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Mini Review

Case about Swyer syndrome (complete, or "pure" gonadal dysgenesis)

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Abstract

46XY - Swyer syndrome (complete, or "pure" gonadal dysgenesis) can be briefly described as a female phenotype in the male genotype. The disease is named for the British endocrinologist Gerald Swyer, who described it in 1955. The full form of dysgenesis is nonsyndromic (not accompanied by extragenital malformations), excludes the duality of sexual development (the presence of male primary sexual characteristics along with female ones), psychological development occurs according to the female type. Congenital pathology occurs in one case in 180,000 individuals with a male karyotype and is recorded more often than other forms of XY-dysgenesis of gonads. Based on these results of the one female, the diagnosis of Swiera syndrome (complete genital dysgenesis: impaired sexual development) was suspected.

Keywords: 46XY; complete gonadal dysgenesis; sweyer syndrome

Introduction

The etiology of Swyer syndrome is not well understood. To date, it is known that the onset of pathology is most often associated with the absence or mutation of the SRY gene located on the short arm of chromosome Y and is mostly responsible for controlling the formation of testicles. Judging by observations of familial cases of the disease, involvement of unknown X-linked or autosomal genes is possible.

Risk factors are also not fully established. In addition to the common mutagenic effects (ionizing radiation and intoxication, viral infections, unbalanced or reduced nutrition) and the aforementioned hereditary burden, it is assumed that the probability of pathology may be directly dependent on the age of the father. Most often, it is not possible to trace the relationship between any effect on the body during gestation and the development of Sweyer syndrome.

Medical history

Outwardly, a 22-year-old girl turned to me with complaints typical of severe depression without psychotic symptoms, but suicidal thoughts. There is no uterus, testicles and no menstruation. Sex chromosomes as in men - XY. But at the same time she has a vagina. A patient, who grew up as a girl born from non-blood relationship, turned to our clinic with primary amenorrhea. The subject had a female sexual identity with normal growth and corresponding secondary sexual characteristics for her age. Past medical, family, and developmental stories did not help. General and system exams were within normal limits; the subject did not have the stigma of Turner syndrome. Breast development was age-appropriate (Tanner III stage). External genitalia were normal; there was no evidence of ambiguity. Pubic hair growth was Tanner's third stage, and axillary hair development was normal. Anthropometric measurements were age-appropriate.

From the anamnesis it is known that until the age of 22, not one of the doctors I was contacting could diagnose me and say what was wrong with me. The first time I went to a doctor at the age of 14 was the issue of delayed sexual development. I had all the examinations and said that I still have to wait and take vitamins.

Two years later, I again turned to the same gynecologist, because menstruation did not begin. I was prescribed a course of hormonal drugs, and, lo and behold, the cycle was established! But, as soon as I stopped taking hormones, everything returned to normal. I did not know what to do, I was confused, and I was overwhelmed by a bunch of emotions! To live without knowing what is happening to you, and even as a teenager, you will not wish this to the enemy.

I decided to omit this question and the next time, at the age of 18, I turned to another doctor. I was prescribed hormones again, having been diagnosed with primary amenorrhea (that is, a complete absence of independent menstruation). After six months of such treatment, I again stopped drinking hormones, because I was not sure that it was right.

"I felt like an alien, I did not understand why this is happening to me, who to contact, so that they explain to me the reason for this state of the body"

I was overwhelmed with depression. I felt like an alien, I did not understand why this was happening to me, who to contact, to explain to me the reason for this state of the body. By this time, I was already

studying at the institute in absentia and got a job at a large bank. I went to work and study with my head, but the thoughts on how to find out what was wrong with me did not leave me for a minute. I buried myself in the thought that no man needed me, that most likely I would never have children, and that I would never get married.

At 22, I finally decided to go to the doctor again, this time on the recommendation of a friend. He examined me and called the head of the department. The dream said that it seemed that I had something serious, and recommended that to the geneticists in the hospital to them. There I donated blood to determine the karyotype (I didn't even suspect what it was then) and waited a month for the results of the analysis.

A month later, I came for the result. The geneticist began to explain to me that I was supposed to be born a boy, but due to a malfunction in the development of the embryo at about the eighth to ninth week of pregnancy, the formation of my body went according to the female type.

To say that I was in shock is to say nothing! A lump was in my throat! At the end of the conversation, they told me that it was urgent to have an operation and remove the gonads, most likely male ones, because of the high probability of their transformation into cancer cells, and also to start taking hormonal drugs.

Due to deep depression, on the advice of my mother and senior sister, we turned to the psychiatrist Professor N. Aliyev. The professor prescribed me antidepressants and tranquilizers, and recommended that I consult with the surgeon for removal. Following the advice of the professor, I decided to remove the gonads in the future. She smoothed a very beautiful, outwardly brunette with blue eyes.

Surgical intervention in the volume of removal of rudimentary gonads and fallopian tubes is performed immediately after the diagnosis of this form of congenital ovarian agenesis. Surgical treatment is performed to prevent neoplasms, the source of which are cells of dysgenetic genital glands. Conservative treatment is not recommended to be started before surgery, since hormone therapy increases the risk of cancer complications.

Comment

In 1955, Svayer described two cases of "pseudohermaphroditism in men." For two women with an XY karyotype with primary amenorrhea, high stature and female external genitalia, although one of the patients had an enlarged clitoris, but a normal vagina [1]. Subsequently, such complete gonodal dysgenesis was called Swyer syndrome.

It affects approximately 1: 30,000 to 1: 80,000 children born who have a female phenotype, there is no genital ambiguity at birth, and Müller's normal structures. This is usually detected in adolescence, with delayed puberty and amenorrhea, since the gonads do not have reproductive or hormonal potential, as is seen in our patient [2].

Since Swyer's syndrome has a high incidence of gonadoblastoma and "malignant germ cell tumors", gonadectomy should be performed as soon as the diagnosis is made, as in the described case.

Pathogenesis

The formation of the reproductive organs comes from the Mueller duct in women and in Wolf in men. In an embryo with a male genotype, the synthesis of male steroid hormones by Leydig cells in the embryonic testes is due to the action of the mother's chorionic gonadotropin. Sertoli cells stimulate the differentiation of Leydig cells and others. They produce anti-Muller hormone, which contributes to the atrophy of the Muller duct. Their normal activity leads to the development of a male - with adequate differentiation of the testicles, atrophy of the Mueller duct.

The noted failures of this mechanism lead to the formation of female reproductive organs from bipotent kidneys, the development of which does not require such complex regulation. The maturation of the male fetus is controlled by the SRY gene. In its absence or mutation, the activity of Sertoli cells is impaired, gonad differentiation does not occur, which entails the development of Sweyer syndrome - a phenotypically female body without full ovaries, which can further stimulate the development of secondary sexual characteristics, but with useless gland, embryos prone to malignancy.

Symptoms

In the pre-pubertal period, the pathology proceeds without any subjective manifestations. During puberty, Swyer syndrome is characterized by the absence of signs of puberty. It is possible to note only meager hair growth in the pubic and armpit areas, but often it is also absent. Menarche does not occur, the mammary glands do not develop or are expressed very weakly. The type of physique during male ovarian dysgenesis is with broad shoulders, a voluminous chest, and a narrow pelvis.

In women with Swyer's syndrome, normal or above average height, developed muscles and a "heavy" lower jaw are more often observed. Sometimes there is a slight hypertrophy of the clitoris, although usually the external genitalia are somewhat underdeveloped. Patients complain of primary infertility, a feeling of discomfort or pain due to insufficient development of the vagina during intercourse or gynecological examination.

Conclusion

This case is important because it draws attention to the exclusion of rare DSD, such as Swyer syndrome, in a subject with primary amenorrhea, since accurate and early diagnosis would allow for conservative treatment and appropriate psychological counseling that can ensure fertility, reduce emotional trauma, and improve patient survival.

Conflict of interest

We declare that there is no conflict of interest that could be perceived as detrimental to the impartiality of the research reported.

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