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Imaging and Diagnostic Challenges in an 11-Year-Old Girl with Vaginal Agenesis: A Case Report

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Data Collection B
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Patient: Female, 11-year-old
Final Diagnosis: Distal 1/3 vaginal agenesis • hematometra et hematocolpos
Symptoms: Cyclic abdominal pain persisting for the last three months
Clinical Procedure: Exploratory laparotomy • omentectomy • vaginoplasty
Specialty: Obstetrics and Gynecology





Objective: Unusual clinical course
Background: The prevalence of female genital tract anomalies is around 4-6.9%. Vaginal agenesis is a form of Müllerian agenesis and defined as the congenital absence of the vagina. It affects 1 in 5000 women. During normal fetal development, the Müllerian ducts combine to form the uterus, fallopian tubes, and most of the vagina. However, in cases of vaginal agenesis, the Müllerian ducts fail to fuse to form the upper part of the vagina. This report presents the case of an 11-year-old girl with vaginal agenesis evaluated using ultrasound.

Case Report: An 11-year-old girl, Tanner stage II, presented with cyclic abdominal pain for 3 months and no menarche. After a hymenectomy for imperforate hymen in January 2023 failed to relieve symptoms, she was referred to our hospital. Physical examination showed a tender lower abdomen with no genital abnormalities. Transabdominal ultrasound was inconclusive for differentiating between a transverse vaginal septum and cervical agenesis with hematometra, and magnetic resonance imaging suggested hematometrocolpos due to a transverse vaginal septum. Intraoperatively, distal vaginal agenesis was identified, and vaginoplasty was performed, evacuating 200 mL of thick blood. The cervix and uterus were normal, and postoperative management included gradual vaginal dilation with a soft mold. This case highlights the diagnostic challenges of distal vaginal agenesis, particularly in patients with a history of imperforate hymen.

Conclusions: This report emphasizes the importance of accurate imaging and clinical evaluation in diagnosing reproductive anomalies such as distal vaginal agenesis. Developing techniques to enhance the specificity of imaging modalities is crucial for distinguishing vaginal agenesis from a transverse vaginal septum, ensuring appropriate surgical management and better patient outcomes.

Keywords: Diagnosis • Hematometra • Magnetic Resonance Imaging • Uterus Bicornis Bicolis with Partial Vaginal Septum and Unilateral Hematocolpos with Ipsilateral Renal Agenesis • Absence of Vagina

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Introduction

Congenital vaginal agenesis has a prevalence of 1 in 4000-10 500 live female births. Among these cases, less than 10% of these patients show normal development of the uterine corpus [1]. Distal vaginal agenesis is a congenital abnormality of the female reproductive tract that causes obstruction. Fibrous tissue replaces the distal section of the vagina, resulting in the obstruction of menstrual blood flow and the impairment of cervical gland secretions [2]. Vaginal agenesis has an incidence of 1 in every 4000 to 10 000 girls. The most common cause of vaginal agenesis is congenital absence of the uterus and vagina, which also is referred to as Müllerian aplasia, Müllerian agenesis, or Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome [3]. Hemato/hydrocolpos is a medical condition in which menstrual blood or secretory fluid accumulates in the vagina as a result of vaginal obstruction. Hemato/hydrocolpos can be caused by congenital urogenital anomalies or be acquired as a result of vaginal occlusion [4].

The typical onset and progression of physical maturation differ significantly according to sex, race, ethnicity, and environmental influences. Tanner stages are employed in pediatric and adolescent practice to advise patients on the timing of expected bodily changes, conduct relevant medical screenings, and track deviations in the normal timing and sequence of physical signs of puberty that may indicate physiological issues. Tanner staging is essential in distinguishing between endocrinological problems, such as hormonal imbalances, and structural problems that may impair normal pubertal development [5,6].

Distal vaginal agenesis defect typically presents during the anticipated onset of menstruation with primary amenorrhea and cyclic lower abdomen pain due to cryptomenorrhea, resulting in progressive hematocolpos and hematometra. The diagnosis is established via a gynecological examination, and the probable aberration is verified through ultrasound testing and magnetic resonance imaging (MRI) [2]. The condition usually can be successfully managed nonsurgically with the use of successive dilators if it is correctly diagnosed and the patient is sufficiently motivated. In addition to accurate diagnosis, effective management entails assessing for related congenital renal or other anomalies, as well as thorough psychological preparation of the patient prior to any treatment or intervention [3].

In the embryological process, the upper parts of the Müllerian tubes do not fuse and form the fallopian tubes. On the other hand, in the lower part of the development process, the Müllerian ducts will unite and eventually form the uterus and the upper part of the vagina. Once the caudal end of the converging Müllerian duct reaches the urogenital sinus, endodermal cells originating from the sinus proliferate and form a solid synovaginal bulb. The synovaginal bulb will develop and

form the vaginal plate. Proliferation continues at the end of the cranial plate [7]. The resulting proliferation will increase the distance between the developing uterus and the urogenital sinus, and the nucleus of the synovaginal bulb will degenerate and form a cavity at 17-18 weeks gestation. In the fifth month of pregnancy, complete canalization occurs in the vaginal plate. The upper part of the vagina originates from the Müllerian duct, and the lower part of the vagina originates from the synovaginal bulb. The fusion of the Müllerian ducts forms the uterovaginal canal, which forms the uterus, cervix, and upper 2/3 of the vagina. The distal contact point of the Müllerian duct with the urogenital sinus is the site of the proliferation of the urogenital sinus epithelium, forming the synovaginal bulb. Moreover, the synovaginal bulb develops into the vaginal plate [4].

Immediately after the thick end of the paramesonephric duct touches the urogenital sinus, 2 protrusions form from the pelvic section of the sinus. Evagination, also known as a synovaginal bulb, is the process by which a dense vaginal plate is formed through the proliferation of cells. Cell division persists at the upper part of the body, causing the gap between the uterus and the urogenital sinus to widen [8]. The vaginal lumen is kept distinct from the urogenital sinus by a delicate layer of tissue called the hymen. This layer is composed of the epithelial lining of the sinus and a thin layer of vaginal cells [9]. If distal agenesis occurs, the synovaginal bulb, which has 2 origins, does not undergo canalization in the vagina. Fibrous tissue replaces the distal section of the vagina, leading to hematocolpos due to the absence of a passage for menstrual blood [2].

The vaginal septum is created due to the failure of the border between the Müllerian ducts and the urogenital sinus to be absorbed. This anomaly will result in the division of the vagina into 2 distinct segments, reducing its functional component. The vaginal septum can have either permeable or impermeable characteristics, and it can impact all aspects of the vagina. The thickness of the vaginal septum varies, with a greater thickness observed higher up, particularly at the point of insertion, but always less than 1 centimeter [10].

MRI is often regarded as the most reliable imaging tool for diagnosing Müllerian and vaginal abnormalities. It is highly recommended to undergo an MRI of the internal genitalia if possible. MRI is a non-invasive method that surpasses computed tomography (CT) in providing a detailed visualization of the Müllerian structures, such as uterine remnants or total agenesis [9]. It can accurately detect the existence of endometrium in uterine remnants. MRI is capable of visualizing both the ovaries and extragenital abnormalities. A pelvic MRI scan confirms the diagnosis by detecting the existence of underdeveloped uterine buds or complete absence of the uterus and vagina [11]. A renal scan, utilizing either ultrasonography or MRI, is

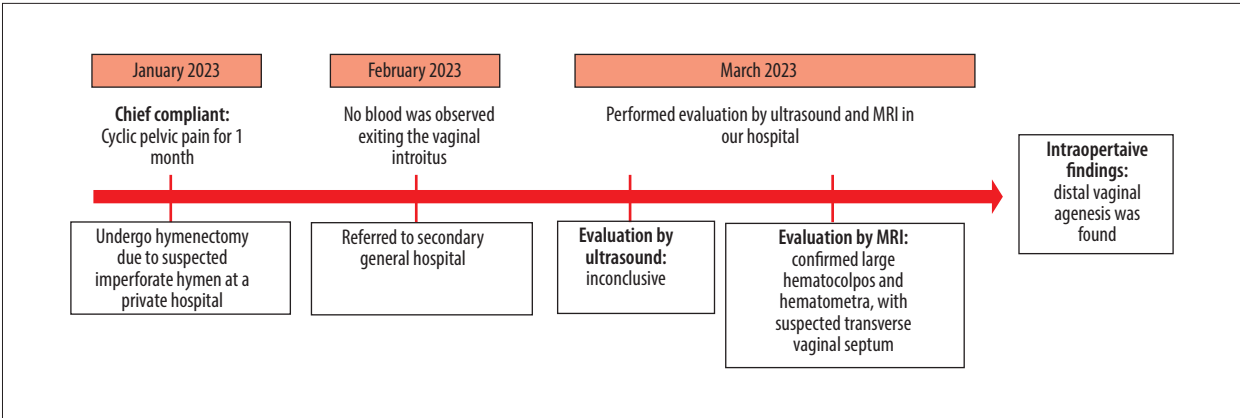


Figure 1. Chronological timeline of the patient. The figure illustrates the diagnostic and treatment journey of the patient from January to March 2023. In January, the patient presented with cyclic pelvic pain for 1 month and underwent a hymenectomy at a private hospital due to suspected imperforate hymen. In February, no vaginal bleeding was observed following the procedure, prompting a referral to a secondary general hospital for further evaluation. In March, an ultrasound examination was inconclusive, but an MRI confirmed the presence of a large hematocolpos and hematometra, with a suspected transverse vaginal septum. During surgery, distal vaginal agenesis was identified as the underlying condition.

recommended to assess the existence of abnormalities, which are identified in approximately 30% of patients [12]. However, the case of a 20-year-old woman published by Petrykowski et al found that misdiagnosis may also occur from MRI images [13]. The present report presents the case of an 11-year-old girl with vaginal agenesis evaluated using ultrasound. The report emphasizes the importance of accurate imaging and clinical evaluation in diagnosing reproductive anomalies such as distal vaginal agenesis (Figure 1).

Case Report

An 11-year-old girl, classified as Tanner stage II based on the Sexual Maturity Rating system, presented with a history of cyclic abdominal pain persisting for the last 3 months, alongside an absence of menarche, in March 2023. Tanner staging, a widely used classification system for secondary sexual characteristics during puberty, assesses the development of breast tissue, pubic hair, and other markers of physical maturation. This system is crucial in evaluating the pubertal status of pediatric patients. In this case, the Tanner stage II classification indicated early pubertal development, with the appearance of breast buds and sparse pubic hair.

The patient had been diagnosed with an imperforate hymen and underwent a hymenectomy in January 2023. However, post-procedure, no blood was observed exiting the vaginal introitus, prompting referral to a secondary general hospital for further evaluation. A physical examination revealed a convex abdomen with tenderness localized to the lower abdominal region. External genital examination showed no abnormalities in the vulva, and the vaginal introitus was present.

Additional diagnostic investigations included a transabdominal ultrasound, which yielded inconclusive results for differentiating between a transverse vaginal septum and cervical agenesis, both associated with hematometra. The patient declined a transvaginal ultrasound examination due to pain and was otherwise uncooperative during the procedure. Visualization through transabdominal ultrasound suggested the presence of a transverse vaginal septum in the proximal vagina, though the thickness of the partition wall remained unclear.

MRI subsequently confirmed hematometrocolpos, caused by a transverse vaginal septum located in the distal third of the vaginal canal (Figure 2). The imaging also revealed that the distal vaginal structure was not visible, with the most distal segment distended to the peritoneum or cutis, measuring up to 2.7 cm. The partition wall was found to be 10 mm in thickness and 7 mm in height. Based on these findings, the pre-operative diagnosis was determined to be transverse vaginal septum.

The main diagnostic challenges from this case report were based on initial suspicion of cervical agenesis because no cervix was found in the ultrasound, but the vaginal septum was present. Further evaluation with MRI was conducted, which found that a large hematocolpos had hindered the ultrasound ability to visualize the cervix. Initially, the distal vaginal agenesis was not visualized clearly by MRI, and the history of previous hymenectomy from imperforate hymen in this case may have distorted the anatomical structure within the vaginal area.

During the operation, the patient was reassessed, and the vaginal canal was not visible. It was concluded that the patient had distal 1/3 vaginal agenesis, and vaginoplasty was performed by a horizontal incision of the anteroposterior part of the vagina.

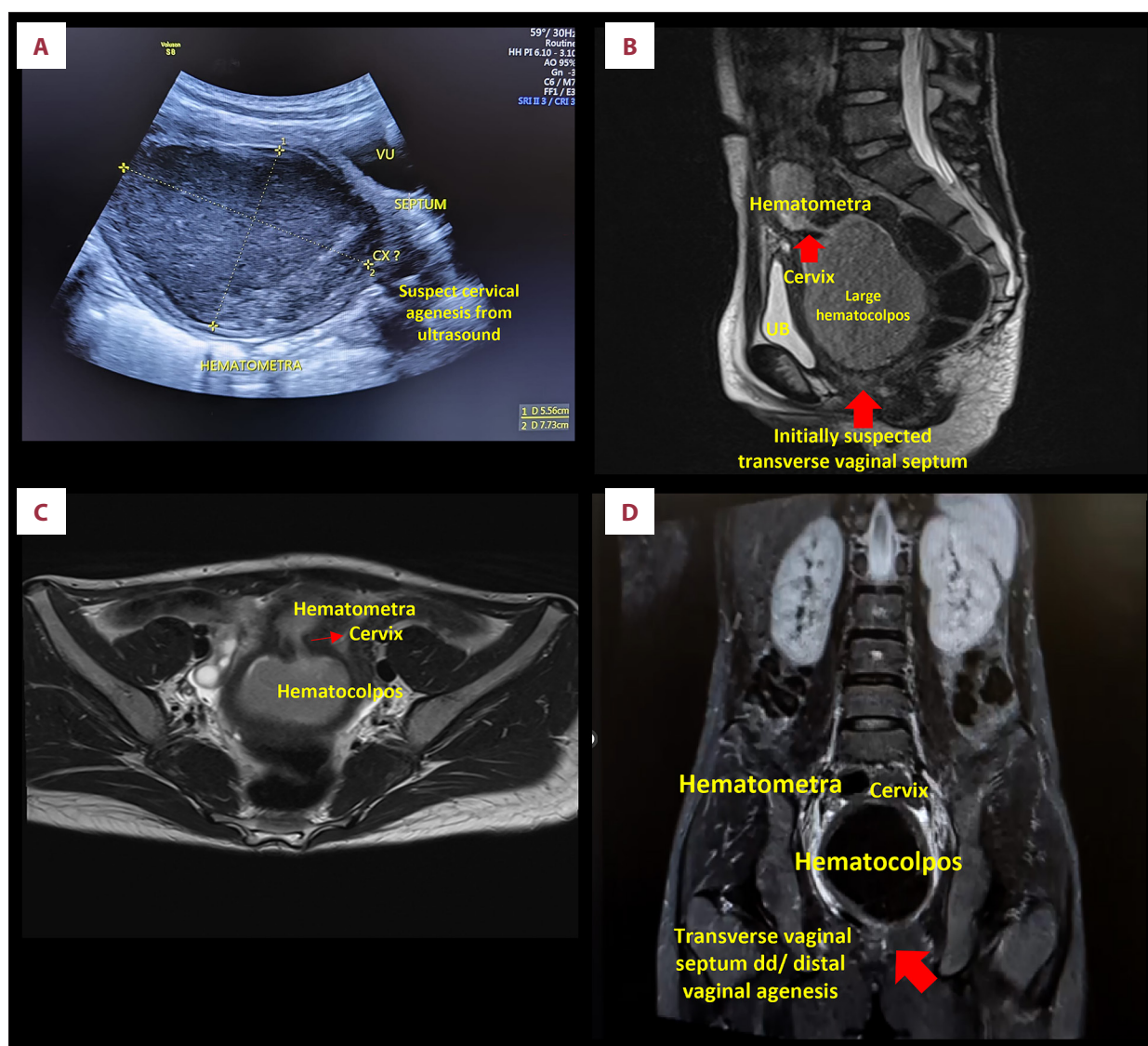


Figure 2. Pre-diagnostic imaging. (A) Transabdominal ultrasound found a hypoechoic mass in the uterine cavity 5.56×7.73 cm. The transverse septum was visualized on the proximal vagina. The cervix was not clearly visualized. (B) MRI scan found hematometrocolpos with transverse vaginal septum on the distal third of the vaginal canal (red arrow). (C) The cervix was visualized (red arrow). (D) A distal vaginal structure was not visualized. The distance between the most distal vaginal canal (distended) and the perineum/cutis was 2.7 cm (red arrow). UB – urinary bladder; CX – cervix.

After the vaginal distal incision with approximately 1 cm depth, 200 ml of reddish-brown thick blood came out. During exploration, the cervix was found to be normal, and the anteroposterior vagina was sutured to the labia minora using a simple interrupted suture technique (Figure 3). During exploration, the uterus, measuring 5x4x3 cm, showed no adhesions to surrounding organs. Both fallopian tubes were within normal limits. Blood clots were observed adhering to the omentum. The decision was made to perform a partial omentectomy. Palpation of the cervix was conducted, and the cervix appeared normal. Postoperative management for vaginoplasty, in this case, is using a gradual dilation technique with the aid of a soft plastic mold.

Discussion

This case highlights the diagnostic challenges of distal vaginal agenesis, particularly in patients with a history of imperforate hymen. Presentation of vaginal agenesis is commonly at menarche with complaints of amenorrhea and cyclic abdominopelvic discomfort secondary to hematometra, hematocolpos, and hematosalpinx. Often, external genitalia are normal, and a tiny vaginal pouch is generally visible with hymenal tissue present. Blood products retained in the uterus may be palpated on a rectovaginal exam [13]. Ultrasonography can be performed to determine the presence and anatomy of the uterus,

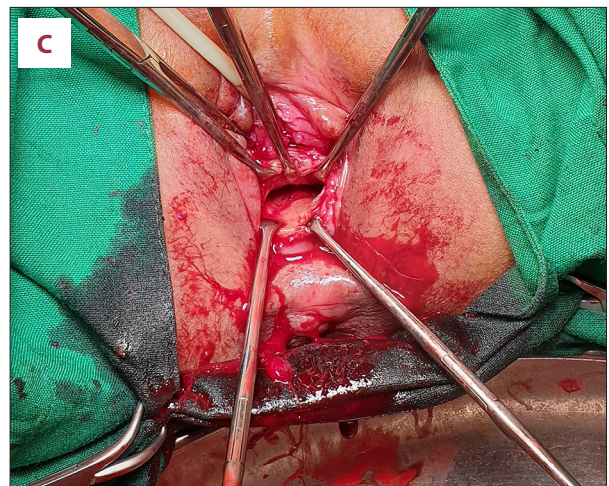
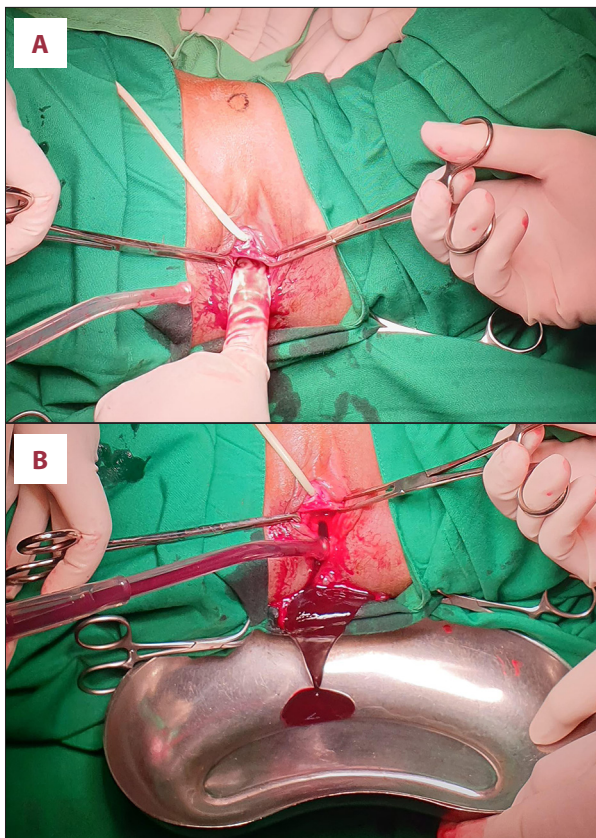


Figure 3. Intraoperative finding and procedure. (A) The vaginal canal was not visible, giving the impression of distal third vaginal agenesis. (B) An incision was made in the distal vagina along 1 cm and then expanded bluntly to the side. From within the body, 200 ml of reddish-brown thick blood came out. (C) Interrupted suturing with PGA 2.0 thread between the anteroposterior vaginal mucosa and the labia minora was performed.

cervix, and upper vagina [14]. MRI may be useful to determine whether the endometrium is functioning [13]. While first-line treatment is ideally non-surgical, the surgical construction of a neovagina is a more immediate option [15].

MRI is recognized as a precise and reliable method for assessing Müllerian duct defects. As a result, it is frequently employed in clinical settings [16,17]. MRI helps identify uterine and vaginal abnormalities and allows for the visualization of commonly linked kidney abnormalities. Despite some challenges to the purported high accuracy of MRI, it is frequently employed as the definitive method for assessing suspected Müllerian duct anomalies. This is particularly true in cases where clinical evaluation is complicated, and there is little clinical suspicion, especially prior to surgery. The doctor utilizes the findings from the MRI test within the framework of a full clinical assessment scheme to arrive at a definitive clinical diagnosis [17]. The highlighted image, in this case, is an MRI scan, which can be used in addition to or in comparison with other imaging techniques, such as ultrasound or laparoscopy. Furthermore, ultrasound results were unable to give conclusive results of the anomalies.

A study conducted by Mueller et al demonstrated a high level of concordance between the identification of uterine anomalies using MRI and clinical diagnosis. Out of the 103 patients

who were examined for uterine anomalies, the MRI diagnosis was inconclusive in 5 cases. Similarly, out of the 12 patients who were evaluated for vaginal anomalies, the MRI diagnosis was questionable in just 1 case [17]. This study highlights the possibility of MRI leading to a misdiagnosis, as demonstrated in this case report.

The primary reasons for the discrepancy between the clinical and radiologic diagnoses were the absence of a unified clinical-radiologic classification system that utilizes identical criteria for diagnosis, as well as the absence of distinct imaging or clinical diagnostic criteria. Errors in radiologic diagnosis occurred due to readers' lack of experience with uncommon anomalies, variable visualization of anatomical features, and the presence of fibroids causing distortion in the uterus. Additional misinterpretations arose from inadequate representation of some structures on MRI scans, resulting from either substandard signal or resolution of the images or insufficient visualization due to diminutive size (eg, atretic uterus) or physiological conditions (eg, collapsed vagina) [17].

The range of Müllerian duct abnormalities exists as a continuous spectrum rather than as separate and distinct entities. Additionally, many complicated anomalies may exhibit characteristics that belong to more than 1 class. There are no definitive clinical or MRI criteria for exact categorization. As a result, different radiologists and physicians sometimes classify these anomalies in different ways. Previous studies have shown a significant association between MRI findings and surgical outcomes in persons diagnosed with MRKH syndrome.

Both cases of cervical agenesis or dysgenesis were wrongly assessed, possibly because the readers lacked knowledge of this rare condition. Due to its infrequency, even experienced MRI radiologists may require assistance in characterizing this aberration. Precise identification of the cervix and endocervical canal is crucial for surgical planning, as the absence of a cervix requires a hysterectomy in young patients. It is not possible to manufacture an artificial cervix [17].

On MRI, it can be observed that a transverse vaginal septum results in a sudden termination of the vaginal canal, which may be accompanied by hydrocolpos or hematocolpos if there is a blockage in the outflow. Identifying an average vagina below the septum may not always be feasible, but it aids in distinguishing it from vaginal atresia [18]. A further study conducted by Kala et al highlighted that, in situations involving a transverse vaginal septum, the challenges in perceiving and understanding the surgical anatomy on MRI can be reduced by inserting a soft vaginal mold into the underdeveloped lower vaginal segment [19]. In our case, because of the history of hymenectomy, the distortion of anatomical structures may confuse the diagnosis of transverse vaginal septum and it was unclear whether vaginal or cervical agenesis was the source of the hematocolpos.

In comparison with our case, a similar case in a 20-year-old woman published by Petrykowski et al found that misdiagnosis may also occur from MRI images. They found that their original assessment suggested a potential imperforate hymen; nevertheless, it was later found that the patient likely had vaginal agenesis. The MRI images may be disrupted by the hematocolpos. As healthcare providers, we must be aware of vaginal agenesis and how to detect it. Vaginal agenesis can appear in various different ways, and because of this, it is often misdiagnosed or undetected. We suggest that clinicians should include vaginal agenesis in their differential diagnosis for individuals presenting with cyclic stomach discomfort and amenorrhea [13].

Advanced radiological imaging techniques, such as radiopaque coils and aqueous gel, can enhance the understanding and analysis of the surgical anatomy and various abnormalities of the vagina [20]. Visualizing the vaginal septum might be challenging in situations of partial agenesis of the vagina or transverse vaginal septum when using transvaginal ultrasonography and contrast MRI scan, as the walls of the vagina may appear collapsed. Therefore, in the investigation of the transverse vaginal septum by Kala et al, a pliable and appropriately sized vaginal mold was inserted into the patient's vagina without applying any force, and the radiologist was asked to perform an MRI scan of the pelvis [19]. Transrectal and transperineal ultrasonography have been proposed as substitutes for transvaginal ultrasound and vaginoscopy in diagnostics [21].

The patient in this case refused transvaginal ultrasound due to pain; therefore, only transabdominal ultrasound was done. Further examination with MRI was then conducted. A case report conducted by Huguelet et al also stated that MRI may be necessary in a patient who refuses gynecological examination. The patient with cyclical pain due to hematocolpos may present with significant pain and this may potentially hinder the diagnosis [22]. Therefore, comprehensive imaging evaluation is necessary.

From a histopathological perspective, the fibrous-tissue septum exhibited a significant presence of blood vessels, tiny clusters of smooth muscle, and nerve tissue. The transverse vaginal septum was flanked by stratified squamous epithelium on both sides, while the other side had no epithelium, which may confirm the diagnosis [23]. However, histopathological examination was not performed because the final diagnosis was distal vaginal agenesis.

Misdiagnosis, in this case report, could have been improved by inserting the vaginal mold. As reported by Kala et al, the utilization of a pelvic MRI with a vaginal mold facilitated the detection of the curved walls of the vaginal septum. It improved the distinction of the distance between the lower end of the cervix and the transverse vaginal septum [19]. Interpreting the distance between the lower end of the cervix and the high transverse vaginal septum was challenging on the contrast MRI scan in the case of a high transverse vaginal septum. This mold effectively separated the walls of the collapsed transverse vaginal septum, facilitating a more accurate assessment of its level. This facilitated the strategic development of reconstructive surgery in a more effective manner [19].

A pliable vaginal mold was utilized intraoperatively to position the skin graft in McIndoe's vaginoplasty, followed by postoperative dilatation. In the aforementioned study, the authors utilized it as a tool in diagnostic radiography to enhance the understanding of the surgical anatomy of the transverse vaginal septum, hence facilitating the planning of reconstructive surgery. It is an inexpensive and tidy substitute for coils and gel [19].

As per the American College of Obstetricians and Gynecologists Committee Opinion No. 728, after undergoing surgical creation of a vagina, regular dilation or sexual intercourse is necessary to ensure the vagina maintains its desired length and width. Therefore, this surgical procedure cannot be used as an alternative to vaginal dilator therapy. Given the high success rate of primary vaginal dilatation, surgery should only be considered for the few patients who are unsuccessful with primary dilator therapy or who choose surgery after a comprehensive discussion about the risks and benefits [24].

Performing postoperative dilation is crucial to avoid substantial neovaginal stenosis and contracture. Consequently, these methods should not be advised if the patient expresses opposition to dilation. The use of dilators should be continued periodically until the patient begins to engage in sexual intercourse on a regular and frequent basis [24].

Conclusions

Vaginal agenesis is commonly diagnosed with the aid of MRI as its criterion standard. However, there are certain pitfalls in MRI interpretation in vaginal agenesis, which include the lack of a comprehensive clinical-radiologic classification system, inadequate visualization of certain structures on magnetic resonance images, the reader's unfamiliarity with uncommon anomalies, and inconsistent portrayal of anatomical structures. Developing techniques to enhance the specificity of imaging modalities is crucial for distinguishing vaginal agenesis from a transverse vaginal septum, ensuring appropriate surgical management and better patient outcomes.

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Informed Consent Statement

The patient has received a comprehensive explanation regarding the case's particulars and the images to be included in the case report. The patients' parents have provided consent in the case report.

Declaration of Figures' Authenticity

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