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Case Report

Comprehensive management of Mayer-Rokitansky-Küster-Hauser syndrome management: A case report

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Abstract

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome, also known as Müllerian aplasia, is a congenital condition characterized by uterine and upper vaginal aplasia. It affects females with a normal female karyotype and typical secondary sex characteristics. The aim of this case report was to highlight the multidisciplinary management approach for MRKH syndrome, focusing on tailored interventions to address physical and psychological challenges and improve reproductive prospects. A 26-year-old married female presented to Acharya Vinoba Bhave Rural Hospital (AVBRH), Sawangi, Wardha, India, in January 2023 with primary amenorrhea. Physical examination revealed a blind vagina and a hypoplastic uterus, indicative of MRKH syndrome. Further evaluation, including pelvic magnetic resonance imaging (MRI), confirmed Müllerian duct abnormalities and bilateral ovarian anomalies. The absence of a functional vagina significantly impacted the patient's quality of life, leading to difficulties with sexual intercourse and emotional distress related to infertility. A collaborative approach involving a gynecologist and a psychiatrist at AVBRH was initiated to address these challenges. The patient underwent vaginoplasty to create a neovagina, enhancing the sexual function and intimate relationship with the spouse. However, due to the hypoplastic uterus, achieving motherhood through traditional means was not possible. Therefore, assisted reproductive techniques, in particular surrogacy, were explored. Normal, functional ovaries were harvested from the patient for use in surrogacy procedures. This comprehensive management strategy exemplifies the challenges associated with MRKH syndrome and underscores the importance of tailored interventions and long-term follow-up. The case highlights the significance of collaborative care in improving the quality of life and reproductive prospects for individuals with MRKH syndrome.

Keywords: Müllerian aplasia, MRKH syndrome, vaginoplasty, surrogacy, assisted reproductive techniques

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Introduction

Müllerian aplasia, commonly known as Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome, is a congenital condition that affects females with normal female karyotype and typical secondary sex characteristics. It is characterized by uterine and upper (2/3) vaginal aplasia (46, XX) [1]. The diagnosis, which has an estimated prevalence of 1 in 5000 live female births, is frequently made during adolescence after tests for primary amenorrhea [2]. Primary amenorrhea, defined as the

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absence of menstruation by age 16 without the presence of secondary sexual characteristics or by age 14 with secondary sexual characteristics, necessitates a comprehensive diagnostic approach [3].

A thorough evaluation typically begins with a physical examination to assess secondary sexual characteristics such as breast development and pubic hair growth [4]. Subsequently, hormonal tests are conducted to measure the levels of key hormones, including follicle-stimulating hormone (FSH), luteinizing hormone (LH), estradiol, prolactin, thyroid-stimulating hormone (TSH), and testosterone. Karyotype analysis, examining chromosomal patterns, helps in detecting conditions such as Turner syndrome, a common cause of primary amenorrhea [5,6]. Imaging techniques such as pelvic ultrasound, magnetic resonance imaging (MRI), or CT scans may be employed to evaluate the internal reproductive organs for structural abnormalities [7]. Additionally, gonadotropin-releasing hormone (GnRH) stimulation tests assess pituitary and hypothalamic function, aiding in distinguishing between different causes of hypothalamic-pituitary dysfunction and ovarian failure. Thyroid function tests and bone density assessments may also be included in the diagnostic process to rule out thyroid disorders and evaluate bone health, respectively. Genetic testing may be pursued in cases where specific genetic conditions are suspected [7,8]. These comprehensive evaluations enable clinicians to identify the underlying cause of primary amenorrhea and guide appropriate treatment strategies tailored to the individual's needs [9].

Although MRKH can occur in isolation (type I), it is more commonly linked to cardiac, renal, and spinal abnormalities (MRKH type II) [10]. Treatment for vaginal aplasia can be provided to enable sexual activity by creating a neovagina [11]. Vaginoplasty is a surgical technique aimed at creating a neovagina, enabling the patient to engage in sexual intercourse and improving overall quality of life [12,13]. The vaginoplasty involves surgical intervention, including the laparoscopic Davydov technique, which utilizes peritoneal flaps to reconstruct the vaginal canal [14,15]. During this technique, two peritoneal flaps are created within the abdominal cavity, then these flaps are then carefully brought together and sutured to form a neovagina, mimicking the anatomical structure of a natural vaginal canal [13,15]. The procedure allows for precise reconstruction while offering advantages such as reduced postoperative pain and shorter recovery times compared to traditional methods [11]. MRKH has been linked to several genetic disorders; however, because the genetic pathways involved in the embryogenetic development of the Müllerian ducts are complicated, precise etiology remains unknown [8,9,16]. The aim of this case study was to highlight the multidisciplinary management approach for MRKH syndrome, focusing on tailored interventions to address physical and psychological challenges and improve reproductive prospects.

Case

A 26-year-old married female presented to Acharya Vinoba Bhave Rural Hospital (AVBRH), Sawangi, Wardha, India, in January 2023 with a history of primary amenorrhea. The patient had never experienced menstruation, indicative of primary amenorrhea. The absence of a functional vagina significantly impacted the patient's quality of life, resulting in difficulties with sexual intercourse and psychological distress related to the inability to conceive.

Evaluation by the gynecologist revealed the presence of a blind vagina with no discernible vaginal orifice. Further physical examinations were conducted to assess secondary sexual characteristics and signs of androgen excess. This examination included Tanner staging for breast development and pubic hair and the evaluation of hirsutism using the Ferriman-gallery score. Tanner stage II (Tanner staging for breast development) indicated that the breast buds were present, and the elevation of the breast and papilla was a small mound. Tanner stage IV (Tanner staging for pubic hair development) indicated that the hair was adult-like, covering a larger area but not extending to the thighs. Hirsutism evaluation using Ferriman-gallery score 1 indicated that the terminal hair was present but less than an average male.

Given the potential association of MRKH syndrome with cardiac anomalies, a cardiologist was consulted to screen for and manage any cardiac abnormalities that may be present, ensuring appropriate intervention or ongoing monitoring. An otolaryngologist was involved in evaluating and managing any anomalies in the ear, nose, and throat region, including hearing loss or

structural abnormalities of the ear canal, which may be associated with MRKH syndrome. Evaluation of the urinary system was conducted by a urologist to rule out associated anomalies and address any urological concerns, ensuring a comprehensive assessment of urinary tract anatomy and function.

Subsequent evaluation through pelvic MRI confirmed the presence of a hypoplastic uterus, bilateral ovarian anomalies, and Müllerian duct abnormalities, notably including a malpositioned right ovary. MRI confirmed a hypoplastic uterus, demonstrating decreased size and underdeveloped myometrium.

A karyotype analysis was performed to accurately diagnose MRKH syndrome and differentiate it from other conditions, such as androgen insensitivity syndrome (AIS) or gonadal dysgenesis. The karyotype analysis of the patient in this case report revealed a 46, XX karyotype.

A collaborative approach involving a gynecologist and a psychiatrist at AVBRH was initiated to address these challenges, providing comprehensive counseling and emotional support to alleviate the patient's distress. In pursuit of restoring sexual function, the patient underwent vaginoplasty, a surgical procedure aimed at creating a neovagina.

Through collaborative efforts and surgical intervention, significant improvements were achieved in the patient's quality of life and reproductive prospects. The patient underwent vaginoplasty to address the blind vagina, thereby enhancing her quality of life and facilitating sexual function. The pre-surgical preparation and intervention plan for vaginoplasty is described in **Table 1**.

Table 1. Pre-surgical preparation and intervention plan for vaginoplasty

Stage	Description
Preoperative assessment	
Comprehensive medical history	Menstrual history Sexual function psychological evaluation
Pelvic examination	Assess vaginal anatomy and dimensions
Imaging studies (MRI pelvis)	Evaluate pelvic structures and anomalies
Hormonal assessment (FSH, LH, and estradiol)	Evaluate ovarian function (FSH=10 IU/L) (LH=05 IU/L) (estradiol=200 pg/mL)
Counselling	Surgical expectations, risks, and postoperative care
Intervention plan	
Multidisciplinary team	Gynecologist, urologist, psychologist
Surgical technique selection	Vaginoplasty is minimally invasive
Preoperative antibiotics	Reduce the risk of infection
Intraoperative monitoring	Vital signs and anesthesia depth
Vaginoplasty procedure	Meticulous tissue handling and hemostasis
Vaginal moulds/stents	Desired vaginal dimensions
Postoperative care	
Pain management	Ensure patient comfort
Psychological support and counselling	Throughout recovery
Follow up appointments	Monitor healing, function, and satisfaction
Long-term outcomes evaluation	Sexual function and quality of life
Wound care and hygiene	Prevent infection
Prophylactic anticoagulation	Reduce thromboembolism risk
Gradual activity resumption	Guided by surgeon recommendations
Postoperative complication management	Vaginal stenosis and hematoma
Continued psychological support	As needed
Long term surveillance	Potential complications and further interventions

FSH: follicle-stimulating hormone; LH: luteinizing hormone; MRI: magnetic resonance imaging

During the vaginoplasty, Davydov surgical technique was employed to reconstruct the vagina and address associated Müllerian duct abnormalities. The procedure involved mobilization and creating a neovaginal canal using peritoneal flaps derived from the patient's peritoneum. The peritoneum was dissected and carefully fashioned into a tubular structure, then positioned as the neovagina. This technique allowed the creation of a functional vaginal canal while minimizing the risk of complications, such as in the case of vaginal stenosis. After neovagina creation, the vaginal canal was closed, and vaginal cake molds were inserted to maintain desired dimensions. Hemostasis was ensured to minimize bleeding, and the incision site was closed in layers. Finally, a sterile dressing was applied to the surgical site. This comprehensive approach to vaginoplasty

was aimed at addressing vaginal aplasia in MRKH syndrome, improving sexual function and quality of life. The laparoscopic Davydov procedure was successful in creating a neovagina, enabling the patient to engage in sexual intercourse and improving the quality of life.

Postoperatively, the patient underwent a period of recovery and postoperative care to ensure optimal healing and adaptation to the newly reconstructed vagina. This care involved the use of vaginal dilators to maintain vaginal depth and width, as well as supportive psychotherapy addressing the challenges related to sexual function and body image. It involved seven psychotherapy sessions, with periodic reassessment to evaluate progress and adjust treatment goals as needed in the case. Following successful surgery, the patient reported the ability to engage in sexual intercourse, thereby enhancing the intimate relationship with the spouse.

Given the patient's desire for motherhood despite the presence of a hypoplastic uterus, assisted reproductive techniques were explored. Normal, functional ovaries were harvested from the patient and subsequently utilized in surrogacy procedures, offering the prospect of biological childbearing. The timeline of key milestones and interventions in managing the patient in this case is presented in **Table 2**.

Table 2. Timeline of events in the case of Acharya Vinoba Bhave Rural hospital (AVBRH)

Date	Event
January 2023	Admission to AVBRH
January 2023	Magnetic resonance imaging (MRI) pelvis scan was conducted
February 2023	Vaginoplasty surgery performed
February 2023 to January 2024	Follow-up after surgery
February 2024	One-year follow-up completed

Discussion

MRKH syndrome is a rare congenital condition affecting females with normal female karyotype and typical secondary sex characteristics. Primary amenorrhea is the absence of menstrual periods by the age of 16 in females who have otherwise undergone normal pubertal development [4]. In this case, the patient had never experienced menstruation, which is a hallmark feature of MRKH syndrome [1]. Upon physical examination, it was observed that the patient had a blind vagina, meaning there was no discernible vaginal orifice. This condition is consistent with the vaginal aplasia characteristic of MRKH syndrome [2]. Imaging studies, including pelvic MRI, revealed the presence of a hypoplastic uterus in the patient [9]. A hypoplastic uterus is underdeveloped in size and may have an incomplete formation of the uterine cavity and myometrium [17]. The absence of a functional vagina significantly impacted the patient's quality of life, leading to difficulties with sexual intercourse [18]. This symptom underscores the physical and emotional challenges faced by individuals with MRKH syndrome. The patient experienced psychological distress related to her inability to conceive due to the reproductive anomalies associated with MRKH syndrome [17]. The presented case exemplifies the intricate management required for MRKH syndrome, underscoring the importance of a multidisciplinary approach, focusing on interventions such as vaginoplasty and assisted reproductive techniques. MRKH syndrome poses significant challenges in terms of both physical and psychological well-being for affected individuals. For instance, a study reported few cases of MRKH syndrome, emphasizing the diversity of associated anomalies and the need for tailored treatment approaches [19]. These comparative analyses aid clinicians in tailoring interventions to maximize patient outcomes and satisfaction.

The management plan of MRKH involved collaboration among gynecologist, reproductive specialist, cardiologist, otolaryngologist, urologist, and other medical professionals to address potential associated anomalies and provide comprehensive care tailored to the individual's needs. Managing individuals with MRKH syndrome requires a multidisciplinary approach to address potential associated anomalies and provide comprehensive care [19].

Furthermore, this multidisciplinary approach can aid in differentiating between type I and type II MRKH syndrome, which are distinguished primarily by the presence or absence of renal anomalies. The main difference between type I and type II MRKH syndrome lies in the presence or absence of associated renal anomalies. Type I MRKH syndrome involves isolated Müllerian

duct anomalies without renal involvement, while type II MRKH syndrome includes Müllerian duct anomalies along with associated renal abnormalities [2,20]. It is important to differentiate between the two types as it can impact the management and counseling of affected individuals, particularly regarding potential renal complications and associated health considerations. This case involved Müllerian duct anomalies without renal involvement, which is type I MRKH syndrome.

Vaginoplasty stands as a cornerstone in addressing primary amenorrhea and enhancing sexual function in MRKH syndrome patients. Various approaches, including the McIndoe, Williams, and Vecchietti techniques, offer advantages and limitations regarding Vaginoplasty. For instance, the Davydov procedure, a laparoscopically assisted neo-vaginoplasty technique, has gained attention for its minimally invasive nature [21,22]. Few case reports describe the successful utilization of laparoscopically assisted neo vaginoplasty like the Davydov procedure in patients with MRKH syndrome, highlighting the efficacy of minimally invasive techniques [22]. Furthermore, a study compared outcomes of different vaginoplasty techniques in MRKH syndrome patients, shedding light on the optimal approach based on individual needs and anatomical variations [23].

The utilization of assisted reproductive techniques, such as surrogacy, highlights the comprehensive nature of care required to address the reproductive aspirations of individuals with MRKH syndrome. Few studies discussing advancements in assisted reproductive technologies specific to MRKH syndrome patients shed light on evolving treatment options and success rates [17,24]. The discussion of MRKH syndrome necessitates consideration of the psychosocial implications and long-term outcomes. Understanding the psychological impact, coping mechanisms, and the role of support groups and counseling services is crucial for holistic patient care. Recent studies have demonstrated the profound psychological impact of MRKH syndrome on individuals, underscoring the importance of psychological support and counseling throughout the treatment journey [25].

Lastly, ensuring comprehensive and diligent long-term follow-up is paramount in managing MRKH syndrome. It not only facilitates the monitoring of surgical outcomes, sexual function, and reproductive outcomes but also plays a crucial role in providing ongoing support and care for individuals affected by this condition [25]. By prioritizing consistent follow-up, healthcare providers can better address evolving needs, optimize treatment strategies, and improve overall well-being and quality of life for those living with MRKH syndrome.

Conclusion

The presented case of a 26-year-old married female with MRKH syndrome underscored its management's complexity and multidisciplinary nature. Through comprehensive assessment and intervention, including vaginoplasty and assisted reproduction via surrogacy, significant improvements were achieved in the patient's quality of life and reproductive prospects. This case highlights the importance of tailored treatment approaches and comprehensive care to address both the physical and psychosocial aspects of MRKH syndrome. Moreover, using minimally invasive techniques exemplifies the evolving landscape of surgical interventions, leading to improved outcomes and patient satisfaction. Continuing research and collaborative efforts are essential to further enhance our understanding of MRKH syndrome and optimize therapeutic strategies, ensuring holistic and effective care for affected individuals.

Ethics approval

The patient provided the written informed consent to be included as a case report.

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Competing interests

All the authors declare that there are no conflicts of interest.

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Underlying data

Derived data supporting the findings of this study are available as part of the article.

How to cite

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