



When salt is needed to grow: Questions

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Case summary

A 50-day-old girl was referred for a history of recurrent vomiting, poor feeding and moderate failure to thrive. She was born by a caesarean section at 41 + 5 weeks of gestation, from non-consanguineous parents. Her birth weight was 3430 g and she was fed with breast milk and formula. Upon admission, her weight was 3700 g (3rd percentile) and blood pressure was normal. Physical examination showed mild dystrophy with poor representation of subcutaneous fat, normal external genitalia and age-appropriate psychomotor development.

Laboratory tests showed mild hyponatraemia (130 mEq/L) and hyperkalaemia (6 mEq/L), normal plasma creatinine (0.26 mg/dl) and metabolic acidosis (pH 7.23, pCO₂ 53 mmHg, HCO₃⁻ 21.8 mmol/L, lactic acid 7.5 mmol/L). Potassium and sodium urinary fractional excretion were 23% and 0.98%, respectively. In the urine sample, no proteinuria, white blood cells, bacteria or haematuria were detected. Abdominal ultrasound results were normal.

Due to the acidosis with persistence of poor feeding and vomiting, a hydration with an intravenous physiological solution was empirically started. While metabolic acidosis was

resolved with the infusion, hyponatraemia and hyperkalaemia persisted. Further analysis revealed a significant increase of plasma renin activity and aldosterone (respectively, > 500 μUI/mL, normal value 4–44; > 1000 ng/dL, normal value 30–50).

Questions

1. What is the most likely diagnosis?
2. What would be an alternative diagnosis?
3. What is the treatment and prognosis of the disease?

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Compliance with ethical standards

Conflict of interest The authors declare that they have no conflict of interest.

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The answers to these questions can be found at <https://doi.org/10.1007/s00467-020-04647-8>.

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