Current Research in MRI

Review Article

Magnetic Resonance Imaging Findings in Mayer–Rokitansky–Kuster–Hauser Syndrome: A Review Article

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Cite this article as: Kadirhan Ö, Fatihoğlu E. Magnetic resonance imaging findings in Mayer–Rokitansky–Kuster–Hauser syndrome: A review article. *Current Research in MRI.* 2022; 1(2): 24-26.

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Received: July 24, 2022 **Accepted:** September 16, 2022

DOI:10.5152/CurrResMRI.2022.221117

Abstract

Mayer–Rokitansky–Kuster–Hauser syndrome is a congenital disorder characterized by uterine aplasia and upper vaginal aplasia in women with normal secondary sex characteristics and a normal female karyotype. Patients often present to the hospital with the complaint of primary amenorrhea in the presence of normal pubertal development and secondary sexual characteristics in adolescence. It is stated that the frequency is approximately 1 in every 5000 live female births. In recent years, neovagina and infertility treatment options have been increasing in the management of these patients. The use of imaging has made the diagnosis and evaluation of this disease much more efficient. This study aimed to highlight the findings of magnetic resonance due to its high efficiency in diagnosing and managing Mayer–Rokitansky–Kuster–Hauser syndrome as it is a non-invasive method.

Keywords: MRKH, mullerian agenesis, uterovaginal agenesis, infertility, neovagina

INTRODUCTION

The Mayer–Rokitansky–Kuster–Hauser (MRKH) syndrome is a condition characterized by congenital aplasia or severe hypoplasia of the uterus, upper vagina, and fallopian tubes, which are tissues originating from the Müllerian duct. It has been found to occur in 1 out of every 4000-5000 newborns.[1] It manifests itself with primary amenorrhea in the presence of normal pubertal development and secondary sexual characteristics due to the normal functioning of the ovaries in adolescent patients. In young women, the MRKH condition has disastrous consequences for fertility and sexual intercourse.

To facilitate clinical and psychologic input, it is critical that the diagnosis be made swiftly and precisely. Despite the fact that magnetic resonance (MR) imaging is widely accepted as a valuable modality, there are few publications in the literature, and they focus on a small number of women.[2] There are little precise descriptions of what to expect in these conditions. It is a widely known misconception that there is no uterine development in patients with MRKH syndrome; consequently, if primitive Mullerian structures are present, diagnostic confusion may arise.[3] Although ultrasonography is the first step in imaging methods, the main role in the diagnosis of patients belongs to MR examination.

History, Embryology, Etiology, and Genetics

This syndrome was named after the research carried out by the German anatomist August Franz Josef Karl Mayer (1829), the Austrian anatomist Carl von Rokitansky (1838), the German gynecologist Hermann Küster (1910), and the Swiss gynecologist Georges Andre Hauser (1961). Years later, Küster was the first to report this case by removing the painful uterine remnant. Hauser and Schreiner described uterovaginal agenesis in 21 cases with normal karyotype and normal genotype. Schmid-Tannwald and Hauser showed that extragenital findings can be added to uterovaginal agenesis. In this way, type 2 classification has emerged in the current literature. Type 1 MRKH is 56%-72%, and no additional findings are observed. Type 2 MRKH is seen at a rate of 28%-44% and has extragenital findings (such as renal and musculoskeletal systems).[4]

The female reproductive system in humans includes the fallopian tubes, uterus, cervix, and vagina. The paramesonephric ducts (PMDs) are the source of the upper two-thirds of the vagina, the cervix, the uterus, and the fallopian tube, while the urogenital sinus is the source of the lower third of the vagina. The uterus, cervix, and upper vagina are formed by fusing the caudal parts of the 2 PMDs, whereas the oviducts are formed from the upper part of the PMD on the side. Mayer–Rokitansky–Kuster–Hauser syndrome is caused by a state of complete agenesis or partial aplasia of PMDs that make up the uterus and upper vaginal wall.[5,6]

Many genetic and environmental factors are considered in the etiology. Based on developmental pathways and linked illnesses, several other potential genes have been disproven to cause MRKH syndrome.[4] The presence of WNT4 mutations has been detected in patients with Müllerian aplasia and virilization. This, however, should be seen as a distinct entity rather than a cause of MRKH syndrome. In addition, it has been discovered that there are genetic polymorphisms in the WNT9B gene, which plays a role in the development of the genitourinary system and acts upstream of

WNT4. A recent study investigating male microchimerism as a possible cause failed to obtain sufficient data to support this conclusion.[7,8]

Clinical Presentation and First Imaging Methods

The most common early MRKH symptoms are primary amenorrhea in a healthy adolescent female. When physical examination results consistent with an incomplete or hypoplastic vagina are obtained, MRKH and complete androgen insensitivity syndrome caused by an inactivating mutation in the androgen receptor can be considered among the differential diagnoses. After MRKH is determined, imaging examinations are very important in determining the extent of abnormalities. Some studies have found that most patients have concomitant anomalies, mainly anomalies of the renal and skeletal systems. Renal agenesis was the most common concomitant renal anomaly.[1,9]

Ultrasound and magnetic resonance imaging are the 2 most common imaging studies. Ultrasound is inexpensive and easy to use, but it is not always successful in detecting underdeveloped Müllerian structures and ovaries of different localization. It has been found that the ovaries are outside the typical location in approximately 16%-19% of patients.[10] According to various studies, MRI is the preferred procedure for detecting uterine abnormalities, including MRKH.[11] According to studies, MRI and laparoscopic examinations are highly associated, and the majority of them are based on bilateral uterine examinations. In recent articles, it has been understood that MRI can adequately show the endometrial tissue in the rudimentary uterus. In addition, it has been found that MRI has sufficient diagnostic power compared to laparoscopy, which is an invasive method.[1] Briefly, previous research has demonstrated that MRI is 100% sensitive and specific in the diagnosis of MRKH syndrome when paired with other diagnostic methods such as ultrasonography or laparoscopy.[12] Accordingly, MRI is the most useful noninvasive method for surgical planning. Sample cases are shown in Figures 1-3.

Magnetic Resonance Imaging Findings

There is no standard MRI protocol determined in the literature. Axial, coronal T1-weighted (T1W); axial, coronal, sagittal T2-weighted; and sometimes fat-printed T2-weighted (T2W) pelvic MRI images were produced using 1, 1.5, and 3 T multiplanar scanners. Three-dimensional SPACE arrays, gradient-required echo, and diffusion-weighted images were also used in some patients. Although intravenous contrast material was used during imaging in some of the patients, it was observed that imaging was performed without contrast material in some of them.[13] The presence and degree of development of the vaginal canal can be assessed by looking at the presence of a low-signal density structure between the urethra–bladder neck and rectum in sagittal and axial T2W and 3D SPACE images. In addition, T2W images of hypointense muscle and fibrous tunica may contrast with T2W images of hyperintense mucosa and mucus in the vaginal lumen.[14] T1-weighted and T2-weighted images should be used to evaluate the presence, location, volume, and differentiation of Müllerian buds into layers. On T1W images, uterine remnants may appear as an elongated or oval solid structure with isointense to low signal intensities. Potential cavitation within the uterine buds can be demonstrated by T2W images. The cavitation occurs as a target pattern with a center region of T2W hyperintense signal intensity indicating the endometrium, surrounded by intermediate signal intensity of the junctional zone and medium to high signal intensity of the muscle layer.[15] The ovaries showed isointense to hypointense signal intensity on T1W images and mixed signal intensity on T2W images with T2 hyperintense ovarian follicles and T2W low signal intensity ovarian stroma.[16] Because of the frequent association of MRKH syndrome with renal agenesis and pelvic renal ectopia, abdominal sequences are also obtained to evaluate the renal compartment in all cases.[17]

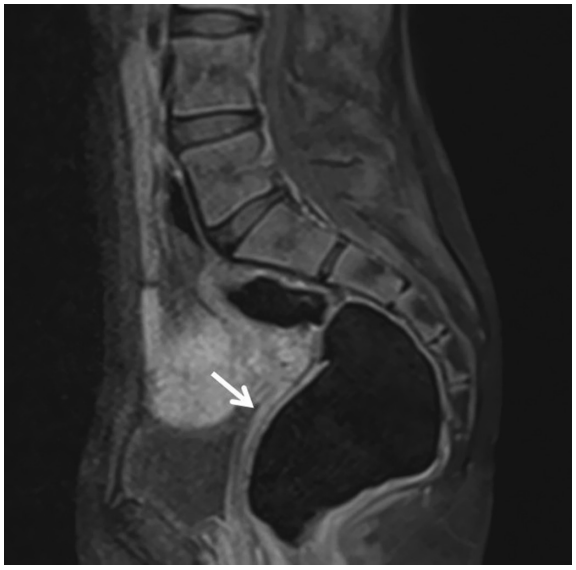

Figure 1. Thin rudimentary uterus is seen between bladder and rectum in postcontrast T1 sequence in the sagittal view (arrow).

The perception in the literature that the uterus is completely agenetic is incorrect because most patients have a rudimentary uterus.[11] Hall-Craggs et al[2] showed the presence of a rudimentary uterus in 93% of

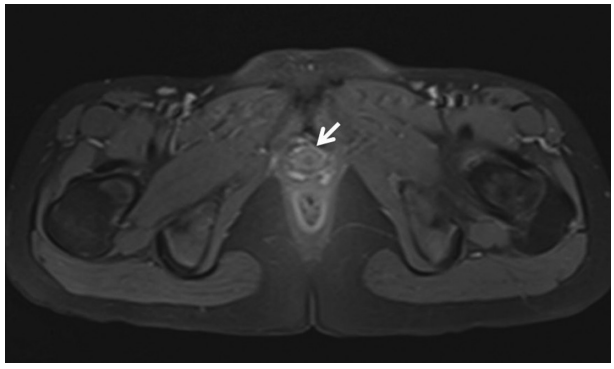

Figure 2. Presence of the distal vagina in the axial view in the post-contrast T1 sequence (arrow).

MAIN POINTS

- Mayer–Rokitansky–Kuster–Hauser (MRKH) syndrome is characterized by congenital complete aplasia or severe hypoplasia of tissues originating from the müller duct.
- Since ovarian function is normal in MRKH, pubertal development and secondary sexual characteristics are normal, but primary amenorrhea is present due to Mullerian anomaly.
- More than half of the patients with MRKH are accompanied by gynecological or extragynecological anomalies, and ultrasound alone is insufficient to detect them.
- Magnetic resonance imaging is the most useful method for detecting underdeveloped Mullerian structures and ovaries and planning surgery.

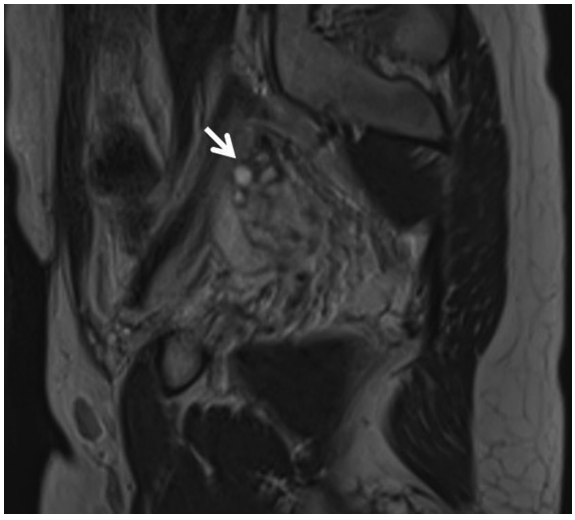

Figure 3. Presence of normal ovarian tissue in the unenhanced T2 sagittal image (arrow).

patients, similar to laparoscopy. However, although the uterine volume is large, it is understood that its parts such as fundus and ostium cannot be organized. The number of patients for whom these departments can be organized is quite limited. It has been shown that the rudimentary uterus is bilateral in 82%, unilateral in 11%, and agenetic in 8%.In most patients, it is located in the midline but only in the inferior caudal margin and laterally in only 1% of the population. In completely agenetic patients, a thin band with a low signal and difficulty to distinguish was noted in the uterine trace. In rare cases, the rudimentary uterus has also been shown to occur in the anterior ovary and inguinal canal. Even when the ovaries were ectopic, all uteri were instantly connected to the caudal edge of their matched ovary, and this association remained constant. Patients with MRKH syndrome may have a large primitive uterus (range, 0.4-80.2 mL) with an average volume of 6.4 mL, which can mimic a normal postpartum uterus. The primitive uterus in patients with MRKH syndrome has varying degrees of layer differentiation (8% are all uterine layers, 13% are 2-layer, 21% are single-layer).[2] It was noted that the rate of selection of the separation of the 3 layers increased in uteri with a volume greater than 20 mL. However, no matter how much the volume increased, the cervix could not be observed in the cases. In addition, it has been shown that the susceptibility to adenomyosis increases as the uterine volume increases in patients. In cases where the ovarian volume is more than 80 mL, blood products indicating that the endometrial tissue is functional were observed. Circular pelvic pain has also been shown to occur in these cases. The presence of bilateral ovarian tissue could be demonstrated in 95% of the patients. In cases where ovarian tissue could not be observed, the diagnosis of ectopic localization was considered as a priority, since there was normal sexual development and gonadotropin levels were sufficient. In the search for ectopic ovarian tissue, first of all, the pelvic anterior-lateral region, around the pelvic bone and around the anterior abdominal wall, should be carefully examined. The mean ovarian volume was observed to be around 10 mL. Vaginal length reaching an average of 2 cm was detected in 70% of the patients, while only vaginal dimple was observed in the remaining population.[2,11]

CONCLUSION

Mayer–Rokitansky–Kuster–Hauser syndrome is a congenital malformation of normal ovaries with agenesis of their tissues of varying degrees of PMD origin, which may be accompanied by extragynecological abnormalities of varying degrees. Patients with MRKH are treated for neovagina formation or reproduction with newer assisted reproductive techniques. Magnetic resonance imaging is a noninvasive method that helps to provide better preoperative counseling in MRKH patients, and knowing its findings is important in diagnosis and treatment planning.

Peer-review: Externally peer-reviewed.

Author Contributions: Concept – O.K., E.F.; Design – O.K., E.F.; Supervision – O.K., E.F.; Fundings – None; Materials – O.K., E.F.; Data Collection and/or Processing – O.K., E.F.; Analysis and/or Interpretation – O.K., E.F.; Literature Review – O.K., E.F.; Writing – O.K., E.F.; Critical Review Contribution Type – O.K., E.F.

Declaration of Interests: The authors have no conflicts of interest to declare.

Funding: The authors declared that this study has received no financial support.

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