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A Case of Congenital Adrenal Hyperplasia with 21Alpha Hydroxylase Deficiency

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Abstract: ***We report a case study of day 3 old female child brought to our NICU,SVRRGGH, Tirupati. with abnormal genitalia, neonate on suspicion of CAH, and on day5 child had dyselectrolytemia, with elevated potassium levels (>7.5meq/l),and weight loss, in view salt wasting crisis, neonate was started on the fludrocortisone and hydrocortisone, child has also elevated levels of 17-OHP levels.***

Keywords: NICU-neonatal intensive care unit, CAH -Congenital adrenal hyperplasia, 21-OH -21 alpha hydroxylase,11beta hydroxylase-11-OH, 3beta hydroxy steroid dehydrogenase- 3beta HSD,DSD -Disorders of sexual development ,17 OHP -17 hydroxyprogesterone**.**

1. Introduction

The most common DSD (Disorders of sexual development) presenting in neonatal period is a 46xx infant with CAH,The most common defect in CAH is 21-OH Deficiency caused by mutations of CYP21A2gene.

CAH is a autosomal recessive disorder, it is characterized by impaired cortisol synthesis, clinically presented as 2 types simple virilizing and salt wasting type.

Most common enzyme deficiency include 21-OH, rare11-OH,3beta HSD.The incidence of 21 alpha hydroxylase deficiency is 1:16,000births.

2. Case Report

A Day 3 old female newborn of term gestation born to primi mother through normal vaginal delivery and APGAR scores of 8 and 9, 10 at 1min, 5min, 10min. All the previous antenatal scans were normal,on day3 New born was referred to Neonatal intensive care unit(NICU), svrrggh, tirupati, in view of ambiguous genitalia ,On examination newborn has hyperpigmentation of axillae and genitalia with has fused labial folds look like scrotal rugae, with Prader stage 2(staging for abnormal genitalia) ,
On further evaluation, USG abdomen shows bilateral adrenal glands bulky suggestive of CAH with Mullerian structures present (uterus), also had dyselectrolytemia, showing elevated potassium levels on day5, newborn was evaluated for 17-hydroxy progesterone levels were elevated, and also newborn has weight loss, new born was confirmed to have CAH with showing 21alpha hydroxylase deficiency.

And newborn was started on hydrocortisone 50mg/m2/day in 3divided doses and after 1week fludrocortisone 100mcg was started and after 2 days electrolytes came to normal, On discharge newborn was advised to intake of salt of 1gm with daily monitoring of weight and electrolytes**.**

Clinical imaging

Figure 1: shows axilary hyperpigmentation

Figure 2: Posterior labial fusion, with cliteromegaly with reduced vaginal orifice.

Investigations:
Elevated potassium levels:>7.5meq/l(normal values:3.5-5.5meq/l)
Elevated 17OHP levels:6.84ng/ml (normal levels are 0.2-0.9)

Discussion:
CAH is inhertied disorders caused to deficiency of enzymes that producing the cortisol,mineralocorticoids, More than 90%of cases of CAH are caused due to deficiency of 21hydroxylase deficency due to the mutations in gene like CY21A2 gene.

They present mainly as two variants :
Such as salt losing crisis,and virilizing types ,and salt losing crisis was usually presents as medical emergency such as shock,hyperkalemia leads to arrhytmias,hyponatermia,if not diagnosed early and treated ,neonates are suspectible to sudden deaths, in our case neonate presented with salt crisis without arrhythmias,and treatment with hydrocortisone shows a good response.

3. Conclusion

In India there are many regions screening for 21 alpha hydroxylase deficiency was not done,so there should be A universalized screening programmed for CAH, in all regions of world,it is so crucial and important to prevent the life threating emergencies and sudden deaths in neonates.

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