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Congenital Adrenal Hyperplasia with Salt Wasting Crisis- A Case Report

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ABSTRACT:

Congenital Adrenal Hyperplasia(CAH) is a recessive group of disorders transmitted autosomally, which is because of impaired cortisol synthesis. 21-Hydroxylase deficiency is the frequent of all the causes. Its onset is before birth usually. It should be identified & treated early to prevent its fatal effects. It can be presented in various forms.The presentation of each type varies depending on the type of enzymaticdeficiency.Infants present as classical form which is further divided as salt-wasting if there is mineralocorticoid deficiency and simple – virilizing if there is no deficiency of mineralocorticoidwhereas as others present as non classical form.This division of sub types is not useful clinically as the children with CAH may lose some extent of salt and their presentations might get overlapped. Clinical features may differ based on the type of CAH and child's gender.It includes vomitingwhich leads to dehydration, salt wasting and death. In conditions of severe virilization,clitoromegalyis seen.There is either precocious puberty or failure of puberty,Hirsutism, irregular Menstruation, infertility. Here,I present a clinical case of one month old boy child with CAH-salt wasting type. I confirmed it on basis of increased 17-OHP levels we managed him symptomatically & necessary treatment given for lifelong basis **.**

KEYWORDS: 17- OHP, CAH, Salt wasting type of CAH,21-Hydroxylase deficiency,DHEAS.

1. INTRODUCTION:

CAH is a disorder which is inherited in the autosomal recessive fashion, is due to deficiency of 21 hydroxylase. This deficiency is resulted by CYP21A deletion or mutation, which is the most frequently occurring form of CAH.Clinical features of salt wasting type include severe dehydration, hyponatremia,hyperkalemia, increased frequency of urination, failure to thrive.Non salt wasting variety presents as hypertrophied clitoris, shallow vagina and ambiguous genitalia. Diagnosis is mainly based on 17-OHP levels, cortisol, testosterone,DHEAS, etc. Early detection & proper management can prevent death . Main stay of treatment is glucocorticoids & mineralocorticoids replacement.

CASE REPORT:

One month old baby boy presented to our OPD with complaints of diarrhea since 3 days, Intractable Vomitings for 3 days which are multiple episodes, poor feeding, lethargy . Baby is passing very less amount of urine. There is no history of fever .Baby was delivered at 40 weeks of gestation by emergency LSCS because of fetal distress. Weight of baby was 3 kg. Baby cried immediately after birth. Baby's APGAR scores were normal. Prenatal checkups done and they are normal. Maternal serological tests were normal. There is no history of any such disease in their family. Baby has no signs of sepsis. He is a product of non consanguineous parents. He is the only child.

On examination, baby was lethargic and also had sunken eye balls, mucosa of oral cavity was dry. Anterior fontanelle(AF) was depressed. Pulse rate , temperature ,respiratory rate are normal. systemic examination was normal. Bp was checked and it was 80/50 mmHg .Baby weighted 3.2Kg ,length 56cm & Head circumference 38 cm. On genital examination , it was normal but darker than the actual skin tone.

Investigations showed deranged electrolytes. Serum Na+ 117meq/l , serum K+=6.3 meq/l ,urea-34mg/dl, creature -0.5 mg/dl ,RBS -77 mg/dl, ABG showed PH- 7.4(7.35-7.45),pco2-24.5(35-45),Hco3-15.8mmol/L , Base excess – 9mmol/L. Liver functions tests were normal.ECG was normal which is done to rule out arrhythmias. USG was normal which is done to rule out pyloric stenosis.

Baby was managed with continuous nebulization with salbutamol (2.5 mg) , 20 ml/kg of 0.9% NS bolus ,I.V. Hydrocortisone (2mg/kg) 6^{th} Hourly.17- Hydroxyprogesterone levels are raised i.e. 456mg/ml which is supposed to be <22 mg/ml. Hence we confirmed Congenital adrenal hyperplasia. He was continuously monitored while in the hospital and slowly got improved around 8 th day. Baby was discharged. we prescribed him prednisolone at 15 mg/ml/day p/o & Fludrocortisone Acetate -0.3 mg/day. We asked him to review and follow up after 7 days in the OPD. Baby weighted 3.6 kgs.Serumelectrolytes , ABG analysis showed normal findings & the treatment has been adjusted according to the reports.

2. DISCUSSION:

[1]As many as 90% of CAH cases are due to deficiency of 21-Hydroxylase. Among affected , around 70% are due to $CYP21A_2$ gene mutation .There are two types of CAH :- classic and non-classic .Incidence of classic variety has been noted around 1:10,000 – 1:20,000. [2,3] In classic variety deficiency , there occurs inadequate hydroxylation of progesterone which leads to aldosterone deficiency hence predisposed to salt wasting crisis. [4]Females with this classic type, which is deficiency of 21-Hydroxylase undergo exposure to more androgens antenatally hence they have features of virilized external genitalia.Non classic variety have relatively less elevated levels of androgen hence they might be asymptomatic.

[5] Management for the persons with genital anomalies should be multi team approach. Diagnosis of CAH in males is difficult because there is no genital ambiguity. [6] This case presented as having acute gastro enteritis with severe dehydration . since in male child physical examination alone does not help in diagnosing CAH unlike females . we diagnosed it only after lab reports showing decreased serum sodium levels with increased potassium levels. [7] Most of the cases with 21-OHD has pigmentation of the skin , masculinised female external, and are with poor sucking reflex & failure to gain weight . If not diagnosed and managed early, there are high chances of sudden death in the children in the first few weeks of their life. [8]The Diagnosis mainly depends on levels of 17-OHP, cortisol, testosterone,Dehydroepiandrosterone-sulphate,etc. In this case 17-OHP levels are raised. Early detection & proper management prevented death in our case . Main stay of treatment is glucocorticoids & mineralocorticoids

replacement. This case was treated with injection hydrocortisone & normal saline bolus. We discharged him with oral prednisolone & fludrocortisones.

3. CONCLUSION:

Based on the above report, it emphasizes the need for multi team approach. It also makes the treating clinicians aware of such entities to enable judicial management of these cases.

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