

# Rare case of bizarre leiomyoma in Mayer-Rokitansky-Küster-Hauser Syndrome

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## Summary

The authors report a case of leiomyoma that developed from an aplastic uterus in a 38-year-old Chinese female with a typical Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome. She had normal female secondary sexual characteristics, normal external genitalia with discernible vagina, primary amenorrhea, and infertility with asymptomatic a left adnexal mass. Pelvic MRI showed a mass of 3.7 cm in the left pelvis and a mass of  $1.5 \times 1.4 \times 2.6 \text{ cm}^3$  in the right pelvis without a normal functioning uterus and vagina between the bladder and rectum. Laparoscopic exploration showed that the bilateral rudimentary uteri located laterally in the pelvis and they had a constant caudal relationship with their paired ovary. The bilateral fallopian tubes and ovaries were normal. A leiomyoma developed from the left rudimentary uteri. Removal of the left leiomyoma, bilateral rudimentary uteri and fallopian tubes was performed with preservation of bilateral ovaries. The histologic diagnosis showed that the bilateral rudimentary uteri had no clear structures of uterine cavity and endometrium, and the bizarre leiomyoma developed from the left rudimentary uterus without necrosis or mitosis.

**Key words:** Leiomyoma; Laparoscopic surgery; Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome; MRI.

## Introduction

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is usually present in the form of primary amenorrhea and congenital abnormalities of the female internal genitalia due to early arrest in the development of the Müllerian ducts. The diagnosis is often made either radiologically or laparoscopically in primary amenorrhea women with normal hormonal tests and 46 XX karyotype. Its etiology and pathophysiology are poorly understood and it may be associated with renal, skeletal, cardiac anomalies, and unilateral auditory defects [1]. Leiomyoma can seldom develop from a rudimentary, non-functioning uterus or the Müllerian remnant tissue in patients with MRKH syndrome [2, 3]. The authors report a rare case of bizarre leiomyoma which developed from an aplastic uterus in a patient with MRKH syndrome.

## Case Report

This study was conducted in accordance with the principles outlined in the Declaration of Helsinki, and approved by the Ethical Committee of Clinical Research of Beijing Obstetrics and Gynecology Hospital, Capital Medical University, China (2019-KY-105-01).

A 38-year-old Chinese female, with primary amenorrhea for 20 years, presented with an incidental asymptomatic left adnexal mass detected by diagnostic images two months ago, without symptoms related to the bladder or the bowel.

She had been treated for her primary amenorrhea and cyclical breast swelling when she was 18 years-old, but no further investigation after that. She got married at the age of 25 years without complaint of difficulty during sexual intercourse with her partner and her family history was negative.

She was phenotypically female, with average height and intelligence. She had no history of loss of appetite or weight, and no complaint of galactorrhea, hirsutism or acne. Examination of her head and neck did not reveal any abnormal facies or a webbed neck. Examination of her extremities did not show any obvious abnormalities, such as syndactyly, polydactyly, or absence of digits. Thyroid, chest, and heart examinations were normal. Her breasts and pubic hair were in Tanner's stage 4 with normal female external genitalia.

In pelvic examination, the uterus was not palpable. Compared with the right accessory area, the left area was slightly thicker. The external genitalia were normal and mature type. Vagina examination revealed shallow vaginal canal of 3.0 cm length and cervix was not palpable.

Transabdominal ultrasonography (TAS) showed a homogenous adnexal mass with the size of  $3.8 \times 3.7 \text{ cm}^2$  beside the left ovary without a normal uterine morphology in either the longitudinal or cross-cutting image in the back of the filling bladder. Contrast enhanced MRI of the pelvis revealed a mass of 3.7 cm in the left pelvis and mass of  $1.5 \times 1.4 \times 2.6 \text{ cm}^3$  in the right pelvis without normal functioning uterus and vagina among the bladder, urethra, and rectum;

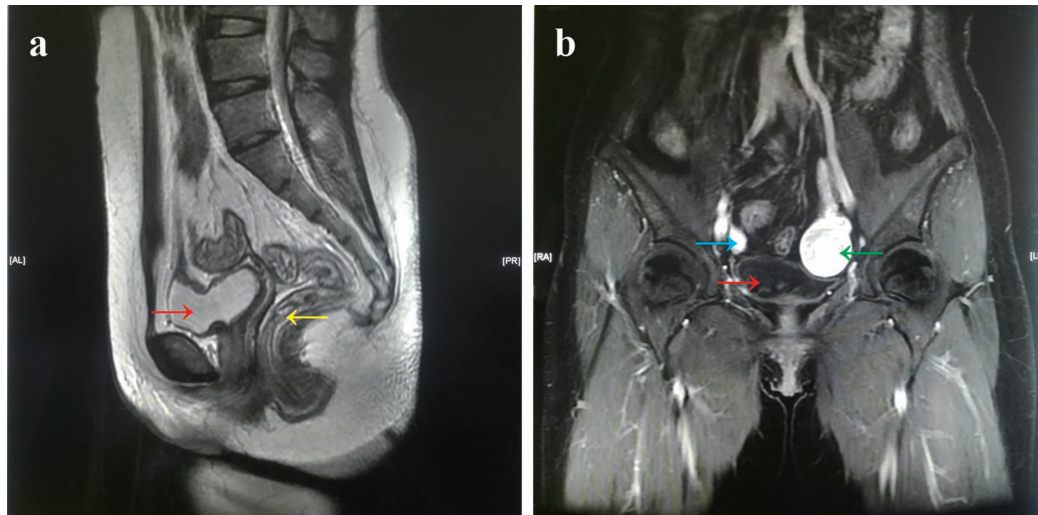


Figure 1. — Magnetic resonance imaging scan. (a) T2WI sagittal view: no obvious uterine tissue is seen between the rectum (yellow arrow) and bladder (red arrow). (b) T1WI coronal post-contrast view: green arrow: left leiomyoma, blue arrow: right rudimentary uterus, and red arrow: bladder.

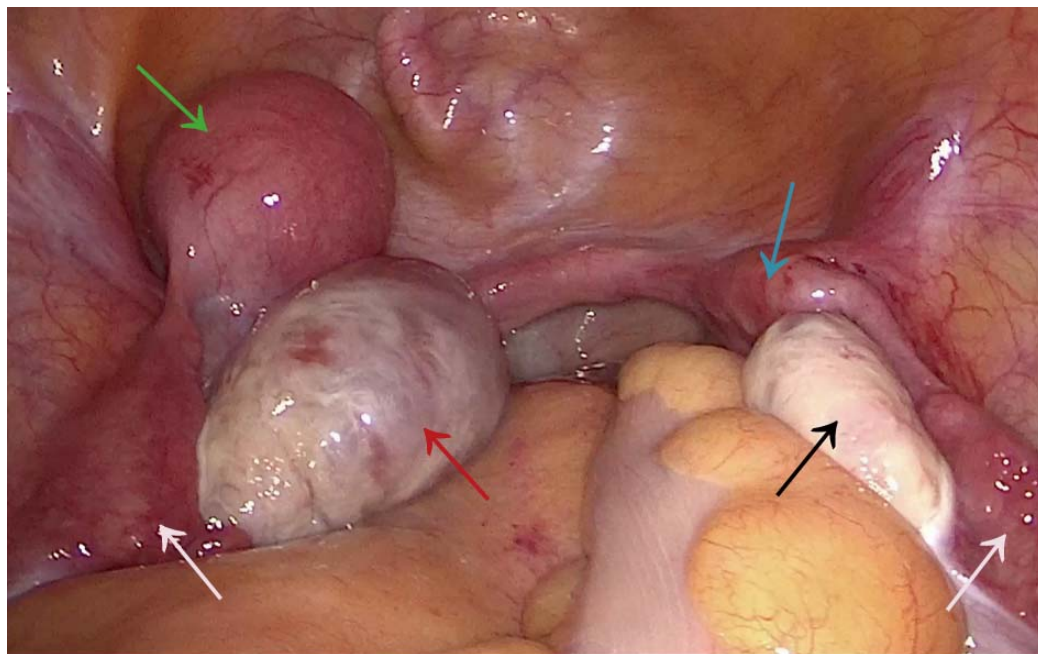


Figure 2. — Laparoscopic view: green arrow: left leiomyoma, blue arrow: right rudimentary uterus, white arrow: bilateral fallopian tube, red arrow: left ovary with the follicular development, and black arrow: right ovary.

the bilateral fallopian tubes and the left ovary were within normal size, position, and signal intensity, while the right ovary was ectopic and located in the right iliac fossa region (Figure 1). Abdominal CT scan showed that the intra-abdominal organs were normal and there were no ascites.

The levels of gonadotropins were normal: follicle stimulating hormone (FSH) was 3.31 U/L and luteinizing hormone (LH) was 2.48 IU/L. The serum levels of tumor markers were also normal: CA-125 was 11.40 U/mL, CA-199 was 4.89 U/mL, and CEA was 0.45 ug/L. The anti-Müllerian hormone (AMH) level was 1.10 ng/mL. Primary

diagnosis of MRKH syndrome associated with pelvic mass (leiomyoma or ovarian tumor) was made and laparoscopic exploration was offered to the patient.

Laparoscopic exploration showed that the bilateral rudimentary uteri had a maximum diameter of 2 cm and located laterally in the pelvis with a constant caudal relationship with their paired ovary. The bilateral fallopian tubes and ovaries were all normal in size, morphology, and position. Follicular development could be seen in the left ovary and there were no abnormal or enlarged vessels noted. The examination showed that a leiomyoma developed from the left



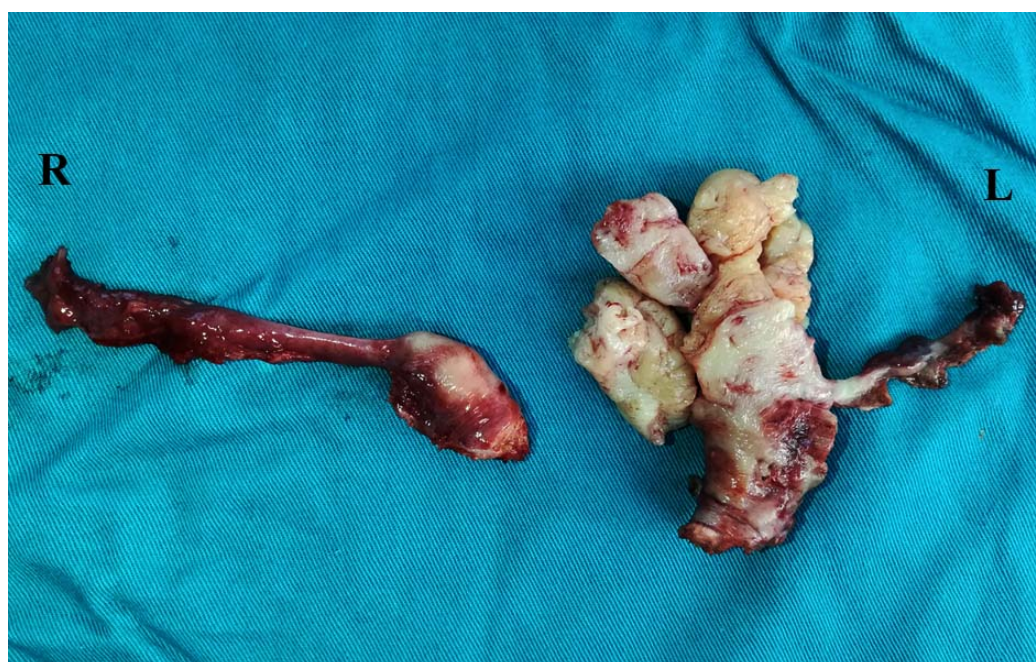


Figure 3. — Cut sections of left leiomyoma, bilateral rudimentary uteri and fallopian tubes.

rudimentary uteri (Figure 2) and it was also complicated by vaginal agenesis. Removal of left leiomyoma, bilateral rudimentary uteri, and fallopian tubes was performed (Figure 3), however, the bilateral ovaries were preserved as the patient was still within child bearing age.

Postoperative pathological examination demonstrated that the bilateral rudimentary uteri did not have clear structures of uterine cavity and endometrium and the bizarre leiomyoma arose from the left rudimentary uterus without necrosis or mitosis.

The patient's recovery was uneventful and was discharged from the hospital in good condition after two days.

## Discussion

MRKH syndrome is a rare congenital disorder and it shows varying degrees of aplasia or hypoplasia of the uterus and the upper two-thirds of the vagina. Müllerian aplasia can be an isolated finding (MRKH type I), although associated anomalies, such as renal, skeletal, auditory, and cardiac anomalies, often coexist (MRKH type II or MURCS association) [4]. Women with MRKH syndrome have a normal karyotype of 46 XX, secondary sexual characteristics, ovarian functions, rudimentary vagina, and endocrine status. These phenomena are due to the different origins of different organs.

The incidence of MRKH syndrome is reported to be one in every 4,000–5,000 female births and is typically diagnosed during puberty [5]. The etiologic factors of this syndrome are not fully understood. Potential reasons are thought to be environmental and genetic factors. It has been suggested that mutations in a major developmental gene or a limited chromosomal imbalance might be involved [4]

with specific genes such as the HOXA7, HOXA9–13, and WNT4 being proposed [6].

Occurrence of leiomyoma of the uterus is very common in the female genital tract. However, occurrence of leiomyoma in MRKH syndrome is extremely rare and only a few cases were reported in the literature. Dimitriadis *et al.* [7] reported a 43-year-old woman with MRKH syndrome who was found to have mitotically active leiomyoma and adenomyosis. Kundu *et al.* [8] reported the first known case of acute surgical presentation secondary to torsion of uterine remnant leiomyoma and ipsilateral adnexa in a woman with MRKH syndrome. In even rarer instances, a leiomyoma can develop in patients with a congenitally absent uterus. Amaratunga *et al.* [3] reported for the first time, a case of an ectopic fibroid in association with congenital absence of a uterus in a 66-year-old white female with MRKH syndrome and unilateral renal agenesis. Salem *et al.* [9] also reported an extrauterine leiomyoma, arising from the intraperitoneal portion of the round ligament in a female with the Müllerian agenesis diagnosed at the age of 40. Here the present authors reported a bizarre leiomyoma originating from the left rudimentary uterus in a 38-year-old woman.

Since the ovaries of the patients with MRKH syndrome are normal, estrogen-dependent pathological conditions including leiomyomas, neoplasms, and adenomyosis can develop in the rudimentary uterus. Similarly, since the proximal ends of Müllerian ducts have smooth muscles, development of leiomyomas in Müllerian remnants is also theoretically possible. However, the incidence of leiomyomas is quite rare probably due to the reduced concentration or sensitivity of estrogen receptors in the Müllerian remnants [10].

In the most recent publications of MRKH syndrome presenting with a pelvic mass, differential diagnosis includes masses originating from female genital tract (leiomyoma, adenomyosis, ovarian tumor, etc.), and deriving from other pelvic organs (gastrointestinal stromal tumor of the intestine and extravesical of the urinary bladder). This can pose a diagnostic dilemma regarding the origin of MRKH. Girma and Woldeyes [11] reported a case of a large leiomyoma with areas of hyaline, edematous, and cystic degenerations originating from the Müllerian remnant, mimicking ovarian tumor in a woman with MRKH syndrome. The diagnostic method needs to be carefully chosen to diagnose the MRKH syndrome combined pelvic mass.

TAS is a simple and non-invasive method, and it can be used to diagnose Müllerian agenesis, and evaluate associated renal anomalies. In TAS examinations, leiomyomas are hypoechoic or heterogeneous masses. Cystic component with internal echogenic material can be seen due to cystic degeneration with hemorrhage or necrosis and the calcifications can be seen as hyperechoic foci. However, TAS is an operator-dependent technique which may fail to identify anatomical structures of the pelvis. CT or MRI is recommended to provide more specific and objective information. In CT examination, leiomyomas are well circumscribed masses with isoto hypodense in the myometrium and show variable enhancement patterns. MRI is more accurate than TAS in defining the exact anatomic characteristics of MRKH syndrome [12]. MRI can sensitively check the malformation of the reproductive, urinary, and skeletal system and it can also depict the distance of an obstructed vagina from the perineum and the thickness of a vaginal septum or atretic segment. MRI is valuable in determining the origin of the mass, defining its relationship with adjacent structures, proposing possible differential diagnosis, and thus guiding further management. In MRI, leiomyomas show low to intermediate signal intensity compared to myometrium in T1 and T2-weighted images. Therefore, the initial diagnosis of MRKH syndrome should utilize the MRI scans to identify possible abnormalities and differentiate the pelvic mass. Although CT or MRI is very useful in the diagnosis of MRKH syndrome and the pelvic mass, surgery might still be the only definitive solution. Laparoscopic or laparotomy surgery allows excellent analysis of a solid pelvic tumor in the patient with MRKH syndrome. Meanwhile, it is also a good treatment method. Removal of the leiomyomas along with the uterine remnants is recommended to reduce risk of recurrence.

## Conclusion

Women with MRKH syndrome who present with abdominal mass, leiomyoma, adenomyosis, or ovarian cysts of Müllerian remnant are recommended for further diagnostic procedures. MRI enables more accurate modalities to delineate the intraabdominal masses before planning for surgery. Complete removal of the masses with the uterine remnant by either laparotomy or laparoscopy is recom-

mended.

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## Conflict of Interest

The authors declare no competing interests.

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