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Original Article

Radiological Approach of Uterine Agenesis caused by Mularian Duct Malformation

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Abstract

Background: Müllerian duct anomalies (MDAs) are a group of congenital malformations affecting the female reproductive tract, leading to significant reproductive challenges. Uterine agenesis, particularly in the form of Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome, is one of the most severe forms, causing primary amenorrhea and infertility. Imaging plays a crucial role in diagnosis and classification of these anomalies. ***Objective:*** The purpose of the present study was to evaluate the radiological approach in diagnosing uterine agenesis caused by Müllerian duct malformations. ***Methodology:*** This hospital-based observational study conducted form February 2024 to January 2025 included 65 patients diagnosed with uterine agenesis caused by MDAs. Radiological evaluations with ultrasound and magnetic resonance imaging (MRI) were conducted, with additional assessments through hysterosalpingography and three-dimensional ultrasound where necessary. Descriptive statistics were used to analyze the clinical and radiological findings. ***Results:*** The mean age of participants was 22.4±3.8 years. Primary amenorrhea was the most common clinical presentation (69.23%), followed by pelvic pain (15.38%) and infertility (13.85%). Uterine agenesis was the most frequent radiological finding (60%), with 40% of cases also showing associated renal anomalies. Class I Müllerian anomalies (hypoplasia/aplasia) were the most common (60%). Surgical success was achieved in 83.08% of cases, and 30.77% of patients who pursued fertility treatments achieved pregnancy. Psychological support improved the psychosocial outcomes in 75.38% of patients. ***Conclusion:*** Radiological imaging, particularly ultrasound and MRI, is essential for the accurate diagnosis and management of uterine agenesis and other Müllerian duct anomalies.

Keywords: :Müllerian duct anomalies; uterine agenesis; MRKH syndrome; ultrasound; MRI; fertility; psychological support; radiological imaging; congenital malformations

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Introduction

Müllerian duct anomalies (MDAs) represent a broad spectrum of congenital malformations resulting from incomplete development, fusion, or resorption of the paired Müllerian ducts during embryogenesis[1]. These anomalies affect the female reproductive tract, leading to varying degrees of uterovaginal defects. Among these, uterine agenesis is one of the most severe forms, characterized by the complete absence of uterine tissue above the vagina[2]. Globally, MDAs occur in approximately 1 to 5.0% of all women, with uterine agenesis seen in about 1 in 5,000 female live births, making it a relatively rare but clinically significant condition[3]. Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome, the most recognized form of uterine agenesis, involves congenital absence of the uterus and.

upper two-thirds of the vagina It has two subtypes: type I (isolated MRKH), affecting only reproductive organs, and type II (MURCS association), which includes renal, cardiac, skeletal, and auditory anomalies[4]. The absence of the uterus in affected individuals leads to primary amenorrhea despite normal ovarian function, often resulting in significant psychological distress and infertility concerns[5]. Embryologically, the Müllerian ducts are responsible for forming the fallopian tubes, uterus, cervix, and upper vagina. Their development begins in early gestation, around the 6th week, and proceeds through a complex process of elongation, fusion, and canalization[6]. Uterine agenesis occurs due to a disruption in the elongation phase, resulting in either complete failure of uterine formation or the presence of rudimentary, nonfunctional uterine remnants[7]. Most patients present during adolescence with primary amenorrhea but normal secondary sexual characteristics, as ovarian function remains intact[8].

Some may experience cyclical pelvic pain from obstructed rudimentary uterine structures with functional endometrial tissue. Imaging is crucial for diagnosis and differentiating from other causes of primary amenorrhea, like androgen insensitivity syndrome or gonadal dysgenesis[9]. Radiological imaging is key in diagnosing and classifying uterine agenesis. Ultrasound, the first-line modality, is favored for its non-invasiveness, accessibility, and cost-effectiveness[10]. Transabdominal or transvaginal ultrasound can detect uterine absence and assess rudimentary structures but may struggle with complex Müllerian anomalies, especially small uterine remnants[11].

Magnetic resonance imaging (MRI) has emerged as the gold standard for diagnosing MDAs, offering superior soft-tissue contrast and multiplanar capabilities. MRI provides a comprehensive assessment of pelvic anatomy, accurately delineating the extent of uterine agenesis, the presence of rudimentary uterine horns, and associated renal anomalies[12]. Other imaging techniques, such as hysterosalpingography (HSG) and three-dimensional ultrasound, may be useful in select cases but are less commonly employed due to their limitations in patients with complete uterine agenesis[13]. In complex cases requiring further anatomical clarification, laparoscopy can serve as both a diagnostic and therapeutic tool, confirming the absence of the uterus and evaluating associated pelvic abnormalities[14]. Management of uterine agenesis focuses on both anatomical and psychological aspects. Patients diagnosed with MRKH syndrome often require counseling to address the emotional impact of infertility[15]. Early diagnosis of uterine agenesis is crucial for appropriate clinical management and addressing reproductive challenges. This study aims to evaluate the radiological approach in diagnosing uterine agenesis caused by Müllerian duct malformation.

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Methodology

Study Settings and Population: This study was a hospital-based observational study assessed the radiological approach to diagnosing uterine agenesis due to Müllerian duct malformation. The study was performed at Monno Medical College Hospital, Manikganj, Bangladesh and Delta Medical College Hospital, Dhaka, Bangladesh over a period of one year. The study included 65 patients with uterine agenesis caused by Müllerian duct malformation. Female patients diagnosed with Müllerian duct anomalies (MDAs) via imaging studies (ultrasound and MRI), patients presenting with primary amenorrhea and suspected congenital uterine anomalies, patients who underwent radiological and/or surgical confirmation of their diagnosis, patients with complete medical records and follow-up data were included int his study. Patients diagnosed with uterine anomalies secondary to acquired conditions such as infections, trauma, or tumors or patients with known chromosomal abnormalities affecting genital tract development like Turner syndrome were excluded from this study.

Study Procedure: The data collection was retrospective and involved the analysis of medical records, imaging findings, and clinical presentations of patients diagnosed with uterine agenesis. Medical records of patients diagnosed with uterine agenesis from February 2024 to January 2025 were reviewed. Data were extracted on demographic details (age, clinical presentation), imaging findings, associated anomalies, and management strategies. Imaging reports were reviewed by two independent radiologists to ensure diagnostic accuracy.

Imaging Assessment: Transabdominal and transvaginal ultrasound was performed as the first-line diagnostic tool to assess the presence of the uterus and associated anomalies. Magnetic Resonance Imaging (MRI) was conducted for detailed anatomical assessment, confirming the absence or presence of rudimentary uterine structures and identifying associated renal or skeletal abnormalities. In selected cases, hysterosalpingography (HSG) and three-dimensional ultrasound were used to further delineate anomalies.

Statistical Analysis: Data were analyzed using SPSS version 26.0. Descriptive statistics were used to summarize demographic and clinical characteristics. Categorical variables were expressed as frequencies and percentages, while continuous variables were presented as mean ± standard deviation (SD).

Ethical Consideration: The study was approved by the Institutional Ethics Committee, and all procedures adhered to the ethical principles outlined in the Declaration of

Helsinki. As this was a retrospective study utilizing anonymized medical records, patient consent was waived. Confidentiality was strictly maintained, and no identifiable information was included in the analysis.

Results

A total of 65 patients were included in the study, with a mean age of 22.4±3.8 years. The majority of participants presented with primary amenorrhea, which was observed in 69.23% of cases, making it the most common clinical complaint. Pelvic pain was reported by 15.4% of the women, while 13.9% presented with infertility issues. A family history of congenital anomalies was noted in 20.0% of the cases, and 15.38% of participants were born of consanguineous marriages (Table 1).

Table 1: Baseline characteristics of the study population (n=65)

Variable	Frequency	Percent
Age (years)	22.4 ± 3.8	
Primary amenorrhea	45	69.2
Pelvic pain	10	15.4
Infertility	9	13.9
Family History of Anomaly	13	20.0
Consanguineous Marriage	10	15.4

Radiological evaluations identified uterine agenesis as the most prevalent anatomical anomaly, affecting 60.00% of the study population. Hypoplastic uterus was diagnosed in 26.2% of patients, while unicornuate uterus and septate uterus were observed in 9.2% and 6.2% of cases, respectively. Associated renal anomalies were presented in 40.0% of participants, highlighted the common coexistence of renal tract abnormalities in patients with Müllerian anomalies. In addition, ovarian abnormalities were found in 15.4% of the cases (Table 2).

Table 2: Distribution by Radiological Findings (n=65)

Radiological Findings	Frequency	Percent
Uterine Agenesis	39	60.0
Hypoplastic Uterus	17	26.2
Unicornuate Uterus	6	9.2
Septate Uterus	4	6.2
Associated Renal Anomaly	26	40.0
Ovarian Abnormalities	10	15.4

Classification based on the American Society for Reproductive Medicine (ASRM) system revealed that Class I Müllerian anomalies (hypoplasia/aplasia) were the most frequently encountered, affecting 60.0% of participants. Class II anomalies (unicornuate uterus) were present in 15.4%, followed by Class IV (bicornuate uterus) in 10.8%, Class V (septate uterus) in 7.7%, and Class III (uterine didelphys) in 6.15%, as illustrated in Figure I.

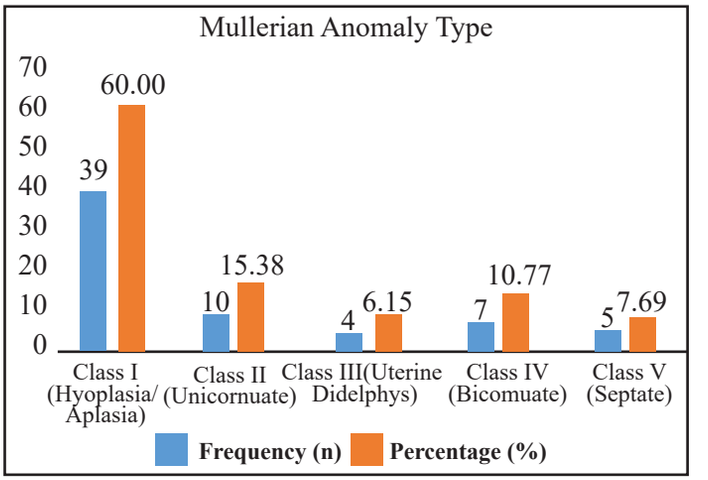

Figure I: Müllerian Anomaly Type among patients (n=65)

In terms of management, 70.8% of patients received counseling and psychological support, emphasizing the importance of addressing emotional and mental health concerns in this population. Vaginal reconstruction surgery was performed in 40.0% of cases, while 29.2% were managed with hormonal therapy. Hysteroscopic surgery was carried out in 9.2%, and 13.9% underwent assisted reproductive techniques (Table 3).

Table 3: Treatment and Management Approaches of Participants (n=65)

Treatment Option	Frequency	Percent
Hormonal Therapy	19	29.2
Vaginal Reconstruction Surgery	26	40.0
Hysteroscopic Surgery	6	9.2
Assisted Reproductive Techniques (ART)	9	13.9
Counseling & Psychological Support	46	70.8

During follow-up, which averaged 4.2±1.5 years, surgical success was achieved in 83.1% of patients. A pregnancy success rate of 30.8% was recorded among those who desired fertility. Furthermore, 75.4% of participants demonstrated psychosocial improvement, reflecting a positive impact of multidisciplinary management strategies over time (Table 4).

Table 4: Prognosis and Follow-Up Data of Women (n=65)

Parameter	Frequency	Percent
Follow-up Duration (years), Mean ± SD	4.2 ± 1.5	
Surgical Success Rate	54	83.1
Pregnancy Success Rate	20	30.8
Psychosocial Improvement	49	75.4

Discussion

Uterine agenesis and Müllerian duct malformations are congenital anomalies resulting from improper development, fusion, or resorption of the Müllerian ducts during embryogenesis. These conditions can significantly impact reproductive health, often presenting with primary amenorrhea or infertility. Radiological imaging plays a crucial role in accurately diagnosing and classifying these anomalies. Early and precise diagnosis guides appropriate clinical management and treatment planning. In our study, the mean age was 22.4±3.8 years. It was comparable with the findings of Hua et al[16], who observed an average maternal age of approximately 29 years among the patients in their study. Primary amenorrhea was the most common presentation (69.2%), aligning with Saravelos et al[17], who reported similar findings among women with Müllerian anomalies. Pelvic pain (15.4%) and infertility (13.9%) were less frequent but comparable to rates observed by Grimbizis et al[18]. A positive family history (20.0%) and consanguinity (15.4%) support possible genetic contributions, consistent with Oppelt et al[19], who noted familial clustering in such anomalies.

This study highlights the radiological patterns of Müllerian duct malformations, with uterine agenesis being the most common finding (60.0%). This aligns with previous studies, such as Oppelt et al[19] and Morcel et al[20], who identified uterine agenesis as the hallmark of Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome. The presence of hypoplastic uterus (26.2%) and unicornuate uterus (9.23%) is also consistent with reports by Gordts[21], reflecting the diverse spectrum of Müllerian anomalies. Septate uterus was observed in 6.2% of cases, slightly lower than in some other populations, possibly due to differences in diagnostic techniques or sample size. Renal anomalies were associated in 40.0% of cases, supporting the well-established embryological link between the reproductive and urinary systems[19]. The current study indicates that Class I Mullerian anomalies (Hypoplasia/Aplasia) are the most prevalent, accounting for 60.0% of cases, followed by Class II (Unicornuate) at 15.4%. This finding contrasts with several previous studies where Class I anomalies were less common. For example, Grimbizis et al reported septate uterus (Class V) as the most frequent anomaly[22], while the American Society for Reproductive Medicine (ASRM, 1988) also noted a higher incidence of septate and arcuate anomalies compared to hypoplasia[23].

The elevated frequency of Class I anomalies in this study may reflect differences in population genetics, diagnostic criteria, or referral patterns. Further large-scale and multicentric studies are necessary to clarify the true prevalence across populations. Regarding treatment, vaginal reconstruction was performed in 40.0% of cases, reflecting the standard management in MRKH patients lacking a functional vaginal canal. Mentessidou and Mirilas[24] emphasized surgical correction as a cornerstone of treatment in adolescent gynecology for vaginal and uterine anomalies, particularly to facilitate sexual and reproductive function. In comparison to the study by Sugi et al, which emphasized the role of Müllerian anomalies in infertility and pregnancy outcomes, our study showed a 30.77% pregnancy success rate among those who pursued assisted reproductive technologies (ART). This is slightly lower but still encouraging given the anatomical limitations in these patients[25].

There are limitations of this study like retrospective design, potentially introducing selection bias and limiting generalizability, reliance on imaging alone for diagnosis, without genetic or histological confirmation, which could affect accuracy and lack of long-term follow-up data on fertility outcomes and psychosocial aspects, limiting comprehensive understanding.

Conclusion

This study highlights the critical role of radiological imaging in diagnosing uterine agenesis and other MDAs, providing valuable insights into the spectrum of these congenital malformations. Early and accurate imaging, particularly with ultrasound and MRI, is essential for timely diagnosis and effective management. The high prevalence of associated renal anomalies emphasizes the importance of comprehensive evaluation. Psychological counseling and multidisciplinary management, including surgical interventions and assisted reproductive technologies, significantly contribute to improving patient outcomes. Future studies should focus on larger, multicenter cohorts to refine diagnostic criteria and treatment strategies, ensuring better clinical outcomes and fertility prospects for affected individuals.

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Conflict of interest: None declared

Ethical approval: The study was approved by the Institutional Ethics Committee.

References

1.Santana González L, Artibani M, Ahmed AA. Studying Müllerian duct anomalies–from cataloguing phenotypes to discovering causation. Disease Models & Mechanisms. 2021 ;14(6):dmm047977.

2.Troiano RN, McCarthy SM. Mullerian duct anomalies: imaging and clinical issues. Radiology. 2004 ;233(1):19-34.

3.Fahmy M. Rare congenital genitourinary anomalies. Springer-Verlag Berlin An; 2016.

4.MedlinePlus. Mayer-Rokitansky-Küster-Hauser syndrome [Internet]. Bethesda (MD): National Library of Medicine (US); 2022 [cited 2025 Mar 25]. Available from: https://medlineplus.gov/genetics/condition/mayer-rokitansky-kuster-hauser-syndrome/

5.Goodman N, Cobin RH. Reproductive disorders. Evidence-Based Endocrinology, 3rd ed.; Lippincott Williams & Wilkins: Philadelphia, PA, USA. 2012:190-228.

6.Falcone T, Hurd WW. Clinical reproductive medicine and surgery [Internet]. 2007

7.Reichman DE, Laufer MR. Congenital uterine anomalies affecting reproduction. Best Practice & Research Clinical Obstetrics & Gynaecology. 2010 ;24(2):193-208.

8.Mentessidou A, Mirilas P. Surgical disorders in pediatric and adolescent gynecology: Vaginal and uterine anomalies. International Journal of Gynecology & Obstetrics. 2023 ;160(3):762-70.

9.Acién P, Acién M. Malformations of the female genital tract and embryological bases. Current Women's Health Reviews. 2007 ;3(4):248-88.

10.e Passos ID, Britto RL. Diagnosis and treatment of müllerian malformations. Taiwanese Journal of Obstetrics and Gynecology. 2020 ;59(2):183-8.

11.Wu MH, Hsu CC, Huang KE. Detection of congenital müllerian duct anomalies using three-dimensional ultrasound. Journal of clinical Ultrasound. 1997 ;25(9):487-92.

12.Sugi MD, Penna R, Jha P, Põder L, Behr SC, Courtier J, et al. Müllerian duct anomalies: role in fertility and pregnancy. Radiographics. 2021 Oct;41(6):1857-75.

13.Raga F, Bonilla-Musoles F, Blanes J, Osborne NG. Congenital Müllerian anomalies: diagnostic accuracy of three-dimensional ultrasound. Fertility and sterility. 1996 ;65(3):523-8.

14.Pitot MA, Bookwalter CA, Dudiak KM. Müllerian duct anomalies coincident with endometriosis: a review. Abdominal Radiology. 2020 ;45:1723-40.

15.Tsarna E, Eleftheriades A, Eleftheriades M, Kalampokas E, Liakopoulou MK, Christopoulos P. The impact of mayer–rokitansky–küster–hauser syndrome on psychology, quality of life, and sexual life of patients: A systematic review. Children. 2022 ;9(4):484.

16.Hua M, Odibo AO, Longman RE, Macones GA, Roehl KA, Cahill AG. Congenital uterine anomalies and adverse pregnancy outcomes. American journal of obstetrics and gynecology. 2011 ;205(6):558-e1.

17.Saravelos SH, Cocksedge KA, Li TC. Prevalence and diagnosis of congenital uterine anomalies in women with reproductive failure: a critical appraisal. Human reproduction update. 2008 ;14(5):415-29.

18.Grimbizis GF, Di Spiezio Sardo A, Saravelos SH, Gordts S, Exacoustos C, Van Schoubroeck D, et al. The Thessaloniki ESHRE/ESGE consensus on diagnosis of female genital anomalies. Human Reproduction. 2016 ;31(1):2-7.

19.Oppelt P, Renner SP, Kellermann A, Brucker S, Hauser GA, Ludwig KS, et al. Clinical aspects of Mayer–Rokitansky–Kuester–Hauser syndrome: recommendations for clinical diagnosis and staging. Human reproduction. 2006 ;21(3):792-7.

20.Morcel K, Camborieux L, Programme de Recherches sur les Aplasies Müllériennes (PRAM) pram-network. coordination@ univ-rennes1. fr, Guerrier D. Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome. Orphanet journal of rare diseases. 2007 ;2(1):13.

21.Gordts S. The ESHRE-ESGE consensus on the classification of female genital tract congenital anomalies. Gynecological Surgery. 2013 ;10(3):163-.

22.Grimbizis GF, Camus M, Tarlatzis BC, Bontis JN, Devroey P. Clinical implications of uterine malformations and hysteroscopic treatment results. Human reproduction update. 2001 ;7(2):161-74.

23.Adhesions AD. The American Fertility Society classifications of adnexal adhesions, distal tubal occlusion, tubal occlusion secondary to tubal ligation, tubal pregnancies, Müllerian anomalies and intrauterine adhesions. Fertil Steril. 1988;49(6):944.

24.Mentessidou A, Mirilas P. Surgical disorders in pediatric and adolescent gynecology: Vaginal and uterine anomalies. International Journal of Gynecology & Obstetrics. 2023 ; 160(3):762-70.

25.Sugi MD, Penna R, Jha P, Põder L, Behr SC, Courtier J, et al Müllerian duct anomalies: role in fertility and pregnancy. Radiographics. 2021 ;41(6):1857-75