Faridpur Med. Coll. J. 2026;21(2):61-63 DOI: https://doi.org/10.3329/fmcj.v21i2.90823

Case Report

Mullerian Agenesis/ Mayer-Rokitansky-Kuster-Hauser (MRKH) Syndrome: A Rare Case Report

ZM Ferdousi[1], K Hossain[2], MH Rahman[3]

Abstract:

Background: *Mullerian agenesis or Mayer- Rokitansky- Kuster-Hauser (MRKH) syndrome is a congenital malformation characterized by failure of the Mullerian ducts to develop resulting in a missing uterus & aplasia of upper vagina.*

Case report: *Here, we are reporting a case of MRKH syndrome type II in a 22-year-old married female who presented with primary amenorrhoea & painful coitus & was referred to us for pelvic ultrasonography. Absent uterus & upper part of vagina with normal ovaries & left sided ectopic (pelvic) kidney on ultrasonography & normal female hormone levels on hormone analyses confirmed the diagnosis of MRKH syndrome type II.*

Conclusion: *MRKH syndrome can occur with normal endocrine function & secondary sexual characteristics. Ultrasonography & hormone analyses play a vital role in confirming the diagnosis.*

Keywords: MRKH syndrome, Mullerian agenesis, Mayer-Rokitansky-Kuster-Hauser syndrome, Primary amenorrhoea.

Introduction:

Mullerian agenesis or Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is a rare congenital disorder with an incidence of 1 in 4500 female birth.[1,2] It is characterized by uterovaginal aplasia with normal secondary sexual characteristics[1,3] and genetic karyotype 46XX.[4,5] They are of two types: type I having only uterovaginal agenesis and type II having uterovaginal agenesis with renal, skeletal & auditory anomalies. The exact etiology of MRKH syndrome is not known. Detailed history, physical examination, ultrasonography & hormone analyses are essential for the diagnosis. Counseling of the patient and neovagina creation is the mainstay for the management.

Case Report:

A 22 years old female, married for 1 year, presented with primary amenorrhoea and painful coitus. She had normal secondary sexual characteristics. There was no history of primary amenorrhoea in her first- & second-degree relatives. Her mother confirmed no known exposure to any medication or maternal illness during pregnancy. The patient had no significant past medical history & had not undergone any surgery. Due to her presenting illness the patient was advised ultrasonography of pelvic organs and referred to the ultrasound division of Institute of Nuclear Medicine and Allied Sciences (INMAS), Faridpur. Gray-scale ultrasonography (USG) of pelvic organs was performed using a USG machine (esaote My Lab 9 medical

1. Muslima Ferdousi, MBBS, Medical Officer, Institute of Nuclear Medicine & Allied Sciences (INMAS), Faridpur. E-mail: muslimaferdousi@gmail.com.
2. Kawsar Hossain, MBBS, Medical Officer, Institute of Nuclear Medicine & Allied Sciences (INMAS), Faridpur. E-mail: kawsarhossain02018@gmail.com.
3. Md. Hafizur Rahman, MBBS, MPhil (Nuclear Medicine), Director & Chief Medical Officer, Institute of Nuclear Medicine & Allied Sciences (INMAS), Faridpur. E-mail: mhrn08@yahoo.com

Address of correspondence:

Muslima Ferdousi, MBBS, Medical Officer, INMAS, Faridpur Medical College Hospital Campus, Faridpur, Bangladesh. E-mail: muslimaferdousi@gmail.com

ultrasound system; Italy) equipped with C 1-8 MHz transducer. The USG revealed absent uterus and upper part of vagina with normal ovaries & left sided pelvic kidney (Figure 1A & 1B). However, for further evaluation, her upper abdomen ultrasound scan was done, and it revealed normal right kidney & empty left renal fossa (Fig. 2A & 2B). Her hormonal levels were all normal (Table 1). So, she was diagnosed as MRKH syndrome with ectopic left kidney that is MRKH syndrome type II. After giving proper counselling, we advised her to go to higher center for further management. She went to Bangladesh Medical University (BMU) & undergone vaginoplasty.

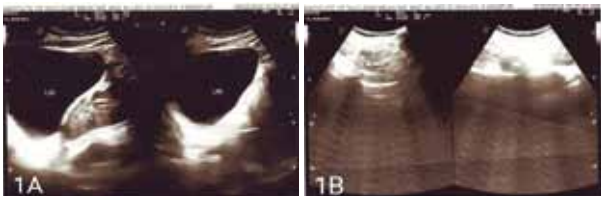

Figure 1: Transabdominal ultrasound image showing A) Urinary bladder (UB) with absent uterus & upper part of vagina along with left sided pelvic kidney (LK-Left Kidney), B) Normal both ovaries (RO- Right Ovary; LO- Left Ovary).

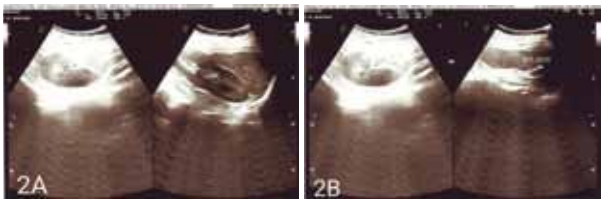

Figure 2: Transabdominal ultrasound image showing A) Normal right kidney in right renal fossa (RK) & ectopic healthy-looking left kidney (LK Ectopic), B) Empty left renal fossa (LRF).

Table 1: Hormone Analyses of patient.

Test	Result	Normal Range
FSH	5.63	Ovulatory Period: 5-23 IU/L
LH	4.1	Follicular Phase: 0.6-7.4 mIU/ml
		Ovulatory Peak: 12-61 mIU/ml
Prolactin	125.1	Female: 80-650 µIU/ml
Estradiol (E2)	149.2	Ovulation: 100-500 pg/ml
T4	9.72	8.56-25 fmol/ml
TSH	0.58	0.35-5.00 µIU/ml
Testosterone & FT3	Not done	

Discussion:

In normal female Mullerian ducts or paramesonephric ducts give rise to uterus, uterine tubes & upper part (2/3) of vagina.[6] In Mullerian agenesis or MRKH syndrome these ducts fail to develop resulting in aplasia of the uterus and the upper part of the vagina.[3] The uterus may be rudimentary as bilateral non-cannulated muscular buds.[7] The vagina is reduced to a more or less deep (2–7 cm) vaginal dimple as the lower portion of the vagina is derived from urogenital sinus, not from the Mullerian ducts.[6] Ovaries are present even when the Mullerian ducts are completely absent as ovaries do not develop from the Mullerian ducts rather from genital or gonadal ridges.[8]

Exact cause is unknown. Changes in several genes that are involved in development before birth have been identified in people with MRKHS. It has autosomal dominant mode of inheritance.[9]

Mullerian agenesis or MRKH syndrome is the second most common cause of primary amenorrhoea after Turner's syndrome[10] & the first clinical signal is generally a primary amenorrhea in patients presenting with a normal female phenotype[3], normal 46XX karyotype[4,5], and normal and functioning ovaries with no sign of androgen excess.[11,12] Patient may also complain painful coitus & cyclical abdominal pain.

This syndrome is subdivided in two types: when only genital system is involved it is classified as MRKH syndrome type I (isolated) or Rokitansky sequence & when other organs are also affected it is classified as MRKH syndrome type II or MURCS association (Mullerian duct aplasia, Renal dysplasia and Cervical Somite anomalies). Common associated malformations in MRKH syndrome type II (MURCS association) are as follows:

A. Associated renal malformations: Unilateral renal agenesis (23–28%), ectopia of one or both kidneys (17%), renal hypoplasia (4%), horseshoe kidney and hydronephrosis.[13,14] Found in about 40% of cases with MRKH syndrome.[15]

B. Associated skeletal abnormalities: Scoliosis (20%),[13] isolated vertebral anomalies (asymmetric, fused, or wedged vertebrae), Klippel-Feil association (fusion of at least two cervical segments, short neck, low hair line, restriction of neck motion)[16] and spina bifida etc.[17]

C. Associated hearing impairment: Conductive deafness due to middle ear malformations, such as stapedial ankylosis,[18] or sensorineural defects of varying severity.[19] Associated with 10 to 20% of MURCS patients.[18]

External examination reveals completed puberty with normal secondary female sexual characteristics (pubic hair and breast development are Tanner stage 5) and normal external genitalia. At the same time, the vagina is reduced to a vaginal dimple.

Transabdominal ultrasonography, magnetic resonance imaging (MRI) & laparoscopy help to confirm the diagnosis.[15]

The main differential diagnosis of Mullerian agenesis is testicular feminization syndrome or androgen insensitivity syndrome. The karyotype in this condition is, however, 46XY. In addition, the hormone profile in Mullerian agenesis will be typically that of a woman.[2,20]

Due to an underdeveloped or absent uterus, people with this condition cannot carry a pregnancy and therefore have uterine factor infertility (UFI). But healthy ovaries make it possible to have a biological child via assisted reproduction.

There are surgical and non-surgical options to treat MRKH:

1. Self-dilatation: it may help to expand the existing vagina over time without any surgery.

2. Vaginoplasty: surgeons can create a functional vagina using a skin graft. It helps to achieve & maintain functional vagina.

3. Uterus transplant: it has been performed in several people with MRKH syndrome. But the surgery is still in the experimental stage.

Conclusion:

MRKH syndrome can occur with normal endocrine function & secondary sexual characteristics. Ultrasonography & hormone analyses play a vital role in confirming the diagnosis. Surgical correction by creating a neovagina is a good treatment option in young females for her sexual life.

Conflict of interest: There is no conflict of interest.

Acknowledgment:

We express our heartfelt gratitude to the physician, who referred this patient. We would like to thank the patient & her family for their cooperation. We owe to our staffs of ultrasonography & in vitro divisions for their dedicated support. And lastly all praise to Allah for being with us.

References:

1. Griffin JE, Edwards C, Madden JD, Harrod MJ, Wilson JD. Congenital Absence of the Vagina: The Mayer-Rokitansky-Kuster-Hauser Syndrome. *Annals of Internal Medicine*. 1976;85(2): 224. DOI: 10.7326/0003 -4819-85-2-224

2. Folch M, Pigem I, Konje JC. Müllerian Agenesis: Etiology, Diagnosis, and Management. *Obstetrical & Gynecological Survey.* 2000;55(10): 644-649. DOI: 10.10 97/00006254-200010000-00023

3. Konar H. *DC Dutta's Textbook of Gynecology*. 8th ed. New Delhi: Jaypee Brothers Medical Publisher; 2020. p.380

4. Azoury RS, Jones HW Jr. Cytogenetic findings in patients with congenital absence of the vagina. *American Journal of Obstetrics & Gynecology*. 1966;94(02): 178-180. DOI: 10.1016/0002-9378(66)90460-1

5. Sarto GE. Cytogenetics of fifty patients with primary amenorrhea. *American Journal of Obstetrics & Gynaecology*. 1974;119(1): 14-23. DOI: 10.1016/0002-93 78(74)90312-3

6. Sadler TW. *Langman's Medical Embryology*. 14th ed. Philadelphia: Wolters Kluwer; 2019.p.272-274

7. Russ PD, Allen-Davis JT, Weingardt JP, Anderson MS, Koyle MA. Mayer-Rokitansky-Kuster-Hauser syndrome diagnosed by magnetic resonance imaging in a 15-year-old girl. *Journal of Pediatric Adolescent Gynecology*. 1997; 10(2): 89-92. DOI: 10.1016/s1083-318 8(97)70058-5

8. Sadler TW. *Langman's Medical Embryology*. 14th ed. Philadelphia: Wolters Kluwer; 2019.p.267-270

9. Opitz JM. Vaginal atresia (von Mayer-Rokitansky-Küster or MRK anomaly) in hereditary renal adysplasia (HRA). *American Journal of Medical Genetics*. 1987;26(4): 873-876. DOI: 10.1002/ajmg.1320260414

10. Konar H. *DC Dutta's Textbook of Gynecology*. 8th ed. New Delhi: Jaypee Brothers Medical Publisher; 2020.p.38

11. Fraser IS, Baird DT, Hobson BM, Michie EA, Hunter W. Cyclical Ovarian Function in Women with Congenital Absence of the Uterus and Vagina. *The Journal of Clinical Endocrinology & Metabolism*. 1973;36(4): 634–637. DOI: 10.1210/jcem-36-4-634

12. Shane JM, Wilson EA, Sciff I, Naftolin F. A preliminary report on gonadotropin responsivity in the Rokitansky-Küster-Hauser syndrome (congenitally absent uterus). *American Journal of Obstetrics & Gynecology*. 1977;127(3): 326-327. DOI: 10.1016/0002-9378(77)90 478-1

13. Pittock ST, Babovic-Vuksanovic D, Lteif A. Mayer–Rokitansky–Küster–Hauser anomaly and its associated malformations. *American Journal of Medical Genetics Part A*. 2005;135A(3): 314-316. DOI: 10.1002/ ajmg.a. 30721

14. 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 Reproduction*. 2006;21(3): 792–797. DOI: 10.10 93/humrep/dei381

15. Strubbe EH, Willemsen WN, Lemenns JA, Thijn CJ, Rolland R. Mayer-Rokitansky-Kuster-Hauser syndrome: distinction between two forms based on excretory urographic, sonographic, and laparoscopic findings. *American Journal of Roentgenology*. 1993;160(2): 331-334. DOI: 10.2214/ajr.160.2.8424345

16. Baird PA, Lowry RB. Absent vagina and the Klippel-Feil anomaly. *American Journal of Obstetrics & Gynecology*. 1974;118(2): 290-291. DOI: 10.1016/0002-9378(74)90 565-1

17. Strubbe EH, Lemenns JA, Thijn CJ, Willemsen WN, van Toor BS. Spinal abnormalities and the atypical form of the Mayer-Rokitansky-Küster-Hauser syndrome. *Skeletal Radiology*. 1992;21(7): 459-462. DOI: 10.1007/BF0019 0992

18. Cremers CW, Strubbe EH, Willemsen WN. Stapedial ankylosis in the Mayer-Rokitansky-Küster-Hauser syndrome. *Archives of Otolaryngology- Head & Neck Surgery*. 1995;121(7): 800-803. DOI: 10.1001/archotol.19 95.01890070086018

19. Letterie GS, Vauss N. Müllerian tract abnormalities and associated auditory defects. *The Journal of Reproductive Medicine*. 1991;36(11): 765-768.

20. Konar H. D*C Dutta's Textbook of Gynecology*. 8th ed. New Delhi: Jaypee Brothers Medical Publisher; 2020. p.373