Severe Systemic Pseudohypoaldosteronism Type 1
5 years of evolution

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Introduction

Type 1 pseudohypoaldosteronism (PHA-1) is a rare syndrome of unresponsiveness to aldosterone, expressed in two forms: renal PHA-1 and systemic PHA-1. In renal PHA-1 the mineralocorticoid resistance is isolated to this organ, so the phenotype is milder and often improves spontaneously due to proximal nephron maturation. Systemic PHA-1 results from autosomal recessive mutations in the genes encoding α, β and γ subunit of epithelial sodium channel (ENaC) that exists in multiple organs (kidney, colon, lung, salivary and sweat glands), and therefore the phenotype is severe. The diagnosis is established by the presence of high levels of serum aldosterone and plasma renin activity associated with hyponatremia, hyperkalemia and metabolic acidosis. Symptoms manifest during the first week of life, requiring prolonged hospitalizations and often lifelong high-salt replacement therapy. The mortality rate is high, especially during the neonatal period.

Case Report

Male; Born at full term; Birth weight of 3010g (10-25% percentile)
No parental consanguinity; 6-year-old sister with Chediak-Higashi syndrome

10th day

Hypovolemic shock NICU

Dehydration

Normal saline bolus

To correct dehydration

Metabolic acidosis

Calcium gluconate

To control hyperkalemia

Sodium bicarbonate

Rectal cation-exchange resin

Numerous life-threatening crises

Congenital Adrenal Hyperplasia was suspected and he started hydrocortisone (max 200 mg/kg/day) and fludrocortisone (max 1.5 mg/day)

In sick newborns a repeated assessment may be required for the diagnosis

Initial 17-OHP was falsely raised (10.76 ng/mL)

Homonymous mutation of intron 3 of the SCNN1A gene c.1052+2dupT

Conclusions:

We report this severe PHA-1 case with poor initial prognosis but with favorable evolution. Management of PHA-1 is challenging since there are no evidence-based recommendations.

Current with 5-years-old

- Failure to thrive (height -2.61SDS, weight -3.53SDS)
- Normal development
- Keeps only oral 20% saline supplement 43mEq/kg/day
- Symptoms became less severe and less frequent with increasing age

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Medication by nasogastric tube until 2-years

- Hydrochlorothiazide from 1.5 to 4-years
- Fludrocortisone reduced from 1.5 to 3-years
- Cation-exchange resin reduced until 3.5-years
- Oral 20% saline ranged from 28 to 55mEq/kg/day

5-months-old

Discharge

Fludrocortisone 1.5mg/day + 20% saline supplement 33mEq/kg/day + cation-exchange resin 1g/kg 6 times/day

Metabolic acidosis

Sodium bicarbonate

To control hyperkalemia

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