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MAYER-ROKITANSKY-KÜSTER-HAUSER SYNDROME WITH PELVIC RENAL ECTOPIA: AN MRI-BASED DIAGNOSIS OF A RARE CONGENITAL ASSOCIATION

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ABSTRACT

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a rare congenital disorder characterized by aplasia of the uterus and vagina in phenotypically female individuals with a normal 46,XX karyotype and functional ovaries. We present the case of a 27-year-old woman referred for primary amenorrhea. This case is particularly instructive due to its unusually delayed diagnosis in adulthood, where pelvic magnetic resonance imaging (MRI) demonstrated a complete absence of the uterus and vagina, bilaterally small but functional ovaries, and an incidental finding of right pelvic renal ectopia. While the association between MRKH syndrome and renal anomalies is clinically recognized, this case highlights important diagnostic challenges, illustrating how multiplanar MRI serves as a critical secondary modality to overcome the limitations of initial pelvic ultrasound, allowing simultaneous delineation of reproductive tract anomalies and exact localization of pelvic ectopic tissue. This report underscores the educational value of systematic screening for concomitant urological anomalies and provides a framework for multidisciplinary counseling.

KEYWORDS

MRKH syndrome, Müllerian aplasia, Pelvic renal ectopia, Primary amenorrhea, Pelvic MRI

MAIN ARTICLE

INTRODUCTION

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a congenital condition with an estimated prevalence of approximately 1 in 4,500 phenotypically female live births [1, 2]. It is defined by congenital aplasia of the uterus and the upper two-thirds of the vagina in individuals with a normal female karyotype (46,XX), normal secondary sexual characteristics, and functional ovaries [1, 3]. Clinically, the syndrome typically manifests during adolescence as primary amenorrhea — the most common complaint prompting medical evaluation — in an otherwise hormonally intact young woman [2, 4].

MRKH syndrome is classically divided into two subtypes. Type I (isolated form) encompasses isolated müllerian aplasia without additional malformations. Type II (MURCS association) involves extra-genital anomalies, most notably renal malformations — including renal agenesis, horseshoe kidney, and renal ectopia — as well as skeletal defects and, less frequently, cardiac or auditory anomalies [3, 8]. The pathogenesis of MRKH syndrome remains incompletely elucidated; current evidence suggests a multifactorial etiology involving disruption of müllerian duct development during embryogenesis, with both genetic and epigenetic contributors [9].

In clinical practice, transabdominal or transvaginal ultrasound constitutes the indispensable first-line imaging modality for evaluating primary amenorrhea due to its accessibility and non-invasive nature. However, magnetic resonance imaging (MRI) has emerged as a preferred secondary imaging modality for the definitive diagnosis and precise classification of complex müllerian anomalies [5]. Thanks to its superior soft-tissue contrast resolution and multiplanar capability, MRI provides detailed characterization when ultrasound findings are ambiguous or limited by anatomical constraints. This report describes the clinical and imaging findings in a 27-year-old woman presenting with an unusually delayed diagnosis of MRKH syndrome, emphasizing the added value of MRI in identifying a concurrent right pelvic renal ectopia and illustrating the importance of comprehensive anatomical evaluation to prevent diagnostic pitfalls.

CASE PRESENTATION :

A 27-year-old phenotypically female patient was referred to the radiology department for pelvic MRI following an initial evaluation for primary amenorrhea. Her medical history was unremarkable, with no prior surgery, no family history of congenital anomalies, and no significant comorbidities. On physical examination, secondary sexual characteristics — including breast development and pubic hair — were fully developed and appropriate for her age. Inspection of the external genitalia revealed normal structures; however, specialized gynecologic examination demonstrated a short, blind-ending vaginal pouch with an severely restricted vaginal introitus. The patient reported no history of urinary symptoms, recurrent urinary tract infections, or cyclic pelvic pain. Systematic clinical screening revealed no signs of skeletal abnormalities or cardiac dysfunction.

Laboratory investigations revealed normal serum levels of follicle-stimulating hormone (FSH), luteinizing hormone (LH), and estradiol, consistent with preserved ovarian function. Karyotyping confirmed a normal 46,XX female karyotype. Serum testosterone levels were within normal female reference ranges, effectively excluding androgen insensitivity syndrome as a differential diagnosis. Furthermore, routine serum renal function tests (creatinine and blood urea nitrogen) were strictly within normal limits.

An initial pelvic ultrasound raised suspicion of uterine agenesis but was limited in fully characterizing the pelvic anatomy and adnexa due to overlying bowel gas and the patient's retroverted pelvic presentation. Consequently, a pelvic MRI was performed on a 1.5 Tesla system using standard pelvic coil sequences, including T2-weighted images in the axial, coronal, and sagittal planes, supplemented by T1-weighted sequences. The key imaging findings are described as follows:

1. Uterine agenesis: No uterine tissue was identified in the pelvis. Neither uterine body nor cervix was visualized on any imaging plane (Figure 1). The typical T2-hypointense junctional zone and the endometrial stripe were entirely absent.
2. Vaginal aplasia: No vaginal canal was discernible. The region between the bladder and rectum was occupied by adipose tissue without any identifiable tubular structure corresponding to a vaginal lumen (Figure 1).

3. Ovaries: Both ovaries were identified within the pelvis, demonstrating normal follicular signal characteristics on T2-weighted sequences. However, ovarian volumes were bilaterally reduced, measuring approximately 2 cm^3 on the right and 1,8 cm^3 on the left (normal reference: 4–14 cm^3 in reproductive-age women). No adnexal mass, hemorrhagic cyst, or torsion was identified.

4. Right pelvic renal ectopia: The right kidney was absent from its expected retroperitoneal location. It was instead identified in the right hemipelvis, anterior to the iliac vessels, displaying preserved corticomedullary differentiation and normal parenchymal signal intensity on all sequences (Figure 2). No hydronephrosis or perinephric collection was observed. The left kidney occupied its normal position and demonstrated unremarkable morphology.

DISCUSSION

The present case exemplifies a classic clinical and imaging constellation of MRKH syndrome type II (MURCS association), characterized by müllerian aplasia associated with a renal anomaly. What makes this case highly instructive is the unusually delayed diagnosis at age 27 in an asymptomatic patient, reinforcing the need for clinicians to remain vigilant when primary amenorrhea is neglected during adolescence. The diagnosis of MRKH syndrome must be distinguished from complete androgen insensitivity syndrome (CAIS), which shares several clinical features. Key differentiating features include the presence of a 46,XY karyotype and the absence of axillary and pubic hair in CAIS, as well as elevated serum testosterone into the male reference range — none of which were present in our patient [3].

While pelvic ultrasound is the universally accepted first-line screening tool for primary amenorrhea, it can sometimes yield incomplete assessments due to technical limitations like intestinal gas or atypical ectopic structures. In this scenario, MRI provides definitive diagnostic value. It serves as an excellent problem-solving tool by clearly differentiating retroperitoneal structures and mapping out the pelvis without radiation. In our patient, MRI not only confirmed the definitive absence of the uterus and upper vagina but also provided unambiguous characterization of the right pelvic renal ectopia, confirming its preserved parenchymal integrity and distinguishing it from an adnexal mass.

The embryological basis of the MRKH-renal ectopia association is well-established. Müllerian (paramesonephric) duct development is closely associated with the mesonephric (Wolffian) duct, which serves as an inducer of müllerian duct formation and guides its caudal migration. The metanephric blastema, which gives rise to the definitive kidney, undergoes cranial migration during normal embryogenesis. Disruption of these shared developmental pathways during the fourth to eighth weeks of gestation can result in concurrent müllerian aplasia and renal malpositions, explaining the well-documented association between MRKH syndrome and urological anomalies [7, 8]. Renal anomalies occur in approximately 30–40% of MRKH cases [2, 7]. Because this coexistence is clinically recognized rather than exceptional, finding any müllerian defect must always trigger a systematic evaluation of the upper urinary tract to prevent long-term silent complications.

The finding of bilaterally small ovarian volumes in our patient, in the context of normal hormonal values and fully developed secondary sexual characteristics, is an interesting feature. While some authors historically suggested that reduced volume might reflect subtle mesodermal variations, this finding should be interpreted cautiously. Given that the patient's hormonal profile and gonadotropin levels are entirely normal, these measurements are highly likely to represent normal physiological variation, early-stage baseline follicular dynamics, or minor measurement variations inherent to MRI multiplanar tracking rather than a profound structural gonadogenesis defect.

From a management standpoint, the diagnosis of MRKH syndrome carries profound psychological and reproductive implications, especially when discovered later in adulthood. Multidisciplinary care — encompassing gynecology, psychology, urology, and genetics — is essential [4]. Options for managing the short vaginal pouch, including progressive vaginal dilation (Frank method or Ingram technique) and surgical approaches (Vecchietti procedure, McIndoe technique, or laparoscopic neovaginoplasty), should be discussed with the patient in a sensitive, individualized manner [6]. Although uterine aplasia precludes natural conception, the preservation of normal ovarian function opens the possibility of assisted reproduction via oocyte retrieval and surrogacy where legally and ethically permitted. The concurrent pelvic renal ectopia, which remains functionally intact with normal renal laboratory values, warrants routine conservative urological follow-up to monitor for urinary tract infections or urinary stasis.

CONCLUSION

This case illustrates the educational value of pelvic MRI as an essential secondary modality in the comprehensive evaluation of MRKH syndrome, particularly when diagnosis is delayed into adulthood. MRI overcomes the regular limitations of first-line ultrasound, enabling precise characterization of müllerian aplasia and simultaneous detection of associated urological anomalies such as pelvic renal ectopia. In any patient presenting with primary amenorrhea, a systematic screening of the urinary tract is mandatory, and a coordinated multidisciplinary approach remains the cornerstone of management to address the anatomical, reproductive, and psychological dimensions of this condition.

FIGURES:

Figure 1. Sagittal T2-weighted MRI of the pelvis demonstrating complete absence of the uterus and vaginal canal. Adipose tissue occupies the vesico-rectal space. The bladder (B) and rectum (R) are labeled for anatomical orientation.

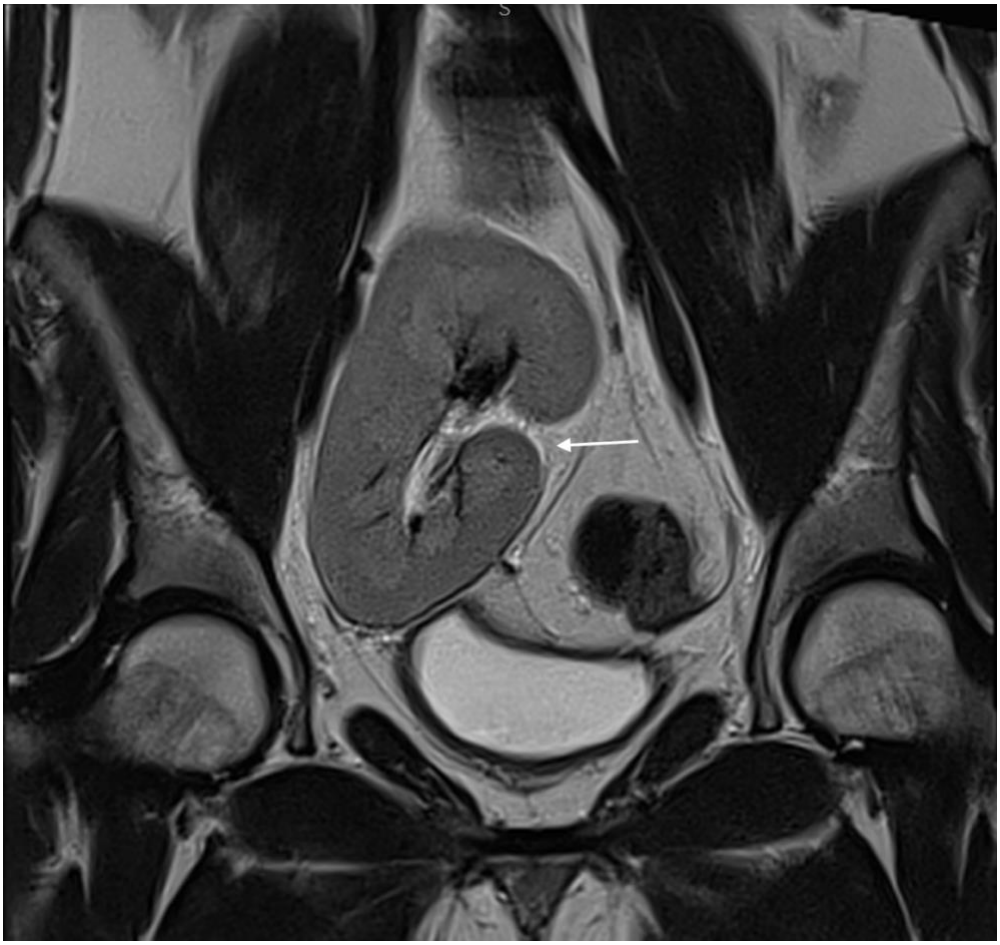

Figure 2. Coronal T2-weighted MRI demonstrating the right kidney (arrow) in an ectopic pelvic position, anterior to the right iliac vessels. Normal corticomedullary differentiation is preserved. The left kidney occupies a normal retroperitoneal position.

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