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Mayer-Rokitansky-Küster-Hauser Syndrome: A Comprehensive Case Report and Review of Clinical, Genetic, and Psychosocial Aspects

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Abstract

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a congenital condition marked by the absence or underdevelopment of the uterus and upper vagina in women with normal karyotype (46, XX) and secondary sexual characteristics. This article presents a case of a young patient diagnosed with MRKH syndrome, detailing clinical presentation, diagnostic approach, and therapeutic interventions. An extensive review of the literature provides insights into genetic factors, phenotypic variability, and the psychosocial challenges faced by affected individuals. Understanding the syndrome's genetic underpinnings and its impact on patients' quality of life is crucial to enhancing management and patient counseling.

Keywords

Rokitansky Syndrome, Müllerian Agenesis, Primary Amenorrhea

1. Introduction

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome, also referred to as Müllerian aplasia, affects approximately 1 in 4500 female births and is the second most common cause of primary amenorrhea [1] [2]. Characterized by the congenital absence or underdevelopment of the uterus and upper vagina, MRKH syndrome can present in isolation (Type I) or with associated extragenital malformations, such as renal and skeletal anomalies (Type II) [2] [3]. Patients typically present during

adolescence, usually due to primary amenorrhea, and are otherwise hormonally and phenotypically female [2].

2. Case Description

Y.K.S.O., an 18-year-old single female student, was referred to the radiology department to investigate primary amenorrhea. The patient had never experienced menarche, despite normal secondary sexual development—including breast and pubic hair growth—beginning around age 13. Her history revealed mild, cyclic pelvic pain starting six months prior to presentation. Additionally, she reported a recent unsuccessful attempt at sexual intercourse due to intense pain and mechanical obstruction, leading to significant distress and social withdrawal.

During the clinical interview, psychological distress was evident. The patient exhibited emotional lability and reported feelings of sadness, lethargy, and inadequacy. Her distress stemmed primarily from the inability to have sexual intercourse and concerns regarding future fertility and her diagnosis.

Physical examination revealed Tanner stage V development, indicating preserved hormonal maturation. External genitalia appeared unremarkable; however, inspection and speculum examination of the vaginal introitus demonstrated a significantly shortened vaginal canal, approximately 3 cm in depth, ending in a blind pouch.

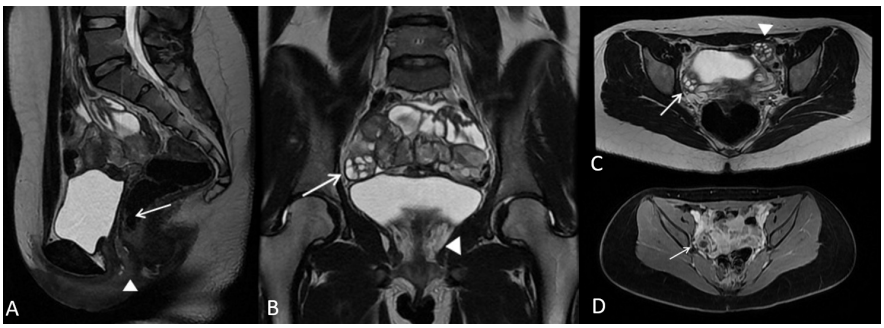

Figure 1. MRI findings. Sagittal T2-weighted sequence (A) demonstrating uterine (arrow) and proximal vaginal (arrowhead) agenesis. Coronal T2-weighted pre-contrast sequence (B) demonstrating the right ovary (arrow) and uterine agenesis (arrowhead). Axial T2-weighted pre-contrast sequence (C) demonstrating the right ovary (arrow) and left ovary (arrowhead) with follicles. Axial T1-weighted post-contrast sequence (D) demonstrating the right ovary with a follicle (arrow) and the absence of T1 hyperintense foci suggestive of hematometra.

The diagnostic workup included laboratory tests, pelvic ultrasound, and pelvic MRI. Among the laboratory tests, the karyotype test was performed and showed a result of 46, XX. MRI confirmed the absence of the uterus, cervix, and upper vagina, with fibrous tissue replacing these structures in their usual anatomical location (see **Figure 1(A)** and **Figure 1(B)**). Bilateral ovaries were visualized in normal positions, with follicles and a corpus luteum on the right, confirming intact ovarian function (see **Figure 1(C)**). Given the history of cyclic pain, a rudimentary uterus with functional endometrium (hematometra) was considered; however,

T1-weighted sequence demonstrated no hyperintense foci, excluding this diagnosis (see **Figure 1(D)**). Therefore, the hypothesis that best explains the cyclical pelvic pain is that of ovulatory pain, since there is a corpus luteum present in the right ovary.

Based on the findings of uterine agenesis, upper vaginal aplasia, and preserved ovarian function, a diagnosis of Mayer-Rokitansky-Küster-Hauser Syndrome Type I was established. Management involved a multidisciplinary approach addressing both surgical and psychological needs. The patient was referred for surgical evaluation regarding possible neovaginoplasty, an alternative to be considered if non-surgical vaginal dilation proves unsuccessful. Concurrent psychological counseling was initiated to assist with coping mechanisms regarding infertility and body image issues.

3. Discussion

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a complex congenital disorder characterized by a range of genetic and phenotypic presentations. Recent studies have significantly advanced the understanding of MRKH's genetic complexity, identifying various candidate genes associated with disrupted Müllerian duct development. Common genetic disruptions occur in PAX8, BMP4, BMP7, and HNF1B, all of which play critical roles in the formation and differentiation of reproductive structures [4] [5]. Additionally, chromosomal variations, such as deletions at 17q12 affecting LHX1, are often observed and are essential for normal uterine development [2] [6]. This variability in genetic presentation contributes to the phenotypic diversity seen across patient cohorts, highlighting MRKH as a heterogeneous syndrome with genetic variability at its core [7].

The clinical manifestations of MRKH syndrome are equally varied, with significant phenotypic heterogeneity. MRKH is typically divided into two types: Type I, which involves isolated uterovaginal agenesis, and Type II, which is associated with extragenital malformations. Type II cases commonly include renal anomalies, such as unilateral renal agenesis, and skeletal anomalies, including scoliosis and fused vertebrae [2] [3]. Radiologic imaging, especially MRI, has been invaluable in the diagnostic assessment of MRKH. MRI allows for detailed visualization of uterine remnants, which, in most patients, appear as bilateral uterine rudiments often connected by a fibrous band. In some cases, rudimentary structures may also present with hematometra or hematosalpinx, requiring further intervention. The current case underscores the diagnostic importance of imaging studies, as they help identify or exclude associated anomalies, thus guiding individualized treatment approaches [8].

The psychosocial challenges associated with MRKH syndrome are profound and should not be underestimated. The diagnosis often has a significant impact on mental health and quality of life, with women frequently experiencing lowered self-esteem, body image issues, and difficulties in intimate relationships. The inability to conceive naturally can also result in feelings of inadequacy and emotional

distress, particularly as patients may compare themselves with peers who do not face fertility challenges. Given these factors, psychological support is essential in MRKH management. Counseling focused on self-worth, intimacy, and coping with infertility can be beneficial, as can support groups that provide a space for sharing experiences and strategies for managing the condition [1] [2].

Management of MRKH syndrome primarily focuses on enabling sexual function and addressing reproductive goals. Non-surgical approaches, such as vaginal dilation, are generally the first line of treatment for sexual function. In cases where dilation is insufficient, surgical options like the McIndoe procedure or the Vecchietti technique may be considered. For patients desiring biological children, uterus transplantation has emerged as a promising, though complex, option, offering the possibility of genetic motherhood. However, this intervention is accessible only in specialized centers due to associated ethical and medical considerations. Gestational surrogacy remains a more widely accessible reproductive option, though legal restrictions may limit its availability in some regions [2] [6].

As research progresses, the understanding of genetic and epigenetic factors involved in MRKH syndrome is likely to improve further. Genome-wide association studies and animal models continue to reveal the molecular mechanisms underlying Müllerian duct anomalies, providing potential pathways for future therapeutic interventions. These insights may allow for earlier diagnosis, better prognostication, and tailored genetic counseling for patients and their families, thereby enhancing clinical outcomes and personalizing patient care [4] [5].

4. Conclusion

The case of this 18-year-old patient highlights the complex nature of Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome. The diagnosis of isolated uterovaginal agenesis (Type I), based on MRI and karyotype findings, confirms the importance of these diagnostic tools for accurate phenotypic differentiation. Beyond the anatomical defect, the emotional distress exhibited by the patient illustrates the psychosocial burden of the condition, directly impacting her body image and reproductive plans. While neovaginoplasty offers a mechanical solution for sexual function, this case demonstrates that surgical intervention alone is insufficient. Psychological support must be an integral component of clinical management. Therefore, a multidisciplinary approach is essential to address both physical and emotional needs, ultimately supporting the quality of life for these women.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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