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Diagnosis and Treatment Approaches in Vaginal Agenesis

Vajinal Agenizde Tanı ve Tedavi Yaklaşımları

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ABSTRACT

The development of the female genital system is a complex process that depends on a series of events involving cellular differentiation, migration, fusion, and recanalization. Failure of any of these processes results in congenital anomalies. Developmental anomalies can occur at various stages, resulting in conditions that impact both the urinary and reproductive systems. In younger patients, such malformations can significantly affect their overall health and quality of life, including aspects such as fertility, sexual function, and psychological well-being. The psychosexual effects of vaginal agenesis should not be overlooked, and clinical care primarily involves comprehensive counseling and support through open communication with the patient. For adult patients, treatment for vaginal agenesis typically starts with therapeutic counseling and education, with non-invasive vaginal dilation being recommended as the first-line approach, or surgery if necessary. Consequently, managing these issues often requires a multidisciplinary approach, engaging specialists such as urologists, gynecologists, endocrinologists, and geneticists, among others. Early detection and timely intervention can greatly enhance the outlook for individuals with these conditions. Besides considering the patient's expectations, the surgeon's experience plays a crucial role in selecting the appropriate surgical technique. This is because the success of the initial surgery is critical to the effectiveness of any subsequent procedures if required. In this review, the evaluation and treatment of vaginal agenesis, which constitutes an important part of congenital anomalies of the vagina, were discussed.
Keywords: Müllerian canal; vaginal agenesis; vaginoplasty.

ÖZ

Kadın genital sisteminin gelişimi, hücresel farklılaşma, göç, füzyon ve yeniden kanalizasyonu içeren bir dizi olaya bağlı olan karmaşık bir süreçtir. Bu süreçlerden herhangi birinin başarısızlığı doğumsal anomalilerle sonuçlanır. Gelişimsel anomaliler çeşitli aşamalarda ortaya çıkabilir ve hem üriner hem de üreme sistemlerini etkileyen durumlarla sonuçlanabilir. Genç hastalarda, bu tür malformasyonlar doğurganlık, cinsel işlev ve psikolojik refah gibi hususlar da dahil olmak üzere genel sağlıklarını ve yaşam kalitelerini önemli ölçüde etkileyebilir. Vajinal agenezinin psikoseksüel etkileri göz ardı edilmemelidir ve klinik bakım öncelikle hastayla açık iletişim yoluyla kapsamlı danışmanlık ve desteği içerir. Yetişkin hastalar için vajinal agenezi tedavisi tipik olarak terapötik danışmanlık ve eğitimle başlar, ilk basamak yaklaşım olarak invazif olmayan vajinal dilatasyon veya gerekirse cerrahi önerilir. Sonuç olarak, bu sorunların yönetimi genellikle ürologlar, jinekologlar, endokrinologlar ve genetikçiler gibi uzmanları içeren multidisipliner bir yaklaşım gerektirir. Erken teşhis ve zamanında müdahale, bu durumlara sahip bireylerin görünümünü büyük ölçüde iyileştirebilir. Hastanın beklentilerini göz önünde bulundurmanın yanı sıra, cerrahın deneyimi de uygun cerrahi tekniğin seçilmesinde çok önemli bir rol oynar. Bunun nedeni, ilk ameliyatın başarısının, gerekirse sonraki prosedürlerin etkinliği açısından kritik olmasıdır. Bu derlemede, vajinanın konjenital anomalilerinin önemli bir bölümünü oluşturan vajinal agenezinin değerlendirilmesi ve tedavisi tartışılmıştır.
Anahtar kelimeler: Müllerian kanal; vajinal agenezi; vajinoplasti.

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INTRODUCTION

Vaginal agenesis is a complex condition affecting the female genital system. Sometimes this condition is an isolated vaginal agenesis and sometimes it is part of a complex disorder of sexual development. To understand why this disorder occurs, it is necessary to understand the embryology of the female genital system.

Sexual development in the female direction is independent of the gonadal or endocrine system. Since the female fetus lacks Müllerian inhibitory factor (MIF), the paramesonephric ducts continue to develop and the caudal part of the duct fuses with the opposite side to form the uterine horns, corpus uteri, and uterine cervix. It is in contact with the uterovaginal canal and the urogenital sinus, which are formed at the end of the 2nd intrauterine month. Afterwards, these parts form the lower part of the vagina which will ensure the continuity of the uterovaginal canal (1).

Genetic factors play an important role in the etiology of vaginal agenesis. However, genetic factors alone are insufficient to explain all cases of vaginal agenesis. In addition to genetics, defective MIF release, estrogen receptor disorders at the caudal end of the paramesonephric duct, exposure to teratogenic agents, mesenchymal induction defects or genetic loss in cytoplasmic receptor proteins of androgenic target cells as in androgen insensitivity syndrome may disrupt normal vaginal canal development (2).

This review aimed to discuss the evaluation and treatment of vaginal agenesis, which constitutes an important part of congenital anomalies of the vagina.

EVALUATION OF VAGINAL AGENESIS CASES

In these patients, basic initial evaluation should be performed in two aspects. The first one is laboratory evaluation including testosterone, follicle stimulating hormone (FSH), and karyotype analysis and the second one is radiological evaluation. Radiological evaluation can be performed with transabdominal, translabial, or transrectal two-dimensional and three-dimensional ultrasound in order to evaluate the internal genital structures. Magnetic resonance imaging (MRI) should be used in addition to radiological evaluation. This allows visualization of rudimentary Müllerian structures, which may be present in most patients with Müllerian agenesis. In addition, MRI can also assess the presence of endometrial activity in the Müllerian structures. Since the evaluation of rudimentary Müllerian structures may be difficult with ultrasound, especially before puberty, MRI is preferred in this age group (3).

In addition, it should be kept in mind that urinary system and skeletal system anomalies may accompany genital system anomalies in these patients (4). In many studies, anomalies including kidney and collecting system, horseshoe kidney, pelvic kidney, renal agenesis, or ureter duplication have been found in approximately one-third of the patients. Therefore, urinary system ultrasonography should be included in the initial evaluation of patients. In addition, scoliosis, hemivertebra, and other vertebral arch disorders involving the skeletal system are encountered more frequently in this group compared to the normal population and X-ray spine radiography may be used in the evaluation (5,6).

PSYCHOSOCIAL COUNSELLING AND SUPPORT

The psychological effects of vaginal agenesis are often underestimated. Many patients with this condition experience intense anxiety and depression. The basis of this is the questioning of their female identity and the feeling of not being able to have children, which they are worried about in their future lives. At the same time, patients sometimes have difficulty in sharing their current situation with their family, friends, or partners. Considering all these factors, appropriate psychological counseling should be offered to these patients. In addition, contact with other patient groups with a similar diagnosis may also be useful (7).

TREATMENT OPTIONS IN PATIENTS WITH VAGINAL AGENESIS

Historically, surgical neovagination in patients with vaginal agenesis was first performed by Hippocrates who lived between 460 and 377 BC (8). However, surgeons did not show much interest in such patients in the following period. In 1898, Abbe succeeded in creating a neovagina long enough to have intercourse with a vaginal mould after surgically opening the perineum in two women (9). In the first half of the 20th century, many surgical and non-surgical methods were described to create neovagina (10).

The aim of treatment of vaginal agenesis is to treat anatomical abnormalities. The methods to be applied for this purpose are passive vaginal elongation or surgical creation of neovagina. In this section, passive methods and surgical techniques will be discussed.

Vaginal Elongation

Passive vaginal elongation with a dilatator should be the first-line treatment option because it can be performed safely and simply under patient control, and it has a lower complication risk and lower cost compared to surgery. The prerequisite for the success of vaginal dilatation is the patient's emotional maturity and willingness to apply this method. Initiation of dilatation at an early age, lack of sufficient knowledge about the diagnosis and anatomical differences of the patients, insufficient understanding of how the dilatation process works, and sociocultural reasons decrease the success of the method.

The dilatation procedure should be performed in an environment adapted to the patient, in which he/she feels comfortable and in which monitoring is possible. Initially, the patient should be thoroughly familiarized with the external genitalia with a mirror so that she can recognize her own clitoris, urethra, and distal vagina. In this way, she learns how to place the dilator in the appropriate location at the appropriate angle. Increasingly larger dilators are inserted into the distal vagina and advanced towards the apex. The application should be between 10 and 30 minutes three times a day. Patients using dilators are evaluated every two weeks to assess the extent of progress. Pain or bleeding may occur during dilatation, patients should be informed about this and encouraged to continue dilatation, lubricants can be used if necessary, or softer dilators can be tried. With passive vaginal dilatation, there is no limit to the vaginal length to be achieved before sexual intercourse. The fact that patients can have vaginal intercourse in a comfortable and functional way is considered as a success (11).

Laparoscopic Vecchietti Vaginoplasty

In this operation, an elliptical body called "olive" is placed at the apex of the distal vagina to provide rapid vaginal elongation. The ropes hanging on the sides of the olive at the vaginal apex are passed through the laparoscopically created vesicorectal and retroperitoneal space in the anterior abdominal wall and fixed to the traction device on the abdominal wall. A vaginal length of 9-10 cm is obtained in approximately one week by daily traction of 1-1.5 cm. Afterward, the olive in the vagina and the traction device on the anterior abdominal wall are removed. Afterward, the patient continues passive vaginal dilatation with a suitable dilator for one month. Sexual intercourse can start one month after the use of the dilator. If there is no sexual intercourse, the patient should continue passive dilatation (12-14). The most common problems encountered are postoperative pain, fever, and urinary tract infections. Rarely, cases of urethral necrosis due to the pressure of the olive have been reported in the literature. At the end of the operation, anatomical success is achieved in 98% of the patients and the average vaginal length is 9.5 cm. Functional sexual intercourse requires 12-16 weeks. In some patients, granulation may develop in the newly formed vaginal tissue and this condition is treated with silver nitrate (15).

Davydov Vaginoplasty

In Davydov vaginoplasty, the vesicorectal space is dissected vaginally to the level of the pelvic peritoneum, then the pelvic peritoneum is mobilized laparoscopically and the free peritoneum is fixed to the introitus with sutures. The proximal side of the peritoneal cavity is sutured and closed, thus creating a neovagene. Afterward, a mould is placed in the neovagina formed with peritoneum for one week. The patient is not mobilized during this period. After the mould is removed, the use of a dilator or regular sexual intercourse is essential. After a successful operation, a vagina length of 7-8 cm can be obtained. Up to 10% of patients experience dyspareunia or dissatisfaction with sexual intercourse. Other rare complications include rectal perforation, rectoneovaginal fistula, bladder injury, and neovaginal prolapse requiring sacrocolpopexy and pelvic adhesions (14,16,17).

Abbe-McIndoe Vaginoplasty

In Abbe-McIndoe vaginoplasty, the vesicorectal space is vaginally dissected to the level of the pelvic peritoneum, usually about 10 cm in length. A full-thickness skin graft, usually from the buttocks or thighs, is placed on a mould that is inserted into the previously dissected vesicorectal. The mould is left in place for one week and then removed in the operating theatre and the condition of the graft is assessed. Patients should then continue passive dilatation and regular sexual intercourse for 6 months. The reported anatomical success of Abbe-McIndoe vaginoplasty is around 80-90%. Pain and cosmetic problems may occur especially in the area where the skin graft is taken. In patients with such concerns, synthetic grafts that will provide neovaginal epithelialization or buccal mucosa can be used instead of skin grafts (18-21).

Intestinal Vaginoplasty

In intestinal vaginoplasty, which is usually performed by pediatric surgeons and urologists, the sigmoid is most commonly used to create a neovagina. The sigmoid colon, which is approximately 10-12 cm long, is mobilized with care to preserve blood flow. The proximal part of the sigmoid colon neovagina is fixed inside the abdomen to prevent prolapse, and the distal part is sutured introitus. There is no need for dilatation after the operation. If there is no regular sexual activity, stenosis may occur in the introitus, but most of these can be opened with dilatation. If neovagina prolapse occurs, sacrospinous fixation can be performed (22).

CONCLUSION

Absolutely, understanding genitourinary embryology is crucial for diagnosing and treating genital malformations. The development of the genitourinary system is a complex process that involves the differentiation of structures from the mesoderm and the interaction of various signaling pathways. Anomalies can arise at different stages of development, leading to a range of conditions that may affect both the urinary and reproductive systems. In young patients, these malformations can have significant implications for their health and quality of life, including issues related to fertility, sexual function, and psychosocial well-being. Therefore, a multidisciplinary approach is often necessary for management, involving urologists, gynecologists, endocrinologists, and geneticists, among others. Referral to specialized centers that have experience in managing complex genital malformations is essential. These centers can provide comprehensive care, including advanced imaging, surgical interventions, and long-term follow-up, ensuring that patients receive the best possible outcomes. Early diagnosis and intervention can significantly improve the prognosis for individuals with these conditions. In addition to the patient's expectations, surgical experience is extremely important in the choice of surgical method. Because the success of the first operation is decisive in the success of subsequent operations if necessary. Whether the patients used passive methods or surgical vaginoplasty, as a result, they had a similar length of a neovagina, and the female sexual function scale scores were similar between the groups. Among the surgical methods, the need for lubricant use during intercourse was seen more in patients who underwent Abbe-McIndoe vaginoplasty compared to other methods.

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