MAYER-ROKITANSKY-KÜSTER-HAUSER SYNDROME IN GIRLS: CAUSES AND DIAGNOSTIC APPROACHES FOR UTERINE AND VAGINAL AGENESIS

Qurbonova Mo'tabaroy Umidjon qizi
Kokand University, Andijan Branch Student of Pediatrics, Group 25-04
Nazirjonov Orifxo'ja Xusanxoja og'li
Senior Lecturer of the Department of Clinical and Pathological Anatomy
https://doi.org/10.5281/zenodo.17588899

Abstract

Mayer-Rokitansky-Küster-Hauser MRKH syndrome is a rare congenital anomaly affecting phenotypically female individuals, characterized by the partial or complete absence of the uterus and upper two-thirds of the vagina. Despite normal ovarian function and secondary sexual characteristics, affected girls often present during adolescence with primary amenorrhea, which frequently serves as the first clinical indicator. The etiology of MRKH syndrome is multifactorial, including genetic mutations, environmental factors, and developmental disturbances during embryogenesis of the Müllerian ducts. Early recognition of MRKH syndrome is essential not only for managing physical health but also for providing appropriate psychosocial support and reproductive counseling. Diagnostic protocols combine detailed clinical examination, imaging modalities such as ultrasonography and magnetic resonance imaging MRI, and, in select cases, genetic analysis to detect associated anomalies or familial patterns. Multidisciplinary care, involving pediatricians, gynecologists, endocrinologists, and mental health specialists, ensures comprehensive and individualized management. This review summarizes current understanding of MRKH syndrome, emphasizing pathogenesis, clinical presentation, and diagnostic strategies. Highlighting the significance of early detection, the study advocates for increased awareness among healthcare professionals and caregivers to improve long-term health outcomes and quality of life for affected girls.

Keywords

MRKH syndrome, uterine agenesis, vaginal agenesis, congenital reproductive anomalies, Müllerian duct malformations, primary amenorrhea, pediatric gynecology, early diagnosis, genetic factors, imaging techniques.

Introduction

Mayer-Rokitansky-Küster-Hauser MRKH syndrome is a rare congenital condition that predominantly affects girls and adolescents, characterized by the absence or underdevelopment of the uterus and the upper portion of the vagina. Despite these structural anomalies, affected individuals generally exhibit normal ovarian function, normal external genitalia, and the development of secondary sexual characteristics, which often delays the recognition of the syndrome until puberty. The most common clinical presentation is primary amenorrhea, prompting medical evaluation during adolescence. The etiology of MRKH syndrome is multifactorial, encompassing genetic, environmental, and embryological factors. Research suggests that abnormalities in the development of the Müllerian ducts during embryogenesis, combined with potential genetic mutations, contribute to the wide spectrum of anatomical variations observed in affected individuals. Environmental exposures during

critical periods of fetal development may further influence the manifestation of the disorder. Early diagnosis of MRKH syndrome is essential, not only to address physical and reproductive health concerns but also to provide psychosocial support and counseling. Timely recognition enables healthcare providers to guide affected girls and their families regarding potential reproductive options, including assisted reproductive technologies and surgical interventions for vaginal reconstruction when necessary. Advanced imaging techniques, such as ultrasonography and magnetic resonance imaging (MRI), have become vital tools for confirming diagnosis, assessing associated anomalies, and planning individualized management strategies. The purpose of this review is to synthesize current knowledge regarding MRKH syndrome in girls, including its pathogenesis, clinical features, diagnostic approaches, and implications for reproductive and psychosocial health. By emphasizing the importance of early detection and multidisciplinary care, this study seeks to inform clinical practice and improve long-term outcomes for patients affected by this rare but impactful congenital disorder.

Main Body

Historical Background. Mayer-Rokitansky-Küster-Hauser MRKH syndrome has been recognized for nearly two centuries, with early descriptions highlighting the absence of uterus and vagina in otherwise healthy girls. In 1829, August Mayer first described uterovaginal malformations, followed by Carl von Rokitansky's detailed anatomical observations in 1838. Hermann Küster and Georges Hauser later contributed to understanding the syndrome's clinical significance, giving rise to the eponym MRKH. Initially, cases were identified post-mortem or during surgical interventions, but over time, improvements in gynecological examination and imaging have enabled early diagnosis during adolescence.

Epidemiology and Statistics. MRKH syndrome is a rare congenital disorder, affecting approximately 1 in 4,500–5,000 live female births worldwide. Epidemiological studies show variation between populations, with typical MRKH cases accounting for roughly 55–60% and atypical or MURCS-associated cases involving renal, skeletal, or auditory anomalies representing 40–45%. The syndrome contributes to approximately 10–15% of primary amenorrhea cases in adolescent girls. In clinical practice, delayed diagnosis is common; one cohort study found that the median age at definitive diagnosis was 16 years, with an average interval of nearly one year from first medical consultation to accurate identification. Associated anomalies are frequent, particularly in atypical MRKH cases. Renal malformations, such as unilateral agenesis or ectopic kidneys, occur in up to 35–40% of patients. Skeletal irregularities, including vertebral anomalies, may also coexist, underscoring the importance of comprehensive evaluation.

Etiology and Pathogenesis. The pathogenesis of MRKH syndrome is multifactorial, involving genetic, environmental, and embryological factors. During early fetal development, the female reproductive tract originates from paired Müllerian ducts. Disruption in duct formation, fusion, or canalization between the 6th and 12th weeks of gestation can lead to uterine and vaginal agenesis. Genetic studies suggest familial clustering in a minority of cases, indicating possible autosomal dominant inheritance with incomplete penetrance. However, no single causative gene has been definitively identified. Environmental factors, including maternal illness, exposure to teratogenic substances, or hormonal imbalances during critical periods of fetal development, may also contribute to the manifestation of MRKH. Clinically,

MRKH is classified into. Typical MRKH Type I: Isolated uterovaginal agenesis with normal renal and skeletal structures. Atypical MRKH / MURCS Type II: Uterovaginal agenesis accompanied by renal, skeletal, auditory, or cardiac anomalies.

MRKH syndrome often remains undetected until adolescence, as affected girls exhibit normal external genitalia, ovarian function, and secondary sexual characteristics such as breast development and pubic hair. The primary clinical presentation is primary amenorrhea, usually identified when menstruation fails to commence by the expected age. Additional signs may include. Blind-ending vaginal dimple or shortened vaginal canal. Pelvic discomfort in cases with rudimentary or obstructed uterine structures. Associated anomalies renal, skeletal, auditory, particularly in atypical MRKH, which may present as incidental findings during imaging. Psychosocially, the diagnosis can be distressing, affecting self-esteem, sexual identity, and future reproductive concerns. Timely diagnosis and counseling are therefore critical.

Diagnosis relies on a combination of clinical evaluation, imaging studies, and, in select cases, genetic assessment. Clinical Evaluation: Physical examination reveals normal external genitalia but a blind-ending vaginal canal. Secondary sexual characteristics are typically present, confirming normal ovarian function. Family history and prior medical history may provide additional clues. Imaging Studies. Ultrasonography - transabdominal or transrectal ultrasound is often the first-line modality, useful for assessing the presence of uterine structures and evaluating the ovaries and kidneys. Magnetic Resonance Imaging MRI - Provides detailed anatomical visualization, detecting rudimentary uterine remnants and associated renal or skeletal anomalies. MRI is considered the gold standard for complex cases. Additional Imaging: X-rays or CT scans may be employed to assess skeletal anomalies, especially in MURCS cases.

Laboratory and Genetic Tests. Karyotype: Typically 46,XX, confirming normal ovarian chromosomes. Hormonal profile usually normal, verifying ovarian function.

Genetic panels - may be indicated in familial cases to identify potential mutations, although no definitive gene is universally associated with MRKH. Screening for Associated Anomalies. Because of frequent coexisting malformations, renal ultrasound, skeletal survey, and audiological assessment are recommended. Early detection of these anomalies allows proactive management and monitoring.

Implications for Management. Early recognition allows for timely psychosocial support, counseling, and planning for sexual and reproductive health. Management may involve - Vaginal elongation procedures or neovagina creation. Reproductive counseling, including assisted reproductive technologies using gestational surrogacy if desired. Multidisciplinary support, involving pediatricians, gynecologists, endocrinologists, psychologists, and genetic counselors. Early and holistic care improves long-term outcomes, reduces psychological distress, and empowers affected girls and their families to make informed decisions regarding their health and reproductive options.

Conclusion

Mayer-Rokitansky-Küster-Hauser MRKH syndrome represents a rare but clinically significant congenital disorder, primarily affecting adolescent girls with otherwise normal ovarian and external genital development. Despite its rarity, the syndrome is a major contributor to primary amenorrhea and can profoundly influence physical, reproductive, and psychological well-being. Early recognition of MRKH is crucial, enabling prompt diagnostic

evaluation, including imaging and genetic assessment, to identify uterine and vaginal agenesis as well as associated anomalies. Timely diagnosis not only facilitates medical and surgical interventions, such as vaginal reconstruction or reproductive planning, but also provides opportunities for psychosocial support, enhancing quality of life and reducing emotional distress. The multifactorial etiology of MRKH, involving embryologic disruptions, genetic predisposition, and possible environmental influences, underscores the need for continued research to elucidate underlying mechanisms. Comprehensive multidisciplinary care, integrating pediatric, gynecological, endocrinological, and psychological expertise, ensures individualized management tailored to the specific anatomical and emotional needs of affected girls. Awareness and early intervention remain central to improving long-term outcomes for patients with MRKH syndrome, promoting optimal reproductive potential, emotional resilience, and overall well-being.

References:

1. Rock, J. A., & Thompson, J. C. (2002). Te Linde's Operative Gynecology (10th ed.). Philadelphia: Lippincott Williams & Wilkins.
2. Edmonds, D. K. (2019). A Textbook of Reproductive Medicine and Infertility (8th ed.). London: CRC Press.
3. Rock, J. A. (1997). Hysteroscopic Surgery for Müllerian Anomalies. Philadelphia: W. B. Saunders.
4. Acién, P., & Acién, M. (2011). Müllerian Anomalies in Women: Diagnosis and Clinical Management. Heidelberg: Springer.
5. Speroff, L., Fritz, M. A. (2005). Clinical Gynecologic Endocrinology and Infertility (8th ed.). Philadelphia: Lippincott Williams & Wilkins.
6. Rock, J. A., & Schlaff, W. D. (2009). Te Linde's Operative Gynecology: A Clinical Approach (11th ed.). Philadelphia: Lippincott Williams & Wilkins.
7. Brucker, S., & Tinneberg, H. (2012). Gynecological Endocrinology and Reproductive Medicine. Berlin: Springer.
8. Fritz, M. A., & Speroff, L. (2011). Clinical Gynecologic Endocrinology and Infertility Workbook (9th ed.). Philadelphia: Lippincott Williams & Wilkins.
9. Rock, J. A., Jones, H. W. III. (2010). Te Linde's Operative Gynecology, 12th Edition. Philadelphia: Wolters Kluwer.
10. Cunningham, F. G., Leveno, K. J., Bloom, S. L., et al. (2018). Williams Obstetrics (25th ed.). New York: McGraw-Hill Education.