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Original Research Article

Mullerian anomalies: from diagnosis to intervention

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ABSTRACT

Background: The aim of the study was to observe prevalence of Mullerian anomalies, their clinical features, various investigation modalities to diagnose anomalies, various treatment modalities to correct anomalies and outcome after correction.
Methods: A prospective study of congenital uterine anomalies and its outcome was performed by using data from women with congenital anomalies presented in OPD at tertiary care hospital. Total 25 women with various congenital anomalies included in this study. In these cases, diagnosis was made on clinical examinations and confirmed by diagnostic modalities like USG, MRI, laparoscopy and hysteroscopy. And then a plan of management was decided, and surgery was planned. And then follow up for 1 year.
Results: In this study, most common anomaly seen was septate uterus followed by Mullerian agenesis. Most common presenting symptom was infertility (primary and secondary) followed by primary amenorrhea.
Conclusions: Mullerian anomalies present themselves in diverse forms. Mullerian anomalies may be difficult to diagnose. For planning treatment and management strategies, establishing an accurate diagnosis is mandatory. The surgical approach for correction of utero vaginal anomalies is tailored to the type of malformation.

Keywords: Amenorrhea, Congenital mullerian anomalies, Infertility, MRI, Surgery, USG

INTRODUCTION

Developmental anomalies of the Müllerian duct system are among the most intriguing disorders faced by obstetricians and gynaecologists.[1] These abnormalities typically arise from errors in organogenesis, although other causes such as deficiencies in steroidogenesis, receptor defects, and genetic abnormalities can also play a role.[2] The Müllerian ducts are essential embryonic structures that develop into key components of the female reproductive system, including the fallopian tubes, uterus, uterine cervix, and the upper part of the vagina.[1]

Müllerian malformations are developmental anomalies that arise from the paramesonephric ducts and are characterized by failures in the fusion of these structures along the midline, where they connect to the urogenital sinus. These malformations result from disruptions in the formation of the upper vaginal and uterine lumens, as well as the incomplete absorption of the septum during duct fusion. The severity of these anomalies can vary widely, ranging from mild forms that are often asymptomatic but may cause significant obstetrical issues, to severe cases such as vaginal and uterine agenesis, including Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome, or vaginal obstructions caused by agenesis or a septum.[3]

The overall prevalence of congenital uterine anomalies is 5.5% in the general population, 8.0% in infertile women, 13.3% in those with a history of miscarriage, and 24.5% in women with both miscarriage and infertility.[4]

Müllerian malformations are often linked with renal abnormalities, such as unilateral renal agenesis, pelvic kidney, hydroureter, and hydronephrosis. These malformations may also be associated with skeletal abnormalities, particularly involving the spine, as well as anorectal abnormalities, cardiac anomalies, and other conditions.[5]

Diagnosing Müllerian anomalies can be challenging due to the wide variability in clinical presentations. While laparoscopy combined with hysteroscopy has been the gold standard for diagnosis, less invasive imaging techniques like ultrasonography, hysterosalpingography (HSG), sonohysterography, and magnetic resonance imaging (MRI) are commonly used for screening, diagnosing, and classifying congenital uterine anomalies.[6] Treatment options are diverse and are typically customized to address the specific utero-vaginal anomaly present.[7]

The study aimed to observe the prevalence of congenital uterine anomalies in our population, their clinical implications, impact on fertility, study various investigation modalities to diagnose anomalies, various surgical treatments to correct anomalies and after treatment their outcomes.

METHODS

A prospective study was performed at our institute to observe the congenital anomalies of the female genital tract in our population. The study period was from March 2023 to March 2024. A total of 25 women with different types of congenital anomalies were detected. Women with congenital anomalies presented with chief complaints of primary amenorrhea, secondary amenorrhea, primary infertility, secondary infertility, cyclical abdominal pain, dysmenorrhea and recurrent pregnancy loss were included in our study. In these cases, diagnosis was made on clinical examinations and confirmed by diagnostic modalities like USG, MRI, Laparoscopy and Hysteroscopy. And then a plan of management was decided, and surgery was planned. And then follow up for 1 year.

Statistical analysis

The data for all parameters were collected, tabulated and frequency and percentage were analysed.

RESULTS

The majority of cases of mullerian anomalies were found in the 20-25 years age group (40%) followed by the 26-30 years age group (33%), which is of the reproductive age group And only 16% of Mullerian anomalies were found in the adolescent age group. Because in reproductive age patients came to OPD with chief complaints of being unable to conceive, recurrent pregnancy loss or wanting to consummate; so that majority of patients are of reproductive age group (Table 1).

Table 1: Age group distribution (n=25).

Age (in years)	No. of patients	Percentage
10-15	3	12
16-20	1	4
20-25	10	40
26-30	8	32
31-35	2	8
36-40	0	0
41-45	1	4

Presenting symptoms consisted of infertility seen in 48% of cases in which 24% of patients were of primary infertility and 24% of secondary infertility while primary amenorrhea seen in 36% of cases and secondary amenorrhea in 4% of cases. 8% patients presented with recurrent pregnancy loss and similar number of cases presented with dysmenorrhea (Table 2).

Table 2: Chief complaints (n=25).

Chief complaints		No. of patients	Percentage
Amenorrhea	Primary amenorrhea	8	32
	Secondary amenorrhea	1	4
Infertility	Primary infertility	6	24
	Secondary infertility	6	24
Recurrent pregnancy loss		2	8
Dysmenorrhea		2	8

Table 3: Investigation modalities (n=25).

Modalities	No. of patients
Ultrasonography	8
Hysterosalpingography	3
MRI	4
Laparoscopy	5
Hysteroscopy	5

In our study, ultrasonography is the main modality used to diagnose Mullerian anomalies followed by laparoscopy and hysteroscopy. MRI was done in 4 patients, in which 3 patients were of Mullerian agenesis who presented to our OPD with primary amenorrhea and in one patient of didelphys uterus with a septum. laparoscopy was carried out on 5 patients, in which 2 patients were of unicornuate uterus with rudimentary horn and presented with dysmenorrhea and 2 patients were of Mullerian agenesis who presented with primary amenorrhea and we had clinical suspicion plus to confirm USG findings; 1 patient was of bicornuate uterus in which laparoscopy was performed who presented with infertility followed by laparotomy done and metroplasty done. Hysteroscopy was performed in 5 patients who presented with infertility and

a septate uterus was found in them which was removed with hysteroscopic scissors. HSG was done in 3 patients who presented with infertility and recurrent pregnancy loss to know the cause of pregnancy loss (Table 3).

It was found in our study that the septate uterus (52%) is the most common uterine anomaly followed by Mullerian agenesis (24%). In Mullerian agenesis 16% cases of uterine agenesis, 8% cases of cervical agenesis and in all cases of mullerian agenesis vaginal agenesis was present. While unicornuate uterus with rudimentary horn present in 8% of cases and didelphys and bicornuate uterus in 4% of cases. Vaginal septum was present in 8% of cases (Table 4).

Table 4: Anomalies (n=25).

	Diagnosis	No. of patients	Percentage
Class 1	Mullerian agenesis	6	24
	Uterine agenesis	4	16
	Cervical agenesis	2	8
	Vaginal agenesis	6	24
Class 2/U4	Unicornuate uterus with rudimentary horn (non-communicating)	2	8
Class 3	Didelphys uterus with oblique vaginal septum	1	4
Class 4/U3C0	Bicornuate uterus	1	4
Class 5/U2	Septate uterus	13	52
Class 6	Arcuate uterus	0	0
Class 7	T shapes uterus	0	0
V3	Vaginal septum	2	8

Table 5: Management.

Surgery		No. of patients	Percentage
Vaginoplasty		6	24
	McIndoe vaginoplasty	4	16
	Pull through vaginoplasty	2	8
Rudimentary horn removal		2	8
Strassmann metroplasty		1	4
Hysteroscopy f/b septal resection		13	52
Vaginal septum resection		3	12

Table 6: Outcomes of surgical treatment (n=25).

	Surgical procedure	Outcome
Mullerian agenesis	Mcindoe vaginoplasty	3 (75%) patients had no coital difficulty
		1 (25) patient's vaginal contracture occurred
	Pull through vaginoplasty	Both patients started menstruating
Unicornuate uterus with rudimentary horn	Laparoscopic rudimentary horn removal	Both patient's pain was relieved
Didelphys uterus with oblique vaginal septum	Vaginal septum resection	Patients have regular menstruation, and their pain was relieved
Bicornuate uterus	Stassmann metroplasty	Patient conceived
Septate uterus	Hysteroscopic septal resection	8 (61%) patients conceived
		5 (38%) patients were not conceived
Transverse vaginal septum	Vaginal septum resection	Both patients started menstruating

Vaginoplasty was done in 24% of patients who presented with vaginal agenesis. McIndoe vaginoplasty was done in 16% of cases and pull-through vaginoplasty was done in 8% of cases in whom cervical agenesis was present. For patients who presented with dysmenorrhea due to a unicornuate uterus with a rudimentary horn (non-communicating), rudimentary horn removal was done. In patients presenting with infertility due to the Septate uterus, resection of the septum was done with the help of hysteroscopy and due to a bicornuate uterus metroplasty was performed, and unification of the uterus was done. And in one case of didelphys uterus with oblique vaginal septum who presented with secondary amenorrhea; resection of septum was done vaginally (Table 5).

After 1 year of follow-up; 4 patients who were operated for vaginal agenesis 3 patients has no coital difficulty and their space created was adequate and 1 patient had contracture of space created due to not following advice correctly. And 2 patients who were operated for cervical agenesis plus vaginal agenesis, both patients started menstruating regularly. Patients presented with dysmenorrhea due to a unicornuate uterus with a rudimentary horn, laparoscopic resection of the rudimentary horn was done and on follow-up patient has no pain during menses. In a patient with bicornuate who presented with secondary infertility in whom Strassmann metroplasty performed, on follow up she was pregnant with 4 months of amenorrhea. And 13 patients with septate uterus in whom hysteroscopic resection of septum was done 7 patients became pregnant and even 1 had full-term normal delivery but 5 patients did not conceive. And in 3 patients of vaginal septum, they have normal menstrual cycle after septum resection (Table 6).

DISCUSSION

The various Müllerian anomalies present themselves in diverse forms and different phases of a woman's life.[8] A careful anamnesis is always the first step, and a thorough clinical exam plays a fundamental role in any decision about the best diagnostic methods and choice of the best treatment.[3]

In our study, the majority of patients who presented with mullerian anomaly were mainly seen in the reproductive age group; 20-30 years who wanted to conceive and consummate. In our study mean age was 24.9 years who presented with mullerian anomalies which was comparable to study done by Mailing Hua Et Al12 who reported mean maternal age of patients around 29 years.[9]

In our study, mainly patients presented with infertility, amenorrhea, recurrent pregnancy loss or dysmenorrhea. Some patients presented with multiple symptoms. 36% of patients presented with amenorrhea and in those 32% patients had primary amenorrhea. In our study, mullerian agenesis was the most common cause of primary amenorrhea. Which was compatible with Tanmahasamut et al (39%).[10] In our study 48% of patients presented with infertility (primary and secondary); which is mainly due to septate uterus (91%) this is in contrast to the study by Raga et al in that the most common cause of infertility was bicornuate uterus (40%).[11] 8% of patients presented with recurrent pregnancy loss, and in them septate uterus was found during hysteroscopy. So in our study septate uterus is main cause of RPL in contrast to Salim et al in that study common cause of recurrent pregnancy loss was the arcuate uterus followed by the septate uterus.[12] 8% patients also presented with dysmenorrhea. During their evaluation unicornuate uterus with rudimentary horn found in laparoscopy.

In our study, USG is the main modality for diagnosing congenital uterine anomalies. USG is a reliable, easily available, noninvasive and inexpensive modality to diagnose uterine anomalies. However, MRI is the most sensitive imaging modality.[4] Sometimes, Mullerian anomalies are incidentally diagnosed during laparoscopy and hysteroscopy during work-up of infertility or amenorrhea.

In our study, septate uterus (52%) is the most common uterine anomaly found followed by Mullerian agenesis (24%) which is compatible with Raga et al in which the most frequent mullerian anomalies were the septate uterus.[9] After diagnosis surgery was scheduled according to anomalies. In complete Mullerian agenesis, Mcindoe vaginoplasty was done (16%) and on follow-up 3 out of 4 patients had no difficulty in coitus. And in cervical agenesis, pull-through vaginoplasty and drainage of hematometra was done (8%) and on follow-up, both patients had regular menstruation and pain was also relieved. Patients who presented with dysmenorrhea, during laparoscopy unicornuate uterus with rudimentary horn (without communication) were found and resection of the rudimentary horn was done. During follow-up pain was relieved. And patients who presented with infertility and recurrent pregnancy loss, the main cause was found to be a septate uterus; which was then removed during hysteroscopy and on follow up 62% of patients was already conceived within 1 year. And in 1 case of bicornuate uterus which was diagnosed during laparoscopy, the procedure was converted to laparotomy and Strassmann metroplasty done. On follow up patient was pregnant with 4 months of amenorrhea. In one patient who presented with secondary amenorrhea MRI was done and the report was suggestive of a didelphys uterus with vaginal septum which was then removed and on follow up patient had regular menses. And lastly, for patients who presented with primary amenorrhea, the transverse vaginal septum was found and then resection of the septum was done patients had normal menstrual cycles during 1 year of follow-up.

This study was conducted at a single centre over short period of time hence the findings of this study cannot be generalised to population. This was the limitation of this study.

CONCLUSION

Müllerian anomalies present themselves in diverse forms and different phases of a woman's life. Because of the wide variation in clinical presentation, Mullerian anomalies may be difficult to diagnose. Clinical suspicion should be there if early diagnosis is not to be missed. For planning treatment and management strategies, establishing an accurate diagnosis is mandatory. The surgical approach for correction of utero vaginal anomalies is tailored to the type of malformation. For most surgical procedures, procedure's value is the patient's postoperative ability to have healthy sexual relation and achieve successful reproductive outcomes.

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