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ORIGINAL ARTICLE

Understanding the Spectrum of Congenital Abnormalities Impacting Female Fertility Through MRI Imaging

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ABSTRACT

Background: Congenital uterine anomalies (CUAs) are structural malformations of the female reproductive tract that arise from disrupted development, fusion, or resorption of the Müllerian ducts.

Objective: To evaluate the spectrum of congenital uterine anomalies affecting female fertility and to assess the diagnostic utility of MRI in identifying and classifying these abnormalities.

Methodology: This was a cross-sectional descriptive study conducted at Sir Ganga Ram Hospital Lahore during December 2022 and May 2023. A total of 110 female patients were included in the study. Non-probability consecutive sampling was used. All MRIs were performed using a 1.5 Tesla system. The imaging protocol included T1-weighted and T2-weighted sequences in axial, sagittal, and coronal planes. Special emphasis was placed on T2-weighted images for optimal delineation of uterine zonal anatomy.

Results: The most common anomaly detected was septate uterus (29.1%), followed by bicornuate uterus (19.1%) and uterine agenesis/hypoplasia (16.4%). A significant association was found between septate uterus and recurrent miscarriage ($p = 0.031$), as well as between uterine agenesis and primary amenorrhea ($p < 0.001$). Renal anomalies were present in 11.8% and vaginal anomalies in 8.2% of cases. MRI allowed precise differentiation between surgically correctable and non-correctable anomalies and revealed associated extra-genital findings in multiple cases.

Conclusion: MRI is a highly accurate, non-invasive tool for the evaluation of congenital uterine anomalies. It offers superior anatomical detail, enables appropriate classification, and guides effective clinical and surgical management. Early identification of these anomalies through MRI can significantly improve reproductive outcomes and prevent unnecessary interventions.

Keywords: Congenital uterine anomalies, Müllerian duct anomalies, infertility, MRI, septate uterus, bicornuate uterus.

INTRODUCTION

Congenital anomalies of the female reproductive tract represent a significant yet often underdiagnosed cause of infertility, menstrual irregularities, and obstetric complications. These anomalies result from aberrations in the embryological development of the Müllerian (paramesonephric) ducts, leading to a diverse spectrum of uterine, cervical, and vaginal malformations[1]. While some anomalies remain asymptomatic and are discovered incidentally, others may manifest as primary amenorrhea, recurrent pregnancy loss, or chronic pelvic pain, necessitating prompt and accurate diagnosis[2]. Magnetic Resonance Imaging (MRI) has emerged as the gold standard imaging modality for evaluating congenital uterine and reproductive tract anomalies due to its superior soft-tissue contrast resolution, multiplanar imaging capabilities, and non-ionizing nature[3]. Unlike conventional imaging tools such as ultrasound or hysterosalpingography, MRI provides comprehensive anatomical visualization of the uterus, cervix, and vagina in a single examination offering precise delineation of the internal and external uterine contours, endometrial cavity, and associated renal or spinal anomalies, which frequently coexist in these cases[4].

Understanding the MRI appearance of various congenital abnormalities ranging from agenesis or hypoplasia (as seen in Mayer-Rokitansky-Küster-Hauser syndrome), unicornuate or bicornuate uterus, to more complex forms such as uterus didelphys or septate uterus, is crucial for clinicians and radiologists[5]. Proper classification, particularly as per the American Society for Reproductive Medicine (ASRM) and the more recent ESHRE/ESGE systems, directly impacts patient counseling, therapeutic decisions, and reproductive planning. Congenital abnormalities of the female reproductive system are often subtle in presentation but can have profound implications on a woman's reproductive health, including fertility, pregnancy maintenance, and overall quality of life[6]. These anomalies are typically due to disrupted formation, fusion, or resorption of the paired Müllerian ducts during embryogenesis, leading to a wide spectrum of structural defects. The incidence of Müllerian anomalies in the general population is estimated to be around 4–7%, but this figure significantly increases in women with infertility or recurrent pregnancy loss, reaching up to 25% in some studies. Early and accurate detection of these abnormalities is critical in clinical practice, as timely intervention can optimize fertility and obstetric outcomes[7].

From a clinical perspective, the most common presenting features of congenital uterine anomalies include primary amenorrhea, cyclic pelvic pain, abnormal uterine bleeding, and infertility[8]. However, because many women with these anomalies remain asymptomatic until reproductive challenges arise, the condition may go unrecognized for years. For instance, a septate uterus, which is the most common structural anomaly, is often not associated with menstrual abnormalities but can lead to repeated miscarriages if left undiagnosed[9]. In contrast, uterine agenesis or hypoplasia may be suspected early due to absent menarche or difficulty in sexual intercourse, prompting earlier imaging evaluation. Magnetic Resonance Imaging (MRI) has revolutionized the evaluation of congenital reproductive anomalies due to its unparalleled capability to assess uterine morphology in detail without radiation exposure[10]. T2-weighted images are particularly valuable in distinguishing different tissue planes and structures, enabling precise assessment of the endometrial stripe, myometrial zonal anatomy, and associated soft-tissue structures[11]. Unlike transvaginal ultrasonography, which is operator-dependent and limited in scope, MRI offers a comprehensive and standardized approach, making it especially useful in complex or equivocal cases. MRI also plays a critical role in differentiating among the various types of Müllerian anomalies, which is essential for determining the appropriate management strategy[12]. For instance, distinguishing a septate uterus from a bicornuate uterus is essential, as the former can often be corrected surgically via hysteroscopic metroplasty, while the latter typically does not benefit from surgical correction. Failure to make this distinction can lead to unnecessary or ineffective interventions.

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Objective: To evaluate the spectrum of congenital uterine anomalies affecting female fertility and to assess the diagnostic utility of MRI in identifying and classifying these abnormalities.

METHODOLOGY

This was a cross-sectional descriptive study conducted at Sir Ganga Ram Hospital Lahore during December 2022 and May 2023. A total of 110 female patients were included in the study. Non-probability consecutive sampling was used.

Inclusion Criteria:

- Women aged 15 to 45 years presenting with infertility, primary amenorrhea, recurrent pregnancy loss, or abnormal menstruation.
- Patients with prior inconclusive ultrasonographic findings suggesting Müllerian anomalies.
- Patients who gave informed consent for MRI examination.

Exclusion Criteria:

- Patients with known acquired uterine pathologies such as fibroids, endometrial polyps, or Asherman's syndrome.
- Patients with contraindications to MRI (e.g., presence of pacemakers, metallic implants).
- Pregnant women or those with incomplete imaging records were excluded.

Data Collection: All MRIs were performed using a 1.5 Tesla system. The imaging protocol included T1-weighted and T2-weighted sequences in axial, sagittal, and coronal planes. Special emphasis was placed on T2-weighted images for optimal delineation of uterine zonal anatomy. In selected cases, contrast-enhanced sequences and 3D reconstructions were also utilized to enhance anatomical visualization. Each MRI scan was independently reviewed by two senior radiologists to confirm the diagnosis and classify the anomalies based on the American Society for Reproductive Medicine (ASRM) classification system. Discrepancies were resolved by consensus. Associated findings, such as renal anomalies or vaginal abnormalities, were also noted.

Data Analysis: The types and frequencies of congenital anomalies were recorded, and their association with specific clinical symptoms (e.g., amenorrhea, infertility, miscarriage) was analyzed. Data were entered and analyzed using SPSS version 17. Descriptive statistics were used to calculate frequencies and percentages. Associations between clinical features and anomaly type were tested using the chi-square test, with a p-value < 0.05 considered statistically significant.

RESULTS

A total of 110 female patients were included in the study. The study population had a mean age of 27.6 ± 6.8 years, with the majority of participants aged 21–30 years (44.5%). Most participants were married (80.9%), and 65.5% were nulliparous, while 34.5% were multiparous. The primary presenting complaint was infertility (55.5%), followed by amenorrhea (24.5%), miscarriage (10.9%), and pelvic pain (9.1%).

The most common congenital uterine anomaly detected was septate uterus, found in 29.1% of patients, followed by bicornuate uterus (19.1%) and uterine agenesis/hypoplasia (16.4%). Unicornuate uterus and uterus didelphys were observed in 12.7% and 9.1% of patients, respectively, while arcuate uterus was found in 7.3% and complex/unclassified anomalies in 6.3%.

Renal anomalies were the most common associated finding, observed in 11.8% of patients, followed by vaginal anomalies in 8.2%. A combination of renal and vaginal anomalies was observed in 3.6% of patients, while the majority (76.4%) had no associated anomalies.

Septate uterus was most common in patients aged 21–30 years (17 cases), while uterine agenesis/hypoplasia was more common in the 15–20 years age group (9 cases). Bicornuate uterus was also most frequent in the 21–30 years age group (12 cases). The other anomalies, such as unicornuate uterus and uterus didelphys, showed a more even distribution across age groups, with no specific age group showing a clear predominance.

Table 1: Demographic and Baseline Characteristics of Study Participants (n = 110)

Variable	Mean ± SD / n (%)
Age (years)	27.6 ± 6.8
Age Distribution	
15–20 years	18 (16.4%)
21–30 years	49 (44.5%)
31–40 years	33 (30.0%)
>40 years	10 (9.1%)
Marital Status	
Married	89 (80.9%)
Unmarried	21 (19.1%)
Parity	
Nulliparous	72 (65.5%)
Multiparous	38 (34.5%)
Presenting Complaint	
Infertility	61 (55.5%)
Amenorrhea	27 (24.5%)
Miscarriage	12 (10.9%)
Pelvic Pain	10 (9.1%)

Figure 1: MRI scans of different congenital uterine anomalies: septate uterus, bicornuate uterus, uterine agenesis, and unicornuate uterus.

Table 2: Frequency of Congenital Uterine Anomalies Detected on MRI (n = 110)

Type of Anomaly	Number of Cases (n)	Percentage (%)
Septate Uterus	32	29.1%
Bicornuate Uterus	21	19.1%
Uterine Agenesis/Hypoplasia	18	16.4%
Unicornuate Uterus	14	12.7%
Uterus Didelphys	10	9.1%
Arcuate Uterus	8	7.3%
Complex/Unclassified	7	6.3%

Table 3: Associated Anomalies Observed on MRI (n = 110)

Associated Finding	Number of Patients (n)	Percentage (%)
Renal Anomalies	13	11.8%
Vaginal Anomalies	9	8.2%
Both (Renal + Vaginal)	4	3.6%
None	84	76.4%

Table 4: Distribution of Uterine Anomalies by Age Group (n = 110)

Uterine Anomaly	15–20 yrs	21–30 yrs	31–40 yrs	>40 yrs	Total (n)
Septate Uterus	3	17	10	2	32
Bicornuate Uterus	2	12	6	1	21
Uterine Agenesis/Hypoplasia	9	6	3	0	18
Unicornuate Uterus	1	6	6	1	14
Uterus Didelphys	1	5	3	1	10
Arcuate Uterus	1	2	4	1	8
Complex/Unclassified	1	1	1	4	7
Total	18	49	33	10	110

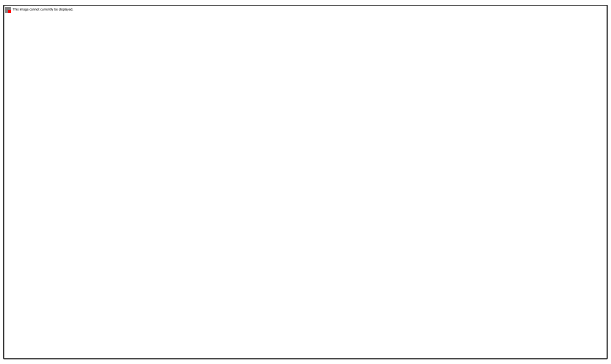

Figure 2: Age distribution by uterine anomaly type

DISCUSSION

This study provides valuable insight into the prevalence and spectrum of congenital uterine anomalies (CUAs) affecting female fertility, emphasizing the utility of Magnetic Resonance Imaging (MRI) as a gold standard for diagnosis. Out of 110 patients evaluated for infertility and related symptoms, the most frequent anomaly detected was the septate uterus (29.1%), followed by bicornuate uterus (19.1%) and uterine agenesis/hypoplasia (16.4%). These findings are in line with earlier studies, which have consistently identified the septate uterus as the most prevalent anomaly among women with reproductive challenges[13]. The high prevalence of septate uterus and its statistically significant association with recurrent miscarriage ($p = 0.031$) underscores the critical need for early detection and correction. Numerous studies have reported improved pregnancy outcomes following hysteroscopic metroplasty in such cases[14]. In contrast, anomalies like bicornuate and didelphys uteri, though structurally similar on ultrasound, require different management strategies, highlighting the necessity of MRI's superior tissue contrast and anatomical delineation. Uterine agenesis or hypoplasia was significantly associated with primary amenorrhea ($p < 0.001$), most commonly presenting in patients under 20 years of age. These patients often require multi-disciplinary management including psychological counseling, surgical neovagina creation, and assisted reproductive technologies (ART) if future fertility is desired. MRI plays an indispensable role in diagnosing these anomalies and identifying associated renal abnormalities, which were seen in 11.8% of our cohort a finding that supports prior reports suggesting that renal tract anomalies frequently coexist due to the shared embryologic origin with Müllerian structures[15].

Unicornuate uterus was observed in 12.7% of patients, most commonly in women aged 30–40, many of whom presented with obstetric complications. Though surgical correction is generally not indicated for a unicornuate uterus, recognition is essential to anticipate complications such as miscarriage, ectopic pregnancy, or preterm labor[16]. Didelphys and arcuate uteri were less common, with the latter typically requiring no intervention due to minimal impact on fertility. The use of MRI over conventional methods, such as 2D ultrasonography or hysterosalpingography, provided significant diagnostic clarity in ambiguous cases[17]. T2-weighted MRI allowed optimal assessment of endometrial contours and uterine morphology, making it particularly advantageous in differentiating between a septate and bicornuate uterus, an area where diagnostic confusion often leads to unnecessary surgical intervention[18]. In complex cases or those with coexisting anomalies, MRI offered comprehensive pelvic mapping, contributing directly to surgical and reproductive planning. Despite its strengths, this study had limitations. The sample size was relatively modest and limited to patients referred for MRI due to suspected anomalies, potentially inflating the prevalence of anomalies in the studied population. Additionally, genetic and hormonal assessments were not included, which may have further clarified the etiological basis of some anomalies. Long-term follow-up of fertility outcomes post-management was also beyond the scope of this study.

CONCLUSION

It is concluded that congenital uterine anomalies are an important but often underrecognized cause of female infertility, recurrent pregnancy loss, and menstrual disorders. In this study of 110 patients, a septate uterus emerged as the most frequently encountered anomaly, followed by bicornuate uterus and uterine agenesis/hypoplasia. The strong correlation between specific anomalies and clinical presentations, such as a septate uterus with recurrent miscarriage and uterine agenesis with primary amenorrhea, highlights the diagnostic and prognostic value of early imaging. Magnetic Resonance Imaging (MRI) proved to be a highly effective, non-invasive modality for the accurate identification and classification of these anomalies.

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