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Mini Case Study

Unveiling Mayer-Rokitansky-Küster-Hauser (MRKH) Syndrome: A Rare Genital Malformation in a 18-Year-Old with Primary Amenorrhea

IZI Zineb*, Basma Beqqali, Tlaite Oubaddi, Oumaima Mesbah, Badr Kabila, Nazik Allali, Siham El Haddad and Latifa Chat

Radiology Department, Children's Hospital, Mohamed V University, Rabat, Morocco

***Corresponding Author:** IZI Zineb, Radiology Department, Children's Hospital, Mohamed V University, Rabat, Morocco.

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Abstract

We report the case of an 18-year-old female presenting with primary amenorrhea. Clinical examination showed normal secondary sexual characteristics. MRI revealed complete agenesis of the uterus and vagina, with normal-appearing ovaries and renal tract. These findings were consistent with Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome. MRI plays a key role in diagnosis and management planning of Müllerian anomalies.

Keywords: *MRKH Syndrome; Müllerian Agenesis; Primary Amenorrhea; Uterine Agenesis; Vaginal Agenesis*

Abbreviation

MRKH: Mayer-Rokitansky-Küster-Hauser

Short Case Report

We report the case of a 18-year-old female patient with no significant medical history, presenting with primary amenorrhea. The clinical examination showed normal external genitalia and well-developed breasts, along with other secondary sexual characteristics. As the patient was a virgin, a vaginal examination was not performed. A pelvic ultrasound failed to detect the uterus, leading to further examination with MRI. The MRI confirmed complete absence of the uterus and vagina. The ovaries appeared normal, showing no signal or morphological abnormalities, and the renal tract was also normal. These findings led to the diagnosis of MRKH syndrome.

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome also referred to as Müllerian aplasia; is a rare congenital anomaly of the female genital tract characterized by uterine and vaginal agenesis with normal ovarian function, with normal female karyotype (46,XX) [1]. It results from the failure of the Müllerian ducts to develop properly during embryonic life. In its typical form, the uterus is reduced to two rudimentary structures, often associated with total or partial vaginal agenesis [2]. The atypical form includes asymmetrical uterine remnants and may involve malformations of the fallopian tubes or kidneys [3].

MRI is the diagnostic tool of choice for MRKH syndrome, as it effectively assesses the development of the uterus and vagina. Rudimentary uterine horns, when present, are best visualized on T2-weighted axial and coronal sequences as hypointense, elongated, or oval structures. MRI also accurately measures the length of the vaginal remnant, crucial for planning potential vaginoplasty, and identifies associated anomalies [4].

This case underscores the importance of MRI in diagnosing MRKH syndrome and highlights the need for comprehensive care involving gynecologists, radiologists, and mental health professionals.

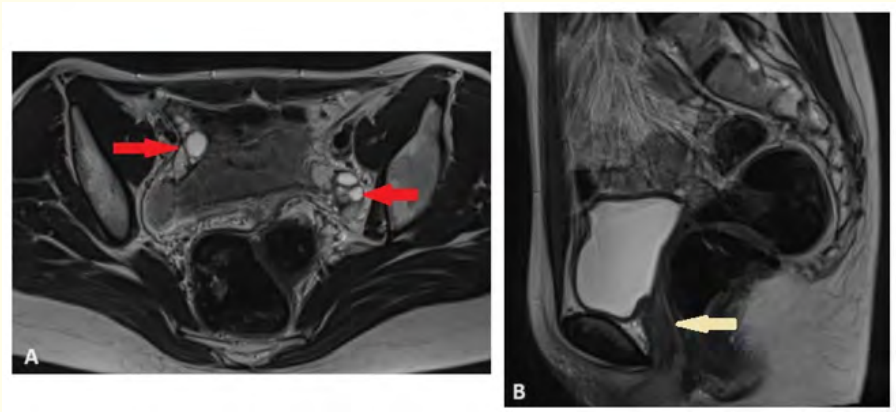

Figure 1: *(A, B) axial and sagittal MRI in T2-weighted sequence showing agenesis of the uterus and vagina (yellow arrow) with both ovaries have normal signal and morphology (Red arrows).*

Conclusion

This case underscores the critical role of MRI in diagnosing Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome, confirming uterine and vaginal agenesis while evaluating ovarian structures and associated anomalies. MRI also aids in planning potential reconstructive procedures and highlights the importance of a multidisciplinary approach, including gynecologic, radiologic, and psychological care.

Conflict of Interest

Declare if any financial interest or any conflict of interest exists.

Bibliography

1. Herlin M., *et al*. "Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome: A comprehensive update". *Orphanet Journal of Rare Diseases* 15.1 (2020): 214.

2. Kobayashi A and Behringer RR. "Developmental genetics of the female reproductive tract in mammals". *Nature Reviews Genetics* 4.12 (2003): 969-980.

3. Herlin M., *et al*. "Prevalence and patient characteristics of Mayer-Rokitansky-Küster-Hauser syndrome: a nationwide registry-based study". *Human Reproduction* 31.10 (2016): 2384-2390.

4. Govindarajan M., *et al*. "Magnetic resonance imaging diagnosis of Mayer-Rokitansky-Kuster-Hauser syndrome". *Journal of Human Reproductive Sciences* 1.2 (2008): 83-85.

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