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CASE REPORT

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Mixed Müllerian and gonadal dysgenesis: A case report

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Abstract

Background: Globally, the incidence of cervical agenesis is estimated at 0.01% and constitutes 3% of all Müllerian anomalies.

Case presentation: A 17-year-old nulliparous presented to the gynecology clinic with a transverse vaginal septum and cyclic lower abdominal pain after two failed vaginal surgical procedures to restore genital tract patency. The examination under anesthesia revealed a 3cm short vagina, and a decision was made for vaginoplasty. Magnetic resonance imaging of the pelvis revealed a bulky uterus. Intraoperatively, locating the cervix through the neo-vagina was difficult, and laparotomy was opted for. The left adnexa and cervix were absent, with an atrophic right ovary and a hydrosalpinx on the fallopian tube. Creating a uterovaginal anastomosis was difficult as the uterus was high in the pelvis. Postoperatively, the patient was counseled, and hysterectomy and right oophorectomy were done four months later.

Conclusion: Vaginoplasty with total hysterectomy is an alternative management method in failed uterovaginal anastomosis.

Keywords: cervical agenesis, hysterectomy, uterovaginal anastomosis, vaginoplasty

Introduction

Cervical agenesis or dysgenesis is a rare congenital anomaly with an estimated prevalence of 1 in 80,000-100,000 women (1,2). The fallopian tubes, the uterus, cervix, and the upper vagina develop from the Müllerian ducts, and anomalies occur due to complete agenesis, abnormal lateral or vertical fusion, or failure of resorption (3). Cervical atresia is an extremely rare abnormality that may be associated with complete or partial vaginal agenesis. According to the American Society of Reproductive Medicine (ASRM) guidelines, cervical dysgenesis is classified as type 1B. Cervical dysgenesis can further be classified as i) cervical fragmentation, ii) fibrous cervical cord, and iii) cervical obstruction (4). Conservative management approaches include cervical canalization, cervical reconstruction, and uterovaginal anastomosis. In failed conservative surgery, total hysterectomy is recommended (5). Uterovaginal anastomosis is the current trend, with the attainment of fertility and livebirths reported in some cases. This is instead of the previous recommendation for hysterectomy for cervical agenesis (5). This is a case of cervical dysgenesis in a 17-year-old nulliparous who successfully underwent total abdominal hysterectomy and right oophorectomy after three failed surgical procedures to restore genital tract patency.

Case presentation

A 17-year-old nulliparous presented to the gynecology clinic at the Kenyatta National Hospital

(KNH) with primary amenorrhea as a referral from a peripheral facility, where two surgical attempts at hymenectomy were made, and a thick septum was encountered. She had a history of cyclic lower abdominal pain without distension. On examination, she was generally fair, not pale or jaundiced. Her secondary sexual characteristics, breasts, and pubic hair were Tanner stage iii. Axillary hair was present with a fine texture and sparse distribution. Pubic hair had a female distribution pattern. The abdomen was nondistended with mild tenderness in the suprapubic region, and no masses were palpable. A pelvic examination revealed normal external genitalia and a short blindly ending vagina measuring 3cm with a fibrous septum. Her complete blood count and renal function tests were within normal ranges.

Magnetic resonance imaging (MRI) of the pelvis revealed a bulky uterus with free endometrial fluid and cervical stenosis. The left ovary was visualized; however, the right adnexa was not visualized. The patient was scheduled for vaginoplasty. Intraoperatively, jungle juice was infiltrated into the vaginal septum. Sharp dissection was perfomed to access the peritoneal cavity, but the cervix could not be located. A decision on an abdominal approach was made. Upon laparotomy, the left ovary and fallopian tube were absent. The right ovary was atrophic (thinned out and flat), and the right fallopian tube was blind-ending with hydrosalpinx (**Figures 1**). The cervix was absent (**Figure 2**). A longitudinal incision was made on the anterior body of the uterus and drained 30mls of the bloody tarry fluid. Drilling to create an outlet inferiorly was done. Attempts at uterovaginal anastomosis were unsuccessful as the uterus was high in the pelvis. Vaginoplasty was performed with the posterior vaginal wall reinforced with a rotational flap from the left groin.

Postoperatively, the patient received antibiotics and analgesics. While in the ward, counseling on the intraoperative findings and procedure (success in creating a vagina, but not at uterovaginal anastomosis) was done. The impact on future fertility was discussed with the patient, and she opted to discuss with her family the two options available; the need for hysterectomy due to the severity of the Müllerian dysgenesis and a second attempt at the uterovaginal anastomosis to be performed later. Four months later, the patient came in as part of a follow-up, ready for total abdominal hysterectomy and right oophorectomy. On laparotomy, peritoneal adhesions with bowel adherence to the uterus and bladder were noted, all of which were released, and a uterine corpus with no cervix and right edematous fallopian tube with a hydatid cyst of Morgagni were found. Hysterectomy and oophorectomy were performed. Her postoperative period was unremarkable. She was discharged home for follow-up. The patient was counseled later on the need for hormone replacement therapy. The histology report showed the endometrium in the normal secretory phase, with endometrial tissues deep in the myometrium. The ovarian section revealed stroma with cysts lined by follicular cells. The patient is currently on long-term follow-up.

Discussion

Mixed cervical and gonadal agenesis are very rare. Pubertal girls with cyclic pelvic pain and amenorrhea should be assessed for genital tract patency. While imperforate hymen and blindly ending vaginal pouch are easily detected clinically, cervical atresia is not. As in this case, the unilateralabsence of the fallopian tube and ovary remains largely unexplained and unclear, with suspicion of torsion or congenital malformations the likely causes. It may be associated with uterine

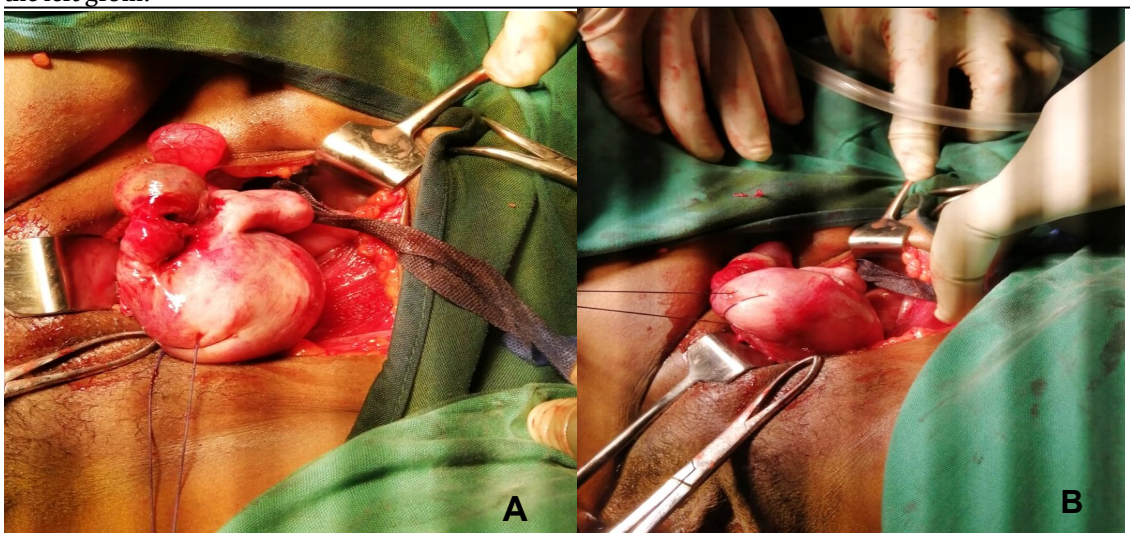

Figure 1: A: Intraoperative findings demonstrating the right-sided hydrosalpinx with atrophic right ovary and bulky uterus; **B:** An absent left fallopian tube and ovary.

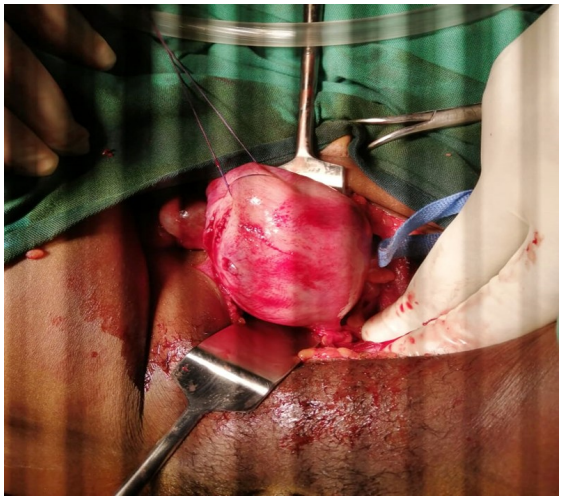

Figure 2: Intraoperative findings demonstrating bulky uterus and absent cervix.

or urinary tract abnormalities, including unilateral renal agenesis (6). This case can be assigned to several categories of Müllerian anomalies according to the latest ASRM guidelines of 2021. The current case could be placed in classes 1 and 2 of Müllerian agenesis and cervical agenesis and their variants or into more classes, given the varied nature of the anomaly.

Adnexal dysgenesis diagnosed in this case is very rare and estimated to affect 1 in 11,240 and is often associated with Müllerian tract anomalies is difficult to estimate exact numbers as most cases are asymptomatic and may be incidental findings during pelvic surgery (7). A thorough preoperative workup of the patients is necessary. The high chance of anomalies in the urinary tract calls for targeted imaging modalities, like renal ultrasound and intravenous urography (8). Unlike the traditional hysterectomy approach, uterovaginal anastomosis is quickly gaining acceptance as the first line of treatment in cervical agenesis (8). However, cervical canalization poses a risk of secondary stenosis in up to 40%-60% of patients (8). Laparoscopic procedures have also been reported, but they require a lot of experience and skill (8). The current case was a referral from a peripheral facility after two failed surgical attempts at hymenectomy. This further highlights the necessity of integrating clinical–radiological classification schemes and thorough knowledge of rare mixed Müllerian duct anomalies.

In the current case, creating a vagina was successful, but uterovaginal anastomosis attempts were unsuccessful as the uterus was high in the pelvis, limiting any future spontaneous pregnancy. However, in the absence of such challenges, attempts can be made for later successful pregnancies. Successful pregnancies have been reported following surgical correction of cervical agenesis (9). With the advent of assisted reproductive technology (ART), in vitro fertilization (IVF) and transmyometrial embryo transfer (TMET) in patients with unsuccessful surgical correction have been done successfully (10). This, therefore, provides hope that fertility is still possible. In cases of unsuccessful uterovaginal anastomosis, subsequent pregnancy would require hysterotomy to evacuate the uterus in the event of miscarriage. Other options that may be explored may include local or systemic methotrexate injection. Successful pregnancies are delivered through cesarean birth (2).

This case consists of three rare Müllerian anomalies: cervical agenesis, vaginal dysgenesis, and adnexal agenesis. Although the medical literature has underscored the possibility of mixed Müllerian anomalies, the previously published studies did not describe similar cases. A combination of all three anomalies is extremely rare, hence the motivation to report this case. Magnetic resonance imaging and three-dimensional (3D) ultrasound are excellent modalities for the characterization and classification of such anomaly types.

Conclusion

Mixed adnexal and cervical agenesis, and vaginal dysgenesis anomalies are rare. The goal of treatment is to restore reproductive performance, including creating sexual function via vaginoplasty. The second goal is fertility restoration through the uterovaginal anastomosis, cervical canalization, or cervical reconstruction. In cases of failed uterovaginal anastomosis, hysterectomy is the alternative method for managing cyclic pain, as was the case for this patient.

Consent for publication

Informed consent for publication was obtained from the patient.

Acknowledgement

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Declarations

Conflict of interests

The authors declare no conflicts of interest.

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