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Case Presentation

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Congenital Adrenal Hyperplasia due to 17α-Hydroxylase Deficiency in a 9-Year-Old Phenotypic Female Presenting with Hypertension: A Case Report

Reham M. Aljudaba[1], Futon A. Akoor[1], Amer O. Alali[2*], Abdullah Alhazmi[2], Abdulrahman Hummadi[2], Fatemah Qumairy[2], Hassan Almalki[2], Mosa Y. Hakami[2], Mohammed Shami[3] and Aida Al Jabri[4]

[1]Medical Intern, Jazan General Hospital, KSA

[2]Pediatric Consultant, Jazan General Hospital, KSA

[3]Pediatric Resident, King Fahad Central Hospital,, KSA

[4]Pediatric Endocrine Consultant, Al-Hassa General Hospital, KSA

[*]**Corresponding author**: Amer Alali, MD, Pediatric Endocrine Consultant, Jazan General Hospital, KSA, Tel: 966560018115

Abstract

Background: Congenital adrenal hyperplasia (CAH) due to 17α-hydroxylase deficiency (17OHD) is an uncommon form of CAH (<1% of cases) resulting from biallelic mutations in CYP17A1. This leads to defective cortisol and sex-steroid synthesis with accumulation of mineralocorticoid precursors, causing hypertension and hypokalemia.

Case report: A 9-year-old phenotypic female presented with chronic headache and persistent hypertension. Laboratory results showed hypokalemia, low serum cortisol, elevated ACTH, and markedly increased progesterone. Imaging demonstrated absence of female internal genitalia and bilateral inguinal testes. Karyotype was 46, XY, and genetic testing confirmed a homozygous nonsense mutation in CYP17A1 (c.987C>G; p.Tyr329*). Treatment with hydrocortisone and captopril normalized blood pressure and serum potassium.

Keywords: Congenital adrenal hyperplasia; 17α-hydroxylase deficiency; Hypertension; CYP17A1; XY female; Saudi Arabia

Introduction

Congenital Adrenal Hyperplasia (CAH) comprises a group of autosomal recessive disorders characterized by defective adrenal steroidogenesis, most commonly 21-hydroxylase deficiency. In contrast, 17α-hydroxylase deficiency (17OHD) is rare, accounting for less than 1% of cases, yet its clinical features are distinctive and clinically significant. Mutations in CYP17A1 impair the dual enzyme activity (17α-hydroxylase and 17, 20-lyase) required for cortisol and sex-steroid biosynthesis. Consequently, accumulation of precursors such as 11-deoxycorticosterone and corticosterone produces a state of mineralocorticoid excess, manifesting as hypertension and hypokalemia.

Individuals with a 46, XY karyotype typically present with female external genitalia and absent internal female organs due to anti-Müllerian hormone activity, whereas 46, XX patients often have normal genitalia but fail to undergo puberty. Because early symptoms are subtle, diagnosis is often delayed. We report a Saudi child with complete 17OHD to highlight the importance of considering endocrine etiologies in pediatric hypertension.

Case Presentation

A 9-year-old phenotypic female from Jazan, Saudi Arabia, presented with a 6-month history of progressive headaches, fatigue, and occasional vomiting. Symptoms intensified over recent weeks, accompanied by skin hyperpigmentation. There was no history of trauma or visual disturbance.

She was born full-term to second-degree consanguineous parents; developmental milestones were normal. Two older sisters had experienced childhood hypertension without diagnosis.

Examination: Blood pressure 175/110 mmHg, hyperpigmentation over elbows and knees, Tanner stage I female genitalia, and a mobile right inguinal mass.

Laboratory findings:

-Serum K^+ 2.4 mmol/L

-Serum cortisol 58 nmol/L (low)

-ACTH 155 pmol/L (high)

-Progesterone 48.3 nmol/L (elevated)

-Normal sodium and glucose

Imaging: Pelvic ultrasound and MRI revealed absent uterus and ovaries, with bilateral inguinal masses suggestive of undescended testes.

Genetic testing: Karyotype 46, XY; CYP17A1 homozygous nonsense mutation (c.987C>G; p.Tyr329*).

Management: Hydrocortisone 12 mg/day (divided doses) and captopril were started, resulting in normalization of blood pressure and potassium. The family received multidisciplinary counseling, and the patient was referred for prophylactic gonadectomy because of malignancy risk in ectopic testicular tissue.

Discussion

17OHD is distinguished by the triad of hypertension, hypokalemia, and sexual infantilism. Unlike 21-hydroxylase deficiency, patients lack virilization and rarely develop adrenal crisis in infancy. Our patient's hypertension and fatigue reflected chronic DOC-mediated mineralocorticoid excess. The phenotypic female appearance despite a 46, XY karyotype results from absent androgen

production in the presence of anti-Müllerian hormone. Partial deficiency forms may produce ambiguous genitalia and variable pubertal development.

Glucocorticoid replacement suppresses ACTH and reduces DOC levels, thereby correcting hypertension and hypokalemia. In our case, hydrocortisone alone with adjunctive ACE inhibition achieved full clinical stabilization. Gonadectomy is recommended for 46, XY females with undescended testes due to the risk of gonadoblastoma. Consanguinity was a notable factor in this family, with two siblings displaying similar symptoms but no diagnosis—emphasizing the need for genetic screening and family counseling in such settings. Reports from Saudi Arabia indicate an increased prevalence of rare autosomal recessive disorders linked to consanguineous marriage.

Conclusion

Although rare, 17α-hydroxylase deficiency should be considered in any child or adolescent with unexplained hypertension, hypokalemia, or absent pubertal development—particularly in consanguineous populations. Prompt diagnosis and appropriate management can prevent serious complications and guide genetic counseling. This case underscores the value of molecular testing in identifying atypical forms of CAH.

Authorship Contribution

All authors contributed to clinical care, data collection, literature review, manuscript drafting, and revision. All approved the final manuscript.

Ethical Considerations

This case was approved by the Institutional Review Board of Jazan General Hospital. Written informed consent was obtained from the patient's parents for publication of the case and accompanying data.

Conflict of Interest

The authors declare no conflict of interest.

Citation of this Article

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