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Case Report

Mullerian agenesis: An invisible illness

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ABSTRACT

Mullerian ageness is commonly known as Mayer-Rokitansky-Kuster-Hauser syndrome. It occurs in 1 among 4000-5,000 females. Mullerian ducts are the anlage of primordial tissue of female reproductive tract, which is rare cause of agenesis or hypogenesis female genital tract. This case presented here is one among the rarest in this tertiary care hospital.

Case History: 11-year-old female presented with complaints of fever and vague abdominal pain since 15 days. On examination, the presence of imperforate hymen was noted; USG revealed hematometra and hematocolpus. Right ovary showed a simple cyst. Left ovary was poorly indiscernible; right kidney was unremarkable while left kidney was not visualized.

Vaginal atresia was noted while doing hymenoplasty; hysterectomy with salpingoophorectomy was done for hematometra with hematocolpus and hematosalpinx and the specimen was sent for HPE examination..

Conclusion : The case presented here is unique example of mullerian agenesis which presented with unusual clinical features of fever and vague abdominal pain. It is also unique in that it is very rare case which occurred in a female with early menarche and cryptomenorrhea that was incidentally diagnosed on pelvic USG. Lastly, its incidence is noted to be rare in this tertiary care teaching hospital that it is the first case of its kind reported in the last five years.

1. Introduction

Mullerian agenesis is commonly known as Mayer-Rokitansky-Kuster-Hauser syndrome. It occurs in 1 among 4000-5,000 females. Mullerian ducts are the anlage of primordial tissue of female reproductive tract,[1] which is rare cause of agenesis or hypogenesis female genital tract. This case is one among the rarest in this tertiary care hospital. Developmental anomalies of the Mullerian Duct System are some of the more fascinating disorders encountered.[2] Mullerian defects represent a broad spectrum of abnormalities ranging from uterine-vaginal agenesis to duplication of the uterus-vagina to minor uterine cavity abnormalities and these are divided among six categories.[2] They are frequently associated with kidney and axial skeletal system abnormalities.[3] Workup for these patients includes ultrasonography and/or hysterosalpingography.[4] If these investigations are not able to conclude the diagnosis, further investigations like MRI and IVP need to be done.[5]

2. Case History

11-year-old female came to the out-patient department with clinical complaints of fever and vague abdominal pain since 15 days. On examination, the presence of imperforate hymen was noted; USG revealed hematometra and hematocolpus. Right ovary showed a simple cyst. Left ovary was poorly indiscernible; right kidney was unremarkable while left kidney was not visualized. Vaginal atresia was noted while doing hymenoplasty; hysterectomy with salpingoophorectomy was done for hematometra with hematocolpus and hematosalpinx and the specimen was sent for HPE examination.

On gross examination, the specimen received consisted of Unicornuate uterus, 4x3x1 cm, with only one ovary that

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measured 2x1 cm and a single rudimentary dilated fallopian tube with multiple paratubal cysts. An outpouching from lower end of cervix measuring 10x6x4 cm was seen.

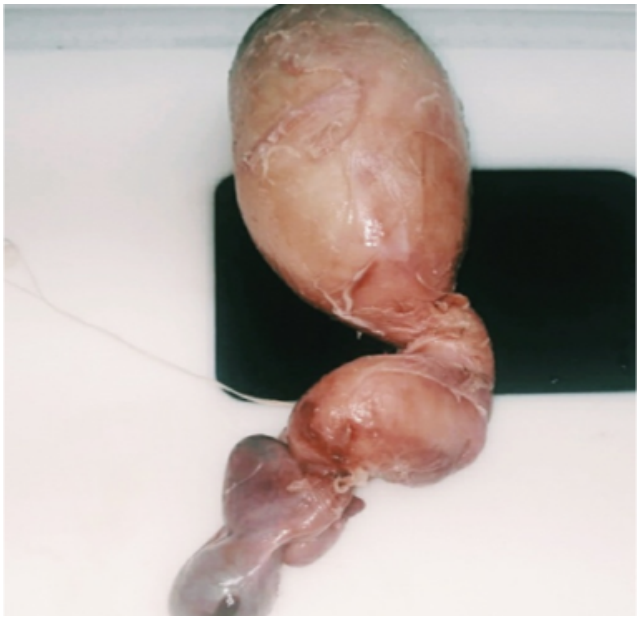

Fig. 1: Gross specimen shows unicornuate uterus with single ovary.

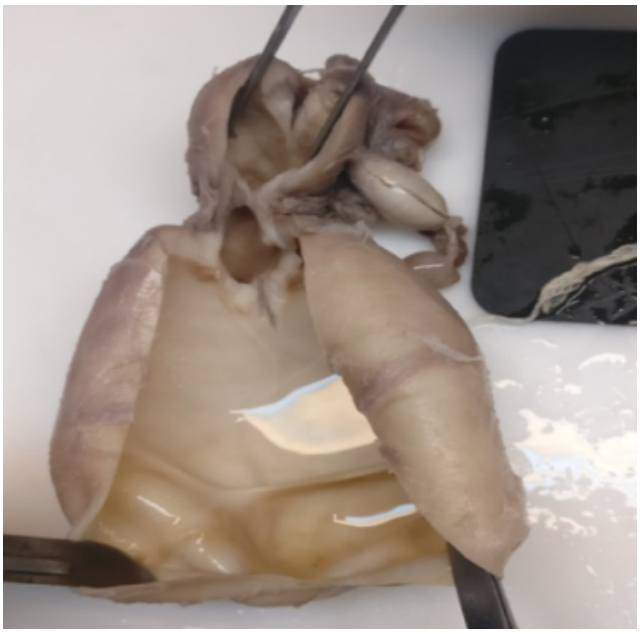

Fig. 2: Single fallopian tube with multiple paratubal cysts. An outpouching from lower end of cervix was seen.

On microscopic examination, the sections from dilated pouch-like cervical structure showed a dilated portion of cervix lined by attenuated tall columnar epithelium interspersed sparsely by few endocervical type of glands; ectocervical epithelium was indiscernible.

Sections from uterus showed dilated uterine cavity lined by attenuated cuboidal epithelium with occasional dispersed endometrial glands; the endometrial cavity showed the presence of abundant haemorrhagic material.

Sections from the only ovary showed few attenuated primordial follicles and occasional cystic ovarian follicles amidst abundant stroma. Sections from the only dilated fallopian tube showed features of haematosalpinx.

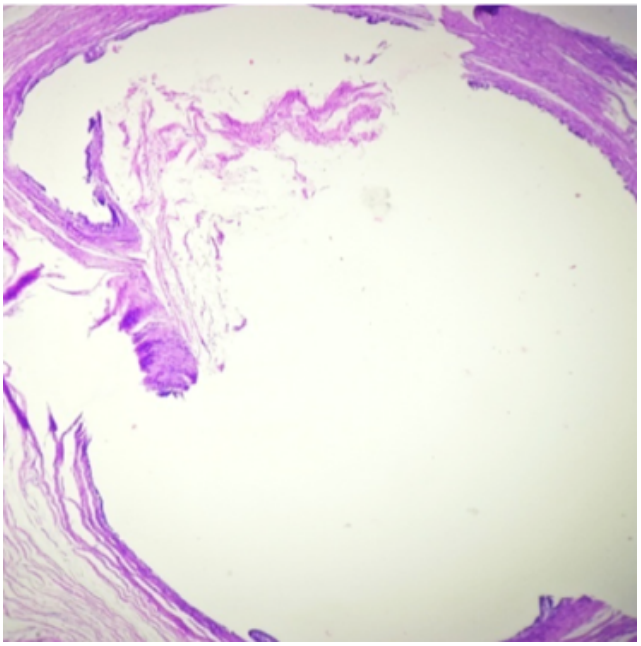

Fig. 3: Dilated fallopian tube lined by cuboidal to flattened epithelium(10X, H and E)

3. Discussion

Patients with Mullerian agenesis are usually diagnosed when they are clinically checked for primary amenorrhoea with otherwise normal growth and sexual development.[2] On physical examination, patients with Mullerian agenesis are found to have normal height, breast, body hair and external genital organs.[2] Atypical midline uterus is generally not found on radiological imaging studies. Other clinical conditions that needs to be differentiated with mullerian agenesis are vaginal or uterine obstructions or 46 XY differences of sexual development.[2] Patients with mullerian agenesis generally have 46 XX karyotype i.e. normal female karyotyping in most individuals.[2]

This case here highlights that Mullerian duct anomalies should be taken into consideration as an important condition in the differential diagnosis of cyclical abdominal pain that responds poorly to analgesics, because if active endometrium is present, patient may experience a cyclic abdominal pain corresponding to her menstrual cycle.[6]

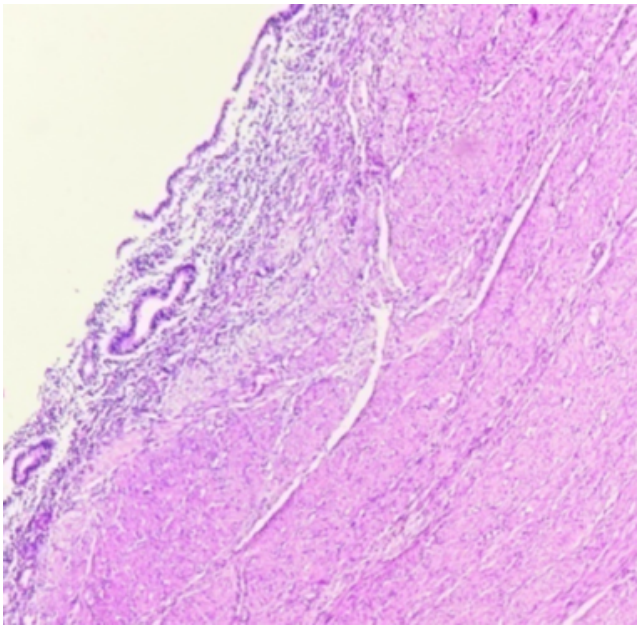

Fig. 4: Dilated uterine cavity showing attenuated cuboidal epithelium

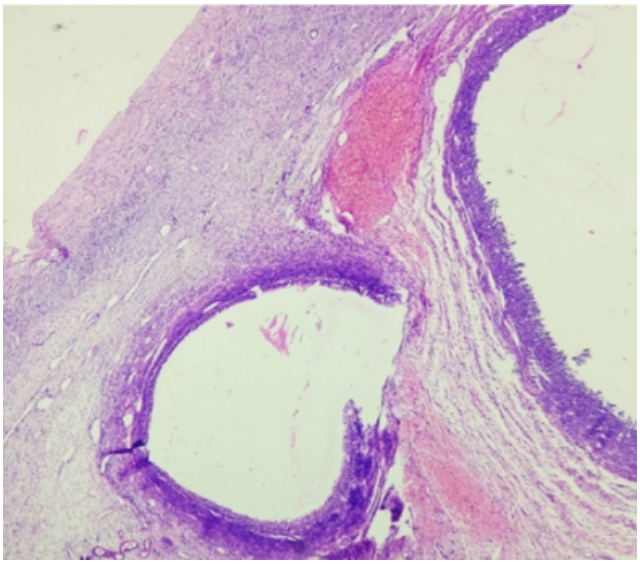

Fig. 5: Ovary showing attenuated and cystic ovarian follicle with abundantstroma.

Though laparoscopic examination is not essential part of evaluation of Mullerian Agenesis, it may be of significant utility value in evaluation of differential diagnosis and management of patients complaining of pelvic pain due to varied clinical conditions.[2]

As developmental anomalies of the urinary tract and Mullerian agenesis commonly co-exist,[7] the former anomaly should be specifically investigated for before elective surgery is carried out.[8,9] Since multiple studies

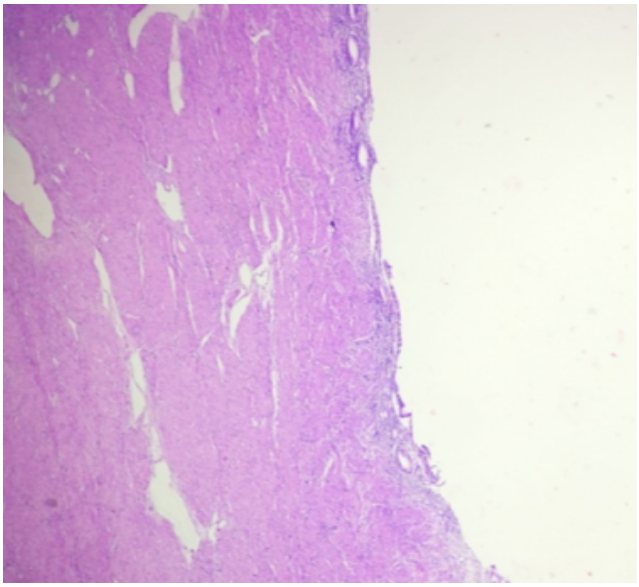

Fig. 6: Dilated pouch-like cervix showing columnar epithelium with fewendocervical glands.

have confirmed the prevalence of renal anomalies in patients with Mullerian Agenesis to be 27-29%,[8,9] ultrasonography of kidney is clinically mandated in all cases. Skeletal anomalies (e.g. scoliosis, vertebral arch disturbances, hypoplasia of wrist) have been reported in approximately 8-32% of patients; hence radiographic examination of spine may reveal an anomaly even in patients with no discernible clinical symptoms.[7–9] A variety of uterine anomalies including mullerian agenesis are often found in VATER/VACTERL association (vertebral anomalies, anorectal malformations, cardiovascular anomalies, traceosephageal fistula, esophageal atresia, renal anomalies, limb defects).[10]

Surgical management of simpler Mullerian duct malformations such as imperforate hymen, transverse vaginal septum, and cervical atresia have been carried out without complications.[11–14] Artificial construction of new vagina/cervix requires more complex operations,[15–18] albeit with limited success, are associated with high morbidity with many of these patients ending up with severe complications ultimately needing hysterectomy. In this patient reconstructive surgery was thought to be unsuitable because of the associated morbidity. Generally accepted mode of treatment of these patients has been to surgically remove the Mullerian structures during the initial stages of operation itself, which could help prevent postoperative complications.[6]

4. Conclusion

The most important steps in effective clinical management of mullerian agenesis include correct diagnosis of the

underlying condition, evaluation of associated congenital anomalies and counseling for psychosocial affectations arising out of such problems, in addition to treatment or clinical intervention to address the functional effects of genital anomalies.[2] All such patients should be encouraged to have close interactions with peer support groups which work to help in alleviating physical and psychological fallouts arising out of such conditions including the consequent problems of, at least in some of these cases, difficulties or inability in future to conceive and bear children.[19,20] It would be prudent to apprise the patient of this significant consequence of these conditions as also the possibility of sexually active women with Mullerian agenesis being at greater risk of getting sexually transmitted diseases of which they be made aware of so that appropriate preventive and regular screening measures could be adopted.[21]

5. Source of Funding

None.

6. Conflict of Interest

None.

References

1. 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.
2. ACOG Committee Opinion No. 728. American College of Obstetricians and Gynecologists. *Obstet Gynecol*. 2018;131:e35–42.
3. 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. *Human Reprod*. 2006;21(3):792–7.
4. Purandare CN, Rashna U. Bandukwalla Vidya, Visarya Nita, Purandare Nikhil. Hematometra in Uterus Didelphys with Right Hemivagina and Right Renal Agenesis. *J Obstet Gynecol India*. 2011;p. 210–2.
5. Rock JA, Jones HW. The double uterus associated with an obstructed hemivagina and ipsilateral renal agenesis. *Am J Obstet Gynecol*. 1980;138:338–42.
6. Gasim T, Jama FEA. Massive Hematometra due to Congenital Cervicovaginal Agenesis in an Adolescent Girl Treated by Hysterectomy: A Case Report. *Case Rep Obstet Gynecol*. 2013;2013:640214.
7. 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. *Human Reprod*. 2006;21(3):792–7.
8. Kapczuk K, Iwaniec K, Friebe Z, Kedzia W. Congenital malformations and other comorbidities in 125 women with Mayer-Rokitansky-Küster-Hauser syndrome. *Eur J ObstetGynecol Reprod Biol*. 2016;207:45–9.
9. Rall K, Eisenbeis S, Henninger V, Henes M, Wallwiener D, Bonin M, et al. 2015.
10. Breech L. Gynecologic concerns in patients with anorectal malformations. *Semin Pediatric Surg*. 2010;19(2):139–45.
11. Oga M, Anai T, Yoshimatsu J, Kawano Y, Hayata T, Miyakawa I, et al. Retrohymenal Vaginal Atresia with Perforate Transverse Vaginal Septum. *Gynecol Obstet Investig*. 1992;34(3):190–2.
12. Quinn T, Erickson V, Knudson MM. Down's syndrome, precocious puberty, and transverse vaginal septum: An unusual cause of abdominal pain. *J Pediatric Surg*. 2001;36(4):641–3.
13. Alborzi S, Momtahan M, Parsanezhad ME, Yazdani M. Successful treatment of cervical aplasia using a peritoneal graft. *Int J Gynecol Obstet*. 2005;88(3):299–302.
14. Garat JM, Martinez E, Aragona F, Gosalbez R. Cervical Uterine Atresia with Hematometra: A Rare Cause of Urinary Retention in a Girl. *J Urol*. 1984;132(4):772–3.
15. Khunda SS. A new approach in the management of lower Mullerian atresia. *J Obstet Gynaecol*. 1998;18(6):566–8.
16. Bugmann P, Amaudruz M, Hanquinet S, Scala GL, Birraux J, Coultre CL. Uterocervicplasty with bladder mucosa for the treatment of complete cervical agenesis. *Fertil Steril*. 2002;77:831–5.
17. Gurbuz A, Karateke A, Haliloglu B. Abdominal surgical approach to a case of complete cervical and partial vaginal agenesis. *Fertil Steril*. 2005;84(1):217.
18. Lee CL, Wang CJ, Liu YH, Yen CF, Lai YL, Soong YK. Laparoscopically assisted full thickness skin graft for reconstruction in congenital agenesis of vagina and uterine cervix. *Human Reprod*. 1999;14(4):928–30.
19. Patterson CJ, Crawford R, Jahoda A. Exploring the psychological impact of Mayer–Rokitansky–Küster–Hauser syndrome on young women: An interpretative phenomenological analysis. *J Health Psychol*. 2016;21(7):1228–40.
20. Ernst ME, Sandberg DE, Keegan C, Quint EH, Lossie AC, Yashar BM. The Lived Experience of MRKH: Sharing Health Information with Peers. *J Pediatr Adolesc Gynecol*. 2016;29(2):154–8.
21. Workowski KA. Centers for Disease Control and Prevention Sexually Transmitted Diseases Treatment Guidelines. *Clin Infect Dis*. 2015;61(RR3):1–137.

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