CASE REPORT

Late Diagnosis of Primary Hyperoxaluria Type 1 Despite Recurrent Kidney Stones and Positive Family History

Jamal Qasem Abumwais

The Martyr Dr. Khalil Sulaiman Hospital, Jenin City, Palestine.

Corresponding author: Dr Jamal Q Abumwais   Email: jamal_abumwais@yahoo.com
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Abstract
Primary hyperoxaluria type 1 (PH1) is the most common form of primary hyperoxalurias. It results in increased synthesis and subsequent urinary excretion of the metabolic end product oxalate and the deposition of insoluble calcium oxalate in the kidney and urinary tract. Individuals with PH1 are at high risk for recurrent nephrolithiasis, nephrocalcinosis, and end-stage renal disease (ESRD). A 13 years-old female presented with fatigue, headache, nausea, vomiting, anorexia, renal colic. Initial clinical and laboratory investigations revealed ESRD and the patient was initiated on hemodialysis. Family history of PH1 and a history of recurrent kidney stones for 4 years was elicited. Subsequently, PH1 was confirmed by, physical examination, medical history, family history, laboratory tests, ultrasound imaging, X-ray, CT scanning and renal biopsy. As this disease is no longer rare in Jenin District of Palestine where this patient was seen, delayed diagnosis can not be justified. In conclusion, early diagnosis and effective treatment of PH1 in areas with high prevalence, such as the Jenin District of Palestine, is warranted as it may delay the progression towards ESRD or systemic oxalosis.

Key words: Primary hyperoxaluria type 1, End-stage renal disease (ESRD), Nephrocalcinosis, Nephrolithiasis.

Introduction
Primary hyperoxaluria type 1 (HP1) is the most common form of primary hyperoxalurias (PH). It is an autosomal recessive disorder caused by deficiency of the liver-specific enzyme alanine: glyoxylate aminotransferase (AGT). This results in increased synthesis and subsequent urinary excretion of the metabolic end product oxalate and the deposition of insoluble calcium oxalate in the kidney and urinary tract. As glomerular filtration rate (GFR) decreases due to progressive renal involvement, oxalate accumulates and results in systemic oxalosis (1). PH is a heterogeneous disease with a variable age of onset and a
A thirteen year-old female was admitted to the emergency unit of The Martyr Dr. Khalil Sulaiman Hospital in Jenin city of Palestine on 1/10/2012 suffering from fatigue, headache, nausea, vomiting, anorexia and renal colic. She had had a 4 year history of recurrent kidney stones. Laboratory tests showed hemoglobin 9g/dl, red blood cells 3.53 m, white blood cells 4.8 k/Ul, urea 155.4 mg/dl and serum creatinine 21.6 mg/dl. Urinalysis showed 3+ proteinuria and 1+ hemoglobinuria. Microscopic examination of urine sediment showed many white blood cells (packed field), 10-12 red blood cells and 8-10 epithelial cells. Depending on these laboratory findings, clinical investigation, and medical history (recurrent kidney stones), the patient was diagnosed initially as ESRD as a result of recurrent kidney stones and she was initiated on hemodialysis 3 times weekly after 3 sessions in the initial 3 days from diagnosis. Abdominal ultrasound showed that both kidneys to be 10 cm in length with marked increase in echogenicity obliterating corticomedullary differentiation. Partial hydronephrosis of the left kidney was also seen with no evidence of solid renal masses identified. On both sides and at positions corresponding to renal papillae there were calcification of medium and large size in keeping with calcified papillae, 4 cm cystic space was noted in the upper pole of left kidney. Review of the history revealed that the patient suffered from renal colic about two months before developing ESRD (on 11/8/2012). A urinary tract ultrasound was done which showed multiple left renal stones with the largest one being about 1.5 cm in size, there was no hydronephrosis. Urinary bladder was normal. Unfortunately, the patient was not duly followed by physicians and family after this. Concerning the right kidney, there were 3 stones, the largest one about 1 cm in size, there was no hydronephrosis.

Case presentation

A thirteen year-old female was admitted to the emergency unit of The Martyr Dr. Khalil Sulaiman Hospital in Jenin city of Palestine on 1/10/2012 suffering from fatigue, headache, nausea, vomiting, anorexia and renal colic. She had had a 4 year history of recurrent kidney stones. Laboratory tests showed hemoglobin 9g/dl, red blood cells 3.53 m, white blood cells 4.8 k/Ul, urea 155.4 mg/dl and serum creatinine 21.6 mg/dl. Urinalysis showed 3+ proteinuria and 1+ hemoglobinuria. Microscopic examination of urine sediment showed many white blood cells (packed field), 10-12 red blood cells and 8-10 epithelial cells. Depending on these laboratory findings, clinical investigation, and medical history (recurrent kidney stones), the patient was diagnosed initially as ESRD as a result of recurrent kidney stones and she was initiated on hemodialysis 3 times weekly after 3 sessions in the initial 3 days from diagnosis. Abdominal ultrasound showed that both kidneys to be 10 cm in length with marked increase in echogenicity obliterating corticomedullary differentiation. Partial hydronephrosis of the left kidney was also seen with no evidence of solid renal masses identified. On both sides and at positions corresponding to renal papillae there were calcification of medium and large size in keeping with calcified papillae, 4 cm cystic space was noted in the upper pole of left kidney. Review of the history revealed that the patient suffered from renal colic about two months before developing ESRD (on 11/8/2012). A urinary tract ultrasound was done which showed multiple left renal stones with the largest one being about 1.5 cm in size, there was no hydronephrosis. Urinary bladder was normal. Unfortunately, the patient was not duly followed by physicians and family after this. Concerning the right kidney, there were 3 stones, the largest one about 1 cm in size, there was no hydronephrosis.

Discussion

Despite the personal history of recurrent kidney stones for 4 years and family history of PH1, recognition of the sad case
was delayed till the patient developed ESRD. Although she had bilateral multiple kidney stones and nephrocalcinosis for long time, she was not screened for PH over the years 2008-2012. This misdiagnosis (i.e. inability to distinguish common types of kidney stones from recurrent kidney stones caused by PH) reflects the difficulty that is faced by some physicians in making a confident diagnosis of PH. This may be due to limited experience in this field, but ignoring family history by the physicians must have contributed to this miss-diagnosis. The patient is from a family with a history of PH (2 maternal cousins died from complications of PH1 and many persons from the patient’s own tribe are either suffering or died from the same disease). There are several reports in the literature in which a delay in the diagnosis of PH for many years occurred and sometimes until ESRD has developed (2,6-9). Furthermore, in some reports, the diagnosis of PH was only reached after failed kidney transplantation (10,11). The patient’s family on many occasions, play an important role in the progression of the patient’s case towards ESRD. Parents must seek second opinion, they should not leave their child’s case of recurrent kidney stones without adequate explanation. This may have been a definite contributing factor in our case.

In conclusion, due to the difficulties that face some physicians or nephrologists in the diagnosis of PH due to rarity of the disease, lack of some medical facilities or limited experience, delay in the diagnosis of the disease may occur. However, in some areas or certain groups physicians must have a high index of suspicion when dealing with increased risk such as the patient’s tribe in Jenin district which has a history of PH. All patients with history of recurrent kidney stones from this tribe must be screened for PH. Therefore, family history remains an important tool in our practice with this regard. Because of the heterogeneity of PH and because it may appears in infancy, some times since birth or after few months from birth, every case of kidney stone in the kids must be explored to find an etiology, family history must always be ascertained and documented.

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References

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