Scholars Journal of Medical Case Reports
Abbreviated Key Title: Sch J Med Case Rep
ISSN 2347-9507 (Print) | ISSN 2347-6559 (Online)
Journal homepage: https://saspublishers.com

OPEN ACCESS

Radiology

Mayer-Rokitansky-Küster-Hauser Syndrome as a Cause of Primary Amenorrhea: A Case Report

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DOI: 10.36347/sjmcr.2024.v12i07.015 | **Received:** 26.05.2024 | **Accepted:** 02.07.2024 | **Published:** 16.07.2024

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Abstract **Case Report**

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a rare congenital disorder marked by the absence of the uterus and upper two-thirds of the vagina in women with normal secondary sexual characteristics. We present the case of a 35-year-old woman with primary amenorrhea, where pelvic MRI played a crucial role in diagnosing MRKH syndrome by revealing a rudimentary uterus and normal ovaries. Imaging, particularly MRI, is essential for accurate diagnosis and assessment of associated abnormalities, ensuring a comprehensive evaluation of this condition.

Keywords: MRKH syndrome, Primary amenorrhea, Pelvic MRI, Congenital malformation.

INTRODUCTION

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome refers to a congenital malformation of the female genital tract due to interrupted embryonic development of the müllerian ducts. It is defined by a congenital aplasia of the uterus and the upper two-thirds of the vagina in women who exhibit normal development of secondary sexual characteristics and a normal karyotype. It remains a rare cause of primary amenorrhea [1].

We report the case of a 35-year-old women who presented with primary amenorrhea, in whom pelvic magnetic resonance imaging revealed Mayer-Rokitansky-Küster-Hauser syndrome.

CASE REPORT

We present a case of a 35-year-old female patient with a history of primary amenorrhea. Clinical examination revealed female external genitalia. Speculum examination revealed vaginal hypoplasia with no visualization of the cervix. To further investigate the patient's condition, a pelvic MRI was performed (Figure 1). Which revealed a rudimentary hypoplasic uterus and the upper two-thirds of the vagina with normal appearance and signal of the ovaries. There were no other associated malformations, particularly of the kidneys. Thus, allowing the diagnosis of Mayer-Rokitansky-Küster-Hauser syndrome type I has been established.

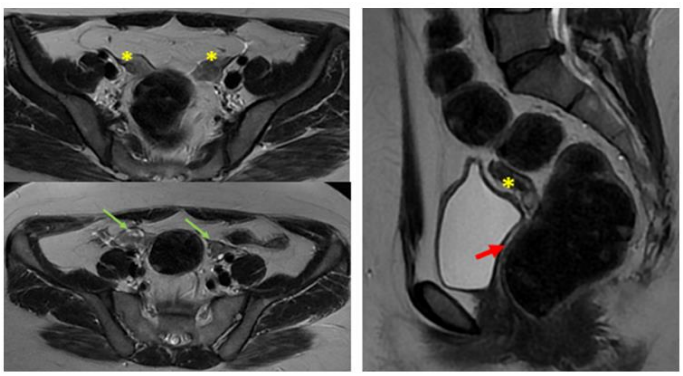

Figure 1: Pelvic MRI in axial an sagittal T2 sequences, showing a rudimentary hypoplasic uterus (yellow Asterix) and the upper two-thirds of the vagina (Red arrow) with normal appearance and signal of the ovaries (green arrows)

DISCUSSION

Mayer-Rokitansky-Küster-Hauser syndrome is a congenital disorder of the female genital tract caused by the maldevelopment of the mullerian ducts. It was initially described by the German anatomist Mayer in 1829, followed by Rokitansky reporting a similar case in 1938. The syndrome was first reviewed by Kuster in 1910. In 1961, Hauser and Schreiner coined the term Mayer-Rokitansky-Küster-Hauser to refer to the condition. The prevalence of the syndrome is estimated to be approximately 1 in 4,500 live births, making it the second most common cause of primary amenorrhea [2].

MRKH syndrome is characterized by varying degrees of underdevelopment of the uterus and vagina. While it was initially thought to be a sporadic disorder, evidence of familial clusters suggests the presence of a specific genetic basis for the syndrome. In general, patients with Mayer-Rokitansky-Küster-Hauser syndrome have a normal female karyotype of 46 XX, normal secondary sexual characteristics, and normal ovarian function [2].

It can occur as an isolated condition (type I) or be accompanied by other malformations (type II), affecting the kidney (unilateral renal aplasia, ectopic kidney, horseshoe kidney, double ureter), skeletal system (vertebral malformations like spina bifida or transitional lombo-sacral or cervical vertebrae). Although less common, cardiac malformations and neurological disturbances have also been reported. An atypical form of MRKH syndrome, known as MURCS syndrome, involves anomalies in all three systems: Mullerian duct aplasia, renal aplasia, and cervicothoracic somite dysplasia [3].

While clinical evaluation, hormonal studies, and karyotyping are essential for diagnosing (MRKH) syndrome, imaging plays a crucial role to provide a clear diagnosis, determining the extent of uterovaginal anomalies, identifying any associated abnormalities, and assisting in surgical planning.

Ultrasonography is a simple and non-invasive procedure commonly employed for initial imaging modality whitch allows for the suspicion of the diagnosis by showing the absence of uterine structures between the bladder and rectum. However, a retrovesical quadrangular structure can mistakenly be identified as a hypoplastic uterus. This structure corresponds to the vestigial fold located beneath the middle part of the transverse peritoneal fold. Additionally, it assists in evaluating for any associated renal anomalies. It may not always provide sufficient anatomical detail in cases of MRKH syndrome. Transvaginal imaging is often not feasible or appropriate for this specific group of patients. Therefore, alternative imaging modalities may be necessary to obtain a comprehensive evaluation of the uterovaginal anatomy in MRKH syndrome cases [4].

MRI is recommanded for obtaining more comprehensive and precise information in cases of MRKH syndrome. It enables a precise diagnosis through the use of sagittal and axial T2-weighted imaging sequences, confirming the uterine aplasia and the upper two-thirds of the vagina, as well as demonstrating the normal appearance of both ovaries. Furthermore, MRI allows for the detection of any associated malformations, such as renal and skeletal abnormalities [5].

Differential diagnosis should first consider vaginal atresia or a transverse vaginal septum, which can be confirmed through careful clinical examination. Another consideration is WNT4 gene mutation syndrome, which presents with a phenotype similar to MRKH syndrome, however, the anomalies are associated with signs of hyperandrogenism, which can be correlated with elevated plasma testosterone levels. Lastly, androgen insensitivity syndrome is a form of male pseudohermaphroditism. The phenotype appears feminine with the presence of testes in the abdominal or inguinal region and high levels of testosterone comparable to male individuals [5].

Treatment involves the reconstruction of a neovagina to enable the patient to have a normal sexual life. Psychological support is crucial for patients with MRKH syndrome [5].

CONCLUSION

In cases of primary amenorrhea in young women with well-developed physical sexual characteristics, it is important to consider Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome as a potential diagnosis. Imaging techniques, such as pelvic ultrasound and MRI, play a key role in confirming the presence of uterovaginal aplasia, evaluating ovarian integrity, and identifying associated malformations. However, it is essential to be knowledgeable about other syndromes with similar uterovaginal anomalies in order to avoid diagnostic confusion.

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