Case Report

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Endometrioma Excision in a Patient with VACTERL Syndrome and a Rudimentary Uterine Horn: a Case Report

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ABSTRACT

Vertebral anomalies, anal atresia, cardiac malformations, tracheoesophageal fistula, renal anomalies, and limb defects (VACTERL) syndrome is a rare congenital condition characterized by multiple organ systems, including the vertebrae, anus, cardiac system, trachea, esophagus, renal system, and limbs. Mullerian duct anomalies are infrequently observed in VACTERL syndrome, with only a few cases reported in the literature. We present a rare case of a 20-year-old female with VACTERL syndrome, presenting with a right adnexal mass diagnosed as an endometrioma. The mass measured 83x64x38 mm and exhibited typical features of endometrioma, including a hypoechoic 'ground glass' appearance. MRI also revealed a right non-communicating rudimentary uterine horn. A laparotomy was performed for the excision of the endometrioma and the removal of the rudimentary uterine horn. The patient had an uneventful postoperative recovery, with discharge on postoperative day three. Follow-up visits showed satisfactory healing and resolution of symptoms. This case highlights the challenges in managing gynecological conditions in individuals with VACTERL syndrome and underscores the need for tailored surgical approaches to address coexisting congenital anomalies.

Keywords: VACTERL syndrome, endometrioma, mullerian duct anomalies, rudimentary uterine horn, surgical management

INTRODUCTION

Vertebral anomalies, anal atresia, cardiac malformations, tracheoesophageal fistula, renal anomalies, and limb defects (VACTERL) association is a rare, non-random constellation of congenital malformations that affects multiple organ systems, including the vertebrae, anus, cardiac structures, trachea, esophagus, kidneys, and limbs.[1,2] While Mullerian duct anomalies are infrequent in patients with VACTERL, the rudimentary uterine horn is a rare form of such anomaly. Often associated with other congenital malformations, it can present significant surgical challenges, especially when complications like endometriosis or endometriomas.[3-5]

Managing endometriomas in patients with VACTERL association is challenging, especially when Müllerian defects like a rudimentary horn are present.[6] Altered reproductive anatomy can complicate surgical access and treatment. The rare coexistence of a rudimentary horn and endometriomas in VACTERL patients requires careful surgical planning.[7]

This report highlights the surgical management of an endometrioma in a patient with VACTERL association and a rudimentary uterine horn. It emphasizes the challenges of diagnosis and treatment, the importance of a multidisciplinary approach and the need for awareness of Mullerian anomalies in patients with VACTERL syndrome, as their presence can significantly impact the clinical course and management of associated gynecological conditions.

CASE REPORT

A 20-year-old female with a known history of vertebral malformations, tracheoesophageal fistula repair in infancy, and renal hypoplasia was referred to our clinic with a complaint of progressively worsening right lower abdominal pain, dysmenorrhea and irregular bleeding over the last few months. Upon review of her medical history, it was found that she had undergone multiple surgical interventions, including tracheoesophageal fistula repair during infancy, anal transposition, and rectal dilation procedures in later years. Additionally, she had been treated for vesicoureteral reflux with cystoscopy and subureteral injections. Other noted anomalies included butterfly vertebrae, left renal hypoplasia, right aortic

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arch, and aberrant left subclavian artery. Based on these findings, the diagnosis of VACTERL association was strongly supported.

On initial examination, the patient was alert, oriented, and afebrile, with stable vital signs. Abdominal examination revealed tenderness in the right lower quadrant without signs of peritoneal irritation. No palpable masses were felt, and the abdomen was soft and non-distended. The pelvic examination did not reveal any abnormalities, though the patient did report mild discomfort upon deep palpation of the right adnexal region. Further evaluation through transabdominal ultrasonography revealed a well-defined, cystic mass measuring approximately 8 cm in diameter located in the right adnexa. The mass demonstrated typical features of an endometrioma, including a homogenous, hypoechoic appearance, with a "ground glass" pattern, a hallmark of endometriotic tissue. No internal vascularity was detected on Doppler imaging, which is consistent with a benign cystic lesion. Additionally, the surrounding ovarian tissue appeared intact, without signs of torsion or rupture. These ultrasonographic findings were suggestive of an endometrioma, which was later confirmed by magnetic resonance imaging (MRI).

The MRI confirmed the presence of an endometrioma and additionally identified a rudimentary uterine horn on the right side. This rudimentary horn with an endometrial cavity and was not connected to the cervix. In contrast, the left uterine horn was normally developed and was found to be connected to the cervix, with a well-formed endometrial cavity, consistent with a unicornuate uterus. The anatomical findings supported a diagnosis of a unicornuate uterus with a non-communicating rudimentary horn, an anomaly rarely seen in VACTERL association.

Given the complexity of the patient's case, consultations with nephrology and anesthesiology were sought to evaluate the associated risks and plan for optimal perioperative care. The nephrology consultation focused on assessing the patient's renal function, as she had a history of renal hypoplasia, and to ensure that her kidney function was adequate for surgical management. The anesthesiology consultation was essential due to her history of tracheoesophageal fistula, which required a thorough assessment of her airway and anesthetic risks. The team was particularly concerned about potential challenges with intubation, given her anatomical anomalies.

Routine preoperative laboratory tests, including hemoglobin, renal function, and coagulation profiles, were within normal limits. The patient received intravenous cefazolin (1 g) prophylactically before surgery to minimize the risk of infection.

The laparotomy was selected due to the patient's altered anatomy and history of previous surgeries, which increased the complexity of laparoscopic access. Under general anesthesia, a Pfannenstiel incision was made. Upon entry into the peritoneal cavity, extensive pelvic adhesions were noted, particularly between the bowel and adnexa. Sharp and blunt adhesiolysis was performed to improve visualization. The right adnexal mass was identified and confirmed as an endometrioma. The cyst was carefully dissected from ovarian tissue, and complete excision was performed using meticulous hemostasis to preserve ovarian function. The right non-communicating rudimentary uterine horn was then mobilized and excised at its base. The excised horn was non-communicating with the endometrial cavity and had a fibrous attachment to the main uterus.

Hemostasis was ensured, and the peritoneal cavity was irrigated with warm saline. A drain was placed in the right adnexal region due to the extent of adhesiolysis. The abdominal wall was closed in layers, and the patient was extubated without complications (Figure 1).

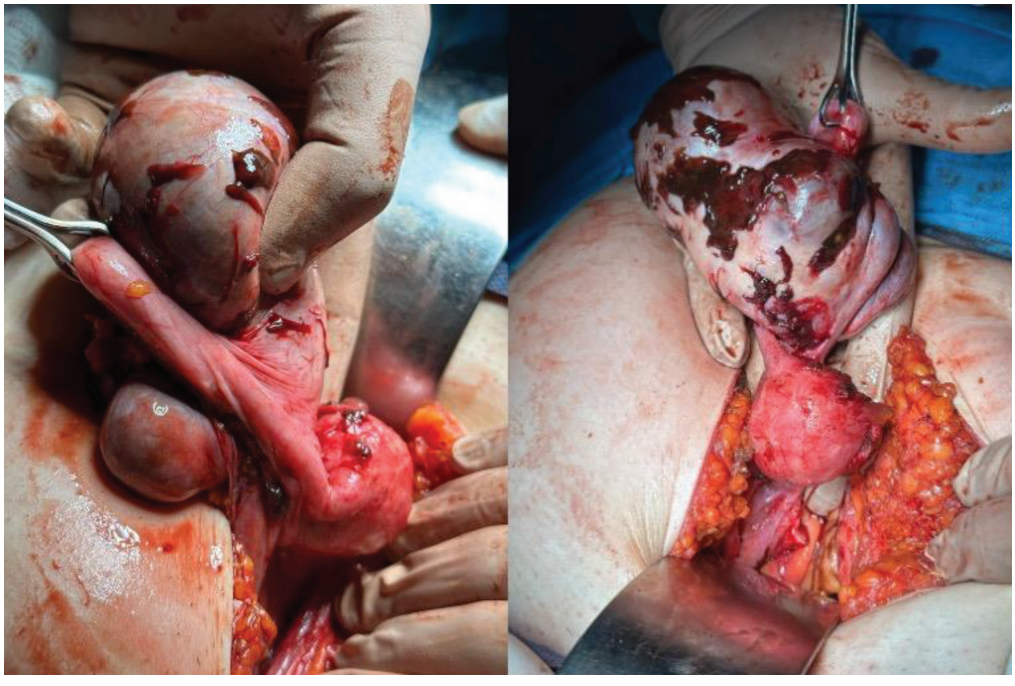

Figure 1. Endometrioma and right rudimentary horn

The patient was monitored in the postoperative unit for 24 hours, with stable vital signs and adequate urine output. Pain control was managed with IV paracetamol and tramadol as needed. Deep vein thrombosis prophylaxis was provided with low molecular weight heparin (enoxaparin 40 mg subcutaneously daily) until discharge. The surgical drain was removed on postoperative day two, with minimal serous output. The patient tolerated oral intake on postoperative day one, ambulated early, and had an uneventful recovery. She was discharged on postoperative day three with instructions for wound care, pain management, and scheduled follow-up. Histopathological examination confirmed an endometriotic cyst and fibromuscular tissue consistent with a rudimentary uterine horn.

The patient was treated with dienogest after surgery to prevent recurrence of endometrioma, reduce inflammation, and manage residual pain. At the one-month follow-up, the patient reported resolution of pain, and at the three-month follow-up, she reported regular menstrual cycles. No complications or recurrent symptoms were noted, indicating a favorable response to treatment. Informed consent was obtained for this case report at the post-discharge outpatient clinic controls.

DISCUSSION

The aim of this case report was to highlight the challenges in diagnosing and managing a patient with VACTERL association who presented with an endometrioma in the presence of a rudimentary uterine horn, a rare Müllerian anomaly. This case expands on the existing literature by presenting a unique combination of congenital anomalies that complicate both diagnosis and surgical management. The objective was to demonstrate the multidisciplinary approach required to address the complexities of this case and to explore how the rare coexistence of these abnormalities in a patient with VACTERL syndrome presents specific clinical challenges.

Several studies have shown that patients with VACTERL association often have reproductive system abnormalities, including uterine malformations like unicornuate uterus or, less frequently, rudimentary horns.[7-14] For example, a 17-year-old girl with VACTERL presented with severe dysmenorrhea and was found to have a unicornuate uterus and a non-communicating rudimentary left horn.[9] After pelvic MRI and surgery, her symptoms resolved. Delayed diagnosis contributed to prolonged symptoms, highlighting the importance of timely intervention. Similarly, a 14-year-old with left renal agenesis and anorectal malformation presented with cyclical abdominal pain, which was later diagnosed as a right unicornuate uterus with a rudimentary left horn and hematosalpinx. Surgical intervention resolved her symptoms.[12] Lavoie et al.[11] reported a 10-year-old with recurrent abdominal pain, eventually diagnosed with uterine didelphys, hematometrocolpos, and hematosalpinx, which was also successfully treated with surgery. These cases emphasize the importance of early imaging and intervention in managing reproductive tract anomalies in VACTERL patients.

However, the combination of VACTERL association, a rudimentary horn, and an endometrioma is an extremely rare clinical scenario that has not been well documented.[7] Bhadwal et al.[7] presented two cases of uterine rudimentary horn and ovarian endometriosis in patients with VACTERL association. One involved a 12-year-old girl with a unicornuate uterus and an obstructed right uterine horn, leading to hematometra, hemosalpinx, and an endometrioma in the right ovary. This case emphasizes the importance of considering Müllerian anomalies in adolescents with gynecological symptoms and VACTERL, as they can lead to complications like hematometra and endometriosis. The second case involved a 14-year-old girl with abdominal pain initially diagnosed as a right ovarian cyst, which was later reclassified as a rudimentary horn with hematometra. This case highlights the need for thorough radiological evaluation, as Müllerian anomalies can be easily overlooked on standard imaging. Both cases underscore the rarity of VACTERL-associated Müllerian duct anomalies and emphasize the need for a high index of suspicion. Management typically involves surgical excision of the obstructed uterine horn and removal of endometriotic lesions to prevent complications like infertility and chronic pain.[7]

A distinguishing feature of our case was the need for a multidisciplinary approach. Given the complexity of the patient's anatomy, consultations with nephrology and anesthesiology were essential for preoperative evaluation. Nephrology assessed the patient's renal function due to the presence of renal hypoplasia, while anesthesiology evaluated potential airway complications arising from her previous tracheoesophageal fistula repair. These consultations were crucial for ensuring the patient's safety during surgery, highlighting the importance of comprehensive preoperative planning in complex cases such as this.

The management of patients with VACTERL syndrome requires a tailored, multidisciplinary approach due to the complexity of the associated anomalies. A combination of gynecological, surgical, and radiological expertise is often necessary to address the challenges presented by these cases. Surgical intervention, as demonstrated in this case, can offer significant symptomatic relief and improve the quality of life for affected individuals.

CONCLUSION

This case presents a unique and rare combination of VACTERL association, a rudimentary uterine horn, and an endometrioma, highlighting the complexities in diagnosis and management when multiple congenital anomalies coexist. The presence of a rudimentary uterine horn in a patient with VACTERL association is infrequently documented, and its coexistence with an endometrioma further complicates both the clinical picture and surgical approach. This report contributes to the limited body of literature on such complex cases and underscores the importance of a multidisciplinary approach in the management of these patients.

Ethics

Informed Consent: It was obtained.

Authorship Contributions

Surgical and Medical Practices: A.A., C.K., Concept: B.K., Design: C.K., Data Collection or Processing: A.A., Analysis or

Interpretation: B.K., C.K., Literature Search: B.K., Writing: A.A.,

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