Case Report

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Paediatrics Section

Congenital Adrenal Hyperplasia Characterised by Electrolyte Imbalance in a Male Neonate: A Case Report

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ABSTRACT

Congenital Adrenal Hyperplasia (CAH) is a group of autosomal recessive disorders of adrenal steroidogenesis, most commonly caused by 21-hydroxylase deficiency. The salt-wasting form of CAH represents a neonatal endocrine emergency. This is a case report of a four-day-old male neonate, who presented on day four of life with lethargy and poor feeding and the systemic examination revealed marked hyper pigmentation especially over the scrotal region but otherwise unremarkable. Investigations revealed hypoglycaemia, hyponatraemia, and hyperkalaemia and hyperpigmentation, however, the septic screen was negative. In view of persistent hypoglycaemia, hypotension and dyselectrolytemia together with hyperpigmentation and diagnosis of CAH was suspected and the baby was worked-up for the same. Biochemical evaluation revealed markedly elevated serum 17-hydroxyprogesterone levels, confirming the diagnosis of classical salt-wasting CAH. This condition is often mistaken for sepsis and the diagnosis is usually delayed. Prompt recognition and initiation of glucocorticoid and mineralocorticoid therapy led to rapid clinical stabilisation. This case highlights the diagnostic challenge of CAH in male neonates and emphasises the importance of early suspicion, timely biochemical testing, and urgent management to prevent mortality.

Keywords: Adrenal insufficiency, Hyperpigmentation, Hypoglycaemia, Newborn, 21-hydroxylase deficiency

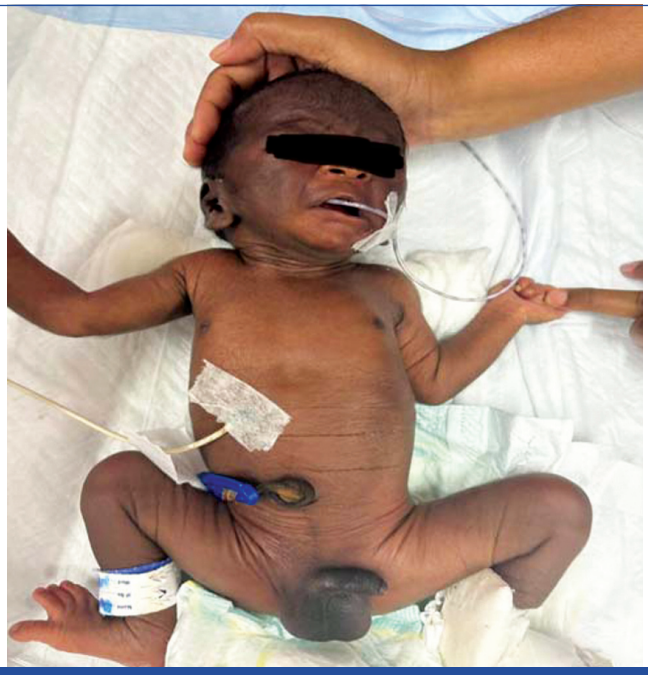
CASE REPORT

A four-day-old male neonate was admitted to the neonatal intensive care unit with complaints of poor feeding, lethargy, and abnormal movements noticed since day 3 of life. He was born at term by normal vaginal delivery with a birth weight of 2.8 kg. The antenatal and perinatal periods were uneventful. The parents were non-consanguineous, and there was no family history of neonatal deaths or known endocrine disorders.

On examination, the neonate appeared lethargic with clinical signs of dehydration. Vital parameters namely heart rate, blood pressure, respiratory rate and SpO_2 were monitored which revealed tachycardia {Heart Rate (HR)>160/min} and borderline hypotension (50/40 MAP 56 mmHg) Capillary blood glucose at admission was 19 mg/dL, indicating severe hypoglycaemia. External genital examination showed a normal male phenotype with a normally formed penis and bilaterally palpable testes with marked hyperpigmentation of the scrotum [Table/Fig-1]. No dysmorphic features or congenital anomalies were noted. Other systemic examination namely respiratory, cardiovascular were normal.

Initial laboratory investigations revealed hyponatraemia (serum sodium 132 mEq/L), hyperkalaemia (serum potassium 5.68–6.0 mEq/L), and persistent hypoglycaemia. Renal function tests were within acceptable neonatal reference ranges [Table/Fig-2,3]. In view of early neonatal onset of hypoglycaemia and electrolyte imbalance, adrenal insufficiency was suspected.

Further hormonal evaluation revealed a markedly elevated serum 17-hydroxyprogesterone level (>300 ng/mL), even after dilution, which was diagnostic of classical CAH due to 21-hydroxylase deficiency [Table/Fig-4]. Serum cortisol levels were inappropriately low for age. In view of hyponatremia, hyperkalemia, and dehydration, differential diagnoses considered included pseudohypoaldosteronism, adrenal hypoplasia, neonatal sepsis, and isolated hypoaldosteronism. Adrenal hypoplasia and pseudohypoaldosteronism were ruled out due to markedly



[Table/Fig-1]: Clinical photograph showing generalised hyperpigmentation with prominent pigmentation over genital region and scrotum in the neonate. (Patient identifiers masked).

Sample	Observed values (Day 4 of life)	Biochemical Reference value
Plasma random glucose	19 mg/dL	80 - 120 mg/dL
Serum electrolytes		
Sodium	132 mEq/L	136 - 145 mEq/L
Potassium	5.68 mEq/L	3.5 - 5.1 mEql/L
Chloride	107.2 mEq/L	96 - 106 mEql/L

[Table/Fig-2]: Laboratory report demonstrating severe hypoglycaemia along with associated electrolyte abnormalities.

Parameters	Observed values	Biochemical reference value
Renal function tests		
Serum urea	17.78 mg/dL	17.19-49.2 mg/dL
Serum creatinine	1.29 mg/dL	Male-0.7-1.3 mg/dL
		Female-0.6-1.1 mg/dL
Serum uric acid	6.30 mg/dL	Male-4.4-7.6 mg/dL
		Female-2.3-6.6 mg/dL
Electrolytes		
Serum sodium	135 mEq/L	136-145 mEq/L
Serum potassium	6.0 mEq/L	3.5-5.1 mEq/L
Serum chloride	104 mEq/L	96-106 mEq/L

[Table/Fig-3]: Renal function and electrolyte report showing hyponatraemia and hyperkalaemia, on Day 2 of hospitalisation.

Immunoassay	Obtained value	Biochemical reference values
17-OH Progesterone, Serum (Serum, CLIA)	>300 ng/mL	Both: 1-12 months: 0.79-16.71 ng/mL Both: 1-13 years: <2.28 ng/mL Male: 0.29-2.06 ng/mL

[Table/Fig-4]: Immunoassay report showing markedly elevated serum 17-hydroxyprogesterone (>300 ng/mL).

elevated serum 17-hydroxyprogesterone (>300 ng/mL) and low serum cortisol levels. Isolated hypoaldosteronism was excluded as it does not cause elevated 17-OHP levels. As the septic screen {blood counts, C-Reactive Protein (CRP) and cultures} were negative neonatal sepsis was ruled out, and the presence of generalised hyperpigmentation and definitive hormonal abnormalities favoured an endocrine cause. Based on clinical features, electrolyte imbalance, and elevated 17-OHP levels, a final diagnosis of classical salt-wasting congenital adrenal hyperplasia due to 21-hydroxylase deficiency was established.

The neonate was managed as an endocrine emergency. Hypoglycaemia was corrected with an intravenous 10% dextrose bolus followed by continuous infusion. Intravenous hydrocortisone was initiated at stress doses (100 mg/m^2/day in divided doses). Electrolyte abnormalities were corrected with appropriate intravenous fluids and sodium supplementation.

Once clinically stable, the neonate was transitioned to oral hydrocortisone (10 mg/m^2/day, in three divided doses) and fludrocortisone (0.1 mg/day) lifelong therapy, along with oral sodium supplementation. Dose titration was performed according to the patient's response to therapy, as assessed by periodic growth monitoring, bone age assessment, and serum biochemical parameters. The infant showed significant clinical improvement with normalisation of blood glucose and electrolyte levels over the next 72 hours. Parents were counselled regarding the nature of the disease, need for lifelong treatment, stress-dose steroids during illness, and regular endocrinology follow-up. The neonate was discharged in stable condition on oral hydrocortisone and fludrocortisone therapy with sodium supplementation. The baby is on regular monthly follow-up, last follow-up was done one month back and he is doing well with age appropriate milestones and with no recurrences.

DISCUSSION

Salt-wasting CAH is a potentially fatal condition if not recognised early. Aldosterone deficiency results in renal sodium loss, dehydration, and hyperkalaemia, while cortisol deficiency leads to impaired gluconeogenesis and hypoglycaemia [1,2]. Male neonates pose a unique diagnostic challenge, as the absence of genital ambiguity often delays suspicion [3,4] until metabolic decompensation occurs.

Reddy NA et al., reported a similar case of devastating salt wasting crisis in a four-month-old male child with congenital adrenal hyperplasia, highlighting the need for high-index of suspicion in neonates presenting with hyponatraemia, hyperkalaemia, hyperpigmentation, also under scoring the need for neonatal CAH screening which is currently not a mandatory practice in several countries including India [5]. Balaji MD et al., reported a similar case of 20-day-old male neonate with classical CAH which was initially mistakenly treated as a case of refractory neonatal sepsis until they arrived at the diagnosis of CAH [6]. This case report highlighted the need to have CAH as one of the differential diagnoses for unresponsive sepsis so that it is not missed.

Hypoglycaemia may be an early and prominent feature, as observed in the present case. Electrolyte abnormalities typically evolve over the first week of life, coinciding with physiological decline in maternal steroid influence [7]. Measurement of serum 17-hydroxyprogesterone is the cornerstone of diagnosis and remains the primary screening and confirmatory test for CAH [8].

Management involves acute stabilisation followed by lifelong hormone replacement. Hydrocortisone is the preferred glucocorticoid in neonates due to its short half-life and minimal impact on growth [9]. Mineralocorticoid replacement with fludrocortisone and sodium supplementation is essential in salt-wasting forms. Education of caregivers regarding stress dosing during illness is critical to prevent adrenal crises [10].

Newborn screening programmes have significantly reduced mortality and morbidity related to CAH in developed countries. However, in many low- and middle-income countries, early diagnosis still depends on clinical vigilance [11,12]. This case reinforces the importance of considering CAH in any neonate presenting with unexplained hypoglycaemia and electrolyte imbalance.

CONCLUSION(S)

Salt-wasting congenital adrenal hyperplasia is a neonatal endocrine emergency with high mortality if untreated. Male neonates may present without obvious genital abnormalities, leading to delayed diagnosis. Early recognition of hypoglycaemia and electrolyte imbalance, followed by prompt hormonal evaluation and treatment, is crucial for survival. This case highlights the need for heightened clinical awareness and supports the implementation of universal newborn screening for CAH.

Patient Consent: Written informed consent was obtained from the parents for publication of clinical details and images. Adequate measures were taken to ensure patient anonymity.

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