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Case Report

Pure Mayer-Rokitansky-Küster-Hauser Syndrome in a 22-Year-Old Woman: A Radiological Case Report

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Received: October 22, 2025; **Published:** November 07, 2025

Abstract

Background: Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a congenital anomaly characterized by agenesis of the uterus and upper vagina in women with a normal 46,XX karyotype. It often coexists with renal or skeletal malformations, but the isolated, or pure, form is rare.

Case Presentation: We report the case of a 22-year-old woman presenting with primary amenorrhea and normal secondary sexual characteristics. Pelvic MRI revealed complete uterine and upper vaginal agenesis, with normal ovaries and kidneys.

Conclusion: This case highlights the crucial role of MRI in diagnosing MRKH and differentiating the pure form from syndromic variants. Multidisciplinary care ensures accurate diagnosis, counseling, and appropriate reproductive management.

Keywords: *MRKH; Rokitansky Syndrome; Primary Amenorrhea; MRI; Uterine Agenesis*

Introduction

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a congenital malformation resulting from the failure of Müllerian duct development, leading to partial or complete uterovaginal agenesis in phenotypically and chromosomally normal females (46,XX) [1]. It affects approximately 1 in 4,500–5,000 women [2].

Two forms are recognized: the type I (pure) or isolated form, limited to Müllerian agenesis, and the type II (syndromic) form, associated with renal, skeletal, auditory, or cardiac anomalies [3].

MRI is considered the gold standard for diagnosis because it offers multiplanar evaluation and high soft-tissue contrast, allowing for detailed visualization of pelvic anatomy and associated anomalies [4].

We report a case of pure MRKH syndrome diagnosed in a 22-year-old woman, emphasizing MRI findings and recent insights into its etiology and management.

Case Presentation

A 22-year-old woman was referred to our department for investigation of primary amenorrhea. She had normal pubertal development, including breast and pubic hair growth. There was no history of pelvic pain or urinary symptoms.

Physical examination revealed normal external genitalia and a short, blind-ending vaginal pouch. No palpable uterus was detected on bimanual examination. Hormonal assays (FSH, LH, estradiol, prolactin, and androgens) were within normal limits for age and pubertal status. Karyotype confirmed a normal 46,XX pattern.

MRI findings

Pelvic MRI was performed using T1- and T2-weighted sequences in sagittal, axial, and coronal planes.

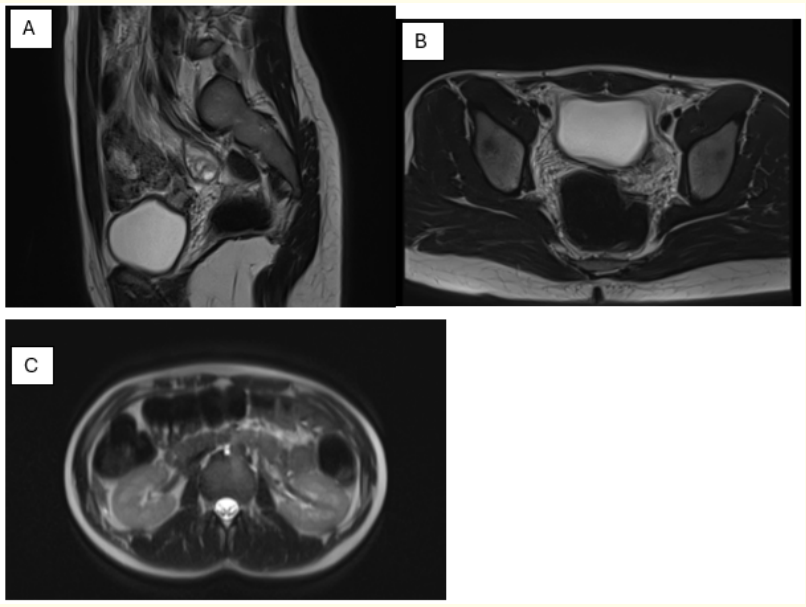

Figure A-C: *A: Sagittal T2-weighted MRI showing complete uterine and upper vaginal agenesis with a short distal vaginal remnant posterior to the bladder. B: Axial T2-weighted pelvic MRI demonstrating bilateral normal ovaries with no identifiable uterine structure. C: Axial T2-weighted abdominal MRI confirming normal kidneys in position and morphology, ruling out renal malformations.*

These findings were consistent with a pure (type I) form of MRKH syndrome.

The patient received psychological counseling and was informed about non-surgical and surgical options for vaginal reconstruction. She was also referred for fertility counseling regarding assisted reproductive possibilities (oocyte retrieval and gestational surrogacy, where legally available).

Discussion

MRKH syndrome results from disrupted embryonic development of the Müllerian ducts between the 5th and 7th gestational weeks [5]. The pure form represents around 60% of cases, while the remainder exhibit additional renal or skeletal anomalies [6].

Imaging role

MRI provides a comprehensive evaluation of Müllerian structures, ovarian morphology, and associated anomalies [7]. It is preferred over ultrasound due to its multiplanar capability and superior soft-tissue contrast, enabling confident diagnosis and differentiation of pure versus syndromic MRKH.

In our case, MRI confirmed complete uterovaginal agenesis with normal ovaries and kidneys—key criteria defining the pure form.

Genetic insights

Recent studies have identified several candidate genes involved in MRKH pathogenesis, including GREB1L, PAX8, and regions on 17q12 and 16p11.2, suggesting a heterogeneous genetic background [8]. These findings highlight the potential for future molecular classification and genetic counseling.

Management

Treatment aims to create a functional vagina to allow normal sexual activity and address psychological well-being. Non-surgical vaginal dilation (Frank method) remains the first-line approach due to its safety and success rate [9]. Surgical vaginoplasty (Vecchietti or McIndoe procedures) is reserved for selected cases.

Regarding fertility, MRKH patients can conceive via *in-vitro* fertilization (IVF) with oocyte retrieval and gestational surrogacy, or through uterine transplantation, an experimental option in select centers [10].

Conclusion

Pure MRKH syndrome should be considered in any woman with primary amenorrhea and normal secondary sexual characteristics.

Pelvic MRI plays a pivotal role in diagnosis, confirming the absence of the uterus and vagina while assessing the ovaries and kidneys.

Recognizing the pure form has significant implications for genetic counseling, reproductive planning, and psychosocial support.

Learning Points

- Pelvic MRI is the gold standard for diagnosing MRKH syndrome and assessing associated anomalies.
- The absence of renal and skeletal anomalies confirms the pure (type I) form.
- Early psychological and reproductive counseling are crucial for optimal outcomes.

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Volume 8 Issue 12 December 2025