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Case Report
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Surgical Management of an Obstructive Müllerian Anomaly in a Patient with VACTERL Association: A Case Report

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Abstract

Background: The incidence of VACTERL association alone has been estimated at around 1 in 10,000 to 1 in 40,000 live born infants. Upon review of existing literature, few if any other cases of VACTERL association in combination with vaginal atresia, uterine anomalies, and cervical agenesis in a patient of pubescent age have been discussed despite a likely association between VACTERL and vaginal anomalies.

Case Report: A 10-year-old female with a history of VACTERL association initially presented for management of nephrolithiasis and pyelonephritis. Following successful treatment, she re-presented six months later with recurrent cyclical abdominal pain. Workup demonstrated proximal vaginal atresia with right hematometrocolpos, hematosalpinx, and uterine didelphys. A complex robotic assisted vaginoplasty was successfully performed. Two years later, the patient re-presented due to recurrent cyclical monthly pelvic pain, nausea, and vomiting and was found to have a left hematometra, hematosalpinx, and ovarian cysts due to cervical agenesis of the left hemi-uterus. Patient underwent left laparoscopic hysterectomy, drainage of the hematosalpinx, and left ovarian cystectomy which led to symptom resolution.

Conclusion: This is a rare case of VACTERL association which presented in combination with distal vaginal atresia, right hematometrocolpos, hematosalpinx, and uterine didelphys, later complicated by left hematometra, hematosalpinx, and ovarian cysts secondary to agenesis of the left hemi-uterus demonstrates an exceedingly complex case of interdisciplinary medical and surgical management. The array of symptoms which were present in a single patient and the numerous surgical interventions which were utilized to improve the patient's quality of life and symptomatology represent a unique case of medical presentation and management.

Keywords: Mullerian Anomaly; Uterine didelphys; Vaginal atresia; VACTERL

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Introduction

The incidence of VACTERL association alone has been estimated at around 1 in 10,000 to 1 in 40,000 live born infants [1]. Upon review of existing literature, few if any other cases of VACTERL association in combination with vaginal atresia, uterine anomalies, and cervical agenesis in a patient of pubescent age have been discussed despite a likely association between VACTERL and vaginal anomalies [2]. We present the case of a 10-year-old female with a history of VACTERL association and Posterior Sagittal Anorectoplasty (PSARP) in infancy, spinal and upper extremity anomalies, and recurrent UTIs presenting for evaluation of multiple episodes of nephrolithiasis and pyelonephritis and subsequent lower abdominal pain.

Case Presentation

Patient information

A 10-year-old female with a history of VACTERL association initially presented to our pediatric urology clinic for evaluation and management of nephrolithiasis and pyelonephritis. In addition to VACTERL association, her medical history was significant for recurrent Urinary Tract Infections

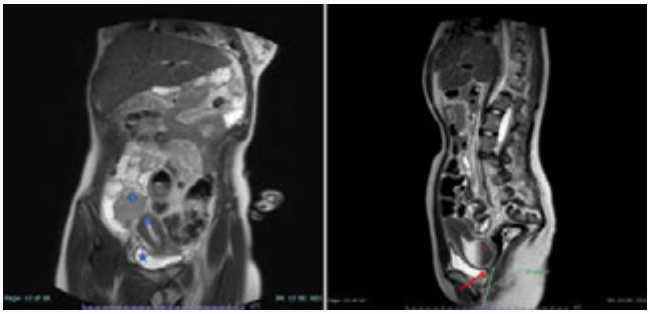

Figure 1: MRI images of initial presentation Left Image: T2 Coronal, showing bladder (*), dilated right hemiuterus (Δ) and hematosalpinx (+) Right Image: T2-weighted Sagittal, demonstrating dilated proximal vagina (*). Vaginoplasty dissection length of approximately 5 cm to bring the proximal vagina to orthotopic location. Arrow demonstrates atretic vaginal canal.

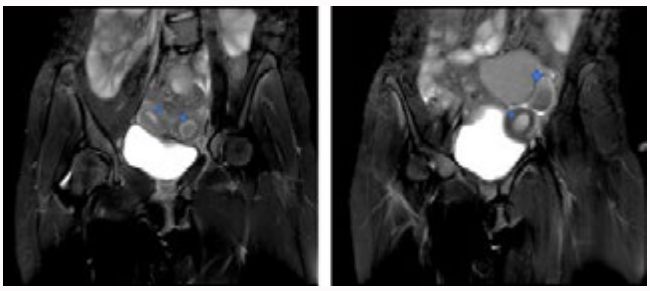

Figure 2: MRI Images of second presentation Left Image: Coronal T2 image showing decompressed right uterine horn (Δ) adjacent to dilated left uterine horn (+) consistent with obstruction Right Image: Coronal T2 image showing dilated left uterine horn (+) consistent with obstruction and left ovary with follicles of different sizes (◊).

(UTIs), imperforate anus status post PSARP in infancy, and spinal and upper extremity anomalies. She underwent an uneventful ureteroscopy to treat her nephrolithiasis. However, six months later she was readmitted for pyelonephritis. After culture-directed treatment, she developed lower abdominal pain which was attributed to a mildly inflamed appendix, and she underwent a laparoscopic appendectomy at an outside hospital. According to the outside hospital operative report, she was found to have hemorrhagic fluid in the posterior cul-de-sac from an unknown etiology. One year later, she was admitted for a febrile UTI and severe cyclical abdominal pain.

Clinical findings

Due to her history, a Magnetic Resonance Imaging (MRI) (Figure 1, left panel) was performed which suggested a Mullerian duct anomaly with a large right uterine horn and rudimentary, left uterine horn; there was evidence of right sided hematometrocolpos, hematosalpinx, and cystic right ovary. On physical exam, the patient was noted to have tenderness to deep palpation in the right lower abdomen, Tanner stage V and unremarkable external genitalia with an intact hymen without obvious hematocolpos.

Therapeutic intervention

Given the clinical scenario, the decision was made to perform a cystoscopy and Exam Under Anesthesia (EUA). EUA demonstrated a normal appearing introitus but proximal vaginal atresia. The cystoscope was advanced into a shallow fibrinous appearing vaginal canal but could only be advanced 5 mm. Cystoscopy demonstrated a smooth bladder with bilateral ureteral orifices laterally inserting. The patient was subsequently taken to the operating room for a complex

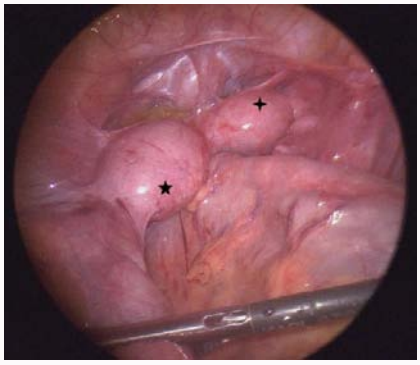

Figure 3: Intraoperative image demonstrating suspected bicornuate uterus with dilated left horn (*) and right horn (+).

robotic assisted reconstruction. Intraoperatively, a single cervical os with a presumed bicornuate uterus was noted. The blind ending proximal vagina was mobilized, and a plane was created between the rectum and bladder deep in the pelvis. A pull-through vaginoplasty was performed to bring the proximal vagina to its orthotopic location in the perineum. Dissection was difficult as the distance from the perineum to the dilated vagina was approximately 5 cm (Figure 1, right panel). A 12 French pigtail catheter was left as a vaginal drain. The patient recovered and was discharged home.

Follow-up and outcome

Postoperative vaginoscopy and vaginogram demonstrated well-healed vaginal mobilization and vaginoplasty. She was then advised to begin self vaginal dilation which was successful. Additionally, she began having regular menses and resolution of cyclical abdominal pain. Approximately two years later, the patient re-presented with recurrence of severe cyclical abdominal pain, nausea, and vomiting. MRI (Figure 2) confirmed the suspected diagnosis of an obstructed and non-communicating left uterine horn with new hematometra and left hematosalpinx with left ovarian cysts, suspicious for uterine didelphys *vs.* bicornuate uterus with non-communicating left horn. Vaginoscopy revealed well healed vaginoplasty with cervical opening only to the right uterine horn. There was no cervical opening to the left uterine horn. She subsequently underwent diagnostic laparoscopy, left laparoscopic hysterectomy, drainage of hematosalpinx, and left ovarian cystectomy (Figure 3). She remains asymptomatic and has recovered fully from these interventions.

Discussion

This case is a complex presentation of VACTERL association with Mullerian duct anomalies. VACTERL association occurs very rarely, with an incidence of approximately 1/10,000 to 1/40,000 infants [1,3]. The condition is characterized by the presence of at least three congenital malformations including vertebral defects, anal atresia, cardiac defects, tracheoesophageal fistula, renal anomalies, and limb deformities [1,3]. The association manifests vertebral and limb anomalies in 60 to 80% of cases, imperforate anus/anal atresia in 55% to 90%, genitourinary anomalies in up to 25% of cases, and renal anomalies in roughly 50% to 80% of patients [4].

During development, the mullerian ducts differentiate and develop into the uterus, fallopian tubes, cervix, and proximal one-third of the vagina [5]. The distal two thirds of the vagina develop from the urogenital sinus, a different embryological precursor [5]. Malformations in the urogenital sinus can also lead to imperforate

hymen and cloacal anomalies when combined with anorectal malformations, which can present together in VACTERL association [6]. A derivative of the same mesoderm is the mesonephros, which gives rise to the renal structures, mullerian ducts and their anomalies can also be associated with renal anomalies, as seen in this case and VACTERL association [5]. The co-occurrence of these congenital anomalies appears to relate closely to the development of a number of the defining anomalies associated with VACTERL association and may indicate further exploration of their connection [2].

Medical management of patients with VACTERL association remains complex and highly individualized to each case. Generally, management is split into two stages, the first stage being immediate intervention for conditions deemed incompatible with life without surgical intervention, such as imperforate anus, and the second being longer term management of additional malformations that may impact functioning and quality of life such as vaginal atresia [1]. Multiple complex surgical interventions may be necessary for both short- and long-term quality of life; some subtle manifestations may become debilitating later in life, such as in this case with the development of nephrolithiasis and pyelonephritis secondary to bilateral vesicoureteral reflux.

There are some similar cases that have been reported previously in the literature, such as that of a 17-year-old girl with VACTERL association who presented with a three-year history of severe dysmenorrhea. She was found to have a unicornuate right uterus and rudimentary non-communicating cavitary left horn with longstanding symptoms, complications, and delayed intervention [7]. Following pelvic MRI and resection of the left communicating horn, left salpingectomy, and left ovarian cystectomy, the patient resumed regular menses and her pain completely resolved; however, the delayed evaluation of her pelvic anomalies likely contributed to the three-year history of her symptoms prior to diagnosis and surgical intervention [7].

Conversely, in our case, the patient presented after a shorter interval of dysmenorrhea and was evaluated with imaging and laparoscopy earlier, potentially preventing further complications such as infection or hemorrhage and providing resolution of discomfort in a more-timely fashion. Ultimately, the surgical intervention was nearly identical. This indicates that early imaging screenings may be necessary in patients with known VACTERL association in anticipation of additional reproductive tract anomalies.

Another similar case describes a 14-year-old girl with a history of left renal agenesis and anorectal malformation status post-PSARP in infancy. She presented with a three-year history of cyclical left-sided abdominal pain which coincided with the start of menarche. Based on MRI and ultrasound studies, she was found to have uterine didelphys with hematocolpos and following laparoscopy was definitively diagnosed with right unicornuate uterus with a dilated, rudimentary, non-communicating left uterine horn and left hematosalpinx. Treatment involved laparoscopic excision of the rudimentary non-communicating left uterine horn and dilated left fallopian tube with sparing of both ovaries. This intervention resulted in resolution of abdominal pain and return of regular menses [8].

The progression of the above case from symptom onset, to evaluation with imaging, and eventual definitive diagnosis and intervention with laparoscopy is consistent with our own reported case and does appear to be the most expedient and effective means of addressing potential vaginal and uterine anomalies in patients with existing VACTERL association. Our case similarly recommends anticipatory and early evaluation of Mullerian structures and the urinary system in patients with known history of vaginal and anorectal anomalies prior to onset of menses. Additionally, a laparoscopic approach for resection of atretic components with sparing of functional tissue where possible provides an effective and minimally invasive option for symptom resolution while preserving critical structures such as the ovaries and facilitating the return of menses. Thus, laparoscopic or robotic management of patients with similar presentations is a successful intervention and should be considered.

In this case, the patient underwent thorough imaging and scope-based interventions with ureteroscopy, cystoscopy, hysteroscopy, vaginoscopy, and MRI to properly diagnose and determine the etiology of her symptoms. Initial imaging and visualization of presenting anomalies is critical to proper management of malformations and proper intervention [9,10]. She then underwent vaginal mobilization and vaginoplasty for vaginal atresia which successfully addressed her hematometrocolpos and hematosalpinx, providing symptomatic relief and improving function. Additionally, the patient underwent further surgical intervention with a subsequent left laparoscopic hysterectomy, drainage of hematosalpinx, and left ovarian cystectomy for recurring abdominal pain and absent menses despite prior reconstruction, which ultimately led to symptom resolution.

As discussed, the adherence to surgical intervention in addressing and managing this patient's symptoms and anatomical findings continues to be the most widely used approach in management of VACTERL and associated anomalies [4,7,8]. The occurrence of additional complications in this patient which eventually necessitated a left hysterectomy also represents the necessity of adequate follow-up, symptom monitoring, and further surgical intervention if complications arise. The numerous surgical interventions which were employed across multiple organ systems make this case an example of successful combined medical management and surgical intervention for patient's presenting with VACTERL and associated complex congenital anomalies with severe symptoms. It also suggests that anticipation of and early screening for congenital anomalies involving the female reproductive tract in patients with VACTERL association may be beneficial.

Conclusion

VACTERL association is a rare condition that can be managed with appropriate multi-specialty care and surgical interventions. The association is often accompanied by other genitourinary anomalies such as vaginal agenesis and renal anomalies, which can manifest as several comorbidities that can ultimately impact patient quality of life and functionality. Some of the manifestations from these anomalies may not appear until later in life, more specifically during adolescence. Careful consideration of a patient's prior history, knowledge of the association and its embryologic basis, as well as appropriate imaging can aid in management. Patient follow-up should be maintained in these complex cases to appropriately address emerging symptoms as early as possible. Additionally, anticipatory imaging in the form of MRI and/or ultrasound prior to the onset of menses in patients with known VACTERL association may prove necessary to fully evaluate reproductive tract anomalies and prevent complications.

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