

# Endometriosis in a Patient with Mayer - Rokitansky - Küster - Hauser Syndrome

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**Abstract:** ***Objective:** To report a case of Mayer - Rokitansky - Ku"ster - Hauser syndrome (MRKH) in which there were two nonfunctional rudimentary uteruses with the presence of ovarian endometrioma, corroborating that there are valid alternative theories to the existence of endometriosis, rather than Sampson's theory alone, such as the coelomic metaplasia theory. **Design:** A case report. **Setting:** A tertiary referral center, which is also a university hospital. **Patient:** A sixteen - year - old patient with MRKH syndrome and endometriosis. **Intervention:** Laparoscopic approach for diagnostic confirmation and treatment of the endometrioma. **Results:** Evidence of endometriosis in a patient with no functional uterus. **Conclusions:** This case report and a few others that are available in the literature reinforce the possibility that coelomic metaplasia could be the origin of endometriosis. Patients with mu"llerian agenesis and pelvic pain should be carefully evaluated, and the presence of pelvic endometriosis should not be excluded.*

**Keywords:** endometriosis, mrkh syndrome, sampson theory, coelomic metaplasia, laparoscopy

## 1. Introduction

Mayer - Rokitansky - Ku"ster - Hauser (MRKH) syndrome is characterized by different degrees of mu"llerian duct abnor - malities, usually with the congenital absence of the upper two - thirds of the vagina, together with uterine agenesis or the presence of a rudimentary uterus. The rudimentary uterus can have a functioning or nonfunctioning endometrium. It is the second most common cause of primary amenorrhea, with a reported incidence of 1 in 4000 female live births [1]. Pelvic pain might be associated with hematometrium in case of a functioning uterus. Distortion of anatomy that accompanies mu"llerian abnormalities predisposes the appearance of endometriosis, and therefore such patients, when presenting pelvic pain, should always be considered for diagnosis.

Endometriosis is defined as the presence of endome - trium - like tissue outside of the uterus, which leads to an inflammatory process. Symptoms include cyclic and acyclic pelvic pain, dysmenorrhea, dyspareunia, and infertility, besides gastrointestinal and urinary complaints that may be present when there is deep infiltrating endometriosis.

Diagnosis should be predominantly based on the character - istics of the pain, combined with imaging methods such as ultrasound and magnetic resonance imaging, although the gold standard is laparoscopic visualization and confirmation through biopsy. Treatment is based on medical therapy, which includes both hormonal and nonhormonal drugs such as analgesics, and surgery is also a very effective approach towards pain [2, 3].

There are a few theories that try to explain the mechanism of endometriosis, a condition that is nowadays estimated to affect 10 to 15% of women in reproductive age [4]. One of

the most accepted theories is the theory proposed by Sampson, in which endometrial implants occur because of retrograde menstruation. Other theories suggest that there may be endometriotic cells originated from other tissues rather than the endometrium and uterus [5], for exam - ple, through coelomic metaplasia, a hypothesis proposed by Meyer that suggests that the original coelomic mem - brane undergoes metaplasia, forming typical endometrial - like glands and stroma. The histologic findings of gradual transition from normal - appearing ovarian surface epithelium and ovary epithelial inclusions to minimal formation of endometrioid glandular epithelia, as well as the transition from normal ovarian stroma to endometriotic stroma, pro - vide direct evidence supporting coelomic metaplasia in the genesis of ovarian endometriosis [6].

However, endometriosis is a complex disease, and most likely a group of distinct factors combined have an important role in its pathophysiology, such as familial aggregation and genetic polymorphisms, and hormonal interference through estrogen and progesterone receptors [7]. Each of these mech - anisms might have a simultaneous part in the etiology of the disease, rather than being individually responsible for endometriosis in different patients. That is, immune and endocrine factors, also called endocrine disrupting chemi - cals, could promote the differentiation of stem cells and cells of the peritoneum into endometriotic cells [5].

A few authors have reported endometriosis in patients with MRKH. Cho et al. reported a case of endometrioma in a patient with MRKH with no uterus [8]. Mok - Lin et al. also reported endometriosis in a patient with complete uterus agenesis [9]. Some other studies also describe endometriosis in patients with MRKH, but they report a rudimentary uterus that might have a functioning endometrium. Such is the case

in the study by Parkar and Kamau, in which there were two horns that did not communicate with the vagina and there was also evidence of functioning endometrium, with adenomyosis, resulting in small hematometra bilaterally [10]. In this case report, we present endometriosis in a patient with Mayer - Rokitansky - Ku"ster - Hauser syndrome, without a functioning uterus. This case reinforces the theory of coelomic metaplasia as having a complementary role in the genesis of endometriosis rather than Sampson's retrograde menstruation theory alone.

## 2. Case Report

A fourteen - year - old patient was seen in 2021 with primary amenorrhea and normal secondary sexual characteristics. Thelarche and pubarche occurred at 12 years of age. Some basic laboratory tests they were all within the normal ranges.

USG s/o hematometra, right ovary not seen properly p/o hemorrhagic cyst or endometriosis, right ectopic kidney, left ovary normal. s/p/o Tuboovarian abscess/hematosalpinx.

MRI was not done as patient was not affording. patient remains asymptomatic till 2023 with age 16 year she appear with c/o cyclical abdominal pain. Physical examination performed s/o imperforated hymen. Hymenal opening done with cruciate incision and after that we found blind vaginal pouch. This procedure followed by laparoscopy.

In a laparoscopy done in March 2023, two rudimen - tary uteruses and an endometrioma in the right ovary were observed). Right sided uterine horn which had cavity was removed, right hematosalpinx and right oophorectomy done for right sided chocolate cyst. left sided ovary normal and left sided rudimentary horn of uterus have no cavity. Further plan explain to patient, vaginoplasty before marriage. The biopsy confirmed endometriosis. The continuous use of an oral contraceptive was established and the patient become asymptomatic.

## 3. Discussion

Endometriosis is defined as the presence of tissue similar to the endometrium outside the uterine cavity, which induces a chronic inflammatory reaction, leading to adhesion for - mation and interference with normal reproductive processes [11]. Multiple hypotheses have been suggested to explain the pathogenesis of endometriosis, including theories of retro - grade menstruation [12], lymphatic and vascular metastases [9], immunologic deficiency resulting in insufficient clear - ance of ectopic endometrial cells [13], and coelomic metapla - sia [14]. None of them alone supply a sufficient explanation that can be applied to all cases. This pathology remains an enigma despite the extensive clinical investigations and expe - rience. It is suggested that an endometrioma is a pseudocyst formed by accumulation of menstrual debris from endome - trial implants adherent to the peritoneal layer of the ovary, which generate from adhesion of active superficial implants in that peritoneum and posterior invagination [15, 16].

According to the Brazilian Society of Endometriosis and Minimally Invasive Gynecology (SBE), this disease has

become, in recent decades, a public health problem, with significant morbidity and unquestionably high costs. Accord - ing to data from SBE from 2009, currently in Brazil about six million women have the disease. For unknown reasons, both the incidence and aggressiveness of the disease have been increasing alarmingly. However, advances of diagnostic methods and laparoscopic techniques may be one of the factors involved in increased incidence [17].

The prevalence of endometriosis in patients with Roki - tansky - Ku"ster - Hauser syndrome (MRKH) without function - ing endometrial tissue appears to be very low. A diagnosis of MRKH involves physical examination and imaging modali - ties. Magnetic resonance imaging (MRI) is the gold standard for the uterus and surrounding structures [18], allowing for better visualization of mu"llerian structures and better delin - eation of endometrium than ultrasound. As with our patient, secondary sexual characteristics and external genitalia are generally normal, as are hormonal levels; a vaginal dimple or short, blind - ending vagina and a imperforated hymen are usually present [19]. In the present case, after imaging methods and laparoscopic visualization and approach, diagnosis was obtained, as well as clinical improvement and a satisfactory outcome for the patient, especially with the use of continuous oral contraception.

According to the literature, it is possible to notice that patients with MRKH syndrome who present acute pelvic pain, endometriotic ovarian cysts, or adenomyotic mu"llerian remnant should be considered for diagnosis; and MRI and laparoscopy are the recommended diagnostic tools and in regard to laparoscopy generally also the treatment. For a safe laparoscopic approach in women with mu"lleri - an abnormalities, according to Will et al., some recommen - dations should be followed; for example, an adequate preop - erative assessment of the urinary tract is imperative, given the high incidence of associated anomalies. Considering the approach of the uterine remnants, medial traction has to be part of the surgical technique to avoid lateral pelvic wall injuries, and the surgeon should be aware of possible vascular anomalous supply. [20]

## 4. Conclusions

There are many theories that try to explain the origin of endometriosis. The most accepted is Sampson's retrograde menstruation theory. This theory is reinforced by some studies that report that obstructive mu"llerian anomalies are more associated with endometriosis, by which an increase in retrograde menstrual flow occurs secondary to obstruction. Sampson's theory is also supported by the observation of the anatomical distribution of endometriotic lesions, which is asymmetric in a way that would be expected after retrograde menstrual flow. This theory cannot explain, however, the appearance of endometriosis in patients with nonfunctioning uteruses, which is the case in some patients with Mayer - Rokitansky - Ku"ster - Hauser syndrome. Even the lymphatic and vascular dissemination theory and the immunologic deficiency theory can be discarded in these cases.

This case report and a few others that exist in the literature

reinforce the possibility that coelomic metaplasia could be the origin of endometriosis, described as the transformation of pluripotential cells in endometrial cells in the peritoneal cavity. Patients with müllerian agenesis and pelvic pain should be carefully evaluated, and the presence of pelvic endometriosis should not be excluded.

### Conflict of Interests

The authors declare that there is no conflict of interests regarding the publication of this paper.

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