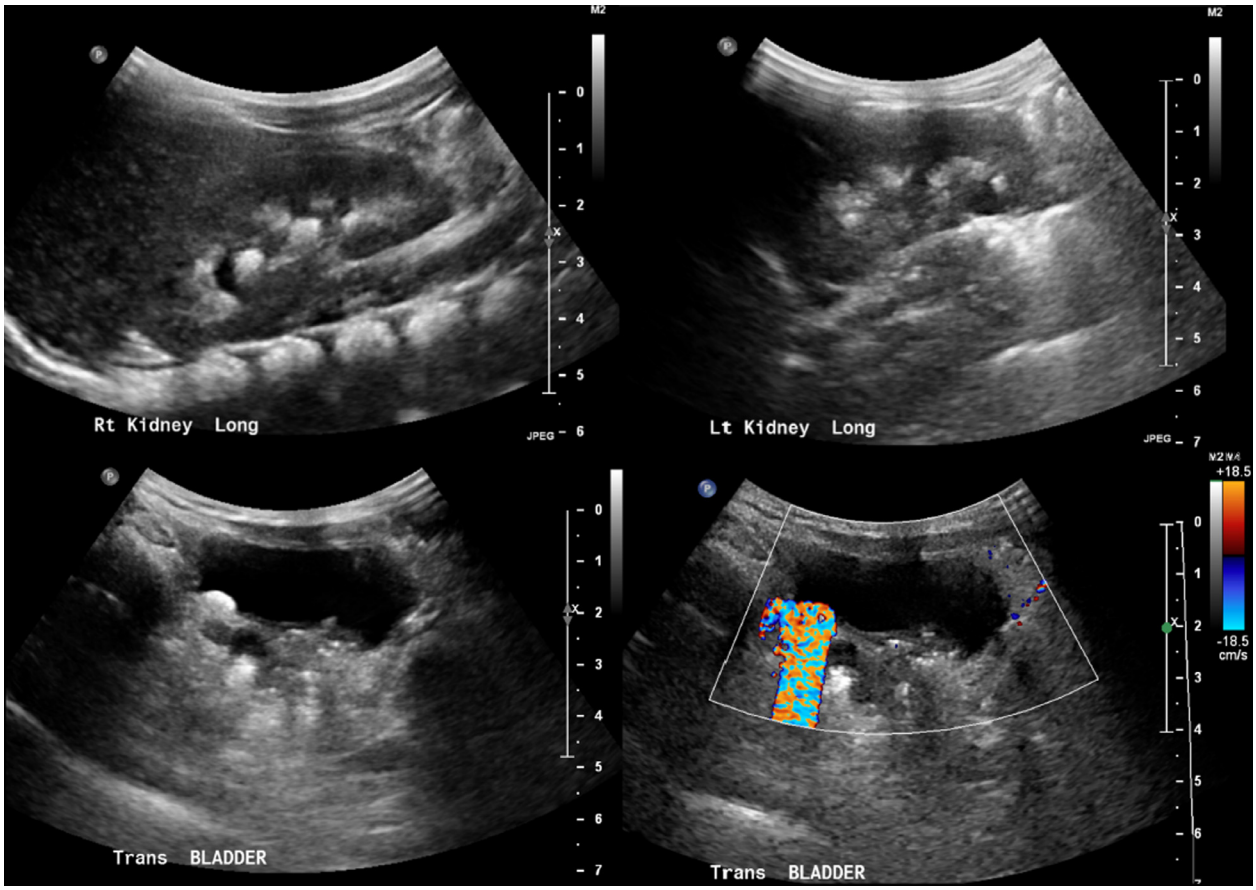

Figure 2 – Ultrasound images of kidneys and bladder on admission.
Grayscale sonographic images of the right (a) and left (b) kidneys demonstrate increased echogenicity involving the medullary pyramids, some of which appear as distinct stones within the renal calyces. Transverse grayscale (c) and color Doppler (d) sonographic images of the bladder demonstrate an echogenic structure layering in the bladder which demonstrates "twinkle" artifact on color Doppler (a finding often associated with urinary tract stones). The distal right ureter is dilated and likely contains layering echogenic material.

Conclusion

In our patient PH1 presented with FTT without accompanying renal failure at the time of diagnosis, suggesting a previously unreported comorbidity in the early stages of the disease. It also describes the course of infantile hyperoxaluria prior to ESRD which has not been well characterized previously. Poor oral intake is a barrier to optimization of care in patients with hyperoxaluria as high fluid intake is required to mitigate precipitation of stones. Future case reports are needed to confirm these findings.

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