A Rare Case of Serous Cystadenofibroma in a Patient with Mayer-Rokitnsky-Kuster-Hauser Syndrome

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Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is characterized by failure of embryologic growth of the mullerian ducts resulting to agenesis or hypoplasia of the uterus and upper part of the vagina while both ovaries and fallopian tubes are normal. Various associated malformations with MRKH syndrome are noted in literature, with a renal system anomaly as the most common. However, adnexal tumors in MRKH syndrome are rare. To date there have been no reported cases of fallopian tube tumors in patients diagnosed with MRKH. This paper presents a case of an 18 year old nulligravida who presented with primary amenorrhea secondary to MRKH syndrome, with an associated Papillary Serous Cystadenofibroma of the right fallopian tube. Management of the case as well as review of related literature are presented.

Keywords: Mayer-Rokitansky-Küster-Hauser, serous cystadenofibroma

Introduction

Mullerian agenesis, also known as Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome, has an incidence of 1 per 4500-5000 females.[1] This syndrome is characterized by failure of embryologic growth of the mullerian ducts resulting to agenesis or hypoplasia of the uterus and upper part of the vagina while both ovaries and fallopian tubes are normal.[2] Women with MRKH syndrome typically have normal ovarian function and a 46,XX karyotype; thus, these patients typically present with primary amenorrhea. The association of malformations in mullerian duct development with other organ systems suggests that crucial genes involved in fetal development and sex differentiation are potential candidates for these congenital malformations. However, the etiology of MRKH syndrome remains unknown.[3]

MRKH syndrome is classically divided into 2 types: type I (isolated) or Rokitansky syndrome, and type II (associated with malformations of organs of the renal, skeletal, cardiovascular, and other systems). A more recent and comprehensive classification is through the use of "vagina-cervix-uterus-adnexa-associated malformation" (VCUAM) genital classification system. Developed in 2005, the VCUAM system provides an accurate description of the phenotypes of female genital malformations.

Various associated malformations with MRKH syndrome are noted in literature, with a renal system anomaly as the most common. Rudimentary uterine horns may be present in MRKH. Cases of uterine remnants complicated by the presence of leiomyoma or adenomyosis have also been reported.[4] However, adnexal tumors in MRKH syndrome are rare. So far, only 6 cases of ovarian tumors in MRKH have been reported. To date, there have been no reported cases of fallopian tube tumors in patients diagnosed with MRKH.

This paper presents a case of an 18 year old nulligravida who presented with primary amenorrhea secondary to MRKH syndrome, with an associated Papillary Serous Cystadenofibroma of the right fallopian tube. Management of the case as well as review of related literature are presented.

The Case

The patient is an 18 years old nulligravid who consulted with a chief complaint of primary

amenorrhea. She did not present with cyclic pelvic pain. There was no family history of menstrual disorder or congenital malformations. The patient has one female sibling who had her menarche at 11 years old. The patient denies any sexual contact.

On physical examination, the patient had stable vital signs and BMI of 30. She had normal secondary sexual characteristics (Tanner staging 4 and 5 for breast and pubis). She was not pale and had pink palpebral conjunctivae. She had a soft flabby abdomen with no palpable mass or tenderness. Pelvic examination revealed normal appearing external genitalia and presence of what seemed to be a hymen. As the patient was a virgin, a bimanual rectal examination was done revealing the presence of a midline, cystic, movable mass measuring approximately 12 cm in widest diameter.

A transrectal ultrasound revealed a right pelvic cyst probably paratubal cyst measuring 14.83cm x 10.06cm x 8.88cm, an infantile uterus and normal ovaries. Ultrasound of the kidney and urinary bladder revealed presence of normal bilateral kidneys and bladder.

Initial working impression was Primary amenorrhea secondary to Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome; Paratubal cyst, right; Obese class I.

The patient underwent exploratory laparotomy with right salpingectomy. Intraoperatively, the right fallopian tube was converted to a unilocular thin-walled cystic mass measuring 11.0cm x 17.0cm x 6.0cm with smooth outer capsule measuring 0.2cm and clear fluid within. The inner capsule was smooth with no solid areas but with papillary excrescences. (Figures 1 & 2).

A uterine remnant measuring about 2cm in widest diameter attached to the broad ligament was noted on each side underneath each ovary. (Figure 3) The left adnexa and the right ovary were grossly normal.(Figure 4)

The patient had an uneventful postoperative recovery and was allowed to go home after 3 days. Histopathologic findings revealed Papillary Serous Cystadenofibroma, right.

Final impression was Primary amenorrhea secondary to Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome; Papillary Serous Cystadenofibroma, right fallopian tube; Obese class I.

Discussion

This is a case of 18 year old nulligravid patient who presented with primary amenorrhea and a

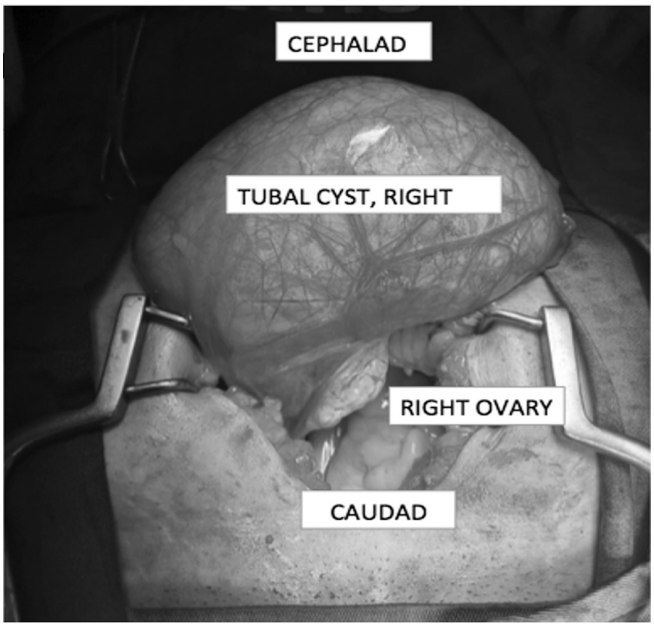

Figure 1. Intraoperatively, the right fallopian tube was converted to a unilocular thin-walled cystic mass measuring 11.0cm x 17.0cm x 6.0cm with smooth outer capsule.

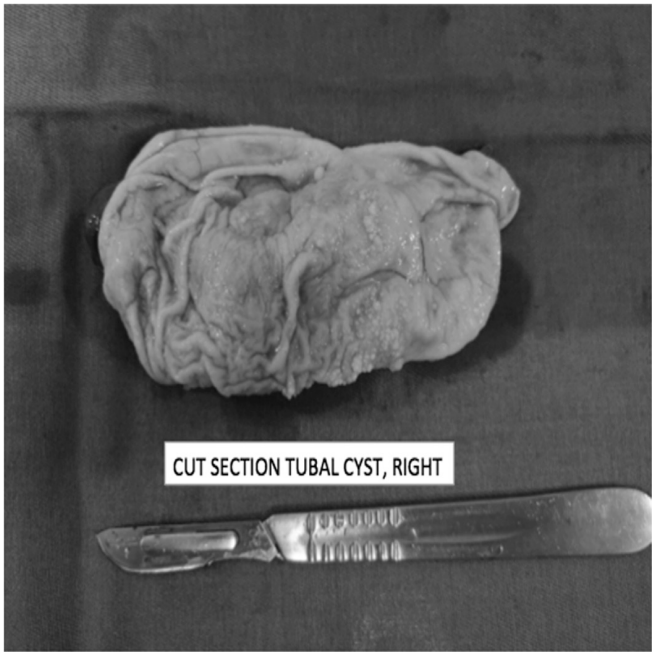

Figure 2. The inner capsule was smooth with no solid areas but with papillary excrescences. Capsule thickness measured 0.2cm and with clear fluid within.

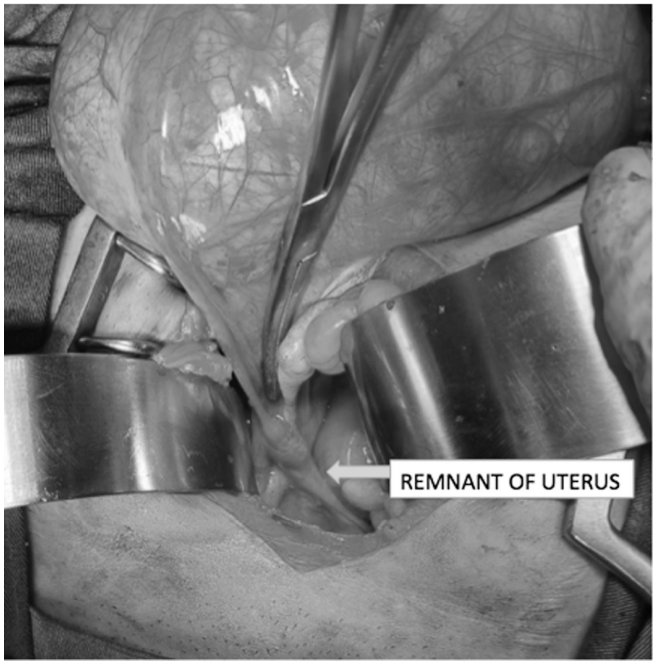

Figure 3. A uterine remnant measuring about 2cm in widest diameter attached to the broad ligament was noted on each side underneath each ovary

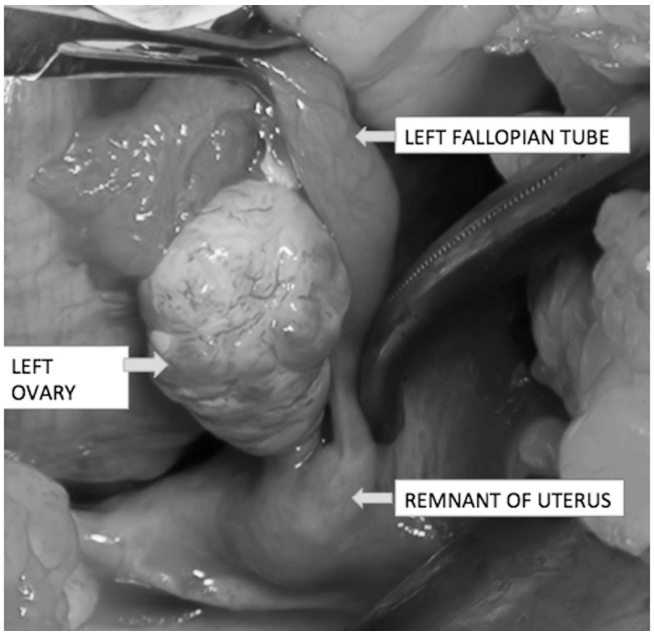

Figure 4. The left adnexa and the right ovary were grossly normal

huge pelvic mass. Preoperatively the pelvic mass was thought to be a paratubal cyst and the patient underwent right salpingectomy. However final histopathologic report revealed it is a rare case of papillary serous cystadenofibroma of the fallopian tube.

MRKH is the most severe anomaly of the Mullerian system characterized by atresia of the upper vagina, uterus or both. The etiology of MRKH is not fully understood. Recently, research has shown environmental and genetic factors play a role in the development of the syndrome. Patients often present with primary amenorrhea, normal development of female secondary sexual characteristics, normal external genitalia, as seen in the patient, and normal female karyotype of 46 XX. As ovarian function is normal, estrogen dependent conditions such as leiomyoma and adenomyosis may occur in patients with MRKH.[2,4,5,6,7]

In 2005, Oppelt and colleagues developed the VCUAM classification system to describe mullerian malformations. This classification is very similar to the system used for oncologic tumors in the TNM classification. The aim of the classification is to provide a description of malformations that is both as individual and as precise as possible, reproducible and clinically practicable. This comprehensive and precise structural assignment of the anomaly concerned, it is possible to obtain a descriptive picture at any time of the extent of the condition and its associated malformations.

An associated adnexal tumor is quite rare in MRKH syndrome and often difficult to diagnose. As of 2018, only 6 cases of MRKH syndrome associated with ovarian tumors have been reported.[4] To the best of the authors' knowledge, no case of fallopian tube tumor associated with MRKH has been reported in literature. In a large retrospective study done by Pan, et al.[3] in 2016, it was noted that 96% of patients with MRKH syndrome had normal adnexae (A0, using VCUAM classification), and none of the remaining 4% manifested with a fallopian tube tumor. An earlier study done by Pittock, et al.[7] reviewed 25 patients diagnosed with MRKH syndrome. Only 4 out of the 25 had adnexal abnormalities (Two were found to have absence of one ovary and two had benign cysts.). None of the patients they studied presented with a fallopian tube mass. Most of the patients presented with renal (MR), cardiac (MC) and skeletal anomalies (MS).

Neoplasms of the fallopian tube, per se, are quite uncommon and among the rarest tumors in the female genital tract. They are often asymptomatic

and majority are incidental findings during a gynecologic workup or surgery. Papilloma, adenofibroma, endometrioid polyp, cystadenoma, and cystadenofibroma are among the benign tumors of the fallopian tube. Papillary serous cystadenofibroma of the fallopian tube is a very rare benign tumor which is usually found at the fimbrial end of the fallopian tube and is considered to be of mullerian origin.[8]

As of 2015, only 18 cases of serous cystadenofibroma of the fallopian tube have been reported. Most measure between 0.5 to 3 cm in diameter, are cystic and with coarse papillary excrescences.[8] These tumors often have a benign course and no malignant transformation has been reported. Histologically serous cystadenofibromas of the fallopian tube are similar to their ovarian counterpart with the cyst being lined by cuboidal epithelial cells.[9,10]

Conclusion

Patients with mullerian agenesis may still develop pelvic tumors often seen in women with normal development of reproductive organs. They should be regularly screened for such tumors and timely psychosocial counseling should be part of the management of these patients.

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