Uncommon cribfellows: an infant with hypercalcemia, nephrocalcinosis, and acidosis: Questions

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Case

A 2-week-old girl presented with emesis and failure to thrive. She was treated with ranitidine and lansoprazole for presumed gastroesophageal reflux. Her birth weight was 3.5 kg (70th percentile). Physical exam was notable for a weight of 3.42 kg (30th percentile) but was otherwise unremarkable. Laboratory studies at 5 weeks of age revealed a serum sodium level of 136 (normal 133–140) mmol/L, potassium level of 4.1 (normal 4.1–5.8) mmol/L, chloride level of 105 (normal 96–106) mmol/L, carbon dioxide concentration of 18 (normal 20–26) mmol/L, creatinine level of 0.2 mg/dL and calcium level of 11.7 (normal 9.3–10.7) mg/dL. Over the next 2 months she had persistent emesis and poor weight gain. Serial evaluation of her electrolytes over that time period demonstrated serum calcium and bicarbonate levels which ranged from 11.2 to 11.7 mg/dL and from 17 to 20 mmol/L, respectively. Urinalysis showed a specific gravity of 1.001, pH of 7.5 and no blood or protein.

At 4 months of age her weight was 4.8 kg (< 3rd percentile). Further evaluation of the hypercalcemia and acidosis revealed a phosphorus level of 6.0 (normal 4.8–8.2) mg/

dL, magnesium level of 2.0 (normal range 1.5–2.5) mg/dL, an intact parathyroid hormone level of < 3.4 (normal range 9–56) pg/mL, a 25-hydroxyvitamin D level of 38.2 (sufficient 30–80) ng/mL and a 1,25 hydroxy vitamin D level of 29.1 (normal range 19.9–79.3) pg/mL. The urine calcium to creatinine ratio was 0.8, and the urine anion gap ($\text{Na}^+ + \text{K}^+ - \text{Cl}^-$) was + 16.7 mmol/L. Renal ultrasound demonstrated normal-sized kidneys (length: right 5.5 cm, left 5.3 cm) with bilateral medullary nephrocalcinosis. At this time, potassium citrate therapy was initiated, and the dose was titrated to a final dose of 3.5 mEq/kg body weight/day over the next year. The gastrointestinal symptoms significantly improved and she had appropriate weight gain. Notably the serum calcium levels dramatically improved after the initiation of potassium citrate therapy, ranging from 9.7 to 10.9 mg/dL. At 15 months of age, her weight was 9.8 kg (50th percentile), serum bicarbonate level was 19 mmol/L and her serum calcium was 10.5 mg/dL.

Questions

1. What is the differential diagnosis for nephrocalcinosis in infancy?
2. Which of these conditions are associated with hypercalcemia?
3. Which of these diagnoses are associated with acidosis?

Compliance with ethical standards

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