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(CASE REPORT)

Mayer–Rokitansky–Küster–Hauser Syndrome: A Case Report and Review of the Literature

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Abstract

Mayer–Rokitansky–Küster–Hauser (MRKH) syndrome is a rare cause of primary amenorrhea. It is characterized by congenital aplasia of the uterus and the upper two-thirds of the vagina in women with normal development of secondary sexual characteristics. The diagnosis relies mainly on magnetic resonance imaging (MRI).

We report the case of a 27-year-old woman presenting with primary amenorrhea and well-developed secondary sexual characteristics. Hormonal investigations revealed normal ovarian function and a normal gonadotropic axis. Pelvic ultrasound and MRI demonstrated complete uterine agenesis, confirming the diagnosis of MRKH syndrome.

This report highlights the importance of suspecting MRKH syndrome in young women with normal secondary sexual characteristics presenting with primary amenorrhea, regardless of its type, and emphasizes the essential role of MRI in identifying specific features of this condition.

Keywords: Primary Amenorrhea; Uterovaginal Aplasia; MRI; Case Report

1. Introduction

Mayer–Rokitansky–Küster–Hauser (MRKH) syndrome is a rare congenital malformation of the female genital tract defined by agenesis of the uterus and vagina in the presence of normal ovaries. Magnetic resonance imaging (MRI) is the diagnostic modality of choice.

MRKH syndrome has a significant psychological impact and requires multidisciplinary management.

2. Patient and Observation

We report the case of a 27-year-old woman with no notable medical history, married for ten years, who was referred for evaluation of primary amenorrhea.

Clinical examination revealed normal external genitalia and well-developed breasts, with the presence of other secondary sexual characteristics. Vaginal examination showed a short vagina ending in a thin blind pouch, without a palpable cervix.

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Pelvic ultrasound revealed non-visualization of the uterus. Further evaluation with MRI confirmed complete uterine agenesis. The ovaries were present, with normal morphology and signal intensity, thus confirming the diagnosis of MRKH syndrome.

3. Discussion

Mayer–Rokitansky–Küster–Hauser syndrome is defined by congenital aplasia of the uterus and the upper two-thirds of the vagina in women with normal secondary sexual characteristics and a normal karyotype (46, XX) [1].

It results from a failure of Müllerian duct migration toward the urogenital sinus during embryonic development. In the typical form, the uterus is reduced to two rudimentary remnants, associated with partial or complete vaginal agenesis. The atypical form is characterized by asymmetric uterine remnants associated with tubal or renal malformations [2].

Endocrine evaluation, including plasma FSH, LH, and 17-estradiol levels, is usually normal, reflecting preserved ovarian function and an intact gonadotropic axis [4]. In our patient, hormonal assessment was normal.

Transabdominal pelvic ultrasound is the first-line imaging modality and may suggest the diagnosis by demonstrating the absence of uterine structures between the bladder and rectum. However, a quadrangular retro vesical structure may be mistakenly identified as a hypoplastic uterus; this corresponds to a vestigial lamina located beneath the transverse peritoneal fold. Associated malformations should always be investigated during ultrasound examination [5].

MRI is the gold standard for diagnosing MRKH syndrome, as it allows accurate assessment of uterine and vaginal development. Rudimentary uterine horns are often difficult to detect and may appear on T2-weighted axial and coronal images as elongated or oval hypointense structures resembling a corn cob [3].

MRI also enables precise measurement of the vaginal remnant, which is particularly important when vaginoplasty is considered, and facilitates the detection of associated anomalies [4]. Renal and skeletal malformations should also be systematically assessed.

MRKH syndrome carries a significant psychological burden, emphasizing the need for comprehensive multidisciplinary care.

4. Conclusion

In cases of primary amenorrhea in young women with well-developed secondary sexual characteristics, Mayer–Rokitansky–Küster–Hauser syndrome should be considered. The diagnosis is mainly based on imaging, particularly pelvic ultrasound and MRI. MRI remains the investigation of choice, as T2-weighted sequences allow confirmation of uterovaginal aplasia, assessment of ovarian integrity, and detection of associated anomalies, especially renal malformations.

Nevertheless, potential confusion with other syndromes involving uterovaginal anomalies necessitates thorough knowledge of differential diagnoses.

Compliance with ethical standards

Disclosure of conflict of interest

No conflict-of-interest to be disclosed.

Statement of informed consent

Informed consent was obtained from all individual participants included in the study.

References

[1] Sarto GE. Cytogénétique de cinquante patientes atteintes d'aménorrhée primaire. Am J Obstet Gynecol. 1974;119(1):14–23. doi : 10.1016/0002-9378(74)90312-3.

[2] Strübbe EH, Willemsen WN, Lemmens JA, Thijn CJ, Rolland R. Mayer-Rokitansky-Küster-Hauser syndrome: distinction between two forms based on excretory urographic, sonographic, and laparoscopic findings. Am J Roentgenol. 1993;160(2):331-4.

[3] Carrington BM, Hricak H, Nuruddin RN, Secaf E, Laros RK Jr, Hill EC. Müllerian duct anomalies: MR imaginga evaluation. Radiology. 1990;176(3):715-20. PubMed | Google Scholar

[4] Selvaggi G1, Monstrey S, Depypere H, Blondeel P, Van Landuyt K, Hamdi M et al. Creation of a neovagina with use of a pudendal thigh fasciocutaneous flap and restoration of uterovaginal continuity. Fertil Steril. 2003;80(3):607-11

[5] Timmreck LS, Gray MR, Handelin B, Allito B, Rohlfs E, Davis AJ, et al. Analyse des mutations du gène CFTR (régulateur de la conductance transmembranaire de la mucoviscidose) chez des patients présentant une absence congénitale d'utérus et de vagin. Am J Med Genet A. 2003;120(1):72–6. doi: 10.1002/ajmg.a.20197.