ORIGINAL ARTICLE

An integrated imaging approach for diagnosis of cervico-vaginal outflow defects and associated genital anomalies

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Cervico-vaginal outflow; MRI; Ultrasound

Abstract  Objective: The objective of this study was to evaluate the diagnosis of cervico-vaginal outflow anomalies using different ultrasound approaches (transabdominal, endoluminal and transperineal) and magnetic resonance imaging.

Subjects and methods: Thirty female patients, their age ranged from 11 to 42 years (mean age 14.4, +/−6.9 s/d) with clinically suspected cervico-vaginal outflow defects presented with amenorrhea, dyspareunia or cyclic abdominal pain. They were subjected to ultrasound and MR imaging examinations. Imaging results were correlated with clinical examination and surgery.

Results: The study included 10/30 cases of aplasia/hypoplasia (33.3%), 4/30 cases of imperforate hymen (13.3%), 6/30 cases of transverse vaginal septum (20%), 4/30 cases of vaginal atresia (13.3%), 3/30 cases of combined cervical/vaginal atresia (10%), 1/30 case of cervical stenosis (3.3%), 1/30 case of cervical atresia (3.3%), and 1/30 case of vaginal atresia with fistula (urogenital sinus syndrome) (3.3%). Obstructed outflow was detected in 17 patients (56.6%). The accuracy of multi-approach US and MRI examinations in diagnosis of cervico-vaginal outflow anomalies was 94.1% and 97.1% respectively.

Conclusion: Ultrasound examination using different approaches remains the initial investigation for all patients with simple müllerian anomalies. MRI examination could be reserved for more complex anomalies, long cervicovaginal atresia and for cases of urogenital sinus syndrome.

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1. Introduction

The true incidence of müllerian anomalies, including obstructive subtypes, is believed to be between 0.1% and 3.8% (1). Multifactorial polygenic and familial factors are involved in their formation. The result may be deficient development, non-fusion, or defective canalization of the müllerian ducts (2).

The modified American Fertility Society (AFS) by Rock and Adam embraces a broader collection of uterine and vaginal anomalies. This classification correlates anatomic anomalies with embryologic arrests. Accordingly, uterovaginal anomalies are categorized as dysgenesis disorders or vertical or lateral fusion defects (3).

If isolated, these anomalies go unnoticed. However, many surface as a result of deviations in menses or sexual capacity. The diagnosis and treatment of urogenital anomalies is challenging and requires a comprehensive and thorough understanding of the condition, which may not be fully appreciated until surgical exploration is undertaken. Counseling and treatment of these conditions often requires a lifelong multimodal approach. Improved patient self-imaging, reproductive potential, and sexual satisfaction are the main goal of treatment (1).

Vaginal agenesis is characterized by an absence or hypoplasia of the uterus and proximal vagina. It occurs in an estimated 1 in 5000 newborn females (4). Vaginal atresia occurs when the urogenital sinus fails to contribute to the inferior portion of the vagina (5). The müllerian structures are usually normal, but fibrous tissue completely replaces the inferior segment of the vagina. Although not müllerian in origin, vaginal atresia can clinically mimic vaginal agenesis and imperforate hymen (6).

Transverse vaginal septum is one of the most rare müllerian duct anomalies, with approximate frequency of 1 case in 70,000 females (7,8). Transverse septa arise from an incompletely canalized vagina and urogenital sinus, and may be complete or incomplete. 46% occurs in the upper vagina, 40% in the midvagina, and 14% in the lower vagina (9).

Failure of the lower müllerian ducts to fuse can result in a longitudinal septum that either partial or complete (10). If a double uterus (uterine Didelphis) exists, one uterine hemicorpus tends to be developed and may be a source of infertility or recurrent abortion. If a partial or fully obstructing septum exists, progressive dysmenorrhea or a pelvic mass may be the presenting factors (1).

Although of different embryological origin, the imperforate hymen is commonly listed with defects of the vertical fusion of the müllerian ducts. Imperforate hymen is the most common obstructive anomaly and has an incidence of 0.1% (1).

Cervical aplasia and dysgenesis are vertical fusion defects that involve complete or partial absence of the cervix and upper vagina. The true incidence of this condition is difficult to determine, but the frequency is reportedly 1 in 4500 live births. Anatomically, cervical anomalies may be subtyped: (1) fibrotic cervical banding with endocervical glands, (2) cervical fragmentation, (3) cervical os obliteration, and (4) midcervical (11). Because of hematometas, patients with cervical dysgenesis are prone to endometriosis and adhesions. Laparoscopic treatment may be less invasive than abdominal surgery, but ultimately vaginal canalization with hysterectomy is required to treat this condition (12).

Ultrasound (US) is recommended as the first line imaging modality. Limitation of pelvic US includes operator dependence and patient obesity (10). Magnetic resonance (MR) imaging is a useful noninvasive tool for demonstrating pelvic anatomy and abnormalities, including anomalies of female genital system (13–15). With development of new software and improved hardware, MR imaging has proved to be helpful tool in the management of uterovaginal anomalies, particularly complex lesions (16).

The purpose of the study was to evaluate the diagnostic accuracy of ultrasound using different approaches (transabdominal, endoluminal and transperineal) compared to magnetic resonance imaging in patients with clinically suspected cervico-vaginal outflow anomalies and also to determine the cause, the level of canalization defect and associated genital anomalies in order to guide for proper surgical management.

2. Patients and methods

Thirty patients with clinically suspected cervico-vaginal outflow defects presenting with amenorrhea, dyspareunia or cyclic abdominal pain were referred to the Women’s Imaging Unit in Radiology department from the Obstetrics and Gynecology Department, Cairo University. Their age ranged from 11 to 42 years (mean 14.4).

Ultrasound and pelvic MRI examinations were performed for all patients.

2.1. Ultrasound examination

Ultrasound examination was performed through different approaches:

(1) Transabdominal scan using a 3.5–5 MHz sector transducer was performed for all patients after adequate bladder distention.

(2) Endoluminal; transvaginal (TVS) (in married individuals, n = 4) or transrectal (TRUS) approach (n = 8) was performed using 7–8 MHz endoluminal transducer.

(3) Transperineal approach using 7–8 MHz endoluminal transvaginal probe was performed for all patients.

2.2. Magnetic resonance imaging

Pelvic MRI was performed for all cases. All the patients were imaged in the supine position using pelvic phased-array coil. Cases were examined by T2-weighted pulse sequences FSE (TR/TE 1600/100 ms) in the axial, sagittal and coronal planes, and matrix 256 × 192; Axial T1-weighted sequence SE (TR/TE 500/40 ms); Slice thickness 4 mm with 1 mm gap. Instilling of endovaginal aqueous gel prior to imaging was performed in one patient for better delineation of vaginal anatomy.

2.3. Image analysis

Ultrasound through different pelvic approaches and MR pelvic images were reviewed to assess cervicovaginal outflow regarding genesis, canalization, obstruction, coexistence of other müllerian duct anomalies or associated pelvic lesions.
The pubertal uterus has adult pear configuration (fundus to cervix ratio = 2/1–3/1) and is 5–8 cm long, 3 cm wide and 1.5 cm thick (17). There are differences in vaginal dimensions among women. The length of the vagina from external OS to introitus ranged from approximately 4.1 to 9.5 cm (18).

2.3.1. Ultrasound image analysis

The cervix and vagina are seen in midline sagittal plane, and the cervix is barrel in shape with central endocervical canal. The vagina is seen as a collapsed, hypoechoic tubular structure with central high-amplitude linear echo between the urethra anteriorly and the rectum posteriorly. The cervix and vagina were best evaluated by endoluminal and transperineal scanning.

Transabdominal approach was effective for identifying uterine and vaginal collections (hematometrocolpos), most often seen as cystic mass with diffuse low level internal echoes. The uterus is differentiated from the cervix by thick myometrial wall and milder distention of the cavity of the uterus, as compared with the thin and often imperceptible wall of the vagina. Transabdominal scan is also used to check renal anomalies.

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2.3.2. MR imaging analysis

- The uterus with its zonal anatomy was best evaluated in T2 sagittal plane with the confirmation of normal cervical dimensions, shape and canalization. In case of obstruction, its level was determined with distended blood filled uterus (hematometra) and blood filled fallopian tubes (hematosalpinx) were noted. The presence of müllerian ducts anomalies was checked.
- The vagina is seen as a tube of intermediate signal intensity between the bladder base and urethra anteriorly and the anal canal posteriorly. It was evaluated in T2 sagittal and axial planes. Vaginal canalization was checked for. The site and extent of vaginal septum – if present – was assessed. The obstruction site of a blood filled vagina was estimated, and any fistulous tract between the uterovaginal canal and perineal level was seen in cases of imperforate hymen.
- Both ovaries should be identified and presence of cysts was reported.

The final diagnosis was based on findings at history, clinical examination, imaging studies and surgery.

3. Results

The study included 30 cases of cervico-vaginal anomalies listed in Table 1.

The diagnosis and accuracy of US and MRI in diagnosis of cervico-vaginal anomalies in our 30 cases are listed in Tables 2 and 3.

Obstructed outflow was seen in 17 patients (56.6%), 13 cases with hematometra and 4 cases with hematometrocolpos. Transabdominal scanning was effective for identifying uterine and vaginal fluid collections; however it did not allow adequate visualization of the entire length of vagina or proper evaluation of the cervix (Fig. 1). Transvaginal US was performed in four married cases, and it was effective in delineating combined vaginal/cervical atresia (n = 3) (Fig. 2) and cervical stenosis in normally canalized cervix (n = 1); however, in more complex anomalies, it failed in the detection of upper vaginal atresia with fistulous tract with the urethra (n = 1).

Transperineal approach was performed in all cases. Vaginal septum is seen as hypoechoic fibromuscular band in 6 cases (Fig. 3), and its thickness was measured, ranged from 4 to 5 mm. In cases with vaginal atresia, by using the least pressure in coronal plane, the length of atretic segment was measured, ranging from 2 to 2.5 cm. Distention of the whole vagina down to perineal level was seen in cases of imperforate hymen.

Transrectal approach was performed in 8 cases. It identified combined cervical/vaginal atresia (n = 1), vaginal atresia (n = 1), and vaginal septum (n = 2). Measurement of the length of atretic segment and septum thickness was performed. Such approach was effective in confirmation of müllerian agenesis in 4 cases.

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<tr>
<th>Table 1</th>
<th>Cases of cervico-vaginal anomalies included in the study.</th>
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<tbody>
<tr>
<td>Cervico-vaginal anomalies</td>
<td>Patient no.</td>
</tr>
<tr>
<td>Müllarian agenesis</td>
<td>9</td>
</tr>
<tr>
<td>Müllarian hypoplasia</td>
<td>1</td>
</tr>
<tr>
<td>Transverse vaginal septum</td>
<td>6</td>
</tr>
<tr>
<td>Vaginal atresia</td>
<td>4</td>
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<tr>
<td>Vaginal atresia with fistula</td>
<td>1</td>
</tr>
<tr>
<td>Cervical dysgenesis</td>
<td>2</td>
</tr>
<tr>
<td>Combined cervical/vaginal atresia</td>
<td>3</td>
</tr>
<tr>
<td>Imperforate hymen</td>
<td>4</td>
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</tbody>
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<tr>
<th>Table 2</th>
<th>US and MRI diagnosis in all 30 cases.</th>
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<tbody>
<tr>
<td>Cervico-vaginal anomalies</td>
<td>Patient no.</td>
</tr>
<tr>
<td>Vaginal anomalies</td>
<td>15/30</td>
</tr>
<tr>
<td>Transverse vaginal septum</td>
<td>6/15</td>
</tr>
<tr>
<td>Vaginal atresia</td>
<td>4/15</td>
</tr>
<tr>
<td>Vaginal atresia with fistula</td>
<td>1/15</td>
</tr>
<tr>
<td>Imperforate hymen</td>
<td>4/15</td>
</tr>
<tr>
<td>Cervical dysgenesis</td>
<td>2/30</td>
</tr>
<tr>
<td>Combined cervico-vaginal anomalies</td>
<td>13/30</td>
</tr>
<tr>
<td>Agenesis/hypoplasia</td>
<td>10/13</td>
</tr>
<tr>
<td>Atresia</td>
<td>3/13</td>
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<tr>
<th>Table 3</th>
<th>Accuracy of multiapproach US and MRI in diagnosis of cervico-vaginal anomalies in 30 cases.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Multiapproach US</td>
<td>MRI</td>
</tr>
<tr>
<td>94.1%</td>
<td>97.1%</td>
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In our study, 4 cases (13.3%) out of 30 were associated with class 3 müllerian duct anomaly, and nonobstructed rudimentary horn could not be seen by any of US approaches in one case. MRI showed gross uterine anatomy with identification of bicornuate anomalies in 3 cases and unicornuate uterus with nonobstructed rudimentary horn in one case. In obstructed anomalies retained blood products were seen as high signal on T1 weighted images and variable signal intensity on T2 weighted images. Secondary processes involving the ovaries and adnexa, such as endometriosis were properly characterized. Cases with atresia, vaginal septum, imperforate hymen and müllerian agenesis were correctly diagnosed with correct measurement of atretic segment (Fig. 4) and thickness of vaginal septum. One case with internal cervical os stenosis in properly canalized cervical canal was not diagnosed by MRI. It was effective in delineation of fistulous tract in one case of urogenital sinus syndrome (Fig. 5).

The accuracy of multi-approach US and MRI examinations in diagnosis of cervico-vaginal outflow anomalies was 94.1% and 97.1% respectively.

4. Discussion

According to the modified AFS classification (19), uterovaginal anomalies are categorized as dysgenesis disorders or vertical or lateral fusion defects. Anomalies are further subcategorized into obstructive or nonobstructive forms, since their treatment differs. Correct diagnosis and classification of uterovaginal anomalies is needed to determine cases requiring interventional therapy (20).

Obstructive uterovaginal anomalies require immediate attention because of retrograde flow of trapped mucus and menstrual blood and increasing pressure on surrounding organs (21). In this study, we included 30 cases of cervico-vaginal anomalies, 17 cases were obstructed, and 13 cases with hematometra and 4 cases with hematometrocolpos, associated hematosalpinx (one case) and endometriomas (9 cases) were detected.

Transverse vaginal septum occurs in approximately 1 in 30,000 to 1 in 80,000 women. These septa may be located at various levels in the vagina; approximately 46% are found in the upper vagina, 35–40% in the middle portion and 15–20% in the lower vagina. The septa are generally less than 1 cm in thickness and may have a small central or eccentric perforation. The majority of transverse vaginal septa have a fenestration and are thus not completely obstructed (22). We had 6 cases (20%) with transverse vaginal septum, 3 were located in upper vagina just below the cervix, other 3 were lower septum (2–3 cm) from perineum. They were complete without fenestration, and the mean thickness was 5 mm. Both US and MRI could delineate the septum, its position and thickness.

![Fig. 1](image_url)

Female 13 year old complaining of cyclic abdominal pain. (A) TAS longitudinal scan revealed distended uterus, cervix and upper vagina. (B) Axial TAS showing obstructed unicornuate uterus with upper transverse vaginal septum, rudimentary horn is seen on the right (arrow). (C, D) Coronal and axial T2 confirmed US findings.
In this study we had 2 cases with complex anomalies of both lateral fusion and vertical defects (one didelphys with one obstructed hemivagina with transverse septum, other unicorneate with rudimentary horn, third bicorneate with high transverse vaginal septum), they had associated renal agenesis, and both are well assessed by US & MRI. Smith and Laufer in 2007 in their research on obstructed hemivagina and ipsilateral renal anomaly (OHVIRA) syndrome found that the mean age of diagnosis was 14 years. Twenty-three patients had ipsilateral renal anomalies, including 20 patients who had renal agenesis. Two had dysplastic ipsilateral kidneys requiring nephrectomy in infancy. Twenty-six patients underwent vaginal reconstruction, and eight of those additionally underwent laparoscopy for clarification of diagnosis. Six required a two-stage vaginoplasty because of incomplete previous resection (n = 1), infection or anatomic distortion (n = 4), or restenosis (n = 2). Vaginal septum adenosis was seen in eight patients (23).

Complex congenital anomalies of the müllerian system can occur in isolation or in association with other developmental disorders involving the cloaca, urogenital sinus and anorectal areas (24). Minto et al. in 2001 confirmed the accuracy of magnetic resonance imaging as a sensitive and specific diagnostic investigation for müllerian anomalies in this complex group.

Fig. 2 Married female 33 year old complaining of primary amenorrhea and cyclic abdominal pain. (A) Transperineal US showing normal canalized lower vagina (2.9 cm in length). (B&C) TVS: showing upper atretic vaginal segment (hypoechoic cord like appearance 1.7 cm), also the cervix is not canalized, uterus showed thickened endometrium. (D&E) Axial and sagittal T2 MRI showing atretic cervix (arrow), the atretic part of vagina is not well delineated. (F) Coronal T1 showed minimal blood in the cavity, associated right endometrioma.
of adolescent patients (16). One of our cases had urogenital sinus syndrome with delayed presentation at the age of 42, such anomalies with upper vaginal atresia and fistulous communication with urethra were properly delineated by MRI examination with instilling of endovaginal aqueous gel, and such findings could not be diagnosed by US approaches.

Congenital atresia of the uterine cervix has become recognized as a clinical entity that is distinct from other müllerian anomalies. The visualization of the cervix and cervical canal is important, as the presence or absence of the cervical canal is considered fundamental in the decision whether or not to conserve the uterus (25). In the study of Fujimoto et al. in

**Fig. 3** Female 13 year old complaining of cyclic abdominal pain. (A) TAS longitudinal scan shows haematocolpos. (B) Transperineal US shows canalized lower vagina with normal mucosa and transverse vaginal septum. (C) Sagittal T2 MRI showing lower transverse vaginal septum, 5 mm in thickness (arrow).

**Fig. 4** Female 18 year old complaining of primary amenorrhea and infertility. (A) Transperineal US showed atretic vagina (in between calibers), anterior hypoechoic strand (urethra*), uterine shadow could not be assessed. (B) Sagittal T2 MRI showing long atretic segment more than that seen on US (arrows), small hypoplastic uterus*, and lower vaginal pouch.
1997, forty-eight percent of their patients had isolated congenital cervical atresia with a normal vagina whereas the reminder had either complete or partial vaginal atresia (25). In our study, cervical atresia was associated with vaginal atresia in three cases. Isolated cervical atresia was seen in one case. Such cases were diagnosed by different US approaches and MRI examination. We had one case with internal os stenosis. It was diagnosed by transvaginal US examination and missed on MRI, and this may be attributed to focal stenosis in normal canalized cervix.

Congenital absence of the vagina is a relatively rare condition most commonly associated with Mayer–Rokitansky–Küster–Hauser (MRKH) syndrome. Historically, several reconstructive techniques have been described to provide for functional vaginal reconstruction on these patients, both operative and nonoperative (26). In this study we had 9 cases of uterine agenesis with lower vaginal pouch ranging from 2 to 3.5 cm as measured by both transperineal US & MRI.

Fedele and his colleagues concluded that transrectal ultrasonography could be considered as a diagnostic procedure of choice in the assessment of vaginal canalization defects (27). TRUS was done in 8 cases. It can replace TVUS with proper delineation of the cervix and whole vagina when such approach cannot be performed.

In conclusion, ultrasound examination using different approaches being noninvasive, cheap and easily available remains the initial investigation for all patients with simple müllerian anomalies. MRI examination could be reserved for more complex anomalies, long cervicovaginal atresia and for cases of urogenital sinus syndrome.

Conflict of interest

None declare.

References


