Case Report

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Congenital Adrenal Hyperplasia: A Case Report

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ABSTRACT

Background: *Congenital Adrenal Hyperplasia (CAH) in children varies in presentation and progression with several challenges in optimal management. Effective treatment is to achieve normal growth and development while avoiding adrenal crisis and hyperandrogenisation though achieving the balance between over treatment and undertreatment remains challenging.*

Case Presentation: *A 24 days old male child presented with repeated vomiting, severe weight loss and had decreased level of activity since 7 days of age. After total clinical, radiological, pathological evaluation it was diagnosed as congenital adrenal hyperplasia.*

Conclusion: *The approach to its diagnosis and management was a combined multidisciplinary effort of the Departments of Pediatrics, Endocrinology and Pathology.*

Key words: Congenital Adrenal Hyperplasia; 17 hydroxy progesterone.

Introduction

Most patients with Classic Congenital Adrenal Hyperplasia (CAH) both cortisol and aldosterone production are impaired while adrenal androgen production is excessive. As a result of lack of the vital hormones, cortisol and aldosterone, patients are susceptible to potentially lethal adrenal insufficiency, if untreated. Thus, emergency and critical care personnel must consider the diagnosis in patients presenting with shock. Excess androgen production, a side effect of 21-hydroxylase deficiency, causes genital ambiguity in females along with various endocrinologic, gynecologic, and reproductive complications. Men with CAH may also have reproductive and endocrine problems, most notably testicular adrenal rest tumors and oligospermia.[1] The present report is written for the purpose of reminding readers of this issue and emphasize the necessity for further research on how to deal with in the best way.

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Case Presentation

'M' a 24 days old male child of nonconsanguinous parents, hailing from Raozan got admitted in NICU of Bangabandhu Memorial Hospital (BBMH) IAHS on 23rd December, 2019 with the complaints of repeated vomiting, severe weight loss and had decreased level of activity from 7 days of age.

Mother of 'M' 22 years old, para 1 +0, blood group B positive was on regular antenatal checkup and her pregnancy was uneventful. She was immunized against tetanus as per EPI schedule. At 39 weeks of her pregnancy she delivered a male baby at Chittagong General Hospital by LUCS due to prolonged labor. A male baby weighing 3100 gm was taken birth. He cried immediately after birth and his APGAR score was 7/10 and 8/10 at 1 and 5 minutes respectively. After the delivery, the baby was on exclusive breast feeding. But from 7days of age he started repeated vomiting and weight loss despite proper feeding and shortly baby became reluctant to feed and then lethergic.

On admission, the baby was very lethargic, severely wasted and dehydrated. Temperature was 98.9°F, respiratory rate 42 breaths/min, heart rate180 beats/min, CRT 4 second, SPO was 76% in room air. BP was 40/30 (Mean pressure 33) mmHg and his reflex and activity were poor. He was grossly wasted and his cumulative weight loss was 51.6% from birth weight. Auscultatory findings on chest and precordium were normal. Penis looked apparently larger in size and scrotum was deeply pigmented. Other systemic examination was normal.

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Patient was initially diagnosed as a case of late onset of neonatal sepsis with hypovolemic shock. Investigations and treatment was given accordingly. But there was no significant improvement after 48 hours of antibiotic therapy, the baby was still lethargic and developed AKI. Investigations revealed hyponatremia, hyperkalemia and sepsis screening appeared to be normal. Na+ correction was done by 3% NaCl solution, but the parents took DORB for to financial reason.

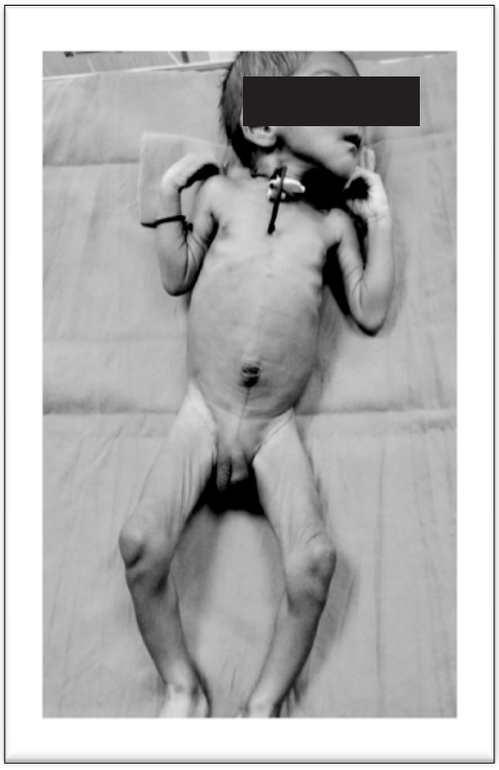

Figure 1 Child patient 'M'

After 2 weeks, baby re-admitted due to same complaints and the investigation revealed profound hyponatremia and hyperkalemia. Again, Na^+ correction was done by 3% NaCl solution and after 24 hours of correction, Na^+ level did not increase to normal level. Na^+ level measured for five times, but the Na^+ level didn't reach to normal. Other investigations were also done at that time. 17α HDPG and S. testosterone was increased, S. cortisol, S. aldosterone were normal. USG of whole abdomen showed no abnormality. Finally, the patient was diagnosed as a case of congenital adrenal hyperplasia and treatment was given with hydrocortisone and fludrocortisone, calcium gluconate, 3% NaCl solution. Patient's condition improved and eventually discharged with oral NaCl tablet 300mg, 2 tablets thrice daily along with advise of general measures.

Figure 2 Child patient with his mother

On subsequent follow up-patient's condition was improving, catchup growth achieved and CT scan of brain and Serum electrolytes, S. aldosterone, 17 HDPG, S. cortisol and S. testosterone were within normal range. At 2 year 2 month of age, baby again came with large penis and with pubic hair and raised serum testosterone, raised 17-OH Progesterone. But serum cortisol was not done because of financial problem. At that time baby was clinically diagnosed as a case of precautious puberty.

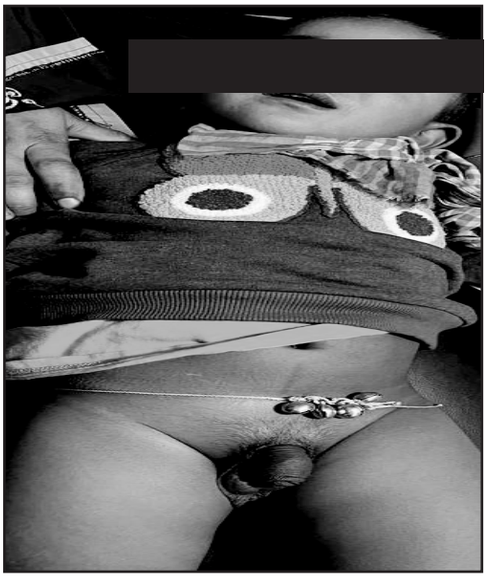

Figure 3 Child patient, a case of precautious puberty

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Investigation Profile

Date 23/12/19					<6.0 mg/dl	
	Serum Electrolyte	Serum Creatinine	CBC	Blood Culture	CRP	IT ratio
25/12/19	Na+:24.6mmol/L K+: 5.9mmol/L Cl: 95.7mmol/L HCo_3: 22.6mmol/L		Hb%-13g/dl ESR -5 TC-11,400 N-44%, PC-156,000	11.5 mg/dl		
26/12/19						
30/12/19				No growth		
14/1/20	Na+: 115 K+: 4.3 Cl:75 HCo_3:25.5	1.3mg/dl	Hb%13.2 ESR: 07 TC: 9000 DC: N: 54% L: 40% PC: 50000D			
15/1/20	Na+: 114.3 K+: 5.35 Cl: 96.2 HCo_3: 21.5	0.85				
16/1/20	Na+: 118.9 K+: 5.98 Cl:97.0 HCo_3:20.0	1.09				
18/1/20	Na+: 121.3 K+: 5.4 Cl:96.0 HCo_3:25.0	0.6				
19/1/20						
20/1/20	Na+: 128.1 K+: 5.71 Cl:96.8 HCo_3:20	0.62				
22/1/20	Na+: 127 K+: 5.0 Cl:98 HCo_3:28	0.78				

Date	TORCH Screening	Serum TSH	Urine Culture	Urine for CMV DNA	Urine Electrolyte	S. creatinine
23/12/19						
25/12/19						
26/12/19	Negative		No growth			
30/12/19						
14/01/20		3.5microIU/ml	No growth			
15/01/20				Negative		
16/01/20					Na+: 65 K+:10 Cl-:50	12.0
18/01/20						
19/01/20						10

Date	17- Hydroxyl Progesteron	USG of whole abdomen	Serum Aldosterone	Serum Testosterone
20/01/20	>44.0 ng/ml	Huge bowel gas		
22/01/20			47.3mg/dl	68.04
26/01/20				Normal

Date	S. Electrolyte	S. Creatinine
10/01/20	Na+: 126.5 K+: 5.3 Cl:97.0 HCo_3:23.0	0.6
16/3/20	Na+: 126.8 K+: 5.28 Cl:96.0 HCo_3:24.0	0.4
29/6/20	Na+: 120 K+: 5.0 Cl:98 HCo_3:25.0	0.4
4/9/20	Na+: 132 K+: 6.2 Cl:97 HCo_3:21	0.5
3/12/2020	Na^+ 143	
23/12/2020	Na^+ 138 K^+ 5.0 Cl^- 103 HCo_3 28.0 PH 7.40	

Discussion

In classic 21-OH deficiency, inadequate hydroxylation of progesterone deoxycorticosterone results 11- in aldosterone deficiency, and a salt-wasting crisis may occur.[2,3] Urinary sodium concentrations may exceed 50mEq/L. The infant cannot maintain blood volume; hyponatremic dehydration begins to develop by the end of first week of life. Potassium and acid excretion are impaired leading to hyperkalemia and metabolic acidosis gradually. Ability to maintain circulation is further limited by the effect of cortisol deficiency. The early symptom is poor weight gain, but most infants with severe CAH develop vomiting, severe dehydration and shock by the second or third week of life which may lead to death of infant if not treated properly.[4]

Objective of treatment is to achieve normal growth, pubertal development, sexual function and fertility. Glucocorticoids are administered to decrease ACTII secretion. It suppresses hyperplastic adrenal gland, stop overproduction of adrenal androgens, thereby preventing progressive virilization. A variety of glucocorticoids (Hydrocortisone, prednisolone, dexamethasone) with dosage schedules have been used.

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Salt-losing crisis should be treated with high doses of hydrocortisone (50 to 100 mg/m²/day in 3-4 divided doses) in addition to intravenous fluids to correct sodium and water depletion. Glucose infusion is needed in presence of hypoglycemia. With correction of electrolyte and fluid depletion and resolution of the adrenal crisis, glucocorticoid dose can be tapered, oral fluids to begin, and intravenous fluid is discontinue. Maintenance therapy for classic 21-OH deficiency is oral hydrocortisone (10 to 20 mg/m²/day) in 3 divided doses. Some clinicians prefer intramuscular cortisone acetate 15 to 20 mg every 3 days for first 2 years of life. Patients with disturbed electrolyte regulation (Salt-wasting) also require a mineralocorticoid and sodium supplementation.[2] Fludrocortisone acetate (Usually 0.05 to 0.1 mg daily irrespective of body size) is the drug of choice. Some also recommend sodium supplementation (I to 5 mEq/kg/day).[3] Increased doses of glucocorticoid is indicated during periods of stress.[2,3] The protocol for monitoring patients varies. Serum 17-OHP, androstenedione, testosterone, and plasma renin activity, preferably measured at 7:30 AM to 8:30 AM, prior to morning medication, provide indices of control. Surgical treatment on genitalia of infant depends on degree of virilization. Initial surgery most often is performed within first year of life; later revision may be necessary.[2] Counselling of the parents is the crucial part of management of CAH. Management of such condition needs multidisciplinary involvement.

Presentations and laboratory findings of the present newborn correlate to CAH of 'salt loosing type'. The infant was attended earlier than many other cases with vomiting, dehydration and virilized external genitalia.[5,6,2] Penis looked apparently larger in size and scrotum was deeply pigmented.[5,7,8,6,2,9-12,4] The newborn responded well with recommended medical treatment

It is evident that the majority of male children presented with SW early on and precocious puberty at later ages. Females mostly presented early with either or both of SW and SV. The results of this study emphasize that screening for CAH may be beneficial in our population because late presentation may be detrimental to a child well-being. The fact that 47.7% of patients are from nonconsanguinous marriages implies that the gene frequency for CAH is probably common in our population. Prenatal diagnosis and treatment are advisable to the high-risk group with a sibling who has CAH. Such neonates should, at least, be investigated by serum 17-OHP analysis some days after birth to rule out the 25% risk for CAH.

Limitations

Long term follow up and surveillance for recurrence could not be done.

Conclusion

Congenital Adrenal Hyperplasia (CAH) is a rare disorder of adrenal corticosteroid biosynthesis. The 'salt loosing' variant is a medical emergency. Prompt treatment is essential to save the life of neonate. Management of such condition needs multidisciplinary involvement. Counseling of parents with follow-up is crucial part of management of CAH.

Recommendation

All the CAH cases diagnosed in BBMH should be recorded briefly and long term follow up should be ensured to know about disease recurrence.

Acknowledgement

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Disclosure

All the authors declared no competing interest.

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