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Innovative Diagnostic Methods: The Role of MRI and Ultrasound in Detecting Uterine and Vaginal Anomalies

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Abstract Congenital anomalies of sexual development often lead to serious disorders of the reproductive system and are associated with an unfavorable prognosis for fertility. However, in recent years, thanks to the active development of assisted reproductive technologies, it has become possible to significantly improve reproductive outcomes in infertile patients. The key factor for successful treatment in such cases remains the timely and accurate diagnosis of congenital anomalies, as well as a well-designed management strategy for such patients. The aim of this study is to summarize modern approaches to the diagnosis of congenital anomalies of the reproductive organs and to analyze the role of genetic factors in their formation. The paper reviews clinical, instrumental, and laboratory diagnostic methods, including ultrasound, MRI, endoscopic methods, and molecular genetic studies, which allow not only to identify the anomaly but also to determine its etiology. The work aims to increase awareness of the importance of early diagnosis and genetic counseling, and emphasizes the importance of a personalized approach when planning treatment and rehabilitation for women with congenital genital defects.

Keywords Congenital anomalies of the reproductive organs, Bicornuate uterus, Genetics, Diagnosis, Uterine and vaginal aplasia

The issue of human reproductive health encompasses biological, medical, social, and philosophical aspects. The expansion and deepening of scientific research in this area is largely due to the relevance of demographic issues [1]. In developed countries, special attention is paid to the early diagnosis and prevention of gynecological diseases, which contributes to their prevention and timely treatment.

In Europe, the practice of early reproductive health screening for women is widespread. If girls aged 12-13 show no signs of puberty, specialists recommend diagnostic measures [2]. According to experts, timely detection of abnormalities in the development of the reproductive organs allows for the correct choice of treatment strategy, which in the future contributes not only to a reduction in the prevalence of gynecological diseases, but also to an improvement in the quality of life of patients [3].

Comprehensive diagnosis of congenital anomalies of the female reproductive system requires the use of both traditional and modern research methods. To obtain an objective picture of pathological changes, a combination of non-invasive and invasive techniques is used, each of which plays a specific role in refining the clinical diagnosis.

One of the basic tools for primary diagnosis is ultrasound examination (2D ultrasound), which is widely used as a screening method for detecting abnormalities in the structure of the reproductive organs. This method is highly accessible and safe, and also provides an initial overview of the structural features of the area being examined. However, its diagnostic accuracy is limited and largely depends on the level of training of the specialist conducting the examination, as well as on the technical characteristics of the equipment [4].

The method of three-dimensional ultrasound reconstruction (3D ultrasound) demonstrates high diagnostic accuracy and reproducibility of the data obtained. Its use provides advanced visualization capabilities, allowing for a more detailed assessment of the anatomical structure of the lower reproductive system, including the cervix and vagina. However, its use is limited by its higher cost and the need for specialized training of the diagnostic physician, which makes it less accessible compared to 2D ultrasound [5].

Magnetic resonance imaging (MRI) is considered the most reliable method for diagnosing congenital anomalies of the female reproductive organs. This method provides three-dimensional visualization of the anatomy of the pelvic organs, with the exception of the fallopian tubes, and allows for the detection of various developmental defects, including obstructive anomalies. Despite its high diagnostic accuracy, MRI is an expensive method that requires a qualified specialist to interpret the images obtained, which limits its widespread use [6].

Echohysterosalpingography (EHSG) is a minimally invasive diagnostic method that allows assessment of the condition of the cervix and uterine cavity. This method is relatively safe, but its accuracy largely depends on the qualifications of the specialist performing the examination. It

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is important to note that incorrect insertion of a rubber catheter into the uterus can cause tissue trauma, and expanding the uterine cavity with air and forming a balloon in the catheter can lead to misinterpretation of the internal contours, which in some cases causes false positive or false negative results.

X-ray hysterosalpingography (X-ray HSG) is used to visualize the uterine cavity and fallopian tubes. Despite its diagnostic value, this method is considered invasive and is accompanied by discomfort or pain for the patient. The main disadvantage of RHSG is its inability to provide information about the external contour of the uterus, as well as the inability to differentiate between an intrauterine septum and a bicornuate uterus. In addition, this method does not allow the detection of an additional closed horn and cannot be used in cases of cervical or vaginal obstruction [7].

Hysteroscopy is a minimally invasive diagnostic procedure that allows for visual assessment of the condition of the uterine cavity mucosa, including the cervical canal and vagina. This method provides highly informative data on the internal structure of the uterus, but does not provide information on the external contours of the organ or the thickness of its walls.

Laparoscopy is used for comprehensive visualization of the reproductive organs, which, through minimal incisions, allows pathological processes to be visualized on a screen and simultaneously corrected surgically. However, laparoscopy does not always provide an accurate assessment of the thickness of the uterine wall.

Thus, to obtain complete information about the condition of the uterus and its structures, it is advisable to use a combination of various diagnostic methods, including hysteroscopy, laparoscopy, and ultrasound examination [8].

In some cases of Mayer-Rokitansky-Küster-Hauser syndrome (MRKH), one or two additional uterine structures are found in the lateral parts of the pelvis. These rudimentary structures may contain functional endometrium, which causes clinical manifestations such as cyclic pain or hematometra formation. Initial diagnosis usually begins with an ultrasound examination (US), which allows the absence of the uterus and vagina to be detected, as well as the degree of vaginal aplasia and the presence of rudimentary uterine structures to be determined. However, magnetic resonance imaging (MRI) is recommended for a more detailed assessment of anatomical features and identification of associated anomalies. MRI provides high-quality images, allowing visualization of rudimentary uterine formations, their structure, and the presence of functional endometrium, as well as assessment of the condition of the ovaries, which are often located high on the wall. In addition, MRI effectively detects associated anomalies in the development of the urinary system, such as unilateral kidney, pelvic kidney dystopia, or duplication of the pyelocaliceal system. Thus, the combined use of ultrasound and MRI provides an accurate diagnosis and allows for the development of optimal management strategies for patients with MRKH. If the horn consists of fibrous tissue, its structure is defined as low-intensity on T2-weighted images. An important aspect of diagnosis is the assessment of the connection between the rudimentary horn and the main uterine cavity, which requires the combined use of HSG and MRI [9].

During hysteroscopic examination, the cavity of a unicornuate uterus appears rounded, unlike the triangular shape characteristic of a normal structure. The single opening of the fallopian tube is a diagnostic marker of this anomaly. In cases where there is a rudimentary horn that is not connected to the main cavity, scar tissue is detected in the area where it branches off. Laparoscopic examination allows additional confirmation of the presence of a rudimentary horn, which is usually smaller than the main uterus and located laterally, slightly above the internal os [10].

A bicornuate uterus is characterized by the presence of two symmetrical cavities that are partially connected at the bottom, mainly in the area of the uterine isthmus. In some cases, the cavities are separated up to the internal os. Regardless of the form of this anomaly, in the presence of a bicornuate uterus, as a rule, a single cervix is determined and communication between the uterine cavities is preserved. The most informative methods for diagnosing this anomaly are ultrasound (US) and magnetic resonance imaging (MRI), which allow clear visualization of both the internal and external contours of the uterus. Unlike hysterosalpingography (HSG), hysteroscopy, and laparoscopy, ultrasound provides a non-invasive and more complete assessment of the anatomical features of the organ, especially when using three-dimensional reconstruction. HSG and hysteroscopy only provide information about the internal contours and boundaries of the uterus, while laparoscopy only provides information about the external boundaries. Therefore, it is recommended to use endoscopic methods in combination with each other to improve the accuracy of diagnosis and select the optimal treatment strategy.

On ultrasound and MRI, a double uterus and vagina appear as two separate uterine bodies that do not touch each other and are located at a distance from each other. It is also necessary to be able to diagnose two cervixes. In some cases, patients are found to have a double vagina with a longitudinal septum between the two channels. For the differential diagnosis of intrauterine septum, magnetic resonance imaging (MRI) allows the external contour of the uterus to be assessed: if the external contour remains uniform, the diagnosis is more likely to be intrauterine septum. In cases where MRI shows a slight concavity in the area of the uterine fundus, this is referred to as a saddle-shaped uterus. If the depression reaches the level of the internal os or penetrates deeper, a bicornuate uterus is diagnosed.

Contrast enhancement during echosalpingography (ESG), when a substance is injected into each of the cervical os, allows visualization of isolated horns that morphologically resemble a unicornuate uterus. At the same time, if the upper contour of the uterus remains continuous, the most likely diagnosis is an intrauterine septum. A slight depression at the bottom of the uterus is interpreted as a saddle-shaped form, and a pronounced cleft reaching the level of the internal os is interpreted as a bicornuate uterus with two cavities and two cervixes.

Ultrasound examination does not always allow a clear distinction between a complete and partial intrauterine septum. To diagnose a saddle-shaped uterus, ultrasound, hysterosalpingography, and MRI are used to determine the smooth expansion and flattening of the bottom of the uterine cavity, which is characteristic of this shape. A saddle-shaped uterus is considered a mild form of bicornuate uterus, but there is disagreement among specialists regarding its interpretation and clinical significance.

Diagnosis of developmental defects is possible using ultrasound, MRI, echo-salpingography (ESE), and hysteroscopy. Laparoscopy, unlike these methods, does not provide information about the presence of structural abnormalities. The introduction of modern imaging methods has contributed to an increase in the detection rate of congenital malformations of the uterus and vagina, especially rare forms [1,2,3,5].

When contrast is injected during echosalpingography into a separate cervix on both sides, it is possible to detect two separate uterine horns that resemble a unicornuate uterus in structure. One of the pathologies that is difficult to diagnose differentially and has a similar visual picture on hysterosalpingography (HSG) is a complete intrauterine septum. The use of magnetic resonance imaging (MRI) allows this condition to be identified with high accuracy, thanks to the ability to assess the outer contour of the uterine fundus, which remains unchanged in the presence of a septum.

Thus, comprehensive diagnosis of congenital anomalies of the reproductive system is of key importance. Comparison of the results of various imaging methods, as well as assessment of their informativeness and accuracy, allows for the formation of an objective picture of the nature of the anomaly and the selection of the optimal management strategy for the patient [1,2,3,5].

Congenital anomalies and malformations of the genitourinary system are characterized by anatomical and structural changes in the organ itself or its absence in the early stages of intrauterine development. The etiology of such anomalies can vary and may be associated with heredity or gene mutations.

Scientists believe that developmental abnormalities arise as a result of endogenous and exogenous factors [1,2,3,5,7,9].

Many scientists say that malformations of the genitourinary system are more related to exogenous factors such as hyperthermia, hypothermia, radiation, chemicals, etc. According to other scientists, 12-24% of developmental abnormalities of the genitourinary system depend on gene mutations, chromosomal abnormalities in the form of translocation or deletion [1,2,3,5], and in 66% of cases, the cause of developmental abnormalities of the genitourinary system remains unclear.

Mayer-Rokitansky-Küster-Hauser syndrome (MRKH) is of considerable scientific interest because it is characterized by the congenital absence of the uterus, cervix, and upper two-thirds of the vagina in phenotypically normal girls with a female karyotype of 46. Agenesis or severe hypoplasia of the female reproductive organs can also be observed in a number of rare genetic syndromes, such as McCusick–Kaufman syndrome (mutations in the MKKS gene, located in the 20p12 region), Bardet–Biedl syndrome (involving MKKS and other genes), Wolf-Hirschhorn syndrome (deletions in the 4p16.3 region), and Goldenhar syndrome. These observations indicate a possible commonality in the pathogenetic mechanisms underlying these disorders [1,6].

The polygenic nature of vaginal and uterine agenesis suggests the presence of molecular abnormalities affecting early embryogenesis during the fusion of the Müllerian ducts. This syndrome is also associated with kidney abnormalities, as the kidneys are also formed from the mesoderm [2,3].

Previously, it was believed that uterine-vaginal aplasia (MRKH syndrome) occurred predominantly sporadically. However, an increasing number of reported familial cases confirm the genetic nature of this condition. In such familial observations, the syndrome is transmitted in an autosomal dominant pattern with variable expression and incomplete penetrance. This indicates the possibility of mutations in key genes regulating embryonic development or limited chromosomal abnormalities. According to the literature, only 68 cases of the familial form of MRKH syndrome have been described [1,8].

According to L.V. Adamyan et al. (2008), genetic factors play a significant role in the pathogenesis of congenital anomalies of the uterus and vagina, as well as in the development of endometriosis [2,4]. The observed association between MRKH syndrome and ectopic endometriosis in a number of clinical cases suggests the presence of a common pathogenetic mechanism. A hypothesis has been put forward about the multifactorial nature of these diseases, based on genetic polymorphisms, hereditary predisposition, and the influence of hormonal background, in particular, the effect on estrogen and progesterone receptors [11].

Scientific literature pays particular attention to the role of genes from the WNT, HOXA, and PAX families involved in the processes of reproductive system embryogenesis. WNT genes, in particular Wnt4 and Wnt9b, play an important role in the formation of the urogenital system: Wnt4 participates in sexual differentiation and regulates the invasion of luminal epithelial cells; Wnt9b is expressed in the epithelium of the Wolffian ducts and contributes to the elongation of the Müllerian duct [1].

In humans, Wnt4 was the first gene identified as being associated with uterine dysgenesis and hyperandrogenism. Mutations in this gene lead to impaired suppression of steroidogenic enzyme activity in the ovaries and pathological expression of 17α-hydroxylase, which causes the development of hyperandrogenism (Biason-Lauber et al.). In a study by Philibert R. et al. (2008), mutations causing increased expression of androgen-synthesizing enzymes were identified in 28 girls with primary amenorrhea and uterine and vaginal dysplasia. These data allow us to consider MRKHS with hyperandrogenism as a separate clinical variant of the disease.

At the same time, a study by Ravel C. et al. (2009) failed to identify mutations in the Wnt7a gene in patients with MRKH syndrome. Later, Wang et al. (2014) first reported mutations in the Wnt9b gene associated with the syndrome in the Chinese population. However, subsequent studies did not confirm a consistent correlation between Wnt9b and this disease. Nevertheless, more recent studies have again pointed to the role of Wnt9b in the etiology of MRKH: five heterozygous missense mutations and one heterozygous nonsense mutation were found in patients with type I syndrome, confirming its pathogenetic significance.

The genes of the homeobox family, in particular the HOX cluster, play an important role in the development of the female reproductive system. Among them, HOXA9–HOXA13 and HOXB9–HOXB13 are of particular interest, as they are considered potential candidates in the formation of MRKH syndrome [3]. In humans, mutations in HOXA13 or deletions of the entire HOXA cluster can have a pronounced effect on the development of the genitourinary system and the musculoskeletal system. Thus, mutations in the coding region of the HOXA13 gene cause palpebral-genital syndrome (HFGS), accompanied by a disruption in the fusion of the Müllerian ducts, which manifests as vaginal septum, cervical duplication, and urinary tract malformations.

Interestingly, deletions of the entire HOXA cluster do not lead to more pronounced urogenital anomalies compared to an isolated mutation in HOXA13. This suggests that dominant mutations in HOXA9, HOXA10, or HOXA11, or a disruption in the regulation of HOXA cluster gene expression, affecting the rate of transcription and the spatiotemporal characteristics of their activity. Recent data on mutations in the HOXA13 promoter region further support this assumption.

The expression of individual HOXA genes varies depending on the anatomical area: HOXA9 is predominantly active in the fallopian tubes, HOXA10 and HOXA11 in the body and cervix of the uterus, and HOXA13 in the distal parts of the vagina. In addition, the WT1, PAX2, HOXA7–13, and PBX1 genes are also considered potential candidates in the etiology of MRKH syndrome. However, experimental studies using mutant mouse models have not yet confirmed their unequivocal involvement in the development of this pathology.

Of particular interest are clinical cases in which MRKX syndrome develops in only one of identical twins, while the other remains phenotypically healthy. This indicates the possible role of not only genetic but also epigenetic and environmental factors in the pathogenesis of the disease [3]. In a study by Rail et al. (2011), significant differences in DNA transcription and methylation levels were found between patients with MRKX and the control group. Genomic analysis identified nine key genes: HOXA5, HOXA9, WISP2, CDH5, PEG10, MFAP5, LRRC32, and RALGPS2. Six of them (CDH5, MFAP5, WISP2, HOXA5, PEG10, HOXA9) demonstrate significant activity in the development of female reproductive system structures. Network bioinformatic analysis showed that WISP2, HOXA5, HOXA9, GATA4, and WT1 may be central regulators, suggesting their leading role in the pathogenesis of MRKH syndrome.

The WT1 and GATA4 genes are involved in regulating sexual differentiation by influencing the expression of anti-Müllerian hormone (AMH). Their activity promotes the production of anti-Müllerian hormone (AMH), which initiates the degeneration of Müllerian ducts and plays a key role in the formation of the female reproductive system [2].

According to Rail et al. (2011), excessive estrogen influence combined with ectopic expression of HOXA cluster genes may contribute to female genital hypoplasia and the development of MRKH syndrome [9]. In turn, De Tomasi F. et al. (2017) described clinical cases of uterine and renal aplasia, as well as developmental disorders of both the urinary and reproductive systems, confirming the importance of the GREB1L gene (formerly known as GREB1F) in the pathogenesis of these anomalies. Later, Herlin M.K. et al. (2019) identified GREB1L as a promising candidate gene involved in the etiology of MRKH syndrome.

Some studies have identified a link between developmental abnormalities, such as bicornuate uterus and Müllerian duct dysplasia, and mutations in the TCF2 gene (also known as HNF1B). Defects in this gene may explain rare cases of congenital aplasia, especially when combined with renal developmental abnormalities or a family history of diabetes, making it an important target for further study of genetic associations with MRKH syndrome.

In addition, some authors point out that a significant number of sexual development disorders are caused by chromosomal abnormalities affecting chromosomes 1–7, 10–18, 22, and the X chromosome. However, according to the summarized results of a number of genomic studies, only five recurring structural rearrangements (deletions and duplications) were identified with high frequency: 1q21.1, 16p11.2, 17q12, 22q11.21, and Xp22. These abnormalities were found in 28 patients with MRKH syndrome and account for about 10% of all reported cases of the disease [11].

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