Case Report

SALT-WASTING CONGENITAL ADRENAL HYPERPLASIA: A CASE REPORT

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ABSTRACT:

Salt-Wasting Congenital Adrenal Hyperplasia (SW-CAH) is a rare genetic disorder characterized by enzyme deficiencies affecting the adrenal steroidogenesis pathway, leading to a decrease in cortisol synthesis. Here presented a case of a 28-day-old male infant presented with symptoms of lethargy and feeding difficulties. Initial evaluations revealed severe electrolyte imbalances, including hyponatremia and hyperkalemia. Further investigations, including hormone tests, confirmed a diagnosis of SW-CAH. The management involved intravenous therapy with steroids, including hydrocortisone and fludrocortisone, to restore hormonal balance. Strict monitoring of sugar levels, blood pressure, serum electrolytes, and cortisol levels was crucial. The case highlights the importance of early diagnosis and intervention, particularly in regions with limited access to healthcare services. This report underscores the need for increased awareness of CAH, particularly SW-CAH, and the potential benefits of implementing newborn screening programs and establishing effective disease databases to improve early detection and management of this condition, potentially saving lives.

Keywords: Congenital Adrenal Hyperplasia, Hyponatremia, hyperkalemia, Glucocorticoids

INTRODUCTION

Congenital Adrenal Hyperplasia (CAH) is an autosomal recessive condition characterized by enzyme abnormalities that affect the adrenal steroidogenesis pathway, resulting in decreased cortisol synthesis. Depending on the kind and degree of the steroid interference, people may experience glucocorticoid, mineralocorticoid, and sexual corticosteroid modifications in production which necessitate substitute hormone treatment (1). Prolonged excessive stimulation of the adrenal cortex as a result of the activity of an enzyme necessary for cortisone synthesis causes a build-up of antecedents close to the enzyme's inhibited phase. The most prevalent type of CAH is brought on by steroid 21-hydroxylase deficiency resulting from CYP21A2 alterations (2).

Congenital Adrenal Hyperplasia is an autosomal recessive disorder that affects about 1 in 15,000 people and is usually caused by 21-hydroxylase insufficiency (3). Given that CAH is a hereditary biological condition, the larger CAH prevalence may be attributable to greater consanguinity, lesser diversity in genes, or additional genetic variables (such as hotspots for abnormalities). There is scarce data on the disorder, additionally, there are limited testing facilities, exacerbating the matter two-folds. Just by establishing an effective disease database and implementing a newborn screening plan, this issue can be addressed (4).

The prevalence of CAH may be directly impacted by cultural norms about consanguineous unions or isolation from the outside world in some regions. The number of confirmed cases and the actual prevalence of CAH could be impacted by the fact that newborn tests for the disease aren't always accessible in numerous impoverished nations (5).

Case Presentation

A 28-day-old male baby was brought to the emergency department with complaints of being reluctant to feed and being lethargic for two days. Initially, the baby was managed in another private hospital for the correction of serum electrolytes.

The baby was afebrile and had no sign of respiratory distress. The chest was clear on examination with no added sounds, and the cardiovascular and central nervous system was intact. The abdomen was soft and no tenderness was observed. The gut sounds were audible.

In the emergency department, serum electrolytes were repeated which showed serum magnesium of 2.4mg/dl, blood urea nitrogen 35mg/dl, serum creatinine 1mg/dl, serum sodium 118/dl and serum potassium of 9.3mg/dl. The patient showed hyponatremia and hyperkalemia. The initial management in the emergency department was intravenous fluids, calcium gluconate, salbutamol nebulization, Insulin and dextrose cocktail and K-oxalate to treat hyponatremia and hyperkalemia.

For further workup, the Adrenocorticotropic hormone, 17 OH progesterone level, Serum cortisol, and renin level were sent to the laboratory for further investigation. 25mg hydrocortisone was given after taking the laboratory samples. The emergency department suspected it was Salt-Wasting Congenital Adrenal Hyperplasia with hyperkalemia. For further management, the baby was then moved to the neonatology unit.

The laboratory profile of the hormones is given below in the Table 1.

S. No	Tests	Range	Results
1	Neonatal TSH	1.3 to 16uIU/ml	0.38 uIU/ml
2	17 OH progesterone	<100	>320 ng/ml
3	Cortisol	5 to 25	10.3 ug/dl
4	ACTH	112- 128	353 pg/ml
5	Renin	2.8- 39.9	>500 uIU/ml
6	Aldosterone	6 to 179ng/dl	84.2ng/dl

In the neonatology unit, the serum electrolytes were repeated which showed sodium 129, potassium 4.3, chloride 96 and urine and blood were sent for culture and sensitivity in which no growth was found. The medical management includes 5% dextrose at the rate of 15ml/hour, intravenous antibiotics cefotaxime 125mg every 8 hourly, injection of hydrocortisone 2mg every 6 hourly and injection of fludrocortisone acetate 0.1mg every 12 hourly.

The baby's condition improved with the management of intravenous steroid therapy. The baby was active and was able to take feed. The baby was discharged and was kept on steroids and the diagnosis was confirmed as Salt-Wasting Congenital Adrenal Hyperplasia.

DISCUSSION

Salt-Wasting Congenital Adrenal Hyperplasia is a rare disease with an uneven ratio of males and females. It additionally brought down the unequal gender proportion, having more females being diagnosed versus males, implying that before screening, certain males had possibly gone undiagnosed and died (6). The critical point is that male babies are usually undiagnosed at the initial visits. Aldosterone production is typically markedly impaired for those with salt-wasting congenital adrenal hyperplasia, necessitating the replenishment of mineralocorticoids as well. The increased production of ACTH levels in the blood causes hyperplasia of the adrenal gland because they are signaling the adrenal gland to produce the cortisol and other hormones which they are unable to produce. Salt wasting occurs due to the low level of aldosterone in the blood.

There is not much research on mineralocorticoid replacement, even though glucocorticoid therapy in SW-CAH is widely used to substitute lost glucocorticoids and manage excessive adrenal androgen (7). The Laboratory findings showed hyponatremia, hypoglycemia, and hyperkalemia with augmented renin activity, increased adrenocorticotropic hormone (ACTH), low aldosterone, low cortisol, and high 17 OH progesterone levels. Literature also says that the salt-losing variety of 21 hydroxylase deficiency is thought to be the most prevalent and dangerous variant, with almost no glucocorticoid synthesis and reduced aldosterone production that causes salt loss, inability to flourish, and possibly deadly low blood pressure and shock (8).

There should be strict monitoring of sugar levels, blood pressure, serum electrolytes and cortisol levels in these patients. In this scenario, the baby was lethargic due to low sugar, low blood pressure and cortisol levels in the

blood. To maintain the sugar level, 5% dextrose was continued at the rate of 15ml/hr. Injection hydrocortisone was prescribed to maintain the cortisol level in the body and fludrocortisone 0.1mg was given to maintain the sodium level in the body. Additionally, according to the literature, the infant has to begin substituting medication with steroids and sodium therapy (hydrocortisone and fludrocortisone), which will have a dramatic impact on their gradual physical and cognitive growth (9). The baby was discharged on oral hydrocortisone and fludrocortisone with regular follow-up and serum electrolytes, sugar and blood pressure monitoring.

CONCLUSION

Congenital Adrenal Hyperplasia is a rare autosomal recessive disorder characterized by enzyme deficiencies that affect the adrenal steroidogenesis pathway, leading to decreased cortisol synthesis. This can result in various hormonal imbalances and necessitate hormone replacement therapy. In our case, the baby was presented with hyponatremia and hyperkalemia. The hormone, 17 OH progesterone, cortisol and aldosterone were also abnormal. Glucocorticoid and mineralocorticoid therapy was started for the baby. Literature also says that SW-CAH is a severe form of CAH characterized by impaired aldosterone and cortisol production, leading to salt wasting. Treatment for SW-CAH involves hormone replacement therapy with steroids (hydrocortisone) and mineralocorticoids (fludrocortisone) to restore hormonal and electrolyte balance. Strict monitoring of sugar levels, blood pressure, serum electrolytes, and cortisol levels is essential for managing such patients.

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