Urol Res (2011) 39:417–419
DOI 10.1007/s00240-011-0364-z

CASE REPORT

Urine alkalinization may be enough for the treatment of bilateral renal pelvis stones associated with Lesch–Nyhan syndrome

Mi Mi Oh · Byeong Kuk Ham · Seok Ho Kang · Jae Hyun Bae · Je Jong Kim · Ki Hwan Yoo · Duck Ki Yoon · Du Geon Moon

Received: 22 July 2010 / Accepted: 28 January 2011 / Published online: 18 February 2011

Abstract Lesch–Nyhan syndrome is a rare sex-linked disorder of purine metabolism that is caused by a mutation in the hypoxanthine–guanine phosphoribosyltransferase (HPRT) gene which causes marked hyperuricemia and hyperuricosuria, with signs of gouty arthritis and uric acid stone disease in early childhood. We report a case of renal pelvis calculi which was dissolved within 10 days of urine alkalinization and hydration.

Keywords Lesch–Nyhan syndrome · Urine alkalinization · Stones

Case report

A 10-year-old boy was admitted due to urinary tract infection (UTI) with gross hematuria. He displayed mental retardation, self-mutilating behavior, and choreoathetosis. He was previously misdiagnosed as cerebral palsy and had never undergone further work up before. A plain X-ray revealed a faint radio-opaque shadow overlying the left renal fossa and abdmino-pelvic computed tomography (CT) showed bilateral renal pelvis stones (Fig. 1). Urine analysis results were RBC > 60/HPF, WBC > 60/HPF, culture; no growth and urine pH; 5.6. Serum BUN and Cr were within normal limit. Serum uric acid level was 7.9 mg/dL. Lesch–Nyhan syndrome was suspected and he had undergone HPRT1 gene mutation analysis and showed a mutation of c.151C>T(p.Arg51X) in HRT1 gene. Allopurinol treatment (5 mg/kg as a single daily dose) with hydration (maintaining urine output more than 2 L) and urine alkalinization (using intravenous sodium 120 mEq/day) was started. As the patient was uncooperative due to his mental status, we decided to perform bilateral pyelolithotomy instead of lithotripsy and percutaneous nephrolithotomy. On 10th day of urine alkalinization and hydration, bilateral pyelolithotomy was performed. During right pyelolithotomy, we identified a stone sized 1 × 1 cm which was decreased compared with previous CT scan. Lt pyelolithotomy was performed and we identified no stone. Intraoperative sonography revealed no remnant stone. Urine PH at the time of operation was 7.0. On 7th postoperative day, a follow up CT showed no remnant stone (Fig. 2). The patient was discharged and was maintained on sodium bicarbonate and allopurinol.

Discussion

Lesch–Nyhan syndrome is a rare sex-linked disorder of purine metabolism that is caused by a mutation in the hypoxanthine–guanine phosphoribosyltransferase (HPRT) gene [1]. Patients with Lesch–Nyhan syndrome classically present with mental retardation, choreoathetoid movements, and self-mutilating behavior [2]. The diagnosis of Lesch–Nyhan syndrome should be considered in any child who presents with mental retardation and uric acid calculi [3]. The enzymatic deficiency of HPRT in Lesch–Nyhan syndrome results in hyperuricemia, which causes hyperuricosuria, uric acid stone formation, and uric acid crystal

M. M. Oh · B. K. Ham · S. H. Kang · J. H. Bae · J. J. Kim · D. K. Yoon · D. G. Moon (✉)
Department of Urology, Korea University Hospital, 80, Guro-dong, Guro-ku, Seoul 152-703, Republic of Korea
e-mail: dgmoon@korea.ac.kr

K. H. Yoo
Department of Pediatrics, Korea University Hospital, 80, Guro-dong, Guro-ku, Seoul 152-703, Republic of Korea

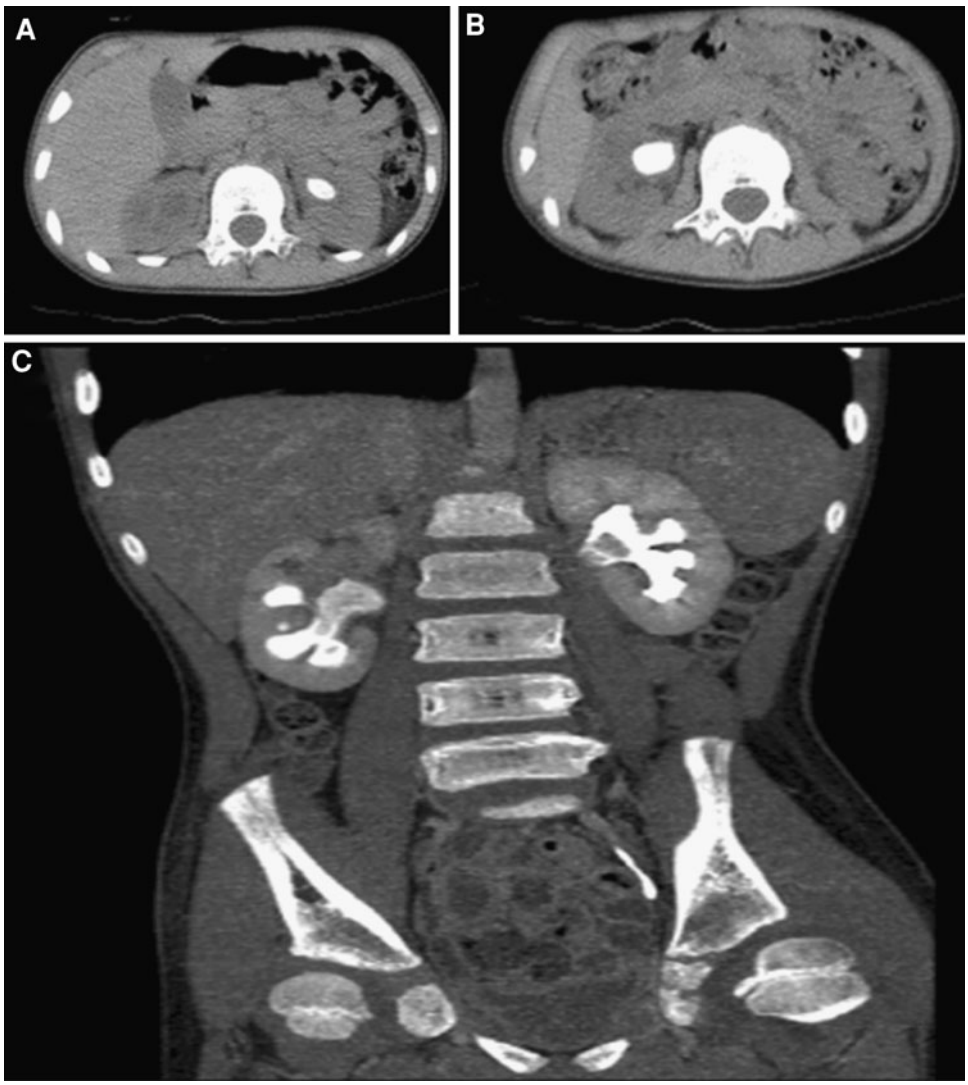

Fig. 1 **a** Non contrast enhanced abdomino-pelvic CT showing left renal pelvis stone. **b** Non contrast enhanced abdomino-pelvic CT showing right renal pelvis stone. **c** Coronal view showing multiple filling defects due to renal pelvis stones after IV contrast enhancement

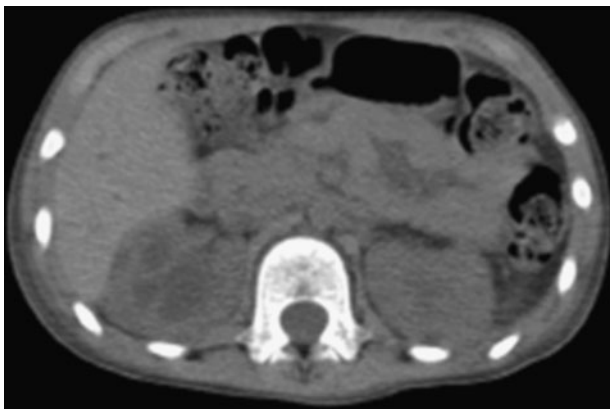

Fig. 2 No remnant stone on follow up abdomino-pelvic CT scan

nephropathy. Death caused by uric acid-related nephropathy is common in children with this disease [4].

The treatment of uric acid stones or uric acid crystal nephropathy in children with Lesch–Nyhan syndrome consists primarily of hydration, urinary alkalinization, and administration of allopurinol. Children who fail, do not tolerate, or are not candidates for this dissolution therapy, are candidates for standard interventions such as shock wave lithotripsy, ureteroscopy, percutaneous nephrolithotomy, and open surgery except in cases in which there is severe obstruction, progressive azotemia, serious infection, or unremitting pain, the initial management of patients with uric acid nephrolithiasis should be medical dissolution therapy because this approach is successful in the majority of cases (70–80%) [5, 6]. It is reported that in most cases, stone dissolution is slow and averages 1.0 cm of loss per month of therapy [1].

We report our case because the rate of stone dissolution was faster than we had expected which occurred within 10 days of urine alkalinization and if we had known the possibility of resolution of renal stones, the surgical treatment could have been avoided.

Children with Lesch–Nyhan syndrome are recommended to take a low purine diet (free of organ meats, fish such as anchovy, herring, mackerel, salmon, sardines and tuna, dried beans and peas) and high fluid intake to maintain a urinary volume of at least 2–2.5 L per day [7, 8]. Maintaining an alkaline urine, with potassium citrate or sodium bicarbonate, is also necessary as prophylaxis against uric acid stone formation [9]. The child's urinary pH should be monitored on a daily basis, with pH paper, and the urinary pH should be maintained between 6.5 and 7.5 [1].

References

1. Ankem M, Glazier DB, Barone JG (2000) Lesch–Nyhan syndrome presenting as acute renal failure secondary to obstructive uropathy. Urology 56:1056
2. Seegmiller JE (1968) Lesch–Nyhan syndrome. Management and treatment. Fed Proc 27:1097
3. Jenkins EA, Hallett RJ, Hull RG (1994) Lesch–Nyhan syndrome presenting with renal insufficiency in infancy and transient neonatal hypothyroidism. Br J Rheumatol 33:392
4. Rosenfeld DL, Preston MP, Salvaggi-Fadden K (1994) Serial renal sonographic evaluation of patients with Lesch–Nyhan syndrome. Pediatr Radiol 24:509
5. Kursh ED, Resnick MI (1984) Dissolution of uric acid calculi with systemic alkalization. J Urol 132:286
6. Moran ME, Abrahams HM, Burday DE et al (2002) Utility of oral dissolution therapy in the management of referred patients with secondarily treated uric acid stones. Urology 59:206
7. Cochat P, Pichault V, Bacchetta J et al (2010) Nephrolithiasis related to inborn metabolic diseases. Pediatr Nephrol 25:415
8. Porena M, Guiggi P, Micheli C (2007) Prevention of stone disease. Urol Int 79(1):37
9. Cameron JS, Moro F, Simmonds HA (1993) Gout, uric acid and purine metabolism in paediatric nephrology. Pediatr Nephrol 7:105