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CASE REPORT

Mayer–Rokitansky–Kuster–Hauser syndrome (MRKH) and Ovarian Cyst : A Rare Case

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Abstract

Background: In the most recent publications on Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome, the uterine remnants and ovaries in patients may develop uterine remnant leiomyoma, adenomyosis, or ovarian tumor, and this can lead to problems in differential diagnosis. Here we summarize the diagnosis methods and available interventions for ovarian tumor in MRKH syndrome, with emphasis on the relevant clinical findings and illustrative relevant case. According to the clinical findings and illustrative relevant case, with the help of imaging techniques, ovarian tumors can be detected in the pelvis in patients with MRKH syndrome and evaluated in terms of size. Laparoscopy could further differentiate ovarian tumors into different pathological types. In addition, laparoscopic surgery not only is helpful for the diagnosis of MRKH combined ovarian tumor, but also has a good treatment role for excising ovarian tumor at the same time. Moreover, laparoscopic removals of ovarian tumor can be considered as a safe and reliable treatment for conservative management.

Case report: 25-year-old nulliparous woman with female phenotype and normal secondary sex development presented with complaints of primary amenorrhea and difficulty in sexual intercourse. Physical examination revealed that there were no vaginal and uterus structure. A transvaginal ultrasound examination showed absent of uterine . normal right ovaries and cystic mass in left adnexa . Laparoscopy cystectomy and amniograft vaginoplasty was performed in this patient. Histopatology result showed Cystadenoma srous ovarii with borderline serous ovarii.

Conclusion: Rarely ovarian cyst was associated with MRKH . Although malformation are associated with MRKH syndrome. Association beetween MRKH syndrome and ovarian cyst can be found

Keywords: *Mayer – Rokitansky – Kuster – Hauser syndrome, primary amenorrhea, ovarian*

INTRODUCTION

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The Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is characterized by congenital hypoplasia of the uterus and the upper part of the vagina. The incidence of MRKH syndrome has been estimated as 1 in 4500 women [1]. The principal character is a primary amenorrhea in women presenting with normal development of secondary sexual characteristics and normal external genitalia, but congenital vaginal or a shallow concave nest in the vaginal mouth, congenital uterine, or uterus aplasia. The ovaries are normal and functional as well as the endocrine status. Karyotype is 46, XX, with no visible chromosome modification. At present, most of the studies suggest that MRKH syndrome has been considered as a genetic disease, and genes such as the HOXA7, HOXA9–13, HOXD9–13, and WNT4 have been considered as possible offenders 2.

The MRKH classification is divided into 3 types below:

- Type I (isolated): the presence of isolated uterovaginal aplasia
- Type II: asymmetric uterovaginal aplasia or hypoplasia, with hypoplasia or absence of one or both fallopian tubes and presence of ovarian and/or renal system malformations.
- Type III or MURCS (Mullerian duct aplasia, Renal dysplasia, and Cervical Somite Anomalies), the most commonly associated anomalies are urological (15-40% of cases) and skeletal anomalies (20-40%) while hearing, cardiac, and digital defects (syndactyly or polydactyly) is rare.[1,3]

In the most recent publications on MRKH syndrome, we could found some cases report about uterine remnant leiomyoma, or adenomyosis, but the ovarian tumor is rare in MRKH syndrome and is difficult to be diagnosed [3–8]. Although most publications about pelvic masses in MRKH are about uterine remnants, adenomyosis, or fibroids, the occurrence of ovarian tumors in MRKH could not be ignored as these patients do have ovaries.

Diagnosis can be made using ultrasound, MRI, laparoscopy, and pyelography. Ultrasound is a simple and non-invasive method and should be the first investigation in evaluating a patient with suspected Mullerian aplasia. This technique reveals the absence of uterine structures between the bladder and rectum. Magnetic resonance imaging (MRI) is a non-invasive technique that is the gold standard in these cases because it produces excellent images of the superficial and deep tissues and can clarify inconclusive ultrasound results regarding uterine cavitation. MRI should be performed when the ultrasound findings are inconclusive or incomplete, as it allows accurate evaluation of uterine aplasia, as well as clear visualization of the rudimentary tubes and ovaries. Uterine aplasia is best characterized on sagittal images, whereas vaginal aplasia is best demonstrated on transverse images. In addition, MRI can be used at the same time to look for malformations of the kidney and associated bones.[3,7,11]

Laparoscopy is an invasive procedure that requires hospitalization and anesthesia. It can be performed in case of a dubious diagnosis after ultrasonography and/or MRI. Laparoscopy is primarily performed for women who are likely to undergo interventional therapy (neo-vaginal construction) to determine the exact anatomic location and abnormality of the uterus, possible fallopian tubes, vestigial lamina, and ovaries. [3,7,11]

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The sexuality of women with MRKH syndrome is also a concern. Many of them prefer anal or oral sex. Thus, most of the medical therapy for MRKH syndrome has focused on surgical and non-surgical techniques to enhance sexual intercourse through the creation of a neovagina. With the increasing number of MRKH patients, there has also been increasing interest in the clinical aspects of patients with MRKH .[9,10]

Laparoscopy is the ideal technique to identify and treat ovarian benign tumor, so it may also be able to treat the ovarian benign tumor in MRKH syndrome. From our illustrative relevant case, we treated a rare case of large ovarian serous papillary cystadenofibroma in a young woman with the MRKH syndrome with laparoscopic surgery. After 6 months of postoperative follow-up, the patients recovered well. To the best of our knowledge, our illustrative relevant case describes the fifth case in which ovarian tumor in MRKH syndrome was removed under laparoscopy confirming that laparoscopy is a powerful tool for treatment as well as diagnosis of these tumor. In addition, cytoreductive surgery and oophorectomy are further needed to treat the ovarian malignant tumor in MRKH syndrome.

CASE REPORT

1. Patien Information

A 25-year-old nulliparous woman presented the gynecology clinic with primary amenorrhea and difficulty in sexual intercourse. Sh has been married for 4 years and has never been pregnant. A review of her pubertal development revealed onset of thelarche at age 10 and pubarche at age of 13. She did not report having cyclic pelvic pain . The patient had no history of taking hormonal drugs and no history of previous gynecological surgery.The patien second child of three sibling. Additionally, There was no history of congenital aomalies among the family members. Her mother’s obstetric history was unremarkable, with no exposure to hormonal therapy or radiation while pregnant and a full term vaginal delivery.

2. Physicel Examination

On physical examination, vital signs were within normal limits, with a normal female body appearance. The patient's BMI of [24.34]. Head and neck examination did not reveal webbed neck or facial malformations. Physical examination of the chest, heart, and abdomen was also normal.

There were no major abnormalities of the extremiti, Breasts were well developed (Tanner 4 M) and had external genitalia. Female, clitoris, labia minor and labia majora appeared normal. Distribution of pubic hair was also Tanner 4.

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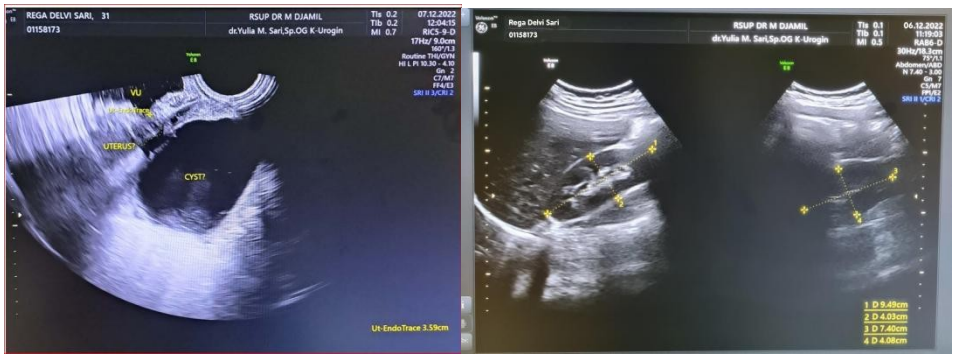

Figure 1 Transvaginal ultrasound: there appears to be a mass in the right adnexa, suggestive of a cyst of the right ovary and both kidneys are in normal size

Genitalia examination revealed only 2 cm frank. Vaginal toucher can not be performed . Rectal Toucher showed uterine band with cystic mass in left adnexa.

3. Diagnostic assesment

Transvaginal ultrasound examination revealed absent of the uterus. However, on the right adnexa, there was an ovary while on the left adnexa, there was cystic mass suggesting a left ovarian cystic. On transabdominal ultrasound examination, revealed both kidneys were normal

4. Theraupetic Intervention

The patient, a diagnostic laparoscopy was performed, a cystic mass appeared on the right adnexa, suggesting a right ovarian cyst, and on the left adnexa, an ovarian cyst with normal size with an intact tube was seen, but no uterus was seen. From the examinations performed, the diagnosis of MRKH syndrome type I was made. The patient underwent a laparoscopic cystectomy and vaginoplasty was performed with shears method modified wit amniograft.

5. Followup Outcome

Histopatology result of the cyst is Cystadenoma serous ovarii with borderline serous ovarii the patient was consulted to a gynecological oncologist, and the results found no abnormalities and the patient was advised to have a routine ultrasound every 1 month A\and after which those patients were advised intermittent self-dilatation till active sexual function was established.The patient was discharge with satisfactory condition.

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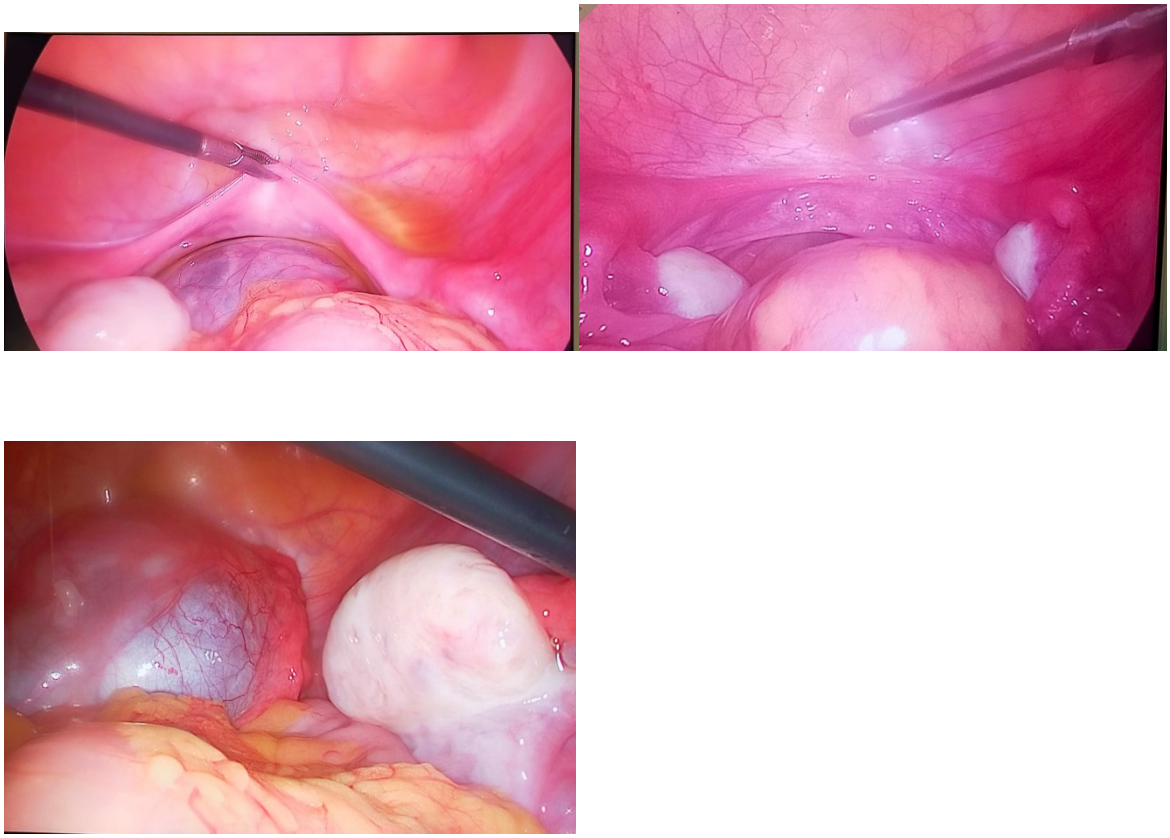

Figure 2 diagnostic laparoscopy (a) uterine band (b) Right and left ovary and tube, (c) left ovary and left ovarian cyst

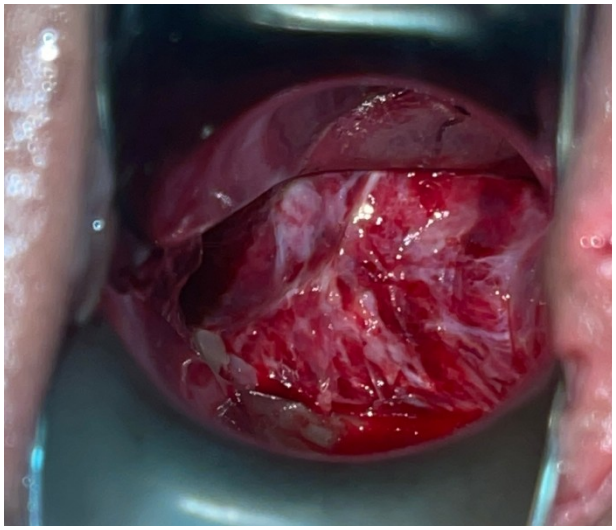

Figure 3. After neovaginaplasty

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DISCUSSION

The prevalence of MRKH syndrome is estimated at 1 per 4,000 female live births in the Caucasians. In addition, the etiology of MRKH syndrome remains unknown.2,7 The majority of cases appear to be sporadic, however, reports of familial cases of MRKH syndrome suggest a genetic component.8 In our patient, we found no family history of this pathology whose prevalence is unknown, and which remains a mystery in our country

The MRKH syndrome is a devastating diagnosis for a young woman to receive, which has considerable medical, psychological, social and reproductive implications. The diagnosis is often made in adolescence in the context of primary amenorrhea with normal puberty.[4]This condition, often suspected in the face of primary amenorrhoea, is usually confirmed radiologically (ultrasound or MRI) or by laparoscopy in patients for whom hormonal examinations and karyotype are normal.[9] The MRKH syndrome was subdivided into 2 types: type I, with malformations of the Müller's canal which are in the form of a superficial vaginal dimple with uterine cervix, uterus and upper vagina absent and is not associated with other abnormalities. Type II with Müller's canal agenesia similar to type I and various degrees of associated congenital renal malformations (renal agenesia and horseshoe kidney), skeletal abnormalities (scoliosis, spina bifida) and in rare cases of unilateral hearing defects, cardiac and extremities abnormalities. [10] In our case, the patient was only 27 years old, and had not yet seen her menarche, which was not a concern for parents.. It was a type I. In the most recent publications on MRKH syndrome, we could find cases reported on leiomyoma, adenomyosis, but ovarian tumor is rare in MRKH syndrome and difficult to diagnose if it is not bulky.[11-14] The incidence of the combination of MRKH with ovarian tumors was not reported. However, a review of the literature demonstrated nearly a dozen reported cases of MRKH syndrome associated with ovarian tumors.Benign tumors were predominant in most studies. This is the case in our observation, or it was a left ovarian benign.

Laparoscopy is the gold standard for the diagnosis and management of benign ovarian tumors. In the literature, in five cases described, associating an ovarian tumor with MRKH syndrome, management was performed under laparoscopy, which confirms the interest of this approach. 15 In addition, conventional surgery by laparotomy is possible, especially in case of large solid or mixed tumor as was the case in our observation. In addition, the patient's care remains holistic, taking into account the psychosocial aspects, vaginoplasty to improve sexual practice and the possibility of medical assistance.

This patient was offered to perform neovaginal formation because one of the patient's complaints was difficulty in sexual intercourse. The goal is to provide sexual satisfaction.

Management of neovagina creation, through surgical and non-surgical methods. The nonsurgical technique consists of dilating the vagina with an appropriate dilator. Also, through proper stretching exercises, it is possible to form a functioning vagina, even from a tiny opening. The most well-known modality is the Frank method, which consists of placing a special dilator into the vestibule area of the vagina. Another well-known method of increasing patient comfort was devised by Ingram et al, who constructed a stool with dilators that fit on the chair.[3,6,12] The most popular surgical techniques for vaginoplasty practiced in Europe today are the Vecchietti and Davydov procedures.[1]

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Infertility is also a common problem for patients, including this patient. Patients with MRKH syndrome have well-functioning ovaries and a normal hypothalamic-pituitary-ovarian axis. From a hormonal and physiological point of view, MRKH patients are capable of reproduction and may have genetic offspring but are unable to bear children on their own. The options for these patients are adoption or hiring a surrogate mother. However, in many countries, including Indonesia, surrogacy is not allowed for ethical, legal or religious reasons. Uterine transplantation has been developed as one of the infertility therapies in MRKH patients.[6]

CONCLUSION

MRKHS syndrome is a rare condition. The association with a voluminous ovarian tumor is even rarer, if not exceptional. The available data and knowledge on the etiopathogenic factors being still limited, it is impossible to define a supposed correlation between these two

Affections Laparoscopy is the gold standard for the diagnosis and management of benign ovarian tumors. In the literature, in five cases described, associating an ovarian tumor with MRKH syndrome, management ovarian cyst was performed under laparoscopy and agenesis vagina was performed with the Sheares' method is suitable for a flat perineum with no pouch.The Sheares' method of vaginoplasty is an easy and safe method to create a neovagina with least complications, like injury to urinary bladder, rectum or bleeding.

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