



MRI in the diagnosis of Mayer-Rokitansky-Kuster-Hauser syndrome

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PURPOSE

The aim of this study was to establish the role of magnetic resonance imaging (MRI) in patients with Mayer-Rokitansky-Kuster-Hauser syndrome (MRKHS).

MATERIALS AND METHODS

Sixteen female MRKHS patients (mean age, 19.4 years; range, 11–39 years) were studied using MRI. Two experienced radiologists evaluated all the images in consensus to assess the presence or absence of the ovaries, uterus, and vagina. Additional urogenital or vertebral pathologies were also noted.

RESULTS

Of the 16 patients, complete aplasia of uterus was seen in five patients (31.3%). Uterine hypoplasia or remnant uterus was detected in 11 patients (68.8%). Ovaries were clearly seen in 10 patients (62.5%), and in two of the 10 patients, no descent of ovaries was detected. In five patients, ovaries could not be detected on MRI. In one patient, agenesis of right ovary was seen, and the left ovary was in normal shape. Of the 16 cases, 11 (68.8%) had no other extragenital abnormalities. Additional abnormalities were detected in six patients (37.5%). Two of the six had renal agenesis, and one patient had horseshoe kidney; renal ectopy was detected in two patients, and one patient had urachal remnant. Vertebral abnormalities were detected in two patients; one had L5 posterior fusion defect, bilateral hemisacralization, and rotoscoliosis, and the other had coccygeal vertebral fusion.

CONCLUSION

MRI is a useful and noninvasive imaging method in the diagnosis and evaluation of patients with MRKHS.

Mayer-Rokitansky-Kuster-Hauser syndrome (MRKHS) is a rare congenital Müllerian anomaly characterized by partial or complete absence of the uterus with an absent or hypoplastic vagina. It was first described by Mayer (1829) and Rokitansky (1838) as agenesis of the uterus and vagina due to abnormal development of the uterine ducts (1). In 1910, Küster described urological associations, and in 1961, Hauser distinguished MRKHS from testicular feminization. Because the external appearances of MRKHS females are normal, the syndrome is difficult to diagnose until puberty. Although patients may occasionally be diagnosed at birth or during childhood due to other health problems, the mean age of diagnosis is between 15 and 18 years (2). The aim of this study was to establish the role of magnetic resonance imaging (MRI) in patients with MRKHS.

Materials and methods

This retrospective study included 16 female patients (mean age, 19.4 years; range, 11–39 years) referred to the radiology department between August 2006 and February 2011. The main complaint during presentation was primary amenorrhea (n=14; 87.5%). The other patients (n=2; 12.5%) presented with abdominal pain (Table 1).

On physical examination, all patients had normal female external genitalia. Completed puberty with normal secondary female sexual characteristics, such as pubic hair or Tanner stage 5 breast development, was confirmed in 13 patients (81.3%). Three of the patients (patients 7, 11, and 13) had incomplete breast development, and breast development was consistent with Tanner stage 2, 3, and 3, respectively. It was thought that incomplete breast development of two patients (patient 7 and 13; both 11-years old) was due to their ages. Although complete secondary sex characteristics were not observed in one patient (patient 13; 16-years-old) during MRI examination, secondary sex characteristics developed completely during the follow-up after one year.

The diagnosis of all patients was based on findings at physical examination and MRI. Additionally, one patient (patient 2) had been diagnosed by laparoscopy. Four patients (patients 1, 12, 15, and 16) had karyotype analysis, and all had a normal 46,XX karyotype. For exclusion of testicular feminization, both inguinal canals were evaluated for the presence of rudimentary ectopic testis by ultrasonography (US) and MRI. None of the patients had ectopic testis.

All patients were evaluated by pelvic MRI. All MRI examinations were performed with a 1.5 Tesla magnet (GE Signa Excite HD, GE Medical Systems, Milwaukee, Wisconsin, USA) using a body coil. The MRI protocols included the axial fast spin echo T1, the fast spin echo T2, the fast spin echo T2 with fat saturation, the coronal fast spin echo T2, and the sagittal

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Table 1. Complaints and physical examination of patients with Mayer-Rokitansky-Kuster-Hauser syndrome

Patient number	Age (years)	Complaint	Additional imaging modality	Secondary sex characters
1	26	Primary amenorrhea	US	+
2	25	Primary amenorrhea	US	+
3	16	Primary amenorrhea	US	+
4	16	Primary amenorrhea	US	+
5	39	Primary amenorrhea	US, CT	+
6	18	Primary amenorrhea	US	+
7	11	Pelvic pain	US	-
8	31	Primary amenorrhea	US	+
9	16	Primary amenorrhea	US	+
10	14	Primary amenorrhea	US	+
11	11	Pelvic pain	US, CT	-
12	15	Primary amenorrhea	US	+
13	16	Primary amenorrhea	US	-
14	18	Primary amenorrhea	US	+
15	16	Primary amenorrhea	US	+
16	22	Primary amenorrhea	US	+

CT, computed tomography; US, ultrasonography.

Table 2. MRI findings of patients with Mayer-Rokitansky-Kuster-Hauser syndrome

	Present n (%)
Ovaries	10 (62.5) ^a
Uterus	11 (68.8) (hypoplastic or remnant)
Vagina	16 (100) (incomplete)
Additional pathology	5 (31.2)

^aIn two patients (patients 5 and 14), ovaries were located at anterior of iliopsoas muscle, and in one patient (patient 13), only left ovary was visible on MRI, the right ovary was absent.

Table 3. Additional abnormalities detected on MRI

Patient number	Type of abnormality
1	Fusion of sacral vertebrae
5	Right renal ectopy, left renal agenesis
8	Right renal ectopy, sacral fusion defect, rotoscoliosis
14	Right renal agenesis
15	Horseshoe kidney

Two experienced radiologists with six and eight years of experience in genitourinary radiology, respectively, evaluated all of the images in consensus to assess MRI results for the presence of uterus or remnant uterus, complete vagina, ovaries, and additional urogenital or other pathologies such as renal ectopia or agenesis.

Results

Of the 16 patients, complete aplasia of uterus was seen in five patients (31.3%) (Fig. 1). Uterine hypoplasia or remnant uterus was detected in 11 of the 16 patients (68.8%). Ovaries were seen clearly in 10 patients (62.5%) (Fig. 2). In two of these 10 patients, no descent of ovaries was detected; instead, the ovaries were located near the iliopsoas muscle (Fig. 3). In five patients, ovaries could not be detected by MRI (Fig. 4). In one patient, agenesis of the right ovary was seen, and the left ovary was in normal shape. MRI findings are shown in Table 2.

All of the patients had a blindly ended vagina on MRI. Vaginal agenesis was not observed in any patient. MRI showed additional urogenital and other pathologies. Of the 16 cases, additional abnormalities were detected in six patients (37.5%) (Table 3), while the other ten (62.5%) had no other abnormalities. Two of the six with abnormalities (patients 5 and 14) had renal agenesis, and one patient (patient 15) had horseshoe kidney (Fig. 5). Renal ectopy was detected in two patients (patients 5 and 8) (Fig. 6), and one patient (patient 4) had a urachal remnant (Fig. 7). Vertebral abnormalities were detected in two patients (patients 1 and 8) on MRI (Fig. 8). One had L5 posterior fusion defect, bilateral hemisacralization, and rotoscoliosis (patient 8), and the other had coccygeal vertebral fusion (patient 1).

From these findings, six of the patients were classified as MRKHS type 1, and ten were classified as MRKHS type 2.

Discussion

MRKHS accounts for approximately 15% of patients with primary amenorrhea, and is the second common cause of primary amenorrhea (3). The incidence of this syndrome is 1:4000 female live births. The cause of MRKHS is still not completely understood, and the etiology of MRKHS is believed to be polygenic and multifactorial.

fast spin echo T2 images. The parameters of the pelvis MRI were as follows: TR/TE 560/15 (T1-weighted image), 4000/126 or 4400/124 (T2-weighted image), 4000/126 (T2-weighted image with fat saturation), 5- or 8-mm slice

thickness, 1- or 1.5-mm intersection gap, 26- or 28-cm field of view (FOV), 2 or 3 number of excitations, and a 384×256 or 320×224 matrix. Intravenous contrast material was not used or needed during the examinations.

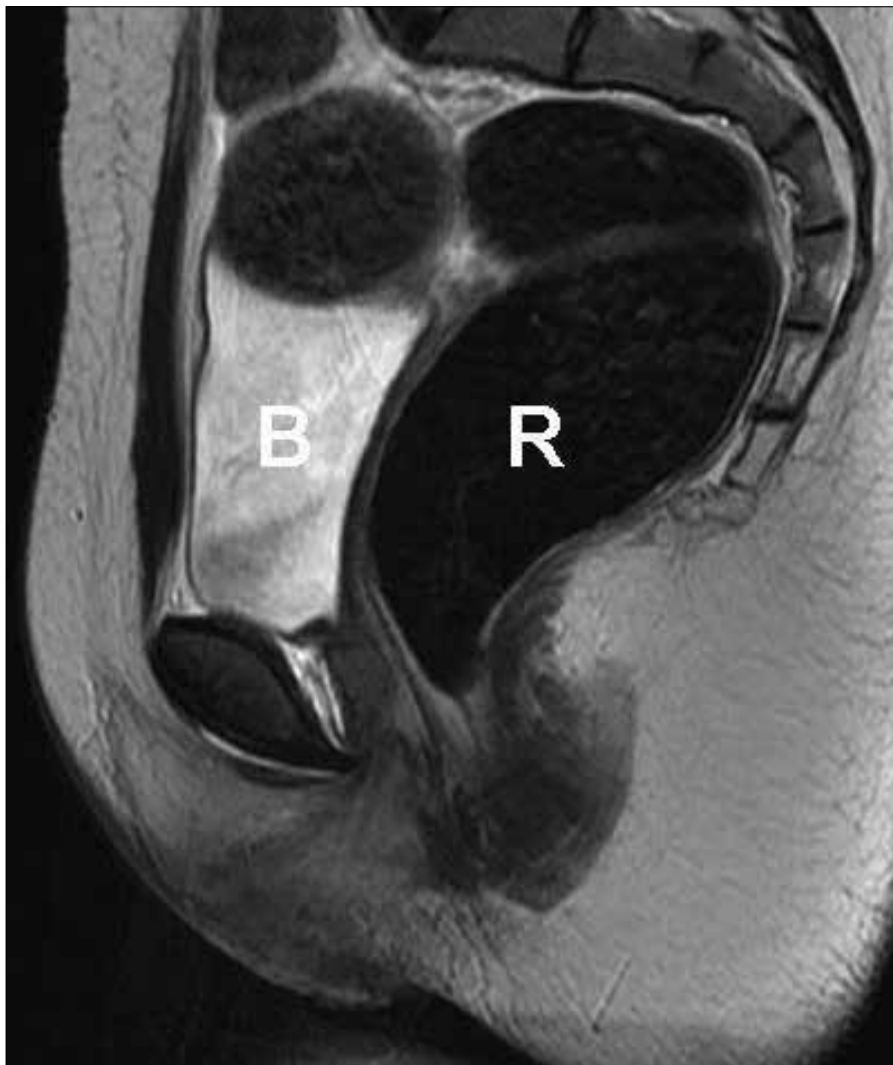


Figure 1. Uterus is not observed on sagittal T2-weighted image. B, bladder; R, rectum.

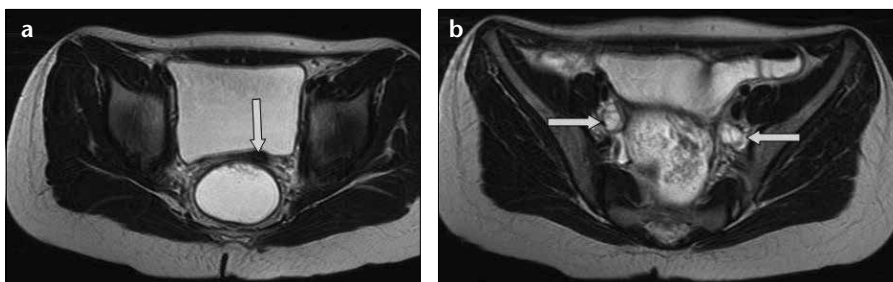


Figure 2. a, b. Axial T2-weighted images show a uterine remnant in its expected location (a, arrow) and both ovaries of normal size in the normal location (b, arrows).

Müllerian ducts are the primordia for the female internal reproductive system that differentiate into Fallopian tubes, uterus, cervix, and the upper part of the vagina during embryogenesis (4). The arrest of Müllerian duct development seven weeks after fertilization results in MRKHS during embryogenesis (5). Müllerian duct anom-

alies are classified according to the system established by the American Fertility Society. MRKHS is the most common form of class 1 (6).

The most common symptom of the patients with MRKHS is primary amenorrhea. Upon physical examination, secondary sexual characteristics are normal due to normal ovary functions,

and there is no sign of androgen excess in MRKHS patients. Due to normally functioning ovaries, the levels of follicle stimulating hormone and luteinizing hormone are normal. Patients with MRKHS have a 46,XX karyotype.

MRKHS has two subtypes: the typical (also called Rokitansky sequence, type I, type A or isolated) and the atypical form (type II or type B) (3). In patients with typical form, the only affected part is the caudal part of the Müllerian duct. The atypical form of MRKHS is associated with other anomalies, including mainly renal anomalies such as unilateral agenesis, ectopia of one or both kidneys, or horseshoe kidney (4). The most common upper urinary tract malformation associated with MRKHS is unilateral renal agenesis (7). In addition, vertebral anomalies (8), hearing defects (9), skeletal abnormalities (10), ovarian cancers (11, 12), and heart malformations (13, 14) have been reported as associated anomalies with MRKHS.

MURCS (Müllerian Renal Cervical Somite) is used to refer to MRKHS with associated abnormalities, and is the most severe form of the disorder (15). Type B or the atypical form of MRKHS is more common than the isolated form. Discrimination of type A and type B is essential because of the associated anomalies that occur only with type B (16).

Although the clinical diagnosis of MRKHS can be easily established, for confirmation of the diagnosis and detection of possible associated abnormalities, further evaluations using laparoscopy, imaging and karyotyping may be needed. US should be the first choice in evaluating MRKHS and can provide information about associated renal malformation. However, US is an operator-dependent imaging method that may generate conflicting results when performed several times (17). In addition, US may not always allow for the recognition of Müllerian buds and ovaries with extrapelvic location, which is essential information for defining the best surgical treatment (18). Thus, differentiation between type A and type B may not always be possible using US.

Although computed tomography may give information about congenital anomalies, it is not routinely performed due to the use of ionizing radiation (2). Detailed anatomical

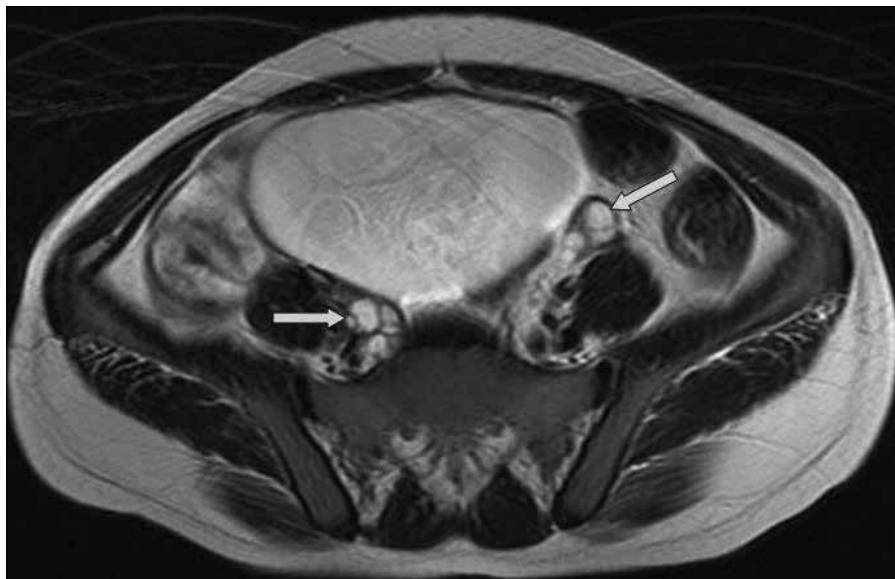


Figure 3. An axial T2-weighted image demonstrates incomplete descent of the ovaries (arrows) adjacent to psoas muscles.

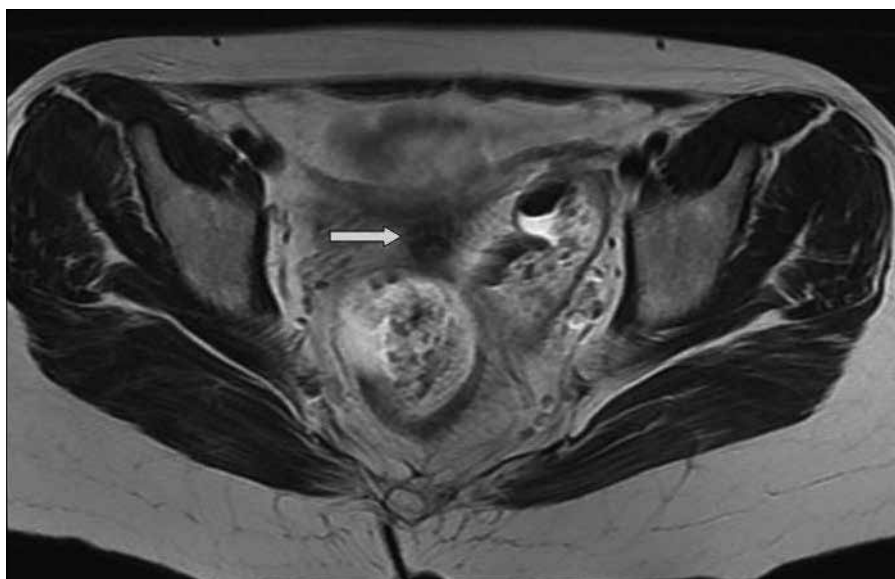


Figure 4. An axial T2-weighted image shows uterine remnant (arrow) in its expected location. Both ovaries are not detected in their normal locations.

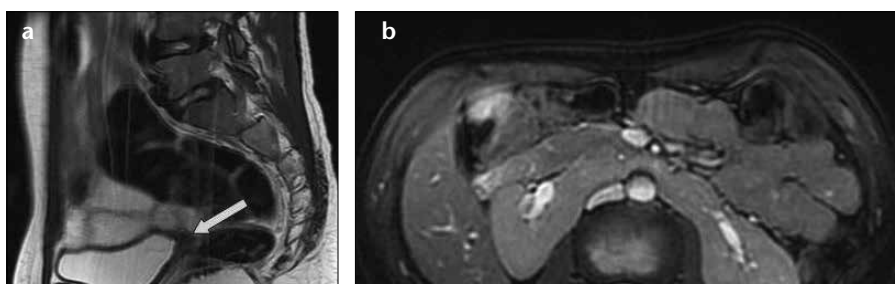


Figure 5. a, b. A sagittal T2-weighted image (a) shows a uterine bud in its expected location (arrow). The axial fat saturated T2-weighted image (b) reveals a horseshoe kidney in the midline anterior to the aorta in the same patient.

information was provided only by laparoscopy until a few years ago. However, diagnostic laparoscopy is

an invasive and expensive diagnostic method compared to MRI. MRI can be more effective due superior soft tissue

contrast resolution as well as the multiplanar capability, noninvasiveness, and lack of ionizing radiation. Because the use of a transvaginal US probe is avoided in pediatric patients, MRI becomes a more important examination method in this group of patients. MRI is also more cost-effective compared to laparoscopy. MRI can clearly demonstrate the anatomy of uterus. Vaginal agenesis is best identified on axial images with no normal vaginal anatomy demonstrated between the rectum and urethra. MRI can also identify associated congenital abnormalities such as renal or anorectal malformations that are essential for differentiation of both types of MRKHS.

In MRKHS, no identifiable uterine tissue or upper two-thirds of the vagina are seen, though due to a different embryonic origin, the lower third of the vagina is always present. However, in approximately 6%–10% of patients with MRKHS, endometrial tissue or variable development of uterus with hematometra may be present, which results in cyclic abdominal pain (16). The rudimentary Müllerian structures can be either functional or nonfunctional (19). If the endometrial layer is functional, the main symptoms will be primary amenorrhea and cyclic abdominal pain due to cryptomenorrhea and hematometra. Patients with complete agenesis of uterine will present with primary amenorrhea.

All information obtained by US, intravenous pyelography, and diagnostic laparoscopy can be given by MRI alone (2). In a study of 56 patients with MRKHS, MRI was judged on the basis of laparoscopic findings and diagnostic sensitivity, and the specificity of MRI was found to be 100% (17). MRKHS is often diagnosed clinically, but in some patients the diagnosis is either radiologically or laparoscopically confirmed. MRI is useful for differentiating between uterine agenesis and hypoplasia, both of which can be best assessed on sagittal images (19). There is no standard MRI protocol for evaluating MRKHS; sequences and acquisition planes vary among centers and investigator preference, and are generally effective in evaluation. Coronal T1-weighted spin echo with a large FOV including the pelvis and upper abdomen can give information on anatomy and associated urinary abnormalities (20). As a general rule,

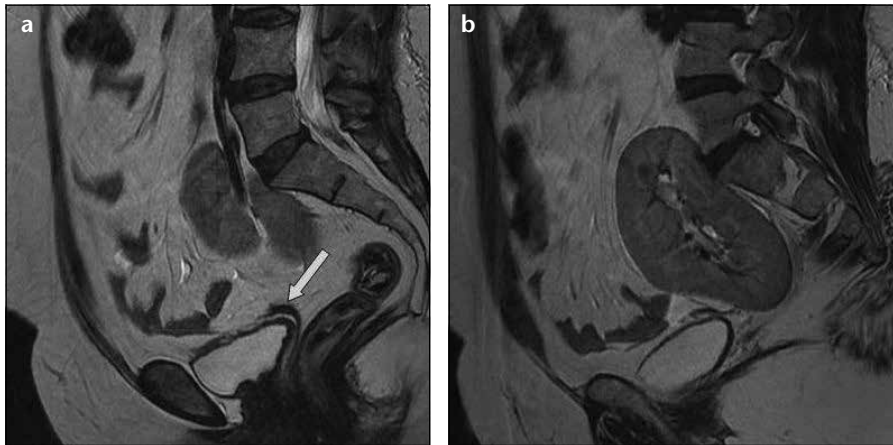


Figure 6. a, b. Sagittal T2-weighted images show a uterine remnant (arrow) in its expected location (a) and an ectopic kidney located in the pelvis (b).

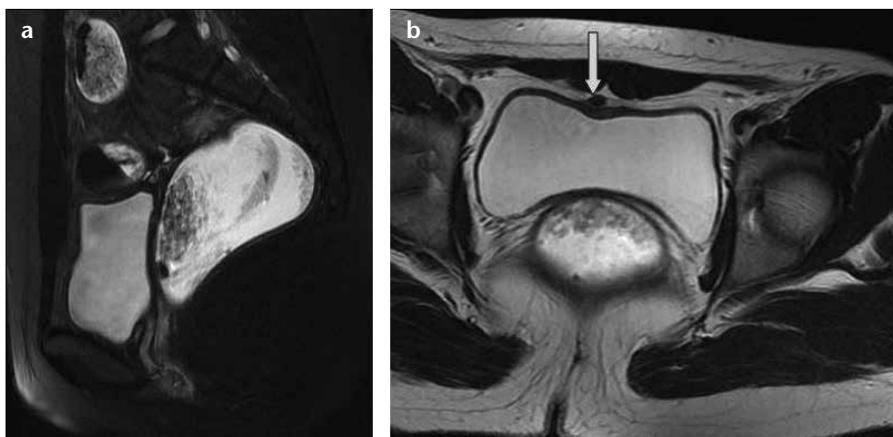


Figure 7. a, b. The uterus is not observed in its expected location on a sagittal fat saturated T2-weighted image (a). An axial T2-weighted image (b) shows an urachal remnant (arrow) in the midline anterior to the bladder

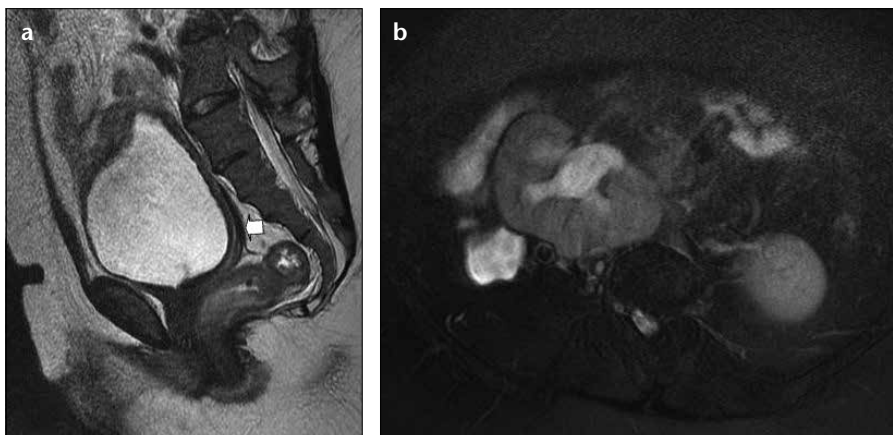


Figure 8. a, b. A sagittal T2-weighted image (a) shows uterine remnant (arrow) in its expected location, and lumbosacral vertebral and spinal abnormalities. A fat saturated T2-weighted image (b) demonstrates right ectopic kidney located in the pelvis and lumbar vertebral abnormalities in the same patient.

T2-weighted sequences are useful sequences in female pelvic imaging due to the ability to demonstrate the zonal anatomy of the uterus (21). Sagittal

T2-weighted spin echo and oblique long axis T2-weighted fast-spin echo images obtained parallel to the long axis of the uterus can be used for di-

agnosis of uterovaginal anomalies (20). Uterine hypoplasia or agenesis is best diagnosed on T2-weighted sagittal images. Normal vagina is seen as a tube of intermediate signal intensity between the base of bladder and urethra anteriorly and the anal canal posteriorly (20). Vaginal agenesis is best demonstrated on axial images of MRI.

In a phenotypic woman with primary amenorrhea and an absent uterus, MRKHS should be differentiated from androgen insensitivity syndrome or testicular feminization syndrome (21). In such cases, MRI can also confirm the presence of rudimentary ectopic testis and the absence of both uterus and ovaries, which are essential findings for differential diagnosis. Detection of normal ovaries is the main finding in the diagnosis of MRKHS. Identification of normal follicles can be helpful as a marker for detection of ovaries.

There were some limitations in our study. First, karyotype analysis was performed in only four patients. Second, only some of the patients had surgical or laparoscopic confirmation of the MRI diagnosis. Finally, the patient number of this study was limited, therefore further studies with larger number of patients are required.

In conclusion, MRI is a useful and noninvasive imaging method in the diagnosis and evaluation of patients with MRKHS.

Conflict of interest disclosure

The authors declared no conflicts of interest.

References

1. Pittock ST, Babovic-Vuksanovic D, Lteif A. Mayer-Rokitansky-Küster-Hauser anomaly and its associated malformations. *Am J Med Genet A* 2005; 135:314–316. [\[CrossRef\]](#)
2. Govindarajan M, Rajan RS, Kalyanpur A, Ravikumar. Magnetic resonance imaging diagnosis of Mayer-Rokitansky-Küster-Hauser syndrome. *J Hum Reprod Sci* 2008; 1:83–85. [\[CrossRef\]](#)
3. Strübbe EH, Cremers CW, Willemsen WN, Rolland R, Thijn CJ. The Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome without and with associated features: two separate entities? *Clin Dysmorphol* 1994; 3:192–199.
4. Guerrier D, Mouchel T, Pasquier L, Pellerin I. The Mayer-Rokitansky-Küster-Hauser syndrome (congenital absence of uterus and vagina)--phenotypic manifestations and genetic approaches. *J Negat Results Biomed* 2006; 27:5:1–8. [\[CrossRef\]](#)

5. Troiano RN, McCarthy SM. Mullerian duct anomalies: imaging and clinical issues. *Radiology* 2004; 233:19–34. [\[CrossRef\]](#)
6. The American Fertility Society classifications of adnexal adhesions, distal tubal occlusion, tubal occlusion secondary to tubal ligation, tubal pregnancies, müllerian anomalies and intrauterine adhesions. *Fertil Steril* 1988; 49:944–955.
7. Oppelt P, Renner SP, Kellermann A, et al. Clinical aspects of Mayer-Rokitansky-Küster-Hauser syndrome: recommendations for clinical diagnosis and staging. *Hum Reprod* 2006; 21:792–797. [\[CrossRef\]](#)
8. Strübbe EH, Lemmens JA, Thijn CJ, Willemssen WN, van Toor BS. Spinal abnormalities and the atypical form of the Mayer-Rokitansky-Küster-Hauser syndrome. *Skeletal Radiol* 1992; 21:459–462. [\[CrossRef\]](#)
9. Strübbe EH, Cremers CW, Dijkers FG, Willemssen WN. Hearing loss and the Mayer-Rokitansky-Küster-Hauser syndrome. *Am J Otol* 1994; 15:431–436.
10. Strübbe EH, Thijn CJ, Willemssen WN, Lappöhn R. Evaluation of radiographic abnormalities of the hand in patients with the Mayer-Rokitansky-Küster-Hauser syndrome. *Skeletal Radiol* 1987; 16:227–231. [\[CrossRef\]](#)
11. Tsaur GT, Lee MH, Su SL, Wu MJ, Huang TW. Mayer-Rokitansky-Küster-Hauser syndrome with immature teratoma of the ovary at age 4 years. *Gynecol Oncol* 1995; 56:456–459. [\[CrossRef\]](#)
12. Ghirardini G, Magnani A. Mayer-Rokitansky-Küster-Hauser syndrome and ovarian cancer. Report of a case. *Clin Exp Obstet Gynecol* 1995; 22:247–248.
13. Fakhri MH, Williamson HO, Seymour EQ, Pai S. Concurrence of the Holt-Oram syndrome and the Rokitansky-Küster-Hauser syndrome. A case report. *J Reprod Med* 1987; 32:549–550.
14. Kula S, Saygili A, Tunaoglu FS, Olguntürk R. Mayer-Rokitansky-Küster-Hauser syndrome associated with pulmonary stenosis. *Acta Paediatr* 2004; 93:570–572. [\[CrossRef\]](#)
15. Braun-Quentin C, Billes C, Böwing B, Kotzot D. MURCS association: case report and review. *J Med Genet* 1996; 33:618–620. [\[CrossRef\]](#)
16. Giusti S, Fruzzetti E, Perini D, Fruzzetti F, Giusti P, Bartolozzi C. Diagnosis of a variant of Mayer-Rokitansky-Küster-Hauser syndrome: useful MRI findings. *Abdom Imaging* 2011; 36:753–755. [\[CrossRef\]](#)
17. Pompili G, Munari A, Franceschelli G, et al. Magnetic resonance imaging in the preoperative assessment of Mayer-Rokitansky-Küster-Hauser syndrome. *Radiol Med* 2009; 114:811–826. [\[CrossRef\]](#)
18. Fedele L, Dorta M. Magnetic resonance imaging in Mayer-Rokitansky-Küster-Hauser syndrome. *Obstet Gynecol* 1990; 76:593–596.
19. Junqueira BL, Allen LM, Spitzer RF, Lucco KL, Babyn PS, Doria AS. Müllerian duct anomalies and mimics in children and adolescents: correlative intraoperative assessment with clinical imaging. *Radiographics* 2009; 29:1085–1103. [\[CrossRef\]](#)
20. Saleem SN. MR imaging diagnosis of uterovaginal anomalies: current state of the art. *Radiographics* 2003; 23:e13. [\[CrossRef\]](#)
21. Marcal L, Nothaft MA, Coelho F, Volpato R, Iyer R. Mullerian duct anomalies: MR imaging. *Abdom Imaging* 2011; 36:756–764. [\[CrossRef\]](#)