ORIGINAL RESEARCH

MULLERIAN DUCT ANOMALY: SPECTRUM OF PRESENTATION AND MANAGEMENT IN A TERTIARY CARE CENTER

[1]Dr.Akrati Jain, [2]Dr.Anjali Kanhere, [3]Dr. Preeti Singh, [4]Dr. Sonal Jain

[1]Assistant Professor, Obstetrics and Gynaecology
[2]Professor and Head of department, Obstetrics and Gynaecology
[3]Senior Resident, Obstetrics and Gynaecology
[4]Associate Professor, Obstetrics and Gynaecology
Chirayu Medical College and Hospital (CMCH), Bhopal

Corresponding author
Dr.Akrati Jain
Assistant Professor, Obstetrics and Gynaecology

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ABSTRACT

Background: Mullerian developmental defects are congenital anomalies of the female reproductive tract that may adversely affect a woman's obstetric and gynaecological health. Uterine anomalies arise if there is agenesis of one or both mullerian ducts or absence of fusion or reabsorption of the septum between these ducts. The defect may be partial or complete and may affect one or multiple parts of the tract.

AIM: This study was done to assess the spectrum of clinical presentation of mullerian duct anomalies and it's reproductive outcome in a tertiary care centre. The role of imaging MRI , diagnostic hysterolaparoscopy, treatment options, surgical interventions, effect of mullerian malformations on reproductive health and fertility was reviewed.

Material and methods: The present study is a retrospective study including all cases diagnosed to have mullerian duct anomalies. Cases were worked-up and investigated to reach a final diagnosis and case-based management was done. All cases were assessed in terms of clinical presentation, classification, impact on reproductive outcome and managed accordingly.

Results: In our study period, we managed total of 49 cases with different mullerian anomalies. The main presenting symptoms were cyclical pain abdomen, primary amenorrhoea, infertility and recurrent pregnancy loss. Most of the patients belonged to the young age. With optimum surgical management, we reported good patient outcome for all the cases.

Conclusion: Cases of mullerian anomalies should be dealt with empathy, proper work-up and case-based management lead to a good patient outcome and improved quality of life.

Keywords: mullerian duct; uterine anomalies; diagnostic hysterolaparoscopy

INTRODUCTION

Mullerian duct anomaly (MDA)encompasses a diverse spectrum of congenital reproductive tract anomalies. The prevalence ranges from 0.001 to 10% in the general population and 8-10% in those with adverse reproductive history[1,2]. While it can pose challenges in terms of fertility and pregnancy outcomes, advances in diagnostic techniques and treatment options have provided individuals with MDA anincreased opportunity to achieve their reproductive goals. A multidisciplinary approach involving gynecologists, reproductive endocrinologists, and mental health professionals is crucial to ensure comprehensive care and support for individuals with Mullerian duct anomalies. Mullerian(Parames one phric duct) anomaliesoccur due to a variety of embryological disruptions during its development. This might be the result of total agenesis, defective vertical or lateral fusion, or resorption failure. A unicornuate uterus and uterine agenesis/hypoplasia is the outcome of organogenesis failure. The uterus is formed by the ducts joining together during fusion. If this fails, it develops as a bicornuate or didelphys uterus. Septal resorption involves resorption of the central septum after the ducts have fused. Failure in this stage result in a septate or arcuate uterus. Mullerian Duct Anomalies are associated with renal anomalies (25%), skeletal

anomalies (12%),and Gastrointestinal tract anomalies (12%) due to their close association during their development[3,4]. The clinical presentation of MDA can vary widely with symptoms ranging fromprimary amenorrhoea, infertility, recurrent pregnancy loss, and abnormal uterine bleeding.Among the cases of Mullerian duct anomalies, a few present to the emergency as acute abdominal pain or urinary retention whereas a majority presented with chronic complaints related to menstruation and fertility. Whenclinicallysuspected,otherinvestigationslikeUltrasonography,Hysterosalpingography(HSG),MRI& diagnostic hysterolaparoscopy are required to confirm the diagnosis. -based retrospective observational analysis conducted in the Obstetrics and Gynaecology Department of Chirayu medical,

AIMS AND OBJECTIVES

1) Study the various clinical presentations of mullerian malformations with associated congenital anomalies.
2) Study the clinical impact of mullerian malformation on the reproductive outcome of the women.
3) Optimize of management in different diagnosed cases.

MATERIALS AND METHODS

The present study is a hospital-based retrospective observational analysis conducted in the Obstetrics and Gynaecology Department of Chirayu medical college and Hospital, Bhopal a tertiary care center from June 2019 to June 2023

Inclusion criteria :

1) All the patients diagnosed with mullerian anomaly (based on history,

clinical examination, radiological imaging, and diagnostic

hysterolaparoscopy)presented in our institute were included in the study

Exclusion criteria:

1)Patients presenting with primary amenorrhea but not due to mullerian duct

anomaly.

RESULTS AND OBSERVATIONS

During our study period, about 49 patients were found to have mullerian ductanomalies.

In our study, 12 patients (24.4%) were between the ages of 10 and 20. A total of 26 patients (53%) were between the ages of 21 and 30; 9patients (18.3%) were between the ages of 31 and 40; and just 2 patients (4.08%), wereover 40. The earliest presentation was at 14 years of age and the latest was at 42 years,thedescription is shown in

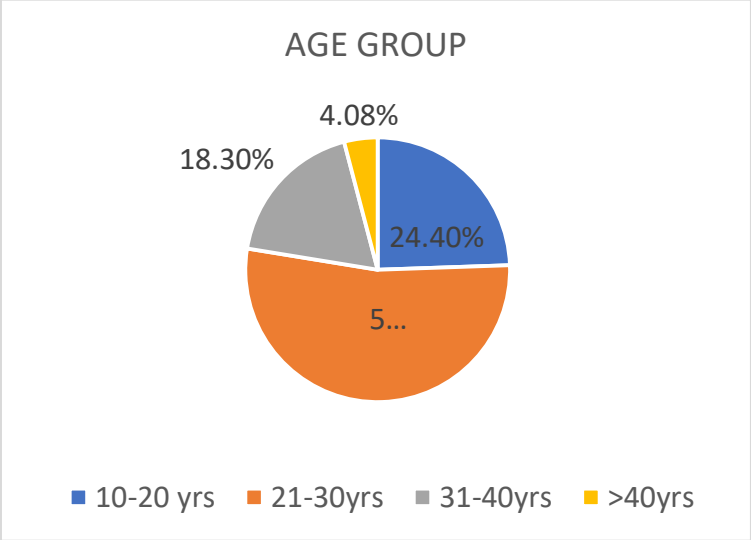

FIGURE 1: AGE DISTRIBUTION

The primary complaint ofpresentation leading to a diagnosis of mullerianductanomaly was evaluated. 19 individuals had primary amenorrhoea without any concomitant pain, and 5 patientshad abdominal pain associated with primary amenorrhoea. 20 of our patients presented with infertility, and 3 patients had a history of recurrent pregnancy loss. Out of the 49, 2 patients in our study had abnormal uterine bleeding (figure2). It is significant to highlight that individuals with primary amenorrhoea and cyclical abdominal pain appearedat a younger age in our study, whereas patients with fertility issues, recurrent pregnancy loss, or abnormal uterine bleeding presented at a later age. options have provided individuals with MDA anincreased opportunity to achieve their reproductive goals. A multidisciplinary approach involving gynecologists, reproductive endocrinologists, and mental health professionals is crucial to ensure comprehensive care and support for individuals with Mullerian duct anomalies. Mullerian(Parames one phric duct) anomaliesoccur due to a variety of embryological disruptions during its development. This might be the result of total agenesis, defective vertical or lateral fusion, or resorption failure. A

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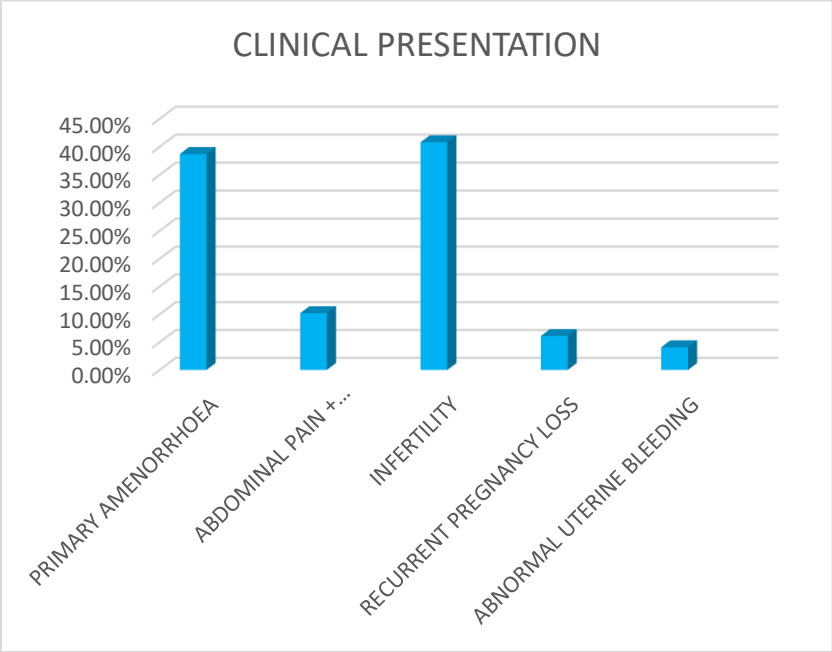

FIGURE 2 : CLINICAL PRESENTATION

12(24.4%) of the 49 patients had uterine agenesis, and 7(14.2%) had hypoplastic uterus.While 3 (6.1%) individuals with unicornuate uteri were detected when they were evaluated for management ofprimary infertility. 2 (4.08%) patients had didelphyicuterusin whom one patient presented with primary infertility, and the second patientpresented with pregnancy in one of the horn of the didelphic uterus.4(8.1%) patients presented with bicornuate uterusin whom 2 had primary amenorrhea and abdominal pain, while the other two had abnormal uterine bleeding. Maximum patients 17 (34.6%) in our research had a septate uterus and were diagnosed with infertility and recurrent pregnancy loss. Only 1(2.04%) patienthad an arcuate uterus, which was diagnosed onhysterolaparascopy during her management for infertility.3(6.1%) of our patients experienced cryptomenorrhea and abdominal pain, 2 patients had transverse vaginal septum with secondary vaginal atresia, and one had a hypoplastic cervix with agenesis of the upper 2/3 of the vagina (table 1).

Multiple classification systems for MDA exist and have been updated with different criteria, thus recognizing the features which guide clinical management is essential. In our study, we classified the patients into different classes based on the classification system proposed by the American fertility society for uterovaginal anomalies.

TABLE 1: DISTRIBUTION OF PATIENTS BASED ON TYPE OF MULLERIAN ANOMALY

TYPE OF MULLERIAN ANOMALY	NO. OF PATIENTS	PERCENTAGE
AGENESIS OF UTERUS	12	24.4%
HYPOPLASTIC UTERUS	7	14.2%
UNICORNUATE	3	6.1%
DIDELPHYS	2	4.08%
BICORNUATE	4	8.1%
SEPTATE UTERUS	17	34.6%
ARCUATE UTERUS	1	2.04%
CERVICAL HYPOPLASIA WITH VAGINAL AGENESIS	3	6.1%

TABLE 2: MANAGEMENT AND FOLLOW-UP

TYPE OF ANOMALY	NUMBER OF PATIENTS	TREATMENT
MRKH 1 SYNDROME	7	VAGINOPLASTY DONE AND COUNSELLED FOR SURROGACY/ADOPTION

	4	REFUSED SURGERY,SO NON -SURGICAL METHOD INVOLVING GRADUAL DILATATION OF VAGINAL DIMPLE WAS ADVISED
	6	PRESENTED AT YOUNG AGE,COUNSELLED FOR VAGINOPLASTY AT LATER AGE (BEFORE MARRIAGE)
MRKH 2 SYNDROME	1	VAGINOPLASTY DONE AND COUNSELLED FOR SURROGACY/ADOPTION
	3	PRESENTED AT YOUNG AGE,COUNSELLED FOR VAGINOPLASTY AT LATER AGE (BEFORE MARRIAGE)
VAN WYK GRUMBACH SYNDROME WITH BICORNUATE UTERUS	1	MEDICAL MANAGEMENT FOR JUVENILE HYPOTHYROIDISM WAS GIVEN FOLLOWED BY SURGICAL DRAINAGE OF HAEMATOMETRA AND HAEMATOCOLPOS
UNICORNUATE UTERUS	3	INFERTILITY MANAGEMENT
BICORNUATE UTERUS	2	HYSTERECTOMY FOR AUB
	1	SURGICAL MANAGEMENT OF HAEMATOCOLPOS,HAEMATOMETRA AND HAEMATOSALPINX
DIDELPHYIC UTERUS	1	INFERTILITY MANAGEMENT
DIDELPHYS UTERUS WITH PREGNANCY	1	ANC,HAD A FULL-TERM VAGINAL DELIVERY.
SEPTATE UTERUS	17	HYSTEROSCOPIC SEPTAL RESECTION
ARCUATE	1	MANAGED FOR INFERTILITY
CERVICAL HYPOPLASIA WITH VAGINAL AGENESIS	3	DRAINAGE OF HEMATOMETRA WITH CERVICOPLASTY AND VAGINAL CANAL EXTERNALIZATION

In our study, there were 19 patients with MRKH syndrome, out of which 15 had MRKH1 while4 patients were MRKH2. A total of6 patients underwent McIndoe's vaginoplasty with split skin graft with counselling of serial vaginal dilatation, remaining 2 refused surgical management, and 3 very youngpatientswerecounselledfor vaginoplasty at a later age (table2).

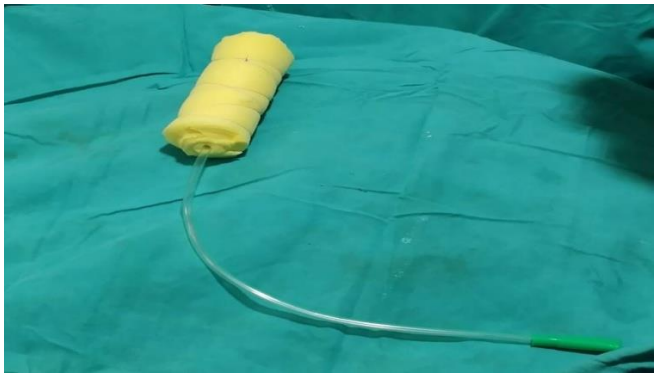

IMAGE 1: AUTOCLAVED VAGINAL MOULD USED FOR VAGINOPLASTY

A spectrum of clinical courses of atypical case presentation and management is discussed here. A 22-year-old, nulligravida presented with primary amenorrhoeawith pain in the inguinal region.MRI suggestive of the uterus,cervix, and upper 2/3rd of vagina absent suggestive of congenital aplasia, left ovary seeninthe pelvic cavity, right ovaryseen just below the anterior abdominal wall in the inguinal region suggestive of ovarian inguinal hernia(canal of nuch hernia) withcrossfused ectopic leftkidney, diagnosed as Type 2 MRKH with an ovarian hernia. She underwent right inguinal mesh hernioplasty and vaginoplasty, vaginal reconstruction with skin grafting, and was discharged well. A 16-year-old girl presented with long-standing untreated hypothyroidismandpainabdomen.Shewas diagnosedto

be a rare case of Von Wyk Grumbach Syndrome with mullerian duct anomaly.On evaluation, MRI showed a bicornuate uterus withhematometraandhaematosalpinx of the left side with a normal right uterine cavity and cervix with normal ovaries. Surgical management of haematocolposand haematometra was done. A case ofa14-year-old girl presented with abdominal distension, pain,andprimaryamenorrhoea.OnCT scanwas diagnosed to have a bicornuate uterus with haematocolpos, hematometra, and haematosalpinx of the right side. The left horn of the uterus appeared normal. The right renal fossa was empty.Diagnostic laparoscopywasdone, and a cruciate incision was taken at the point of maximum bulge, drainage of around 200ml of viscidbloodwas done, and eversion of the cruciate incision was done. The patient reported back with pain inabdomen after a month andonMRI was foundto have a bicornuate uterus with small residual hematometra in the right horn of the uterus andlargeaematosalpinx.Sheunderwentlaparotomyand removal of haematosalpinx and was discharged on cyclical progesterone pills. In another case, a 16-year-old girl presented with complaints of pain in the lower abdomen and primary amenorrhoea. On per abdomen examination, 16 weeks size mass was felt andon local examination, transverse vaginal septum was seen.On MRI large hematometra and right haematosalpinx was seen. Sheunderwent drainage of haematopyometra, and cervicoplasty with vaginal canal reconstruction, followed by dilatation of the vaginal canal. The patient was counselled and trained forfrequentvaginaldilatation on discharge.

One more patient with a similar diagnosis, a 22-year-old female married for3 years presented with secondary amenorrhoea and pain in the abdomen. She had a history of primary amenorrhoea with cryptomenorrhea at the age of 15 years for which she underwent canalization. She had irregular menses for a few years followed by prolonged secondary amenorrhoea with cyclical abdominal pain andunderwent laparotomy with drainage of left haematosalpinx, left endometriotic cystectomy, and cervicoplasty. One more such case,a14-year-old girl presented with a complaint of primary amenorrhea with cyclical abdominal pain. MRI was suggestive of hematometra with bilateral haematosalpinx.Thepatient was taken up for EUA(examination under anaesthesia)and on local examination was found to have a blind vaginal pouch,cervix was not appreciatedseparately(cervical agenesis) and ultrasound guided drainage of hematometra was done.Patient reported back with a complaint of pain in the abdomen after a month and was found to have a tense tender 14 weeks size mass on abdominal examination and imaging showed residual hematometra with bilateral haematosalpinx,so was taken up for drainage of the same.As drainage through the vaginal route was not possible due to very thick and fibrosed septum,decision for laparotomy was taken,bilateralhematosalpinx were removed with drainage of hematometra and intrauterine foley's catheter was put and patient was started on high dose hormonal pills.

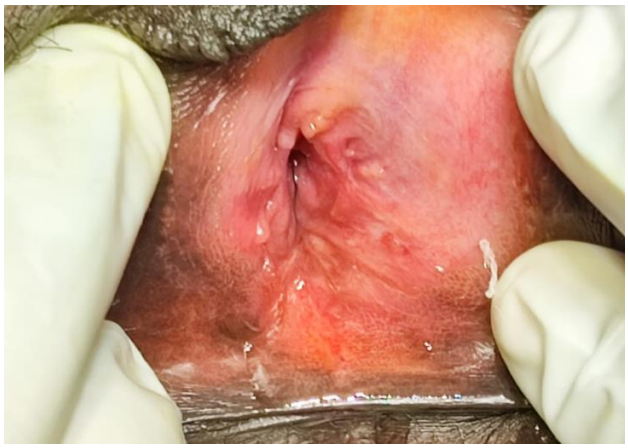

IMAGE 2: BLIND VAGINA WITH MALPOSITIONED URETHRAL OPENING

Management of patients with obstructive uterovaginal anomalies requires comprehensive evaluation and stabilization. Surgical management is dependent on the type of obstructive anomaly. All such patients have risk of stenosis and may require ongoing dilation or additional surgical intervention[5]. In our study three patients presented with infertility were diagnosed to have unicornuate uterus by HSG and diagnostic hysterolaparoscopy. progesterone pills. In another case, a 16-year-old girl presented with complaints of pain in the lower abdomen and primary amenorrhoea. On per abdomen examination, 16 weeks size mass was felt andon local examination, transverse vaginal septum was seen.On MRI large hematometra and right haematosalpinx was seen. Sheunderwent drainage of haematopyometra

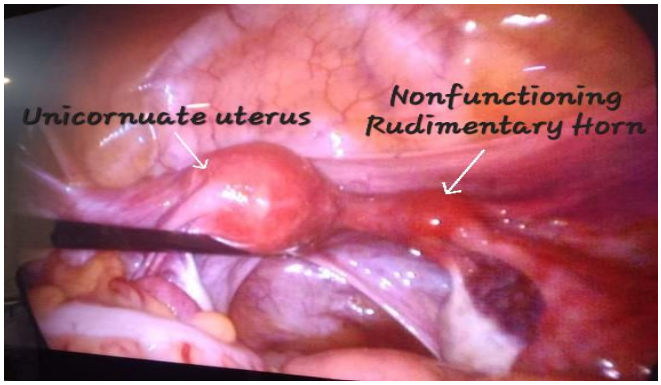

IMAGE 3: UNICORNUATE UTERUS WITH NON-FUNCTIONING RUDIMENTARY HORN

Two of the cases in our study presented at a later age of > 40 years with the complaint of abnormal uterine bleeding(AUB). Both of them were incidentlydiagnosed to have bicornuate uterus on imaging. Both underwent surgical management, one had Total Laparoscopic Hysterectomy and other abdominal hysterectomy.

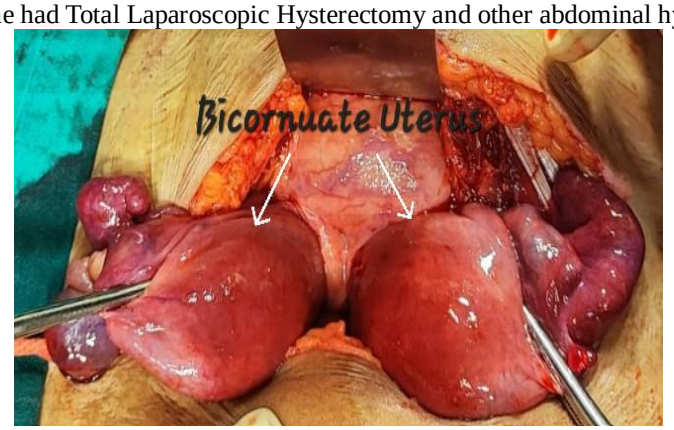

IMAGE 4: BICORNUATE UTERUS

Didelphys uterus was diagnosed in another patient, a 21-year-old primigravidawith spontaneous conception. She presented at 19 weeks gestation with foul-smelling vaginal discharge and underwent drainage for the same.She had an uneventful antenatal period with full-term vaginal delivery.This was one of the rare case of mullerian malformation,didelphic uterus with healthy intrauterine pregnancy in one horn and anechoic collection in the other horn with absent kidney on left side.

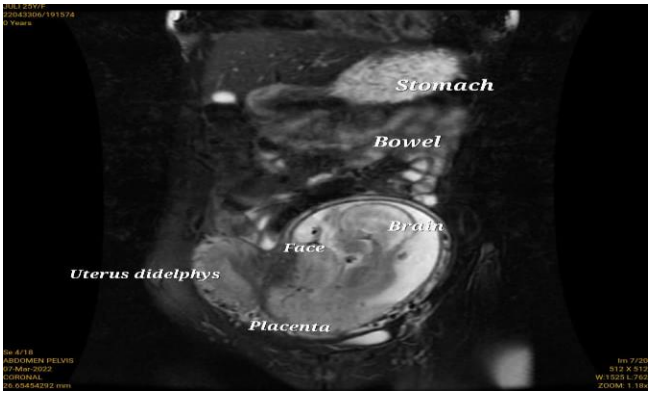

IMAGE5: MRI IMAGE OF PREGNANCY IN ONE HORN OF DIDELPHYIC UTERUS

A 35 year old patient with infertility was diagnosed with didelphic uterus and complete vaginal septum .

IMAGE 6: COMPLETE VAGINAL SEPTUM IN DIDELPHIC UTERUS

Majority of the patients who were diagnosed with septate uterus (17) presented with infertility and recurrent pregnancy loss. They were managed on hysterolaproscopy by septal resection and are under follow up for infertility.

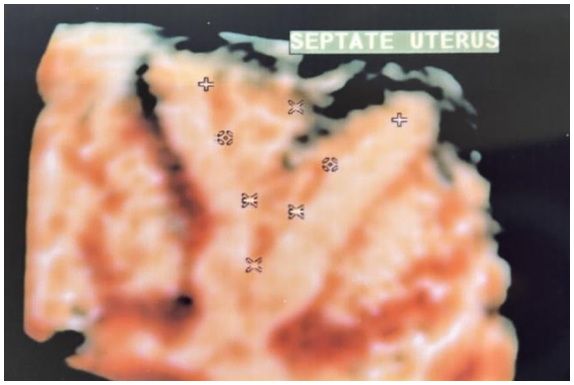

IMAGE 7: 3 -D SCAN OF SEPTATE UTERUS

DISCUSSION

Congenital abnormalities of mullerian ducts are relatively common, occurring in 7-10% of all women,and contributing to the problems of infertility,recurrent pregnancy loss, and poor pregnancy outcomes that occur in approximately 25% of women with uterine anomalies[6] In our study majority of patients presented in the age group of 21-30 years followed bythe adolescent group.In another notable study by Mane *et al.*, the mean age of presentation was typically at 17 years[7].Study by Reindollar *et al*., showed that the cases of mullerian abnormalities typically presented at adolescent age group[8] The patients in our study mostly presented with infertility 20(40.8%) followed by primary amenorrhoea 19(38.7%). In Banerjee et al study the patients mostly presented with chronic abdominal pain (37%), closely followed by primary amenorrhea (31.5%) and inability to conceive (31.5%)[9]. In the study done by Jeon *et al*., these again were the three most common presenting complaints with incidence rate of 10.8%, 12.4% and 8.6% respectively[10]. We found 19 patients with MRKH syndrome in our study. The treatment goal in these women is the creation of an artificial vagina, either conservatively or surgically, that allows sexual functioning.McIndoe's procedure refers to the surgical creation of an artificial vagina using a split skin graft or an amnion graft.In our study, we had also performed McIndoe vaginoplasty using autologous skin graft in 8patients . All patients had a successful recovery . Chaudhary *et al*.[11] conducted a study on eight patients with MRKH and performed McIndoe vaginoplasty using amnion grafts with similar results in all patients. Didelphys uterus is a very rare uterine anomaly. The data available on the reproductive outcome in these patients is mixed, owing to its rare occurrence. In our studydidelphys uterus was pregnant with singleton .Nohara *et al*.[12] reported a case of a woman with didelphys uteri that was pregnant with twins while Mashiach *et al*.[13] reported triplet pregnancy in a case with didelphys uterus. On the contrary, a retrospective study done by Zhang *et al*.[14] demonstrated that patients with didelphys uterus required infertility treatments more frequently than with other anomalies. Ghi*et al*.[15] documented that 40% of patients with septate uterus presented with recurrent abortions. A study conducted by Nouri *et al*.[16] documented a 60% pregnancy rate after hysteroscopic management of septate uterus . In our study 34.6% of patients presented with septate

uterus and managed accordingly. Xie *et al*.[17] conducted a retrospective study on 32 patients with CervicoVaginal Agenesis, wherein 84.3% had a successful outcome with utero-cervical canalization and vaginal reconstruction procedure while 15.6% of cases had to undergo a hysterectomy. While in our study we had 4 patients with obstructive cervicovagianl anomaly who were managed surgically.

CONCLUSION

The various mullerian anomalies present themselves in diverse forms and in different phases of a woman's life. A careful anamnesis is always the first step , a thorough clinical examination and preoperative evaluation of reproductive and pelvic anatomy is crucial for any decision about the best diagnostic method and treatment.A multidisciplinary team work, well counselled and documented optimal treatment pathway is required for better reproductive , psychological as well as social outcome.

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