Magnetic Resonance Imaging Findings in Mayer-Rokitansky-Küster-Hauser (MRKH) Syndrome: A Report of Two Cases

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INTRODUCTION

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome, commonly known as Müllerian duct agenesis, is an uncommon congenital condition. MRKH syndrome has a prevalence of 1 in 4000 women and ranks as the second most frequent cause of primary amenorrhea. This condition arises from the incomplete development of the Müllerian ducts. The primary characteristic of this disease is the lack or limited growth of the uterus and upper two-thirds of the vagina (resulting in a missing or shallow vaginal pouch), while maintaining normal external genitalia, functioning ovaries, and a normal karyotype for women (46, XX).¹⁻³

MRKH syndrome is classified into two distinct categories. The usual form, known as Type I, is distinguished by abnormalities that are confined to the reproductive system. On the other hand, the atypical form, referred to as Type II, is accompanied by non-gynecological anomalies in the urological, skeletal, vertebral, hearing, or cardiac systems. Some urological abnormalities often seen in individuals with type-II MRKH syndrome include ectopic kidney, horseshoe kidney, and sometimes unilateral renal agenesis. Anomalies in the musculoskeletal system include a spectrum of abnormalities, including fused vertebral segments, scoliosis, and irregularities in the radius, carpals, phalanges, and femoral head epiphysis.¹⁻³

The etiology of MRKH syndrome remains uncertain. The correct development of the Müllerian ducts, kidneys, and bone structure is regulated by many genes. Estimates suggest that irregularities in the expression of the HOXA and WNT4 genes are the primary factors contributing to the developmental defects of female internal genitalia.¹⁻⁵

The MRKH condition has a substantial impact on both fertility and the physical and emotional well-being of women. Thoroughly examining the intricate structure of the patient’s reproductive system is crucial in order to determine the diagnosis and develop an appropriate treatment strategy accurately. Consequently, MRI is the preferred method that does not need invasive procedures, enabling exact assessment of uterine aplasia and enhanced imaging of the rudimentary horn and ovaries. We describe two exceptional examples of MRKH syndrome in girls who had primary amenorrhea and intact secondary sexual features.⁶ This case report aims to elaborate on MRI findings in females with typical and atypical types of MRKH syndrome.

CASE DESCRIPTION

Case 1
A female patient, aged 18, was sent to the radiology department to investigate...
primary amenorrhea. The secondary sexual traits seemed to be within the expected range of development. The patient has no history of marriage or pregnancy. There was no familial precedent of a comparable ailment. The patient had undergone a menstrual stimulation test administered by a doctor in the past. However, she did not thereafter have menstruation.

Upon initial physical examination, the weight was measured at 41 kg, and the height was 155 cm. Secondary sexual characteristics showed Tanner stage M5P3 with axillary and pubic hair appropriate for her age. External genitalia, labia majora, and minora with intact hymen were visible. Digital vaginal and speculum examinations were not performed because of virginity.

Normal plasma testosterone levels (36.56 ng/dL) indicated no signs of endocrine hyperandrogenism. The karyotyping report states 46 XX. A transrectal ultrasound examination showed no uterus.

The patient then underwent an abdominal-pelvic MRI, which showed there were no visible structures of the cervix, uterus, or upper 2/3 of the vagina. A short tubular blind end structure resembling a vagina, visualized between the lower part of the bladder and rectum, measured +/- 2.9 cm long (average vaginal length: 7-10 cm) (Figure 1a). Both ovaries were found in the pelvic cavity and contained sub-centimetres follicles (Figure 1b & 1c). Both kidneys were in their usual location. There was no visible soft tissue structure suggestive of Müllerian remnant. This examination then concluded as agensis of the uterus and proximal vagina, without gynaecological anomalies, which supported the diagnosis of type I Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome.

Case 2

A female patient, aged 26 years, was referred for primary amenorrhea. The patient has never had menstruation. Breasts and pubic hair have appeared normal. The patient has never been married or pregnant but already had a history of sexual intercourse. The patient had previously been treated with hormonal therapy for one cycle, but no menstrual bleeding occurred. Family history of the same complaint was denied. During pregnancy, the patient’s mother also did not take teratogenic medication.

Upon initial physical examination, the body weight was measured at 48 kg, and the height was 153 cm. There is no impression of a man’s face or an Adam’s apple. Secondary sexual characteristics showed Tanner stage M5P5 with axillary and pubic hair appropriate for her age. Genitalia external examination showed normal labia majora/minora, vaginal orifice, and urethra. A digital vaginal examination revealed a shallow blind-end vagina.

Transvaginal ultrasound examination failed to evaluate the existence of the uterus. Testosterone laboratory results were normal (25.22 ng/dL). Complete blood count and kidney function tests were within normal limits (BUN 9.4 mg/dL, creatinine 0.81 mg/dL, EGFR 100.23). The karyotyping report states 46 XX.

Whole Abdomen MRI with contrast Gadolinium showed the absence of normal morphology of the uterus, cervix, and upper two third of the vagina between the bladder and rectum. A short blind end vaginal structure was visible, measured +/- 4.3 cm long (N: 7-10 cm) (Figure 2). Both ovaries were displaced superolateral to the pelvic sidewall (Figure 3 & 4). The right kidney is of normal size, without dilatation of the pelvicalyceal system.
No left renal structures were found in the left renal fossa (Figure 5). The other abdominal organs and bones exhibit no abnormalities. The examination revealed the absence of the uterus and proximal vagina, along with the misplacement of both ovaries and the absence of the left kidney. These findings corroborate the diagnosis of type II Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome.

**DISCUSSION**

The MRKH syndrome was first documented by Mayer in 1829 and Rokitansky in 1838. In 1961, Hauser and Schreiner identified distinguishing characteristics that set MRKH syndrome apart from androgen insensitivity syndrome. In 2021, the American Society of Reproductive Medicine released an updated categorization of Müllerian duct abnormalities. Instead of using numbers, the anomalies are now defined using descriptive terms, with one such phrase being Müllerian agenesis.$^1,7$

Patients with a typical female appearance, normal 46 XX genetic makeup, and healthy ovaries without symptoms of excessive male hormones often have primary amenorrhea. The physical examination revealed full puberty with distinctive female secondary sexual traits (pubic hair and breast growth at Tanner stage 5) and normal external genitalia. However, the depth of the vagina decreases, resulting in the formation of a vaginal dimple of 2-7 cm.$^6$

Transabdominal ultrasonography (US) is a simple and non-invasive technique that should be used as the first assessment for individuals suspected of having Müllerian duct agenesis. The ultrasound reveals the lack of uterine tissues in the space between the bladder and the rectum in the United States. The United States should also ascertain the presence of any renal abnormalities.$^6$

MRI is a diagnostic procedure that uses advanced imaging techniques to examine the body without the need for intrusive measures. It is known to have a higher level of sensitivity and specificity compared to ultrasound. Magnetic resonance imaging (MRI) is recommended in cases when ultrasound results are equivocal or partial because of the challenges in accurately seeing uterine tissues, Müllerian rudiments, or ovaries. MRI enables precise assessment of uterine aplasia and enhanced visibility of the rudimentary horn and ovaries. The optimal visualization of uterine aplasia is achieved with a sagittal scan, whereas
vaginal aplasia is most effectively seen using a transverse imaging. The uterine remnant is seen as a dense, elongated ovoid structure on the T1-WI sequence. It has an intensity that is isointense to hypointense and forms a target pattern on T2-WI. The assessment also includes the examination of the existence, structure, and position of the ovaries and any ovaries located outside the pelvic region. The signal intensity on T2-weighted imaging shows a combination of hyperintense ovarian follicles. The vaginal canal may be clearly seen on sagittal T2-weighted imaging (T2-WI), axial T2-WI, and 3D SPACE. It appears as a dark structure with the urethra and bladder neck in front and the rectum behind it. In addition, MRI might potentially reveal concomitant renal and skeletal abnormalities.\(^1,6\)

The differential diagnosis of MRKH syndrome is androgen insensitivity syndrome (AIS). AIS is an X-linked recessive disorder in which affected males have female external genitalia, with female breast development, an occluded vagina, no uterus and female adnexa, and abdominal or inguinal testicles. Karyotype examination exhibits genotype 46 XY. Other differential diagnoses are isolated vaginal atresia and WNT4 defect.\(^1,6\)

The psychological well-being of patients is significantly impacted by MRKH syndrome, due to its effects on sexual relationships and fertility. The management of MRKH syndrome requires a comprehensive strategy including gynecologists, radiologists, clinical specialists, psychologists, and, if available, other professionals from relevant fields. Initially, the primary objective in managing people with MRKH syndrome is to construct a neo-vagina that is both functional and of sufficient size to facilitate sexual intercourse. This therapy is only provided to patients who have reached a state of readiness to engage in sexual activity and have also attained emotional maturity. The therapy may be either surgical or non-surgical, but the selected approach must be customized to suit the specific requirements of the person, the patient’s level of motivation, and the available choices. It is important to grasp the fact that they are unable to conceive via natural means. Presently, there are accessible choices for fertilization help, including assisted reproduction techniques, surrogate pregnancy, and uterine transplantation.\(^1,3,9,10\)

**CONCLUSION**

MRKH syndrome is a congenital disorder with varying degrees of urovaginal agenesis and functional ovaries. Radiological imaging is essential in the diagnosis, classification, and surgical planning of MRKH syndrome. Due to excellent soft tissue resolution, MRI is currently the best imaging technique for accurately evaluating pelvic anatomy, uterine aplasia, uterus development, and location of the ovaries. Identifying normal and functional ovaries is essential to differentiate MRKH syndrome from another differential diagnosis. The presence of MRKH should provide alertness to look for other non-gynaecological disorders.

**CONFLICT OF INTEREST**

The writers confirm that they possess no conflict of interest.

**RESEARCH ETHICS**

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**REFERENCES**