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FEATURES OF REPRODUCTIVE SYSTEM DEVELOPMENT IN GIRLS WITH CONGENITAL ADRENAL CORTEX DYSFUNCTION

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ANNOTATION

Congenital adrenal cortex dysfunction (CACD) is a chronic endocrine disorder associated with impaired synthesis of glucocorticoids, mineralocorticoids, and androgens in the body. This condition, especially in girls, leads to various pathological changes in the development of the reproductive system. The disease can be diagnosed in infancy or childhood, but in some cases, clinical symptoms may intensify or appear for the first time during reproductive age.

Reproductive system changes: Excess androgen production in girls leads to the development of male secondary sexual characteristics (virilization). There may be ambiguous or abnormal development of external genitalia, clitoromegaly, hypertrophy of the labia majora, menstrual irregularities, amenorrhea, or oligomenorrhea. These conditions can also negatively affect gender identity and psychosexual development. During puberty, secondary sexual characteristics may develop late or remain underdeveloped. Short stature and limited growth due to early epiphyseal closure are observed. Disruption of ovulation, follicular cycle cessation in the ovaries, and chronic anovulation directly affect reproductive health. Effects of hyperandrogenism: In many cases, chronic anovulation, a clinical picture similar to polycystic ovary syndrome, infertility, hair loss, acne, oily skin, and hirsutism are observed. These patients often consult a gynecologist or endocrinologist due to menstrual irregularities, infertility, or male-pattern physical features. Diagnosis: In addition to clinical examination, hormone levels are determined through laboratory tests: ACTH, cortisol, androgens (testosterone, DHEA-S), 17-OH progesterone, etc. Ultrasound is used to assess the condition of the ovaries and uterus. CT or MRI may be performed to evaluate the size and structure of the adrenal glands.

Treatment approach: Treatment in girls with CACD is long-term and complex, managed by gynecologists and endocrinologists. Hormone replacement therapy is used to maintain normal cortisol levels, which suppresses ACTH secretion and limits androgen production. At reproductive age, ovulation-inducing therapy or assisted reproductive technologies (e.g., IVF) may be applied if necessary.

The purpose of the study: To identify developmental changes and assess the main clinical features of the reproductive system in girls with congenital adrenal cortex dysfunction.

Materials and Methods: The study involved 30 girls aged 10 to 18 years diagnosed with congenital adrenal cortex dysfunction (CACD). All patients were under the supervision of a gynecologist-endocrinologist and were examined due to specific clinical signs related to reproductive system development. The aim of the examination was to determine the degree and nature of reproductive and sexual development disorders in girls with CACD.

Clinical examination included:

- Assessment of height, body weight, body mass index (BMI), and body proportions.
- Gynecological examination (if necessary) for external genital development and virilization signs (clitoromegaly, labia majora hypertrophy).
- Hirsutism grading based on the Ferriman-Gallwey scale.
- Skeletal development assessed by bone age comparison using hand and wrist radiography.

Laboratory tests:

- Adrenocorticotropic hormone (ACTH) – to evaluate pituitary activity and adrenal stimulation.
- Cortisol – to assess glucocorticoid production.
- 17-OH-progesterone – the key marker for CACD.
- Testosterone and DHEA-S – to determine the level of hyperandrogenism.

• Aldosterone and plasma renin activity (PRA) – to evaluate mineralocorticoid function.
• Electrolytes (Na^+, K^+) – to identify salt-wasting forms of the disorder.
All samples were collected in the morning on an empty stomach and analyzed in a clinical laboratory using specialized equipment.

Results:
Clinical and laboratory findings revealed several specific abnormalities in girls aged 10–18 with CACD:Disorders of sexual development: 60% of participants had delayed or incomplete development of secondary sexual characteristics. 30% showed signs of virilization (clitoromegaly, labia majora hypertrophy). Menstrual irregularities: 70% had delayed menarche, oligomenorrhea, or amenorrhea. 20% had not yet experienced menarche. Signs of hyperandrogenism: Moderate to severe hirsutism was observed in 75%, acne and oily skin in 50%, and male-pattern hair loss in 20% of the girls. Laboratory findings: Elevated 17-OH-progesterone in most patients. ACTH levels were high in the majority. Testosterone and DHEA-S were above normal in 65% of cases. Hypokalemia and reduced PRA were noted in 40% of cases. Instrumental findings: Polycystic ovary changes were detected in 55% via ultrasound.
Bone age exceeded chronological age in 60%. Adrenal hypertrophy was found in 25% of patients.

Conclusion:
Congenital adrenal cortex dysfunction (CACD) has a significant impact on sexual and reproductive development in girls. Hyperandrogenism, menstrual disorders, accelerated skeletal development, and signs of virilization are key indicators of the disease. Timely diagnosis and a comprehensive treatment approach help alleviate the consequences, restore menstrual regularity, and preserve reproductive health.

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