

# Neonatal Hypercalcaemia Associated with Congenital Adrenal Hyperplasia

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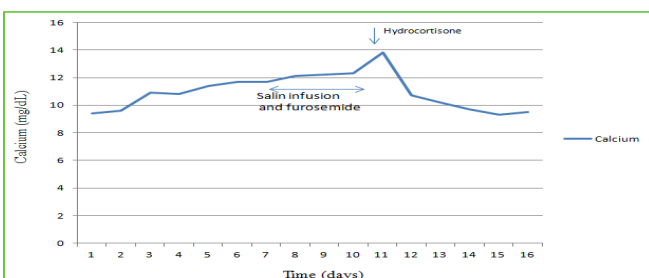
## Objective and Hypotheses

Adrenal insufficiency is an important and potentially life-threatening condition, and it is also known as a rare cause of hypercalcaemia. We report a case of congenital adrenal hyperplasia (CAH) due to 11-hydroxylase deficiency presenting with neonatal hypercalcaemia.

## Methods & Patient

A 18-day-old male infant was born at 36 weeks gestation to a 30-year old healthy mother by emergency cesarean section for fetal distress and intrauterine growth retardation, was consulted to our paediatric endocrinology clinic due to resistant hypercalcaemia. On physical examination, the infant was appearing ill with the vital signs, heart rate of 160 beats/min; respiration rate of 68/breaths/min and he was found to have hypertension (90/65 mm/Hg). Genital examination revealed bilateral palpable gonads, a 3 cm phallus and mild scrotal hyperpigmentation.

**Figure 1: Serum calcium variation in a newborn diagnosed with congenital adrenal hyperplasia secondary to 11-beta hydroxylase deficiency.**



## Results

The laboratory evaluation revealed a glucose level of 58 mg/dL; calcium, 13.8 mg/dL; phosphorus, 6 mg/dL; sodium, 134 mEq/L; potassium, 6.8 mEq/L; alkaline phosphatase, 1100 U/L; ACTH, 104 pg/mL; morning (08<sup>00</sup>) cortisol, 3.4 µg/dL; 17-OHP, 1900 ng/dL, DHEA-S, 1000 µg/dL; total testosterone, 506 ng/dL; progesterone, 4.05 ng/mL; androstenedion, 3.4 ng/mL. At a standard-dose ACTH test peak cortisol level was 12 µg/dL and the peak 17-OHP level and 11-deoxycortisol were 4800 ng/dL and 37.1 ng/mL, respectively. The diagnosis of 11 beta hydroxylase deficiency was established. Further evaluation revealed a serum 25-OH vitamin D level of 10.5 ng/mL a PTH level of 4.8 pg/mL. It was noted that, the patient had been treated with intravenous (IV) saline followed by IV furosemide 1 mg/ kg/dose every 12 hours, however the serum calcium levels could not normalised with this therapy. Oral hydrocortisone treatment at the dose of 20 mg/m<sup>2</sup>/day was commenced. Hypertension and hypercalcaemia were corrected by hydrocortisone replacement therapy in three days (Figure 1). On the follow-up the patient did not develop hypercalcaemia or hypertension.

## Conclusion

This case report confirms that, though rare, CAH and adrenal insufficiency should be considered in cases of hypercalcaemia that can be corrected by corticosteroid administration. The patient reported herein presented with only mild scrotal hyperpigmentation and no other findings remarkable for CAH. Neonatologists should consider CAH as a differential diagnosis of neonatal hypercalcaemia.