DS (Child) H J 2020; 36(1): 74-79

CASE REPORT

A Male Neonate with Congenital Adrenal Hyperplasia: A Case Report

Ruma Parvin, Nobo Krishna Ghosh, Sharmin Mahbuba, Farhana Jaya Chudhury, Sultana Amena Ferdoucy

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 inincreased levels of progesterone and 17-hydroxyprogesterone which is converted into androstenedione and then to testosterone.[1] 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-â hydroxylase.[2] Female newborns with CAH can be diagnosed early due to genital ambiguity but male with CAH are usually asymptomatic at birth and are usually diagnosed after life threatening adrenal crisis or they die unsuspected.[3] We reported a male neonate with CAH salt-losing form. The case is reported to orient clinicians so that they may be able to manage the problem timely.

Case report

A 11 days old male infant, 3rd issue of a consanguineous parent from Brahmanbaria district was admitted in the SCANU of Dr. MR Khan Shishu Hospital & ICH with lethargy, less feeding, and weight loss. Mother, 25 years old, was under regular antenatal check up and her pregnancy was uneventful. The baby was delivered by caesarian section at term, without any adverse perinatal events. Weight of the baby was 3200 gram which falls on 50th percentile on growth chart. Breastfeeding was started within 1 hour of delivery and he was on exclusive breastfeeding. He was ok for the initial one week, then he became weak, unable to suck breast properly and losing weight gradually. Mother also noticed his body was becoming black. Mother gave history of 2 sib death. First baby was female, apparently normal, died at 9 months of age with unknown etiology. 2nd baby, male died at 12th days of life with severe sepsis at hospital. There was no history of similar disorders in the families either of parents. He was treated with injectable antibiotics in a local hospital for 2 days with the diagnosis of sepsis but his condition didn't improve and admitted in our hospital.

On admission, the infant was dehydrated, hypotonic and tachypnoeic. Respiratory rate was 66 breaths per minute. His genitalia was examined and was noted normal male genitalia but scrotal hyper pigmentation present. His reflex activity was moderate. Other examination findings were normal. His weight was 2800 gram and calculated weight loss was 12.5%.

Supportive measures were taken. Septic screening

1. Associate Professor, Department of Neonatology, Dr. MR Khan Shishu Hospital & Institute of Child Health.
2. Professor, Department of Paediatrics, Dr. MR Khan Shishu Hospital & Institute of Child Health.
3. Assistant Professor, Department of Paediatrics, Dr. MR Khan Shishu Hospital & Institute of Child Health.
4. Assistant Professor, Sylhet Womens Medical College Hospital, Sylhet
5. Assistant Professor, Department of USG & Radiology, Dr. MR Khan Shishu Hospital & Institute of Child Health.

Correspondence to: Dr. Ruma Parvin, Associate Professor, Department of Neonatology, Dr. MR Khan Shishu Hospital & Institute of Child Health. Cell: 01711119127, E-mail: ziaruma66@yahoo.com

Received 8 April 2020; **Accepted**: 26 May 2020

Fig 1 *Photograph of congenital adrenal hyperplasia patient on day-1*

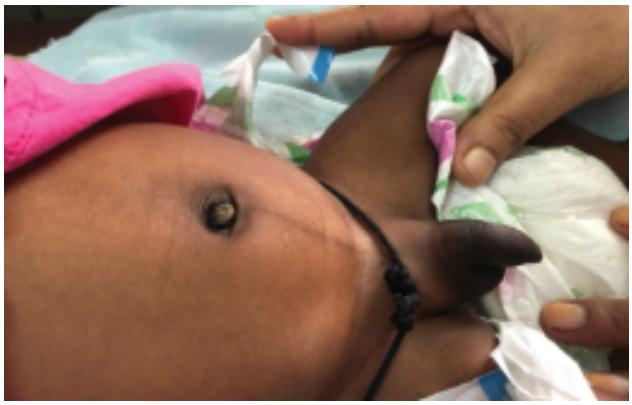

Fig 2 *Hyperpigmentation of genitalia*

was normal. Blood sugar was normal on several occasions. Serum electrolytes showed hyponatremia (Serum sodium - 125mmol/L), hyperkalemia (Serum potassium - 7.9 mmol/L, hypochloremia (Serum chloride - 84 mmol/L), renal profile was normal (Serum creatinine - 0.75 mg/dl and urea - 50mg/dl). Sodium correction was done by fluid therapy (increase sodium = 12meq/l/24 hours) and hyperkalemia was treated with injection calcium gluconate, nebulization with salbutamol and insulin glucose infusion but his serum sodium and potassium was not corrected. So our provisional diagnosis was CAH. An ultrasound of abdomen with special attention to adrenal gland was normal. Serum 17-hydroxyprogesterone (17-OHP) was >50ng/ml - highly suggestive of CAH. Seruma aldosterone was normal (141.70pg/ml) but Serum cortisol level was low (99.07nmol/L) and plasma Renin is high (46.09pg/ml). Serum testosterone (0.46nmol/L) and ACTH (54.12pg/ml) was high. Urinary sodium concentration is high (67meq/l). So the diagnosis of CAH (classic, salt-losing variety) was made. Then he was started on replacement therapy (hydrocortisone 100 mg/m^2/day, fludrocortisone 150 µg/day). Within 5 days of therapy there was significant improvement both in clinically and biochemically. On discharge his weight was 3.7 kg. The steroids were gradually tapered over the next one week and he was discharged on maintenance doses of oral steroids (hydrocortisone 20 mg/m^2/day and fludrocortisones 100 µg/day and sodium chloride 1mmol/kg twice daily). Parents were counseled about the disease and an instruction to his parents was given to double the dose of his oral hydrocortisone if the baby has intercurrent illness (e.g. fever, cough, vomiting and diarrhea). They were also given an emergency card that he can be assessed and managed immediately by the pediatric team whenever he presents to the hospital.

The baby came at 45 days of her age for follow up visit. His growth and development was normal. His weight was 5.3 kg and he is very alert and active. His Serum electrolyte was within normal range.

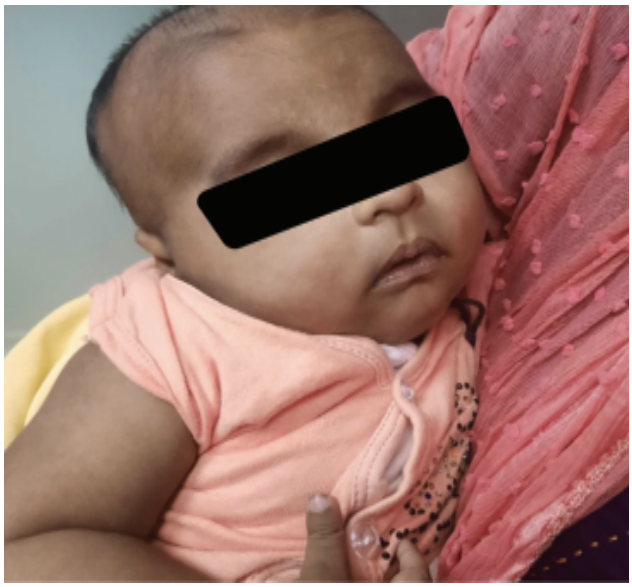

Fig 3 *Photograph of the congenital adrenal hyperplasia patient at 45 days of age*

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.[4] 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. Urinary sodium concentrations may exceed 50 meq/l. The infant can't maintain blood volume; hyponatremic dehydration begins to develop by the end of first week of life. Potassium and acid secretion are impaired leading to hyperkalemia and metabolic acidosis gradually. The early symptom is poor weight gain, but most infants with severe CAH develop vomiting, severe dehydration, and shock by the 2nd or 3rd week of life.[5] Females with classic 21-hydroxylase deficiency are exposed to excess androgens prenatally and are born with virilized external genitalia.[6] If routine screening test of CAH was not done, the male infant may remain undiagnosed as males have no genital ambiguity to alert physician and presented with life threatening salt loosing crisis.[3,7] Infants of the salt-wast-ing type were typically characterized by skin pigmentation, likely related to abnormal hor-mone levels.[8] If CAH is not diagnosed and treated early,neonates are susceptible to sudden death in the first few weeksof life.[9] In this case, salt loosing crisis were reported and he presented with less feeding, unable to suck and dehydration and genital pigmentation. The infant was attended earlier.

It is important to consider this disorder in all cases of otherwise unexplained electrolyte abnormalities during the first few weeks of life.[7] In this case, remarkable electrolyte abnormality was found. Diagnosis of 21-OHD is confirmed by steroid analysis in newborn screening or later on. Standard medical treatment consists of oral glucocorticoid and mineralocorticoid administration in order to suppress adrenal androgens and to compensate for adrenal steroid deficiencies.[10] In the index case, the high concentration of 17-hydroxyprogesterone(17-OHP) is suggestive of CAH. While on replacement therapy, the child should be closely followed up for growth, development, biochemical and radiological parameters to monitor the effect to titrate the dose of the replaced steroids.[11] The newborn responded well with recommended medical treatment and the baby gained weight during discharge. During follow up, the baby was found to be growing appropriately.

Conclusion

The salt-loosing variant of congenital adrenal hyperplasia (CAH) is a rare disorder and is a medical emergency. A male infant with CAH is usually remaining undetected at birth and the mortality rate for boys with CAH is thus higher than that of girls. So routine neonatal screening is essential for diagnosis of CAH in male infant if there is a history of parental consanguinity or presence of other affected siblings. Prompt treatment is essential to save the life of neonate. Counseling of parents with follow up is crucial part of management of CAH.

References

1. Qureshi S, Roohi M, Khaliq M. Congenital adrenal hyperplasia. *Professional Med J* 2008;**15**:403-06.
2. Das JC, Khanom M, Islam R. Congenital adrenal hyperplasia: A case report. *JCMCTA* 2013;**24**:44-47.
3. Nordenstrom A, Ahmed S, Jones J. Female preponderance in congenital adrenal hyperplasia due to deficiency in England: implications for neonatal screening. *Horm Res* 2005;**63**:22-28.
4. White PC, Speiser PW. Congenital adrenal hyperplasia due to 21-hydroxylase deficiency. *Endocr Rev* 2000;21:245-91.
5. Trakakis E, Loghis C, Kassanos D. Congenital Adrenal Hyperplasia because of 21-Hydroxylase deficiency. A genetic disorder of interest to obstetricians and gynecologists. *Obstet Gynecol Surv* 2009;**64**:177-89.
6. Fleming L, Knafl K, Van Riper M. How the child's gender matters for families having a child with congenital adrenal hyperplasia. *J Fam Nurs* 2017;**23**:516-33.
7. Bhakri BK, Biswas K. Congenital Adrenal Hyperplasia: Presentation in a male infant. *Journal of Clinical and Diagnostic Research* 2010;**4:**2910-12.
8. Bonfig W. Growth and development in children with classic congenital adrenal hyperplasia. *Curr Opin Endocrinol Diabetes Obes* 2017;**24**:39-42.
9. Ishii T, Anzo M, Adachi M. Guidelines for diagnosis and treatment of 21 hydroxylase deficiency (2014 revision). *Clin Pediatr Endocrinol* 2015;**24**:77-105.
10. Riepe FG, Sippell WG. Recent advances in diagnosis, treatment, and outcome of congenital adrenal hyperplasia due to 21-hydroxylase deficiency. *Reviews in Endocrine and Metabolic Disorders* 2008;**8**: 349-63.
11. Livak K, Foster PJ, Thalange N. Short term growth in children with congenital adrenal hyplasia. *Horm Res* 2009;**71**:142-47.