

Congress Abstract

Systemic Pseudohypoaldosteronism Type 1: From Salt-Wasting Crisis to Genetic Confirmation

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ABSTRACT

Background: Systemic pseudohypoaldosteronism type 1 (PHA1B; OMIM 264350) is a rare autosomal recessive disorder caused by biallelic pathogenic variants in SCNN1A, SCNN1B, or SCNN1G, which encode epithelial sodium channel (ENaC) subunits. Aldosterone resistance causes neonatal salt wasting, hyponatremia, severe hyperkalemia, metabolic acidosis, and potentially multiorgan dysfunction. Because the presentation mimics salt-wasting congenital adrenal hyperplasia (CAH) and sepsis, early differentiation is critical. We report a genetically confirmed, life-threatening neonatal case of PHA1B.

Case Presentation: A full-term girl (38 weeks + 2 days) was admitted on day 8 of life in critical condition with an 11% weight deficit, severe hypoxic-ischemic encephalopathy, subarachnoid and intraventricular hemorrhage with cerebral edema, bilateral pneumonia, and grade II respiratory failure. In the pediatric intensive care unit, respiratory failure progressed, requiring mechanical ventilation. Cardiac manifestations included bradyarrhythmia to 80 beats/min, ventricular ectopy, and complete right bundle branch block. Laboratory tests showed K⁺ 13.6 mmol/L, Na⁺ 110 mmol/L, pH 7.20, and base excess -17.4 mmol/L.

Salt-wasting CAH was initially suspected; however, hydrocortisone and fludrocortisone produced no clinical or biochemical response. Cortisol (27.53 µg/dL), 17-hydroxyprogesterone (1.2 ng/mL), and adrenocorticotrophic hormone (5.0 pmol/L) were within reference ranges, while plasma aldosterone was markedly elevated at 10,355 pg/mL (reference range, 28-376 pg/mL), supporting aldosterone resistance. Whole-exome sequencing identified a homozygous pathogenic nonsense variant in SCNN1A (exon 10; c.1474C>T; p.Arg492Ter), confirming systemic PHA1.

Management included mechanical ventilation, intravenous calcium, intensive enteral and intravenous sodium chloride replacement, antibacterial therapy, and peritoneal dialysis for refractory life-threatening hyperkalemia. Potassium decreased to 8.1 mmol/L, sodium increased to 128-135 mmol/L, and the metabolic crisis initially improved. Despite treatment, the patient died from progressive multiorgan failure and neurological complications.

Conclusion: PHA1 should be considered in neonates with severe hyponatremia, hyperkalemia, and metabolic acidosis, particularly when CAH markers are normal and glucocorticoid/mineralocorticoid therapy is ineffective. In this setting, markedly elevated aldosterone supports mineralocorticoid resistance. Prompt sodium replacement and emergency management of hyperkalemia, including early dialysis when medical therapy fails, are essential. Molecular testing confirms the systemic form, informs prognosis, and enables family genetic counseling.

Keywords: Pseudohypoaldosteronism Type 1; Hyperkalemia; Neonatal Salt Wasting; Hyponatremia
