Journal of Gynecological Oncology

Case Report
Published: 01 Oct, 2022

Endometrioid Adenocarcinoma Occurring in a Patient of Mayer Rokitansky Küster Hauser Syndrome - A Rare Occurrence

*Ritu Thakur**

Department of Surgical Oncology, All India Institute of Medical Sciences, India

Introduction

Mayer Rokitansky Küster Hauser syndrome is a rare congenital developmental anomaly (Mullerian agenesis) characterized by congenital absence of uterus or rudimentary uterus with absence of upper two third of vagina with normal development of secondary sexual characteristics and a normal 46 XX karyotype [1]. Incidence of MRKH syndrome is estimated to be 1 in 4,500 women [2]. MRKH syndrome has been subdivided into 2 subtypes: Type A has isolated Mullerian duct anomaly which include shallow vaginal dimple or pouch with absent cervix, uterus, upper third of vagina with no other organ anomaly. Type B include in addition to type A Mullerian duct anomaly, varying degree of associated congenital renal malformations (renal agenesis and horseshoe kidney), skeletal abnormalities (scoliosis, spina bifida and sacral lumenization) and unilateral auditory defects [3]. Infrequently isolated cases of ovarian tumors in MRKH syndrome have been reported in the past. Of these Serous adenocarcinomas are more commonly. We report only second case of endometroid adenocarcinoma occurring in a patient with MRKH syndrome.

Case Presentation

A 57-years old married, nulligravida female with history of primary amenorrhea presented with complaints of lower abdominal pain for 4 months. She was evaluated for primary amenorrhea and infertility 15 years ago. Diagnostic laparoscopy was done and absent uterus was confirmed, no further work up was done at that time. Her past medical history was unremarkable for other medical conditions. On physical examination she had normal secondary sexual characteristics and her external genitalia were normal in appearance. The urethra was normal in appearance. There were no other cardiovascular, respiratory or skeletal anomalies in the patient. On her abdomen examination ill-defined abdomino-pelvic mass was palpable in midline. On per vaginum examination, while vagina was approx. 6 cm in length and two fingers in breadth, uterus and cervix were not palpable. However a palpable mobile extraluminal pelvic mass was noted (Figure 1).

In view of patient's presentation, transabdominal ultra sound was done which showed 9.4 cm × 6.6 cm hypoechoic mass in right side of pelvis, uterus and right ovary not visualized separately. A follow up CECT whole abdomen was done which showed 8.3 cm × 7.4 cm × 9.5 cm right adnexal mass with internal non enhancing areas of cystic degeneration reaching up to right iliac fossa, uterus and cervix not visualized separately, Moderate ascites, no nodes, bilateral kidney normal. There were no peritoneal/omental deposits. Liver and all other solid and hollow organs were normal. Her pre -operative CA-125 was raised: 361 U/ml (N: 0-35 U/ml) (Figure 2).

Elective staging laparotomy revealed a solid cystic mass of size 10 cm × 8 cm in right adnexa, right ovary not visualized separately; left ovary and tube appeared to be normal and attached to rudimentary uterus. On Histopathology right ovary was enlarged measuring 9.3 cm × 6.2 cm × 5 cm with no capsular invasion, in cut section it was predominantly solid with few cystic areas. On microscopy the tumour was mainly in the form of glands. On immunohistochemistry it showed a wild type pattern of immunopositivity for P53 and was immunonegative for WT1. So a final diagnosis of endometroid adenocarcinoma Grade 1 with uterine agenesis was given (Table 1 and Figure 3).

Discussion

Mayer Rokitansky syndrome is a rare syndrome with an estimated prevalence of 3.2% in all women [17]. It is characterized by the utero vaginal aplasia of variable degrees with a normal

OPEN ACCESS

Correspondence:
Ritu Thakur, Department of Surgical oncology, All India Institute of Medical Sciences, New Delhi, India, Tel: +91-8120698544;
E-mail: ritu.thakur380@gmail.com

Received Date: *06 Sep 2022*
Accepted Date: *27 Sep 2022*
Published Date: *01 Oct 2022*

Citation:
Thakur R. Endometrioid Adenocarcinoma Occurring in a Patient of Mayer Rokitansky Küster Hauser Syndrome - A Rare Occurrence. J Gynecol Oncol. 2022; 5(2): 1076.

Copyright *© 2022 Ritu Thakur. This is an open access article distributed under the Creative Commons Attribution License, which permits unrestricted use, distribution, and reproduction in any medium, provided the original work is properly cited.*

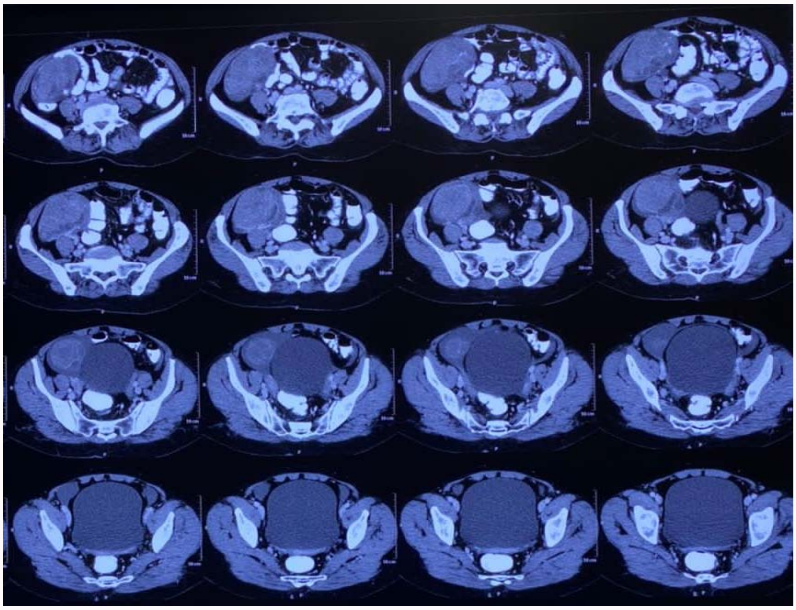

Figure 1: CT film (axial cuts) suggestive of rudimentary uterus with adnexal mass.

Table 1: Summary of previously reported cases of MRKH syndrome coexisting with ovarian carcinoma.

S. No	Article	MRKH Type	Age at onset	Ovarian tumour type	Ovarian tumour stage (FIGO)
1	Koonings et al. [4]	I	26	Yolk sac tumour	I
2	Larsen et al. [5]	II	34	Sertoli cell	I
3	Pommerenke et al. [6]	II	79	Bilateral Sertoli cell	NA
4	Ghirardini and Magnani et al. [7]	I	68	Undifferentiated carcinoma	III
5	Tsaur et al. [8]	I	4	Immature teratoma	I
6	Takeuchi et al. [9]	NA	8	Yolk sac tumour	I
7	Mishina et al. [10]	NA	35	Dysgerminoma	I
8	Kavallaris et al. [11]	II	48	Mixed epithelial carcinoma	III
9	Ko et al. [12]	II	37	Serous carcinoma	III
10	Bae et al. [13]	II	31	Serous papillary	III
11	Nusrath et al. [14]	II	65	Endometriod carcinoma	I
12	Juusela et al. [15]	I	72	Sex cord stromal	I
13	Huepenbecker et al. [16]	NA	64	Serous carcinoma , serous tubal carcinoma	III
			63		III
14	R.villa et al. [17]	I	33	Bilateral serous carcinoma	II
15	Current case	I	57	Endometroid carcinoma	I

development of secondary sexual characteristics and a normal 46XX karyotype. Though the ovaries in these patients are normal and functional, the co-occurrence of ovarian cancers with MRKH syndrome is still very rare. Only few cases (14 cases) of ovarian cancer in MRKH syndrome have been reported in literature so far.

Ovarian cancer is the second most common gynecologic malignancy in developed countries and third in developing countries. Several risk factors including increasing age, infertility, polycystic ovarian syndrome, endometriosis, and various ovarian cancer susceptibility genes like *BRCA1*, *BRCA2*, Mismatch repair genes associated with lynch syndrome have been associated with ovarian cancer.

Koonings et al. [4] in 1991 first described the occurrence of endodermal sinus tumor in a patient with MRKH syndrome [4]. Following which 14 more cases including 8 epithelial, 4 germ cell and 3 sex cord stromal tumors of ovary were reported subsequently. Out of 14 reported cases 7 cases were below 40 yrs of age. Median age of onset was 55 years (range 31-68) for epithelial tumors, 30 years (4-79) for non-epithelial tumors [16]. Our patient presented at the age of 57 years with primary amenorrhea and pain abdomen.

Table 1 Summarizes current available literature on MRKH syndrome associated ovarian cancer. Amongst all, eight developed epithelial cancer, (of which five were serous adenocarcinoma and only two were of endometroid adenocarcinoma subtype) four developed germ cell tumour and 3 developed sex cord stromal tumour of ovary.

Majority of cases reported in literature are sporadic in origin. The etiological basis of MRKH syndrome is still unclear. Frequently found to be sporadic in origin few studies have also reported familial

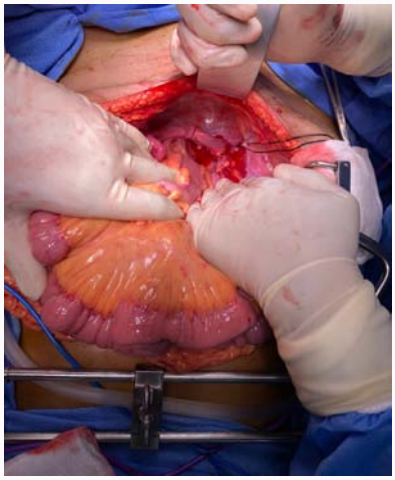

Figure 2: Intraoperative Picture showing cord like rudimentary uterus.

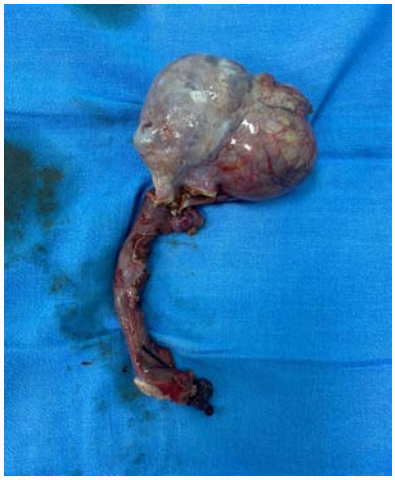

Figure 3: Specimen of cord like rudimentary uterus with right adnexal mass of approximately 5 cm × 5 cm size.

clustering in these cases suggesting a putative genetic origin of this syndrome. Fontana et al. [18] studied the genomic alterations in MRKH and observed that these patients have polygenic patterns of inheritance [18]. A definite genetic basis of this syndrome is yet to be established. Owing to rarity of ovarian carcinoma coexisting with MRKH syndrome very limited data is available on genetic basis of these ovarian tumors. Interestingly in 2017, when Huepencker et al. [16] reported occurrence of ovarian cancer in MRKH syndrome in two sisters, 63 and 64 years of age, a possibility of a shared genetic origin of the two conditions was speculated and a genetic testing was performed [16]. Both these cases however tested negative for *BRCA1/2* and other 31 cancer susceptibility genes. Since no information on cancer family history is available for the reported cases it is still difficult to rule out genetic basis to this infrequent concurrence.

Conclusion

We report only second case of ovarian endometroid adenocarcinoma occurring in a patient of MRKH syndrome and provided an updated literature review of all the reported cases so far. Due to limited available data and rarity of co-occurrence of ovarian cancer in MRKH syndrome, the possible risk factors, etiological and genetic basis of the disease are yet to be established. None the less the possibility of this relatively infrequent complication must also be kept in mind while managing MRKH syndrome patients.

References

1. Morcel K, Camborieux L, Guerrier D. Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome. Orphanet J Rare Dis. 2007;2(1):13.
2. Morcel K, Guerrier D, Watrin T, Pellerin I, Levêque J. The Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome: Clinical description and genetics. J Obstet Gynecol Reprod Biol. 2008;37(6):539-46.
3. Bombard DS, Mousa SA. Mayer-rokitansky-kuster-hauser syndrome: Complications, diagnosis and possible treatment options: A review. Gynecol Endocrinol. 2014;30(9):618-23.
4. Koonings PP, Al-Marayati L, Schlaerth JB, Lobo RA. Mayer-rokitansky-kuster-hauser syndrome associated with endodermal sinus tumor of the ovary. Fertil Steril. 1991;56(3):577-8.
5. Larsen ET, Chang-Poon V, Benediktsson H, Robertson DI, Stuart GCE. Bilateral ovarian tumors in a woman with Mayer-Rokitansky-Kuster-Hauser syndrome. Adolesc Pediatr Gynecol. 1992;5(3):198-200.
6. Pommerenke F, Tessmann D, Meyer K. Giant Sertoli cell tumor of the ovary in mayer-rokitansky-küster-hauser syndrome. Zentralbl Gynakol. 1994;116(5):295-300.
7. Ghirardini G, Magnani A. Mayer-Rokitansky-Küster-Hauser syndrome and ovarian cancer. Report of a case. Clin Exp Obstet Gynecol. 1995;22(3):247-8.
8. Tsaur GT, Lee MH, Su SL, Wu MJ, Huang TW. Mayer-Rokitansky-Kuster-Hauser syndrome with immature teratoma of the ovary at age 4 years. Gynecol Oncol. 1995;56(3):456-9.
9. Takeuchi K, Oomori S, Oda N, Maeda K, Kaji Y, Maruo T. Coexistence of Mayer-Rokitansky-Küstner-Hauser syndrome and yolk sac tumor of the ovary in a prepubertal girl. Acta Obstet Gynecol Scand. 2006;85(2):245-7.
10. Mishina A, Gladun E, Petrovici V, Iakovleva I. Ovarian dysgerminoma in Mayer-Rokitansky-Kuster- Hauser syndrome to. Eur J Obstet Gynecol Reprod Biol. 2007;131(1):103-5.
11. Kavallaris A, Sait KH, Marar EA, Shakra RA, Diedrich K. Ovarian cancer and Mayer-Rokitansky-Kuster-Hauser syndrome. Gynecol Surg. 2011;8(2):197-9.
12. Ko AR, Kim JH, Park SY, Jeon YE, Kim SJ, Rhim CC, et al. A case of ovarian cancer developed in Mayer-Rokitansky-Küster-Hauser syndrome: Innate carcinogenesis of ovarian cancer. Korean J Obstet Gynecol. 2012;55(12):1001.
13. Bae HS, Ryu MJ, Kim IS, Kim SH, Song JY. Cancer of the supernumerary ovary in Mayer-Rokitansty-Küster-Hauser syndrome: A case report. Oncol Lett. 2013;5(2):598-600.
14. Nusrath S, Goel V, Varma P, Pawar S, Murthy S, Raju KVVN. Ovarian cancer and Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome a rare combination. Indian J Gynecol Oncol. 2016;14(2):1-4.
15. Juesela, Alshiek J, Garcia B, Minassian VA, Iglesia CB, Clark A, et al. Vaginal energy based devices. Female Pelvic Med Reconstr Surg. 2020;26(5):287-98.
16. Villa R, Azzollini J, Peissel B, Manoukian S. Co-occurrence of Mayer-Rokitansky-Küster-Hauser syndrome and ovarian cancer: A case report and review of the literature. Gynecol Oncol Reports. 2019;28:68-70.
17. Neisani Samani E, Falahian M. Leiomyoma arising from the rudimentary uterus. Iran J Pathol. 2007;2(4):187-9.
18. Fontana L, Gentilin B, Fedele L, Gervasini C, Miozzo M. Genetics of Mayer–Rokitansky–Küster–Hauser (MRKH) syndrome. Clin Genet. 2017;91(2):233-46.