Case Report

A Case of Mayer–Rokitansky–Küster–Hauser Syndrome with a Fused Pancake-shaped Pelvic Kidney

Ali Reza Eftekhari Moghadam, Ghasem Saki, Mahin Taheri Moghadam, Seyed Mohamad Hossein Mohseni[1], Vajieh Heidari[2], Mohammad Hossein Jamshidi[3]

From the Department of Anatomical Science, Faculty of Medicine, Ahvaz Jundishapur University of Medical Sciences, [1]Golestan Medical Imaging Center, Ahvaz Jundishapur University of Medical Sciences, [3]Department of Radiologic Technology, Faculty of Paramedicine, Ahvaz Jundishapur University of Medical Sciences, Ahvaz, Iran, [2]School of Health and Biomedical Science, Royal Melbourne Institute of Technology University, Melbourne, Australia

Address for correspondence:
Dr. Ali Reza Eftekhari Moghadam, Department of Anatomical Science, Faculty of Medicine, Ahvaz Jundishapur University of Medical Sciences, Ahvaz, Iran. E-mail: eftekhari.ar@ajums.ac.ir

Abstract

Mayer–Rokitansky–Küster–Hauser (MRKH) Syndrome is a female reproductive system disorder. It is characterized by a defect in the Müllerian ducts development, and it causes the absence of the uterus in variable degrees in upper vaginal hypoplasia. In addition, it is often associated with the unilateral renal dysplasia. Müllerian agenesis affects 1 in 4500 newborn girls and is considered as a sporadic anomaly. Women with MRKH Syndrome have a normal female chromosome pattern 46, XX with normal ovarian function. The presence of bilateral kidney agenesis with a pelvic pancake-shaped kidney is a rare condition, and a few cases have been reported in medical journals. This case study focuses on a case of MRKH Syndrome with bilateral renal agenesis and a pancake-shaped kidney.

Keywords: *Mayer–Rokitansky–Küster–Hauser Syndrome, Müllerian ducts, renal agenesis*

Introduction

During the first 6 weeks of gestation, the Müllerian ducts (or paramesonephric ducts) begin to form laterally to the mesonephric ducts, in both male and female embryos. The caudal tips of the Müllerian ducts grow further to connect with the developing of the pelvic urethra. Before the development of the pelvic urethra, the tips of the Müllerian ducts adhere to each other. The cranial ends of the Müllerian ducts form funnel-shaped openings into the coelom.[1] In the absence of SRY gene, the mesonephric ducts disappear, and the Müllerian ducts do not develop properly. The wall of the pelvic urethra, at the end of the Müllerian ducts, thickens forming a paramesonephric tubercle. The tips of the Müllerian ducts fuse and connect with the paramesonephric tubercle. Then, the Müllerian ducts begin to fuse from their caudal tips cranially to form a short tube with a single lumen. This tube, called the uterovaginal canal, becomes the uterus and cranial part of the vagina. The unfused, cranial portions of the Müllerian ducts become the uterine tubes.[2] The primary evolution of the paramesonephric ducts rely on Wnt-4 signaling; thus, Wnt-4 deficiency can lead to the absence of paramesonephric ducts formation. It seems that Wnt-7a is involved in the maintenance of the expression of a sequence of homeobox (Hox) genes (Hoxd-10, Hoxd-13, and the Hoxa paralogues), which are expressed throughout the reproductive tract. Hoxa-9 is expressed in the fallopian tubes, Hoxa-10 in the uterus, Hoxa-11 in the uterus and cervix, and Hoxa-12 in the upper vagina.[3] In the absence of Hoxa-10, the cranial part of the uterus transforms into the uterine tube. In contrast to other regions of the body, the Hoxa gene expression throughout the female reproductive tract continues into adult life.[3,4]

Mayer–Rokitansky–Küster–Hauser (MRKH) Syndrome is a disorder that affects the female reproductive system. Although the external genitalia are normal, this condition prohibits the vagina and uterus development. Affected women usually do not have menstrual periods due to the absent uterus. Consequently, the first noticeable sign of MRKH Syndrome is the nonappearance of menstruation by the age of 16 years (primary amenorrhea). Women with MRKH Syndrome have a normal female chromosome pattern (46, XX) and normal ovaries. They also have normal breasts, pubic hair development, and other secondary sexual characteristics. The incidence of the congenital absence of the uterus and vagina is unknown. The

How to cite this article: Eftekhari Moghadam AR, Saki G, Taheri Moghadam M, Hossein Mohseni SM, Heidari V, Jamshidi MH. A Case of Mayer–Rokitansky–Küster–Hauser Syndrome with a Fused Pancake-shaped Pelvic Kidney. Adv Biomed Res 2019;8:35.

Received: December, 2018. **Accepted:** April, 2019.

Access this article online

Website: www.advbiores.net

DOI: 10.4103/abr.abr_97_18

Quick Response Code:

incidences of MRKH Syndrome vary from 1 in 4000 to 5000 newborn girls.[5] The examination reveals an absence or hypoplasia of the upper portion of the vagina and uterine agenesis. There are three types of MRKH Syndrome: the typical (Type I), the atypical form (Type II), and MURC (Type III), which consist of Müllerian duct (MU) aplasia, renal (R) aplasia, and cervicothoracic somite dysplasia. The typical form is characterized by Müllerian oddment (muscular buds) and normal uterine tubes (Rokitansky sequence); however, the caudal part of the Müllerian duct is affected.[6] The atypical form, in addition to Type I characteristics, is demonstrated by asymmetric hypoplasia of Müllerian remnants, with or without dysplasia of the uterine tubes. It is often related to other abnormalities such as renal defects (unilateral agenesis or ectopic kidneys and a horseshoe kidney in 40%–60% of patients). Almost 20% of affected women have hearing problems, cervicothoracic scoliosis, and Klippel-Feil syndrome.[7] These complications show the distinction between the MRKH Syndrome from other abnormalities of the genital tract, such as testicular feminization syndrome or Turner's Syndrome.[8,9] There have been few cases of a shapeless pelvic kidney, combined with bilateral renal agenesis or obstacle vagina. This study examines the Type 2 of MRKH Syndrome, with a pancake-shaped pelvic kidney, which is supplied by the median sacral artery.

Case Report

A 28-year-old married woman with a primary amenorrhea was referred to our medical imaging department for further evaluation of a shapeless pelvic mass, and to investigate the possibility of having functional ovaries and uterus. Initial gynecological studies such as height, weight, body mass index, and secondary sexual characteristics were performed by a gynecologist. Secondary sexual characteristics of the patient indicated the presence of pubic and axillary hair (Tanner Stage III), her breast examination showed Tanner Stage IV, and the vaginal examination showed a blind vagina. The uterus was not obvious by physical examination. Larche and pubarche, in this case, started at around the age of 13 years with no history of amenorrhea in her family. The laboratory tests, including the hormonal serum profile, showed the normal range of follicle stimulating hormone (FSH) (2.5 mIU/ml), luteinizing hormone (3.5 mIU/ml), serum creatinine (0.6 mg/dl), and prolactin (9.45 ng/ml). However, the concentration of anti-Müllerian hormone (AMH) was below the normal range (1.5 ng/ml). Moreover, a positive buccal smear, (for determination of gender by observing Barr bodies in the exfoliative cells of both men and women), was also normal. A bilateral renal agenesis with a 95 mm × 72 mm ectopic pancake-shaped kidney on the left side of the pelvic cavity was seen in the ultrasound examination. In addition, the complete absence of the uterus, the cervix, and the upper part of the vagina was also identified during the ultrasound procedure. The size of the ovaries was normal with the echo pattern of follicles between 10 and 14 mm in diameter.

The patient also underwent a pelvic magnetic resonance (MR) imaging at 1.5T (Magnetom Avanto Siemens Medical Solution, Erlangen, Germany). The conventional MR images consisted of: a sagittal T_2-weighted turbo spin-echo (TSE) (turbo spin-echo), 3 mm thickness, a small field of view (sFOV), 4500/120 ms (repetition time/echo time), with 320 × 320 matrix sagittal T_2W, fat saturation, the (3 mm thickness sFOV, 3500/120 ms), T_2W coronal oblique images (3 mm thickness sFOV, 4000/120 ms), and T_1W TSE with fat saturation axial oblique (3 mm, sFOV, 400/25 ms) with 256 × 256 MATRIX. After intravenous administration of gadobutrol 0.1 mmol/kg body weight (Gadovist, Bayer Pharma, Germany), the contrast-enhanced T1W and TSE images were obtained in axial, coronal, and sagittal planes. MR images confirmed ultrasound findings, including the uterovaginal atresia with normal ovaries in shape and size [Figures 1 and 2]. The coronal section revealed an ectopic pancake-shaped kidney with no evidence of hydronephrosis [Figure 3]. An interesting feature in this patient was the existence of the middle sacral artery in the blood supply of the pelvic kidney. An MR angiogram (time of flight technique) showed that the middle sacral artery was the principal artery that supplied the ectopic pelvic kidney [Figure 4].

Discussion

The Müllerian ducts are indispensable for the development of the female internal genital system. The Müllerian ducts fuse and become the uterine tube, the corpus, and the cervix of the uterus and upper portion of the vagina. The lower third of the vagina derives from the urogenital sinus. Müllerian ducts and urogenital sinus are attached through the sinovaginal bulb.[10] The presence of the AMH is an essential factor for the maturation of Müllerian ducts to the previously mentioned parts of the female internal

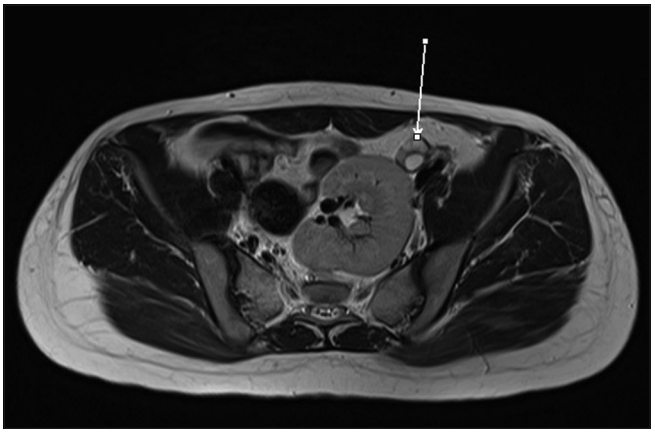

Figure 1: Axial T_2-weighted magnetic resonance image of the pelvis shows a complete absence of the uterus, the cervix and the vagina, with normal ovarian signal intensity. The white arrow shows the left ovary

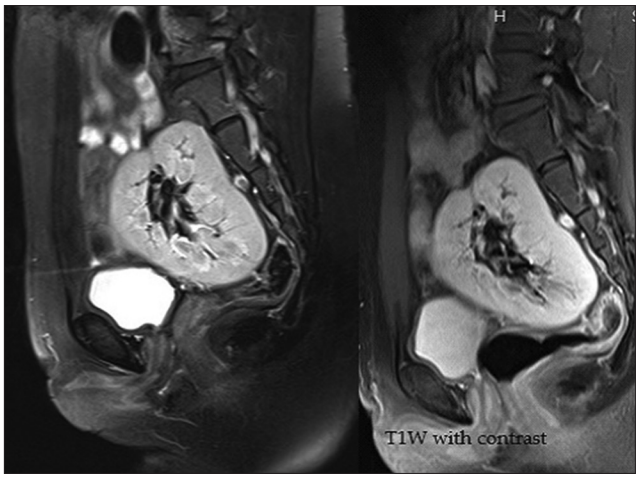

Figure 2: Sagittal T_2-weighted with fat saturation and T_1-weighted with contrast show uterovaginal atresia

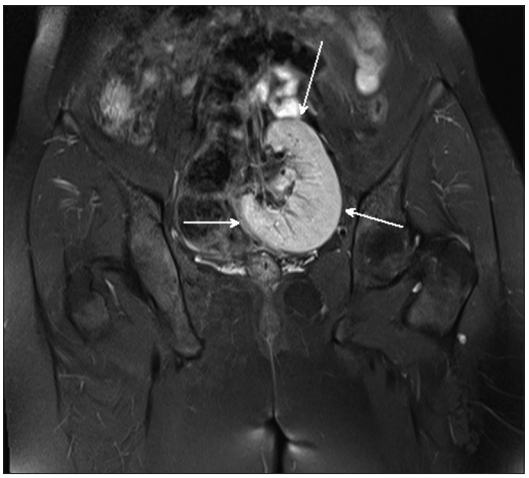

Figure 3: Coronal section magnetic resonance imaging of the pelvis (proton density with fat saturation) shows a left side pancake-shaped kidney. The white arrow shows the ectopic kidney

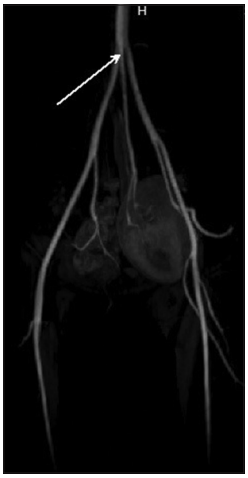

Figure 4: Magnetic resonance angiography (anterior view) of pelvic main vessels shows that the median sacral artery supplies the pelvic kidney. The white arrow shows the median sacral artery

genital system. A lack of AMH and the fusion of the paramesonephric duct in a local area or the line of fusion, cause abnormalities such as uterine and vaginal agenesis or duplication of the uterus and the vagina.[11] MRKH Syndrome is a congenital malformation that is the cause of 15% of cases with primary amenorrhea.[5] Because the ovaries do not develop from the paramesonephric ducts; women with this syndrome might have normal secondary sexual characteristics.[12] There may be different kinds of abnormalities in the Müllerian ducts, ranging from total aplasia to small anatomical changes. Patients with Müllerian agenesis have the normal 46, XX pattern of chromosomes and the visual characteristics of the chromosomes, but primary amenorrhea is seen in adolescents.

Chromosomal abnormality in Müllerian aplasia is uncommon.[3] It is estimated that 40%–60% of women have renal disturbances such as unilateral, horseshoe, and ectopic kidneys. Moreover, 20% of women with MRKH Syndrome have bone changes such as vertebral spine fusion, scoliosis in addition to hearing and cardiac abnormalities. MRKH Syndrome has different grades: (1) the typical form of MRKH includes uterovaginal aplasia or hypoplasia; (2) The atypical MRKH comprises uterovaginal aplasia or hypoplasia with renal malformation, along with ovarian dysfunction; and (3) MURC Syndrome, which consists of uterovaginal aplasia and hypoplasia with renal, skeletal, and heart abnormalities.[7] Oppelt *et al*. showed that 64% of patients with MRKH had a typical form, 24% an atypical, and 12% had MURC Syndrome.[7] No vertebral body malformation was detected in the patient examined. The diagnostic assessment for the MRKH Syndrome included an abdominal and pelvic cavity ultrasound, MR imaging, hormonal assay for FSH, luteinizing hormone, testosterone, estradiol, and AMH. In our view, based on the correlated symptoms, an audiogram or electromyogram should have been carried out. The biochemical study of the patient was normal and showed that she had normally functioning ovaries. Although the development of MRKH Syndrome is well demonstrated, its cause remains unknown.[13] Approximately 2%–7% of patients have a nonfunctioning uterus, while most women with the Müllerian agenesis have a mostly functioning uterus.[11] The treatments usually comprise surgical or nonsurgical procedures when these women are ready to commence sexual activity. Vecchietti surgery is a combination of surgical and nonsurgical techniques for creating a neovagina and has been used as an appropriate treatment over the last 20 years.[11] *In vitro* fertilization and surrogate pregnancy could be used as an alternative for patients who want to have a child in the future.

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 name and initial will not be published and due efforts

will be made to conceal identity, but anonymity cannot be guaranteed.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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