ejpmr, 2025, 12(4), 621-623

SJIF Impact Factor 7.065

EUROPEAN JOURNAL OF PHARMACEUTICAL AND MEDICAL RESEARCH

www.ejpmr.com

Case Study

ISSN 2394-3211

EJPMR

UNILATERAL CYSTINE STONE IN FEMALE ADULT PATIENT: CASE REPORT

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Article Received on 23/02/2025 **Article Revised on 14/03/2025** **Article Published on 04/04/2025**

ABSTRACT

Cystinuria, a rare genetic disorder, leads to the formation of cystine stones, with a subset of patients developing stones exclusively in one kidney. While cystine stones are more frequently observed in bilateral formations, unilateral cases do occur, and understanding why stones form unilaterally in some individuals remains unclear. We report a case of a 35-year-old female presented with right flank pain and lower urinary tract symptoms and was found to have a 5 cm right renal stone. Following open surgical removal at Tishreen University Hospital, the stone was confirmed to be pure cystine. Her 24-hour urine cystine level was 620 mg/L. Post-operatively, she was treated with potassium citrate and increased fluid intake.

KEYWORDS: Cystinuria, cystine stone, unilateral, adult.

INTRODUCTION

Urinary stone disease is a common global ailment, affecting 1-15% of individuals at least once in their lifetime, with variations based on several factors[1], and stone recurrence occurs in approximately 50% of patients.[2]

Despite the importance of metabolic evaluation for identifying predisposing factors and preventing recurrence, especially in high-risk patients, studies indicate that only a small percentage (7.4%) of high-risk stone formers undergo such evaluations.[3]

Cystine stones are infrequent, representing 1-3% of all urinary stones, but account for 10% of stones in children.[4]

Amino acids are normally filtered in the kidney glomeruli and almost entirely reabsorbed in the proximal convoluted tubules. This amino acid transport in the kidney and jejunum is facilitated by the Heteromeric amino acid transporter (HAT). The cystine transporter is encoded by the SLC3A1 gene on chromosome 2 and the SLC7A9 gene on chromosome 19.[5,6]

Cystinuria is a recessive autosomal genetic disorder, rarely autosomal dominant with incomplete penetrance, classified into three types by the International Cystinuria Consortium (ICC)[7]:
Type A: mutation on the chromosome 2 gene.
Type B: mutation on the chromosome 19 gene.
Type AB: mutations on both genes.

Cystinuria is characterized by the presence of four amino acids in the urine: Cystine, Ornithine, Lysine, and Arginine (COLA), but cystine's low solubility makes it the primary stone-forming culprit.[8]

Cystine stone formation is primarily governed by the degree of cystine supersaturation in the urine and the urinary pH, as there is no known inhibitor of cystine crystal deposition.[9] Cystine solubility is highly pH-dependent: at pH 5, solubility is approximately 300 mg/L; at pH 7, it increases to 400 mg/L; and at pH 9, it reaches 1000 mg/L.[10] The coexistence of other metabolic disorders, such as hypercalciuria or hypocitraturia, can lead to the formation of mixed stones like calcium-cystine stones.[11]

CASE SUMMARY

A 35-year-old female presented to Tishreen University Hospital with right flank pain, which was relieved by analgesics, and lower urinary tract symptoms including urinary infection and urinary urgency. Her past medical history was unremarkable, with a surgical history significant only for two Cesarean sections.

Initial laboratory analysis showed a hemoglobin level of 12.3 g/dL, a white blood cell count of 7800/µL, a creatinine level of 0.8 mg/dL, and a urea level of 27 mg/dL. Ultrasound imaging revealed a 5 cm kidney stone in the right kidney, extending from the renal pelvis to the inferior calyx, without hydronephrosis. A subsequent computed tomography (CT) scan confirmed the presence of a stone in the right renal pelvis extending to the inferior calyx, with a density of 840 Hounsfield Units

(HU).

The patient underwent open surgical removal of the right kidney stone under general anesthesia. A right DJ (double-J) ureteral stent was placed, along with a drain for peri-renal drainage. She was discharged 48 hours post-operatively.

Gross examination of the extracted stones revealed a rough, hard texture and a yellowish color. Laboratory analysis confirmed the stones were composed of pure cystine, and 24-hour urine collection confirmed cystinuria, with a cystine concentration of 620 mg/L.

Six weeks post-surgery, the ureteral stent was removed. The patient was started on prophylactic treatment with potassium citrate at a dose of 20 mEq three times daily to alkalinize the urine, along with instructions to increase fluid intake to more than three liters per day orally.

Cystine stones are more difficult to treat. Management includes increased fluid intake, and urinary alkalization with potassium citrate, which increases cystine solubility.

DISCUSSION

This case presents a rare occurrence in medical literature, as pure cystine stones are more frequently diagnosed in children compared to adults, whereas in this instance, the stone was first diagnosed in the fourth decade of life.

Cystine stones are commonly bilateral due to the underlying issue of excessive cystine excretion in the urine surpassing the saturation level; however, in this case, the stones were exclusively located in the right kidney. Furthermore, cystine stones in adults are typically non-pure, with calcium oxalate being a common component, but in this case, the cystine stones were pure.

In discussing this study in the context of existing literature, it is necessary to explore how these findings correlate with or diverge from other significant research in the field of urology about to urinary stones.

The study by Usawachintachit et al.(2018) conducted a retrospective case-control study to evaluate clinical outcomes in cystinuria patients with unilateral versus bilateral cystine stone formation. The study aimed to understand why some cystinuria patients consistently form stones in only one kidney despite seemingly similar contralateral renal anatomy. The study results indicated that most patients presented with bilateral cystine stones, and the specific cause for unilateral versus bilateral stone formation remained unclear. It is believed that the affected kidney has an unclear effect on cystine stone formation in patients with cystinuria.[12]

CONCLUSION

This case highlights the presentation, diagnosis, and management of a relatively uncommon condition, cystinuria, leading to cystine stone formation. While open surgical removal was required in this instance due to the size and location of the stone, long-term management focuses on medical strategies, including urine alkalinization with potassium citrate and increased fluid intake, to prevent recurrence. This case underscores the importance of considering cystinuria in patients presenting with renal calculi, especially when the stones have an unusual composition or when there is a history of recurrent stone formation, to ensure appropriate diagnosis and long-term management.

Declarations

Ethical approval and consent to participate:
Ethical approval to study was obtained from the Scientific Research Ethics Committee at Tishreen University on November 2024 in accordance with the Declaration of Helsinki.

Consent for publication

Not applicable.

Availability of Data and Materials

All the data generated or analyzed during this study are included in this published article. The datasets used and/or analyzed during the current study are available from the corresponding author upon reasonable request.

Competing interests

None.

Funding

None.

Author Contribution

Yousef Bada, collected the data, checked the quality of the data collection, analyzed and interpreted the data, designed and coordinated the study, undertook and checked the quality assessment, produced the first draft of the manuscript, wrote and edited the manuscript and approved the final manuscript before submission.

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