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DIAGNOSIS OF 21-HYDROXYLASE DEFICIENCY IN CONGENITAL ADRENAL HYPERPLASIA IN UZBEKISTAN

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ABSTRACT

Congenital adrenal hyperplasia (CAH) is a group of autosomal-recessive diseases characterized by a defect in one of the enzymes or transport proteins involved in the synthesis of cortisol in the adrenal cortex. Of the seven described forms of CAH, 21-hydroxylase deficiency is the most prevalent. This study aims to evaluate and enhance diagnostic methods for CAH in female patients in Uzbekistan.

Keywords: *Congenital adrenal hyperplasia, 21-hydroxylase deficiency, virilization, neonatal screening, Uzbekistan.*

Introduction

CAH results from mutations in genes encoding enzymes involved in steroidogenesis. The most common form, 21-hydroxylase deficiency, accounts for approximately 95% of CAH cases and leads to impaired cortisol and aldosterone synthesis, resulting in androgen excess. Early diagnosis and appropriate management are crucial to preventing complications such as ambiguous genitalia, virilization, and metabolic crises.

This study focuses on improving diagnostic accuracy for CAH in Uzbekistan by analyzing clinical presentations and current diagnostic protocols.

Materials and Methods

The study included 30 female patients diagnosed with CAH or androgenetic syndrome, treated at the Regional Endocrinological Dispensary in Samarkand city between 2021 and 2023. Patients underwent:

Clinical evaluation: Assessment of menstrual history, physical examination, and gynecological examination.

Laboratory analysis: Hormonal profiling, including serum 17-hydroxyprogesterone, testosterone, and cortisol levels.

Imaging: Ultrasound to assess internal reproductive organs.

Patients were stratified by age:

2 to 10 years: 8 patients (20%)

11 to 20 years: 14 patients (35%)

21 to 33 years: 18 patients (45%)

Results and discussions

Demographic and Clinical Features

Age distribution - patients were divided into 3 groups based on their age:

Neutral age (2-10 years): 5 patients (20%)

Pubertal age (11-20 years): 8 patients (15%)

Reproductive age (21-33 years): 17 patients (55%)

Menstrual cycle patterns, which included absent menstruation (amenorrhea), extremely light menstrual blood flow (known also as hypomenstrual syndrome) and painful menstruation (algodismenorrhea):

Amenorrhea: 14 patients (35%)

Hypomenstrual syndrome: 6 patients (15%)

Algodismenorrhea: 5 patients (15%)

Physical and Androgenic Features:

Body composition changes - broad shoulders, narrow pelvis, and lack of fat deposits on thighs and buttocks. Hirsutism - hair growth observed along the white line of the abdomen, thighs, perineum, and lower legs.

Gynecological Examination revealed:

Clitoral enlargement: 30 patients (100%)

Clitoral scarring (post-clitorectomy): 8 patients (27%)

Vaginal stenosis: 10 patients (30%)

21-hydroxylase deficiency results in:

Cortisol Deficiency: Leading to adrenal insufficiency and increased adrenocorticotropic hormone (ACTH) production.

Androgen Excess: Causing virilization, ambiguous genitalia in newborn females, and secondary sexual characteristics such as hirsutism.

Aldosterone Deficiency: Resulting in salt-wasting crises in severe cases.

The findings align with global studies indicating a high prevalence of virilization and menstrual irregularities in CAH patients. The consistent presence of clitoral enlargement underscores the importance of early intervention to minimize psychosocial impacts.

Diagnostic Approaches

Elevated 17-hydroxyprogesterone remains the cornerstone for diagnosing CAH. Neonatal screening programs, although unavailable in Uzbekistan, would facilitate early detection. Pelvic ultrasound aids in assessing internal genital structures and detecting associated anomalies. A thorough assessment of physical characteristics and menstrual history is vital for diagnosis.

Conclusion

The study highlights the importance of integrating comprehensive clinical evaluations with laboratory diagnostics for CAH. Neonatal screening programs should be implemented to improve early diagnosis and management in Uzbekistan.

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