DOI: 10.7860/NJLM/2016/18926:2133

Case Report

Obstetrics and Gynaecology # Pathology Section

Uterine Adenomyosis in Mullerian Remnant with Transverse Vaginal Septum with Haematocolpos in Mayer-Rokitansky-Kuster-Hauser Syndrome: A Case Report

KAVERI GUPTA, MANISHA SHARMA, REKHA JAIN, TARUN SARIN, NAMRATA

ABSTRACT

The Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is a rare disorder characterised by failure of the uterus and vagina to develop properly in women who have normal ovarian function and normal external genitalia and secondary sexual characters. Mullerian agenesis accounts for approximately 10% cases of primary amenorrhoea. We report a case of 40-year-old female with MRKH syndrome with primary amenorrhea who presented with lower abdominal pain. She had well developed secondary sexual characters. Her ultrasound showed bulky and heterogeneous uterus like mass with ill defined endometrial-myometrial interface with multiple uterine fibroids. Primary amenorrhea with normal secondary sexual characters and phenotype of 44XX lead us to the diagnosis of MRKH syndrome with uterine myoma in mullerian remnant.

Surgical intervention showed the presence of transverse vaginal septum with haematocolpos while the histopathology of the remnant showed adenomyosis. Because of rarity of the condition we report this case.

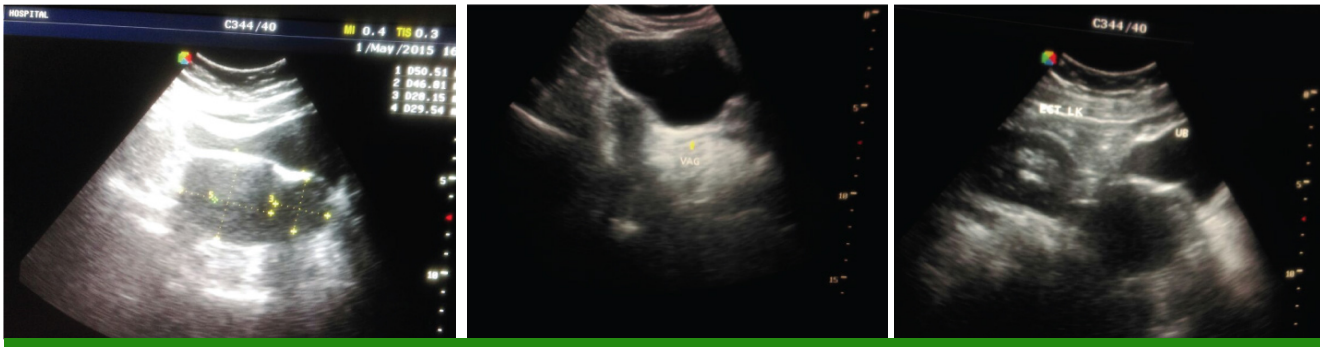
Keywords: Endometrial mass, Mullerian agenesis, Primary amenorrhea

CASE REPORT

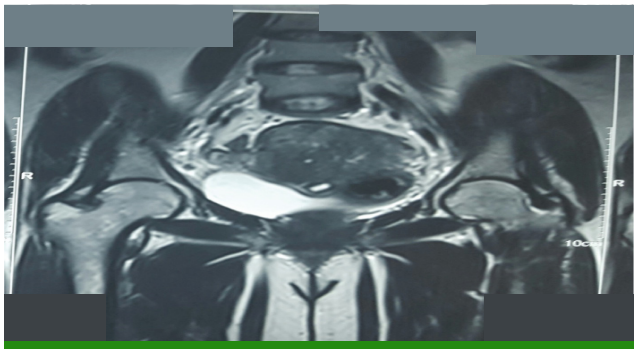
A 40-year-old female with primary amenorrhea presented with lower abdominal pain for past 15 years. The pain was dull aching in nature and radiated to thighs which improved with analgesics. The pain was occasional initially but for past 4-5 years it became cyclical in nature occurring for 4-5 days every month with increased intensity for which she seeked medical help. There was no association of pain with bladder or bowel. She was married for past 20 years with active sexual life. She did not have any sexual problem but never conceived. There was no history of congenital anomalies in the family. On admission her vitals were stable and there was no pallor, icterus, oedema or any lymphadenopathy. Her breasts were normally developed with normal axillary and pubic hair. No abnormality was detected on respiratory, cardiovascular, central nervous system or per abdominal examination. On per vaginum examination, there was a 6-7 cm long blind vagina with no cervix. A uterus like mass of 10 weeks gestation size was felt which was irregularly enlarged, firm in consistency, mobile and non tender. Both the fornices were free. Findings were confirmed on per rectal examination.

Her investigations-complete blood count with ESR, urine examination, blood sugar, LFT and KFT were within normal range. HIV, HBsAg and VDRL were non reactive. Her buccal smear showed presence of barr bodies. On ultrasound examination uterus was anteverted, bulky and heterogeneous with ill defined endometrial-myometrial interface with multiple hypoechoic lesions within anterior and posterior walls suggestive of fibroids (largest of size 4.4 x 3 cm) [Table/Fig-1]. A well defined hypoechoic predominantly cystic lesion 2 x 3 cm was seen just inferior to uterine body in midline and towards left. Cervix was not clearly visualised and vagina was blind [Table/Fig-2]. Bilateral ovaries appeared normal. No free fluid in pelvis. A reniform structure was seen in left hemipelvis suggestive of left sided pelvic kidney [Table/Fig-3].

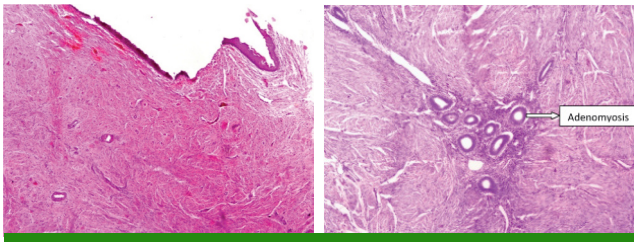
Findings were confirmed by intravenous pyelogram which showed ectopic left kidney and left ureter with normal bilateral renal function. MRI report also showed similar findings with anteverted, bulky uterus with multiple varying sized hypointense lesions with largest being 4.4 x 3 mm. Endometrium was imperceptible. Vaginal canal appeared normal with hypoplastic cervix. Bilateral ovaries were normal size and left sided pelvic kidney was seen [Table/Fig-4].



[Table/Fig-1]: USG showing anteverted bulky uterus with multiple fibroids. **[Table/Fig-2]:** USG showing blind vagina along with cystic lesion. **[Table/Fig-3]:** USG showing left ectopic kidney.



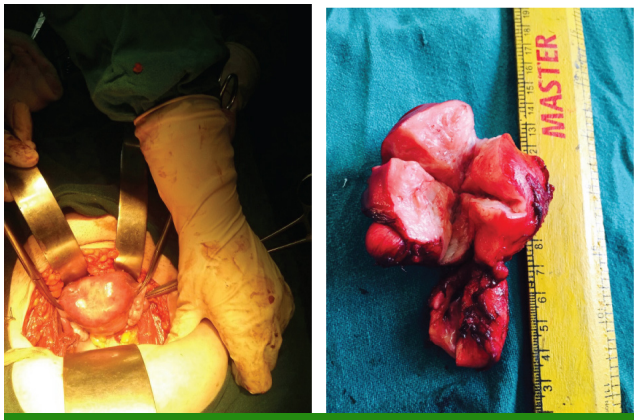
[Table/Fig-4]: MRI showing uterus like mass with multiple fibroids.



[Table/Fig-7]: Histopathology slide showing stratified squamous epithelium in the wall of small mass with cavity (part of vagina above transverse septum). **[Table/Fig-8]:** Histopathology slide showing adenomyosis.

The diagnosis of Mayer-Rokitansky-Kuster-Hauser syndrome was made and decision for exploratory laparotomy with removal of uterine remnants was taken. All pre anaesthetic check-up, investigations were found within normal limits. Exploratory laparotomy was done with removal of uterine remnants. Per operatively, uterine mass of 12 cm in size, firm in consistency with multiple fibroids was seen [Table/Fig-5]. Cervix was not localised. Another rounded mass of 2 x 3 cm size with cavity attached to lower end of solid mass with old collected chocolate coloured blood approximately 5 to 10cc was present. Cavity was thin walled with smooth surface. Bilateral tubes and ovaries were normal.

On cut section, no endometrium or cavity was seen within the uterine mass. There was no growth or papillae in the



[Table/Fig-5]: Uterus like mass with multiple fibroid and bilateral normal ovaries. **[Table/Fig-6]:** Cut section showing solid uterus looking mass with no endometrial cavity or cervix.

cavity of the rounded mass which was attached to lower end of solid mass. It was filled with old collected chocolate coloured blood [Table/Fig-6].

The specimen was sent for histopathological examination which on gross reported already cut open specimen of distorted uterus measuring 7x6x3.5 cm [Table/Fig-6]. Endometrial cavity could not be made out. Two intramural nodules 1-2 cm in diameter were identified. Cervix could not be made out. Micro section examined from distorted uterus showed multiple foci of endometrial glands and stroma surrounded by hyperplastic muscle bundles throughout myometrium. No endometrial cavity or lining could be identified. Micro section from the lower end with cavity show flattened stratified squamous epithelium and fibrovascular stroma showing mild mononuclear cell infiltration (vaginal tissue). Endocervical lining or gland could not be detected on multiple sections [Table/Fig-7,8].

DISCUSSION

The MRKH syndrome is a rare anomaly characterised by congenital aplasia of the uterus and vagina in women showing normal development of secondary sexual characters and normal 44XX karyotype [1]. They have normal ovarian function and normal external genitalia of female. It is a

rare disorder occurring in about 1 in 4000 to 5000 female births and is a common cause of primary amenorrhoea [1]. Mullerian agenesis accounts for approximately 10% of cases of primary amenorrhoea [2].

Our patient presented with primary amennorhoea with cyclical pain along with normal secondary sexual characters and presence of uterus like mass, with blind vagina but absent cervix. On exploratory laparotomy there was absence of uterine cavity and thus she is classified under the main class of aplastic uterus (U5) with co-existent cervical aplasia (C4) and transverse vaginal septum (V3) according to ESHRE/ESGE classification of female genital tract congenital anomalies [3]. Aplastic uterus is a formation defect and at times is associated with functional cavity in an existent rudimentary horn in the mullerian remnant (class U5a of uterine anomaly). Presence of this functional cavity in the rudimentary horn is responsible for the cyclical bleed and pain [3]. In our patient there is a possibility of presence of a small functional cavity in the rudimentary horn of aplastic uterus which was responsible for cyclical bleed and thus formation of hematocolpos in the upper portion of vagina above the transverse vaginal septum as the outflow tract was blocked. The associated cyclical pain can also be explained by this. Cyclical bleed would have been very small in early years which went unnoticed and so she came for medical help late in life.

Of the patients with MRKH syndrome, 15% have an absent, pelvic or horseshoe kidney, 40% have a double urinary collecting system and 5% to 12% have skeletal abnormalities. MRKH syndrome is associated with abnormal galactose metabolism also [2]. There was presence of ectopic left kidney and left ureter in our patient though skeletal X-ray showed no abnormality.

Uterine adenomyosis is a benign disorder characterised by the extension of endometrial glands and stroma into the myometrium [4].The possibility of presence of minimal endometrial lining in our case and the presence of transverse vaginal septum which acted as an obstruction to outflow tract might be responsible for the adenomyosis.

CONCLUSION

The present case is an evidence of Mayer-Rokitansky-Kuster-Hauser syndrome with uterine adenomyosis with transverse vaginal septum and haematocolpos. The histogenesis of adenomyosis in this patient may be direct invasion of eutopic endometrium.

REFERENCES

[1] Mungadi A, Ahmad Y, Ismail S. Mayer-Rokitansky-Kuster-Hauser syndrome: surgical management of two cases. *Journal of Surgical Techniques and Case Report*. 2010; 2: 39-43.

[2] Baker VL, Schillings WJ, McClamrock HD. Amenorrhoea (Chapter 30). In: Berek. JS; Berek DL, editors. Berek and Novak's Gynecology. 15th ed. Philadelphia (PA): Wolters Kluwer India Pvt Ltd; 2012. p. 1035-65.

[3] Grimbizis GF, Gordts S, Sardo ADS, Brucker S, Angelis CD, Gergolet M et al. The ESHRE/ESGE consensus on the classification of female genital tract congenital anomalies. *Human reproduction*. 2013; 28(8): 2032-44.

[4] Chun S, Kim YM, Ji Y. Uterine adenomyosis which developed from hypoplastic uterus in postmenopausal woman with Mayer-Rokitansky-Kuster-Hauser syndrome: a case report. *Journal of Menopausal Medicine*. 2013; 19(3): 135-38.

AUTHOR(S):

1. Dr. Kaveri Gupta
2. Dr. Manisha Sharma
3. Dr. Rekha Jain
4. Dr. Tarun Sarin
5. Dr. Namrata

PARTICULARS OF CONTRIBUTORS:

1. Senior Resident, Department of Obstetrics and Gynaecology, NDMC Medical College and Hindu Rao Hospital, NDMC, Delhi, India.
2. Senior Specialist, Department of Obstetrics and Gynaecology, NDMC Medical College and Hindu Rao Hospital, NDMC, Delhi, India.
3. Chief Medical Officer (NFSG), Department of Obstetrics and Gynaecology, NDMC Medical College and Hindu Rao Hospital, NDMC, Delhi, India.
4. Medical Officer, Department of Radiology, NDMC Medical College and Hindu Rao Hospital NDMC, Delhi, India.
5. Senior Specialist, Department of Pathology, NDMC Medical College and Hindu Rao Hospital, NDMC, Delhi, India.

NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:

Dr. Manisha Sharma,
26, SFS DDA Flats, Mukherji Nagar, Delhi-110009, India.
E-mail: drmanishasharma63@gmail.com

FINANCIAL OR OTHER COMPETING INTERESTS:
None.

Date of Publishing: **Jul 01, 2016**