

Mayer-Rokitansky-Küster-Hauser Syndrome with Scoliosis - A Case Report

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Abstract: *The Mayer-Rokitansky-Küster-Hauser syndrome (MRKH syndrome) is a congenital condition that is characterized by the absence of the uterus and vagina but ovaries are present and the external genitalia are normal. It affects at least 1 out of 4500 women. MRKH may be isolated (Type I) but it is more frequently associated with renal, vertebral and to a lesser extent auditory and cardiac defects (Type II or MURCS - Mullerian duct aplasia, Renal dysplasia and Cervical Somite anomalies). There are very few cases of MRKH syndrome with vertebral defect reported in the literature. To help establish the best criteria for early diagnosis and treatment options for a comprehensive therapeutic approach to MRKH patients, we report a case of MRKH syndrome with scoliosis who presented to us with primary amenorrhoea.*

Keywords: MRKH (Mayer-Rokitansky-Küster-Hauser syndrome), primary amenorrhoea

1. Introduction

Mayer-Rokitansky-Küster-Hauser syndrome is an uncommon condition, with an incidence of 1 in 4000-5000 female births^[1,2] and is the second most frequent cause of primary amenorrhoea after gonadal dysgenesis.^[3] Mayer-Rokitansky-Küster-Hauser syndrome (MRKH syndrome) is named after its most famous discoverer Baron Karl von Rokitansky (Czechoslovakia, 1804-1878) a physician and professor at the University of Vienna. In 1829 and 1838, Mayer-Rokitansky described a syndrome that includes agenesis of the uterus and vagina, while Küster then observed a correlation with urological defects.

The reproductive abnormalities of MRKH syndrome are due to incomplete development of the Müllerian duct. This structure in the embryo develops into the uterus, fallopian tubes, cervix and the upper part of the vagina. This mullerian duct abnormality is characterized by congenital aplasia of the uterus and the upper part (2/3) of vagina, in young women presenting otherwise with normal endocrine status. It may include absence or hypoplasia of the uterus and fallopian tubes. The patients present with normal secondary sexual characteristics, as the functional ovaries are present, but menstruation is absent.^[4]

Syndrome is classified into three types according to the involvement of other systems than reproductive system. The typical syndrome (type I) is represented by abnormalities restricted to the reproductive system. The second one (type II) is an atypical syndrome, with the presence of asymmetric uterine remnants and abnormal uterine tubes. Such syndrome type may be associated with ovarian disease, congenital renal, bone abnormalities and hearing defects. A third one, the so called MURCS type (Mullerian duct aplasia, Renal dysplasia and Cervical Somite anomalies) involves utero-vaginal hypoplasia or aplasia, renal, bone and cardiac malformations^[1-3].

Renal malformations include: unilateral agenesis, horse-shoe kidney, renal hypoplasia, ectopic kidneys and

hydronephrosis. Bone malformations occur particularly in the vertebrae, most commonly with vertebral fusion (particularly cervical vertebrae), Klippel-Feil syndrome and scoliosis. Cardiac alterations and digital alterations such as syndactyly and polydactyly are rarer than those previously mentioned^[1].

The cause of syndrome remains unknown, but the increased number of cases in familial aggregates raises the hypothesis of a genetic cause^[1].

2. Case History

- 17 year old unmarried female presented with no onset of menstrual cycle (primary amenorrhoea).
- There was no history of parental consanguinity and no other sibling had similar complaints.
- No significant past history.

On Examination

- Secondary sexual characters revealed presence of pubic hair, axillary hair, normal breast development (Tanner stage 3) and external genitalia.
- Scoliosis was noted.
- Uterus could not be felt on per vaginal and per speculum examination.
- Respiratory, cardiovascular, per abdomen and central nervous system examination were normal.
- Vitals - stable

Investigations

- Routine investigations were within normal limits.
- The hormonal profile (Serum FSH, LH, prolactin, ovarian cancer antigens) was within normal limits.
- Gene karyotyping showed 46XX.

USG of abdomen and pelvis

- Hypoplastic / infantile uterus measuring 3.8 x 1.4 x 2.6 cm in size (Image 1)
- Normal bilateral ovaries (Image 2)

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- Hypertrophied left kidney and a small right kidney (Image 3 and Image 4)

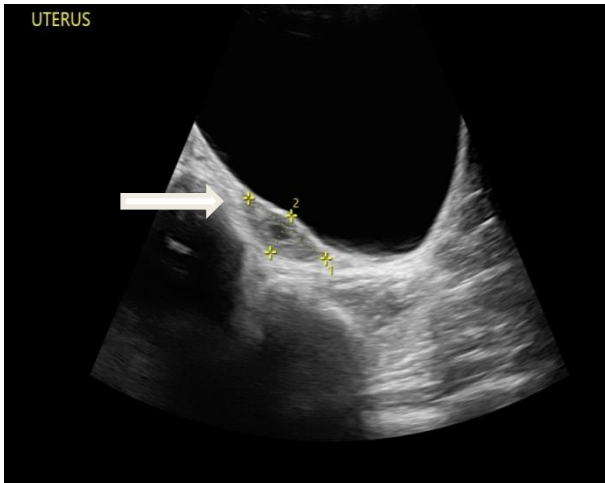


Image 1: USG showing small and hypoplastic uterus

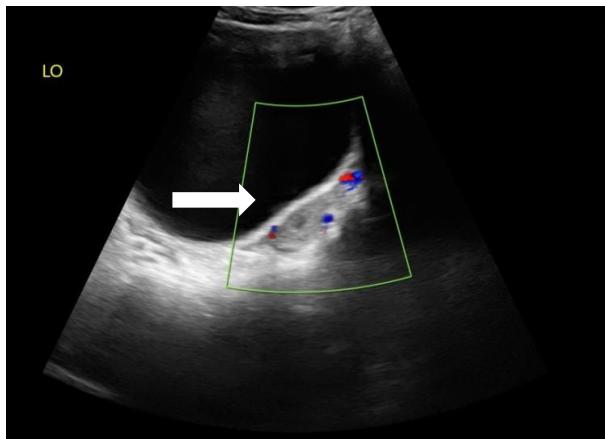


Image 2: USG showing normal left ovary

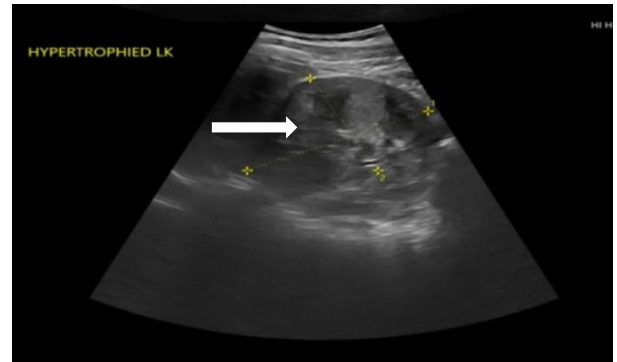


Image 3: USG showing hypertrophied left kidney

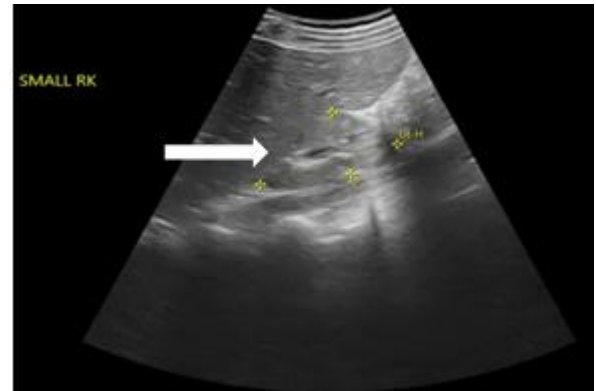
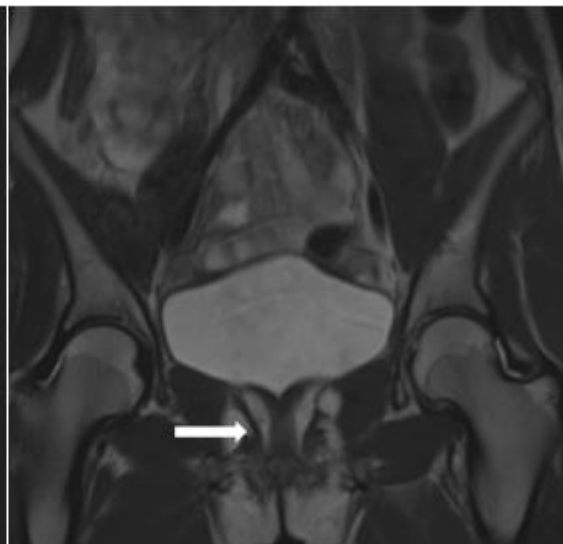


Image 4: USG showing small right kidney

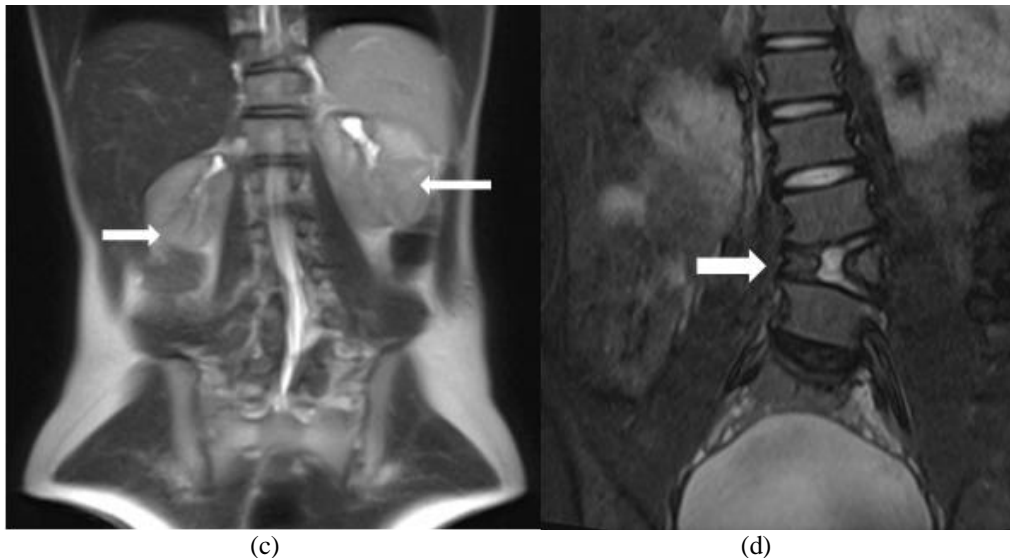
MRI



(a)



(b)



T2WI showing (a) Hypoplastic / infantile uterus (b) Bicornuate uterus (c) Hypertrophied left kidney and a small right kidney (d) Scoliotic spine deformity with butterfly vertebra

From the above history, examination and investigation findings a diagnosis of MRKH syndrome was made.

3. Discussion

The typical clinical presentation of this syndrome is primary amenorrhea in an adolescent with secondary sexual characteristics compatible with age with no signs of virilisation. Gynaecologic examination may detect either absence of the vaginal canal or vaginal shortening [1,2,5-8].

Imaging studies such as ultrasonography and magnetic resonance imaging, in association with or without laparoscopy, are necessary to allow the determination of the anatomic characteristics of the syndrome. Ultrasonography is the initial method of choice. This method can demonstrate the absence of the uterus between the bladder and the rectum [1,5,9]. Magnetic resonance imaging is the most sensitive and specific imaging method in the evaluation of this syndrome, not only for allowing the acquisition of multiplanar images, but also for allowing the acquisition of sequences with fat saturation. It allows a good definition of anatomical alterations such as uterine agenesis, as well as evaluating ovaries, vagina and associated anomalies [1,4,5,9].

Laparoscopy is indicated only in cases where the evaluation by the two previous imaging methods is inconclusive and provided this method allows the definition of a therapeutic strategy. Once the diagnosis of Mayer-Rokitansky-Kuster-Hauser syndrome is established, a clinical investigation should be undertaken to identify possible associated malformations [1,4].

The final diagnosis is achieved by the association of the imaging findings with the presence of the karyotype 46, XX. The differential diagnosis should be made with other situations where the patient presents primary amenorrhea and normal secondary sexual characteristics, such as congenital absence of uterus and vagina, isolated vaginal atresia with androgen insensitivity syndrome and transverse vaginal septum with imperforate hymen [1]. Because of the typical anatomic alterations, MRKH syndrome generates

anxiety and psychological distress with consequences on the patient's quality of life, thus requiring a multidisciplinary approach [5,6]. The indicated anatomic treatment is the surgical or non-surgical creation of a neovagina, which may allow these patients to have a normal sex life [1-6]. As the surgical approach is chosen, uterine remnants can be removed to avoid future endometriosis [1].

Patients who want to have children should be encouraged to adopt, or the possibility of having biological children by means of assisted reproduction techniques should be suggested, considering that the presence of functional ovaries in these women allow the production of normal ovaules. [10]

Even with advanced management of this syndrome, its diagnosis causes significant psychological distress, affecting the patients' quality of life because of the absence of menstruation and impossibility of pregnancy. The distress caused by the diagnosis may be alleviated by surgical or non-surgical treatments, counselling, family's support and with help of support groups. [10]

4. Treatment

The findings and implications regarding potential fertility and child bearing were explained to the patient. After initial non-operative treatment which included psychological counseling, the patient underwent vaginoplasty.

5. Conclusion

MRKH syndrome is one of the most common causes of primary amenorrhea and Ultrasonography is useful for diagnosing any associated renal anomalies. MRI is more precise than USG and less invasive and expensive than laparoscopy, contributing significantly to treatment planning and patient management. Although this condition has psychologically devastating consequences today anatomical defects can be surgically treated allowing a normal sexual function and reproduction thanks to the assisted techniques,

so correct evaluation of these patients and proper management is mandatory.

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