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CONGENITAL ADERNAL HYPERPLASIA WITH SALT WASTSING CRISIS A CASE REPORT

Paediatrics

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KEYWORDS

INTRODUCTION

Congenital Adrenal Hyperplasia is a group of autosomal recessive disorders due to deficiencies of enzymes involved in steroidogenesis. The most common form is 21-hydroxylase deficiency which can be classical or non-classical. The severe form, Classical Congenital Adrenal Hyperplasia is usually detected after birth to infantile period. If Congenital Adrenal Hyperplasia is not diagnosed and treated early, neonates are susceptible to sudden death in the early weeks of life. We report a case of forty-five days old male with a salt-wasting variant of congenital adrenal hyperplasia. The diagnosis was based on an elevated blood level of 17- hydroxyprogesterone. He was managed accordingly and was treated with fludrocortisone and hydrocortisone. prescribed.

Case Report

A 45 days old male child, who is a 2nd issue of twin gestation, born of non consanguineous marriage was brought with complaints of not accepting feeds since 2 days and lethargy

Past History

The patient was admitted in NICU at birth for Preterm (33 weeks) with twin gestation with low birth weight and refractory hypoglycaemia with late-onset sepsis for 30 days .

Physical Examination

- awake and afebrile
- HR – 130/Min
- RR -48/Min
- PP – WELL FELT
- AF – OPEN at level
- Pallor +
- Parietofrontal bossing
- Systemic examination
- CVS- S1 S2 Normal, Soft Systolic Murmur
- RS- B/L Vesicular Sounds+
- P/Abd – Soft NontenderBS+
- CNS – Looks around, AF at level,
- Tone – NORMAL.

Investigation

1	Serum Insulin	< 0.5 ug/ dl
2	Plasma Lactate	12.97 mg/dl
3	Plasma Ammonia	375 ug/dl
4	Growth Hormone	20.52 ng/ml
5	Serum Cortisol	19.34 ug/dl
6	Urine for reducing substances Urine Ketones	Absent
7	ABG	Compensated Metabolic Acidosis
8	Serum Electrolytes Na+ K+	132 4.1
9	17 OHP	2000 ng/dl

Course In Hospital

After initial stabilization, the patient was started on GDR6. The patient was not maintaining BSL, hence critical sampling done which was within normal limits. Urine for reducing substances and urine ketones were negative. 17 OHP was sent which was >2000 ng/ml but for confirmation of CAH. ACTH stimulation test and plasma renin level were planned but not done due to poor financial condition of relatives. Patient responded well to hydrocortisone and fludrocortisone. Thus, the patient was labelled as Congenital Adrenal Hyperplasia.

DISCUSSION

CAH can present as refractory hypoglycemia with clinical sepsis. Diagnosing a male infant is particularly difficult, owing to apparently normal physical features as compared to a female infant with congenital adrenal hyperplasia. Supportive laboratory finding and screening with 17- hydroxyprogesterone is helpful in making a diagnosis and early management of the patient to prevent fatality. Poor understanding and non affordability of parents were limiting factors for adequate diagnostic evaluation and treatment of such a rare inborn illness in this case.

REFERENCES:

1 Speiser PW, White PC. Congenital adrenal hyperplasia. *N Engl J Med.* 2003 Aug;349(8):776–88. doi: 10.1056/NEJMra021561
2 Kriplani A, Lunkad A, Agarwal N, Kulshreshtha B, Ariachery CA. A success story in congenital adrenal hyperplasia. *J Obstet Gynaecol India.* 2012 Dec;62(Suppl 1):78–80
3 Kovacs J, Votava F, Heinze G, et al. Lessons from 30 years of clinical diagnosis and treatment of congenital adrenal hyperplasia in five middle European countries. *J Clin Endocrinol Metab.* 2001 Jul;86(7):2958–64
4 Canlas JF, Ponmani C. Congenital adrenal hyperplasia with salt-wasting crisis and arrhythmia: a case study. *BMJ case Rep.* 2019 Jan 29;12(1):pii: e227565.