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Case Report

Pristine Cystine Urolithiasis

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Abstract

Urolithiasis in children is rare with a reported incidence of 1.8 per 1000 children. Cystine calculi are still rarer with a prevalence of 1:7000 and the typical age of onset is the second decade of life. We present a 3 years old girl with recurrent cystine stones in kidney.

INTRODUCTION

Cystine was described as an unusual chemical component in bladder calculi by Wollaston[1] about two centuries ago. He termed it cystic oxide. This nomenclature was later changed by Berzelius [2] to cystine because of the absence of an oxide component in it. The chemical structure of cystine was elaborated by Friedman[3] in the early 20th century. Garrod hinted as early as 1908 that cystinuria might be caused by an inherited disorder.[4]

The autosomal recessive pattern of cystinuria was first accurately described by Harris in 1955.[5] Its primary manifestation is repeated stone formation. Prevalence of cystine calculi is 1:7000 and the typical age of onset is in the second decade of life. [6,7] Urolithiasis per se is rare in children with reported incidence of 1.8 per 1000 children.[8] Cystine stones constitute 1–2% of all urinary calculi and 6–8% of pediatric renal calculi.[2] High recurrence rate of 60% with the accompanying risk of progressive renal impairment[9] in cystine urolithiasis justifies regular follow-up examination of patients with cystinuria.

We report a 3 years old girl with recurrent cystine calculi of the kidney and bladder. To our knowledge pure cystine renal calculi in children has rarely been reported in Indian medical literature.

CASE REPORT

A 3-year-old girl presented with abdominal pain and difficulty during micturition for 6 months. She was diagnosed with urinary bladder calculus along with left renal calculus. Patient underwent open cystolithotomy followed by left open pyelolithotomy after 3 weeks. After one year interval, she presented with similar complaints, investigation revealed left sided staghorn calculi causing pelvic-ureteric junction obstruction. She again underwent left pyelolithotomy. Postoperative recovery was uneventful.

Renal stone analysis showed 100% cystine stone. (Fig 1) Urine was found to be highly acidic with a

pH ranging from 3 to 4. Twenty-four hours urine analysis did not show cystine crystals. X-ray diffraction of the renal stone showed presence of cystine. (Fig 2)

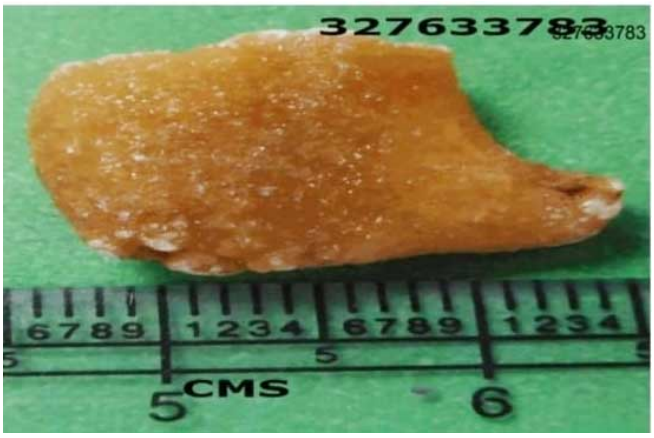

Fig 1. *The cystine stone.*

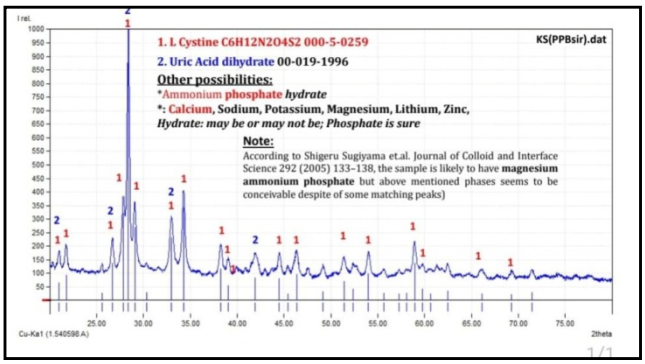

Fig 2. *Report of X ray diffraction analysis of the stone*

DISCUSSION

Renal stones may form at any age; more than 80% of patients develop their first stone within the first 2 decades.[10,11] Early formation of stones is more likely in males than in female patients. Our patient presented with cystine stone at a young age of 3 years.

Cystine is poorly soluble at physiological urine pH between 5 and 7. Stones are formed especially when urinary cystine levels exceeds 240–300 mg/l (1.33–1.66 mmol/l). Higher pH value of >8 leads to threefold increase in solubility of cystine crystals thus preventing stone formation.[12] Consequently, urine alkalinization is the main goal of pharmacotherapy. Oral alkalinization treatment is a safe and effective way of keeping the urine pH between 7.5 and 8.0. Potassium citrate is safer than the sodium compound (starting at 60–80 mEq/day).[13] In our patient, urinary pH was found to be between 4 and 5; after citrate supplement it was 8. Cystine stones must be suspected in patients, with family history of cystinuria.[14] In our patient the family history of cystinuria or recurrent urolithiasis was absent.

Microscopic examination of the first voided urine may reveal typical hexagonal cystine crystals, that confirm the diagnosis. However, such crystals are detectable in only 20–25% of urine specimens in patients with cystinuria.[11,13] Cystine stones typically have a homogeneous structure without striations and are visualized easily on plain radiographs, but they are less radio-dense than struvite or calcium oxalate stones.[15] Stag-horn cystine calculi are common.

Primary goal of conservative treatment is (in children and adults) to increase the solubility of urinary cystine. It includes increased fluid intake, low-salt diet and urinary alkalinization. Maintaining a high urine output of more than 120 ml/h is essential for therapeutic success regardless of drug treatment.

Compliance of these patients with medical treatment is often poor and most of them experience recurrent episodes of stone formation, requiring multiple interventions. Frequent ultra-sonography should be done to identify early recurrence. Family members of patients with cystinuria should undergo screening.[16,17]

REFERENCES

[1] Wollaston WH. On Cystic Oxide, a New Species of Urinary Calculus. Med Phys J. 1811 Mar; 25(145): 254-259.
[2] Ahmed K, Dasgupta P, Khan MS. Cystine calculi: challenging group of stones. Postgrad Med J. 2006 Dec; 82 (974): 799-801.
[3] Friedmann, E. Der Kreislauf des Schwefels in der organischen Natur. Ergebnisse der Physiologie 1, 15–31 (1902).

[4] WDH. Inborn Errors of Metabolism. Nature 1902; 81, 96–97.
[5] Harris H, Mittwoch U, Robson EB, Warren FL. Phenotypes and genotypes in cystinuria. Ann Hum Genet. 1955 Aug; 20(1): 57-91.
[6] Asanuma H, Nakai H, Takeda M, Shishido S, Kawamura T, Nagakura K, Yamafuji M. [Clinical study on cystinuria in children--the stone management and the prevention of calculi recurrence]. Nihon Hinyokika Gakkai Zasshi. 1998 Sep; 89(9): 758-65. (Japanese).
[7] Gentili A, Ria P, Lupo A, Fabris A. [Cystinic nephrolithiasis: clinical experience and new diagnostic and therapeutic perspectives]. G Ital Nefrol. 2016 May-Jun; 33(3): gin/33.3.5. Italian.
[8] Habbig S, Beck BB, Hoppe B. Nephrocalcinosis and urolithiasis in children. Kidney Int. 2011 Dec; 80(12): 1278-91.
[9] Gambaro G, Favaro S, D'Angelo A. Risk for renal failure in nephrolithiasis. Am J Kidney Dis. 2001 Feb; 37(2):233-43.
[10] Leusmann DB, Blaschke R, Schmandt W. Results of 5,035 stone analyses: a contribution to epidemiology of urinary stone disease. Scand J Urol Nephrol. 1990; 24(3):205-10.
[11] Dello Strologo L, Pras E, Pontesilli C, Beccia E, Ricci-Barbini V, de Sanctis L, Ponzone A, Gallucci M, Bisceglia L, Zelante L, Jimenez-Vidal M, Font M, Zorzano A, Rousaud F, Nunes V, Gasparini P, Palacín M, Rizzoni G. Comparison between SLC3A1 and SLC7A9 cystinuria patients and carriers: a need for a new classification. J Am Soc Nephrol. 2002 Oct; 13(10): 2547-53.
[12] Dent CE, Senior B. Studies on the treatment of cystinuria. Br J Urol. 1955 Dec; 27(4): 317-32.
[13] Ng CS, Streem SB. Medical and surgical therapy of the cystine stone patient. Curr Opin Urol. 2001 Jul; 11(4): 353-8.
[14] Knoll T, Zöllner A, Wendt-Nordahl G, Michel MS, Alken P. Cystinuria in childhood and adolescence: recommendations for diagnosis, treatment, and follow-up. Pediatr Nephrol. 2005 Jan; 20(1): 19-24.
[15] Dretler SP. Stone fragility--a new therapeutic distinction. J Urol. 1988 May; 139(5): 1124-7.
[16] Singer A, Das S. Cystinuria: a review of the pathophysiology and management. J Urol. 1989 Sep;142(3):669-73.
[17] Barbey F, Joly D, Rieu P, Mejean A, Daudon M, Jungers P. Medical treatment of cystinuria: critical reappraisal of long-term results. J Urol. 2000 May; 163(5): 1419-23.

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