



The Spectrum of Müllerian Anomalies Presented in a Tertiary Care Centre: Three-Year Experience

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Abstract

Objective: Developmental anomalies of female genital tract are not very common to be seen in Gynaecological practice, incidence reported in the literature is 5-7%. Various systems of classification had been proposed according to the site of involvement, each having its own merits and demerits. Here, we present our experience of Müllerian anomalies for three years in a tertiary care Centre and classified them as per the most recent classification proposed by the European Society of Human Reproduction and Embryology (ESHRE).

Material and methods: Present study is a Prospective Cohort Study including all cases diagnosed to have Müllerian duct anomalies. Cases were worked-up and investigated to reach a final diagnosis and classified according to CONUTA system and case-based management was done for each case. All cases were assessed in terms of the final outcome and are being followed-up.

Results: In the defined time, we managed total of 33 cases with different Müllerian anomalies. Main presenting symptoms were cyclical pain abdomen, primary amenorrhoea, infertility and recurrent miscarriage. Most of the patients belonged to the adolescent age group. With optimum surgical management, we reported good patient outcome for all the cases.

Conclusion: Cases of Müllerian anomalies should be dealt with empathy, as most of the patients are adolescents. Proper work-up and case-based management lead to a good patient outcome and improved quality of life.

Keywords: Müllerian; Müllerian ducts; Vaginal anomalies; Uterine anomalies

Introduction

Female genital tract develops from a pair of ducts called Müllerian or paramesonephric ducts [1]. Any deviations from normal development of these ducts result in congenital defects in the anatomy

of the female reproductive tract, which can lead to many health problems mainly with menstruation and reproduction. Different systems had been proposed time to time to classify these abnormalities [2], the most commonly used one is given by American fertility society (AFS) [3] which is being used to classify these defects for years. But it's difficult to classify the complex Müllerian duct anomalies [4-7], which has more than one site involved as per the American Fertility Society (AFS) classification. Hence, to overcome this difficulty, ESHRE has developed a new system of classification-Congenital uterine anomalies (CONUTA) [8] classification, which can classify even complex Müllerian anomalies.

Aims and Objectives

To assess various clinical presentations of Müllerian duct anomalies.

To classify various Müllerian duct anomalies according to CONUTA system.

To follow up these cases for work up, diagnosis, management and outcome.

Materials and Methods

The present study is a Prospective Cohort Study conducted in a tertiary care centre of Eastern India in between December 2015 to December 2018. During this period, patients who were diagnosed to have Müllerian duct anomalies were included in the study. Work-up of these cases including patient's presenting complaints, relevant clinical findings and detailed investigation like radiological assessment-Pelvic USG, MRI (if needed), S. FSH levels (other hormones as indicated), Karyotyping, IVP and X-ray lumbosacral spine (for associated anomalies) were done and based on the findings, final diagnosis was made. Cases were classified as per CONUTA SYSTEM. Cases were managed individually with optimum surgical management (Figures 1 and 2). The outcome was evaluated, and patients are being followed up till date.

Results

During the defined period, more than 6,000 patients visited the outpatient department for various gynaecological complaints. Out of them, 33 patients were found to have some Müllerian duct anomaly. The most common presentation was pain abdomen (cyclical) followed by primary amenorrhoea, inability to conceive and recurrent miscarriage.

Most of the patients belong to the adolescent age group, 36.36% in the age group of 18-21 years. However, it is important to note that patients with cyclical abdominal pain tend to present at an earlier age and patients with fertility issues tend to present at a later age (Table 1).

Age group	N	Percentage (%)
15-17	10	30.30%
18-21	12	36.36%
22-25	9	27.27%

>25	2	6.89%
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Table 1: Distribution of cases according to age (n=33).

33.33% of patients presented primarily with pain abdomen (associated with amenorrhoea in eight out of 11 cases, 70%). 27.27% of patients presented with primary amenorrhoea without any associated pain. 21.21% of patients presented with infertility and an almost equal number with recurrent miscarriage (Table 2).

Chief complaint	No. of cases (n=33)	Percentage (%)
Pain abdomen ± amenorrhoea	11	33.33%
Primary amenorrhoea	9	27.27%
Infertility	7	21.21%
Recurrent miscarriage	6	18.18%

Table 2: Distribution of cases as per clinical presentation (n=33).

All the 9 patients presented with primary amenorrhoea without any pain abdomen, diagnosed as MRKH syndrome (Table 3).

Diagnosis	Number of cases	Management	Outcome
MRKH syndrome	9	Mcindoe vaginoplasty	Patent vagina with normal sexual life

Table 3: Details of patients presented with primary amenorrhoea.

Among 11 patients presented with abdominal pain with or without menarche attained. Only one patient had imperforate hymen for which hymenectomy was done, one with a transverse vaginal septum which was excised and three with the longitudinal vaginal septum and uterus didelphys, where septum was excised, all three patients resumed menses after surgery. Three patient had cervico-vaginal agenesis along with uterine malformation; all three of them underwent cervico-vagino-plasty with a positive outcome. Out of four patients with unicornuate uterus with rudimentary horn, excision was needed in two cases in whom it was functional and presented with hematometra, one of them had huge hematosalpinx as well, which was also excised. One patient of cervico-vaginal agenesis was diagnosed to have a large ventricular septal defect (VSD) with the bi-directional flow, i.e. Eisenmenger's syndrome. As the pregnancy is posing a threat to life in these patients, hence, the decision of hysterectomy was taken by family, and the patient was counselled to get vaginoplasty done later when planning to start sexual activity (Tables 4 and 5).

Diagnosis	Number of cases	Management	Outcome
Imperforate hymen	1	Hymenectomy	Resumed menses
Transverse vaginal septum (TVS)	1	Vaginal septum excision	Resumed menses
Upper vaginal atresia	1	Vaginoplasty	Resumed menses
Cervico-vaginal agenesis	1	Cervico-vaginoplasty	Resumed menses

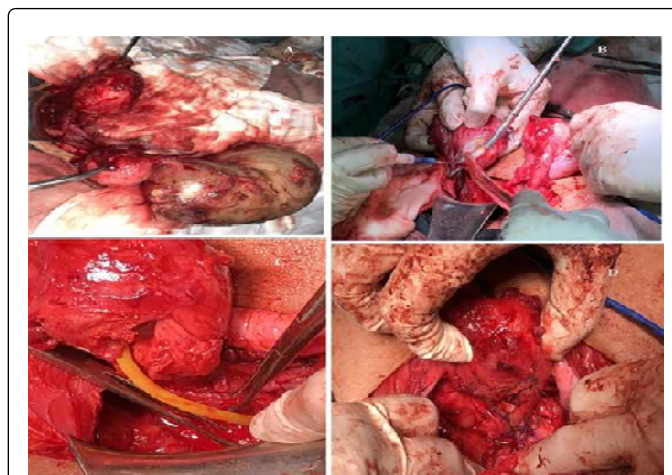


Figure 1: A. cervical agenesis with rudimentary horn with hematosalpinx. B. Passing a Hegar's dilator through uterine fundus and creating two flaps at lower margin of uterus. C. Passing a Foley catheter from newly created cervix to vagina. D. Final picture after suturing uterine and vaginal ends and closing uterine incision

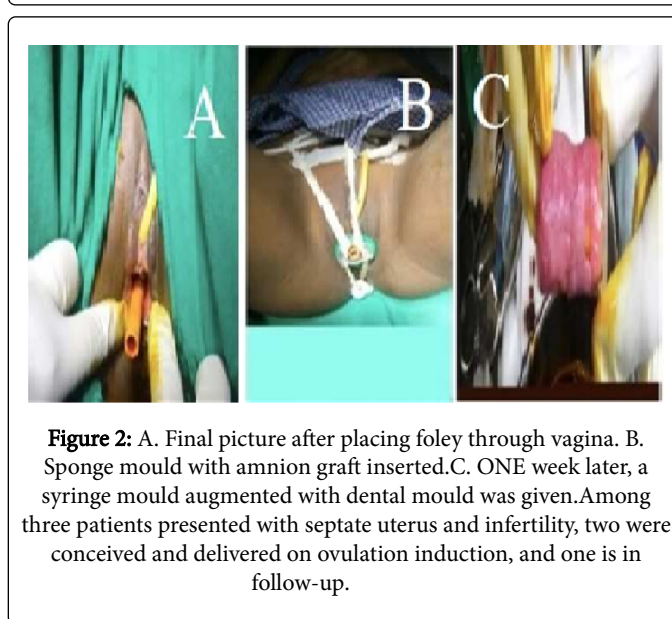


Figure 2: A. Final picture after placing foley through vagina. B. Sponge mould with amnion graft inserted. C. ONE week later, a syringe mould augmented with dental mould was given. Among three patients presented with septate uterus and infertility, two were conceived and delivered on ovulation induction, and one is in follow-up.

TVS+cervical agenesis	1	Septum excision+cervicoplasty	Resumed menses
Cervico-vaginal agenesis+uni-cornuate uterus with functional horn+hematosalpinx	1	Salpingectomy+rud. Horn excision+cervico-vaginoplasty	Resumed menses
OHVIRA Syndrome	3	Vaginal septum excision	Pain relieved
(one with normal kidney)			
Unicornuate uterus with a functional horn	1	Rudimentary horn excision	Pain relieved
Unicornuate uterus with rudimentary horn with absent cervix and vagina with Eisenmenger's syndrome	1	Hysterectomy	Pain relieved

Table 4: Details of cases presented with cyclical pain ± amenorrhoea (n=11).

Diagnosis	No.of cases	Management	Outcome
Arcuate uterus	3	Conservative	Conceived with ovulation induction
Septate uterus	3	Hysteroscopic septal resection	
Unicornuate uterus with non-functional rud horn	1	Conservative	Follow-up

The bicornuate uterus was reported in one case that was presented with recurrent miscarriage; this patient was managed conservatively. Close follow-up was done throughout the pregnancy with cervical length, progesterone supplementation was given and the patient delivered at term by caesarean section for breech presentation (Table 6).

Table 5: Distribution of cases presented with infertility (n=7).

Diagnosis	Number of cases	Management	Outcome
Septate uterus	4	Hysteroscopic septal resection	Two conceived and delivered
Bicornuate uterus	1	Conservative	Conceived and delivered
Unicornuate uterus with non-functional rudimentary horn(with fibroid)	1	Myomectomy	Conceived and delivered

Table 6: Distribution of cases presented with recurrent miscarriage (n=6).

Cases were classified according to CONUTA System proposed by ESHRE, where all organs of female genital tracts have been taken into account (U=Uterus, C=Cervix, V=Vagina). Anomalies are classified on

the scale of 0 to 5; 0 being normal and 5 depicts absence of that particular organ [8] This system enables us to classify complex Müllerian anomalies involving more than one organ (Table 7).

CONUTA System Classification	Interpretation	No. of cases	Management
U5C4V4	MRKH syndrome	9	Mcincoe vaginoplasty
U0C0V3	Imperforate hymen/TVS	2	Hymenectomy/septal resection
U3C2V2	Obstructing longitudinal vag septum with a didelphys uterus	3	Vaginal septal resection
V3C4U4B	TVS+Cx agenesis+UC uterus non-com. Rud. Horn with no cavity	1	Septum excision+cervico-vaginoplasty
U0V4C4	Cervico-vaginal aplasia	1	Cervico-vaginoplasty
U4A C4V4	Cervico-vaginal aplasia, unicornuate ut. With rudimentary horn(non-comm, functional)	1	Rudimentary horn excision +cervico-vaginoplasty

U4AC4V4	Cervico-vaginal aplasia with unicornuate uterus with hematometra with Eisenmenger's syndrome	1	Hysterectomy
U2COVO	Septate uterus	7	Hysteroscopic septal resection
U3C0V0	Dysfused uterus	1	Conservative
U4AC0V0	Unicornuate ut. With non-communicating horn with the cavity	1	Rudimentary horn excision
U4BC0V0	Unicornuate uterus with rudimentary horn with no cavity	2	Conservative
U1C0V0	Arcuate uterus	4	Conservative

Table 7: Distribution of cases according to Conuta system (n=33).

Discussion

Müllerian anomalies are not very common to see in gynaecology outpatient department, prevalence being 6.7% in the general population, 7.3% in the infertile population and 16.7% in recurrent miscarriage population [9]. In our case series, it was 33 cases of Müllerian anomalies diagnosed in three years with almost 6,000 patient attending Gynaecology OPD, making the prevalence of about 0.55% the age at presentation is typically the adolescent age group [10]. In this series, the mean age of presentation was 19.3 years.

Meyer-Rokitansky-Kuster-Hauser (MRKH) syndrome was the most common diagnosis for the cases presented with primary amenorrhoea, all were managed by Mc Indoe vaginoplasty with favourable outcome and leading normal sexual life. The results were in consistency of other studies reported in literature [11-13]. Among cases presented with cyclical pain, most of the patients were having complex Müllerian anomalies barring two, with imperforate hymen [14-16] and transverse vaginal septum and responded well with hymenectomy and septum excision respectively. Other studies [17,18] also shows good results with the same treatment. Three patients presented with obstructing longitudinal vaginal septum with uterus didelphys and hematocolpos, got relieved of symptoms after septum excision and menstruating normally [19,20]. Out of four patients with absent cervix [21,22], with or without other associated malformation, three underwent cervico-vaginoplasty were able to achieve menstruation and are under follow-up. One patient presented with unicornuate uterus with hematometra and rudimentary non-functional horn and absent cervix and vagina had associated large ventricular septal defect with the bi-directional flow with severe pulmonary artery hypertension and Eisenmenger's syndrome at presentation. For her, in consultation with a cardiologist and cardiac surgeon, and with the consent of parents, hysterectomy was done in view of severe heart disease and unable to carry a pregnancy afterwards, the patient is under follow-up in cardiology and gynaecology OPD.

Among three patients presented with septate uterus and infertility, two were conceived and delivered on ovulation induction, and one is in follow-up. Out of four patients of recurrent miscarriage with septate uterus, two had conceived and delivered at term; patients were given injectable progesterone depot preparation weekly till 34 weeks of pregnancy. Previous studies also show similar outcome with the same kind of treatment [23,24].

Since Müllerian anomalies tend to be associated with other systemic anomalies also as renal, skeletal, ano-rectal and cardio-vascular

anomalies [25,26]. In this series, we report three cases of single kidney, two with OHVIRA syndrome and one with bicornuate uterus, one patient with the ectopic kidney. One patient had associated kyphoscoliosis. One case of unicornuate uterus with cervico-vaginal agenesis had large VSD with severe PAH with Eisenmenger's syndrome. In the literature review, we could find case report of patent foramen ovale and small atrial septal defects associated with Müllerian anomalies, but none was large and presented with bi-directional shunt as in our patient [27,28].

Conclusion

Müllerian anomalies are not very uncommon to have in Gynaecology practice. Patients are mostly adolescent girls and need to be treated with genuine care and empathy. Proper diagnostic work-up is essential to reach a diagnosis. Optimum management of the condition results in a good patient outcome.

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