Imaging for Residents – Answer

A 19-year-old Female with Primary Amenorrhea: Mayer– Rokitansky–Küster–Hauser Syndrome

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Section 2 - Answer

CASE

A 19-year-old female patient presented with primary amenorrhea to the gynecology outpatient clinic. On examination, her height and weight were within the normal limits, and she had normal-appearing external female genitalia and breasts along with normal axillary and pubic hair development. There was no significant past, medical, or family history. The patient's blood investigations, including serum hormonal profile, were within the normal limits. Serum hormonal profile included normal serum-luteinizing hormone, follicular-stimulating hormone, and androgen levels.

In view of primary amenorrhea, ultrasonography of the whole abdomen was performed, which showed the absence of uterus and cervix. Bilateral ovaries were normal in appearance but were present in bilateral iliac fossae. The left kidney was not visualized in the left renal fossa. The right lumbar region showed the presence of two kidneys fused with each other forming a shape of L [Figure 1].

The patient was further subjected to magnetic resonance imaging (MRI), which showed a complete absence of uterus, cervix, and proximal vagina; the lower 1/3rd of the vagina was visualized and measured 2.7 cm in length. Bilateral ovaries, which were normal in size, shape and had maturing follicles, were seen in iliac fossae. Crossed-fused renal ectopia (L shaped) was seen in the right lumbar region [Figure 2]. There were no vertebral body anomalies. A diagnosis of Mayer–Rokitansky–Küster–Hauser (MRKH) syndrome Type II with bilateral ovarian maldescent was made based on clinical, laboratory, and radiological findings. The patient and her parents were counseled for further course of action. As she was about to get married, she consented for vaginoplasty. She

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underwent vaginoplasty under general anesthesia and had an uneventful postoperative course and recovery.

DISCUSSION

MRKH syndrome is a Class-I congenital Mullerian anomaly, with an incidence of 1 in 4000 females. The syndrome is divided into two types. There is an isolated absence of the uterus and proximal two-thirds of the vagina in Type I MRKH syndrome. There are associated malformations in Type II MRKH syndrome, including vertebral, cardiac, urological (upper tract), and otological anomalies. MRI is the noninvasive imaging modality of choice for detailed anatomical evaluation of the uterus, ovaries, and vagina. Surgery is necessary to restore normal sexual function; even reproduction may be possible if assisted reproductive techniques are performed.^[1,2]

The MRKH syndrome was first described by Mayer and then by Rokitansky, whereas Hauser and Schreiner described the distinguishing features of MRKH syndrome from androgen insensitivity syndrome (AIS).^[3] The associations of MRKH syndrome are genital, renal, and ear syndrome and Mullerian, Renal, and Cervical Somite dysplasia.^[3]

The MRKH syndrome arises due to the arrested development of Mullerian ducts 7 weeks after fertilization. The Mullerian ducts form the uterus, cervix, upper 2/3rd of the vagina, and fallopian tubes. MRKH syndrome has two subtypes: The typical (also called Rokitansky sequence, Type I, Type A or isolated) and the atypical form (Type II or Type B). In patients with typical form, the only affected part is the caudal part of the Müllerian duct. The atypical form of MRKH syndrome is associated with other anomalies, including renal anomalies

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305

Khanna and Saran: Radiological evaluation of a young female with primary amenorrhea



Figure 1: Ultrasound pelvis showing (a) the absence of uterus in its expected location, urinary bladder is normally outlined; (b) Left kidney was not visualized in left renal fossa, ultrasound of right lumbar region showing the presence of two kidneys fused with each other forming a shape of L (calipers); (c and d) Bilateral ovaries are normal in appearance (calipers) present in bilateral iliac fossae. f: Dominant follicle

such as unilateral agenesis (most common), ectopia of one or both kidneys, or horseshoe kidney. In addition, vertebral anomalies, hearing defects, skeletal abnormalities, ovarian cancers, and heart malformations are other associations. Type B or the atypical form of MRKHS is more common than the isolated form.^[1]

Ultrasonography is the initial investigation, followed by MRI, which adds more information about uterovaginal anatomy and associated anomalies and helps in planning for the surgery.^[2,4]

The MRKH syndrome should be differentiated from AIS and isolated vaginal hypoplasia or atresia. In AIS, a genotypically male (46XY) fetus will develop end-organ resistance to androgen, resulting in the virilization of the external genitalia resulting in the female phenotype of baby with the development of female secondary sexual characteristics and undescended testes. MRI can detect the absence of uterus and ovaries with the presence of rudimentary undescended testis in AIS. Normal serum-luteinizing hormone and follicular-stimulating hormone with no sign of androgen excess are the other features that differentiate between MRKH syndrome and AIS.^[5]

The treatment approach in MRKH syndrome concentrates on two aspects: Restoration of normal sexual function and assisted reproduction. Restoration of normal sexual function is achieved by creating neovagina by two means: surgical and nonsurgical. In nonsurgical technique (Frank's Technology), neovagina is created by vaginal dilators, whereas in the surgical method, it is formed by either sigmoid neovaginoplasty or Vecchietti operation. Assisted reproduction is achieved by fetching normal ovarian follicles from the patient and use them for



Figure 2: Magnetic resonance imaging pelvis with (a) T2 Fat sat sequence in the sagittal plane showing normal urinary bladder with the absence of uterus and cervix; (b) coronal plane showing L-shaped kidney (arrows); (c and d) normal ovaries present in bilateral iliac fossa (circles)

in vitro fertilization and surrogacy. Through this technique, genetically related offspring can be obtained.^[6]

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given her consent for her images and other clinical information to be reported in the journal. The patient understands that her name and initials will not be published, and due efforts will be made to conceal the identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

REFERENCES

- Strübbe EH, Willemsen WN, Lemmens JA, Thijn CJ, Rolland R. Mayer-Rokitansky-Küster-Hauser syndrome: Distinction between two forms based on excretory urographic, sonographic, and laparoscopic findings. AJR Am J Roentgenol 1993;160:331-4.
- Boruah DK, Sanyal S, Gogoi BB, Mahanta K, Prakash A, Augustine A, *et al.* Spectrum of MRI appearance of Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome in primary amenorrhea patients. J Clin Diagn Res 2017;11:TC30-5.
- Duncan PA, Shapiro LR, Stangel JJ, Klein RM, Addonzizio JC. The MURCS association: Müllerian duct aplasia, renal aplasia, and cervicothoracic somite dysplasia. J Pediatr 1979;95:399-402.
- Fiaschetti V, Taglieri A, Gisone V, Coco I, Simonetti G. Mayer-Rokitansky-Kuster-Hauser syndrome diagnosed by magnetic resonance imaging. Role of imaging to identify and evaluate the uncommon variation in development of the female genital tract. J Radiol Case Rep 2012;6:17-24.
- Khan S, Mannel L, Koopman CL, Chimpiri R, Hansen KR, Craig LB. The use of MRI in the pre-surgical evaluation of patients with androgen insensitivity syndrome. J Pediatr Adolesc Gynecol 2014;27:e17-20.
- Michala L, Cutner A, Creighton SM. Surgical approaches to treating vaginal agenesis. BJOG 2007;114:1455-9.