Case Report

A case study of congenital adrenal hyperplasia

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ABSTRACT

Congenital adrenal hyperplasia (CAH) is a rare condition with a variety of symptoms. The most serious "salt losing" scenario is a medical emergency. A lack of 21 a-hydroxylase causes more than 90% of cases of CAH (21aOH). The adrenals produce excess sex hormones rather than cortisol. The vast majority of patients are unable to produce enough aldosterone. Girls are virilized, there is rapid somatic growth with the early epiphyseal fusion in both sexes, and there is even life-threatening hyponatremic dehydration. The following case report evaluates the clinical features, signs, and symptoms of CAH. A 1-day-old baby was admitted to the Neonatal Intensive Care Unit at the Department of Pediatrics, New Civil Hospital, Surat, for ambiguous genitalia. Various studies were conducted, and karyotyping suggestive of 46xx 17OH progesterone decreased. The sodium level was 114.64 mmol and the potassium level was 4.81 mmol on electrolyte analysis. CAH is an autosomal recessive disorder, in which 21-hydroxylase deficiency is the most common cause. In children, hydrocortisone is the preferred treatment.

Key words: Ambiguous genitalia, Congenital adrenal hyperplasia, Karyotyping

Congenital adrenal hyperplasia (CAH) refers to a group of abnormalities in adrenal steroidogenesis (CAH). CAH, an autosomal recessively inherited condition caused by a deficiency in 11-hydroxylase, is characterized biochemically by elevated levels of deoxycorticosterone, 11-deoxycortisol, and delta-4-androstenedione and decreased plasma renin concentration. Clinically, it has been linked to genital ambiguity and hypertension in females [1]. It could take years for approximately two-thirds of 11-hydroxylase-deficient people to develop hypertension [2]. Hypertension is the only clinical feature that distinguishes 11-hydroxylase deficiency from 21-hydroxylase deficiency, the most common type of CAH accounting for 90% of cases [3]. In a prepubescent child with hypertension and virilization, 11-hydroxylase deficient CAH should be considered in the differential diagnosis. The purpose of this article is to raise CAH awareness among health professionals, emphasize the importance of the early diagnosis and treatment to prevent gender identity problems from developing later, and discuss difficult ethical and management issues [4].

CASE SUMMARY

On September 27, 2022, a 1-day-old baby was admitted to the Neonatal Intensive Care Unit, Department of Pediatrics, New Civil Hospital, Surat. The baby was born through vaginal delivery, weighed 2.8 kg, and cried immediately after birth. On general examination, the patient was conscious and cooperative, with ambiguous genitalia. Blood tests revealed that the serum sodium level was 114.61 mmol and the serum potassium level was 7.33 mmol. All other blood tests, such as CBC, LFT, and RFT, were within normal limits. The liver and kidneys were normal in size, and the spleen was 3.7 cm in size with normal echotexture on an abdominal-pelvis ultrasound examination. The uterus was approximately 2.7*1.1*11 cm3 in size, and both ovaries were not visible. Karyotyping suggests 46XX in chromosomal analysis. The patient was fed with a measured spoon. The patient's relatives were counseled and informed about the condition as well as long-term treatment options. The opinion of a specialist endocrinologist was sought. Injectable hydrocortisone (5 mg every 8 h for the first 3 days, then 2.5 mg every 12 h) and injectable fludrocortisone (0.1–0.3 mg daily in divided doses) were started. The patient has been successfully discharged.

DISCUSSION

A lack of the enzyme 11-hydroxylase is the second most common cause of CAH [4]. The enzyme's activity is reduced or rendered inactive when the CYP11B1 gene is mutated, which reduces the conversion of 11-deoxycorticosterone to corticosterone and 11-deoxycortisol to cortisol [5]. Cortisol reduction now initiates a feedback loop that increases ACTH production. Precursors near the enzyme deficiency are overproduced and serve as substrates

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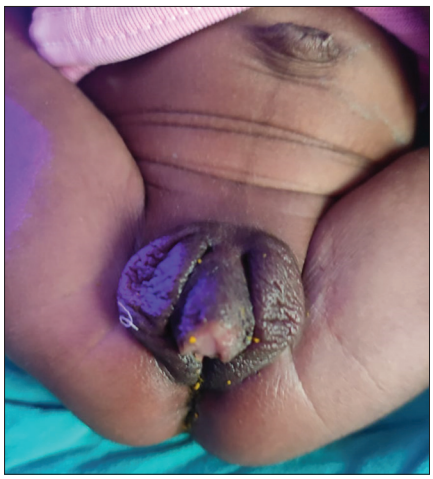

Figure 1: Ambiguous genetalia

Figure 2: Patient of congenital adrenal hyperplasia

for the accelerated androgen pathways, increasing androgen output from the adrenal glands. An excess of fetal adrenal androgens interferes with the development of the female external genitalia *in utero*, resulting in ambiguous genitalia. Females are so virilized at birth that their external genitalia, including the penile urethra and fused labioscrotal folds, resemble males' [6]. Multiple mutations in CAH caused by the 11-hydroxylase deficiency may affect heme binding sites. R374W and R448H/C heme binding site mutations resulted in high Prader scores (4/5), severe hypertension, and significantly accelerated bone age [7]. If the diagnosis is delayed, it is difficult to decide whether to change the gender. The option is determined by the patient's age at the time of diagnosis. To protect fertility, most authorities recommend female assignment in all 46, XX CAH cases [8].

In CAH, the body lacks an enzyme that causes the adrenal gland to release cortisol and aldosterone. More than 90% of CAH cases are caused by CYP21A2 gene mutations that result in 21-hydroxylase deficiency [9]. Salt-losing crises are the most common CAH subtype. Without treatment, these people risk developing a potentially fatal "salt-wasting" crisis due to an inability to produce enough aldosterone to maintain sodium homeostasis. The amount of salt in the urine could exceed 50 mEq/l. Hyponatremic dehydration has developed by the end of the 1st week of life as a result of the infant's inability to maintain blood volume. Hyperkalemia and metabolic acidosis progress as a result of impaired potassium and acid secretion. The first sign of severe CAH is poor weight growth, but by the second or 3rd week of life, the majority of newborns are vomiting, severely dehydrated, and in shock [10]. Females with a typical 21-hydroxylase deficit are born with virilized external genitalia as a result of prenatal exposure to high levels of androgens. If a standard CAH screening test had not been performed, the male newborn may not have received a diagnosis because males do not present with life-threatening symptoms, have no genital ambiguity to warn doctors, and are presented with a salt loss emergency [11].

CONCLUSION

CAH is a hereditary condition that causes insufficient cortisol production. The salt-losing variant of CAH is a rare condition and medical emergency. As a result, whether there is a history of parental consanguinity or the presence of additional afflicted siblings, regular newborn screening is critical for detecting CAH in a male infant. Immediate medical attention is required to save the newborn's life. Parental counseling with follow-up is an essential component of CAH treatment.

REFERENCES

1. Byne W. Developmental endocrine influences on gender identity: Implications for management of disorders of sex development. Mt Sinai J Med 2006;73:950-9.
2. Nimkarn S, New MI. Steroid 11β-hydroxylase deficiency congenital adrenal hyperplasia. Trends Endocrinol Metab 2008;19:96-9.
3. New MI. Inborn errors of adrenal steroidogenesis. Mol Cell Endocrinol 2003;211:75-84.
4. Pant V, Baral S, Shrestha B, Tumbapo A. A child with hypertension and ambiguous genitalia-an uncommon variant of congenital adrenal hyperplasia: A case report. J Med Case Rep 2017;11:168.
5. Zhao LQ, Han S, Tian HM. Progress in molecular-genetic studies on congenital adrenal hyperplasia due to 11β-hydroxylase deficiency. World J Pediatr 2008;4:85-90.
6. Júnior RT, de Souza HS, Tosatti A. Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency. Arq Bras Cardiol 2005;85:421-4.
7. Khattab A, Haider S, Kumar A, Dhawan S, Alam D, Romero R, *et al*. Clinical, genetic, and structural basis of congenital adrenal hyperplasia due to 11β-hydroxylase deficiency. Proc Natl Acad Sci U S A 2017;114:E1933-40.
8. Hughes IA, Houk C, Ahmed SF, Lee PA; Lawson Wilkins Pediatric Endocrine Society/European Society for Paediatric Endocrinology Consensus Group. Consensus statement on management of intersex disorders. J Pediatr Urol 2006;2:148-62.
9. White PC, Speiser PW. Congenital adrenal hyperplasia due to 21-hydroxylase deficiency. Endocr Rev 2000;21:245-91.
10. Trakakis E, Loghis C, Kassanos D. Congenital adrenal hyperplasia because of 21-hydroxylase deficiency. A genetic disorder of interest to obstetricians and gynecologists. Obstet Gynecol Surv 2009;64:177-89.
11. Fleming L, Knafl K, Van Riper M. How the child's gender matters for families having a child with congenital adrenal hyperplasia. J Fam Nurs 2017;23:516-33.

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