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Beyond Primary Amenorrhea Delayed Diagnosis of Mayer Rokitansky Küster Hauser Syndrome in a Young Married Woman A case report

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ABSTRACT

Background: Mayer-Rokitansky-Küster-Hauser syndrome is a congenital Müllerian anomaly characterized by uterine and upper vaginal agenesis or severe hypoplasia in individuals with normal secondary sexual development and functional ovaries. Diagnosis usually occurs during adolescence during evaluation for primary amenorrhea. Delayed recognition until marriage may intensify concerns about sexual function, identity, and fertility. Case presentation: A 22-year-old Indonesian woman was referred for primary amenorrhea. She reported normal breast and pubic hair development, no cyclical pelvic pain, and difficulty with sexual function after marriage. Her Female Sexual Function Index score was 20.8. Examination showed Tanner stage V breast development, Tanner stage IV pubic hair, normal external genitalia, and a vaginal pouch approximately 1 cm deep. Transabdominal and transvaginal ultrasonography could not adequately identify the uterus or ovaries and suggested Müllerian agenesis. Diagnostic laparoscopy showed no macroscopically identifiable uterus, while both ovaries and fallopian tubes appeared normal. The clinical and operative findings strongly supported Mayer-Rokitansky-Küster-Hauser syndrome. The patient received counseling regarding the diagnosis, sexual function, psychological support, and reproductive options. Conclusion: Primary amenorrhea accompanied by normal pubertal development should prompt early assessment for Müllerian agenesis. This case emphasizes the consequences of delayed diagnosis and the need to integrate anatomical assessment with psychosexual care and reproductive counseling. Complete diagnostic evaluation, including endocrine testing, karyotyping, and screening for associated anomalies, remains important when these assessments were not completed before operative confirmation.

Keywords: primary amenorrhea; Müllerian agenesis; Mayer-Rokitansky-Küster-Hauser syndrome; vaginal agenesis; sexual function; diagnostic laparoscopy

ABSTRAK

Latar Belakang: Sindrom Mayer–Rokitansky–Küster–Hauser merupakan kelainan kongenital duktus Müllerian yang ditandai oleh agenesis atau hipoplasia berat uterus dan bagian atas vagina pada perempuan dengan perkembangan karakteristik seksual sekunder yang normal serta fungsi ovarium yang tetap baik. Diagnosis umumnya ditegakkan pada masa remaja ketika dilakukan evaluasi terhadap amenorea primer. Keterlambatan diagnosis hingga pasien menikah dapat memperberat permasalahan yang berkaitan dengan fungsi seksual, identitas diri, dan fertilitas. Presentasi Kasus: Seorang perempuan Indonesia berusia 22 tahun dirujuk untuk evaluasi amenorea primer. Pasien melaporkan perkembangan payudara dan rambut pubis yang normal, tidak mengalami nyeri pelvis siklik, serta mengeluhkan kesulitan dalam fungsi seksual setelah menikah. Penilaian menggunakan Female Sexual Function Index menunjukkan skor 20,8. Pemeriksaan fisik memperlihatkan perkembangan payudara Tanner stadium V, pertumbuhan rambut pubis Tanner stadium IV, genitalia eksterna yang tampak normal, serta kantong vagina (vaginal pouch) dengan kedalaman sekitar 1 cm. Pemeriksaan ultrasonografi transabdominal dan transvaginal tidak dapat mengidentifikasi uterus maupun ovarium secara memadai dan mengarah pada dugaan agenesis Müllerian. Laparoskopi diagnostik menunjukkan tidak adanya struktur uterus yang dapat diidentifikasi secara makroskopis, sedangkan kedua ovarium dan tuba falopi tampak normal. Temuan klinis dan intraoperatif tersebut sangat mendukung diagnosis sindrom Mayer–Rokitansky–Küster–Hauser. Pasien selanjutnya memperoleh konseling mengenai diagnosis, fungsi seksual, dukungan psikologis, serta berbagai pilihan reproduksi yang tersedia. Kesimpulan: Amenorea primer yang disertai perkembangan pubertas normal perlu mendorong dilakukannya evaluasi dini terhadap kemungkinan agenesis Müllerian. Kasus ini menegaskan dampak keterlambatan diagnosis serta pentingnya mengintegrasikan evaluasi anatomis dengan pelayanan psikoseksual dan konseling reproduksi. Evaluasi diagnostik yang komprehensif, termasuk pemeriksaan endokrin, analisis kariotipe, dan skrining terhadap kelainan kongenital penyerta, tetap diperlukan apabila pemeriksaan tersebut belum dilakukan sebelum diagnosis dikonfirmasi melalui tindakan operatif.

Kata kunci: amenorea primer; agenesis Müllerian; sindrom Mayer–Rokitansky–Küster–Hauser; agenesis vagina; fungsi seksual; laparoskopi diagnostik

INTRODUCTION

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is characterized by congenital aplasia or

marked hypoplasia of the uterus and upper vagina in phenotypic females with normal ovarian function and secondary sexual development. It affects approximately 1 in 4,000-5,000 females and is the second most frequent cause of primary amenorrhea after gonadal dysgenesis.1,2 MRKH may occur as an isolated genital anomaly (type I) or together with renal, skeletal, auditory, or cardiac abnormalities (type II or the MURCS spectrum).1,3

The diagnosis is commonly established during adolescence when menstruation fails to occur despite otherwise normal pubertal development. Clinical examination and pelvic ultrasonography constitute the initial assessment. Magnetic resonance imaging can define Müllerian remnants, ovaries, and associated urinary tract abnormalities when ultrasonographic findings are uncertain. Karyotyping and appropriate endocrine testing are important for distinguishing MRKH from conditions such as complete androgen insensitivity syndrome.2,4

The clinical impact extends beyond absent menstruation. Patients may experience difficulty with vaginal penetration, altered body image, psychological distress, and concerns about biological parenthood.5,6 Indonesian data on women undergoing surgery for congenital reproductive tract disorders also indicate that anatomical treatment does not uniformly restore sexual function, underscoring the importance of counseling and individualized follow-up.7 We report a young married woman whose MRKH syndrome was recognized only after presentation with primary amenorrhea and sexual concerns. The case highlights diagnostic delay, the role and limitations of diagnostic laparoscopy, and the need for comprehensive psychosexual and reproductive care.

CASE PRESENTATION

A 22-year-old nulligravid Indonesian woman was referred to the gynecology service of Dr Zainoel Abidin Hospital, Banda Aceh, because she had never menstruated. She had experienced normal breast, axillary hair, and pubic hair development. She denied spotting, cyclical lower abdominal pain, galactorrhea, hirsutism, deepening of the voice, hot flushes, night sweats, vaginal dryness, or a marked change in body weight. Urination and defecation were normal. She had no history of chronic illness, pelvic surgery, chemotherapy, radiotherapy, or long-term medication use. Her mother had experienced menarche at 13 years, and no family history of primary amenorrhea, infertility, or congenital genital anomalies was reported.

The patient had married at 22 years of age and reported sexual activity. Her total Female Sexual

Function Index score was 20.8, suggesting possible sexual dysfunction. The duration of marriage recorded in the source clinical document was inconsistent and therefore requires verification before submission. She had never been pregnant and had never used contraception.

On examination, her height was 155 cm, weight 55 kg, and body mass index 22.8 kg/m^2. Blood pressure was 115/73 mmHg, pulse rate 76 beats/min, respiratory rate 20 breaths/min, temperature 36.9 °C, and oxygen saturation 98% on room air. Breast development was Tanner stage V, and pubic hair development was Tanner stage IV. There were no clinical signs of virilization. The external genitalia, including the labia and clitoris, appeared normal. A vaginal pouch approximately 1 cm deep was identified. Speculum and digital vaginal examinations were not attempted because the short pouch made these procedures unlikely to provide additional information and increased the risk of pain or mucosal trauma. Rectal examination did not identify a pelvic mass.

Routine hematological evaluation showed a hemoglobin concentration of 12.6 g/dL, leukocyte count of 6.59 × 10^3/µL, and platelet count of 301 × 10^3/µL. The mean corpuscular volume was mildly reduced at 79 fL. Transabdominal and transvaginal ultrasonography did not adequately visualize the uterus or either ovary. The kidneys were reported as normal, no free pelvic fluid was detected, and the sonographic impression was suspected Müllerian agenesis.

Because pelvic anatomy remained uncertain after ultrasonography, diagnostic laparoscopy was performed under general anesthesia. Examination under anesthesia again demonstrated a short vaginal pouch with resistance to gentle probing, consistent with vaginal canal agenesis. Laparoscopic exploration showed no macroscopically identifiable uterus. Both ovaries and fallopian tubes appeared anatomically normal. The estimated blood loss was 10 mL, and no intraoperative complication occurred. The operative findings, together with primary amenorrhea, normal secondary sexual characteristics, and vaginal agenesis, strongly supported MRKH syndrome.

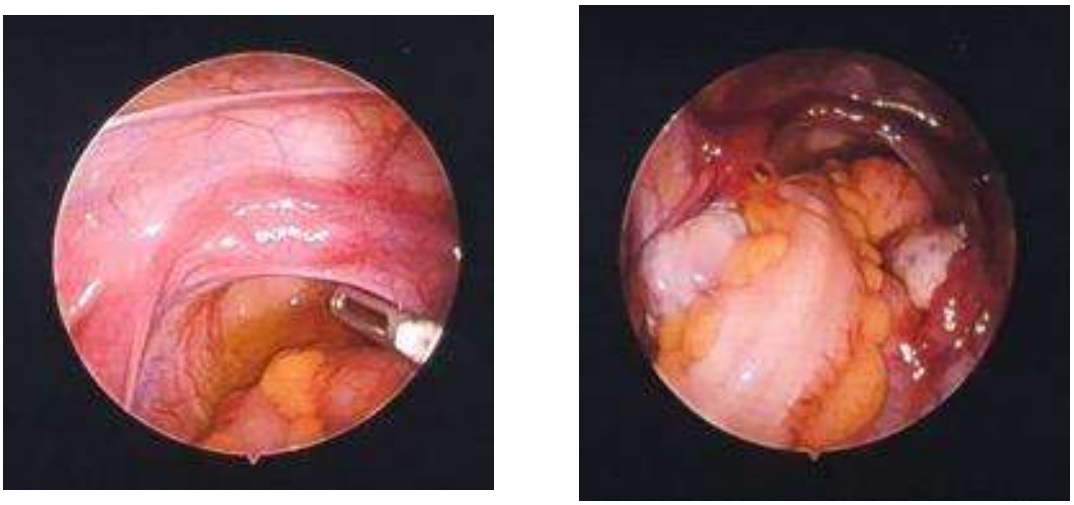
The patient received postoperative monitoring, analgesia, and counseling. The counseling explained that uterine agenesis accounted for the absence of menstruation, while apparently preserved ovaries were consistent with maintained ovarian endocrine function. Sexual rehabilitation, psychological support, and future reproductive counseling were discussed. No

vaginal reconstructive procedure was performed during the reported admission. Follow-up outcomes and the patient's treatment preference regarding progressive vaginal dilation or later vaginoplasty were not documented in the source material.

Table 1. Clinical Timeline

Time	Clinical event	Key finding or action
Since puberty	Primary amenorrhea	Normal secondary sexual development; no cyclical pelvic pain
After marriage	Sexual concerns	FSFI total score 20.8; duration of marriage requires confirmation
25 June 2026	Laboratory and ultrasound assessment	Routine blood results largely unremarkable; uterus and ovaries not adequately visualized; Müllerian agenesis suspected
30 June 2026	Gynecological assessment	Normal external genitalia and an approximately 1 cm vaginal pouch
30 June 2026	Diagnostic laparoscopy	Uterus not identified; bilateral ovaries and fallopian tubes appeared normal
Postoperative period	Counseling and recovery	Diagnosis, sexual function, psychological support, and reproductive implications discussed; longer-term follow-up not available

Intraoperative Findings



A B

Figure 1. Representative laparoscopic views showing no macroscopically identifiable uterus and preserved adnexal structures. Images may be used only after written patient consent for publication has been confirmed.

DISCUSSION

The combination of primary amenorrhea, normal breast development, normal external genitalia, a markedly shortened vagina, and an absent uterus strongly suggests MRKH syndrome. Ovarian function and secondary sexual development are usually preserved because the ovaries arise from the gonadal ridge rather than the Müllerian ducts.1 The bilateral ovaries and fallopian tubes observed during laparoscopy further supported a Müllerian developmental anomaly in this patient.

The differential diagnosis of primary amenorrhea with an absent uterus includes complete androgen insensitivity syndrome. Pubic and axillary hair are often sparse in complete androgen insensitivity syndrome, whereas this patient had Tanner stage IV pubic hair and structures described intraoperatively as normal ovaries. Nevertheless, the diagnostic workup was incomplete because serum gonadotropins, estradiol, testosterone, anti-Müllerian hormone, and a 46,XX karyotype were not documented. These tests should be obtained or the limitation should remain explicit in the final manuscript.2

Ultrasonography is generally the first imaging modality, but the uterus and ovaries may be difficult to identify because of bowel gas, ectopic ovaries, or small Müllerian remnants. MRI provides more detailed noninvasive assessment of uterine remnants, vaginal length, ovarian location, and associated renal or skeletal anomalies.4 In this case, ultrasonography was inconclusive and diagnostic laparoscopy directly established the internal anatomy. Although laparoscopy provided useful confirmation, contemporary evaluation would usually favor MRI before an invasive diagnostic procedure when it is available and when no concurrent therapeutic intervention is planned.

The patient was diagnosed at 22 years of age after marriage, which is later than the usual adolescent presentation. Delayed recognition can magnify distress because the diagnosis may emerge together with difficulty during intercourse and unexpected information about infertility. Qualitative and systematic-review evidence describes anxiety, grief, altered body image, concerns about femininity, and impaired sexual well-being among individuals with MRKH.5,8 The FSFI score of 20.8 in this case signals possible dysfunction, but it should not be treated as a definitive diagnosis. The duration and context of sexual activity, relationship factors, pain, cultural expectations, and emotional response to the new diagnosis may all affect the score.

Management should reflect the patient's goals and readiness. When creation or lengthening of a functional vagina is desired, supervised progressive dilation is recommended as first-line therapy because it avoids surgery and achieves satisfactory anatomical and functional outcomes in many patients.2 Surgery should be considered when dilation is unsuccessful, unacceptable, or unsuitable after informed shared decision-making. The timing of any intervention should be chosen by the patient rather than determined solely by marital status or social pressure.

Long-term outcomes should include more than vaginal length or technical success. Arfita and colleagues reported that surgery for congenital reproductive tract anomalies may improve symptoms and organ function, but sexual-function outcomes can vary and require counseling about realistic expectations.7 Other studies similarly show that satisfactory anatomy does not automatically eliminate psychosexual distress.5,6 For the present patient, structured follow-up should therefore assess comfort with sexual activity, pain, lubrication, emotional well-being, relationship concerns, and preferred reproductive goals.

The absence of a functional uterus results in absolute uterine factor infertility, but preserved ovaries may allow biological parenthood through oocyte retrieval with gestational surrogacy where legally and ethically permitted. Uterus transplantation is an emerging option in selected centers but remains complex, resource intensive, and dependent on national regulation.3 Counseling in Indonesia must therefore be individualized and should distinguish technically possible options from those that are legally accessible and locally available.

MRKH type II cannot be excluded solely because renal ultrasonography was reported as normal. A complete assessment should include the urinary tract and consideration of vertebral, skeletal, auditory, and cardiac anomalies according to clinical findings. Recent cohort evidence suggests that associated malformations are sometimes insufficiently investigated.3 Genetic counseling may also be considered because MRKH has a heterogeneous genetic background involving several developmental pathways, although a causative variant is not identified in most patients.9,10

This report has several limitations. MRI, endocrine testing, and karyotyping were not documented; systematic screening for associated anomalies was incomplete; and longer-term follow-up after counseling was unavailable. The source record also contained conflicting information about the duration of marriage. These limitations reduce diagnostic completeness but offer an important clinical lesson: anatomical confirmation should be integrated with a standardized amenorrhea workup, assessment for associated anomalies, and longitudinal psychosexual care.

CONCLUSION

MRKH syndrome should be considered promptly in patients with primary amenorrhea, normal secondary sexual characteristics, and a short or absent vagina. In this young married woman, inconclusive ultrasonography was followed by laparoscopy showing uterine agenesis and apparently normal bilateral adnexa. The delayed diagnosis and low FSFI score highlight the need to address sexual well-being, psychological adaptation, and reproductive goals alongside anatomical findings. A complete endocrine, genetic, and malformation assessment should be documented before the final manuscript is submitted.

PATIENT PERSPECTIVE

[To be completed after obtaining the patient's voluntary account. A brief statement may describe her experience of delayed diagnosis, the explanation she received, and her priorities for future sexual and reproductive care. Do not infer or construct a patient perspective without interviewing her.]

DECLARATIONS

Competing interests: The authors declare that they have no competing interests.

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Data availability: The clinical data supporting this report are contained within the article. Additional information cannot be shared publicly because of patient confidentiality.

Author contributions: Ika Arfita: conceptualization, manuscript drafting, clinical supervision, interpretation, critical manuscript revision, and correspondence. Muhamad Kelvin Maulana: data collection, initial case documentation, and manuscript drafting. All authors: final approval and accountability for the published work.

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