Clinical Quiz
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A Rare Case of Renal Salt Wasting Associated with Metabolic Acidosis and Hyperkalemia

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Abstract

In the evaluation of patients presenting with chronic hyperkalemia hyponatremia and normal anion gap acidosis, pseudohypoaldosteronism types I and II are within the spectrum of differential diagnosis. Recognition of these patients is important to prevent inappropriate mineralocorticoid therapy. Using clinical scenarios we aim to illustrate clinical mimics and dissimilarities to differentiate theses disorders.

Keywords: Hyperkalemia; Hyponatremia; Metabolic Acidosis; Pseduohypoaldosteronism.

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Case study

A 14-year old female with 5-year history of diabetes mellitus-type 1 was referred for the management of hyperkalemia. She complains of light-headaches. Past medical history was significant for recurrent urinary tract infections during childhood. Her father had similar problems with hyperkalemia when he was young. On examination, her blood pressure is 126/79 mmHg supine and 124/81 mmHg standing. Laboratory evaluation revealed sodium 120 mEq/L, potassium 6.3 mEq/L, chloride 105 mEq/L, bicarbonate 15 mEq/L, creatinine 0.9 mg/dL, and BUN 21 mg/dL Estimated glomerular filtration (eGFR) using Schwartz equation is 87.7 mL/min/1.73 m^2. Urine dipstick showed pH 5.2, specific gravity 1.014, trace protein and small blood. The urine sediment showed 5-10 red blood cells/HPF with no cellular or granular casts. Urine culture was sterile. A random urine protein-to-creatinine ratio was 0.56. Urine electrolytes showed sodium 105 mEq/L, potassium 8 mEq/L, chloride 88 mEq/. Plasma renin activity and serum aldosterone level were both increased. A renal ultrasound was normal.

Which of the following statements about this patient is correct (select all may apply)?

A. Skin rash and recurrent pneumonia are likely present
B. Therapy to lower hyperkalemia and prevent salt-wasting is likely to decrease over time
C. The patient is likely to have type IV renal tubular acidosis due to diabetes mellitus-type 1.
D. The sweat chloride test in this patient is likely to be increased.
E. The clinical and laboratory findings in this patient are compatible with autodomal dominant form of pseudohypoaldosteronism type 1.
F. Gordon's syndrome

Comments

The correct answers are B and E. This patient present with evidence of hyperkalemia, normal anion gap metabolic acidosis, renal-salt wasting, impaired renal function, and elevated plasma renin level and serum aldosterone concentration. In addition, patient's father had increased serum potassium

level that later resolved, suggesting a familial disorder. These findings best fit with autosomal dominant, type I pseudohypoaldosteronism (PHA-I) or renal PHA-I. This disorder is caused by mutations on the gene encoding for the aldosterone receptor (1-4). The clinical manifestations tend to resolve over time. In contrast, autosomal recessive PHA-I (systemic PHA-I) is caused by inactivating mutation of the epithelial sodium channel (ENaC) and is characterized by generalized salt loss in many organs including kidneys, lung, colon, salivary and sweat glands (5) Extrarenal manifestations including skin rush and frequent bouts of pulmonary infections (Choice A) are features of the autosomal recessive Type I PHA, in which the defect is at the level of the epithelial sodium channel. The skin involvement in this form of the disease has been attributed to increased sodium concentration making choice D unlikely (6).

The development of type IV renal tubular acidosis (Choice C) due to diabetes mellitus-type 1 is usually associated with hypertension and low level of plasma renin, and reduced serum aldosterone concentration (7). Familial Type II pseudohypoaldosteronism and Gordon's syndrome or familial, is characterized by autosomal dominant transmission of high blood pressure, hyperkalemia, hyperchloremic metabolic acidosis without renal failure (1,6,7). Associated findings include low plasma renin activity and serum aldosterone level, short stature, stiff spine and deformities of hands and feet. Affected patients respond well to thiazide, suggesting a primary defect in potassium secretion is in the distal nephron (8,9).

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