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ROKITANSKY-KÜSTER SYNDROME

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Rokitansky-Küster (RK) syndrome is a disorder that causes the vagina and uterus to be underdeveloped or absent. Without a uterus, affected women do not have menstrual periods. Often, the first noticeable sign of MRKH syndrome is that menstruation does not begin by age 15 (primary amenorrhea). Girls and women with MRKH syndrome have a 46,XX chromosome pattern. They have normal external genitalia and functioning ovaries. They also have normal breast and pubic hair development. Although women with this condition are unable to carry a pregnancy, they may be able to have children with the help of assisted reproductive technologies or a uterus transplant.

Key words: uterine and vaginal aplasia, sigmoid colon

Relevance. Rokitansky-Küster syndrome (or uterine and vaginal aplasia) in its classic form is characterized by vaginal and uterine aplasia with normally functioning ovaries. With this pathology, the external genitalia retain their normal appearance, which is why this disease is rarely diagnosed at birth.

In modern literature, three types of RK are distinguished:

I – typical (simple or isolated), where only the absence of the uterus and vagina is noted.

II – atypical (complex). With a combination of uterine and vaginal aplasia and excretory or musculoskeletal pathology [1; 2].

III – RK associated symptoms, characterized by the presence of Müllerian duct aplasia, impaired kidney development, and pathological changes in the cervical-

thoracic spine [2], as well as renal agenesis and abnormalities of the cervical-thoracic spine.

Some of our colleagues combine the atypical variant of RK and associated conditions [3]. Other scientists believe that the typical variant of RK and associated conditions are diametrically opposed syndromes. RallK. et al. (2015) believe that the typical form of RK is found in approximately 64% of patients, the atypical form in 24%, and RK-associated pathologies in 12% of patients [5]. Colleagues Burns JL et al (2012) believe that defects in the formation of the uterus and vagina in approximately 4 out of 5 cases manifest with abnormalities of other organs and systems, and in 1 out of 3 cases are characterized by the presence of pathologies of the urinary system [2, 3]. Skeletal abnormalities in RK are expressed in various phenotypes, ranging from scoliosis to Klippel-Feil syndrome, with patients having a congenital malformation of the cervical and upper thoracic vertebrae, resulting in a short and immobile neck. Abnormalities in the development of the excretory system are characterized by the presence of kidney localization disorders (pelvic, lumbar), lack of kidney development, underdevelopment of one or more kidneys, horseshoe kidney, absence of one kidney, and synoptic urinary tract. Also, with KR, there are three types of uterine absence (right, left, central cylindrical ridge; two muscle ridges and total absence of uterine rudiments. During ultrasound examination of this category of patients, the endometrium is detected in a quarter of those examined, and its activity is preserved in every tenth patient (6–10%). Many researchers testify to the role of genetics in the etiology of this pathology, which is confirmed by the relatively high incidence of the disease in some families. It has been found that gene transfer is characterized by an autosomal dominant type of inheritance with incomplete further manifestation of pathological signs in the individual's phenotype and degree of phenotypic expression. These phenomena are caused by the presence of mutations in vital genes and/or an imbalance in chromosomes. RK syndrome is a relatively rare heterogeneous pathology accompanied by aplasia of the genital tract and upper two-thirds of the vagina in 46XX women.

If the patient intends to become sexually active, primary treatment with surgical correction of the defect by forming a neovagina is recommended [4]. In cases of KR syndrome after unsuccessful conservative treatment, or if the woman herself wishes to receive appropriate care, surgical reconstruction of the vagina is the optimal method that can be used in these situations. It should be noted that there is currently no universally accepted surgical approach to this problem. Methods of surgical treatment of the above-mentioned defect, involving the creation of a neovagina, can vary widely depending on the technical capabilities available at a particular institution, the experience of the surgical staff, and the specific form of the defect itself. On average, the effectiveness of these methods is satisfactory in 4 out of 5 cases of surgical treatment of KR, but this indicator does not differ significantly from the results of conservative methods [4]. Both approaches (conservative and surgical) have undergone significant changes and modifications in line with the requirements of the times as technology has developed.

The sigmoid colon is an excellent tissue for vaginal replacement in women with vaginal aplasia. The cosmetic and functional results of sigmoid colon vaginoplasty are good. The advantages of this method are: (1) reduction in the length of the neovagina, (2) preservation of the width and depth of the neovagina without the need for a long-term vaginal stent, (3) spontaneous mucus secretion, facilitating sexual intercourse, (4) prevention of unpleasant odor, (5) texture and appearance similar to a natural vagina. Recently, some authors have confirmed the possibility of vaginoplasty with sigmoid colon using minimally invasive access either by laparoscopy or robotics [8; pp. 274-280, 73; pp. 198, 111; pp. 115-119].

Conclusions: Thus, surgical treatment of this group of patients presents a serious technical challenge, the outcome of which affects both the physical and psychosocial health of patients. The ideal reconstructive procedure should provide a vagina of adequate length and require minimal dilation, if any.

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