Distal Vaginal Agenesis Presenting with Fecal Retention from an Abdominopelvic Mass

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Abstract

Distal vaginal agenesis (DVA) is a rare form of female genital tract malformation that presents as cryptomenorrhea. It results from the failure of the urogenital sinus to form the caudal portion of the vagina. Through a thorough history, physical examination and appropriate imaging studies, an accurate diagnosis is integral in selecting the correct intervention for the patient. This is a case of distal vaginal agenesis in a 10-year-old nulligravid, who presented with fecal retention from an abdominopelvic mass. The patient had no bowel movement for four days, and abdominal enlargement. On inspection, there was a 12.0cm x 10.0cm palpable abdominal mass. Inspection of the external genitalia, the introitus appeared concave, with no appreciable introital opening. On digital rectal examination, an anterior bulge was palpated 0.5 cm from the anal verge. A pull-through vaginoplasty was performed with an unremarkable post-operative course. The patient was discharged with a patent vagina and resolution of her gastrointestinal symptoms. On follow-up, the patient had monthly menstruation after surgery with no recurrence of her gastrointestinal symptoms.

Key words: vaginal agenesis, congenital abnormalities, vaginoplasty

Introduction

Distal vaginal agenesis (DVA) is a congenital anomaly of the female genital tract resulting from failure of development of the caudal vagina derived from the urogenital sinus, while the uterus, cervix, and proximal vagina usually remain intact. Vaginal agenesis is one of the rare forms of female genital tract anomalies, with an estimated incidence of 1:5000 for those with an isolated vaginal agenesis.[1,2] Incidence in the Philippines is unknown due to the paucity of studies reporting the mean incidence of this disease entity.[3] Because DVA represents an obstructive anomaly of the genital outflow tract, delayed recognition may lead to complications including hematocolpos, hematometra, hematosalpinx, endometriosis, and impairment of reproductive function.

Though uncommon, it is important for clinicians to be aware of its presentation, as well as differentiating this from other abnormalities which may present similarly. This is due primarily to the differences in the surgical management of these cases. A good history and physical examination are integral to prevent its sequelae, along with imaging modalities such as ultrasonography or MRI.

Presented here is a case of distal vaginal agenesis in a 10-year-old patient with an atypical presentation and history, and highlight the key points in managing cases of distal vaginal agenesis. This case underscores the role of comprehensive clinical evaluation in establishing the diagnosis and guiding appropriate surgical management.

The Case

This is a case of a 10 year-old nulligravid, who presented in the emergency room with no bowel

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movement for four days and abdominal pain. She has no co-morbidities nor previous surgeries. The family medical history is unremarkable. The patient is a Grade 5 Elementary School student, with no vices, no sexual history and no history of contraceptive use. She has not had menarche, however she has had cyclic hypogastric pain starting 4 months prior to her admission.

Eight months prior to admission, the patient presented with sudden onset of vomiting of previously ingested food for 5 days, with no note of abdominal pain, fever, or abdominal distention. She consulted with a private pediatrician wherein she was diagnosed with gastroesophageal reflux disease (GERD) and prescribed with Omeprazole for two weeks, providing resolution of symptoms. Seven months prior to admission, the patient noted recurrence of vomiting of previously ingested food, still lasting for 5 to 7 days. She consulted at a local hospital where she was admitted for 3 days and treated for a diagnosis of GERD with urinary tract infection. She completed 2 weeks of antibiotics and Omeprazole which provided relief of symptoms. During the interim, she noted monthly episode of hypogastric pain, associated with vomiting, and again treated with Omeprazole, providing relief of symptoms. One month prior to admission, there was note of persistence of vomiting and hypogastric pain, now with constipation, hence consult at a tertiary government hospital wherein she was prescribed again with Omeprazole for 2 weeks. Eight days prior to admission, patient consulted at the Pediatrics Outpatient Department, now presenting with an abdominal mass. A whole abdominal ultrasound was requested which showed the following result: dilated sigmoid colon and intestinal segment filled with fecal material and fluid, with wall thickening suggestive of infectious/inflammatory process. An abdominal X-ray revealed fecal stasis.

On the day of admission, patient presented with hypogastric pain with a pain score of 10/10 with inability to walk, persistent vomiting and no bowel movement for four days. At the ER, the patient was initially managed as a case of partial gut obstruction probably secondary to fecal impaction, rule out gynecologic pathology. She was placed on nothing per orem, and a nasogastric tube was inserted. She was then referred to the OB-GYN General Service for co-management.

On physical examination by the OB-GYN General Service, the patient had stable vital signs, with a BMI of 17.2, and a Tanner stage of 3 for both breast and pubic hair. On abdominal examination, she had a 12cm x 10cm palpable mass on the hypogastric region extending up to the umbilicus, with tenderness on palpation. On pelvic examination, the external genitalia had a concave tissue covering the introitus, with no bluish hue and no bulge on Valsalva maneuver (Figure 1). On digital rectal examination, the patient had good sphincteric tone, intact rectal vault, with a palpable bulge 0.5 cm beyond the anal sphincter. There was no septum appreciated on digital rectal examination, and there was no stool or blood per examining finger.

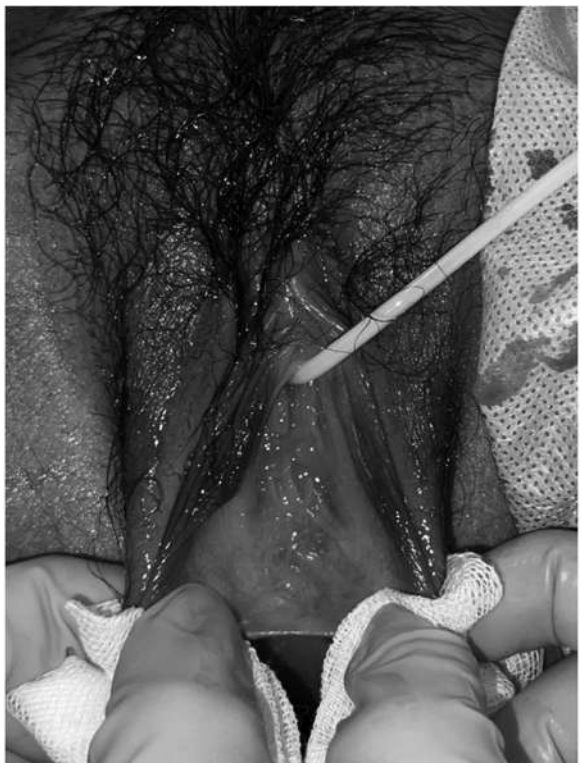

Figure 1. On pelvic examination, the external genitalia had a concave tissue covering the introitus, with no bluish hue and no bulge on Valsalva maneuver.

A transrectal ultrasound showed that the vagina is markedly dilated by a low-level echo fluid measuring 10.8cm x 7.4cm x 7.6cm (volume: 317.1cc). The inferior pole of the vagina ends as a membrane covering the introitus with bulging of the echo fluid. The cervix is open and effaced from which the fluid in the vaginal canal is seen to egress.

The uterus is pushed superiorly into the abdominal cavity with smooth contour and homogeneous echopattern. The uterine corpus measured 8.5cm x 6.9cm x 4.9cm. The endometrium is uniform, hyperechogenic measuring 0.1 cm anteriorly and 0.1 cm posteriorly. The endometrial cavity is dilated by low level echo fluid measuring 7.4cm x 5.3cm x 3.9cm (volume 79.0cc). The right ovary measured 2.6cm x 2.1cm x 2.6cm (volume 4.6cc). Inferior to the ovary is a tubular structure which measured 8.2cm x 4.9cm x 2.6cm (volume 55.0cc). The left ovary measured 1.4cm x 2.2cm x 1.1cm (volume 1.8cc). Inferolateral to the ovary is a tubulocystic structure which measured 5.6cm x 4.4cm x 4.0cm (volume 60.6cc) with low level echo fluid and incomplete septations. The bilateral renal pelves and calyces were not dilated. There was no free fluid in the cul de sac. Sonographic impression during this time was imperforate hymen with hematotrachelocolpos, thin endometrium, normal ovaries and bilateral adnexal masses, consider hematosalpinges (Figure 2). Abdominal X-ray revealed fecal retention (Figure 3).

Referral was made to the Reproductive Endocrinology and Infertility Service and was assessed as a case of Distal vaginal agenesis and normal uterus/cervix (ASRM 2021), ESHRE/ESGE U0 C0 V4. She underwent pull-through vaginoplasty under general anesthesia. A needle was inserted in the center of the vaginal dimple (Figure 4A). The syringe was withdrawn to confirm the presence of hematocolpos (Figure 4B). While overriding the needle, a surgical blade was used to puncture the atrophic distal vagina and distended proximal portion of the lower vagina, then extended laterally (Figure 4C). On incision, 400 cc of mucoid, viscous blood was evacuated from the opening. Mucosal edges were then grasped with Allis clamps and pulled to the perineum (Figure 4D). The proximal vagina was trimmed and widened to accommodate 2 fingers (Figure 4E). Interrupted sutures using Vicryl 2/0 were used to appose the proximal vagina to the introitus at the 3, 6, 9 and 12 o'clock positions, and between the 12 and 3, 3 and 6, 6 and 9, 9 and 12 o'clock positions (Figure 4F). Internal examination revealed the proximal vagina to be smooth (Figure 5), with the cervix 1 cm dilated. Corpus descended to the pelvis and returned to its normal size post-operatively. The patient tolerated the procedure

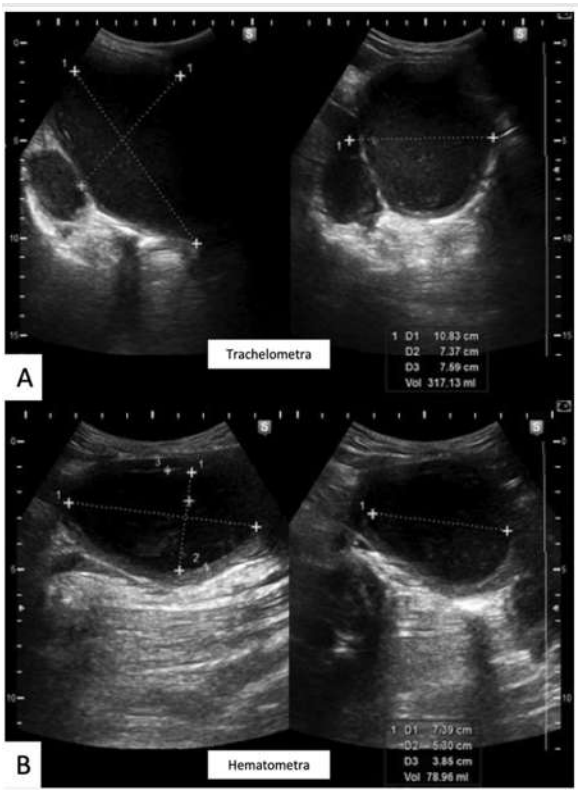

Figure 2. Preoperative ultrasound showed imperforate hymen with hematotrachelocolpos, thin endometrium, normal ovaries and bilateral adnexal masses.

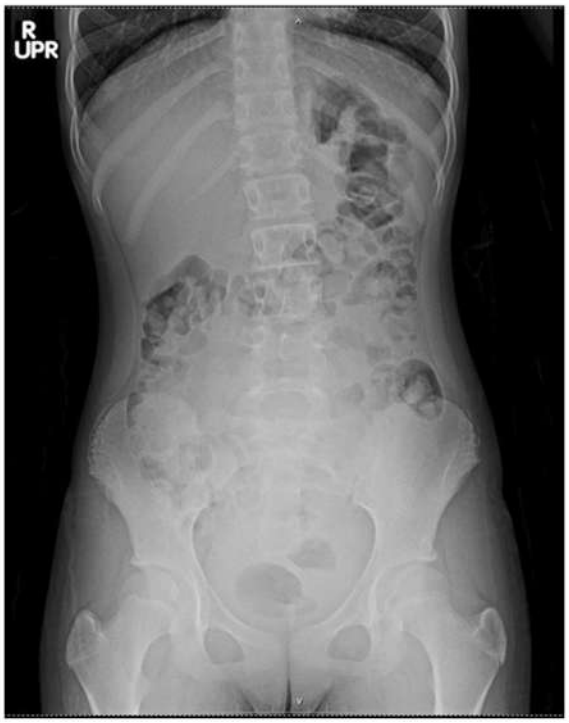

Figure 3. Abdominal X-ray hematosalpinges revealed fecal retention.

well, and eventually had bowel movement on the first post-operative day. The final diagnosis was:

Distal vaginal agenesis and normal uterus/cervix (ASRM 2021), ESHRE/ESGE U0 C0 V4
Status post pull-through vaginoplasty under general anesthesia
Fecal retention secondary to pain and pressure effects from the abdominopelvic mass (hematometrocolpos), resolved
Bilateral hematosalpinges
Nulligravid

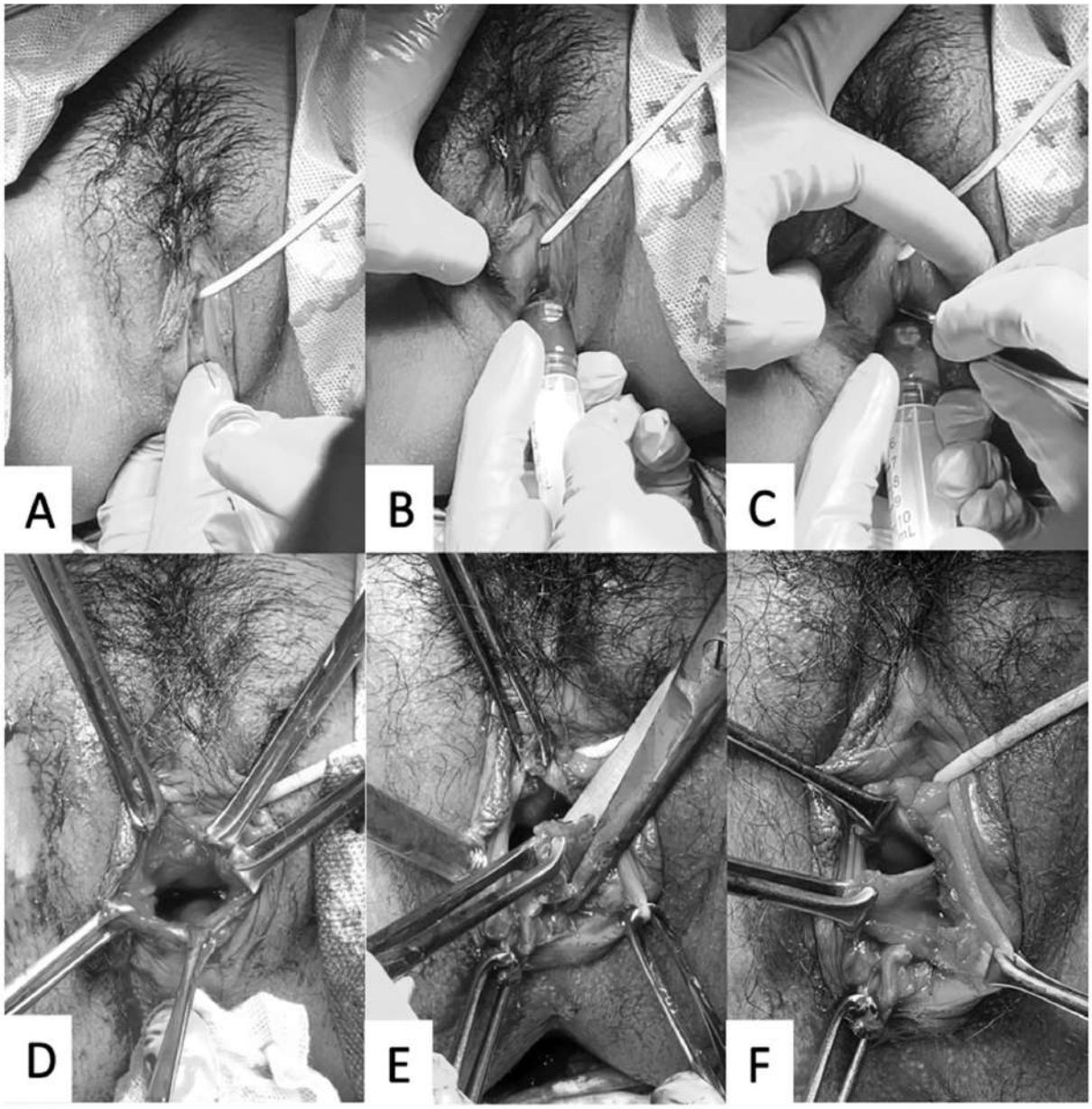

Figure 4. Pull-through vaginoplasty. A needle was inserted in the center of the vaginal dimple (Figure 4A). The syringe was withdrawn to confirm the presence of hematocolpos (Figure 4B). While overriding the needle, a surgical blade was used to puncture the atrophic distal vagina and distended proximal portion of the lower vagina, then extended laterally (Figure 4C). On incision, 400 cc of mucoid, viscous blood was evacuated from the opening. Mucosal edges were then grasped with Allis clamps and pulled to the perineum (Figure 4D). The proximal vagina was trimmed and widened to accommodate 2 fingers (Figure 4E). Interrupted sutures using Vicryl 2/0 were used to appose the proximal vagina to the introitus at the 3, 6, 9 and 12 o'clock positions, and between the 12 and 3, 3 and 6, 6 and 9, 9 and 12 o'clock positions (Figure 4F).

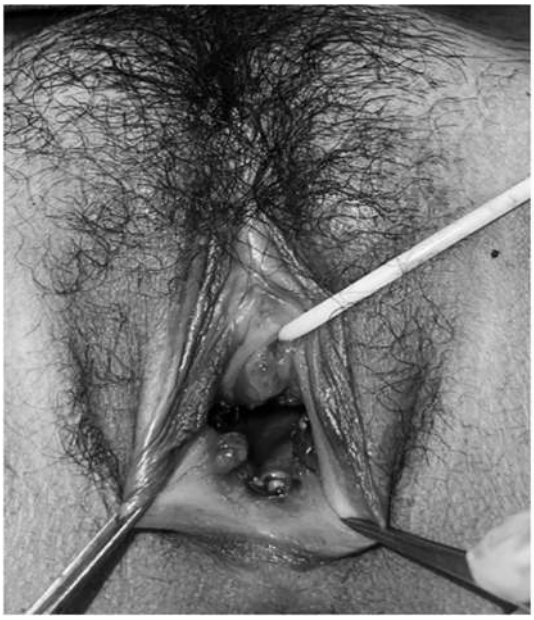

Figure 5. Post-operative examination revealing smooth proximal vagina with intact sutures

The patient was sent home well and followed-up closely in the Outpatient Clinic, with note of patent vagina with a diameter of around 1.5cm on each visit, with no note of abnormal discharge and intact suture sites. The patient was also advised to insert a makeshift vaginal dilator using a 3cc syringe and sterile gloves. She eventually had menses one month post-operatively, and since then, reported having regular bowel movement and resolution of her gastrointestinal symptoms. On the 5th postoperative month, pelvic examination revealed normal external genitalia, patent vagina, no visible sutures, with good healing of suture sites (Figure 6). A fingertip could be inserted in the vagina with ease and no pain. The vaginal canal was smooth, cervix measured 2cm x 2cm, corpus was small, and there was no adnexal mass nor tenderness. Repeat transrectal/ transabdominal ultrasound revealed regression of the previously seen bilateral hematosalpinges. Inferior to the right ovary was a tubuloystic structure measuring 2.1cm x 2.4cm x 1.1cm (volume: 4 cc) with low level echofluid, and no solid areas or papillary excrescences seen (Figure 7). This right adnexal mass previously measured 8.2cm x 4.9cm x 2.6 cm (volume: 55.0 cc). The previously seen left adnexal mass measuring 5.6cm x 4.4cm x 4.0cm was no longer present. The patient was advised to continue using the vaginal dilator twice a week, monitor signs of development of endometriosis, and was started on continuous combined oral contraceptive pills for menstrual suppression.

Figure 6. 5th post-operative month: Pelvic examination revealed normal external genitalia, patent vagina, no visible sutures, with good healing of suture sites. A fingertip could be inserted in the vagina with ease and no pain.

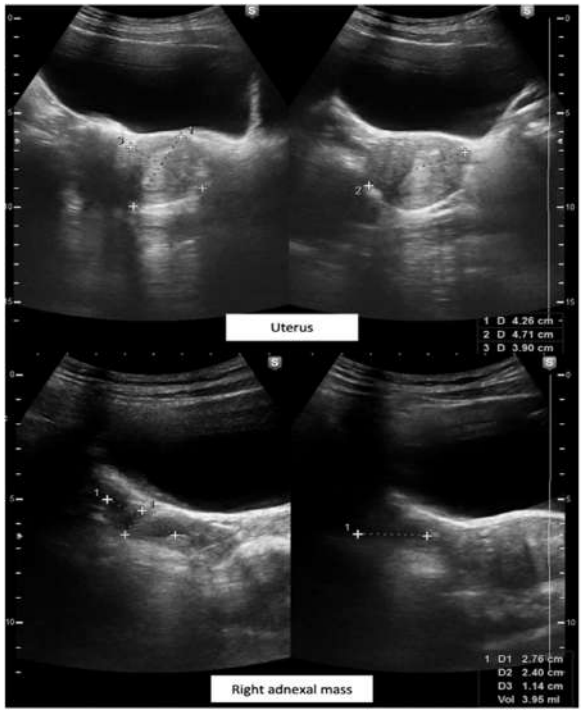

Figure 7. Ultrasound on the 5th post-operative month: Notable regression of the previously seen bilateral hematosalpinges. Inferior to the right ovary was a tubuloystic structure measuring 2.1 cx 2.4cm x 1.1cm (volume: 4 cc) with low level echofluid, and no solid areas or papillary excrescences seen.

Discussion

Formation of the uterus and the vagina entails key processes such as differentiation, migration, fusion, and canalization; disruption of which may lead to a variety of abnormalities.[1] Vaginal development results from the coordinated formation and canalization of two embryologic structures: the fused Müllerian ducts, which form the upper portion of the vagina, and the urogenital sinus, from which the distal vagina originates.[1] Failure of the urogenital sinus to form the caudal portion of the vagina leads to vaginal agenesis, and fibrous tissue replaces the vaginal opening during embryologic development, with the uterus cervix and upper vagina developing normally.[2,4] Patients with distal vaginal agenesis will typically have normal growth and development, as seen in the index case.[3] In the latest American Society for Reproductive Medicine Müllerian Anomalies Classification released in 2021, distal vaginal agenesis was classified under "Transverse Vaginal Septum", with a preferred term as "Distal Vaginal Agenesis and Normal Uterus/Cervix", and classified as U0C0V4 using the ESHRE/ESGE Classification of Female Genital Tract Anomalies.

Patients with distal vaginal agenesis commonly present with primary amenorrhea.[2] In a case study of 39 patients, 71.8% (28/39) presented with primary amenorrhea, followed by 41% (16/39) with cyclic pelvic pain, and abdominal pain 36% (14/39).[5] Patients may also present with an abdominal mass, dyspareunia, periodic fever.[5] In the case presented, the index patient presented atypically with no bowel movement and abdominal pain in the Emergency Department, although on history has had cyclic hypogastric pain beginning 4 months prior to her admission, and gastrointestinal symptoms in the months prior. The hematotrachelocolpos was severe enough to impair normal bowel function of the patient resulting to fecal retention. Intestinal obstruction and constipation from mechanical compression are rare presentations of DVA, but a possible complication of accumulation of blood in the uterus and vagina resulting to hematocolpos, hematometra or hematosalpinx.[6] This highlights the importance of a complete physical examination in arriving at the correct diagnosis, as other abnormalities may present similarly. In the case presented, it is possible that cryptomenorrhea was missed by previous doctors since mean age of menarche is at 12.9 years, with the proportion of those having their menarche at less than 12 years old at 13%.[11] However, in patients presenting with cyclic pelvic pain, it would be prudent to rule out a possible gynecologic problem. It is important to be able to diagnose obstructive female genital tract anomalies because of potential reproductive consequences such as damage to the fallopian tubes, as with the index case who manifested with bilateral hematosalpinges.

It is important to distinguish DVA from other vaginal abnormalities that may present with primary amenorrhea, cyclic pelvic pain and an abdominal mass, such as an imperforate hymen and a transverse vaginal septum. Meticulous examination of the introitus is essential for diagnosing distal vaginal agenesis, which is characteristically marked by a concave perineal surface with a small central dimple and absence of a visible vaginal opening. In contrast, an imperforate hymen usually manifests as a bulging membrane with a characteristic bluish discoloration due to hematocolpos, whereas patients with a transverse vaginal septum generally have a variable length of distal vagina below the level of the obstruction.[2] Although transrectal and transabdominal ultrasonography initially favored a diagnosis of imperforate hymen, the pelvic examination findings were not compatible with this diagnosis, as the characteristic bulging, bluish hymenal membrane was absent. This discrepancy underscored the importance of correlating imaging findings with a meticulous physical examination. It is imperative that an accurate diagnosis is determined prior to surgical management, to avoid any unnecessary procedures that may produce complications such as loss of a surgical plane for eventual reconstructive surgery because of scar tissue formation.[2]

Although distal vaginal agenesis is a rare congenital anomaly, clinicians caring for pediatric patients should be familiar with its clinical presentation and be able to distinguish it from other causes of genital outflow tract obstruction. Imaging modalities such as ultrasonography and magnetic resonance imaging (MRI) play an important role in establishing the diagnosis. MRI is considered the gold standard because it provides superior resolution, is not operator-dependent, and is less affected by

interference from bowel gas or overlying bowel loops.[7] Despite its diagnostic advantages, the use of MRI may be limited by its cost and availability. In the present case, MRI was not performed because of financial constraints, necessitating reliance on clinical findings and ultrasonography for diagnosis and surgical planning. Nevertheless, combining the ultrasonographic images, history, and physical examination of the index patient, an accurate diagnosis was met and appropriate surgical intervention was done.

Various techniques have been described in literature regarding the approach to the surgical intervention of cases of DVA. Selecting the appropriate technique highly depends on understanding the anatomy of each individual case.[8] Historically, cases were treated with a hysterectomy. However, techniques have been developed to preserve the fertility of the patient and allow for normal sexual and reproductive function, in addition to relieving the obstruction.[1] Surgical correction is generally recommended at the onset of menarche, when hematocolpos has resulted in adequate vaginal distention but before the development of substantial hematometra. This approach facilitates surgical dissection, allows for the creation of a tension-free anastomosis using well-developed vaginal tissue, and minimizes the risk of postoperative vaginal stenosis.[2,10] Several surgical techniques have been described for the management of distal vaginal agenesis, although no consensus exists regarding the optimal approach. These include neovagina creation using skin grafts, labial or introital skin flaps, use of vaginal dilators, and local pull-through vaginoplasty.[2,8] The choice of procedure should be individualized and guided by the patient's anatomy and the extent of the vaginal defect. In the index case, a local pull-through vaginoplasty was performed because the length of the agenetic segment was 2 cm or less, making it amenable to this reconstructive approach.

Vaginal stricture or stenosis remains the most common postoperative complication following surgical correction of distal vaginal agenesis. Therefore, patient and caregiver counseling regarding postoperative vaginal dilation is essential to maintain vaginal patency and minimize the risk of restenosis. In the index case, a makeshift vaginal dilator fashioned from a 3-mL syringe covered with a sterile glove was successfully utilized as part of the postoperative management plan.[2,9]

Long-term surveillance is crucial following surgical correction of DVA to evaluate vaginal patency, menstrual function, sexual health, and future fertility potential.[5] Given the risk of retrograde menstruation and endometriosis associated with obstructive genital tract anomalies, patients should be routinely screened for symptoms suggestive of endometriosis, including chronic or cyclic pelvic pain, dysmenorrhea, dyschezia, dysuria, and dyspareunia. Clinical follow-up should include pelvic, vaginal, or rectal examinations when appropriate, together with imaging modalities such as ultrasonography or MRI to assess for recurrent obstruction, residual hematosalpinx, or endometriosis.[12]

Conclusion

Distal vaginal agenesis is a rare congenital anomaly of the female genital tract that should be considered in the differential diagnosis of patients presenting with genital outflow tract obstruction. This case highlights the importance of obtaining a thorough history, performing a meticulous physical examination, and utilizing appropriate diagnostic investigations in patients presenting with atypical manifestations such as fecal retention, abdominal pain, and a palpable hypogastric mass. A concave introitus with a vaginal dimple on physical examination is a key diagnostic finding suggestive of distal vaginal agenesis and may be further evaluated using ultrasonography or magnetic resonance imaging.

Accurate characterization of the anomaly is essential for selecting the most appropriate surgical approach, as management depends largely on the individual patient's anatomy. Surgical options include neovagina creation using skin grafts, labial or introital flaps, progressive vaginal dilation, and local pull-through vaginoplasty. In the present case, the patient successfully underwent pull-through vaginoplasty, resulting in restoration of menstrual flow, resolution of gastrointestinal symptoms, and maintenance of vaginal patency during follow-up. Early recognition and timely surgical intervention are crucial to prevent complications such as hematosalpinx, endometriosis, and future

reproductive sequelae, while optimizing long-term gynecologic and reproductive outcomes.

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