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Laparoscopic intervention for solid pelvic tumours in Mayer-Rokitansky-Küster-Hauser syndrome: a case of bilateral uterine adenomyomas of the rudimentary uterus

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SUMMARY

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome, a condition caused by Müllerian anomalies, is characterised by congenital vaginal aplasia and a rudimentary uterus. Case reports concerning uterine fibroids associated with MRKH syndrome are limited, and differentiating between uterine fibroids and ovarian solid tumours prior to surgical intervention is often challenging. Here, we present the case of a patient with MRKH syndrome and asymptomatic bilateral pelvic solid tumours located close to both ovaries. Based on intraoperative and histopathological findings, the tumours were diagnosed as adenomyomas of the rudimentary uterus. This is the first reported case of a uterine adenomyoma associated with MRKH syndrome. Moreover, our report highlights the fact that diagnostic laparoscopy is a valuable method to evaluate pelvic tumours in MRKH syndrome.

BACKGROUND

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome, a spectrum of Müllerian anomalies, occurs in approximately 1 in 4500 women and is characterised by congenital absence of the uterus and vagina.[1]

Although the uterus is often poorly developed or rudimentary in patients with MRKH, secondary sexual characteristics are present, as chromosomes include 46XX normal karyotypes with normal ovaries and fallopian tubes. MRKH syndrome is associated with urinary tract, skeletal and auditory abnormalities.[1]

In MRKH syndrome, there is no normal endometrium development in the rudimentary uterus, and complications such as uterine fibroids and adenomyosis are rare.[2] Therefore, when uterine fibroids or adenomyosis is suspected in the rudimentary uterus, it is essential to differentiate these from a solid ovarian tumour.[3–8]

Here, we present the case of a patient with known MRKH syndrome who was incidentally found to have elevated tumour marker levels and asymptomatic bilateral pelvic solid tumours noted on transabdominal ultrasound. Laparoscopic surgery led to a definitive diagnosis of uterine adenomyoma. To our knowledge, this is the first case report of MRKH syndrome complicated with uterine adenomyoma.

CASE PRESENTATION

A single woman in her 40s initially presented to our hospital with primary amenorrhoea at the age of 25 years and was diagnosed with MRKH syndrome. She had a complete vaginal defect but did not wish to undergo vaginoplasty. Urinary tract and ovarian functions were normal, and she was under observation without periodic visits.

Recently, during a general physical examination, two solid pelvic masses were noted on transabdominal ultrasound examination. She was referred to our hospital for further examination. Blood test results showed elevated tumour marker levels (CA19-9, 113.6 U/mL; CA125, 88.3 U/mL). Although an increase in the CA19-9 level was observed, she had previously undergone colonoscopy at the health examination centre when she had a general physical examination, and there were no abnormal findings. Therefore, we did not conduct another colonoscopy at our facility. MRI revealed two substantial tumours of different sizes (3 cm and 4 cm on the left and right sides, respectively) in the bilateral adnexal regions (figure 1). The presence of normal ovaries on both sides and a rudimentary uterus in the pelvic cavity led to a preoperative diagnosis of bilateral ovarian solid tumours or uterine fibroids. Laparoscopic surgery was performed to confirm the diagnosis.

There were no adhesions in the pelvic cavity, and no endometriotic lesions were detected. Corpus uteri were absent, and rudimentary uterine horns were observed, fused medially and extending laterally along the pelvic wall. The round ligament was attached to the rudimentary uterine horn. The bilateral adnexa were unremarkable and attached to the rudimentary uterine horns on each side. A solid tumour was observed on both sides, between the round ligament and fallopian tube, covered by the peritoneum (figure 2). Based on the tumour findings, a diagnosis of uterine myomas was made, and a laparoscopic myomectomy was planned.

After visualising the ureter, the solid tumours from the rudimentary uterus were removed, leaving the round ligament on the pelvic wall. The rudimentary uterus was preserved to the maximum extent possible. The tumours were collected in a bag and removed from the abdominal cavity by in-bag manual morcellation using a scalpel. The total tumour weight was 70 g.

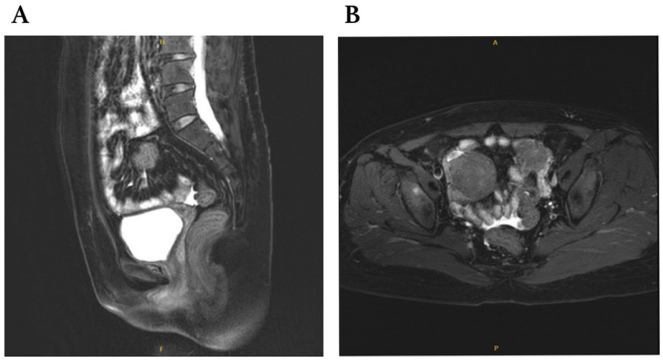

Figure 1 (A) MRI T2 sagittal view. The vagina is absent, and the rudimentary uterus is located between the bladder and rectum. (B) MRI T2 axial view. Bilateral solid tumours are observed in the pelvic cavity.

Histopathological findings showed endometrial tissue inside the myoma nodules (figure 3), indicating that the tumours were adenomyomas of the rudimentary uterus.

OUTCOME AND FOLLOW-UP

Postoperatively, the tumour marker levels decreased to the normal range. The patient was discharged from the hospital after 3 days without any complications. She was able to resume daily activities within a few days after discharge. At the 2-week postoperative follow-up examination, she had no complications and was in a good condition. She returned to work at 2 weeks postoperatively and was followed up at 1, 6 and 12 months postoperatively, with no recurrence of adenomyoma observed on ultrasound. At the patient's request, the follow-up was completed at 12 months.

DISCUSSION

The uterus in patients with MRKH syndrome is hypoplastic, and a functional endometrium is rarely seen.[2 9] Generally, invasion of the endometrial tissue into the myometrium results in the development of uterine adenomyosis.[2] However, the endometrium is often absent in patients with MRKH syndrome. Alternatively, the pathogenesis of adenomyosis in MRKH syndrome may be attributed to ectopic endometrial growth in the myometrium of the uterus.[10] Adenomyotic lesions in the uterus that develop in a focal and localised manner are defined as adenomyomas.[11] In this case, the origin of the lesion could not be identified, as the rudimentary uterus had not been removed. However, as the patient had no menstruation-related symptoms, we assumed that the focal growth of the adenomyoma was caused by the ectopic endometrial tissue that had developed in the rudimentary uterus, as hypothesised above.

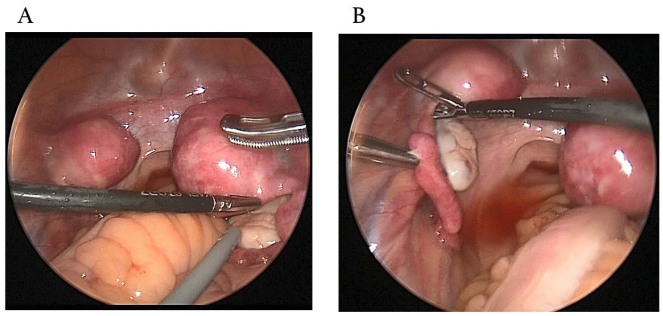

Figure 2 Laparoscopic findings: a solid tumor is observed between the round ligament and the fallopian tube on each side, covered by the peritoneum. Bilateral adnexa are unremarkable.

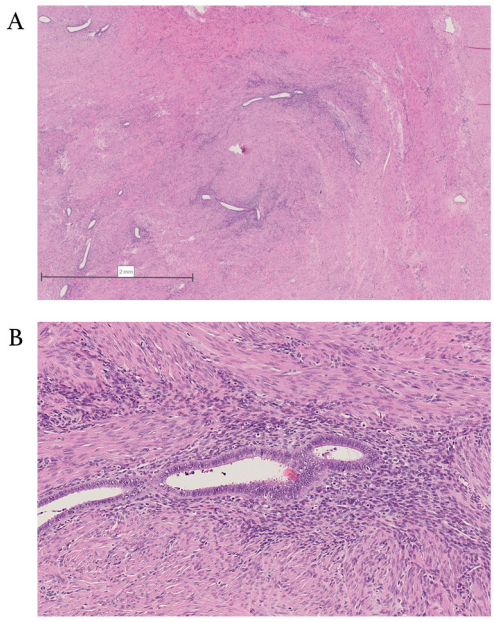

Figure 3 Histopathological findings. (A) Endometrial tissues are observed inside the myoma nodules. (B) Magnified image of figure A.

The diagnosis of pelvic tumours associated with MRKH syndrome is often difficult in the absence of a normal uterine corpus.[3–8 12 13] Non-invasive imaging modalities, such as ultrasound, CT and MRI, are the first choices for the diagnosis of pelvic tumours.[12–14] However, in cases of uterine fibroids arising in a rudimentary uterus, the tumour may be located very close to the adnexa, often making evaluation based on imaging findings alone difficult and necessitating surgical intervention. Laparoscopic observation is an effective means of diagnosis.[4 7 12] In our case, it was challenging to differentiate between uterine myoma and ovarian tumours using imaging alone, because the tumours were accompanied with elevated tumour marker levels and were located close to the bilateral adnexa. Direct observation of the pelvic cavity using laparoscopy enabled us to confirm that the tumours originated from the rudimentary uterus and not from the ovaries, thus resulting in appropriate treatment.

Regarding the treatment of myoma and adenomyosis in MRKH syndrome, previous reports have recommended surgery for symptomatic cases, including differentiation from ovarian tumours. However, currently, there is no consensus on the treatment methods. In the most cases, resection of the entire uterus in asymptomatic patients to prevent recurrence[15 16] and removal of the entire uterus in symptomatic patients are preferred. In addition to our case, only four other cases of tumour removal alone have been reported.[12 17 18] Anatomical structures may differ in women with MRKH syndrome; therefore, there is a risk of surgical complications, such as damage to the surrounding blood vessels, ureter and bladder, when removing the rudimentary uterus. We consider that tumour removal alone is sufficient, especially when there are no symptoms or suspicion of malignancy.

However, some cases of MRKH syndrome complicated with malignant ovarian tumours have been reported despite the condition being rare.[19–23] Their findings indicate that if prior findings of suspected malignancy are observed

on imaging, total hysterectomy and bilateral salpingo-oophorectomy may be considered.

In conclusion, this is the first report of a uterine adenomyoma in a patient with MRKH syndrome. In uterine adenomyomas, similar to other pelvic tumours in MRKH syndrome, laparoscopic surgery and histopathology can facilitate a diagnosis when imaging studies alone are insufficient.

Patient's perspective

I already knew that I do not have a uterus and vagina but have normal ovaries due to my condition. When I was told that there were tumours in my pelvic cavity that looked like ovarian tumours, I was worried that I would lose my ovaries too. However, after surgery, I was relieved to know that the tumours originated from the uterus and not from my ovaries. In addition, I learnt that adenomyomas in MRKH syndrome are rare. Therefore, I wanted to help in making medical advancements in this field by agreeing to publish the findings of my case.

Learning points

- This is the first reported case of uterine adenomyoma associated with Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome.
- Similar to other pelvic tumours in MRKH syndrome, uterine adenomyomas may be difficult to diagnose using imaging alone, and diagnostic laparoscopy may be useful.
- Direct observation of the pelvic cavity using laparoscopy could confirm that the tumours had originated from the rudimentary uterus and not from the ovaries, leading to appropriate treatment.
- Histopathological findings showed endometrial tissue inside the myoma nodules, indicating that the tumours were adenomyomas of the rudimentary uterus.
- Tumour removal alone is sufficient, especially when there are no symptoms or suspicion of malignancy.

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Case reports provide a valuable learning resource for the scientific community and can indicate areas of interest for future research. They should not be used in isolation to guide treatment choices or public health policy.

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