

Mayer-Rokitansky-Kuster-Hauser Syndrome with Pelvic Fused Kidney. A Case Report

Síndrome de Mayer-Rokitansky-Kuster-Hauser asociado a riñón pélvico fusionado. Presentación de caso

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Palabras clave (DeCS)

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Summary

We present a case of Mayer-Rokitansky-Kuster-Hauser Syndrome type II associated with fused pelvic kidneys diagnosed by MRI. While MRKH Syndrome is a well-recognized entity associated with various Mullerian structure malformations, we did not find any similar cases in our review of the literature associated specifically with fused pelvic kidney ectopy. A brief review of the syndrome and its main findings will be done by imaging.

Resumen

Se presenta un caso de Síndrome de Mayer-Rokitansky-Kuster-Hauser tipo II asociado a riñón pélvico fusionado diagnosticado por resonancia magnética. Si bien el síndrome de MRKH es una entidad bien reconocida asociada a diversas malformaciones de las estructuras müllerianas, en esta revisión de la literatura no se han encontrado casos similares asociados específicamente a ectopia renal pélvica fusionada. Se hará una breve revisión del síndrome y sus principales hallazgos por imagen.

Introduction

Mayer-Rokitansky-Kuster-Hauser syndrome (MRKH) is a rare congenital anomaly of the Müllerian ducts, which is characterized by the absence of the uterus and the upper two thirds of the vagina, with an incidence of 1 per 4,000 to 5,000 female births (1). The patients have normal functional ovaries and therefore a normal phenotypic appearance of the external genitalia, while the vagina may occasionally constitute a small pocket 2 to 4 centimeters long or be completely absent (2).

MRKH syndrome is divided into two subtypes depending on whether it is isolated or associated with other malformations. MRKH type 1 syndrome refers to isolated utero-vaginal aplasia. MRKH type 2 syndrome refers to incomplete aplasia associated with other malformations, within which renal and skeletal malformations and hearing defects have been described. The most frequently described renal abnormalities in the literature include unilateral renal agenesis, renal ectopy, and horseshoe kidney (1, 3-5).

The typical clinical presentation is a woman around age 16 with primary amenorrhea. These patients present a normal female phenotype with development of secondary sexual characteristics due to normal ovarian function.

Case presentation

16-year-old female patient consulting for primary amenorrhea without any other associated symptoms.

Physical examination reveals normal appearance of secondary female sexual characteristics. A pelvic ultrasound was performed in which the uterus and ovaries were not found. Magnetic resonance imaging (MRI) was obtained, revealing the absence of a uterus, cervix, and proximal vagina (Figure 1) and the presence of right (Figure 2) and left (Figure 3) ovaries high in the abdominal cavity. Additionally, pelvic kidney was demonstrated with two separate renal units and hilum, with ectopic pelvic kidney fused (Figure 4). A T11 vertebral body segmentation anomaly consistent with butterfly vertebra was also identified.

Discussion

It has been established that MRKH type II syndrome is associated with various renal abnormalities, of which unilateral renal agenesis, pelvic kidney, and horseshoe kidney are most frequently described in the literature (1, 3-5).

In a study conducted by Hall-Craggs and collaborators (4) in which 52 patients with a previous diagnosis of MRKH syndrome had MRIs, they found abnormal kidneys in 13 women. Among the renal abnormalities described were renal agenesis, renal ptosis, horseshoe kidney, pelvic kidney, atrophic kidney, and bilateral renal scarring. In another study carried out by Oppelt and collaborators (6), MRI was performed on 53 patients with MRKH syndrome, in which abnormal kidneys were found in 19 women. The renal abnormalities found were:

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12 cases of unilateral renal agenesis, 9 cases of unilateral pelvic kidney, and two sclerotic kidneys.

Among the differential diagnoses that should be considered are Mullerian aplasia and hyperandrogenism (WNT4 syndrome) and androgen insensitivity syndrome, in which the absence of a uterus and upper vagina is also described: in the first case associated with hyperandrogenism and masculinized ovaries, and in the second case associated with the presence of testicles. Another diagnosis to take into account is isolated vaginal atresia (3, 5, 7).

The treatment of this condition is focused on the creation of a neo-vagina to achieve a normal sexual life (initially by non-surgical methods using vaginal dilators and, if necessary, by surgical procedure). Patients who have a desire for fertility have the option to resort to in vitro fertilization with their own oocytes and the surrogate mother's uterus. The prognosis of this condition is good; however, psychological support is recommended due to the stress it may cause in the patient's life (2, 3, 4).

Despite the fact that this entity and its association with renal anomalies is widely described in the literature, we present a unique case in which MRKH syndrome is associated with fused pelvic renal ectopy.



Figure 1. Pelvic MRI, with sagittal T2 information: pelvic kidney (black arrow). Absence of uterus, cervix and proximal vagina (white arrow).



Figure 3. Abdominopelvic MRI with T2 coronal information: fused pelvic kidney (black arrow), absence of uterus and vagina, left ovary in high position (white arrow).

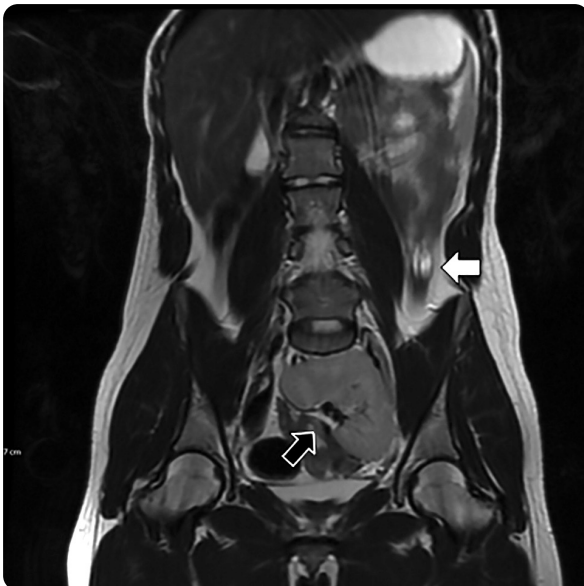


Figure 2. Pelvic MRI with coronal T2 information: right ovary in high position (white arrow) and fused pelvic kidney (black arrow).

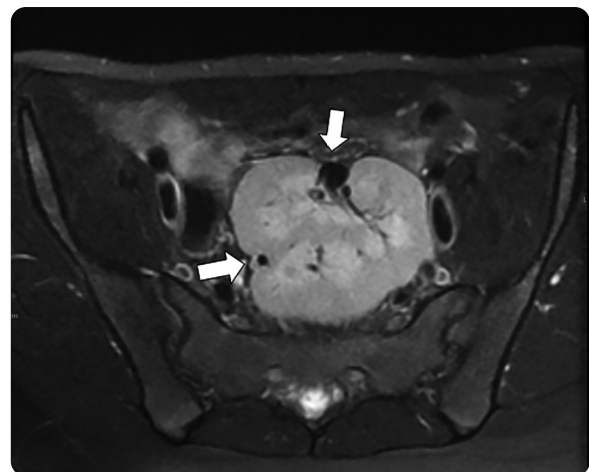


Figure 4. MRI of pelvis STIR axial sequence: pelvic kidney with two renal units and two independent threads (white arrows): fused pelvic kidney.

References

1. Strübbe EH, Willemsen WN, Lemmens JA, et al. Mayer-Rokitansky-Kuster-Hauser syndrome: distinction between two forms based on excretory urographic, sonographic, and laparoscopic findings. *Am J Roentgenol.* 1993;160:331-4.
2. Giusti S, Fruzzetti E, Perini D, et al. Diagnosis of a variant of Mayer-Rokitansky-Kuster-Hauser syndrome: useful MRI findings. *Abdominal Imaging.* 2011;36(6):753-5.
3. Morcel K, Camborieux L, Guerrier D. Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome. *Orphanet J Rare Dis.* 2007;2(1):13.
4. Hall-Craggs MA, Williams CE, Pattison SH, et al. Mayer-Rokitansky-Kuster-Hauser syndrome: diagnosis with MR imaging. *Radiology.* 2013;269(3):787-92.
5. Boruah DK, Sanyal S, Gogoi BB, et al. Spectrum of MRI appearance of Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome in primary amenorrhea patients. *JCDR.* 2017;11(7):TC30.
6. Oppelt P, Renner SP, Kellermann A, et al. Clinical aspects of Mayer-Rokitansky-Küster-Hauser syndrome: recommendations for clinical diagnosis and staging. *Human Reproduction.* 2006;21(3):792-7.
7. Biason-Lauber A, Konrad D, et al. A WNT4 mutation associated with Müllerian-duct regression and virilization in a 46, XX woman. *New Eng J Med.* 2004;351(8):792-8.

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