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"Uterine Agenesis (Mayer-Rokitansky-Küster-Hauser Syndrome) Diagnosed At Queen Mary Hospital, Lucknow"

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Abstract
Background: *Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a congenital disorder characterized by uterine agenesis and upper vaginal aplasia in individuals with a 46,XX karyotype. It is a significant cause of primary amenorrhea. This case report describes the diagnostic process and initial management of MRKH syndrome in a young woman presenting to a tertiary care center in Northern India.*
Case Presentation: *An 18-year-old nulliparous woman presented to Queen Mary Hospital, Lucknow, India, with primary amenorrhea. She had normal secondary sexual characteristics and no significant past medical or family history. Physical examination revealed normal external genitalia but a short vaginal dimple. Hormonal assays were consistent with normal ovarian function. Pelvic ultrasound and MRI confirmed the absence of the uterus and cervix, with normal ovaries. Karyotyping revealed a 46,XX karyotype. A diagnosis of MRKH syndrome type 1 was established. Management included extensive counseling regarding the diagnosis, prognosis, and reproductive options. Vaginal dilatation therapy (Frank's method) was initiated, and psychological support was offered.*
Conclusion: *This case highlights the importance of considering MRKH syndrome in young women presenting with primary amenorrhea and normal secondary sexual characteristics. A systematic diagnostic approach including clinical examination, hormonal evaluation, imaging, and karyotyping is crucial. Comprehensive management necessitates a multidisciplinary approach with psychological support, sexual health counseling, and discussion of reproductive alternatives. This case from a tertiary center in Lucknow underscores the need for increased awareness and accessible care for MRKH syndrome in diverse healthcare settings.*
Keywords: *Uterine Agenesis, MRKH Syndrome, Primary Amenorrhea, Congenital Anomaly, Case Report, India, Vaginal Dilatation*

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I. Background

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a congenital anomaly affecting approximately 1 in 4500-5000 female births, making it a significant cause of primary amenorrhea [1]. Characterized by the agenesis or hypoplasia of the uterus and the upper two-thirds of the vagina in individuals with a 46,XX karyotype and normal ovarian function, MRKH syndrome presents with primary amenorrhea in adolescence despite the presence of normal secondary sexual characteristics [2]. Early and accurate diagnosis is paramount to facilitate appropriate management, including addressing psychological well-being, sexual function, and reproductive aspirations [3]. Queen Mary Hospital, a tertiary care center in Lucknow, Uttar Pradesh, India, manages a wide spectrum of gynecological conditions, including complex congenital anomalies. This case report aims to illustrate the clinical presentation, diagnostic pathway, and initial management strategies employed for a patient diagnosed with uterine agenesis at our institution.

II. Case Presentation

An 18-year-old nulliparous woman presented to the outpatient department of Gynecology and Obstetrics at Queen Mary Hospital, Lucknow, with the chief complaint of primary amenorrhea. She reported never having experienced menstruation.

Clinical Findings Menstrual History: Primary amenorrhea since the expected age of menarche (13 years, based on average Indian menarche age [4]).
Past Medical History: Unremarkable, with no history of chronic illnesses, surgeries, or medications.

Developmental History: Reached normal developmental milestones. Normal breast development (Tanner stage 4), and pubic and axillary hair development consistent with her age.

Family History: No reported family history of menstrual irregularities, infertility, or congenital anomalies.

Sexual History: Not sexually active.

Psychosocial History: Student, expressing concern and anxiety regarding the absence of menstruation and its potential impact on future fertility.

General Physical Examination: Appeared healthy, well-nourished, and of average build. Vital signs were within normal limits. Systemic examination revealed normal cardiovascular, respiratory, gastrointestinal, and neurological systems.

Pelvic Examination: External Genitalia: Normal female external genitalia with well-developed labia majora and minora, clitoris, and urethra. **Vaginal Examination:** Absent vaginal opening or a very short vaginal dimple (approximately 1-2 cm in depth). No cervix palpable. **Rectal Abdominal Examination:** Uterus was not palpable. Adnexa were not distinctly felt.

Diagnostic Evaluation

- **Hormonal Profile (Day 3 of expected cycle):**
  - FSH: 6.5 mIU/mL (Normal follicular phase range)
  - LH: 5.0 mIU/mL (Normal follicular phase range)
  - Estradiol: 45 pg/mL (Normal follicular phase range)
  - Serum Prolactin: 12 ng/mL (Normal range)
  - Thyroid Stimulating Hormone (TSH): 2.0 mIU/L (Normal range)
- **Abdominal and Pelvic Ultrasound:** Revealed the absence of the uterus and cervix. Ovaries were visualized bilaterally and appeared normal in size and morphology. Kidneys were visualized and appeared normal.
- **Pelvic Magnetic Resonance Imaging (MRI):** Confirmed the absence of the uterus and cervix. The vagina was noted to be a blind-ending pouch of approximately 2 cm in length. Ovaries were normal. No evidence of hematocolpos or hematometra. No associated renal or skeletal anomalies were visualized on this imaging.
- **Karyotype:** 46, XX (Normal female karyotype)
- **Renal Ultrasound:** Performed to rule out associated renal anomalies, and was found to be normal.

Timeline

- **Age 18:** Presentation to Queen Mary Hospital with primary amenorrhea.
- **Within 1 week of presentation:** Hormonal profile, pelvic ultrasound, and karyotype performed.
- **Within 2 weeks of presentation:** Pelvic MRI and renal ultrasound performed. Diagnosis of MRKH syndrome established. Initial counseling and management plan initiated, including referral for vaginal dilatation therapy.

Diagnostic Challenges

The diagnosis of MRKH syndrome in this case was relatively straightforward due to the classic presentation of primary amenorrhea with normal secondary sexual characteristics and the clear findings on imaging. The diagnostic challenge primarily lay in excluding other less common causes of primary amenorrhea, which was achieved through the comprehensive workup.

Differential Diagnosis

The differential diagnosis for primary amenorrhea includes:

- **Constitutional delay of puberty:** Ruled out by age and normal secondary sexual characteristics.
- **Hypogonadotropic hypogonadism:** Ruled out by normal FSH and LH levels.
- **Hypergonadotropic hypogonadism (Turner syndrome, premature ovarian failure):** Ruled out by normal FSH and LH levels and 46,XX karyotype.
- **Androgen Insensitivity Syndrome (AIS):** Ruled out by 46,XX karyotype and female phenotype.
- **Imperforate hymen:** Ruled out by physical examination.
- **Vaginal agenesis/transverse vaginal septum (other than MRKH):** Considered, but imaging strongly favored MRKH due to uterine agenesis.

Diagnosis

Based on the clinical presentation, hormonal profile, imaging findings, and karyotype, a diagnosis of **Uterine Agenesis (Mayer-Rokitansky-Küster-Hauser Syndrome, Type 1)** was made.

Prognosis

The prognosis for overall health and longevity is excellent in MRKH syndrome. Patients have normal ovarian function and can achieve pregnancy through assisted reproductive technologies such as surrogacy. With vaginal dilatation therapy or surgical vaginoplasty, a functional vagina for sexual intercourse can be created, leading to a good prognosis for sexual function [5].

Rationale for Treatment

The primary goals of management are to address the patient's concerns regarding primary amenorrhea, sexual function, and reproductive options. Vaginal dilatation therapy is the first-line treatment to create a functional vagina. Psychological support is crucial to address the emotional impact of the diagnosis. Counseling regarding reproductive options is essential for future family planning.

Treatment

- **Counseling:** Extensive counseling was provided to the patient and her family regarding the diagnosis of MRKH syndrome. The etiology, prognosis, implications for fertility, and available management options were discussed in detail. Emphasis was placed on the preservation of ovarian function and the possibility of genetic counseling, although MRKH is often sporadic. Psychological support resources within Queen Mary Hospital were offered.
- **Vaginal Dilatation Therapy:** Vaginal dilatation therapy (Frank's method) was recommended as the initial approach to create a functional vagina. The patient was referred to a specialist gynecologist at Queen Mary Hospital experienced in this technique. Educational materials and ongoing support were provided.
- **Further Investigations and Referrals:** Long-term follow-up for renal function was advised, although initial renal ultrasound was normal. Referral to a genetic counselor was offered to discuss the genetic aspects of MRKH syndrome.
- **Reproductive Options Counseling:** The patient was informed about reproductive options, including adoption and surrogacy. The complexities of surrogacy laws in India were explained [6].
- **Follow-up:** Regular follow-up appointments were scheduled at Queen Mary Hospital to monitor progress with vaginal dilatation, provide ongoing psychological support, and address any concerns.

Follow-up and Outcomes

At the time of case report submission, the patient was undergoing vaginal dilatation therapy and attending regular follow-up appointments. The focus of follow-up was on monitoring the progress of vaginal dilatation, providing ongoing psychological support, and addressing any emerging concerns. The outcome of vaginal dilatation therapy and the patient's long-term adjustment to the diagnosis will be monitored in subsequent follow-up visits.

Patient Perspective

The patient expressed initial distress and anxiety upon receiving the diagnosis of uterine agenesis, particularly regarding its impact on her future fertility and sexual function. However, after extensive counseling and understanding the management options, she expressed a sense of hope and motivation to pursue vaginal dilatation therapy. She appreciated the comprehensive and compassionate care provided at Queen Mary Hospital.

Informed Consent

Written informed consent was obtained from the patient for publication of this case report, including clinical details and imaging findings.

III. Discussion

This case report from Queen Mary Hospital, Lucknow, illustrates the typical presentation, diagnostic workup, and initial management of MRKH syndrome in a young woman presenting with primary amenorrhea. The diagnosis was established through a systematic approach encompassing clinical examination, hormonal evaluation, pelvic imaging (ultrasound and MRI), and karyotyping, consistent with established diagnostic guidelines [7].

MRKH syndrome, though considered a rare condition, is a significant cause of primary amenorrhea. It is crucial for clinicians in tertiary care settings like Queen Mary Hospital to be familiar with its presentation and diagnostic modalities. The normal secondary sexual characteristics and hormonal profile in these patients often

point towards a Müllerian anomaly, necessitating imaging to confirm uterine agenesis [8]. MRI is considered the gold standard for imaging in MRKH syndrome to accurately delineate the anatomy and rule out associated anomalies [9].

The management of MRKH syndrome is multidisciplinary and patient-centered. Psychological support is paramount from the time of diagnosis, as the condition can have a significant emotional impact on young women regarding their body image, sexual identity, and reproductive potential [10]. Vaginal dilatation therapy, as initiated in this case, is the first-line approach to create a functional vagina, with high success rates reported [5]. For patients desiring biological children, reproductive options such as adoption and surrogacy are important considerations, requiring sensitive and comprehensive counseling, particularly in the Indian socio-cultural context.

This case highlights the role of tertiary care centers like Queen Mary Hospital in providing comprehensive diagnostic and management services for rare conditions like MRKH syndrome in resource-constrained settings. Increased awareness among healthcare providers and accessible psychological and sexual health support are essential to improve the quality of life for individuals with MRKH syndrome.

IV. Conclusion

This case report details the successful diagnosis and initial management of uterine agenesis (MRKH syndrome) in an 18-year-old woman at Queen Mary Hospital, Lucknow. The case underscores the importance of a systematic diagnostic approach and multidisciplinary care, including psychological support, vaginal dilatation therapy, and reproductive options counseling. Early diagnosis and comprehensive management are crucial for addressing the physical and psychological needs of patients with MRKH syndrome, improving their overall well-being and quality of life.

List of Abbreviations

- MRKH: Mayer-Rokitansky-Küster-Hauser
- FSH: Follicle-Stimulating Hormone
- LH: Luteinizing Hormone
- TSH: Thyroid Stimulating Hormone
- MRI: Magnetic Resonance Imaging

Declarations

Ethics approval and consent to participate

This case report involves a single patient and describes routine clinical care. Formal ethics approval was not required as per institutional guidelines for case reports. Written informed consent to participate was obtained from the patient.

Consent for publication

Written informed consent was obtained from the patient for publication of this case report and any accompanying images/clinical details. A copy of the consent form is available for review by the journal editors.

Availability of data and materials

All relevant data and materials pertaining to this case are included in the manuscript. Further anonymized data may be available from the corresponding author upon reasonable request.

Competing interests

The authors declare that they have no competing interests.

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Authors' contributions

- **Lokesh Kumar Sharma:** Conceptualization, Data collection, Patient management, Manuscript writing, Review & Editing.
- **Tanima Verma:** Patient management, Data interpretation, Manuscript review.
- **Suman Lata:** Nursing care, Patient counseling, Manuscript review.
- **Pooja Singh:** Review & Editing.

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