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CASE REPORT

Congenital Adrenal Hyperplasia: Presentation In A Male Infant

Bhakhri BK, Biswas K

ABSTRACT

We describe the clinical course of a male infant presenting with recurrent dehydration and salt loosing crises, diagnosed as Congenital Adrenal Hyperplasia. After initial crises management, the child was continued on replacement therapy. During the two year follow up, he was found to be growing appropriately and achieving normal milestones for age.

Key Words: male CAH, salt loosing crises, 17-hydroxy-progestrone (17 OHP)

Bhakhri BK*, Biswas K
***Corresponding author**
Dr. Bhanu
drbhanu04@yahoo.co.in

Introduction

The autosomal recessively inherited classic form of congenital adrenal hyperplasia (CAH) due to 21 α hydroxylase deficiency is detected in approximately 1 in 16,000 births [1]. Due to genital ambiguity which is evident at birth, this disorder is suspected early in female newborns. However, in regions where newborns are not routinely screened, affected males are usually diagnosed after life threatening adrenal crises or they die unsuspected [2]. We report a male infant with CAH, who was managed for salt loosing crises and was subsequently followed up on replacement therapy.

Case Report

This forty day old infant was admitted to our hospital with failure to gain weight and recurrent vomiting. He was born to a 25 year old, primigravida mother, with a regularly supervised and apparently uneventful antenatal period. There was no history of similar disorders/ unexplained foetal loss/ genital ambiguity in the families of either of the nonconsanguinously married parents. He was delivered vaginally at term, without any adverse perinatal events. His birth weight was 2500 grams and he was started on exclusive breast-feeding. After the initial three uneventful postnatal weeks, he started having recurrent vomiting and was also noticed to have lost weight. He was admitted twice in private hospitals during next three weeks, with documented dehydration and was given appropriate correction treatment for the same.

At the time of admission to our hospital, the child was severely dehydrated and lethargic. Slight hyperpigmentation was noted at the nipples and the scrotum. He was given supportive measures in the form of temperature maintenance and normal saline boluses to a total of 20ml/kg. He was started on antibiotics to cover possible sepsis and meningitis. His investigations showed normal blood sugar, hyponatremia (Serum Na-120 meq/l), hyperkalmia (Serum K-6.2 meq/l) and moderate metabolic acidosis (pH-7.21, serum bicarbonate- 16 meq/l). Accordingly, he was started on fluid therapy with targeted rehydration and increase in serum sodium by ~12 meq/l/24 hrs. The condition of the child improved gradually over the next 48 hours in terms of activity, feed intake and urine output. Serum sodium raised to143 meq/l with normokalmia and improved acidosis. Subsequently, intravenous fluids were stopped.

Investigations for the screening of sepsis, blood culture and CSF analysis did not reveal any evidence of sepsis.

After 24 hours of stopping parentral fluids, the child was again noticed to be lethargic and severely dehydrated; however, with apparently preserved urine output. His serum sodium was decreased to 118 meq/l, with serum potassium -6.4 meq.l, with metabolic acidosis and hypernaturia. He was restarted on parenteral fluid therapy. In the view of the recurrent documented hyponatremia, hyperkalmia and metabolic acidosis, the possibility of salt loosing CAH was considered and after taking blood samples, he was started on the management of the adrenal crises (hydrocortisone 100mg/sqm/24 hrs, fludrocortisone 150 µg/24 hrs and twice maintenance volume of saline in 5% dextrose). He gradually improved in the next two days. His serum cortisol was 2.72 µg/dl (normal- 4.3-22.4 µg/dl), with elevated unstimulated serum 17 –hydroxy-progesterone (17 OHP) – 40.5 ng/ml (normal- <0.60 ng/ml) and with elevated serum testosterone 0.5 ng/ml (normal <0.1 ng/ml). On the basis of these findings, the diagnosis of classic salt loosing CAH was made. The steroids were gradually tapered over the next one week and the child was discharged on maintenance doses of steroids (hydrocortisone 15mg/sqm/24 hrs and fludrocortisone 100 µg/24 hrs). Salt replacement was given, with expressed breast milk.

During his 2 years of follow up, the hyperpigmentation disappeared. He was found to be growing adequately for his genetic potential and achieved desired developmental milestones as expected. Weaning was started from 6 months of age and thereafter, salt supplementation was stopped. The doses of hydrocortisone and fludrocortisone are being titrated with body surface area, serum17 OHP levels and serum electrolytes respectively. At present, serum 17OHP is adequately suppressed (1.1ng/ml) and bone age is advancing, corresponding to the chronological age. During his clinical visits, he is being screened with negative results so far, for cushingoid features and for the presence of palpable mass in/ abnormal enlargement of (for testicular adrenal cell rests) the testis. His parents are following the instructions regarding stress dosing in the case of illness and are provided with detailed instructions for stress management and for contacting the paediatrician in case of emergency adrenal crises.

Discussion

Classic CAH is often left undiagnosed in male infants in the absence of routine screening programmes, in spite of being designated as the most common cause of the ambiguity of the genitalia in genetically female newborns and most of the males presented with life threatening salt loosing crises [2].

It is important to consider this disorder in all cases of otherwise unexplained electrolyte/ metabolic abnormalities during the first few weeks of life. Appropriate timing for sampling, adequate dosing of deficient steroids and fluid replacement is the key for the successful diagnosis and management of adrenal crises due to classic CAH [3].

While on replacement therapy, the child should be closely followed up for growth, developmental, biochemical and radiological parameters to monitor the effect and to titrate the dose of the replaced steroids [4],[5]. The child should also be monitored for cushingoid features due to excessive steroid intake and for the development of adrenal rest tumours in the testis, a well-known complication in male children and adults with classic CAH [6]. Information to parents regarding drug adherence, anticipated problems and stress dosing is vital for the appropriate management of these boys [7].

Conclusion

In the absence of routine neonatal screening, a high index of suspicion is essential for the diagnosis of CAH in males. Close

monitoring of clinical and investigational parameters, along with adequate information to the parents is required for appropriate management.

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