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A Case Report of Coital Trauma to the Genital Tract in a Patient with Mayer-Rokitansky-Küster-Hauser (MRKH) Syndrome: Diagnostic Imaging and Surgical Management

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Running Title Mayer-Rokitansky-Küster-Hauser (MRKH) Syndrome as a Müllerian Duct Anomaly

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ABSTRACT

Müllerian duct anomalies (MDAs) are congenital malformations resulting from abnormal development of the Müllerian ducts. Among these anomalies, Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome, characterized by uterine and vaginal agenesis or aplasia, is a severe form often presenting with primary amenorrhea. This case involves a 25-year-old woman with vaginal bleeding, diagnosed by MRI as having MRKH syndrome with cervicovaginal atresia and undescended ovaries. The patient underwent hysteroscopy and improved post-surgery. MRKH syndrome requires a multidisciplinary approach for management, including surgical intervention, psychological support, and fertility counseling, highlighting the importance of early diagnosis and personalized care.

Introduction

Müllerian duct anomalies (MDAs) are congenital abnormalities resulting from improper development, fusion, or resorption of the paramesonephric (Müllerian) ducts during embryogenesis. Their prevalence ranges from 0.1% to 3.5% in the general population [1] and 5–10% in women with infertility and recurrent pregnancy loss[2]. MDAs are often associated with renal and skeletal anomalies due to their shared embryological origins, with renal malformations occurring in nearly 29% of cases [3,4].

The female reproductive tract begins forming between the fifth and sixth weeks of gestation from the paired Müllerian ducts, which later differentiate into the fallopian tubes, uterus, cervix, and upper two-thirds of the vagina [5, 6]. The absence of the Y chromosome and hormonal factors regulate this differentiation [7]. Disruptions in this process can

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result in various MDAs, including agenesis, incomplete development, or abnormal fusion and resorption of the ducts.

MDAs are classified into seven categories, with the American Society for Reproductive Medicine (ASRM) and the European Society of Human Reproduction and Embryology and the European Society for Gynaecological Endoscopy (ESHRE/ESGE) providing widely accepted classification systems. The spectrum of Müllerian duct anomalies includes conditions such as Müllerian agenesis, cervical agenesis, unicornuate uterus, uterine didelphys, bicornuate uterus, septate uterus, arcuate uterus, and T-shaped uterus (the latter associated with diethylstilbestrol [DES] exposure), as well as more complex anomalies like Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome [8]. Among these, the septate uterus poses the highest risk for adverse pregnancy outcomes [9]. MRKH syndrome, also known as Müllerian aplasia or congenital absence of the uterus and vagina, is one of the most severe uterine malformations [10]. This disorder is characterized by the agenesis or aplasia of the uterus and upper two-thirds of the vagina in females with a normal female karyotype (46, XX). External genitalia are usually normal, and most patients with MRKH syndrome have normal reproductive endocrine function. They typically experience puberty with the usual signs of thelarche and pubarche. Primary amenorrhea, often presenting during adolescence, is the most common symptom. MRKH syndrome accounts for approximately 16% of cases of primary amenorrhea, making it the second most common cause after ovarian failure. In some instances, Müllerian aplasia may be associated with other extragenital abnormalities, particularly involving the kidneys and skeletal system [11].

Advancements in imaging have improved MDA diagnosis. Three-dimensional ultrasonography is an effective initial tool, while magnetic resonance imaging (MRI) remains the gold standard due to its high resolution and multiplanar capability [12]. Hysterosalpingography (HSG) is useful for assessing tubal patency but has limitations in evaluating uterine morphology [13].

The exact etiology of MDAs is not fully understood, though they are believed to have polygenic and multifactorial origins [3]. Environmental factors such as prenatal exposure to teratogens, ionizing radiation, and viral infections may contribute [14]. Genetic studies suggest possible roles for anti-Müllerian hormone mutations and apoptotic pathway disruptions in their development [15]. We present a case of Müllerian duct anomaly, with a focus on the diagnostic process and management approach.

Case Presentation

A 25-year-old woman was referred to Firoozgar Hospital from a local hospital near Tehran due to vaginal bleeding following coitus. The patient presented with primary amenorrhea and had no other significant past medical history. She underwent a series of paraclinical tests to assess her overall health status and rule out other potential causes of her symptoms. Based on the results of these tests, creatinine, serum sodium, serum potassium, liver enzymes (SGOT, SGPT), total and direct bilirubin, MCV (mean corpuscular volume), and platelet count were all normal. Alkaline phosphatase and WBC (white blood cell count) were elevated (251 U/L and 13.2 × 10^3/µL, respectively), indicating possible infection or bone abnormality. RBC (red blood cell count), Hb (hemoglobin), and Hct (hematocrit) were below the normal range, suggesting mild anemia (3.58 million/µL, 11.8 g/dL, and 34.7%, respectively). On gynecologic examination, the patient demonstrated an absent or blind-ending vaginal canal consistent with cervicovaginal atresia. The external genitalia appeared normal. No palpable uterine body was appreciated on bimanual examination. Abdominal palpation did not reveal any masses. Secondary sexual characteristics were normal.

The initial differential diagnosis included a possible vaginal anomaly. To confirm the diagnosis, a pelvic MRI was performed, both with and without contrast.

The pelvic MRI revealed significant findings consistent with a complex Müllerian duct anomaly. The imaging showed two widely separated uterine horns with normal myometrial signal intensity, but notably, no obvious endometrial cavity. At the expected location of the lower uterine segment and cervix, two low-signal intensity fibrotic bands were observed. These fibrotic tissues extended into the vaginal region, connecting at the level of the vagina, and extended into the upper two-thirds of the vaginal canal (Figures 1 and 2).

The lower third of the vagina appeared normally developed; however, a rupture in its wall was noted, with associated fluid collection extending into the perineum (Figures 3 and 4).

Undescended ovaries were noted in the retroperitoneum, both containing normal follicles (right ovary: 27 × 18 mm; left ovary: 25 × 17 mm).

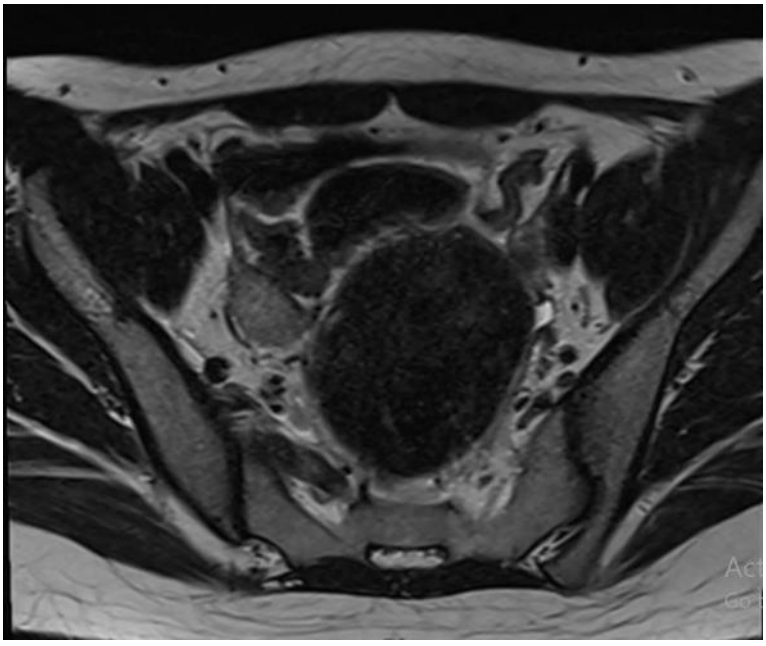

Fig. 1. MRI showing right uterus horn

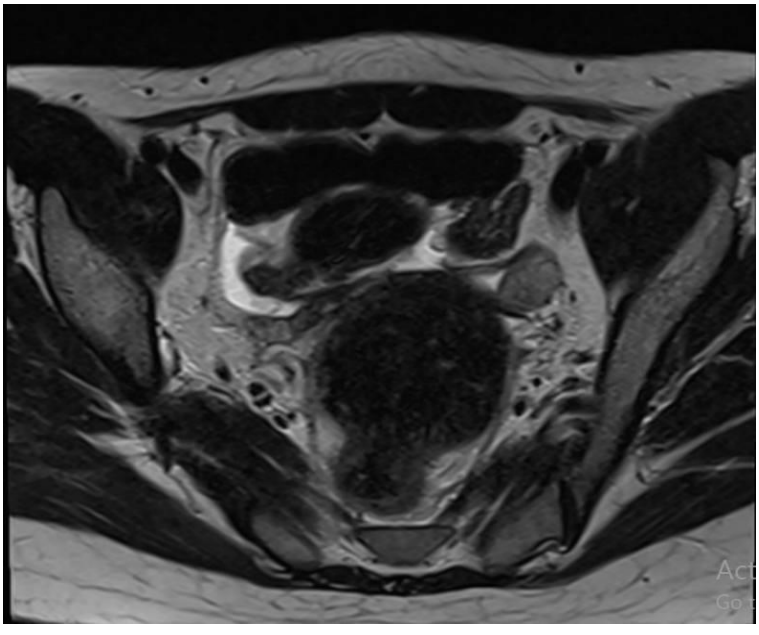

Fig. 2. MRI showing left uterus horn with fibrotic bands

The bladder appeared normal, with no abnormalities in wall thickness or intraluminal filling defects. The rectum also showed normal appearance, without any mass lesions. No significantly enlarged lymph nodes were seen. The visible bony structures and muscles were intact.

The findings were consistent with a complex Müllerian duct anomaly, characterized by divergent, widely separated rudimentary uterine horns without an endometrial cavity, along with cervicovaginal atresia, which is compatible with Mayer-Rokitansky-Küster-Hauser syndrome.

The patient underwent vaginoscopy under anesthesia for direct visualization and assessment of the uterine anatomy. During the procedure, vaginal fibrotic bands were addressed, and the vaginal wall rupture was repaired. Postoperatively, the patient showed significant improvement in her condition.

To further address the identified Müllerian duct

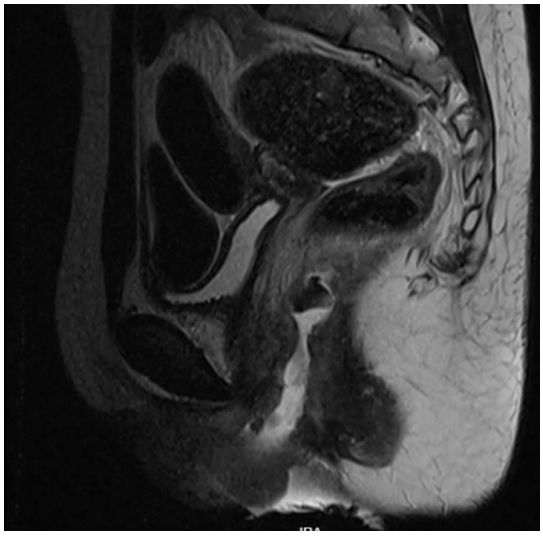

Fig. 3. MRI showing fluid collection

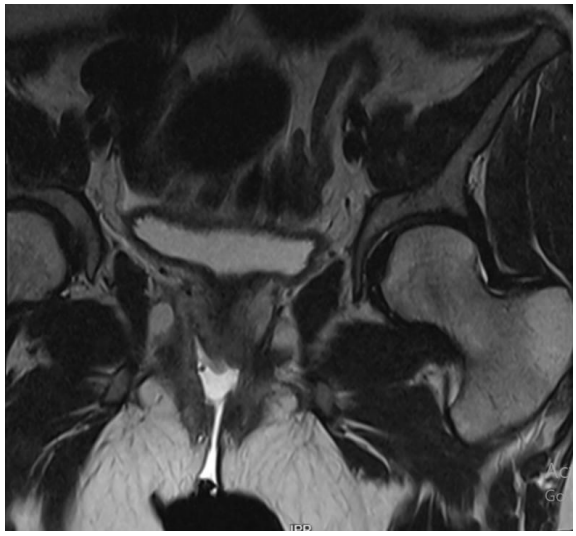

Fig. 4. MRI showing fluid collection

anomaly and enhance the patient's quality of life, a plan for additional surgical intervention, specifically vaginoplasty, was discussed. However, the patient stated that due to her upcoming migration to another country, she could not undergo surgery and preferred to seek treatment in her new location.

Discussion

Müllerian duct anomalies (MDAs) are a diverse group of congenital malformations resulting from the abnormal development, fusion, or resorption of the Müllerian ducts during embryogenesis [16]. Among the various types of MDAs, Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is one of the most severe forms, characterized by the agenesis or aplasia of the uterus and the upper two-thirds of the vagina in women with a normal 46, XX karyotype. The syndrome is typically associated with primary amenorrhea, as in the case of our patient, and is responsible for approximately 16% of all cases of primary amenorrhea, making it the second most common cause after ovarian failure [11].

The diagnosis of MRKH syndrome is often suggested based on clinical findings, such as primary amenorrhea and the absence of menstrual periods in otherwise healthy women, typically during adolescence [17]. Our patient, a 25-year-old woman, presented with vaginal bleeding after coitus, which is an unusual but not uncommon presentation in MRKH syndrome, potentially resulting from vaginal atresia or other associated anatomical anomalies. Initial laboratory tests revealed mild anemia and elevated white blood cell count, suggestive of an inflammatory process, likely due to the rupture in the vaginal wall and associated fluid collection, as seen on MRI.

By reviewing the pre-existing literature, we found some few cases, reporting such complex condition. For example, Pesikhani et al. reported a 13-year-old girl with MRKH syndrome and cervicovaginal agenesis, where both ovaries were normal [18]. Another study by Jaha et al. presented a 25-yer-old female with a history of primary amenorrhea and two episodes of menstruation at menarche. Imaging revealed two widely separated noncommunicating uterine horns with a hypoplastic cervix and upper vagina, while the lower one-third of the vagina and ovaries were normal [19]. Our case can be particularly significant due the simultaneous presence of MRKH syndrome, cervicovaginal atresia and undescended ovaries. The detection of undescended ovaries is especially important, as it needs critical implications for both fertility preservation strategies and the assessment of potential malignancy risk, given the atypical positioning and possible disruption of normal hormonal and anatomical development. Furthermore, the coexistence of these congenital abnormalities introduces substantial complexity in the diagnostic process, necessitating detailed imaging. It also complicates surgical planning and long-term gynecologic care, underscoring the need for a multidisciplinary approach to optimize outcomes in such patients.

MRI remains the gold standard for diagnosing MDAs due to its high resolution and multiplanar imaging capabilities [20]. In this case, MRI findings were consistent with MRKH syndrome, showing two widely separated uterine horns without an endometrial cavity, which is typical of this anomaly [21]. The

fibrotic bands observed at the cervicovaginal junction and the rupture in the lower third of the vagina, with fluid extending into the perineum, further supported the diagnosis of cervicovaginal atresia. Additionally, the presence of undescended ovaries containing normal follicles is commonly seen in MRKH syndrome, as ovarian function is usually unaffected [22]. These findings highlight the complexity of the syndrome and the importance of comprehensive imaging in confirming the diagnosis.

The management of MRKH syndrome is multifaceted, addressing both the anatomical and psychological aspects of the disorder. In this case, hysteroscopy was performed to manage the vaginal bleeding and to assess the vaginal anatomy. The patient responded well to the procedure, and post-operative care was aimed at preventing infection, addressing potential complications, and promoting recovery. Antibiotic prophylaxis with ciprofloxacin and metronidazole was prescribed, and the patient was given clear post-operative care instructions, including hygiene recommendations and restrictions on sexual activity to allow for proper healing.

In addition to surgical management, fertility counseling and psychological support are integral components of care for patients with MRKH syndrome [23]. Although affected individuals are typically infertile due to the absence of a functional uterus, many retain normal ovarian function, and assisted reproductive technologies (ART) may provide avenues for family building [24]. Furthermore, surgical options, such as neovagina creation for those desiring vaginal functionality, can improve the quality of life and sexual health for these patients. Emotional and psychological support is critical, as the diagnosis of MRKH can have significant social and emotional implications, particularly related to body image, fertility, and sexual function [25].

Conclusion

This case highlights the importance of proper diagnosis, careful imaging, and a multidisciplinary approach in the management of Müllerian duct anomalies. With appropriate interventions, patients with MRKH syndrome can lead fulfilling lives, and advances in reproductive medicine offer hope for future family-building possibilities.

Ethical Considerations

Compliance with ethical guidelines

There were no ethical considerations to be considered in this article.

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Conflict of Interests

The authors have no conflict of interest to declare.

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