International Journal of Biomedicine 14(2) (2024) 348-351
http://dx.doi.org/10.21103/Article14(2)_CR4

CASE REPORT

INTERNATIONAL JOURNAL OF BIOMEDICINE

The Role of MRI in Diagnosing Mayer-Rokitansky-Kuster-Hauser Syndrome: A Case Study

Diar Kabashi[1,3], Kreshnike Dedushi[2], Gojart Ferati[4], Argjira Juniku-Shkololli[3], Besiana Kabashi[5], Jeton Shatri[1,2*]

[1]*Institute of Anatomy, Faculty of Medicine, University of Prishtina, Prishtina, Kosovo*
[2]*Clinic of Radiology, University Clinical Centre of Kosovo, Prishtina, Kosovo*
[3]*Clinic of Gastroenterology, University Clinical Centre of Kosovo, Prishtina, Kosovo*
[4]*Clinic of Gynecology and Obstetrics, University Clinical Centre of Kosovo, Prishtina, Kosovo*
[5]*Clinic of Biochemistry, University Clinical Centre of Kosovo, Prishtina, Kosovo*

Abstract

The Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is a rare condition that results from the disturbance of embryonic paramesonephric duct development, which gives rise to varying degrees of malformation of reproductive organs. It is characterized by uterovaginal aplasia with normal secondary sexual characteristics and 46,XX karyotype. We report a 15-year-old female patient with MRKH. Pelvic MRI revealed cervical and uterine agenesis with the absence of the vagina. The diagnosis was confirmed based on radiological findings. The correct clinical and radiological diagnosis of MRKH by MRI is crucial for long-term management. **(International Journal of Biomedicine. 2024;14(2):348-351.)**

Keywords: Müllerian anomaly • primary amenorrhea • magnetic resonance imaging

For citation: Kabashi D, Dedushi K, Ferati G, Juniku-Shkololli A, Kabashi B, Shatri J. The Role of MRI in Diagnosing Mayer-Rokitansky-Kuster-Hauser Syndrome: A Case Study. International Journal of Biomedicine. 2024;14(2):348-351. doi:10.21103/Article14(2)_CR4

Introduction

The Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is a rare disorder characterized by congenital aplasia of the upper 2/3 of the vagina together with partial or total aplasia of the uterus in women with normal development of secondary sexual characteristics and a normal 46,XX karyotype. It is a rare congenital disorder with an incidence of 1 in 4,500 women.[1] Various assumptions exist for its etiology, which can be multi-factorial, such as genetic factors or the use of drugs such as diethylstilbestrol and thalidomide,[2] but to this day the etiology remains unknown. Triantafyllidi et al.[3] identified 76 studies describing multiple genetic defects that potentially contribute to the pathogenetic mechanism of MRKH syndrome. The most reported chromosomal regions and the possible genes implicated are: 1q21.1 (RBM8A gene), 1p31-1p35 (WNT4 gene), 7p15.3 (HOXA gene), 16p11 (TBX6 gene), 17q12 (LHX1 and HNF1B genes), 22q11.21, and Xp22. Usually, the first signs with which patients present to the doctor are primary amenorrhea, with well-developed secondary sexual characteristics. The associated abnormalities of this syndrome can include urological abnormalities (25%-50%), including renal agenesis, pelvic kidney or horseshoe kidney, other abnormalities of the collecting system as well as skeletal abnormalities (10-15%), including the spine, ribs, and extremities.[4]

MRI imaging of the uterus, cervix, and vagina offers in-depth insights into the anatomy of the uterovaginal region, with a specific focus on examining the external contours of the uterine fundus and the shape of the cavity, and it also allows tissue characterization of the possible septa, thus providing a complete classification of the specific anomaly.[5,6] At present, MRI boasts the utmost accuracy in diagnosing uterine anomalies, achieving a nearly perfect rate of nearly 100%, attributed to its outstanding resolution for soft tissue and its ability to visualize structures from multiple perspectives.[7,8] T1-weighted (T1W) and T2-weighted (T2W) MRI provide excellent zonal anatomy of the uterus, i.e., endometrium, junctional zone, and myometrial anatomy.[9,10]

***Corresponding author:** Prof. Ass. Dr. Jeton Shatri, University of Prishtina, University Clinical Centre of Kosovo, Prishtina, Kosovo. E-mail: jeton.shatri@uni-pr.edu*

Case Presentation

A 15-year-old female patient presented to the Radiology Clinic for an MRI of the abdomen and pelvis, ordered by her gynecologist, to whom she presented with primary amenorrhea and abdominal pain. Medical history showed only childhood diseases. Her mother was not a smoker, she rarely consumed alcohol, but she denies consuming alcohol during pregnancy. The woman also denied illness and exposure to medications during pregnancy. The patient's mother had menarche at the age of 12. Regarding the secondary sexual characteristics, the patient was normal for her chronological age. The hematological and biochemical laboratory examinations were all within the normal range (Table 1).

Table 1.

The hematological and biochemical tests.

Test name	Result	Range	Unit
Leukocytes	8.9	3.50-9.50	$\times 10^9$/L
Erythrocytes	4.8	3.80-5.80	$\times 10^{12}$/L
Platelets	250	125-350	$\times 10^9$/L
Hemoglobin	125	115-175	g/L
Hematocrit	38.2	35.0-50.0	%
Glucose	4.8	4.40-6.00	mmol/L
Urea	5.2	1.70-8.30	mmol/L
Creatinine	72	53-115	mmol/L
Ionized calcium	1.15	1.12-1.32	mmol/L
Potassium	140	132-146	mmol/L
Kalium	4.3	3.4-5.5	mmol/L
Progesterone	2.85	0.87-3.37	ng/mL
Testosterone	12.8	6.00-52.00	ng/dL
FSH	6.23	0.3-10	IU/L
LH	3.22	0.60-16.3	IU/L
Prolactin	15.20	4.04-23.30	ng/mL

The patient's weight and height at the time of the examination were 57kg and 155cm, respectively. A gynecological examination showed pubic hair, labia majora, labia minora, vagina opening, all these well-developed. The vagina was visible only at the entry, so only 1cm long with a blind end. Pelvic examination with MR examination technique included T2W pre-contrast images in axial/sagittal/coronal planes and axial T1FS images. DWI images for diffusion-weighted imaging were also obtained. Post-contrast images were obtained with fat suppression T1W axial, sagittal, and coronal planes (Figure 1). During the processing of the images acquired in a T2W sagittal plane, it was observed that there is a complete absence of the uterus and the upper 2/3 of the vagina (Figure 2). The small amount of free fluid in the pelvis can also be seen in these images. The images obtained in a T2W coronal plane clearly show that there is a lack of 2/3 of the upper part of the vagina; while in this view the vaginal remnant (distal part) can be evaluated, it was measured and found to be 1cm long. A T1W MRI scan in the axial plane showed normal, well-formed ovaries.

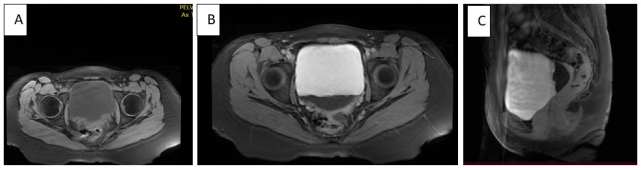

Fig. 1. *Magnetic resonance of pelvis. A) axial T1 (FS)-weighted images, B) axial and C) sagittal contrast-enhanced T1 (FS) - weighted images contrast-enhanced T1-weighted images.*

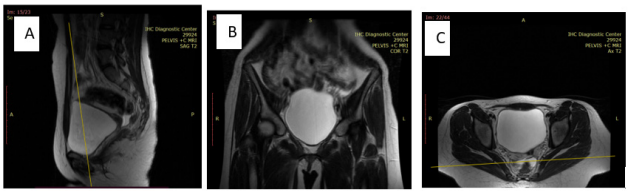

Fig. 2. *Magnetic resonance of pelvic. A) sagittal, B) coronal, and C) axial T2-weighted images show the absence of the uterus and vagina. Also, the presence of free fluid in the pelvis.*

Discussion

The MRKH syndrome manifests in two distinct forms. The typical form (Type 1) is defined by the congenital absence of the uterus and upper vagina, while the ovaries and fallopian tubes appear normal. The atypical form (Type 2) encompasses Müllerian anomalies in addition to non-gynecological anomalies affecting the urological, skeletal, vertebral, or cardiac systems.[11] In our case, it is presented as MRKH type 1. This syndrome was first described by Mayer in 1829, and later, in 1838, the description was completed by Rokitansky, who noted uterine and vaginal agenesis. Later, Kuster added renal abnormalities (renal agenesis, renal ectopy) and skeletal abnormalities. And finally, in 1961, Hauser separated MRKH from testicular feminization. Most cases appear to be sporadic.[11] A retrospective cohort study conducted from 1997 to 2011 at the University of Michigan in 2013 consisting of 48 MRKH patients found that 48% had a primordial uterus.[12] In a review in 2020,[13] it was estimated that 48%–84% of MRKH patients had a primordial uterus.

As for the clinical presentation, it was characterized by the normal development of secondary sexual characteristics and primary amenorrhea. The development of the ovaries and their function was normal. The levels of follicle-stimulating hormone (FSH) and luteinizing hormone (LH) were within normal ranges, and there were no indications of androgen excess, which helps differentiate it from androgen insensitivity syndrome.[14]

The diagnosis is usually made through clinical symptoms and clinical examination, also with imaging or laparoscopic confirmation, as a condition of the normal hormonal values and normal karyotype. It usually starts with

simple imaging procedures, such as 2D or 3D ultrasound, which are non-invasive methods and easy for the patient. As far as can be seen in ultrasound, it can be concluded from our study that there was a lack of development of the uterus. Usually, during the ultrasound examination, abnormalities such as renal ectopia, renal agenesis, or any other abnormality that is assumed to be related to MRKH syndrome are also looked for. Then, usually, patients are advised to undergo more detailed imaging procedures, such as CT or MRI. An MRI stands out for its superior efficacy, thanks to its multiplanar capacity and unparalleled soft tissue contrast. It surpasses all other imaging methods while avoiding the need for ionizing radiation.[15] By means of magnetic resonance, the abnormality of the relevant organs is assessed, through a higher sensitivity and specificity than other imaging methods. It provides accurate details regarding the anatomical positioning and any abnormalities within the uterus, potential tubal remnants, vestigial lamina, and ovaries.[16]

Usually, patients, after receiving the news that they have been diagnosed with MRKH, suffer from mental stress knowing that they do not have a uterus and vagina. This is exactly what the beginning of the treatment is, that is, the consultation with the patient before starting the treatment steps. The next steps include the creation of neovagina, which can be done after non-surgical treatment and surgical treatment. The non-surgical treatment includes the Frank's Method, which involves wearing vaginal dilators for at least 2 hours a day, which affects the increase in the width and length of the vagina. While the surgical treatment, of which there are different methods, consists in creating a channel that plays the role of the vagina. In a study by Motta and D'Alberton,[17] of 108 patients with MRKH syndrome from 1955 to 2003, 53 chose the option of creating neovagina with dilation (functional method), while 55 chose the surgical option. Of the patients who chose the non-surgical method, 83% expressed that they were satisfied with their treatment: 75% of the patients had an optimal result, 13% had an acceptable result, and 12% had a poor result. As for the group that chose the surgical option, 76% were satisfied with these methods, while 68% of the anatomical creation of the vagina were successful.[18,19]

Conclusion

The first point when there is suspicion of MRKH is the clinical presentation and gynecological examination. Imaging examinations, starting with ultrasound, are needed to verify the diagnosis. For a more detailed evaluation, an MRI is needed, which, in addition to showing genital abnormalities, can give us more information about whether it is Type 1 MRKH or Type 2 MRKH by showing other organs as well.

Competing Interests

The authors declare that they have no competing interests.

References

1. Hall-Craggs MA, Williams CE, Pattison SH, Kirkham AP, Creighton SM. Mayer-Rokitansky-Kuster-Hauser syndrome: diagnosis with MR imaging. Radiology. 2013 Dec;269(3):787-92. doi: 10.1148/radiol.13130211. Epub 2013 Oct 28. PMID: 23942608.
2. Bjørsum-Meyer T, Herlin M, Qvist N, Petersen MB. Vertebral defect, anal atresia, cardiac defect, tracheoesophageal fistula/esophageal atresia, renal defect, and limb defect association with Mayer-Rokitansky-Küster-Hauser syndrome in co-occurrence: two case reports and a review of the literature. J Med Case Rep. 2016 Dec 21;10(1):374. doi: 10.1186/s13256-016-1127-9. PMID: 28003020; PMCID: PMC5178070.
3. Triantafyllidi VE, Mavrogianni D, Kalampalikis A, Litos M, Roidi S, Michala L. Identification of Genetic Causes in Mayer-Rokitansky-Küster-Hauser (MRKH) Syndrome: A Systematic Review of the Literature. Children (Basel). 2022 Jun 27;9(7):961. doi: 10.3390/children9070961. PMID: 35883945; PMCID: PMC9322756.
4. Morcel K, Camborieux L; Programme de Recherches sur les Aplasies Müllériennes; Guerrier D. Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome. Orphanet J Rare Dis. 2007 Mar 14;2:13. doi: 10.1186/1750-1172-2-13. PMID: 17359527; PMCID: PMC1832178.
5. Fukunaga T, Fujii S, Inoue C, Mukuda N, Murakami A, Tanabe Y, Harada T, Ogawa T. The spectrum of imaging appearances of müllerian duct anomalies: focus on MR imaging. Jpn J Radiol. 2017 Dec;35(12):697-706. doi: 10.1007/s11604-017-0681-4. Epub 2017 Sep 18. PMID: 28921452.
6. Troiano RN, McCarthy SM. Mullerian duct anomalies: imaging and clinical issues. Radiology. 2004 Oct;233(1):19-34. doi: 10.1148/radiol.2331020777. Epub 2004 Aug 18. PMID: 15317956.
7. Pan HX, Liu P, Duan H, Li PF, Chen RL, Tang L, Luo GN, Chen CL. Using 3D MRI can potentially enhance the ability of trained surgeons to more precisely diagnose Mullerian duct anomalies compared to MR alone. Eur J Obstet Gynecol Reprod Biol. 2018 Sep;228:313-318. doi: 10.1016/j.ejogrb.2018.07.007. Epub 2018 Jul 9. PMID: 30075412.
8. Console D, Tamburrini S, Barresi D, Notarangelo L, Bertucci B, Tamburrini O. Il valore della RM nella valutazione delle anomalie dei dotti di Müller [The value of the MR imaging in the evaluation of Müllerian duct anomalies]. Radiol Med. 2001 Oct;102(4):226-32. Italian. PMID: 11740449.
9. Boruah DK, Sanyal S, Gogoi BB, Mahanta K, Prakash A, Augustine A, Achar S, Baishya H. Spectrum of MRI Appearance of Mayer-Rokitansky-Kuster-Hauser (MRKH) Syndrome in Primary Amenorrhea Patients. J Clin Diagn Res. 2017 Jul;11(7):TC30-TC35. doi: 10.7860/JCDR/2017/29016.10317. Epub 2017 Jul 1. PMID: 28893003; PMCID: PMC5583891.
10. Shivalingappa SS, Shetty SB. Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome with unilateral pulmonary agenesis-a rarity indeed: radiologic review. BJR Case Rep. 2016 Feb 3;2(2):20150157. doi: 10.1259/bjrcr.20150157. PMID: 30363603; PMCID: PMC6180887.

11. Carson SA, Simpson JL, Malinak LR, Elias S, Gerbie AB, Buttram VC Jr, Sarto GE. Heritable aspects of uterine anomalies. II. Genetic analysis of Müllerian aplasia. Fertil Steril. 1983 Jul;40(1):86-90. PMID: 6862043.
12. Marsh CA, Will MA, Smorgick N, Quint EH, Hussain H, Smith YR. Uterine remnants and pelvic pain in females with Mayer-Rokitansky-Küster-Hauser syndrome. J Pediatr Adolesc Gynecol. 2013 Jun;26(3):199-202. doi: 10.1016/j.jpag.2012.11.014. PMID: 23785713.
13. Committee on Adolescent Health Care. ACOG Committee Opinion No. 728: Müllerian Agenesis: Diagnosis, Management, And Treatment. Obstet Gynecol. 2018 Jan;131(1):e35-e42. doi: 10.1097/AOG.0000000000002458. PMID: 29266078.
14. Creighton S. Common congenital anomalies of the female genital tract. Rev Gynecol Pract. 2005;5:221–26
15. Siegel M. Pediatric applications. In: Computed body tomography with MRI correlation on CD-ROOM. Philadelphia: Lippincott-Raven; 1998.
16. Morcel K, Camborieux L; Programme de Recherches sur les Aplasies Müllériennes; Guerrier D. Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome. Orphanet J Rare Dis. 2007 Mar 14;2:13. doi: 10.1186/1750-1172-2-13. PMID: 17359527; PMCID: PMC1832178.
17. D' Alberton A, Reschini E, Motta T. La sindrome di Rokitansky. Aggiornamento Del Medico. 1987;11.
18. Bianchi S, Von Wunster S, Marchini M, D' Alberton A, Motta T. Long-Term Satisfaction following Formation of a Neovagina By Regular Sexual Intercourses. Seattle, Wash, USA: The American Society For Reproductive Medicine; 1995
19. Motta T, Von Wunster S, De Giorgi O, D'Alberton A. The "functional method" as initial approach to the formation of a neovagina. In: Monduzzi, editor. Proceedings of the 6th European Congress on Pediatric and Adolescent Gynecology. Budapest, Hungary; 1993.