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# 46XX Congenital Adrenal Hyperplasia with Salt Wasting Crisis: A Case Report

**Dr. Avinash K[1], Dr. Pritikar Dowerah[2]**

[1]Post Graduate Resident, Department of Pediatrics, Assam Medical College, Dibrugarh, Assam, India
[2]Professor, Department of Pediatrics, Assam Medical College, Dibrugarh, Assam, India

**Abstract:** ***Congenital Adrenal Hyperplasia (CAH) are any of several autosomal recessive diseases resulting from mutations of genes for enzymes mediating the biochemical steps of production of mineralocorticoids, glucocorticoids or sex steroids from cholesterol by the adrenal glands (steroid genesis). This study Present a case report of a 01 month 11 days old infant with 46XX congenital adrenal hyperplasia (CAH).46XX CAH is a rare case.***

**Keywords:** Congenital adrenal Hyperplasia, CAH, 46XX CAH

## 1.Introduction

Congenital adrenal hyperplasia (CAH) is an autosomal recessive disorder related to deficiency of enzyme needed to the biosynthesis of cortisol and aldosterone. More than 90% of cases of CAH are due to deficiency of 21-hydroxylase resulting increased levels of progesterone and 17-hydroxyprogesterone which is converted into androstenedione and then to testosterone. The net effect is prenatal virilization of girls and rapid somatic growth with early epiphyseal fusion in both sexes known as simple virilization form. Most of patients are unable to synthesize sufficient aldosterone to maintain sodium balance and are termed salt-losing forms. This predisposes them to episodically develop potentially life-threatening hyponatremic dehydration. Besides this 8-9% of cases, there may be a nonclassic mild late onset forms of CAH due to deficiency of 11-a hydroxylase. Female newborns with CAH can be diagnosed early due to genital ambiguity[1]. 21 Hydroxylase deficiency is the commonest form of CAH accounting for over 90% of all cases. This disorder is associated with diminished synthesis of the cortisol and aldosterone. Low cortisol levels stimulate ACTH synthesis. Elevated ACTH level causes accumulation of steroid precursor. Depending on the severity of enzyme deficiency, the disease forms a spectrum of presentation[2].

## 2.Case Presentation

01 month 11days old infant came to hospital with history of ambiguous genitalia since birth, hyperpigmentation on trunk since birth and episodes of vomiting of 5 days duration born to non-consanguineous couple. . Mother had history of taking thyroxin tablet in her antenatal period. She was diagnosed as gestation hypertension at 7th month of pregnancy. Mother and father were 26, 31years at the time of conception respectively. Antenatal scans were normal. Elective cesarean section done at 38 weeks in view of pregnancy induced hypertension. Baby cried immediately after birth, weighed 2.9kg, head circumference of 34.5cm and length of 47.5cm. Baby had past history of admission to NICU in view of neonatal jaundice at 11th day of life stayed in NICU for one day duration, received phototherapy and got discharged. Baby was brought to our center with above mentioned complaints. The vitals at admission were PR: 128bpm, RR: 48cpm, SpO2: 97% in room air, CRT <3 SEC; head to toe examination showed hyperpigmentation over neck, bilateral axillary and bilateral lateral side of thoracic regions. Examination of Respiratory, cardiac, per abdomen and nervous system were within normal limits.

Baby was orally allowed. Inj cefotaxime (150mg/kg/day) was started in view of suspected sepsis. His sodium and potassium were 120mmol/L and 6.10 mmol/L respectively. Provisional diagnosis of congenital adrenal hyperplasia with salt wasting crisis was made. Baby was treated inj hydrocortisone (5mg/kg), fludrocortisone (0.1mg/day) and hyponatremia was corrected with 3% NaCl. Hyperkalemia was treated with potassium free fluid.

A diagnosis of CAH with salt wasting crisis was made in view of elevated levels of 17-hydroxyprogesterone (98.50ng/ml). Ultrasonography of abdomen and pelvis showed uterus and testes were not seen. Her karyotype showed 46XX. vitals, glucose and serum electrolytes were monitored daily during hospital stay and baby was clinically improved.

Baby was discharged with oral hydrocortisone (15mg/m2/day), fludrocortisone (0.1mg/day) and salt (1-2gram/day). The parents were advised to bring the child for regular follow up.

**Figure 1:** Infant with congenital adrenal hyperplasia with the skin hyperpigmentation

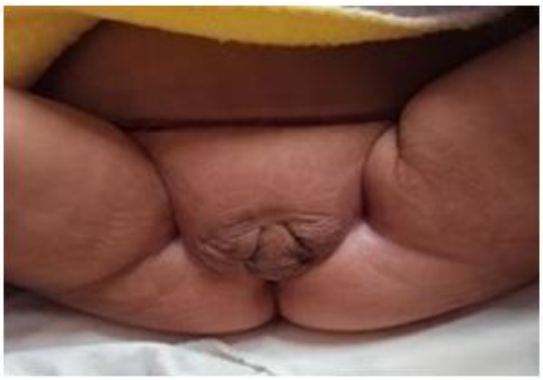

**Figure 2:** ambiguous genitalia

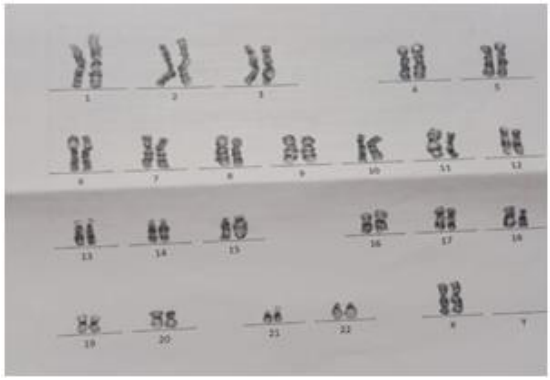

**Figure 3:** Chromosomal report showing 46XX

**Investigative Work-Up:**

- **Complete blood count:** Hemoglobin 16.9g/dl, total counts of 10600/MM3, Platelet count 2.8 lakhs/CUMM.
- **Serum electrolytes:** sodium 118mmol/L, pottassium 6.10mmol/chloride 78mmol/L, calcium 7.3mg/dl
- **Renal function test:** creatinine 0.8mg/dl, Urea 47mg/dl, uric acid 3.83.8mg/dl
- **Hormonal evaluation:** 17 Hydroxyprogesterone [17 OHP] 98.50 ng/ml
- **Radiological workup:** Abdominal ultrasonography showed presence of uterus, continuation of vagina could not be established, testes were not seen.
- **Genetic Workup:** Karyotyping showed 46XX

**Treatment:**

- Initially baby was treated with inj cefotaxime [[at]150mg/kg/day], inj hydrocortisone iv [at]5mg/kg.
- Hyponatremia was treated with 3% Nacl infusion and hyperkalemia was treated with potassium free fluids administration.
- Tab Hydrocortisone [[at]15mg/m2/day] started
- Tab fludrocortisone [[at]0.1mg/day] was started.

## 3.Discussion

In CAH, the body is missing an enzyme that stimulates the adrenal gland to release cortisol and aldosterone. More than 90% of cases of CAH are caused by 21-hydroxylase deficiency due to mutations in CYP21A2 gene. The salt losing crisis is the most important variant of CAH. These patients cannot synthesize sufficient aldosterone to maintain sodium balance and may develop potentially fatal 'salt wasting crisis ' if not treated[3].

Salt wasting form: These patients are the most severely affected and present in the neonatal period with virilization and salt wasting. Abnormal genital appearance should prompt the diagnosis in girls. Diagnosis is often missed in boys as they lack specific clinical features. They present after second week of life with failure to thrive, polyuria, hyperpigmentation and shock. Early diagnosis is mandatory to prevent mortality. 21 hydroxylase deficiency should be suspected in neonates with ambiguous genitalia, polyuria, shock, recurrent vomiting and feature of sepsis with negative sepsis screen. The diagnosis is confirmed by measurement of blood levels of 17 Hydroxyprogesterone [17-OHP][2].

Infants of salt wasting crisis were typically characterized by skin pigmentation likely related to abnormal hormone levels. Most infants with severe CAH develop vomiting, severe dehydration and shock. In this case, infant presented with ambiguous genitalia with skin pigmentation, vomiting with salt wasting crisis. The infant responded well with medical treatment and discharged on fludrocortisone and hydrocortisone. During follow up, the infant was found to be growing normally and skin pigmentation was resolved.

## 4.Conclusion

CAH is a hereditary condition that causes insufficient cortisol production. The salt-losing variant of CAH is a rare condition and medical emergency. Immediate medical attention is required to save the life. Parental counseling with follow-up is an essential component of CAH treatment.

## References

[1] Ruma Parvin, Nobo Krishna Ghosh, Sharmin Mahbuba, Farhana Jaya Chudhury, Sultana. A Male Neonate with Congenital Adrenal Hyperplasia: A Case Report. DS (Child) H J 2020; 36 (1): 74-79
[2] Ghai O P Ghai Essential Pediatrics: Endocrine and Metabolic disorders.10th edition 2023. p.547
[3] Canlas JF, Ponmani C. Congenital adrenal hyperplasia with salt-wasting crisis and arrhythmia: a case study. BMJ Case Rep.2019 Jan 29; 12 (1): e227565. doi: 10.1136/bcr-2018-227565. PMID: 30700462; PMCID: PMC6352754