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Rare cause of hyperkalemia in infant: Report of case of systemic Pseudohypoaldosteronism Type 1

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Abstract

Our patient diagnosed as systemic PHA type I which is characterized by salt wasting crises, hyperkalemia, metabolic acidosis, with markedly elevated plasma aldosterone and high plasma renin activity and the diagnosis confirmed by gene study. Treatment of PHA type 1 comprises adequate hydration, replacement of salt loss, and correction of hyperkalemia and acidosis.

PHA1 should be kept in mind as a rare cause of electrolyte emergency in infants.

Background

PHA type I was reported by Cheek and Perry in 1958 [1]. It is a hereditary salt-wasting syndrome which usually starts in early infancy and is characterized by a diminished renal tubular responsiveness to aldosterone, resulting in hyponatremia, hyperkalemia, metabolic acidosis. Markedly elevated plasma aldosterone and high plasma renin activity.

Case presentation

2-month-old baby girl presented with a history of poor feeding, vomiting and loose motion. Physical examination showed an ill looking, severe dehydration and abdominal distention, blood pressure: 88/53mmHg, heart rate:168/min, respiratory rate: 54/min, SPO2: 99 %, temperatutre:37 ^{0}C

She was born full-term with birth weight 2.9kg to consanguineous parents of Saudi origin, with no complication during pregnancy and delivery.

She had normal genitalia with no clitromegally labial fusion or hyperpigmentation and no palpable abdominal mass.

Investigation

Hyponatremia NA: 124mEq/L (normal range:135-145mEq/L), hyperkalemia K:9.8mEq/L (normal range:3.5-5.5mEq/L), chloride : 98 meq/L (normal range: 96-106mEq/L), metabolic acidosis in venous blood gas PH: 7.32, PCO2:30.8, HCO3:16.

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Due to low serum sodium and high serum potassium RTA type 4 and hypoaldosteroinsm were suspected. Renal function test showed: creatinin: 0.5 mg/dl (normal range:0.3-0.7mg/dl), BUN: 13 mg/dl (normal range: 4-15mg/dl)

Urine culture and analysis revealed no growth and Renal ultrasound showed no deformity.

Serum aldosterone: 132ng/dl (normal 2-70ng/dl), plasma renin activity: 55.0 ng/ml/hr (normal range: 2-35ng/ml/hr) compatible with hyperreninemic hyperaldosteronism.

Normal serum cortisol: 849 nmol/L (normal >500nmol/L), so 17 hydroxyprogestrone was not requested and congenital adrenal hyperplasia was excluded clinically and biochemically.

Gene study revealed a homozygous variant of uncertain significance in SCNN1A

Treatment

Hyponatremia and dehydration were managed aggressively with large fluid and sodium intake. Sodium balance was only achieved with 40 mEq/kg/day of sodium chloride. Hyperkalemia was managed with glucose and insulin infusion, calcium gluconate, sodium bicarbonate and nebulized Salbutamol then tapered and stopped and now potassium controlled with high dose of sodium polystyrene sulfonate.

Outcome and follow up

The patient currently 9 months old and her growth, in weight and length are below the third percentile. At high doses of sodium bicarbonate, 3% saline and sodium polystyrene sulfonate. She has been admitted to the pediatric intensive care unit frequent times due to recurrent respiratory tract infection.

Discussion

The differential diagnosis of severe salt wasting in infancy usually includes conditions such as congenital adrenal hyperplasia, PHA, aldosterone biosynthesis defect, cystic fibrosis and chronic kidney injury with salt wasting. Pseudohypoaldosteronism (PHA) type I is a rare salt wasting syndrome caused by aldosterone resistance with two distinct forms: an autosomal dominant type, caused by a mutation of the mineralocorticoid receptor with sodium wasting in the kidney, and an autosomal recessive form, resulting from mutations in one of the 3 subunits (alpha, beta, or gamma) of the epithelial Na+ channel (ENaC) [2,3].

The autosomal dominant form, also known as renal PHA type I, is confined to the kidney with less severe disease and no systemic involvement. In the autosomal recessive form, also known as systemic PHA type I, patients have a multisystem disease with salt loss from organs expressing ENaC, including the kidneys, salivary and sweat glands, and the colon. As a result of decreased sodium-dependent liquid absorption in the lungs, children often develop pulmonary symptoms including congestion, wheezing, and recurrent pulmonary infections.

Patients typically present shortly after birth with electrolyte findings mimicking adrenal crisis. The most common age at presentation is from 5 days to 7 months and the syndrome is more common in males [4].

The electrolyte abnormalities of hyponatremia, hyperkalemia, and metabolic acidosis are similar in CAH and PHA. However, in contrast to the CAH, serum cortisol and aldosterone levels are elevated in PHA indicating normal glucocorticoid production and resistance to aldosterone activity. The patient presented here clinically fits the picture of systemic PHA1. Systemic PHA1 is also associated with other clinical features including skin changes [5], increased risk of respiratory infections [6], polyhydramnios [7], and cholelithiasis.

A mutation in the subunit genes (SCNN1A, SCNN1B, SCNN1G) of epithelial sodium channel and the NR3C2 gene encoding the mineralocorticoid receptor, result in systemic PHA1 and renal PHA1, respectively [8]. In our case, gene mutation was detected and the diagnosis of type 1 PHA was confirmed.

Sick infants should be treated urgently; saline infusion to correct volume depletion and hyponatremia, and ion exchange resin or emergency dialysis to control hyperkalemia. After recovery from acute crises, patients can be managed with high dose of oral salt supplements and chronic ion exchange resin therapy, and most patients require lifelong salt supplementation [2]. Long-term treatment consists of oral sodium chloride supplementation for hyponatremia, oral sodium bicarbonate for metabolic acidosis as well as additional sodium supplementation, and oral Kayexalate to prevent hyperkalemia. Infants that have difficulty tolerating these treatments may require gastrostomy tube placement [9]. Outpatient follow-up of such patients involve close contact with family for monitoring of general health, electrolytes and weight-gain.

Learning points

(1) The differential diagnosis of severe salt wasting in infancy is congenital adrenal hyperplasia, PHA, aldosterone biosynthesis defect, cystic fibrosis and chronic kidney injury with salt wasting.

(2) Pseudohypoaldosteronism characterized by three essential features:

(1) Hyperkalemia, (2) metabolic acidosis, (3) abnormally elevated plasma aldosterone concentration

(3) Two different forms of pseudohypoaldosteronism1 can be distinguished on the clinical and genetic level, showing either renal or systemic form of mineralocorticoid resistance.

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