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Confronting the unknown: Diagnosis of an ovarian tumor in Mayer–Rokitansky–Küster–Hauser type II: A rare case report

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Abstract:

Mayer–Rokitansky–Küster–Hauser (MRKH) syndrome is a rare congenital disorder characterized by the absence or underdevelopment of the uterus and upper part of the vagina in females with a normal 46, XX karyotype. It affects approximately 1 in 4500–5000 female live births and ranks as the second-most common cause of primary amenorrhea. This case report describes a 28-year-old nulligravid woman who presented with primary amenorrhea, difficulties during sexual intercourse manifesting as pain and resistance, and an incidental finding of a right ovarian new growth. Physical examination revealed normal secondary sexual characteristics and a blind-ending vagina measuring 5 cm in depth. Transvaginal ultrasound confirmed the presence of a transverse vaginal septum with hematocolpos, an infantile uterus with endometrium and cervix, a right ovarian new growth, and a normal left ovary. Both kidneys appeared normal, and hormonal assays were within normal limits. Karyotype analysis confirmed a genotype of 46, XX, indicating a normal chromosomal complement for a female without any detectable structural or numerical chromosomal abnormalities, consistent with typical female development. She subsequently underwent ultrasound-guided excision of the transverse vaginal septum combined with laparoscopic oophorocystectomy. Intraoperatively, findings included a normal left ovary, a right ovarian new growth, absence of fallopian tubes, and an infantile uterus. Histological analysis confirmed a serous cystadenoma in the right ovary. Karyotype analysis confirmed a genotype of 46, XX. The index case was diagnosed with MRKH type II (atypical), characterized by the absence of fallopian tubes and a right ovarian new growth without associated renal, skeletal, or cardiac anomalies.

Keywords:

Mayer–Rokitansky–Küster–Hause syndrome type II, ovarian tumor, primary amenorrhea, serous cystadenoma

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Introduction

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome affects approximately 1 out of 4500-5000 women.[1] It is a malformation of the female genitalia caused by interrupted embryonic development of the Müllerian (paramesonephric) ducts in otherwise chromosomally, phenotypically, and endocrinologically normal females. It ranks second to Turner's syndrome as a cause of primary amenorrhea. This type of Mullerian anomaly belongs to Class I of the classification of Mullerian Anomalies by the American Fertility Society and Class 5 based on the European Society of Human Reproduction and Embryology and European Society of Gynaecological Endoscopy.[2,3] The etiology of MRKH syndrome remains unknown and is typically diagnosed when evaluating individuals presenting with primary amenorrhea.

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MRKH syndrome is broadly subdivided into type I (typical), characterized by symmetric uterine remnants and normal fallopian tubes, and type II (atypical), which features asymmetric uterine buds, abnormally developed fallopian tubes, presence of adnexal pathologies, and other organ system anomalies.[4]

Recent publications on MRKH syndrome mainly report cases of uterine remnant leiomyoma or adenomyosis, whereas ovarian tumors are rare and difficult to diagnose. The possibility of ovarian tumors in MRKH patients should not be overlooked, as these patients typically have well-functioning ovaries. However, their ovaries are often positioned more cranially and laterally to the external iliac arteries, likely due to the absence of fallopian tubes. Ovarian anomalies occur in approximately 5%–10% of cases.[5]

Ovarian tumors associated with MRKH syndrome can be classified as serous, endometrioid, mucinous, clear cell, or mixed types. They may present as cystic (single or multicystic), solid cystic, or solid structures, with varying proportions of cystic and solid fibrotic tissue. The presence of ovarian tumors in MRKH patients complicates both diagnosis and treatment. Despite advancements in understanding MRKH syndrome, the incidence of MRKH with ovarian tumors is not well-documented. A literature review identified six case reports of MRKH syndrome with ovarian tumors, which were predominantly benign, and laparoscopy was commonly used for tumor removal.[5-7]

Case Report

A 28-year-old nulligravid Filipino woman, single, presented with complaints of primary amenorrhea, difficulties in sexual intercourse characterized by pain and resistance, and incidental findings of a right ovarian new growth. The patient had not undergone a gynecologic consultation previously. Reviewing her pubertal development, the onset of thelarche occurred at age 12 and pubarche at age 13. She did not report cyclic pelvic pain. At the age of 9, she underwent an exploratory laparotomy due to persistent abdominal distension, where she was diagnosed with pelvic tuberculosis. She completed 6 months of medical management with isoniazid, rifampicin, pyrazinamide, and ethambutol (HRZE) but was subsequently lost to follow-up. The patient has no other known medical conditions. There is no history of congenital anomalies among her family members. Her mother had an uneventful obstetric history without hormonal therapy or radiation exposure during pregnancy. The patient exhibited normal breast development and typical body hair distribution, including pubic and axillary hair. She measured 147 cm in height and weighed 46 kg, resulting in a normal body mass index of 21.2 kg/m^2. Breast and pubic hair development were consistent with Tanner Stage 5. Her extremities appeared grossly normal.

During abdominal examination, a vertical midline incision from a previous surgery was noted. A palpable, mobile, cystic, nontender mass in the right pelvoabdominal area was identified, measuring approximately 10 cm from the symphysis pubis. Pelvic examination revealed normal, well-estrogenized vulva, labia minora and majora, and clitoris. Speculum examination showed smooth vaginal mucosa ending in a blind pouch. Internal examination indicated a vaginal canal approximately 5 cm in length with no palpable cervix [Figure 1]. A smooth, cystic mass measuring around 10 cm was palpable in the right adnexal area. Rectovaginal examination demonstrated good sphincter tone, smooth rectal mucosa, pliable parametria, and no cul-de-sac fullness.

Transvaginal ultrasonography showed an infantile uterus measuring 3.3 cm × 4 cm × 1.7 cm, with a thin intact endometrium measuring 0.31 cm. The cervix measured 2.8 cm × 2 cm × 2.4 cm, and dilating the endocervical canal was a cystic area with ground glass echoes and sediments measuring 3.7 cm × 3.4 cm × 2.3 cm, suggestive of hematometra with a transverse vaginal septum measuring 0.13 cm [Figure 2]. Anterior to the uterus was a unilocular cystic structure with low-level echoes measuring 9.6 cm × 9.1 cm × 7.9 cm, with a volume of 369 mL, suggestive of an ovarian new growth with benign sonologic features. The left ovary was normal, measuring 2.6 cm × 2.8 cm × 1.9 cm, with a volume of 7.95 cm^3, and containing small follicles [Figure 3]. No cul-de-sac fluid was noted. There were no urinary tract anomalies on transabdominal ultrasonography. Blood routine and renal function tests were normal. The hormone profile included measurements of

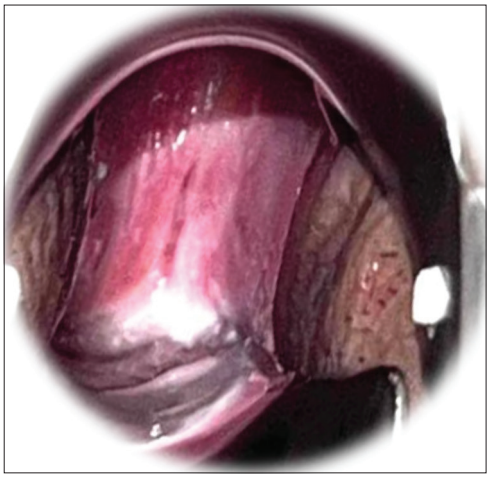

Figure 1: Speculum exam showed smooth vaginal mucosa, ending in a blind pouch with a depth of 5 cm, and no visible cervix

follicle-stimulating hormone, luteinizing hormone, estradiol, and prolactin, all of which were normal, indicating a normal hypothalamic–pituitary–ovarian axis. The chromosomal study indicated a normal (46, XX) female karyotype. The impression at that time was MRKH type II with a transverse vaginal septum and a pelvoabdominal mass, with a plan to proceed with ultrasound-guided drainage of the hematocolpos, excision of the vaginal septum, diagnostic hysteroscopy, and laparoscopic right oophorocystectomy.

Intraoperatively, an initial vaginal depth of 5 cm with a transverse vaginal septum was visualized and grasped using Allis forceps [Figure 4]. An ultrasound-guided excision of the transverse vaginal septum was performed [Figure 5]. Inferior to the external cervical os was a cystic structure with low-level echoes suggestive of a hematocolpos measuring 3.3 cm × 3.8 cm × 2.9 cm. An isoechoic structure between the vaginal probe and the hematocolpos, approximately 0.15 cm in thickness, was suggestive of a transverse vaginal septum. The excision of the transverse vaginal septum was done using Metzenbaum scissors and electrocautery [Figure 6]. Approximately 30 cc of nonfoul-smelling, whitish mucoid fluid was drained. Simple interrupted sutures were then made on the borders with the proximal and distal vaginal tissue [Figure 7].

A diagnostic hysteroscopy using a 5 mm, 30° rigid hysteroscope was attempted; however, due to the anteflexed uterus, we were not able to advance further than the endocervical canal. We then proceeded to laparoscopic surgery for a right oophorocystectomy. Upon inspection, filmy avascular omental adhesions were observed on the left upper quadrant. The liver parenchyma was covered in filmy avascular adhesions extending toward the anterior abdominal wall [Figure 8]. Occupying the pelvic cavity was the right ovarian new growth, measuring 10 cm, with a whitish-gray thin-walled cystic structure [Figure 9]. Upon inadvertent rupture, controlled decompression of the cyst contents was performed, revealing straw-colored serous fluid. An oophorocystectomy was then completed. The uterus was small, the left ovary was grossly normal, however, bilateral fallopian tubes were not visualized [Figure 10].

Histologic examination of the vaginal tissue showed blood vessel-rich fibromuscular tissue with nonspecific chronic inflammation. The right ovarian cyst wall showed a benign serous cyst with a fibrous cyst wall containing a few blood vessels. The tissues were lined by a single layer of cuboidal tubal-type epithelium with no significant atypia.

The postoperative course was unremarkable. The patient was advised to perform postoperative self-dilatation

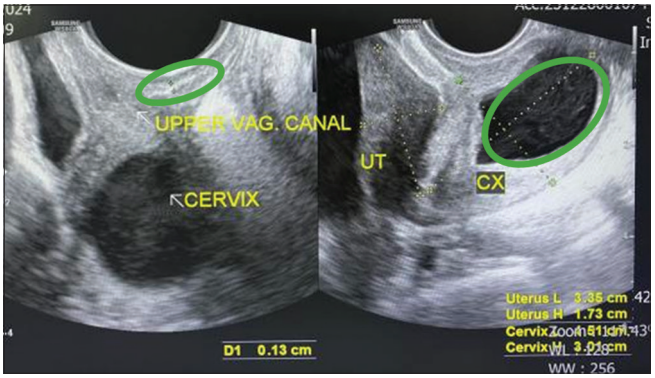

Figure 2: Transvaginal ultrasound showed dilating the endocervical canal was a cystic area with ground glass echoes and sediments measuring 3.7 cm × 3.4 cm × 2.3 cm, suggestive of hematometra with a transverse vaginal septum measuring 0.13 cm

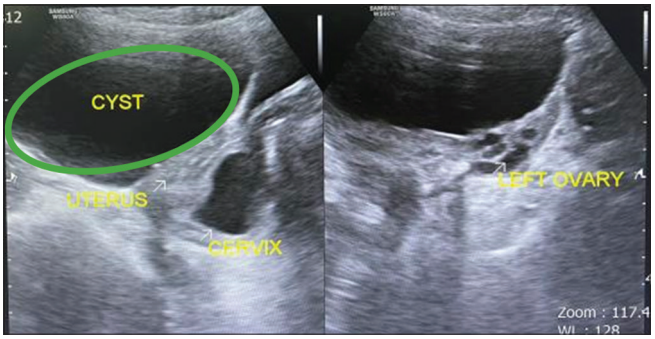

Figure 3: Transvaginal ultrasound showed unilocular cystic structure with low-level echoes measuring 9.6 cm × 9.1 cm × 7.9 cm, with a volume of 369 mL, suggestive of an ovarian new growth, benign sonologic features

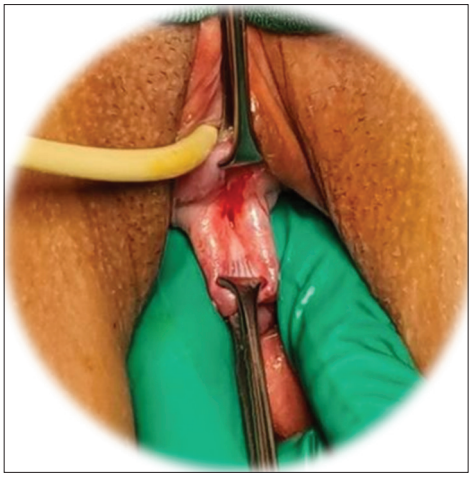

Figure 4: Intraoperatively: The transverse vaginal septum was grasped using Allis forceps

using a modified molded candle with a condom, which she was instructed to manually place at the vaginal apex for 10–30 min, 1–3 times daily, as she currently does not have a partner. The patient received counseling about her condition and reproductive potential. Chromosome analysis revealed a normal female karyotype of 46, XX, with no evidence of chromosomal abnormalities.

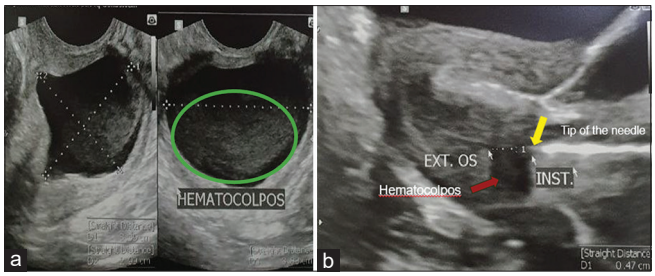

Figure 5: Ultrasound-Guided Excision of Transverse Vaginal Septum. Intraoperative ultrasound-guided excision of the transverse vaginal septum was performed. (a) Inferior to the external cervical os, a cystic structure with low-level echoes suggestive of a hematocolpos measuring 3.3 × 3.8 × 2.9 cm is visible. (b) The hematocolpos is indicated by the red arrow, while the hyperechoic structure, as indicated by the yellow arrow, represents the tip of the spinal needle aspirating the hematocolpos. Source: Based on the authors' original intraoperative findings

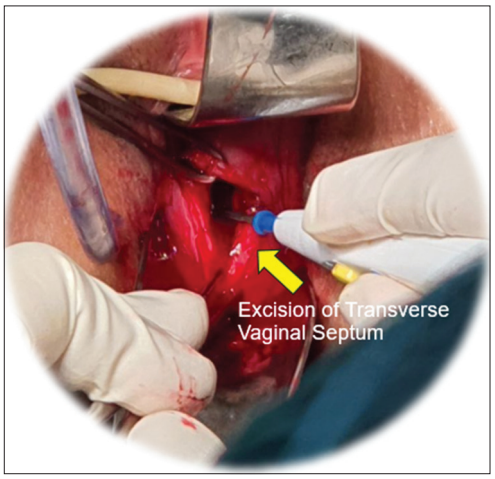

Figure 6: Transverse Vaginal Septum. As indicated by the yellow arrow, the transverse vaginal septum was excised using Metzenbaum scissors and electrocautery. Approximately 30 cc of non-foul-smelling, whitish mucoid fluid was drained. Source: Based on the authors' original intraoperative findings

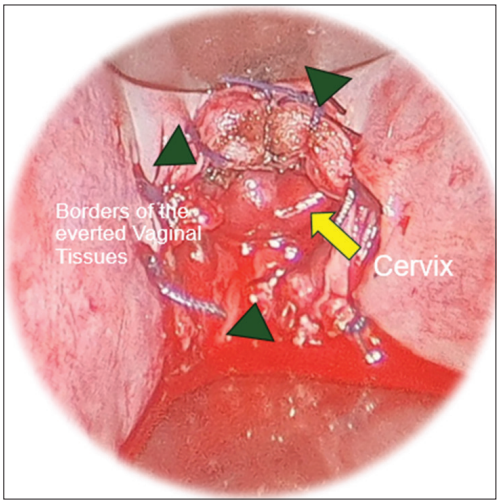

Figure 7: Placement of sutures. As indicated by the green arrowheads, a series of simple interrupted sutures were placed along the borders of the proximal and distal vaginal tissue. The cervix is marked by the yellow arrow. Source: Based on the authors' original intraoperative findings

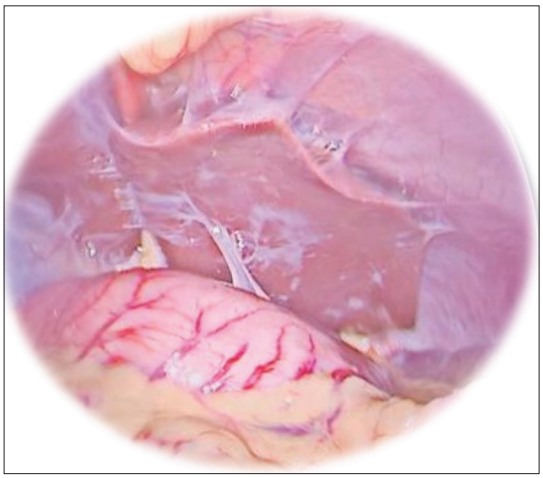

Figure 8: Liver parenchyma was covered in filmy avascular adhesions extending toward the anterior abdominal wall

Case Discussion

The MRKH syndrome is typically diagnosed when a patient presents with primary amenorrhea and inability to have sexual intercourse due to vaginal aplasia or hypoplasia. It is the second-most common cause of amenorrhea after gonadal dysgenesis in Turner's syndrome.[8] In the literature, MRKH syndrome is divided into two types:

1. Type A (Typical): Characterized by symmetric uterine remnants and normal fallopian tubes
2. Type B (Atypical): Characterized by asymmetric uterine buds, abnormally developed fallopian tubes, and other organ system anomalies.[9]

Another classification describes:

1. Type I (Isolated or Rokitansky sequence): No associated anomalies
2. Type II (Müllerian agenesis, Renal agenesis, and Cervicothoracic Somite anomalies [MURCS] association): Includes Mullerian duct aplasia, renal dysplasia, and cervical somite anomalies, often with additional urologic, vertebral, cardiac, or otological abnormalities.[10]

In their analysis of 521 cases, Oppelt *et al*. found 64% of patients with MRKH type 1, 24% with type 2, and 12% with MURCS syndrome.[11] Malformations in the ovaries and fallopian tubes are rare in patients with MRKH syndrome and can vary in their severity. Hypoplasia or aplasia of one or both fallopian tubes and ovarian anomalies have been described before in atypical MRKH syndrome.[12,13]

In MRKH syndrome, both ovaries are typically present and function normally, although they are often positioned higher and laterally to the external iliac arteries due to the lack of fallopian tube development. Ovarian anomalies are rare, occurring in about 5%–10% of cases. Ovarian tumors

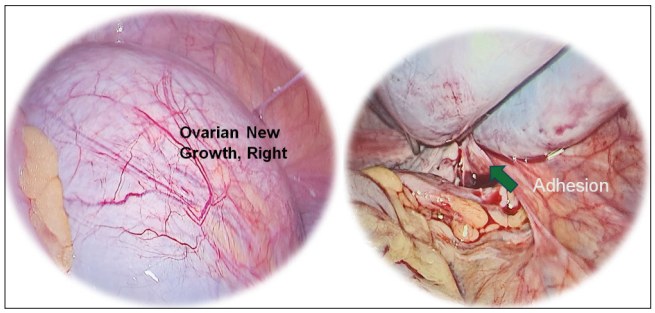

Figure 9: Right Ovarian New Growth and Adhesions. The pelvic cavity revealed a 10 cm right ovarian new growth with a whitish-gray, thin-walled cystic structure. The left image shows the mass, with adhesions marked by a green arrow

associated with MRKH can be serous, endometrioid, mucinous, clear cell, or mixed types, presenting as cystic or solid structures with varying proportions of cystic and solid fibrotic tissue. The presence of ovarian tumors complicates both diagnosis and treatment in MRKH patients. Despite improved understanding, the frequency of MRKH with ovarian tumors remains poorly documented. A review identified six cases of MRKH syndrome with ovarian tumors, mostly benign, treated commonly with laparoscopy. Differential diagnoses of MRKH syndrome combined with pelvic masses include those originating from the female genital tract (such as uterine fibroids, uterine adenomyosis, and ovarian tumors) and those from other pelvic organs (intestinal tract, mesentery, and retroperitoneal tumors). Careful selection of diagnostic methods is crucial for accurately diagnosing MRKH syndrome combined with pelvic masses.[5,11,14]

Epithelial neoplasms of the ovary account for 60% of all ovarian tumors and 40% of benign tumors. The two most frequent types of cystadenomas are serous and mucinous cystadenomas. Benign serous tumors of the ovary represent 16% of all ovarian epithelial neoplasms and account for two-thirds of benign ovarian epithelial tumors and the majority of serous ovarian tumors. They occur in adults of all ages.[15] They are bilateral in 10%–20% of the cases. Most serous cystadenomas are polyclonal, but monoclonal cystadenomas occur. They develop as a hyperplastic expansion from epithelial inclusions.[16]

The diagnosis is typically based on clinical information, laboratory results, and ultrasonographic findings. Among imaging techniques, ultrasonography is the first line of investigation due to its advantages of being easily available, low-cost, and providing straightforward information regarding the status of the uterus and ovaries, as well as allowing simultaneous assessment of the kidneys in the same sitting. If these findings are inconclusive, magnetic resonance imaging should be performed. However, in our case, ultrasound alone was sufficient to diagnose MRKH.

Laparoscopy is the ideal technique to identify and treat benign ovarian tumors, and it can also be effective

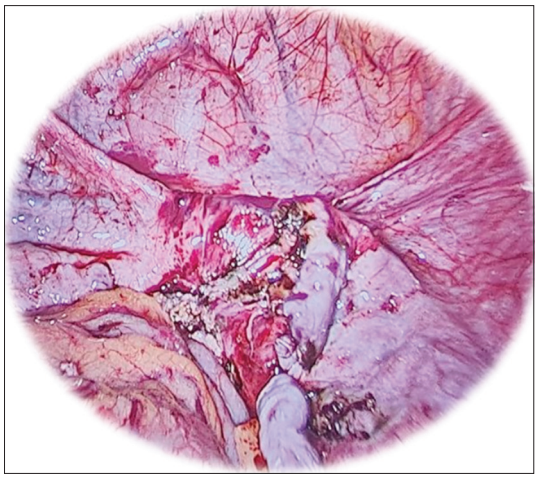

Figure 10: The uterus was small, the left ovary was grossly normal, however, bilateral fallopian tubes were not visualized

for treating such tumors in patients with MRKH syndrome. In our case, we treated a rare occurrence of a large ovarian serous cystadenoma in a young woman with MRKH syndrome using laparoscopic surgery. To the best of our knowledge, our case represents the sixth documented case of an ovarian tumor in MRKH syndrome being removed laparoscopically, confirming that laparoscopy is a powerful tool for both the diagnosis and treatment of these tumors.

Infertility remains the most significant problem in MRKH syndrome. Assisted fertility techniques, including surrogacy, enable women without a uterus to have genetic offspring.[17] This is why genetic characterization of the syndrome is of major importance to exclude other syndromes that clinically mimic MRKH but are not compatible with assisted reproduction due to the absence of normal gonads. Hence, chromosomal analysis was done in our case.

Our presented case is rare because it involves an atypical presentation of MRKH syndrome with a right ovarian serous cystadenoma, confirmed by histology, despite the absence of fallopian tubes. This combination of findings is uncommon and underscores the complexity of diagnosing and treating MRKH patients with concurrent ovarian tumors. The use of laparoscopy not only facilitated accurate diagnosis but also enabled effective surgical management of the ovarian tumor, highlighting its role as the gold standard technique in such cases.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will

be made to conceal their identity, but anonymity cannot be guaranteed.

Authorship contributions

Ma. Carmella Calvelo - Involved in the conceptualization, visualization, writing of the original draft, and review and editing.

Adonis Blateria - Involved in conceptualization, visualization, supervision, and review.

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Conflicts of interest

There are no conflicts of interest.

References

1. Guerrier D, Mouchel T, Pasquier L, Pellerin I. The Mayer-Rokitansky-Küster-Hauser syndrome (congenital absence of uterus and vagina) – Phenotypic manifestations and genetic approaches. J Negat Results Biomed 2006;5:1.
2. Fontana L, Gentilin B, Fedele L, Gervasini C, Miozzo M. Genetics of Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome. Clin Genet 2017;91:233-46.
3. Committee on Adolescent Health Care. ACOG committee opinion no. 728: Müllerian agenesis: Diagnosis, management, and treatment. Obstet Gynecol 2018;131:e35-42.
4. Dasgupta S, Mukhopadhyay P, Sharma PP, Begum N, Kalra A. Mayer-Rokitansky-Kuster-Hauser type-B anomaly with MURCS association and gonadal dysgenesis. J Obstet Gynaecol India 2012;62:83-4.
5. Miao Y, Wen J, Huang L, Wu J, Zhao Z. Diagnosis and management of ovarian tumor in Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome. Biomed Res Int 2018;2018:2369430.
6. Girma W, Woldeyes W. Leiomyoma arising from Mullerian remnant, mimicking ovarian tumor in a woman with MRKH syndrome and unilateral renal agenesis. Ethiop J Health Sci 2015;25:381-4.
7. Lanowska M, Favero G, Schneider A, Köhler C. Laparoscopy for differential diagnosis of a pelvic mass in a patient with Mayer-Rokitanski-Küster-Hauser (MRKH) syndrome. Fertil Steril 2009;91:931.e17-8.
8. Morcel K, Guerrier D, Watrin T, Pellerin I, Levêque J. The Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome: Clinical description and genetics. J Gynecol Obstet Biol Reprod (Paris) 2008;37:539-46.
9. Strübbe EH, Cremers CW, Willemsen WN, Rolland R, Thijn CJ. The Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome without and with associated features: Two separate entities? Clin Dysmorphol 1994;3:192-9.
10. Duncan PA, Shapiro LR, Stangel JJ, Klein RM, Addonizio JC. The MURCS association: Müllerian duct aplasia, renal aplasia, and cervicothoracic somite dysplasia. J Pediatr 1979;95:399-402.
11. 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;21:792-7.
12. Kumar A, Mishra S, Dogra PN. Management of an unusual case of atypical Mayer-Rokitansky-Kuster-Hauser syndrome, with unilateral gonadal agenesis, solitary ectopic pelvic kidney, and pelviureteric junction obstruction. Int Urogynecol J Pelvic Floor Dysfunct 2007;18:823-5.
13. Gorgojo JJ, Almodóvar F, López E, Donnay S. Gonadal agenesis 46, XX associated with the atypical form of Rokitansky syndrome. Fertil Steril 2002;77:185-7.
14. Herlin M, Bjørn AM, Rasmussen M, Trolle B, Petersen MB. Prevalence and patient characteristics of Mayer-Rokitansky-Küster-Hauser syndrome: A nationwide registry-based study. Hum Reprod 2016;31:2384-90.
15. Hunter SM, Anglesio MS, Sharma R, Gilks CB, Melnyk N, Chiew YE, *et al.* Copy number aberrations in benign serous ovarian tumors: A case for reclassification? Clin Cancer Res 2011;17:7273-82.
16. Seidman JD, Mehrotra A. Benign ovarian serous tumors: A re-evaluation and proposed reclassification of serous "cystadenomas" and "cystadenofibromas". Gynecol Oncol 2005;96:395-401.
17. Morcel K, Camborieux L, Programme de Recherches Sur Les Aplasies Müllériennes, Guerrier D. Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome. Orphanet J Rare Dis 2007;2:13.