# Large leiomyoma in a woman with Mayer-Rokitansky-Kuster-Hauser syndrome

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ABSTRACT

Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is a rare congenital anomaly characterized as aplasia or hypoplasia of uterus and vagina in women with normal development of secondary sex characteristics. It affects 1 in 4000-5000 female births. Women with this syndrome present with primary amenorrhoea. MRKH syndrome may be associated with renal, skeletal, cardiac and auditory anomalies. Women with MRKH syndrome may develop leiomyoma from a rudimentary uterus, though very rare. Initial investigation in women having MRKH syndrome with leiomyoma is ultrasonography (USG). However, CT and MRI are more accurate to evaluate the pelvic anatomy and pathologies.





## CASE REPORT

### CASE REPORT

A 35 year-old married nulliparous presented with complaints of primary amenorrhoea, mass and pain in the lower abdomen for 2 years. There was no history of cyclical vaginal bleeding, urinary or bowel complaints. She had no history of loss of appetite or weight. Patient had a past history of laparoscopy at the age of 19 years which reported rudimentary uterus with normal bilateral fallopian tubes and ovaries. Physical examination revealed normal bilateral breasts, normal axillary and pubic hair patterns. External genitalia were normal with a blind vaginal pouch of 1cm length. Her abdominal examination revealed a large mobile mass of approximately 30x20 cm size arising from the pelvis. There were no hepato-splenomegaly or fluid thrill. Laboratory investigations revealed normal hormonal profile. CA-125 was 40.5 U/mL (normal value <35 U/mL).

Transabdominal ultrasonography revealed a large pelvic mass having a cystic area with internal echogenic contents (Figure 1). Uterus and ovaries could not be seen on USG.

CT revealed a large heterogeneous mass with cystic degeneration, measuring 17x11cm in size in the pelvis, a small cyst measuring 3x2cm in size in the right ovary and a cyst measuring 6x4cm in size in the left adnexal region (Figure 2 & 3). Uterus and left ovary could not be seen on CT.

MRI confirmed the mass of low to intermediate signal intensity with cystic degeneration on T2-weighted images in the pelvis, a small cyst in the right ovary and a cyst adjacent to the left ovary (Figure 4, 5 & 6). MRI also revealed absence of uterus and vagina (Figure 7).

On the basis of CT and MRI findings, a provisional diagnosis of a large leiomyoma with MRKH syndrome was made. Chromosomal study of the patient revealed a normal karyotype of 46, XX.

The patient was then taken up for laparotomy and abdomen was opened with a midline vertical incision. A large pelvic mass was seen adherent to the rudimentary uterus. Rudimentary right horn of uterus was seen but left horn of uterus could not be seen. There was a 2x2 cm size cyst in the right ovary and right fallopian tube was normal. Left fallopian tube and left ovary were normal and attached to the left side of the rudimentary uterus. A 6x4 cm size left retroperitoneal cyst was seen adherent to the rectosigmoid colon. Myomectomy, right ovarian cystectomy and retroperitoneal cystectomy were performed. The removed large pelvic mass was 25x18x12cm in size and had an intact capsule and a bosselated outer surface

(Figure 8). Cut section of the mass revealed a large cystic cavity measuring 12 cm in diameter and filled with brown coloured fluid and necrotic material. Rest of the mass was grey white in colour and had whorled appearance. Histological features (Figure 9) were suggestive of leiomyoma. Specimen also had two cysts measuring 6.0 and 2.0 cm in diameter filled with straw coloured fluid and diagnosed as benign retroperitoneal cyst and simple right ovarian cyst. The patient was discharged after 5 days in good general condition.

## DISCUSSION

Mayer-Rokitansky-Kuster-Hauser (MRKH ) syndrome is a rare disorder described as aplasia or hypoplasia of uterus and vagina due to early arrest in development of Mullerian duct. The incidence reported is one in 4000-5000 female births [1]. MRKH syndrome is the second most common cause of primary amenorrhoea after gonadal dysgenesis. Women with this syndrome are characterized by presence of 46 XX karyotype, normal female secondary sex characters, normal ovarian functions and underdeveloped vagina [2]. In addition, women with MRKH syndrome may have renal [3], skeletal [4], hearing and cardiac anomalies. Presence of leiomyoma in MRKH syndrome is very rare and only few cases have been reported in the literature. Here we report a patient of MRKH syndrome with a large leiomyoma originating from the rudimentary uterus.

The female reproductive system develops from two embryologic structures - Mullerian ducts and urogenital sinus (UGS). The Mullerian ducts are mesodermal in origin and UGS is endodermal in origin. The development of the urinary system is closely related to the reproductive system; hence its anomalies are also associated with reproductive system abnormalities [3]. Both male and female embryos, at 6weeks of development, have two sets of paired genital ducts- the paramesonephric (Mullerian) ducts (PMDs) and the mesonephric (Wolffian) ducts (MDs). The paired PMDs, initially separated by a septum, fuse and form a Y-shaped structure, uterovaginal primordium (UVP). The UVP and UGS thereafter differentiate to form the female reproductive system [5, 6].

Mullerian duct anomalies have been estimated to occur in approximately 3% of women [7]. As many as 25% of women with Mullerian duct anomalies have reproductive problems as compared with only 10% of general female population [7].

The American Fertility Society (AFS) classification, based on uterine anomalies, is most commonly used to classify Mullerian duct anomalies (Table 1). Anomalies of vagina, tubes and urinary tracts are described as associated malformations. This classification system comprises seven classes - I, uterine hypoplasia and agenesis; II, unicornuate uterus; III, uterus didelphys; IV, bicornuate uterus; V, septate uterus; VI, arcuate uterus; VII, diethylstilbestrol (DES)-related anomalies [8].

MRKH syndrome is a class I Mullerian duct anomaly and it has been subdivided into typical or type A and atypical or type B [9]. Patients with type A syndrome have symmetric muscular buds and normal fallopian tubes. Patients with type B have asymmetric muscular buds (aplasia of one or both buds or, when both buds were found, one bud was smaller than the other one), abnormal fallopian tubes and may be associated with other congenital anomalies like renal agenesis, syndactyly and cardiac defects. The case, we are reporting here is type B MRKH syndrome and class I Mullerian duct anomaly.

Incidence of leiomyoma of uterus is very high in the general female population. But only few cases of leiomyoma have been reported in women with MRKH syndrome [10-12]. Parikh has stated in his review that leiomyomas and adenomyosis rarely develop in the rudimentary uterus in women with MRKH syndrome [13]. The etiopathogenesis of leiomyoma from smooth muscle cells of normal uterus is not known. Their growth has been associated with genetic predisposition, hormones and few growth factors. Leiomyomas of uterus are estrogen dependent tumours. Mullerian ducts have smooth muscle cells at their proximal ends, which probably may give rise to the growth of leiomyoma from the rudimentary uterus in MRKH syndrome. However, the exact etiopathogenesis of leiomyoma from the rudimentary uterus in MRKH syndrome is not known.

Diagnosis of MRKH syndrome is delayed till late puberty. In MRKH syndrome patients usually complain of primary amenorrhea, infertility and pelvic pain. First diagnostic modality is USG in such patients. On USG examination, leiomyomas are hypoechoic or heterogeneous masses. Cystic component with internal echogenic material may be seen in the leiomyomas due to cystic degeneration with necrosis or haemorrhage, like in our case. Calcifications may be seen as hyperechoic foci. Uterus and ovaries may not be seen in patients with a large pelvic mass. CT and MRI are very useful in the diagnosis of MRKH syndrome with leiomyoma and in providing road-map for surgery, latter being very sensitive and specific. On computed tomography (CT), leiomyomas are well circumscribed masses iso- to hypodense to myometrium and show variable enhancement patterns. Cystic areas and calcifications may be seen. MRI has nearly 100% accuracy in diagnosis of Mullerian duct anomalies because of its excellent soft tissue resolution [14]. Uterus and vagina are best evaluated on T2-weighted sagittal and axial MR images. However, diagnostic laparoscopy is the gold standard for definitive diagnosis of MRKH syndrome [15]. MRI is also very useful and accurate in diagnosis and characterization of pelvic masses. On MRI, leiomyomas show low to intermediate signal intensity compared to myometrium on T1 and T2-weighted images. Cystic areas are hyperintense on T2-weighted image.

Differential diagnosis of leiomyoma of rudimentary uterus in MRKH syndrome includes ovarian fibroma, gastrointestinal stromal tumour (GIST) of intestine and extravesical leiomyoma of urinary bladder.

Ovarian fibromas are seen in middle aged women and patients are generally asymptomatic. On USG, fibromas are hypoechoic masses. Cystic areas may be seen. On CT, fibromas are hypoattenuating masses and show minimal contrast enhancement. Fibromas are well circumscribed masses with low signal intensity on T1 and T2-weighted MR images. Cystic areas are hyperintense on T2-weighted images.

Gastrointestinal stromal tumour (GIST) can affect patients of any age but most patient are over 40 years of age. GISTs commonly arise from stomach (70%) and intestinal GISTs are less common. On USG, GISTs are heteroechoic masses. GISTs demonstrate heterogeneous attenuation and moderate to marked contrast enhancement on CT. Cystic areas may be present. These tumours show low signal intensity on T1-weighted MR images and intermediate signal intensity on T2-weighted MR images and moderate to marked contrast enhancement.

Extravesical leiomyoma of urinary bladder is a rare tumour. It presents with complaints of frequency and urgency of micturition. It generally occurs in the 3rd to 6th decade of life. On ultrasonography, it is a well circumscribed hypoechoic mass. Cystic areas may be seen in large tumours. On CT, it is a homogeneous or heterogeneous attenuation mass with variable contrast enhancement. It shows intermediate signal intensity on T1-weighted and low signal intensity on T2-weighted MR images, and homogeneous or heterogeneous contrast enhancement.

Treatment for patients of MRKH syndrome with leiomyoma is myomectomy or hysterectomy and vaginal reconstruction (vaginoplasty). There are many types of vaginoplasty - Frank's dilatation method, Williams vaginoplasty, Vecchietti procedure, Abbe-McIndoe vaginoplasty and Devydov technique. With advances in minimal access surgery, the Vecchietti and Devydov procedures can now be performed laparoscopically. Psychological counselling is important to emphasize the fertility potential, though most patients may never become pregnant, but still they can become a mother with surrogate pregnancy.

In this case report, we described a case of MRKH syndrome with a large leiomyoma originating from a rudimentary uterus and role of imaging in diagnosis and planning of surgery.

## TEACHING POINT

Mayer-Rokitansky-Kuster-Hauser syndrome is a rare Mullerian duct anomaly and women with this syndrome present with primary amenorrhoea and can be diagnosed at puberty with normal secondary sexual characteristics and normal hormonal and genetic profiles. Leiomyoma in women with Mayer-Rokitansky-Kuster-Hauser syndrome is very rare and only few cases have been reported in the literature till date. Ultrasonography is the first modality to evaluate intra-abdominal masses and genitourinary system. CT and MRI are more accurate modalities to evaluate the anatomy and intra-abdominal masses.

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FIGURES

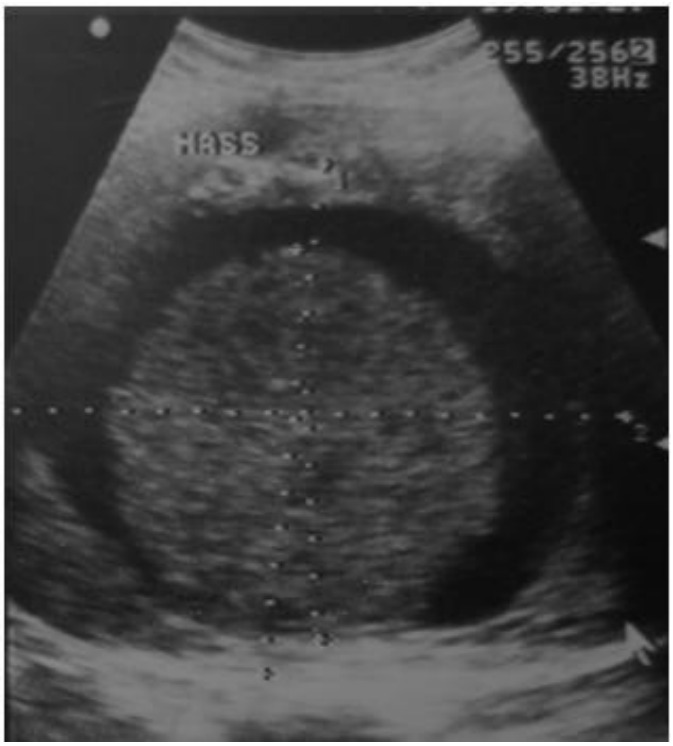



**Figure 1:** 35 year old woman with a large leiomyoma arising from rudimentary uterus in Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome. Ultrasound, transverse transabdominal image shows a large mass having a cystic area with internal echogenic contents in lower abdomen. (Hitachi Aloka ProSound 3500 SX, curved transducer, 3.5-5 Mhz)

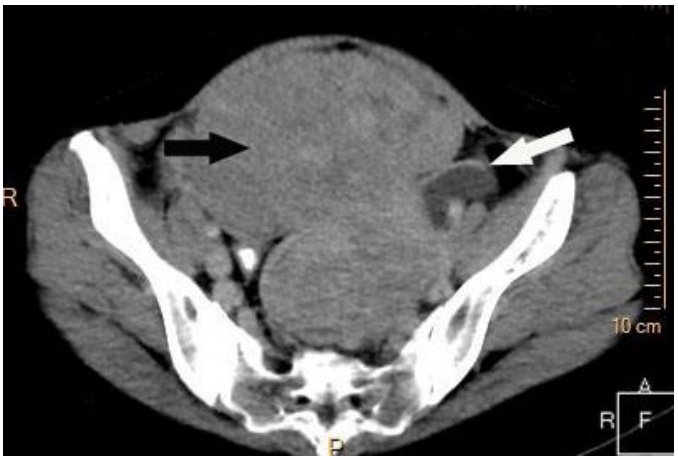



**Figure 2:** 35 year old woman with a large leiomyoma arising from rudimentary uterus in Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome. Axial contrast enhanced CT image of the abdomen in the venous phase showing a large mass (black arrow) measuring 17x11cm in size and a cyst (white arrow) measuring 6x4cm in size on the left side in pelvis. (Philips, Ingenuity, 128-slice CT, Philips Medical Systems, The Netherlands. Protocol: 160 mAs; 120 kv; slice thickness 1mm. 80 ml intravenous non-ionic contrast Optiray 300, Ioversol, Tyco Healthcare Canada Inc.).

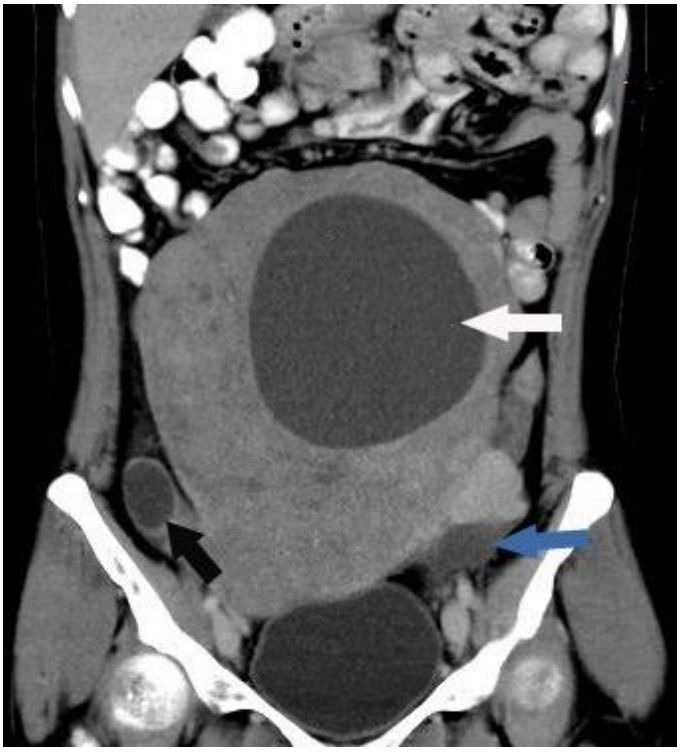


**Figure 3:** 35 year old woman with a large leiomyoma arising from rudimentary uterus in Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome. Coronal contrast enhanced CT image of the abdomen in venous phase reveals a large mass measuring 17x11cm in size in the lower abdomen. Mass shows central hypoattenuating area (white arrow) suggestive of cystic degeneration. Image also shows a small cyst measuring 3x2cm

in size in right ovary (black arrow) and a cyst (blue arrow) measuring 6x4cm in size on the left side of pelvis. (Philips, ingenuity, 128-slice CT, Philips Medical Systems, The Netherlands. Protocol: 160 mAs; 120 kv; slice thickness 1mm. 80 ml intravenous non-ionic contrast Optiray 300, Ioversol, Tyco Healthcare Canada Inc.).

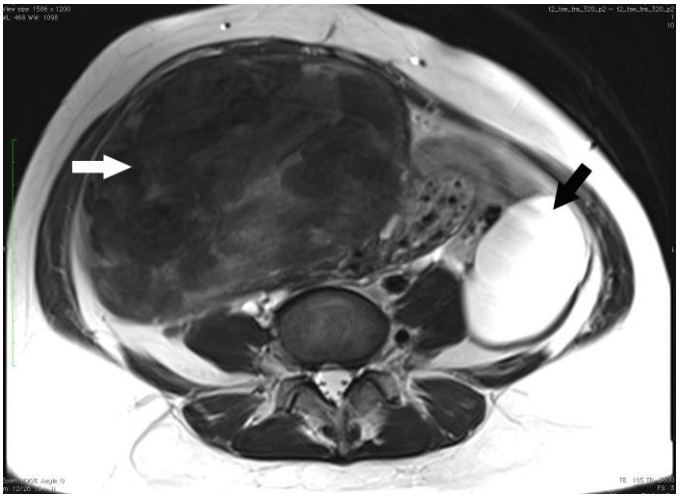


**Figure 4:** 35 year old woman with a large leiomyoma arising from rudimentary uterus in Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome. MRI T2-weighted axial image reveals a large low to intermediate signal intensity mass (white arrow) measuring 16x11cm in size and a cyst (black arrow) measuring 6x4cm in size in the pelvis. (3.0 T MRI Magnetom Verio, Siemens, Munich, Germany. Protocol: TR 3800 ms; TE 105; slice thickness 7mm. Non-contrast MRI).

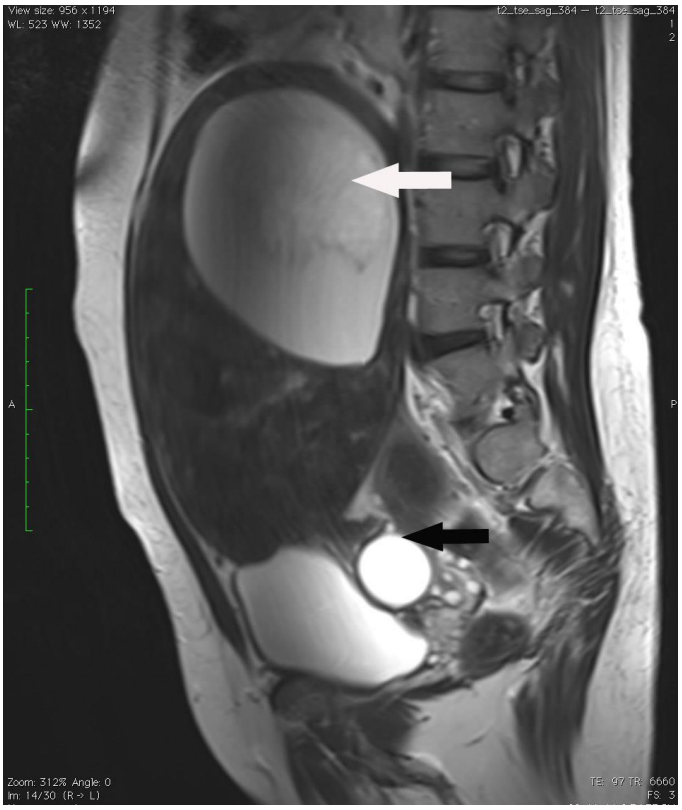


**Figure 5:** 35 year old woman with a large leiomyoma arising from rudimentary uterus in Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome. MRI T2-weighted sagittal image shows a large low to intermediate signal intensity mass measuring 16x11cm in size with a central hyperintense area (white arrow) suggestive of cystic degeneration in lower abdomen and a cyst measuring 3x2cm in size in right ovary (black arrow). (3.0 T MRI Magnetom Verio, Siemens, Munich, Germany. Protocol: TR 6660 ms; TE 97 ms, slice thickness 5mm. Non-contrast MRI).

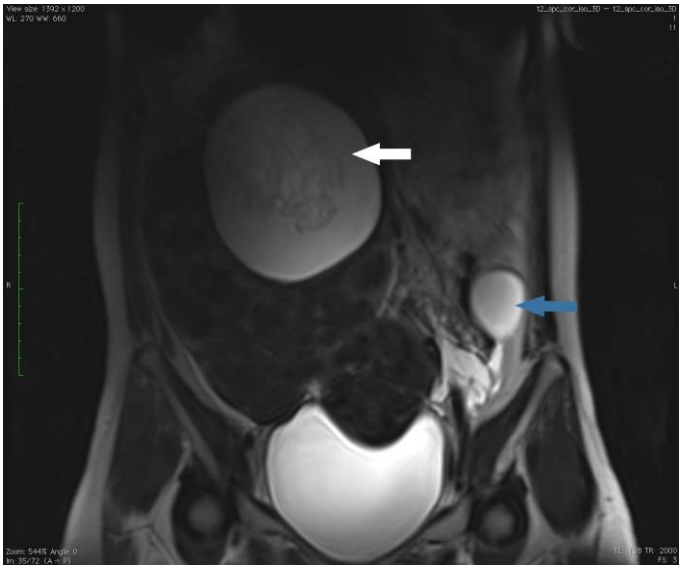


**Figure 6 (left):** 35 year old woman with a large leiomyoma arising from rudimentary uterus in Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome. MRI T2-weighted coronal image reveals a large low to intermediate signal intensity mass measuring 16x11cm in size with a hyperintense area suggestive of cystic degeneration (white arrow) and a hyperintense signal cyst measuring 6x4cm in size on the left side (blue arrow) in lower abdomen. (3.0 T MRI Magnetom Verio, Siemens, Munich, Germany. Protocol: TR 2000 ms; TE 126 ms; slice thickness 3mm. Non-contrast MRI).

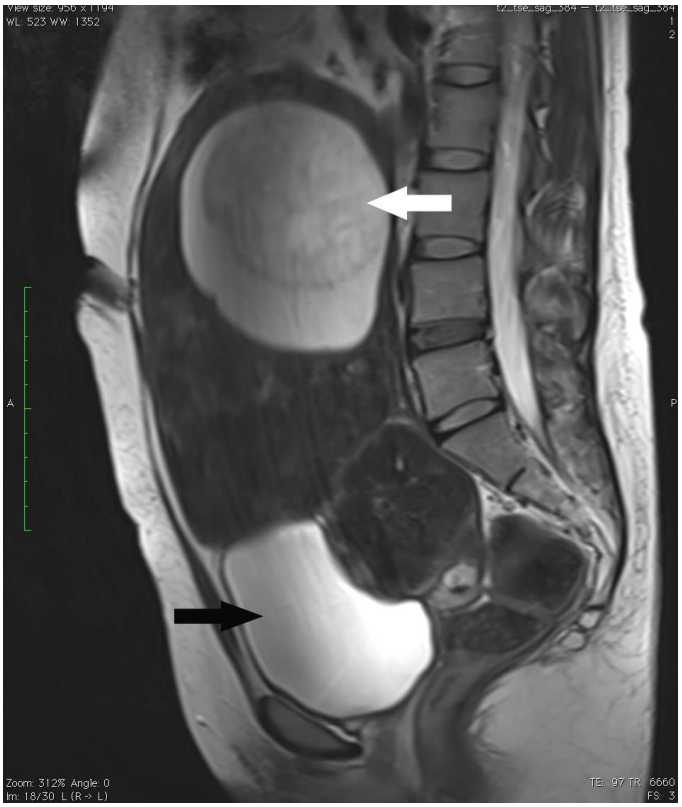
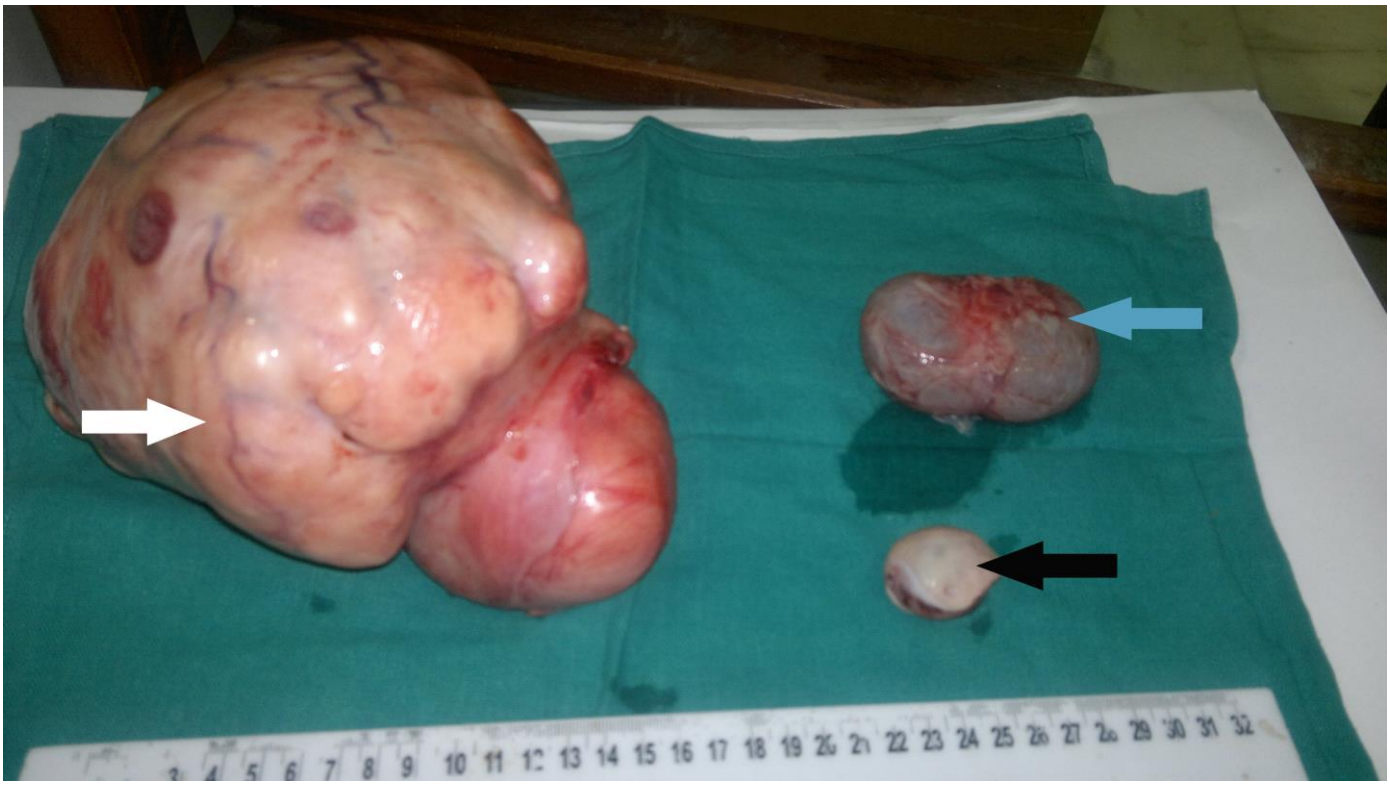



**Figure 7 (left):** 35 year old woman with a large leiomyoma arising from rudimentary uterus in Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome. MRI T2-weighted sagittal image reveals a large mass of low to intermediate signal intensity measuring 16x11cm in size with a hyperintense signal area suggestive of cystic degeneration (white arrow). Image also shows urinary bladder (black arrow) and absence of uterus and vagina. (3.0 T MRI Magnetom Verio, Siemens, Munich, Germany. Protocol: TR 6660 ms; TE 97 ms; slice thickness 5mm. Non-contrast MRI).

**Figure 8 (bottom):** 35 year old woman with a large leiomyoma arising from rudimentary uterus in Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome. Photograph of surgical specimen reveals a large leiomyoma (white arrow) measuring 25x18x12cm, a right ovarian cyst (black arrow) measuring 2x2cm in size and a left retroperitoneal cyst (blue arrow) measuring 6x4cm in size.



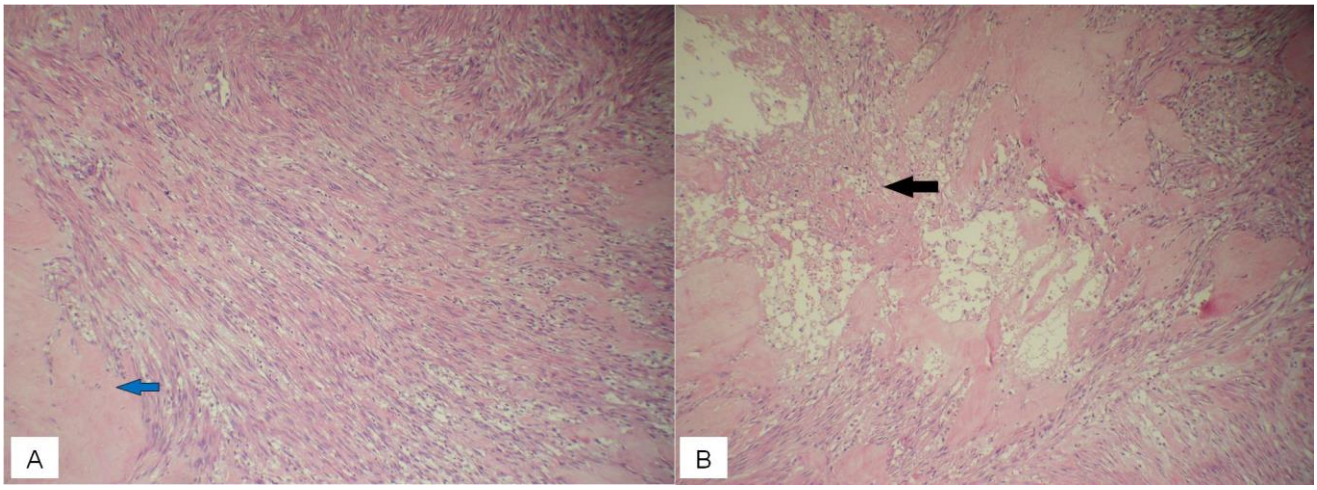



**Figure 9:** 35 year old woman with a large leiomyoma arising from rudimentary uterus in Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome. Microscopic images (haematoxylin-eosin, original magnification x 100) show tumour of smooth muscle cell origin, arranged in whorls. 9A. (haematoxylin-eosin, original magnification x 100) shows areas of hyalinization (blue arrow). 9B. (haematoxylin-eosin, original magnification x 100) shows smooth muscle cells with areas of necrosis (black arrow).





| | |
|---|---|
| **Class I** | Hypoplasia and agenesis<br>a) vaginal, (b) cervical, (c) fundal, (d) tubal, (e) combined |
| **Class II** | Unicornuate<br>(a) communicating, (b) noncommunicating, (c) no cavity, (d) no horn |
| **Class III** | Didelphys |
| **Class IV** | Bicornuate<br>(a) partial, (b) complete |
| **Class V** | Septate<br>(b) partial, (b) complete |
| **Class VI** | Arcuate |
| **Class VII** | Diethylstilbestrol (DES) drug related |

**Table 1:** American Fertility Society (AFS) classification of Mullerian duct anomalies

| | |
|---|---|
| **Etiology** | Etiology of Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is not known. Etiology of uterine leiomyoma is also not known but they are estrogen dependent. |
| **Incidence** | Incidence of Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is one in 4000-5000 female births. Incidence of leiomyoma in women with MRKH syndrome is very rare. |
| **Gender ratio** | Only females. |
| **Age predilection** | Adults. Leiomyomas normally begin to grow in 20s. |
| **Risk factors** | Genetic predisposition and endogenous and exogenous estrogen. |
| **Treatment** | Myomectomy or hysterectomy for leiomyoma and vaginal reconstruction for vaginal aplasia or hypoplasia. |
| **Prognosis** | Myomectomy and vaginal reconstruction can help the woman to live a normal life. However, her fertility will remain compromised for her entire life but she can become a mother with surrogate pregnancy. |
| **Findings on imaging** | ***USG***<br>• A large mass having cystic area with internal echogenic contents in lower abdomen.<br>• Uterus and ovaries could not be seen due large pelvic mass.<br>***CT***<br>• A large mass with cystic degeneration in pelvis, a cyst in right ovary and a cyst in left adnexal region.<br>• Left ovary is not seen separately.<br>***MRI***<br>• A large well circumscribed mass in the pelvis with low to intermediate signal intensity as compared to myometrium with hyperintense cystic area in T2-weighted images.<br>• Absence of uterus and vagina.<br>• Both ovaries are seen. A cyst in right ovary and another cyst adjacent to left ovary. |

**Table 2:** Summary table for leiomyoma of rudimentary uterus in Mayer-Rokitansky-Kuster-Hauser syndrome

| | USG | CT | MRI | Pattern of contrast enhancement |
|---|---|---|---|---|
| **Leiomyoma of rudimentary uterus in Mayer-Rokitansky-Kuster-Hauser syndrome** | -Well defined hypoechoic mass<br>-Large leiomyoma may show cystic degeneration | -Well defined heterogeneous mass<br>-Variable attenuation pattern may be seen<br>-Cystic areas are seen due to degeneration | -Well defined low to intermediate signal intensity mass<br>-Cystic areas are hyperintense on T2-weighted image | -Homogeneous or heterogeneous enhancement |
| **Ovarian fibroma** | -Well defined solid hypoechoic mass | -Well defined low attenuation mass | -Well defined low signal intensity mass on T1 and T2-weighted images | -Mild homogeneous enhancement |
| **Gastrointestinal stromal tumour(GIST) of intestine** | -Well defined heteroechoic mass<br>-Cystic areas may be seen | -Large heterogeneous exophytic mass<br>-Small mass can be homogeneous<br>-Cystic degeneration, necrosis or haemorrhage may be seen | -Low signal intensity mass on T1-weighted image<br>-Intermediate signal intensity mass on T2-weighted image | -Hypervascular tumour<br>-Moderate to marked enhancement |
| **Extravesical leiomyoma of urinary bladder** | -Well circumscribed hypoechoic mass<br>-Cystic areas may be seen in large mass | -Homogeneous or heterogeneous mass along the wall of urinary bladder<br>-Cystic areas may be seen | -Homogeneous intermediate signal intensity mass on T1-weighted image<br>-Low signal intensity mass on T2-weighted image<br>-Cystic areas are hyperintense on T2-weighted image | -Homogeneous or heterogeneous enhancement |

**Table 3:** Differential diagnosis table of leiomyoma of rudimentary uterus in Mayer-Rokitansky-Kuster-Hauser syndrome





## ABBREVIATIONS

CT - Computed Tomography
MD - Mesonephric ducts
MRI - Magnetic Resonance Imaging
MRKH syndrome - Mayer-Rokitansky-Kuster-Hauser syndrome
PMD - Paramesonephric ducts
UGS - Urogenital sinus
USG - Ultrsonography
UVP - Uterovaginal primordium

## KEYWORDS

MRKH syndrome; mullerian duct anomalies; leiomyoma; cystic degeneration

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