An infant with hypercalcemia and hyperammonia: inborn error of metabolism or not? Questions

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Received: 20 June 2018 / Revised: 26 June 2018 / Accepted: 28 June 2018 / Published online: 19 July 2018
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Keywords Hypercalcemia · Hyperammonia · Distal renal tubular acidosis

Case report

A 44-day-old patient was admitted to our emergency room due to complaint of feeding intolerance of 1 week duration. She was born 2940 g weight at 38 weeks gestation. There were no specific findings on her history. There was no history of renal disease in her family and no consanguinity. On physical examination, body weight was 3270 g (3–10 percentile), body height 53 cm (10–25 percentile), head circumference 36 cm (10 percentile), body temperature 36.5 °C, respiratory rate 36/min, and heart rate 122/min. Mild dehydration was detected on her physical examination. On laboratory examination, urea was 23 mg/dL, serum creatinine 0.4 mg/dL, serum sodium level 140 mmol/L (normal 135–145 mmol/L), serum potassium level 2.3 mmol/L (normal 3.5–5 mmol/L), serum chloride level 113 mmol/L (normal 98–107 mmol/L), serum calcium level 13 mg/dL (normal 8.5–10.7 mg/dL), and serum phosphorus level 5.9 mg/dL (4–5.6 mg/dL). On urine examination, pH 8 with blood gas analysis was pH 7.20 and HCO₃ 11.3. Anion gap analysis was 15; urine anion gap was 22. Serum ammonia level was 187 mcg/dL (normal 45–80). Parathyroid hormone (PTH) level was 0.6 pg/mL (normal 14–72 pg/mL) and 25

OHD vitamin level was 51 ng/mL. Abdominal ultrasonography revealed urolithiasis. Hearing test was normal. Urine and blood amino acid analysis and urine organic acid analysis were normal and urine for reducing substances was negative. Tubular phosphate reabsorption test was 92%. Urine sodium, potassium, and chloride levels were 11, 25, and 14 mmol/L, respectively. Urine calcium/creatinine ratio was 2.6 mg/mg which was indicating hypercalciuria. Bone radiographs were normal.

Questions?

1. What is the diagnosis of this patient?
2. What is the differential diagnosis of hyperammonia in infant?
3. What is the differential diagnosis of hypercalcemia?
4. What is the cause of hyperammonemia and hypercalcemia in this patient?

Compliance with ethical standards

Conflict of interest The authors declare no conflict of interest.

The answers to these questions can be found at <https://doi.org/10.1007/s00467-018-4022-3>.

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