A rare cause of urolithiasis in an infant: Questions

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A 13-month-old boy was admitted with red-brown spots on his diaper. He was a term infant born 3600 g and there was no prenatal feature in his history. His mother and father were first degree cousins; none of the family members had any kidney disease. His height was 79 cm (50–75%) and weight 10.1 kg (25–50%). There was no abdominal tenderness on palpation. Physical examination was unremarkable. His laboratory examination results were as follows: blood urea nitrogen 9.7 mg/dL (N : 9–23 mg/dL), serum creatinine 0.17 mg/dL (N : < 0.5), sodium 141 mEq/L (N : 136–145), potassium 4.7 mEq/L (N : 3.5–5.1), calcium 10.2 mg/dL (N : 9.0–11.0), phosphorus 5.1 mg/dL (N : 2.5–5.5), magnesium 2.2 mg/dL (N : 1.7–2.5), bicarbonate 23.1 mmol/L (N : 20–31), and serum uric acid 3.6 mg/dL (N : 3.4–7.0). On urinalysis, pH 5, density 1017, protein trace, hemoglobin 2+, and 132 erythrocytes/HPF were seen on microscopic analysis of the sediment. On ultrasonography, 3–4 hyperechogenic stones, the largest having a diameter of 6 mm on the middle and upper calyces of the left kidney and a stone with a diameter of 5 mm on the middle calyx of the right kidney, all having echogenic twinkling

artifact were seen. No radiopaque stone was found on abdominal X-ray. Urine calcium/creatinine was 0.1 mg/mg (N : < 0.6), urine oxalate/creatinine: 0.02 mg/mg (N : < 0.11), urine uric acid/creatinine: 0.84 mg/mg (N : < 1.9), urine magnesium/creatinine: 0.15 mg/mg (N : > 0.13), and urine citrate/creatinine: 1.41 mg/mg (N : > 0.42). Urinary excretion of cystine was also within normal ranges. A therapy with potassium citrate in addition to hydration and dietary recommendations were given. On follow-up ultrasonographic evaluations, an increase in the number of stones was detected. A kidney computerized tomography scan done at the age of 3.5 years showed 7–8 stones in the renal pelvis (the largest having a diameter of 5 mm) and 4–5 stones on the lower calyx (the largest having a diameter of 5 mm) on the left kidney, 3–4 stones in left distal ureter (the largest having a diameter of 4 mm) and two stones with 2 mm in the bladder. Afterwards, a percutaneous nephrolithotomy operation was performed, in which a large number of milimetric stones were removed. Chemical analysis of the stones yielded 6-amino-2,8-purinedione.

The answers to these questions can be found at <https://doi.org/10.1007/s00467-020-04910-y4>.

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Questions

1. What is your preliminary diagnosis?
2. Which exam would you want to elaborate the diagnosis?
3. How should this patient be managed?

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