Case Report

Partial Cervical Agenesis and Complete Vaginal Atresia

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A B S T R A C T

Background: The objective of this study was to report 2 cases of the combined congenital anomalies of complete vaginal atresia and partial cervical agenesis, and highlight the limitations of magnetic resonance imaging for definitive initial diagnosis, and consequently the importance of early definitive management, to avoid life-threatening sepsis. Herein we provide a retrospective case audit of two patients with congenital abnormalities between 2005 and 2013 who were treated in a quaternary statewide pediatric and adolescent gynecology center.

Cases: Two patients with the combined congenital anomalies of complete vaginal agenesis and partial cervical agenesis highlight the difficulties encountered with the limitations of magnetic resonance imaging in accuracy of diagnosis, as well as development of life-threatening sepsis that requires hysterectomy. Both patients were initially imaged as having distended endometrial cavities and cervical canals with what was thought to be an obstructive upper vaginal septum and absent lower vagina. Both required initial neovagina creation, however the cervices were never clinically or surgically visualized.

Summary and Conclusion: Partial cervical agenesis is a relatively rare form of Müllerian abnormality which, if not diagnosed and definitively treated early, can have significant morbidity and mortality. Although magnetic resonance imaging is the diagnostic imaging gold standard for Müllerian abnormalities, it is important to recognize the limitations of this modality, the potential sequelae of these limitations, and to appreciate the importance of early accurate diagnosis and treatment of this condition. Importantly, if the imaging diagnosis does not completely correlate with the clinical and surgical findings, then a high suspicion of complete or partial cervical agenesis is prudent, because the consequences of nondefinitive early treatment can be life-threatening and potentially fatal.

Key Words: Cervical agenesis, Vaginal atresia, Müllerian abnormality

Introduction

The combination of partial cervical agenesis and complete vaginal atresia is a rare variation of obstructive Müllerian abnormalities in patients who could present with primary amenorrhea and cyclical pelvic pain. The challenges of accurate diagnosis of complete or partial cervical agenesis using magnetic resonance imaging (MRI), and subsequent complexity of balancing surgical and nonsurgical management, can lead to delay in definitive diagnosis and consequent significant morbidity and mortality.

We present two cases of complete vaginal atresia with partial cervical agenesis diagnosed using histology specimens after hysterectomy. Initial MRI diagnostic scans showed distended endometrial cavities and cervical canals with what was thought to be an obstructive upper vaginal septum and absent lower vagina. Clinical correlation of these imaging findings were consistently difficult, because the complete cervix could not be visualized. The incongruence of clinical, surgical, imaging, and histological diagnosis of cervical anatomy made it difficult to definitively treat the patient in the initial period. Counseling was limited by the inability to reach a complete and accurate diagnosis in the first instance and by the fact that both patients were teenagers for whom a hysterectomy on the basis of a nondefinitive diagnosis was unacceptable.

In this report we aim to highlight the difficulties associated with interpretation of diagnostic imaging for this combination of Müllerian abnormalities, and the importance of early definitive treatment in these cases. We also emphasize the management complexities in relation to counseling of patients and their parents to proceed to hysterectomy early, to prevent what can be life-threatening sepsis.

Cases

Case 1

A highly intellectual 14-year-old female adolescent presented with a history of cyclical abdominal pain and primary amenorrhea. She was initially examined and noted to have normal external genitalia. An initial ultrasound showed an obstruction at the cervical level with blood in the endometrial cavity.

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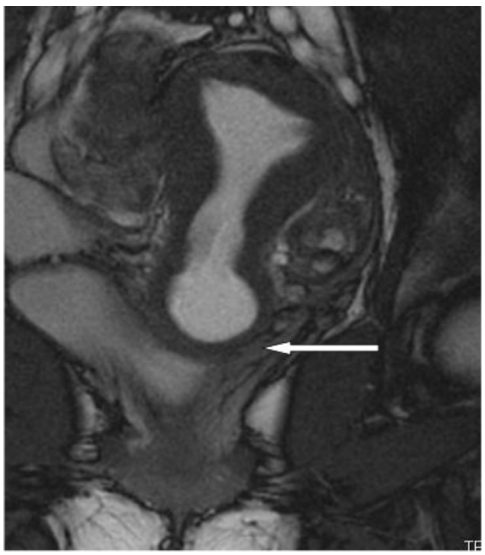

Figure 1. In patient 1, initial magnetic resonance imaging scan showed blood products distending and endometrial cavity and cervical canal. The collection was thought to be the result of a transverse septum in the upper vagina (arrow). The lower vaginal canal was clinically thought to be atretic.

An initial MRI scan showed a dilated, fluid-filled endometrial cavity and cervical canal and what was presumed to be an upper vaginal transverse septum (Figure 1).

She underwent an examination under anaesthesia, and concurrent laparoscopy and was found to have complete vaginal agenesis and a likely atretic cervix. It was also noted that she had extensive endometriosis, normal ovaries and fallopian tubes, and two normally located kidneys. A neovagina was created by dissection and was connected to the presumed very small upper vagina with laparoscopic guidance. It was unclear at this initial surgery whether a cervix was palpable at the conclusion of the surgery. A normal cervix could not be identified intraoperatively (Figure 2).

The patency of the neovagina was maintained with regular use of vaginal dilatation. There was no graft placed, which allowed neoepithelialization to progressively occur, and maintain the patency of the vagina. This patient started use of the combined oral contraceptive pill and experienced regular menses after this initial surgery. Her menses however, ceased within 6 months. She was found clinically to have had closure of the neovagina at the apex with no visible patency into the previously created passage between the uterus and the neovagina, at the site of the suspected cervix.

Over the next 6 years, she underwent 4 surgical attempts to maintain a patent connection between her cervix and neovagina. A Pap smear was performed during one of these procedures, which showed endocervical cells but no ectocervical component was identified. This provided some reassurance, albeit false, that a completely formed cervix existed. Despite numerous surgeries to maintain a patent tract, she experienced only the occasional menses. Three years after her initial surgery, this patient presented to the emergency department with a pelvic infection.

An MRI scan was performed at this stage, and showed a uterine cavity distended with menstrual fluid and a fluid-filled cervical canal measured at a normal length, with again what was thought to be a transverse vaginal septum (Figure 3).

At this stage, the potential need for a hysterectomy was discussed with her, however she was reluctant to undergo such radical treatment because of the lack of a definitive diagnosis.

She was treated with intravenous antibiotics and had complete resolution of her infection. After her admission, her menses were suppressed. She was followed-up regularly at 3- to 6-month intervals by the pediatric and adolescent gynecology service for surveillance of any further suggestion of sepsis. She continued to be counseled about the potential need for a hysterectomy because the

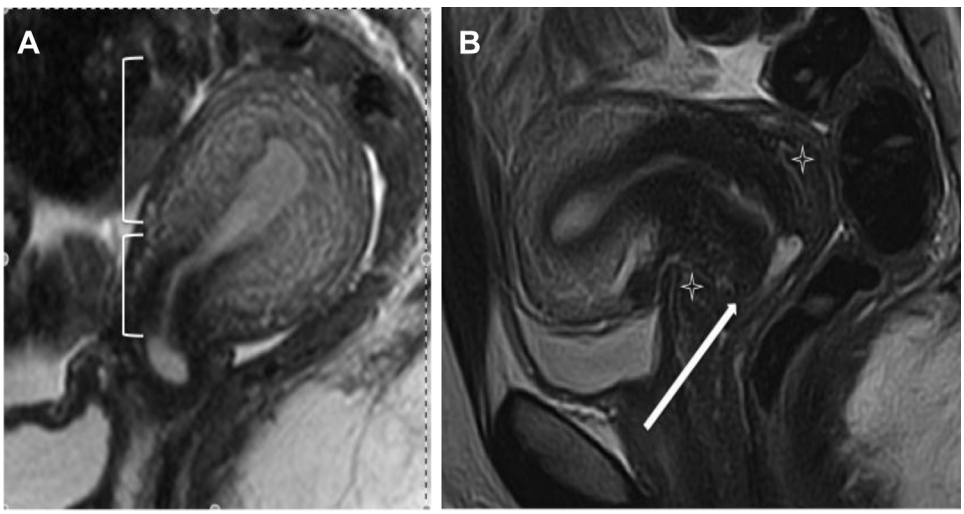

Figure 2. (A) After surgery, the cervical length was approximately one-third of the total length of the uterus and cervix and the zonal anatomy of the endocervical canal showed normal magnetic resonance imaging characteristics. The anterior and posterior vaginal fornices were not well visualized, which was presumed to be the result of the upper vaginal septum that caused distension and distortion of the anatomy in this region. (B) A normal cervix of a patient of the same age, as a comparison, shows well demarcated fornices (stars) and clear contours of the ectocervix (arrow). The length and zonal anatomy of the cervical canal are similar to that seen in patient 1.