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CONGENITAL ADRENAL HYPERPLASIA

Pediatrics

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ABSTRACT

Primary adrenocortical insufficiency due to congenital hypoplasia or atrophy of the glands has been estimated to have a frequency of 1 in 15000 births1 .CAH includes a group of autosomal , recessively inherited diseases with impaired cortisol synthesis. Five different enzymatic conversions in sequence are essential for the production of cortisol from cholesterol .As patients now survive into adulthood , adult health care providers must also be familiar with this condition. Here we report a 55 days old female child with chief complaints of vomiting since 1 month , not gaining weight and hypopigmented patches over buttocks and thighs .

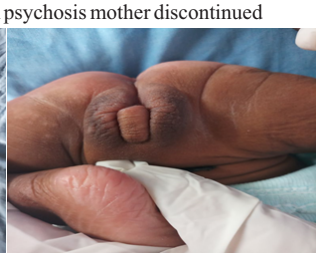
KEYWORDS

INTRODUCTION :

Congenital adrenal hyperplasia is inherited disorder of adrenal steroidogensis. The common functional defect in each disorder is impaired cortisol secretion, resulting in hypersecretion of CRH and ACTH and consequent hyperplasia of adrenal glands. 90% cases of CAH are caused by a defect in enzyme 21 alpha hydroxylase deficiency (21OHD)[2]. Depending on severity of the enzyme deficiency, 21OHD is defined as classic or nonclassic form. Approximately 75% of patients who have classic form also have salt wasting due to inadequate aldosterone production, fruther subdividing into classic simple virilizing and classic slat wasting forms[3].

CASE REPORT:

A 55days old FCh born out of non consanguineous marriage delivered at bheemavaram government hospital brought with c/o vomiting since 1month, not gaining weight and hypopigmented patches over buttocks and thighs. Since birth to 1month of life baby was on exclusive breast feeding, later due to post partum psychosis mother discontinued



breastfeeding .Thereafter baby was on bottle feeding with lactogen-1 with improper dilution. The baby had repeated episodes of vomiting after each feed. Baby developed hypopigmented patches since 1week over buttocks and thighs which is progressively increasing in number. Urine output is normal. At time of presentation baby had hypoglycemic episodes and baby appeared dehydrated .

- **On examination** – AF- sunken, PF- open, hypopigmented patches over buttocks and thighs, eyes sunken, lips- cheliosis, whitish patches over tongue and palate, clitoris is enlarged, pluses are feeble, cool peripheries .
  Diagnosis**:** i/v/o FTT, dehydration, feeble pulses, on examination clitoromegaly(virilisation), hypoigmented patches, B.P <50th percentile, hyponatremia , hyperkalemia, metabolic acidosis. Suspected as CAH and following investagtions were sent
- **Investigations** -Sodium-100meq/l, pottasium-6.5meq/l, calcium-8.9meq/l, 17-OHP-321.8ng/l, androstenedione->10ng/ml .
- **Management** – Intially baby treated with intravenous fluids for dehydration, given sodium correction with hypertonic saline and dextrose normal saline . in view of hyperkalemia potassium supplementation was not given in maintainance fluids and there were no electrocardiogram changes of hyperkalemia . After primary care baby started with Tab.fludrocortisone-0.1mg/dose/OD, T.hydrocortisone 15mg/m^2/day, for oral candidiasis candid mouth paint was given. Slowly feeds were started by nasogastric tube and gradually hiked .After baby started accepting feeds orally baby was given multivitamin supplementations .Baby gained weight and activity improved got discharged.

- **Maintainance therapy :**

baby was discharged on hydrocortisone , fludrocortisone and sodium supplementation , multivitamins.

- **Follow up:**

During follow up the baby showed adequate weight gain and vitals are stable . there was no hypertension and hepatomegaly which maybe consequences of overdose of fludrocortisone. There were no signs of cushingoid features .Electrolytes were repeated every monthly and hormonal profile were repeated every 3 monthly .There was reduction in clitoromegaly on followup

Their parents were counselled regarding the nature of disease , chronicity of treatment required ,the complications of poor compliance and were explained regarding the role of steroids in stressful conditions .Their parents were educated to rear baby as female child and were asked to refer to pediatric surgeon for vaginoplasty if required .

DISCUSSION:

The newborn infant with ambiguous gentalia is a medical emergency and appropriate investigations must be started quickly .The problem is a distressing one for the parents and they must be given a careful explanation of what will be done to determine the sex of the infant and how long this will take . It is important to note that the commonest cause of ambiguous genitalia of the newborn is CAH and the commonest cause of CAH is 21-hydroxylase deficiency [4].To confirm this diagnosis two investigations are essential :measurement of plasma 17 hydroxy progesterone concentration and determination of peripheral karyotype .

Other clinical parameters of control in CAH include measurement of growth velocity and skeletal maturation (bone age), signs of hypercortisolism (striae, weight gain , hypertension),and nothing the pattern of mensus in post pubertal women[5] .These are essential to monitor regularly but the parameters are insensitive indicators of control in the short term and are of necessity retrospective.Irregular mensus and infertility in both sexes have been additional problems[6] .

Treatment in CAH should aim to ensure normal growth in infancy and childhood , the development of puberty at the appropriate age and later , the acquisition of adult reproductive potential .This goal needs to be achieved using a minimum amount of glucocorticoid required to suppress excess androgen production . There is delicate balance to strike between the prevention of androgen induced rapid growth with advanced skeletal maturation on the one hand and the inhibition of normal growth from excessive glucocorticoid replacement on the other .several glucocorticoid preparations have been tried as regular replacement treatment in CAH [7] .Continued use of hydrocortisone is recommended through out childhood and puberty until statural growth is almost complete , dexamethasone is an effective form of treatment to use in the longterm .

Particular attention must be made to delineate the size of vagina and most importantly the level of confluence of the vagina with the USG . This information is invaluable in preoperative planning as well as counseling parents regarding the extent of surgery required for vaginoplasty[8,9]. Multidisciplinary one stop patients centered care with endocrinology , gynaecology and psychology expert teams are now gradually becoming the cornerstone of care for CAH

CONCLUSION

CAH is an uncommon chronic disorder which will present in the newborn nursery only about once evry 2 years .A high clinical index of suspicion should be maintained for all infants with abnormal genitalia .it is a team work approach.

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