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CLINICAL QUIZ

Failure to thrive and nephrolithiasis in a boy with congenital cyanotic heart anomaly—questions

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Case summary

A 3-year-old boy from Kosovo was referred to us for evaluation of failure to thrive and an episode of macroscopic hematuria. He was the son of healthy non-consanguineous parents. His past medical history was notable as he had been born with a cyanogenic heart anomaly (truncus arterious communis) which had been treated medically with furosemide and captopril until successful cardiac surgery, performed at 2 years of age. Following cardiac surgery, echocardiography with Doppler studies confirmed a successful repair, with no mixing of the arterial and venous blood. His cyanosis resolved and medical treatment was discontinued. Despite this apparently good outcome, he failed to thrive.

Upon admission to our unit, the boy was tachypneic, dehydrated and listless, with significant growth retardation [height 80 cm (−4.5 SD; weight 9.0 kg (−4.9 SD)]. There was no cyanosis. Cardiac examination revealed a systolic murmur, grade III/IV, at the precordium. Ultrasound of the abdomen revealed no hepatosplenomegaly, and both kidneys were of normal size without dilatation of the pelvicaliceal system. A single non-obstructive stone measuring 10 mm was seen in the lower pole of the left kidney (Fig. 1) without evidence of generalized nephrocalcinosis. A repeat echocardiogram confirmed adequate repair of the cyanotic anomaly.

Laboratory investigations included a full blood count, revealing: hemoglobin 13.4 g/dl, erythrocytes 4.66 × 10^{12}/l, leukocytes 8.6 × 10^{9}/l, hematocrit 42.7 vol%, platelets 344 × 10^{11}/l. The results of the arterial blood gas analysis were: pH 7.12, pCO_2 2.45 kPa, pO_2 12.7 kPa, O_2 saturation 98%. Serum electrolytes and liver and bone biochemistry test results were: HCO_3 10.1 mmol/l, urea 4.1 mmol/l, creatinine 9 μmol/l, uric acid 116 μmol/l, Na 145 mmol/l, K 3.3 mmol/l, Ca 2.23 mmol/l, P 0.74 mmol/l, Mg 1.0 mmol/l, Cl 115 mmol/l, alkaline phosphatase 762 U/l, parathyroid hormone 33.3 pg/ml (normal 10–65), glycemia 4.6 mmol/l, total protein 78 g/l,

The answers to these questions can be found at http://dx.doi.org/10.1007/s00467-011-1790-4.

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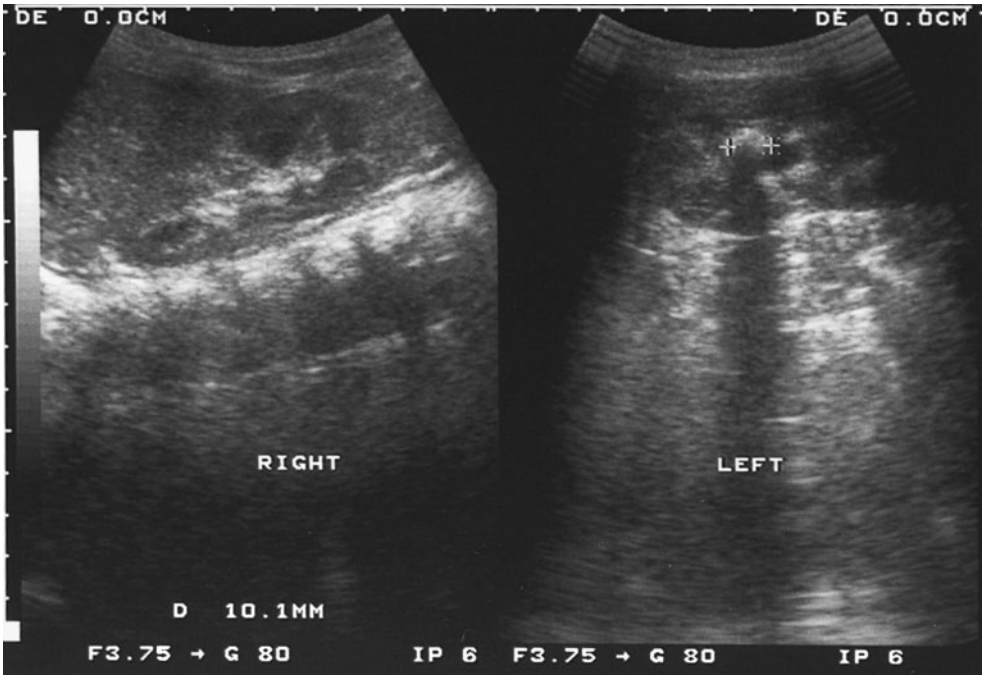

Fig. 1 Non-obstructive calculus in the left kidney

albumin 49 g/l, bilirubin 7 μmol/l, aspartate aminotransferase 32 U/l, alanine aminotransferase 17 U/l, glutamic oxaloacetic transaminase 26 U/l, lactate dehydrogenase 570 U/l, creatine phosphokinase 20 U/l.

The spot urine examination revealed: protein 2+ (1.07 g/l, electrophoresis of urinary proteins showed complete tubular proteinuria), blood 1+, pH 7.17 (with electrode). Urinary electrolytes were as follows: Na 58 mmol/l, K 37 mmol/l, Cl 56 mmol/l. The urinary calcium:creatinine ratio was elevated at 1.82 mmol/mmol (normal <0.70). Tubular reabsorption of phosphate and uric acid was 70 and 78%, respectively. The nitroprusside reaction for urinary cystine was negative. The urinary oxalate:creatinine ratio was 215 mmol/mol (normal 35–126), urinary citrate:creatinine ratio was 80 mmol/mol (normal 150–1007), urinary glycolate: creatinine ratio was 24 mmol/mol (normal 17-103), and urinary glycerate was non-detectable. There was a generalized hyperaminoaciduria.

In addition to the above clinical findings, the findings of an ophthalmological examination were normal, and audiologic examinations excluded any sensorineural deafness. The patient was treated with oral potassium and alkali supplementation (K-citrate 10 mmol/day+$NaHCO_3$ 20 mmol/day). At review 3 months later, the acid–base status, serum phosphate and uric acid had normalized as had the urinary excretion of proteins, oxalate, citrate, amino acids, calcium, phosphate and uric acid.

Questions

1. What is diagnosis of his tubular dysfunction?
2. What is the link between a cyanotic heart anomaly, failure to thrive and nephrolithiasis?
3. What is the explanation for the proximal renal tubular abnormalities?