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Mayer-Rokitansky-Küster-Hauser Syndrome: MRI Findings from Three Young Adult Cases

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Abstract | **Case Series**

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome, also known as Müllerian aplasia, is a rare congenital disorder characterized by agenesis of the uterus and upper two-thirds of the vagina in 46, XX women with normal secondary sexual characteristics. Accurate classification of MRKH subtypes and identification of associated anomalies are essential for appropriate management. This study aimed to illustrate the clinical and MRI features of MRKH syndrome through three cases with different presentations. We retrospectively reviewed the clinical and MRI findings of three young adult females (aged 20–21 years) who presented with primary amenorrhea and normal external genitalia. All patients underwent pelvic ultrasound, followed by pelvic MRI for definitive diagnosis and evaluation of associated anomalies. In Case 1, MRI confirmed complete agenesis of the uterus and upper vagina with normal bilateral ovaries, consistent with typical MRKH (Type I). Case 2 demonstrated uterine agenesis with a left ectopic pelvic kidney and normal ovaries, consistent with atypical MRKH syndrome (type II). Case 3 revealed complete uterine and upper vaginal agenesis. No skeletal anomalies were observed. All patients had normal secondary sexual characteristics. These three cases highlight the clinical and radiological spectrum of MRKH syndrome and the central role of MRI in accurately characterizing Müllerian structures and their associated anomalies. Early diagnosis and clear classification are crucial for guiding multidisciplinary management and for patient counseling.

Keywords: MRKH syndrome, uterine agenesis, MRI, primary amenorrhea.

INTRODUCTION

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a rare congenital anomaly characterized by agenesis or severe hypoplasia of the uterus and upper two-thirds of the vagina in phenotypically normal females with a 46XX karyotype. It results from the arrested development of the Müllerian ducts during early embryogenesis. This condition is classified as a Class I Müllerian duct anomaly and occurs in approximately 1 in 4,000 to 5,000 live female births [1]. MRKH is divided into two types: type I (typical), which involves isolated uterovaginal agenesis with normal ovaries and fallopian tubes, and type II (atypical), which is associated with renal, skeletal, and auditory anomalies. Patients typically present during adolescence with primary amenorrhea despite having normal secondary sexual characteristics. MRI is the imaging modality of choice, enabling precise anatomical assessment and identification of associated malformations [2]. This case series aimed to illustrate the clinical and MRI features of MRKH syndrome through three distinct cases, demonstrating the diagnostic role of MRI and the variability in associated anomalies.

MATERIALS AND METHODS

We retrospectively reviewed three consecutive cases of young adult women (aged 20–21 years) presenting with primary amenorrhea between. All patients had normal development of secondary sexual characteristics and external genitalia. The initial evaluation included clinical examination and pelvic ultrasound. MRI of the pelvis was then performed in all patients for definitive diagnosis and assessment of associated anomalies. The MRI protocols included T2-weighted sequences in axial, sagittal, and coronal planes, with additional fat-saturated sequences when indicated. All patients provided informed consent for the use of anonymized data and images in this publication.

RESULTS

Case 1 (Typical MRKH – Type I)

A 20-year-old woman with no significant medical history presented with primary amenorrhea. Physical examination revealed Tanner stage V breast and pubic hair development, and normal external genitalia. Pelvic ultrasound did not visualize the uterus but revealed normal-appearing ovaries. Pelvic magnetic resonance imaging (MRI) confirmed complete agenesis of the uterus and upper vagina (Figure 1) and bilaterally normal ovaries (Figure 2). No renal or skeletal anomalies were observed.

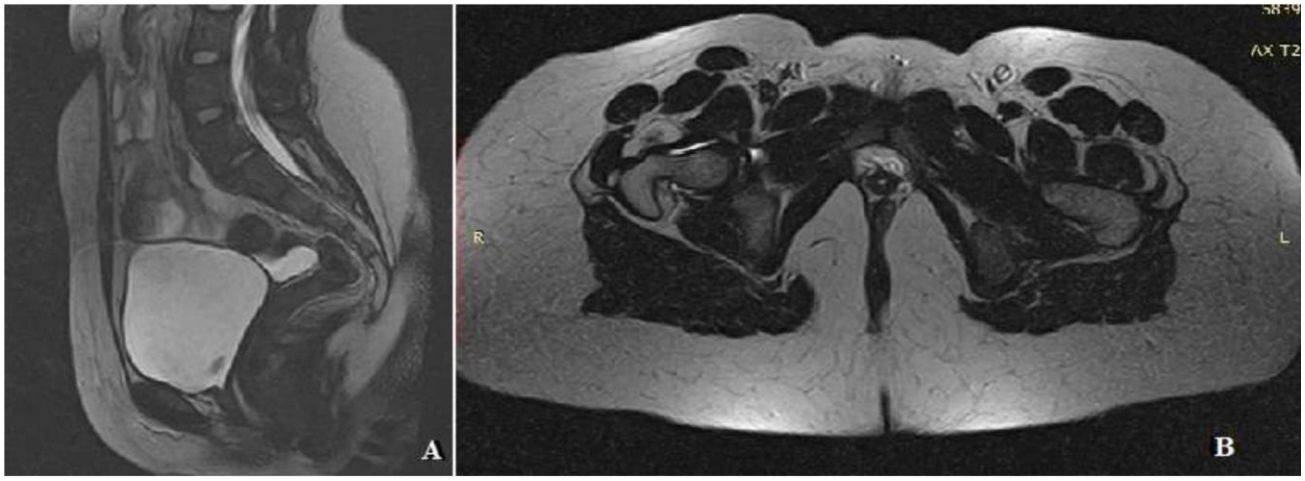

Figure 1: (A, B) Sagittal and axial T2-weighted MRI images from Case 1 showing agenesis of the uterus and upper vagina. A thin rim of nonspecific peritoneal fluid is also noted

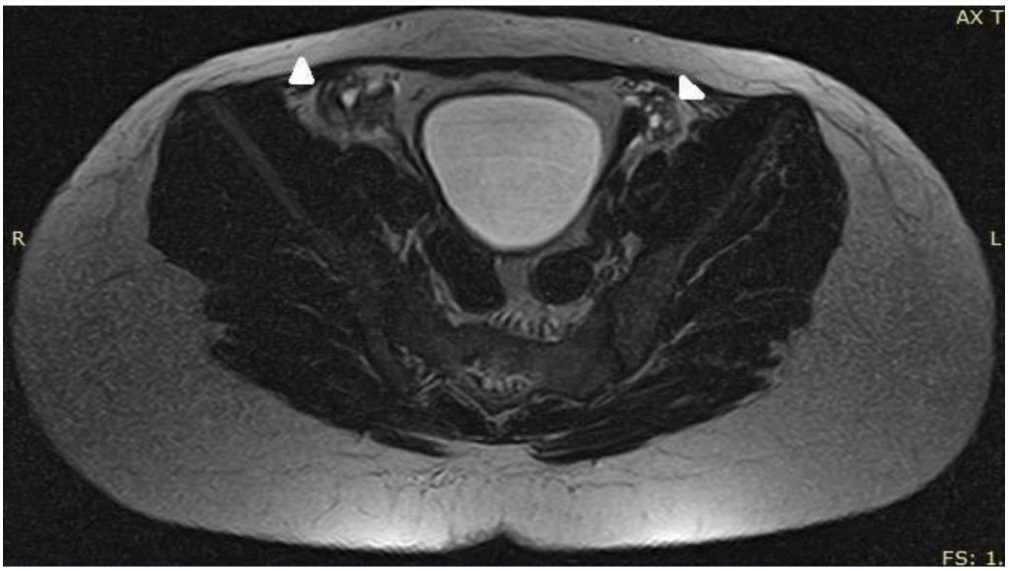

Figure 2: Axial T2-weighted MRI image from Case 1 showing both ovaries with normal signal intensity and preserved morphology (arrowheads). Both ovaries appear normal in signal and morphology

Case 2 (Atypical MRKH – Type II with Renal Anomaly)

A 20-year-old woman presented with similar complaints. Pelvic ultrasound revealed the absence of the uterus and a left pelvic ectopic kidney. MRI confirmed complete uterine agenesis (Figure 3), normal bilateral ovaries (Figure 4), and a left pelvic kidney with the right kidney in its normal position in the lumbar region (Figure 5). No skeletal anomalies were observed.

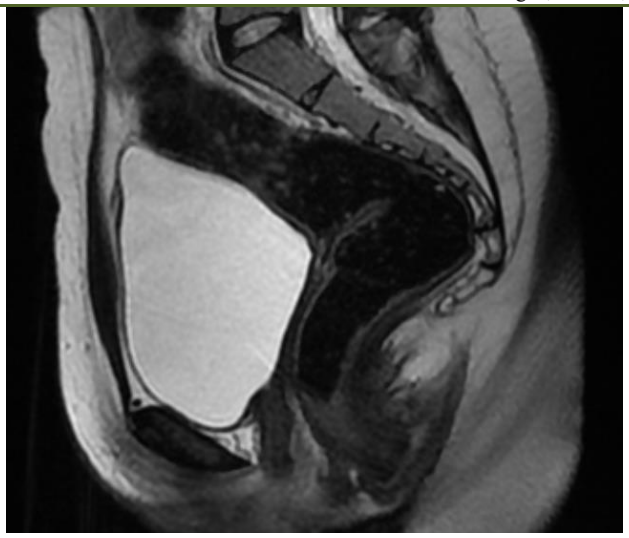

Figure 3: Sagittal T2-weighted MRI image from Case 2 demonstrating complete agenesis of the uterus and upper two-thirds of the vagina

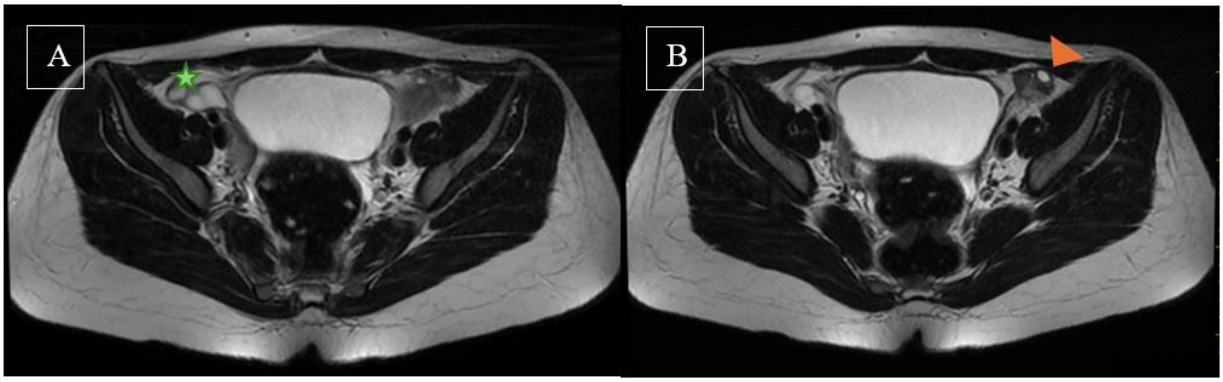

Figure 4: (A, B) Axial T2-weighted MRI images from Case 2 showing the right ovary (asterisk) in (A) and the left ovary (arrowhead) in (B). Both ovaries appear normal in signal and morphology

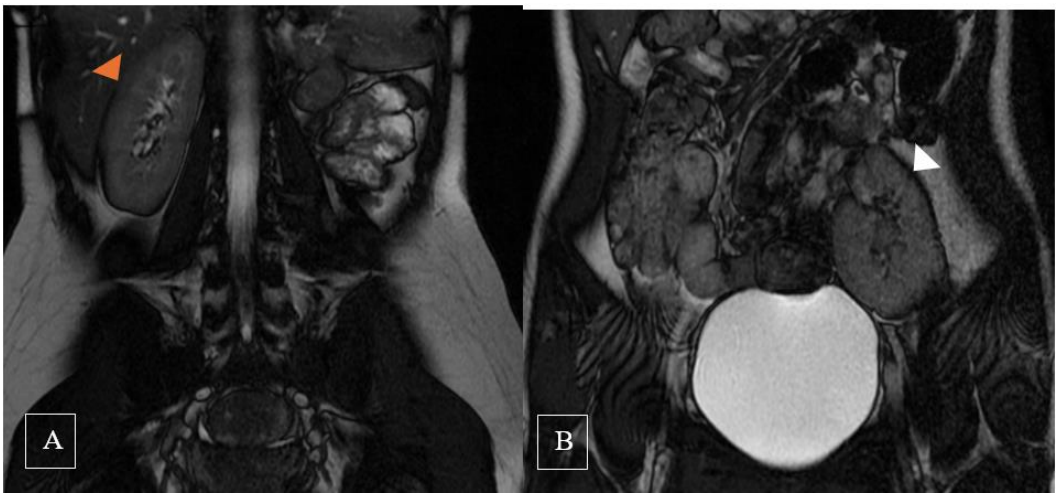

Figure 5: (A, B) Coronal 2D FIESTA MRI images from Case 2. (A) shows the right kidney in a normal lumbar position (orange arrowhead), and (B) shows the left kidney ectopically located in the pelvis (white arrowhead)

Case 3 (Atypical MRKH – Type II)

A 21-year-old female presented with primary amenorrhea. She had normal external genitalia and secondary sexual development. Pelvic ultrasound failed to visualize the uterus and clearly identify the ovaries of the patient. Pelvic MRI revealed complete agenesis of the uterus and upper two-thirds of the vagina (Figure 6), with normal ovaries bilaterally. No renal or skeletal anomalies were observed.

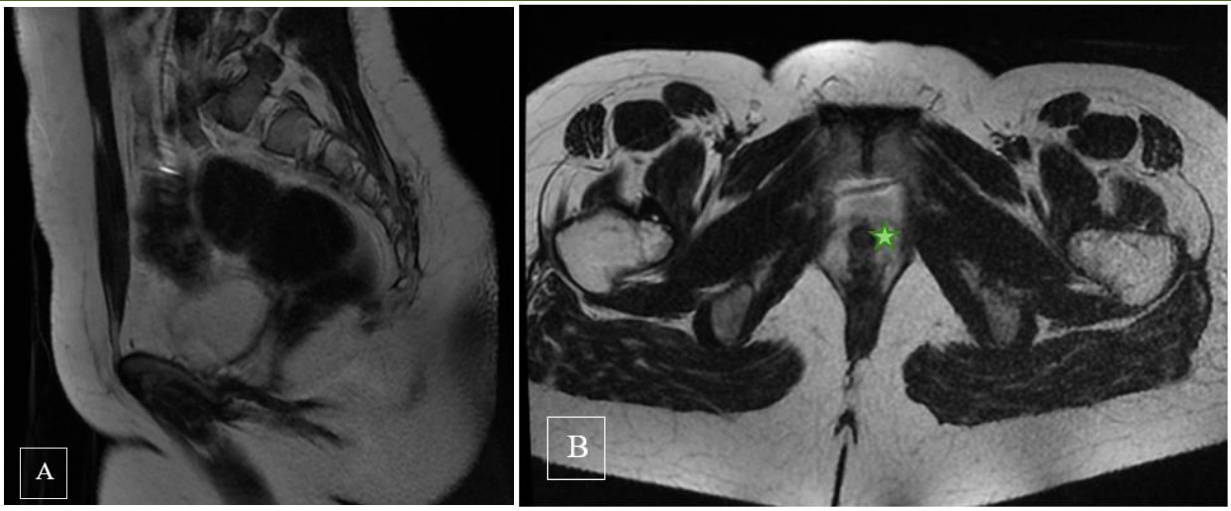

Figure 6: (A) Sagittal T2-weighted MRI image from Case 3 demonstrating complete agenesis of the uterus and upper two-thirds of the vagina. (B) Axial T2-weighted MRI image showing the hypoplastic vaginal remnant. (asterisk)

DISCUSSION

MRKH syndrome is the second most common cause of primary amenorrhea, after gonadal dysgenesis [1]. Embryologically, the paramesonephric ducts fail to develop or fuse during the 6–12 weeks of gestation. While the ovaries, derived from the genital ridge, develop normally and secrete estrogen, Müllerian structures (uterus, fallopian tubes, and upper vagina) are absent or rudimentary. Renal anomalies occur in 30–40% of cases and include renal agenesis, ectopic kidneys, and horseshoe kidneys. In our second patient, the diagnosis of a left pelvic ectopic kidney was made using MRI. The skeletal system can also be involved in 10–20% of cases, although no skeletal abnormalities were observed in either patient. Differential diagnoses include androgen insensitivity syndrome (AIS), in which 46, XY individuals present with similar external genitalia but have undescended testes and absent Müllerian structures. In contrast, patients with MRKH syndrome have a normal female karyotype and ovarian function [3]. MRI is the gold standard for MRKH diagnosis. It accurately identifies uterovaginal agenesis, characterizes the ovaries, and detects associated anomalies [4]. It is also helpful in identifying uterine remnants with or without functioning endometrium, which has implications for management in patients with pelvic pain. Management requires a multidisciplinary approach, including gynecology, radiology, endocrinology, psychology, and reproductive medicine. Non-surgical creation of a neovagina via progressive dilation is the first-line approach, with surgery reserved for refractory cases. Reproductive options remain limited but include surrogacy or uterine transplantation in selected centers [5].

CONCLUSION

MRKH syndrome is a rare but significant congenital anomaly with substantial clinical and psychological implications for patients. This condition should be considered in any adolescent female presenting with primary amenorrhea and normal secondary sexual characteristics. MRI is essential for confirming the diagnosis, classifying the subtype, and identifying associated anomalies that may impact management. These three cases highlight the spectrum of clinical and imaging findings and the importance of a comprehensive, patient-centered approach.

Research Highlights

- MRI is the preferred imaging modality for diagnosing MRKH and identifying associated anomalies.
- Case series includes both typical and atypical presentations, with rare associations like pelvic ectopic kidney.
- Highlights the importance of early diagnosis and multidisciplinary care.

Limitations

- Small sample size limits generalizability.
- Retrospective design without genetic confirmation for all patients.
- No long-term follow-up data.

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Funding and Policy Aspects

No funding was received for this study. The authors declare no financial or policy-related conflicts.

Abbreviations

MRKH – Mayer-Rokitansky-Küster-Hauser
MRI – Magnetic Resonance Imaging
AIS – Androgen Insensitivity Syndrome

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