Case Report

Mayer–Rokitansky–Küster–Hauser Syndrome: A Rare Cause of Primary Amenorrhea

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Abstract

Mayer–Rokitansky–Küster–Hauser (MRKH) syndrome is a highly unusual congenital anomaly characterised by agenesis of the uterus and upper two-thirds of the vagina in individuals with normal secondary sexual characteristics and a normal 46XX karyotype. It commonly presents as primary amenorrhea during adolescence. We report a case of MRKH syndrome in a 14-year-old girl who presented with primary amenorrhea. Imaging revealed a rudimentary, non-communicating uterus with normal ovaries. Comprehensive counseling was done regarding the diagnosis and it's psychological implications, infertility issues, and available surgical options. Early diagnosis, appropriate imaging, and comprehensive psychological support are cornerstone of management in adolescents with MRKH syndrome.

Keywords: Mayer–Rokitansky–Küster–Hauser syndrome, MRKH, primary amenorrhea, vaginal agenesis, counselling.

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Introduction

Mayer–Rokitansky–Küster–Hauser (MRKH) syndrome is a very unusual congenital malformation of the female reproductive tract. Estimated incidence is around 1 in 4,500–5,000 female births. This condition is distinguished by congenital agenesis/hypoplasia of the uterus and upper two-thirds of the vagina, with 46xx karyotype, normal functional ovaries, external genitalia and appropriate secondary sexual characteristics[1-3]. MRKH syndrome is classified into type I (typical or isolated uterovaginal agenesis) and type II (atypical), which is associated with other congenital anomalies [3,4] . Majority are sporadic, with occasional studies indicate genetic component as well, but exact aetiology is still not clear.[3,5] This diagnosis is usually associated with considerable psychological distress, in view of amenorrhea, sexuality, and infertility, making the requirement to diagnose it as early as possible and provide appropriate counselling.[6,7]

Case Presentation

A 14-year-old girl came to pediatric OPD with complaints of amenorrhea. She also reported episodic nausea and vomiting, which were relieved with intravenous medication. There was no history of cyclic abdominal pain. Physical examination revealed normal female external genitalia. Sexual maturity rating (SMR) staging showed age-appropriate pubertal development and normal secondary sexual characteristics.

USG of the abdomen and pelvis showed a hyper-echoic collection within the endometrial cavity suggestive of hematometra.[2] Magnetic resonance imaging (MRI) demonstrated a rudimentary, non-communicating uterus with normal bilateral ovaries and fallopian tubes, consistent with MRKH syndrome.[1,8] Karyotype analysis confirmed a normal female chromosomal pattern (46XX).[3]

Investigations

Investigation	Findings
Ultrasonography (abdomen & pelvis)	Hyper-echoic collection in endometrial cavity (hematometra) [2]
MRI abdomen & pelvis	Rudimentary non-communicating uterus; normal ovaries and tubes [1,8]
Karyotype	46XX [3]
SMR staging	Normal pubertal development without menarche

Management and Outcome

Extensive counseling were done regarding the characteristics of the condition, prognosis, fertility, and treatment options.[4,9] Psychological counselling was emphasised to address anxiety and emotional distress associated with the diagnosis. Surgical therapy (vaginoplasty) was likewise carefully explained.[6] The patient was referred to a tertiary care centre, where she was underwent vaginoplasty surgery. It was recommended to continue psychological support to help in coping with body

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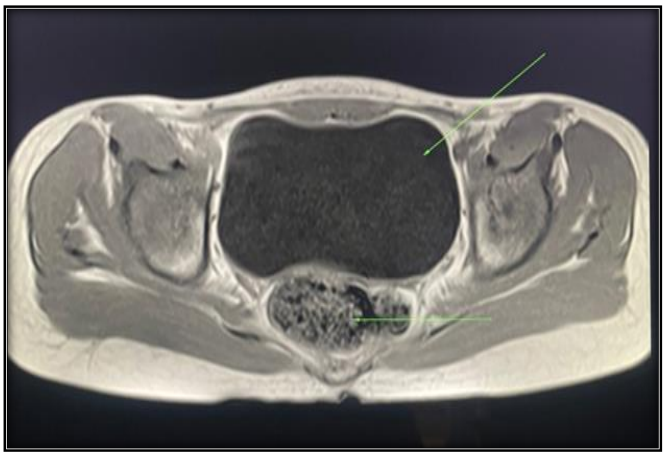
image issues and subsequent reproductive decision-making.



Discussion

MRKH syndrome should be suspected on the list of diagnoses in adolescents with primary amenorrhea and normal secondary sexual developent.[1-3] Ultrasonography and magnetic resonance imaging are vital for confirming the diagnosis and detecting related abnormalities, while karyotyping is also used to rule out chromosomal anomalies.[3,5]

The unusual type can be related to defects and abnormalities of the kidneys and bones, but the most common type of MRKH syndrome is with isolated uterovaginal agenesis and normal ovaries; hence, there is a significant psychological framework in the syndrome.[3,4]

In addition to the anatomical abnormalities, the MRKH syndrome is characterised by a high burden on the psychological system. Teenagers have a lot of emotional discomfort connected to body image, femininity, sexual functioning, and infertility.[10]

As such, holistic management is necessary, as a multidisciplinary approach involving paediatricians, gynaecologists, psychologists, and surgeons is required. Non-surgical or surgical methods of creating a neovagina have been proven to have a significant positive impact on the quality of life and sexual functioning in patients with an affected vagina.[7,9]

Despite the reason that a woman with MRKH syndrome cannot bear a baby, assisted reproductive technologies like in vitro fertilisation through gestational surrogacy provide some possibilities of earning biological parenting.[6,7]

Conclusion

MRKH syndrome must be given sincere consideration in girls who present as primary amenorrheic, yet they have reached puberty. It is necessary to diagnose in a timely and accurate manner using ultrasonography, MRI, and karyotyping. It is also crucial to examine the psychological effects of the condition by undertaking prompt counselling for both the family and the patient. They should also be provided with a multidisciplinary approach and be supportive in choosing vaginoplasty, fertility treatments, and the long-term psychosocial well-being.

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Conflicts of interest

There are no conflicts of interest.

References

1. Shimizu H, Kobayashi H, Shimizu Y, Adachi H. Mayer–Rokitansky–Küster–Hauser syndrome: case report and review. BMJ Case Rep. 2023;16(5):e254262.
2. Muthu Kumar A, Menon P, Mane S. Mayer–Rokitansky–Küster–Hauser syndrome: a case report. Cureus. 2024;16(8):e67606.
3. Herlin MK, Petersen MB, Brännström M. Mayer–Rokitansky–Küster–Hauser (MRKH) syndrome: a comprehensive update. Orphanet J Rare Dis.2020;15:214.
4. Soekersi H, Trihadrian R. MRKH syndrome type II with crossed fused renal ectopia: a rare case report. Radiol Case Rep. 2023;18(5):1877–1881.
5. Schiau C, Csutak C, Ciurea AI, et al. Mayer–Rokitansky–Küster–Hauser syndrome: from radiological diagnosis to further challenges. Diagnostics (Basel). 2026;16(1):138.
6. Villa R, Azzollini J, Peissel B, Manoukian S. Genetic and clinical considerations in MRKH syndrome. GynecolOncol Rep.2019;28:68–70.
7. Blontzos N, Iavazzo C, Vorgias G, Kalinoglou N. Mayer–Rokitansky–Küster–Hauser syndrome: diagnosis and management. ObstetGynecol Sci. 2019;62(4):294–297.
8. Ray U, Adhikari S, Dhital R, et al. Mayer–Rokitansky–Küster–Hauser syndrome: a rare case report from Nepal. Ann Med Surg.2022;82:104725.
9. Mungadi IA, Ahmad Y, Yunusa GH, et al. Mayer–Rokitansky–Küster–Hauser syndrome: surgical management of two cases. J Surg Tech Case Rep. 2010;2(1):39–43.
10. Boruah DK, Sanyal S, Gogoi BB, et al. Psychological impact of MRKH syndrome in adolescents. J ClinDiagn Res. 2017;11(7):TC30–TC35.