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# Mayer Rokitansky Kuster Hauser (MRKH) Syndrome: A Case Series in Northeast India

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**Abstract**

**Background**

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a congenital disorder characterized by the absence or hypoplasia of the uterus and the upper two-thirds of the vagina, while ovarian function remains intact so as the famine nature. It is a rare disorder with an incidence of 1 in 5000 females. It is classified into two types: Type A, with isolated Müllerian duct anomalies, and Type B, which is associated with renal and/or skeletal abnormalities. Diagnosis is typically made during adolescence when patients present with primary amenorrhea. Radio-diagnosis can play a very important role in early diagnosis of this condition and thus helps in rehabilitation in very early age of women.

**Keywords:** Mayer-Rokitansky-Küster-Hauser Syndrome, Congenital Müllerian duct anomaly, Agenesis of uterus, Vaginal agenesis, Primary amenorrhea.

**CASE 1**

An 11-year-old girl presented with primary amenorrhea. Secondary sexual characteristics were in early stages of development, with Tanner stage II for breasts and Tanner stage I for pubic hair. There was no familial history of similar reproductive disorders.

On pelvic ultrasound there were absence of the uterus and upper two-thirds of the vagina. Ovarian structure was normal. A congenital absence of the uterus and upper vagina was confirmed, with a small vaginal remnant. No associated renal or skeletal anomalies were observed IN MRI. On karyotyping 46, XX—normal female karyotype seen.

On the basis of the above findings we diagnosed the case as type A Mayer-Rokitansky-Küster-Hauser Syndrome as there were no skeletal or renal abnormalities.

Psychological counselling was provided to support the patient in understanding her condition and its implications. A plan for vaginoplasty was established, with a goal of addressing the anatomical anomaly in the future. Fertility preservation options were discussed with the family, including egg preservation.

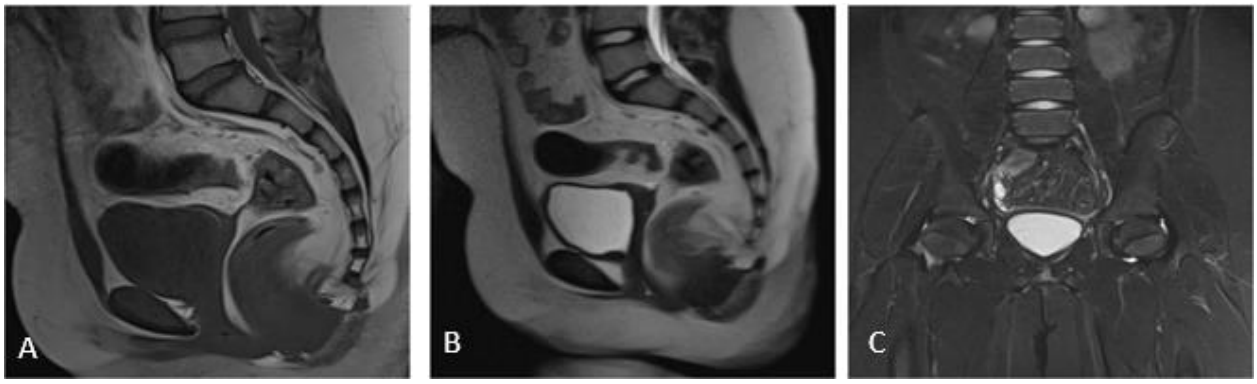


**Figure 1 (A) No uterus visible on T1WI sequence (B) Small remnant of lower part of vagina visible in T2WI sequence (C) Bilateral normal ovaries can be visualized in T2 FAT SAT COR sequence.**

**CASE 2**

A 15-year-old female presented with primary amenorrhea. She had normal secondary sexual characteristics (Tanner stage III for breasts, Tanner stage II for pubic hair). The patient was concerned about her lack of menstruation and potential reproductive issues.

In the initial USG whole abdomen, the uterus was not visualized. Ovary like structures seen in pelvic cavity. Following this patient was advised MRI which revealed absent uterus & upper 2/3 of vagina. No skeletal abnormalities are seen & no renal abnormalities were present.

On the basis of those findings we diagnose that case as as type A Mayer-Rokitansky-Küster-Hauser Syndrome.

Following diagnosis, fertility preservation options were discussed, including egg preservation. The patient was referred for vaginoplasty, and psychological counseling was initiated to address her concerns regarding sexual health and body image.

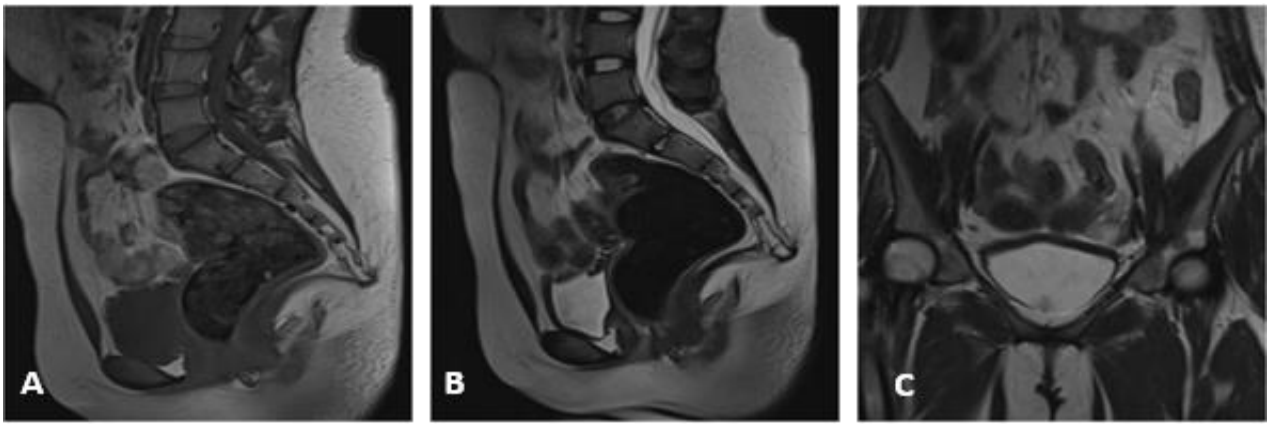


**Figure 2 (A) Uterus could not be visualized on T1WI SAG sequence (B) Lower part of vagina can be visible in T2WI SAG sequence (C) Bilateral normal ovaries can be visualized in T2 FAT SAT COR sequence.**

**CASE 3**

In our 3rd case we got an 18 years old unmarried women with symptoms of recurrent UTI. On taking detailed history we found that she is having primary amenorrhoea for which she was using some ayurvedic medicine.

On USG whole abdomen uterus was not visualized. More over the bilateral kidneys were found in the pelvic cavity, which were fixed together & malrotated. After that she was advised MRI where the uterus & upper 2/3 of vagina were absent. Bilateral kidneys were found in the pelvic cavity at L2-L4 level which were fused together with their hilum facing anteriorly. Bilateral ovaries were visualized as normal.
On the basis of above findings, we diagnosed this case as type B Mayer-Rokitansky-Küster-Hauser Syndrome.
Following this psychological counselling was initiated to address her concerns regarding sexual health & she was referred to nephrology for renal anomaly.

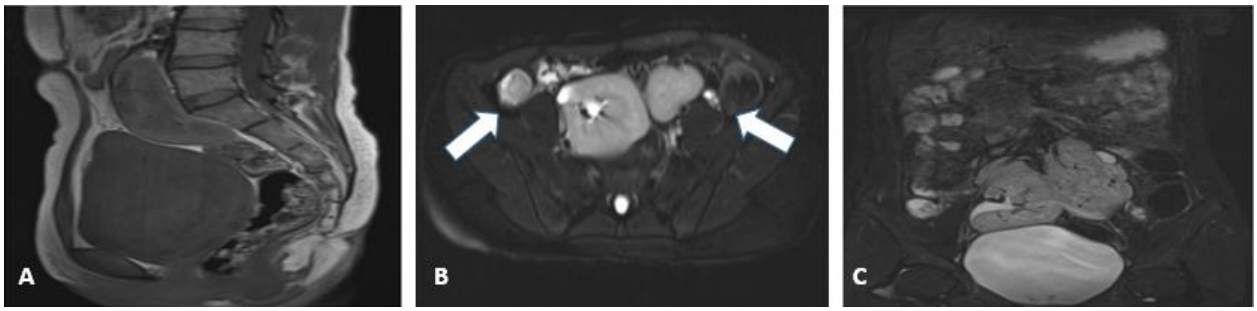


**Figure 3 (A) Uterus not be visualized on T1WI SAG sequence (B) Bilateral ovaries can be visualized in T2WI AXIAL FAT SAT sequence (C) Bilateral fused malpositioned & malroteted kidneys can be visualized in T2 FAT SAT COR sequence.**

## DISCUSSION

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a rare congenital disorder characterized by the partial or complete agenesis of the uterus and upper two-thirds of the vagina, while the external genitalia, ovarian function, and secondary sexual characteristics remain intact. MRKH syndrome is typically diagnosed in adolescence, when patients present with primary amenorrhea, as in the three cases described in this series. Although the condition is often associated with psychological distress due to its impact on reproductive health and body image, appropriate medical and psychological management can greatly enhance the quality of life of these patients.
MRKH syndrome is classified into two types: Type A (isolated form): The primary defect is confined to the Müllerian ducts, leading to agenesis or hypoplasia of the uterus and upper two-thirds of the vagina, without any associated malformations in other organs. Type B (syndromic form): In addition to the Müllerian duct malformation, Type B MRKH is often associated with renal anomalies (e.g., renal agenesis, horseshoe kidneys) and skeletal abnormalities (e.g., scoliosis, hemi vertebrae). It is hypothesized that Type B MRKH results from a disruption in the embryological development of the paramesonephric ducts and other structures during early fetal development.
The precise etiology of MRKH syndrome remains unclear, though it is believed to arise from a combination of genetic, environmental, and epigenetic factors. Studies have suggested a potential genetic predisposition, particularly involving mutations in genes responsible for Müllerian duct development, such as the **WNT4** and **LHX1** genes. However, the low incidence of MRKH and the variability in clinical presentation complicate the identification of specific genetic causes.
The hallmark presentation of MRKH syndrome is primary amenorrhea, typically in patients with otherwise normal puberty and secondary sexual development. As seen in all three of our cases, patients had normal breast and pubic hair development (Tanner stage II and III, respectively), which is suggestive

of intact ovarian function. In fact, ovarian function is typically preserved in MRKH syndrome, and affected individuals have normal levels of estrogen, which contributes to the development of secondary sexual characteristics. However, the absence of menstruation is often the first indication that prompts medical evaluation.

The diagnosis of MRKH syndrome is typically made through imaging studies. Pelvic ultrasound is usually the first modality to reveal the absence of the uterus and upper vagina, though it may not provide sufficient detail regarding the full extent of the malformation. MRI is the gold standard in confirming the diagnosis, as it provides detailed information about the uterine and vaginal anatomy and can assess for associated renal or skeletal anomalies, particularly in Type B cases. In our series, all patients were diagnosed with MRKH through a combination of pelvic ultrasound and MRI, which revealed characteristic findings such as the absence of the uterus and a short vaginal remnant. Furthermore, no renal or skeletal abnormalities were observed in our Type B patient, demonstrating that the absence of associated anomalies does not rule out a Type B diagnosis. Genetic testing, such as karyotyping, was also used in our patients to confirm their 46,XX status, ensuring that the patients had normal female chromosomes.

**Management and Treatment Options**

Management of MRKH syndrome is multidisciplinary, encompassing both medical and surgical aspects. As MRKH syndrome is a congenital anomaly, there is no cure, but various treatment options can help improve the quality of life for affected individuals.

While MRKH patients are unable to carry a pregnancy due to the absence of the uterus, ovarian function remains intact, and they may retain the ability to produce eggs. Fertility preservation, including egg freezing, is an important consideration for patients who wish to pursue biological children in the future. In our series, fertility preservation counseling was provided to all patients, with discussions on the possibility of gestational surrogacy as an option for future childbearing.

Vaginal reconstruction, or vaginoplasty, is often recommended to create a functional vaginal canal, allowing for sexual intercourse and improving quality of life. There are various surgical techniques for vaginoplasty, including the use of skin grafts, bowel segments, or the creation of a neo-vagina from the perineum. The timing of surgery is usually deferred until the patient has reached a level of emotional maturity and is ready for the procedure. Vaginoplasty was recommended for the patients in our cases, and future surgical interventions will be planned as they reach appropriate age and psychological readiness.

Comprehensive psychosocial care is crucial for MRKH patients. Patients often experience anxiety, depression, and body dysmorphia, and these issues need to be addressed through counseling and support. Furthermore, support groups and peer networks can provide a valuable source of understanding and shared experiences. The provision of emotional and psychological support throughout the course of treatment is key to ensuring optimal long-term outcomes.

**CONCLUSION**

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a rare but serious condition that can present as Type A or B. In our cases, the diagnosis was confirmed at the Department of Radio-Diagnosis, Agartala Government Medical College (AGMC) in 2023-24 through proper history USG, and MRI. Early diagnosis and counselling led to a favourable outcome for those patients. Those cases highlight the critical role of radiological imaging, particularly MRI in diagnosing this syndrome.

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