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Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency: Case Report

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Abstract | **Case Report**

There are three interrelated errors of steroidogenesis that are responsible for congenital adrenal hyperplasia (CAH) of the urogenital system, the most prevalent of which is 21-hydroxylase.we report case of 21-hydroxylase deficiency.

Keywords: Ambiguous, genitalia**,** adrenal hyperplasia**,** urogenital system.

INTRODUCTION

Ambiguous genitalia are a rare disorder the urogenital system. This condition is caused by a disorder of sexual development, which results in a defect at birth where the external genitals don not look like those of a girl or a boy [1]. An inherited defect in any of the five steps required to synthesize cortisol from cholesterol causes congenital adrenal hyperplasia (CAH) [2]. Clinical symptoms may occur when an essential step is malfunctioning, not only because cortisol and other steroid hormones cannot be synthesized successfully, but also because precursor steroids accumulate and are shunted into other pathways, especially for androgen [2]. In congenital adrenal hyperplasia (CAH) is most commonly caused by one of three interlinked errors of steroidogenesis that affect children, 21-hydroxylase (the most prevalent), 11*-hydroxylase, and 36-hydroxysteroid dehydrogenase deficiencies [3]. The prevalence and pattern of CAH are not precisely known in Saudi Arabia; however, clinical experience suggests that this is not a rare condition [4, 5].

CASE

A full term baby boy was born by spontaneous vaginal delivery, with an estimated Apgar score of 7 in one minute and 9 in 5 minutes. In which there was no further concern about activity and respiration. Weight at birth was 3.1 kg. Upon admission to peripheral hospital due to Rh- incompatibility, metabolic screening was done as part of routine workup, and after three days of life, the baby was discharged home. It was raised by the parents that the other sibling, who is two years and five months old, has been diagnosed with CAH and is receiving treatment. On the 13th day of life, the metabolic result of the child was conducted to the hospital, which showed high 17-OH progesterone (220ng/ml), and the family was contacted for a second confirmation sample.

Upon arrival in the emergency room, the baby is looked critically ill and dehydrated. Urgent serum electrolytes were done which revealed Na: 116. K: 10 and the other renal parameters were normal. Patient was admitted to NICU and a dose of 100mg/m2/day divided by 4hydrocortisone was started.

Repeated serum Na: 118, K: 8.9, and patient received another bolus of normal saline.

A moderate level of dehydration was observed during the arrival of NICU Upon NICU stay genital examination showed a hyperpigmentedrugae Scrotum and empty (Figure 1) with and phallic length of 2.6cm with opening urethral meatus on glans (Figure 2) – Other systemic examination normal- blood pressure: 91/56 – mean arterial blood pressure (48) which is normal for his age. Throughout his stay in neonatal intensive care unit the patient received a bolus of normal saline followed by dextrose 10 normal saline to correct his hydration status.

An intravenous hydrocortisone was initiated along with fludrocortisone.

The result of serum electrolytes was showing sodium120 mmol\L and potassium 7.5 mmol\L. The repeated result of after six hours showing, sodium 130 mmol\L and potassium 6.8 mmol\L.

During that course, a septic screening was started and broad spectrum antibiotics were initiated, they were discontinued after blood culture and C-reactive protein came to be negative. On the third day of admission, the baby had a reached full feed and his serum electrolytes were back to normal. After that weaning off of intravenous hydrocortisone to 50mg/m2/day was started and baby was shifted to mother. On day four, intravenous hydrocortisone shifted to oral – 20mg /m2/day three times daily. On day five, the serum sodium decreased to 125- potassium: 7.9 so the baby was transferred back to the neonatal intensive care unit, and treatment for the adrenal crisis began. The ultrasound of pelvis was showing a uterus and absent testis (Figure 3). A repeated metabolic screening reviled 17-OH –progesterone to be 380 ng/ml. Karyotyping reviled 46, XX genotype.

Figure 1

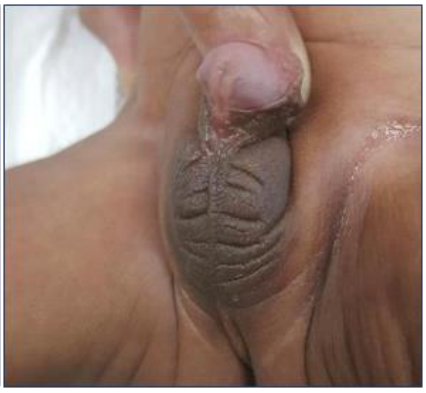

Figure 2

Hyperpigmentedrugaeempty scrotum

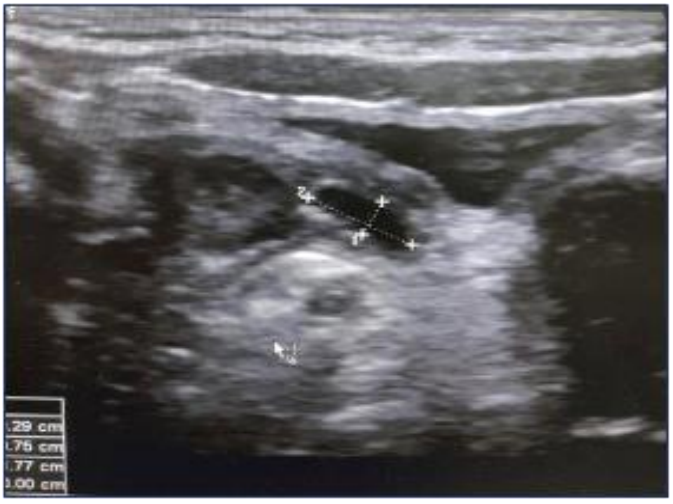

Figure 3: Ultrasound of pelvis: showing a uterus and absent testis

DISCUSSION

The congenital adrenal hyperplasia (CAH) is an autosomal recessive disorder that disrupts the production of adrenal steroids. It consists of three specific enzyme deficiencies; the most common form is 21-hydroxylase deficiency (21-OHD) due to mutations in the 21-hydroxylase (CYP21A2) gene. Other virilizing forms include 3β-hydroxysteroid dehydrogenase and 11β-hydroxylase deficiencies associated with mutations in the 3β-hydroxysteroid dehydrogenase (HSD3B2) and 11β-hydroxylase (CYP11B1) genes. Approximately 1 in 10,000 live births are estimated to be affected worldwide, and the incidence varies considerably by geographic region from 1 in 409 (Yupik Eskimos) to 1 in 67,000 in North America [6, 7]. No real data reported from Saudi Arabia, however Al Jurayyan *et al.*, [8]. In 90%-95% of cases, congenital adrenal hyperplasia is caused by 21-alpha hydroxylase deficiency, which accounts for the most common form of congenital adrenal hyperplasia. The CYP21A2 gene, which encodes a cortisol and aldosterone producing enzyme, is mutated in congenital adrenal hyperplasia due to a mutation located at chromosome 6p21.3 [9].

Lubni *et al.*, [10] has reported an incidence of 1 in 9,000 live births from neighboring Kuwait. Although we are referring to a highly selective group,

the pattern of enzyme deficiency in our series is somewhat different, in that 21- hydroxylase deficiency remains the most common but it accounts for 80%. Among them, 90% were salt-losers. In Saudi Arabia, Al Meshari *et al.*, [11], El Mouzan [12], Al Hazmi *et al.*, [13] and Saedi-Wong Et our population displayed a high level of consanguinity [14]. High parity rates and the involvement of several siblings within a family coupled with high consanguinity rates may contribute to this problem.

CONCLUSION

A full term baby boy delivered by normal spontaneous vaginal delivery, to gravida 3 and para 2 mother whose medically free birth weight 3.1 kg. The baby was discharged home and metabolic screen on day 3 showed elevated 17- hydroxyprogesterone (380 ng/ml) and the baby was moderately dehydrated. On examination, the scrotum was empty, and penile length was 2.6 cm with an opening urethral meatus. Ultrasound pelvis showed absence of testis. Karyotyping reviled 46, XX genotype.

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