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Urolithiasis and psoas abscess in a 2-year-old boy with type 1 glycogen storage disease

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ORIGINAL ARTICLE

Urolithiasis and psoas abscess in a 2-year-old boy with type 1 glycogen storage disease

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Abstract We report on a pyogenic psoas abscess secondary to an impacted calcium oxalate ureteric stone in a 2-year-old boy with glycogen storage disease type 1 (GSD-1). The patient had a drainage of the abscess through a flank incision followed by percutaneous nephrostomy and open ureterolithotomy. Metabolic acidosis, hyperuricemia, hypocitraturia, and hypercalciuria appear to be significant in the pathogenesis of urolithiasis in patients with GSD-1. Regular ultrasonography of the abdomen along with optimal metabolic control may delay or prevent urolithiasis and its complications in GSD-1 patients.

Keywords Glycogen storage disease · Nephrocalcinosis · Psoas abscess · Urolithiasis

Introduction

Glycogen storage diseases are inherited disorders affecting enzymes involved in the synthesis and degradation of glycogen. Type 1 glycogen storage disease (GSD-1), also known as von Gierke's disease, is rare (incidence; 1 in 100,000 live births) and is characterized by the absence or deficiency of glucose-6-phosphatase in the liver, kidney, and intestinal mucosa [1, 2]. Clinical features include growth retardation, hepatomegaly, fasting hypoglycemia, lactic acidemia, hyperuricemia, and hyperlipidemia [1, 3, 4]. Despite advances in therapy, renal disease continues to be an important cause of morbidity and usually determines the long-term outcome [5]. Renal manifestations include nephromegaly, glomerular hyperfiltration and segmental glomerulosclerosis, and proximal and distal tubular dysfunctions causing a Fanconi-like syndrome, nephrocalcinosis, and nephrolithiasis [1, 3–6]. Calcium oxalate urolithiasis is rare in GSD-1 and can lead to obstruction and infectious complications [3, 7].

We report here a 2-year-old boy with GSD-1 who developed psoas abscess secondary to an impacted calcium oxalate ureteric stone and extravasation of urine from the ureteropelvic junction.

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Case report

A 2-year-old male, product of a consanguineous marriage, presented with 15-day history of fever (39°C), abdominal distension, and an inability to walk. According to the parents abdominal distension dated back to early infancy, and he was not growing well. There was no history of trauma, gastrointestinal, or urinary symptoms. Upon examination, he had a protuberant belly, "round doll face", and the height and weight were below the fifth percentile (height: 72 cm; weight: 8.3 kg). The liver was enlarged to 8 cm below the costal margin, and the right lower abdomen was erythematous and tender. Ultrasonography (US) and computerized tomography (CT) of the abdomen showed moderate hepatosplenomegaly, an abscess measuring 9.8×6.5×5.5 cm along the right psoas region, and right-sided hydroureteronephrosis due to a calculus in the lower ureter (Fig. 1a,b). Preliminary investigations revealed anemia (hemoglobin: 7.1 gm/dl; hematocrit: 23%), leukocytosis (38.3×10^9/l), neutrophilia (84%), and elevated C-reactive proteins (10.4 mg/dl; normal: 0–1 mg/dl). Blood urea nitrogen, creatinine, and serum

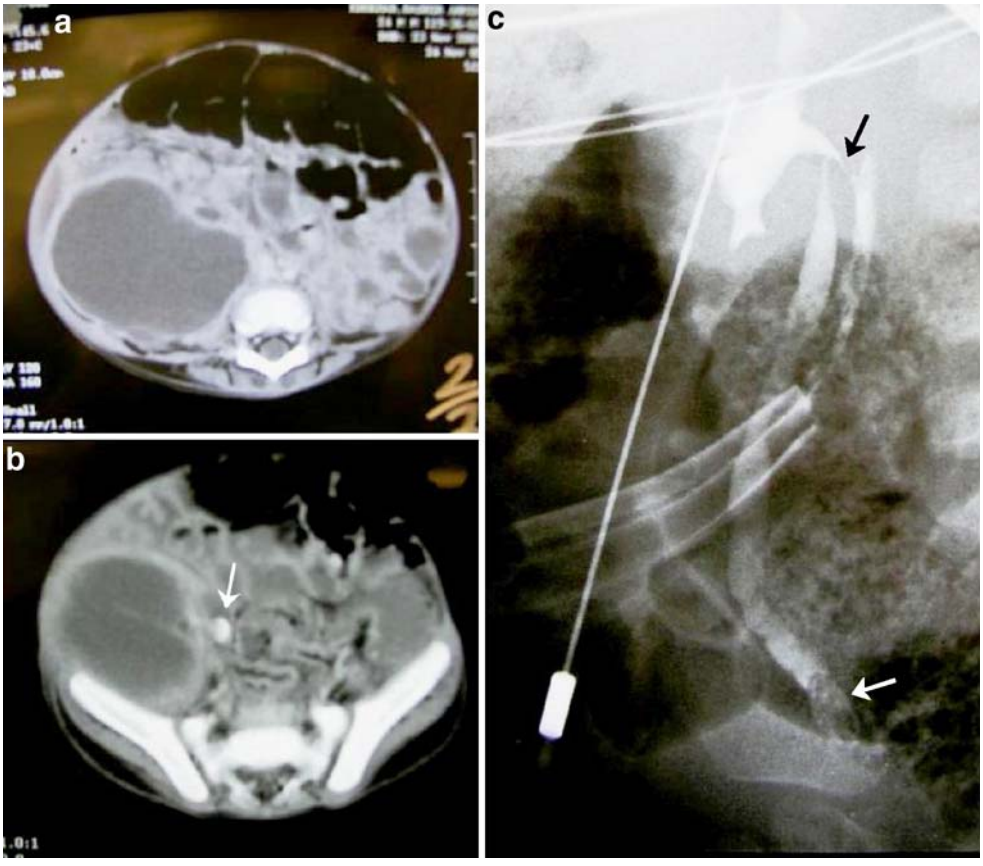

Fig. 1 **a**, **b** Computerized tomography (CT) scan showing a large abscess along the right psoas region and a lower ureteric stone (*white arrow*), **c** antegrade pyelogram demonstrating extravasation of contrast at the pelviureteral junction (*black arrow*) and the stone in lower ureter (*white arrow*)

electrolytes were within normal limits. He received broad-spectrum antibiotics (clindamycin: 40 mg/kg/day; amikacin: 15 mg/kg/day), and drainage of the abscess through a flank incision revealed 120 ml of thick yellowish pus. Culture of the pus revealed *Escherichia coli*. Blood and urine cultures showed no growth of microorganisms. On the third postoperative day urine started leaking through the incision, and an US-guided percutaneous nephrostomy was performed. A contrast study showed extravasation of contrast at the ureteropelvic junction and a 1.5-×1.5-cm stone in the right lower ureter (Fig. 1c). The leakage of urine stopped, and the patient became apyrexial. Subsequent investigations revealed normal sweat chlorides, low-fasting blood sugar (45 mg/dl), and normal random blood sugar (120 mg/dl). The patient had nocturnal hypoglycemic episodes and required carbohydrate supplements. An arterial blood sample revealed lactic acidosis (pH 7.3; lactate: 5.2 mmol/l, normal: 0.5–1.6 mmol/l; base excess: −4.0 mmol/l, normal: −0.2 ± 2.0 mmol/l). Serum proteins, liver function tests, and serum cholesterol were normal, whereas serum triglycerides and total lipids were elevated to 350 mg/dl (normal: 46–236 mg/dl) and 1234 mg/dl (normal: 500–1000 mg/dl), respectively.

An open ureterolithotomy and liver biopsy were performed. Histological analyses of the liver showed minimal fibrosis and a universal distension of hepatocytes by glycogen and fat when stained with periodic acid-Schiff (PAS). Chemical analysis of the stone revealed a mixture of calcium (20%), oxalate (60%), phosphate (15%), and urate (5%). Results of further investigations were as follows. Neutrophil function tests [phagocytic index, nitroblue tetrazolium (NBT) test] were normal; serum uric acid was raised (8.2 mg/dl; normal: 6.5–7.5 mg/dl); sodium, potassium, calcium, phosphate, and parathyroid hormone levels were normal; urinary pH was 6.5; microscopy of urine sediment showed calcium oxalate and urate crystals. A 24-h urine sample revealed hypocitraturia (0.8 mg/kg/day; normal: >2.0 mg/kg/day), hyperuricosuria (15.5 mg/kg/day; normal: <10.7 mg/kg/day), hypercalciuria (7 mg/kg/day; normal: <4 mg/kg/day), and hyperoxaluria (2.9 mg/kg/day; normal: 0.57 mg/kg/day). Allopurinol (50 mg/day) and potassium sodium citrate (Uralyt-U; 5 gm/daily) were administered with carbohydrate and protein supplements on a regular basis. At the 6-month follow-up, the patient was stone-free, normoglycemic, and gaining weight.

Discussion

Nephrocalcinosis and nephrolithiasis are not uncommon conditions in patients with GSD-1, with a reported incidence of up to 65% in adults with GSD-1 [8]. Renal calcification or kidney stones were observed in 14% of

children with a mean age of 4 years followed in a collaborative European study [2]. The development of a calcium oxalate stone and the complications reported here in a patient 2 years of age is unusual. It is worth mentioning – although it may just be a coincidence – that Pakistan, is located in a region known as the stone belt [7]. The contributing factors leading to urolithiasis can be chronic acidosis, hyperuricemia, hypocitraturia, and hypercalciuria due to the delay in diagnosing GSD-1 [1, 3, 4, 6]. Hyperuricemia and hypercalcemia were reported in 29–33% of the patients followed in the European study [2]. Restaino et al. studied 11 patients with GSD-1 (age range: 2–24 years), and renal calculi were present in five of these (45%): five of the 11 patients had hypercalciuria, and nine patients had impaired renal acidification; low urinary citrate concentration was observed in all of the patients [4]. Urinary citrate is a strong chelator of calcium oxalate and calcium phosphate, and low urinary citrate is a recognized cause of calcium oxalate nephrolithiasis [3, 4, 8, 9]. Hyperoxaluria of renal and dietary origin in malnourished children can also promote calcium oxalate stone formation [9]. Severe acidosis in patients with GSD-1 influences urinary citrate excretion (by reduced uptake by the proximal tubular cell in intracellular acidosis and by directly influencing the proximal tubular transport of citrate by the brush border membrane in response to a low luminal pH) and causes bone dissolution and hypercalciuria [10]. In malnourished children, low oral calcium absorption can allow oxalate to be absorbed, and this may explain the hypercalciuria and hyperoxaluria in the reported patient. Periodic abdominal US is recommended in patients with GSD-1 to detect renal calcification and liver tumors [6, 11]. There is some evidence that optimal metabolic control, alkalization of urine, and citrate supplementation may be beneficial in preventing or ameliorating nephrocalcinosis and the development of urinary stones in patients with GSD-1 [2, 11].

Stones in the urinary tract can cause obstruction and may be a source of infection. Disruption of the ureteropelvic junction and the development of psoas abscess secondary to an impacted ureteric stone are rare [7]. Psoas abscesses in infants and children are usually due to primary infection of the retroperitoneal lymph nodes by *Staphylococcus aureus* [12]. Secondary psoas abscesses can occur due to trauma, peritonitis, and gastrointestinal or renal disease [13]. Psoas abscesses of renal or ureteric etiology are unusual in children [14, 15]. A literature review of psoas abscesses of renal etiology included only one child, and in a series of 104 cases of non-tuberculous psoas abscesses in children, none had underlying renal pathology [12, 16]. To our knowledge, this is probably the first case of pyogenic psoas abscess secondary to ureteric lithiasis in a young patient with GSD-1. Given the variable data available on the incidence of nephrocalcinosis and nephrolithiasis in patients with GSD-1, it is difficult to propose definitive recommendations regarding the age at which screening should be initiated. Following the diagnosis of GSD-1, yearly US of the abdomen and regular estimations of urine sediment, urine creatinine, calcium, and citrate, as suggested by European study on glycogen storage disease type I (ESGSD I), are recommended.

In conclusion, we report a case of psoas abscess secondary to a complicated calcium oxalate ureteric stone in a 2-year-old child with GSD-1. Children with GSD-1 are at increased risk for nephrocalcinosis and urolithiasis. Regular abdominal sonography and optimal metabolic control from an early age may delay or prevent urolithiasis in patients having GSD-1.

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