

CASE REPORT

Leiomyoma Arising from Mullerian Remnant, Mimicking Ovarian Tumor in a Woman with MRKH Syndrome and Unilateral Renal Agenesis

Wubishet Girma¹, Wondimagegnehu Woldeyes¹

ABSTRACT

BACKGROUND: *Leiomyoma with secondary changes arising from Mullerian remnant may mimic ovarian tumor in women with Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome in patients with pelvic mass and urologic abnormalities.*

CASE DETAIL: *The patient was a 40 years old known case of MRKH, presented with lower abdominal pain and swelling for over one year. On physical examination, large pelvic mass (about 15cm by 10cm) was found and a diagnosis of ovarian tumor was made by imaging which also showed absent right kidney. Finally, the mass was found to be a big leiomyoma with areas of hyaline, edematous and cystic degenerations.*

CONCLUSION: *Although leiomyoma arising from rudimentary Mullerian bulb is a rare condition, it should be considered in differential diagnosis of pelvic mass in patients with MRKH. Possibility of urologic abnormalities should also be considered in these patients and appropriate work up per local context should be made.*

KEYWORDS: *Leiomyoma, Mayer-Rokitansky-Kuster-Hauser syndrome, Mullerian remnant, renal agenesis*

DOI: <http://dx.doi.org/10.4314/ejhs.v25i4.14>

INTRODUCTION

Congenital anomalies of the Mullerian system are common defects, reported in up to 3.2% of all women (1). The prevalence of congenital uterine anomalies appears to be 6.7% in fertile population, and it is found in 7.3% of an infertile population (2). Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome, the most severe anomaly of Mullerian system, was first described by Mayer, Rokitansky, Kuster and Hauser (3). In MRKH syndrome, the vagina and uterus are congenitally absent, both ovaries are of normal size and fallopian tubes are normal; rudimentary uterine horns may be present in this syndrome (4-6). The etiologic factors of this syndrome are not fully understood, and environmental and genetic factors are thought to play a role (4-6).

Leiomyomas are a rather common occurrence in the normal uterus that can arise from remnant uterus. Occurrence of myoma arising from mullerian remnant is an extremely

rare finding and only few cases reported in literatures so far (1, 2, 6, & 7).

Mullerian aplasia can be an isolated finding although associated anomalies often coexist. The incidence of associated urologic abnormalities ranges between 15-40%, and skeletal anomalies such as congenital fusion or absence of vertebra occur in approximately 12-50% of cases (7-9). Unilateral renal anomalies are associated with 50% of the patients. The various urinary tract anomalies reported are renal agenesis, pelvic kidney, fusion anomaly like horse-shoe kidney and vesicoureteric reflux (5, 9).

Concurrent association of pelvic mass with mullerian agenesis can be a diagnostic dilemma. Here, we report a patient of MRKH syndrome with a large leiomyoma originating from the rudimentary uterus. This case is unique in that our patient as associated unilateral renal agenesis, and to the best of our knowledge there is no similar case report from Ethiopia so far.

¹Department of Obstetrics and Gynecology, Jimma University, Jimma, Ethiopia

Corresponding Author: Wubishet Girma, Email: gwubdz@yahoo.com

CASE REPORT

A 40 years old nulligravida presented to Jimma University Specialized Teaching Hospital, Gynecology OPD with a complaint of lower abdominal pain and swelling progressively worsening over the past 1 year. At 18 years of age, she was told to have no uterus when she presented for primary amenorrhea, failure to conceive, cyclical abdominal pain, breast pain and swelling, which were present till time of her current presentation. She divorced at 30. Her appetite remained normal, and there was no weight loss. She did not have symptoms related to the bladder or the bowel. She had neither excessive growth of

body hair nor a change in her voice. Her physical examination showed a female body contour and normal hair pattern. Her height was 165cm and she weighed 67kg. Thyroid was not enlarged. There was no galactorrhea. On abdominal examination, she was found to have a 15 x 10cm mass arising from the pelvis. The mass was firm, irregular, mobile, non-tender occupying most of the right iliac fossa and hypogastric region. There was no sign of fluid collection nor did she have hepatosplenomegally. On pelvic examination, the external genitalia was normal mature woman type. There was 3cm vaginal pouch with cervix absent and the mass was palpable on rectal examination.

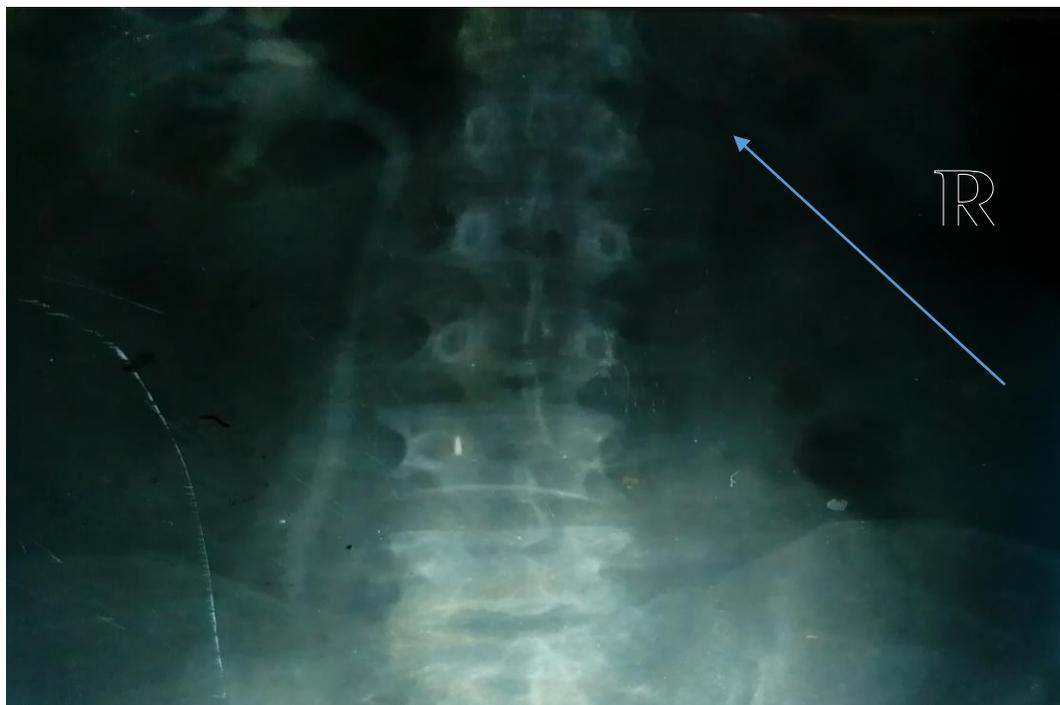


Figure 1: Intravenous pyelogram showing absent right kidney and ureter

Trans-abdominal ultrasonography revealed a large pelvic solid mass (9.5 x 13.5cm) having cystic areas with internal echogenic contents with irregular margins in the pelvis, suspicious of ovarian neoplasm. Uterus is not seen and the right kidney is also not visualized, with the left kidney being enlarged. Other intra-abdominal organs were normal and there was no ascites. Intravenous pyelogram (IVP) confirmed absent right nephrogram and pyelogram, and functional left kidney (Figure 1). Chromosomal investigation of our case indicated a normal karyotype of 46, XX.

With a provisional diagnosis of MRKH syndrome associated with ovarian tumor, she was taken up for laparotomy. On opening the abdomen through a midline vertical incision, a huge tumor occupying the whole pelvis, measuring 18 x 10 x 8 cm and was seen encroaching the right broad ligament ovary and right mullerian bulb. The left ovary (atrophic) and the left mullerian bulbs were seen with fimbriated end (Figure 2). There was minimal free fluid but no metastatic lesions seen. The whole tumor with the right ovary and mullerian bulbs was removed uneventfully.

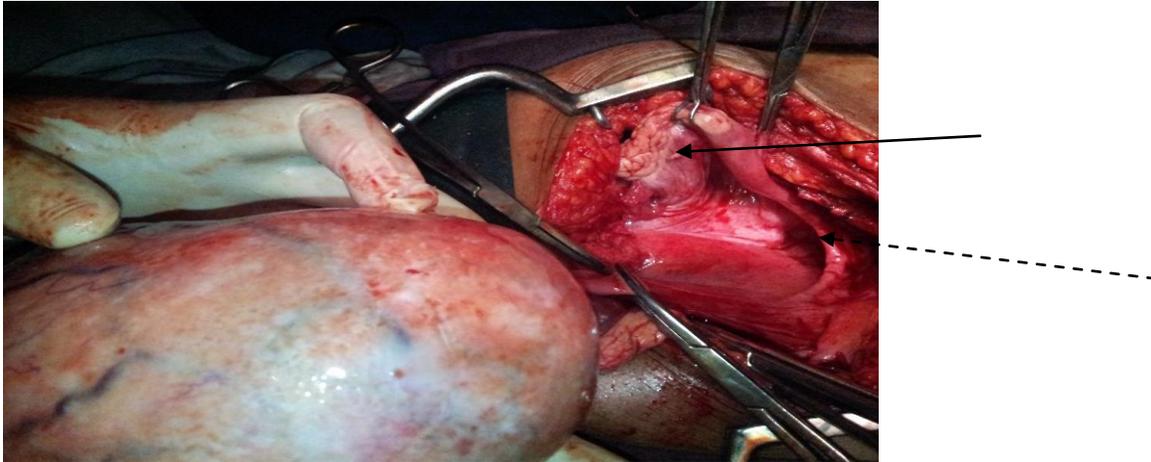


Figure 2: Laparotomy: right adnexal mass with the right ovary closely attached to it; solid arrow shows left ovary, and dashed arrow shows left sided uterine horn with absent uterus

The cut section of the mass showed grey-white whorly appearance with focus cystic degeneration and intact capsule. And, histopathology of the tumor showed subserosal myoma with foci of hyaline, edematous and cystic degenerations. The attached ovary revealed normal cortical and medullary tissues with large corpus albicanses.

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 and is typically diagnosed during puberty (4, 5). It is the second most common cause of primary amenorrhoea after gonadal dysgenesis (6).

Women with this syndrome are characterized by presence of 46 XX karyotype, normal female secondary sex characters, normal ovarian functions absent or undeveloped uterus and upper part of the vagina (4, 5, & 9). Failure of fusion and development of Mullerian ducts around 7th to 8th post-conception age results in muscular thickening at the proximal end of each tube that are joined in the midline by a visible and palpable cord resembling hypoplastic bicornuate uterus without an endometrial lining (4, 5, & 8).

The American Fertility Society's (AFS) classification, based on uterine anomalies, is most commonly used to classify Mullerian duct anomalies. 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 and II), diethylstilbestrol (DES)-related anomalies. MRKH syndrome is a class I Mullerian duct anomaly (5).

The extent of MRKH syndrome is variable, and it is associated with various additional malformations. This is reflected in the classification, which is subdivided depending on each additional malformation that is present into typical when tubes, ovaries, and renal system are generated and developed; atypical, when malformations in the ovary or renal system are present; and MURCS (Müllerian aplasia, renal aplasia, and cervicothoracic somite dysplasia) association, when malformations are in the skeleton and/or heart; muscular weakness, renal malformations (4) which puts our patient under classification of atypical type of MRKH syndrome. Rarely, an active endometrium can exist with uterine anlage, which becomes active in the presence of well estrogenised state (9).

Diagnosis of MRKH syndrome is often delayed until late puberty. The symptoms for presentation are amenorrhea, infertility and pelvic pain. These patients have the ovaries and fallopian tubes of normal functions and most of them have also two uterine remnants of different sizes. Incidence of leiomyoma of uterus is very high in the general female population. However, only few cases of leiomyoma have been reported in women with MRKH syndrome (1, 2). As ovarian function is normal, estrogen-dependent pathological

conditions can develop in the rudimentary uterus, including myomas, neoplasms and adenomyosis (9, 12).

The exact pathogenesis of neoplastic transformation of uterine smooth muscle in a patient with normal uterus is not known. Cytogenetic abnormalities in the form of spontaneous chromosomal rearrangements are known to occur in uterine leiomyomas. These chromosomal arrangements may be responsible for the initiation and progressive growth of the leiomyomas (1, 2, 6, 10,11).

As the proximal ends of Mullerian ducts have smooth muscles, the presence of myoma in a case of Mullerian agenesis is a theoretical possibility. However, occurrence of leiomyoma in a rudimentary uterine bulb has been rarely reported (1, 2, 6, 10, 11, & 12). The possible reason for this uncommon occurrence could be a decreased concentration or sensitivity of the estrogen receptors (11).

On ultrasound 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.

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 (6). Although myoma arising from a rudimentary uterine anlage is a rare finding, it should be considered in the differential diagnosis of pelvic mass in patients with MRKH syndrome.

About 30 to 50% of patients with Mullerian agenesis are associated with significant urologic abnormalities, including unilateral renal agenesis, unilateral or bilateral pelvic kidneys, horseshoe kidney, hydroureter, hydronephrosis and ureteral duplication (4, 5, 9). Our patient too had unilateral renal agenesis which confirmed by imaging studies and intra-operatively. Considering high association between Mullerian and urologic abnormality, possibility of urologic abnormalities should also be considered in these patients and appropriate workup per local context should be made.

REFERENCES

1. Elham Neisani Samani, Masoumeh Fallahian. Leiomyoma Arising From The Rudimentary

Uterus. A case report. *Iranian Journal of Pathology*, 2007;2(4): 187-189.

2. Salman S, Bozkurt M, Yumru AE, Bozyigit A, Kavsi B, et al. Laparoscopic Management of Leiomyoma Developing from Rudimentary Horn in Mayer- Rokitansky-Küster-Hauser Syndrome. *J Androl Gynaecol*, 2013; 1(2): 2.
3. Rokitansky K. Über die sogenannten Verdoppelungen des Uterus. *Med JB Ost Staates*. 1838;26:39-42.
4. Oppelt P, Renner SP, Kellermann A, Brucker S, Hauser GA, Ludwig KS, et al. Clinical aspects of Mayer-Rokitansky-Kuester-Hauser syndrome: recommendations for clinical diagnosis and staging. *Hum Reprod*, 2006; 21(3):792-7.
5. The American Fertility Society classification of adnexal adhesions , distal tubal occlusion, tubal occlusion secondary to tubal ligation, tubal pregnancies, mullerian anomalies and intrauterine adhesions. *Fertil Steril*, 1988; 49(6): 944-955. PMID: 3371491.
6. Rawat KS, Buxi T, Yadav A, Ghuman SS, Dhawan S. Large leiomyoma in a woman with Mayer-Rokitansky-Kuster-Hauser syndrome. *J Radiol Case Rep*, 2013;7: 39-46.
7. Deligeoroglou E, Kontoravdis A, Makrakis E, Christopoulos P, Kountouris A, et al. Development of leiomyomas on the uterine remnants of two women with Mayer- Rokitansky- Kuster- Hauser syndrome. *Fertil Steril*, 2004;81: 1385- 1387.
8. Morcel K, Guerrier D, Watrin T, Pellerin I, Leveque J. The Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome: clinical description and genetics. *J Gynecol Obstet Biol Reprod (Paris)*. 2008; 37(6); 539-546. PMID: 18723299.
9. Gupta N P, Ansari M S. Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome - a review. *Indian J Urol*, 2002; 18:111-6.
10. Singh S, Chakravarty B, Chakravarty M, Chakravarty A. Large fibroid arising from mullerian bulb mimicking ovarian tumor in a woman with MRKH. *Int J Infertility Fetal Med*, 2012; 3(1): 30-32.
11. Roy Kallol K. Lal Suman. Banerjee Neelam. Large leiomyomas in Mayer-Rokitansky-Küster-Hauser syndrome. *J Obstet Gynecol India*, 2005;55(2) : 183-184.
12. Edmonds DK. Multiple fibroids in a postmenopausal women with Mayer-Rokitansky-Kuster-Hauser syndrome. *J Pediatr Adolesc Gynecol*, 2003; 16(2): 65-66. PMID: 12742138.