

Congenital Anomalies of Female Genital Tract: Review of Current Management

*Begum J,¹ Hossain MA,² Sarker SA³

To observe the congenital anomaly of female genital tract in our population and their clinical implication we performed a study in Dinajpur Medical College Hospital and a private clinic, at Dinajpur. The study period was from January 2004 to December 2011. This is a prospective observational study. All women with congenital anomalies attended in out patient department and private chamber, were admitted in the hospital or clinic with complications or operative treatment or during caesarean section or incidental diagnosis during laparotomy. Total 69 women with different types of congenital anomalies were detected. They produced various symptoms like primary amenorrhea, apareunia, infertility, haematocolpos, haematometra, abortion, ectopic pregnancy, malpresentation and uterine prolaps. Some cases were diagnosed incidentally. Among 69 patient 27(39.13%) were treated at these centers, 32(46.37%) patient needed no treatment, 2 (2.89%) patients referred to higher center, 2 (2.89%) patients refused treatment 6(9.68%) patient remain for observation. Müllerian anomalies are a morphologically diverse group of developmental disorders that involve the internal female reproductive tract. Establishing an accurate diagnosis is essential for planning treatment and management strategies.

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Introduction

An understanding of congenital anomalies as they are encountered in clinical practice is greatly enhanced by not only a knowledge of normal embryology and the mechanism of formation of normal infants but also an insight into the processes that result in the development of anomalies.¹⁻⁴ Müllerian duct anomalies consist of a set of structural malformations resulting from abnormal development of the

paramesonephric or Müllerian ducts. The prevalence of these anomalies ranges from 0.001-10% in the general population and from 8-10% in women with an adverse reproductive history.^{5,6} The embryological development of the female reproductive system is closely related to the development of the urinary system, and anomalies in both systems may occur in up to 25% of these patients.⁷ Other associated malformations may affect the gastrointestinal tract (12%) or

1. *Dr. Jahanara Begum, Assistant Professor, Department of Gynae and Obstetrics, Dinajpur Medical College, Dinajpur. aftabmunni@yahoo.com
2. Dr. Md. Aftab Hossain, Assistant Professor, Department of Anesthesiology, Dinajpur Medical College, Dinajpur
3. Dr. S M Wares Ali, Assistant professor, Department of Paediatrics, Dinajpur Medical College, Dinajpur

***For correspondence**

musculoskeletal system (10-12%).^{7,8} Gonad formation begins between the fifth and sixth weeks of pregnancy, with the appearance of the urogenital ridge developing from the intermediate mesoderm and the migration of the germinative cells originating in the coelomic epithelium. Female development is determined by the absence of the Y chromosome (and consequent absence of the factor that determines testicle development) and by the presence of two X chromosomes.^{7,8} At around the ninth week, the ovaries are formed and the Wolffian (mesonephric) and Müllerian ducts coexist. Absence of testosterone leads to involution of the Wolffian duct, whereas absence of anti-Müllerian hormone allows differentiation of the Müllerian duct. The caudal portions of these ducts merge to form the uterovaginal canal, which later gives rise to the cervix and uterus, as well as to the upper third of the vagina. Complete development of the vagina occurs through fusion of the structures of the urogenital sinus and the Müllerian tubercle. The cranial portion of the duct, i.e. the part that does not fuse, opens into the peritoneal cavity, giving rise to the Fallopian tubes.^{7,8} The causes of Müllerian anomalies have yet to be fully clarified. The karyotypes are normal (46 XX) in 92% of the women with Müllerian anomalies and abnormal (sex chromosome mosaicism) in 8% of these women. The majority of these developmental abnormalities are infrequent and sporadic, and are thus attributed to polygenic and multifactorial causes.⁹ A recent study attributed persistence of the intrauterine septum to a deficiency in the antiapoptotic protein Bcl 2, which is responsible for the process of apoptosis and absorption of the septum.⁹ 75% women with Mullerian abnormality will remain asymptomatic.⁷ The remaining 25% will present with various symptom. Nevertheless, a history of pelvic pain following the menarche, dysmenorrhea and an increase in abdominal volume are

complaints suggestive of uterine anomalies. In addition, primary amenorrhea and changes to menstrual flows may be present.¹⁰ Among the ductal differentiation malformations, vaginal agenesis presents with primary amenorrhea and dyspareunia. In cases of uteri with a functional endometrium, haematometra and haematocolpos are frequent findings. A unicornuate uterus is seldom symptomatic unless associated with other malformations. If a rudimentary, noncommunicating uterine horn is present together with a functional endometrium, haematometra and sometimes hematosalpinges may be found.¹¹ Uterine septum is generally an asymptomatic condition and is often only diagnosed when couples with a history of repeated miscarriage or infertility are undergoing investigation. Likewise, lateral fusion defects, which are responsible for uterus didelphys and bicornuate uterus, are often detected only when women undergo imaging tests.¹² Anomalies resulting from failure in vertical fusion, such as cervical agenesis, transverse vaginal septum and imperforated hymen, are associated with primary amenorrhea, haematocolpos and hematometra.¹³ Due to the complexity of presentations, diagnosing of Müllerian malformations requires the use of more than one imaging method in 62% of the cases.^{14,15} Proper assessment of these abnormalities needs combination of clinical examination, examination under anesthesia, USG, HSG, hysteroscopy and laparoscopy.¹⁶ However MRI should now replace HSG and diagnostic laparoscopy.¹⁷ The aim of this study is to observe the congenital abnormalities of the female genital tract those are found in our population and their clinical implications and also to share the experiences regarding their diagnosis, complications and management.

Methods

This is a prospective observational study which was undertaken in Dinajpur Medical college Hospital and a private clinic from January, 2004 to December, 2011. All women with congenital anomalies are attended in out patient department and private chamber, were admitted in the hospital or clinic with complications or operative treatment or during caesarean section or incidental diagnosis during laparotomy. All women underwent clinical examination and some needed examination under anaesthesia. USG was done in every patient to assess the uterine condition any other pelvic pathology and to exclude the renal abnormalities. Some patient needed HSG. Few cases underwent laparoscopy in other centre of the country or abroad. Treatment was given as per diagnosis and availabilities of facilities. Follow up of all patients was not possible due to not come back after diagnosis and treatment. Some patient came for follow up who were treated here.

Results

During the study period, a total of 69 cases of congenital abnormalities of genital tract were diagnosed. The age of the women varies from 14 years to 45 years. Imperforated hymen was found in 14 girls. All were unmarried. Among them 5 girls were admitted at emergency basis due to retention of urine. Another 9 were attended at out patient department and private chamber with the primary amenorrhea, haematocolpos and haematometra.

Müllerian agenesis was found in 12 women. Among them 8 women were unmarried with primary amenorrhea, had normal secondary sexual characteristics, vagina absent. Other 4 were married with primary amenorrhea, had normal secondary sexual characteristics, infertility, within 2 were specious vagina and no coital difficulties. Another two also presented with apareunia.

Arcuate uterus was found in 8 women. 4 women were diagnosed incidentally during caesarean section. Another 4 women were diagnosed by HSG, 1 woman was primary infertility, others were secondary infertility.

Unicornuate uterus was found in 7 women. All were identified during caesarean section. Bicornuate uterus was found 7 cases. 4 women were diagnosed at the time of caesarean section due to transverse lie. Two patients were diagnosed at HSG due to repeated pregnancy loss and infertility. In one woman who was referred at BIRDEM hospital due to 5 times mid trimester pregnancy loss where she underwent hysteroscopy and laparoscopy examination and found bicornuate uterus. Another woman had a long history. She was attended my private chamber with the history of 8 weeks pregnancy and per vaginal bleeding. USG shows ruptured ectopic pregnancy with alive foetus, but clinical finding not correspond. Again USG was done from another centre, but report was same. Diagnostic laparotomy was done and found both fallopian tubes, ovaries were normal, no collection in pouch of Douglas, uterus was 8 weeks pregnancy size and bicornuate. Both the sonologist attended in operation theatre and express their sorrow for misdiagnosis. After 7 days D&C was done due to incomplete abortion. After 2 years this patient underwent caesarean section due to breech presentation.

Unicornuate uterus with noncommunicating horn was found in 3 women. All were identified during laparotomy. Ist case was ruptured ectopic pregnancy. 2nd case was admitted in the hospital with 20 weeks missed abortion. Then she was induced by Folys catheter but failed. Again she was given oxytocin drip but uterus was ruptured. Immediate laparotomy was done and it was found to be a case of ruptured corneal pregnancy. In 3rd case, D&C was attempted

for missed abortion when the uterus was found empty. Careful bimanual examination was done and revealed a adnexal mass. Again USG was done at operation theater by a portable USG machine and shows a pregnancy sac out side the uterine cavity. Laparotomy was done and found unruptured ectopic pregnancy.

Septed uterus was found in 5 women. Four of them were diagnosed during caesarean section. Another one was diagnosed during D&C.

Subsepted uterus was found in 4 cases. Two were found during caesarean section due to transverse lie. Another case was diagnosed during manual removal of placenta. Uterus didelphys was found in 3 women. In 1st case was diagnosed by USG missed abortion with uterus didelphys, which was again confirmed during D&C and found uterus didelphys with double vagina, pregnant uterus continuous with specious vagina. Another woman came to me with 10 weeks pregnancy with 5 times midtrimester pregnancy loss. History, clinical and USG shows cervical incompetence. At 14 weeks of pregnancy McDonald operation was done. During this operation it was found that there was a small cervical like opening present on right fornix. Her pregnancy period was unevenful. She underwent caesarean section for breech presentation and at that time it was diagnosed a case of uterus didelphys. In 3rd patient presented with menorrhagia and lump in the lower abdomen. Her age was 45 years with 3 children, all are normal vaginal delivery, on examination and USG reveals a case of fibroid uterus. Laparotomy was done for hysterectomy and it was found that double uterus, double cervix but single vagina. Interesting thing is that right uterus was normal size, no myoma, well developed cervix, left uterus enlarge about 18 weeks

pregnancy size small cervix, so it is true uterus contain foetus or fibroid.

Congenital elongation of cervix was found 2 women. Both were married complaining with something coming down per vagina. Longitudinal vaginal septum was found 3 cases. One woman complain something coming down pervagina. Another case was diagnosed during labour. Incidentally and 3rd woman for pelvic pain.

Cervical aplasia was found in one case. Patient admitted in the hospital with severe lower abdominal pain. retention of urine and lump in the lower abdomen.



Figure 1. Uterus didelphys

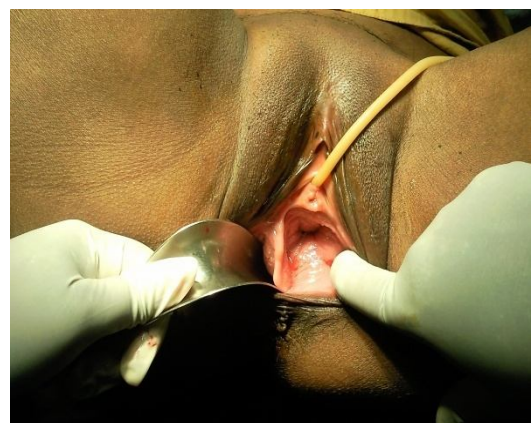


Figure 2. Septed vagina

Table I: Different congenital anomaly with clinical presentation

Abnormality	Number	Clinical presentation
Imperforated hymen	14	Retention of urine, Haematometra, Haematocolpos, Lump in lower abdomen
Müllerian agenesis	12	Primary amenorrhea, Aparaunia, Infertility
Arcuate uterus	8	Incidental diagnosed, Infertility
Unicornuate uterus	7	Incidental diagnosis
Bicornuate uterus	7	Mal presentation, Abortion
Unicornuate uterus with noncommunicating horn	3	Ectopic pregnancy
Septed uterus	5	Incidental diagnosis, Abortion.
Uterus didelphys	3	Abortion. Menorrhagia Lump in the lower abdomen
Congenital elongation of cervix	2	Something coming down per vagina.
Longitudinal vaginal septum	3	Something coming down per vagina. Incidental diagnosis
Cervical aplasia	1	Lower abdominal pain, Lump in the lower abdomen Retention of urine
Total	69	

Müllerian agenesis with left small pelvic kidney, but right kidney normal was found in one case. Another case of Müllerian agenesis with right pelvic kidney but left kidney absent. Müllerian agenesis with vaginal anal opening was found in one case. Uterus didelphys with double vagina with duplex left kidney but right kidney normal was found in one case.

Table II: Associated other Abnormalities

Associated abnormalities	Number	Müllerian abnormalities
Pelvic kidney	2	Müllerian agenesis
Duplex kidney	1	Uterus didelphys
Absent kidney	1	Müllerian agenesis
Vaginal anus	1	Müllerian agenesis

Discussion

Haematocolpos and haematometra produced by obstructive Müllerian abnormalities if untreated. Like imperforated hymen transverse vaginal septum or cervical aplasia. Tubal damage and endometriosis may eventually occur.¹⁸ Incidence of ectopic pregnancy will increase.¹⁹ In this study 14 cases were found imperforated hymen all were excised with precaution. All the patient in this series were presented 2 to 5 years following menarche.

Müllerian agenesis with absent vagina, creation of neovagina is the choice of treatment. Proper counselling is needed before surgery, specially menstrual function and fertility. Habiba et al studied 45 cases of Müllerian agenesis from 1965 to 2002 at BIRDEM hospital Dhaka.²⁰ Among them only two women were married with adequate vagina and needed no treatment. Plan was made for the remaining 43 patient to create a neovagina when they decide to go for conjugal life. In this series 12 women were found Müllerian agenesis. Four women were married, two of them were specious vagina, so no treatment needed but counselled them regarding the menstrual function and fertility. Vaginoplasty was done another two women. Remaining 8 women were unmarried. Two of them were created neovagina and they married with married person (having child). Both of them satisfied with their conjugal life. Two patients were refuse treatment. Plan was made for the remaining 4 patient to create a neovagina when they decide to go for conjugal life.

The arcuate uterus results from near-complete resorption of the uterovaginal septum. It is characterized by a small intrauterine indentation shorter than 1 cm and located in the fundal region. It is the most commonly observed uterine anomaly detected by HSG.^{21,22} Arcuate uterus was found mainly incidentally. No treatment was given.

Unicornuate uterus accounts for approximately 2.4-13% of all Müllerian anomalies.²³⁻²⁵ The unicornuate uterus may occur alone, but it is frequently associated with a rudimentary horn.^{23,26,27} Management plans have not significantly advanced. Women with unicornuate uterus are not generally considered for reconstruction metroplasty.^{7,28,29} In this study unicornuate uterus were found incidentally and due to malpresentation, no treatment was given.

The bicornuate uterus is formed when the Müllerian ducts incompletely fuse at the level of the uterine fundus. In this anomaly, the lower uterus and cervix are completely fused, resulting in 2 separate but communicating endometrial cavities, a single-chamber cervix and vagina. A muscular intrauterine septum is also present, and this defect corresponds externally to an indentation or groove at the fundus. The depth of the groove and length of the uterine septum depend in the adult uterus on the length of their completely fused Müllerian ducts in the fetus.²⁹ Bicornuate uterus seldom requires surgical reconstruction.³⁰ The benefits of metroplasty have never been studied in a prospective trial with data from most reports obtained from observational studies.²⁹ Metroplasty should be reserved for women who have experienced recurrent spontaneous abortions, midtrimester loss, premature birth, and in whom no other etiologic factor has been identified.^{29,31} In this study 4 women with bicornuate uterus were diagnosed during caesarean section due to malpresentation. They were counselled regarding their uterine anomaly and advised

proper antenatal check up during her next pregnancy. Another woman who had laparotomy due to misdiagnosed ectopic pregnancy. Next time she underwent cesarean section due to breech. One woman was referred to BIRDEM hospital. Another woman was advised to become pregnant with ovulation induction.

Pregnancy in a noncommunicating horn is uncommon and is thought to be due to transperitoneal sperm migration into the fallopian tube of the rudimentary horn. Most obstetric complications occur in the first 20 weeks and can result in abortion, uterine rupture, or maternal death (0.5%).^{32,33,34} In the event a pregnancy occurs in a noncommunicating horn, laparoscopic excision of the pregnant horn is advocated. All women in this study were excised noncommunicating horn.

In cases of complete uterine septum, resection of the cervical septum may be related to cervical incompetence and secondary infertility. A randomized controlled trial performed to evaluate the safety and efficacy of resection of the cervical septum during hysteroscopic metroplasty showed that this procedure was safer and easier with resection than with preservation of the cervical septum.³⁵ In this series 9 women were found septed and subsepted uterus. Among them 6 women were diagnosed during caesarean section due to malpresentation so no treatment was given. Two were during manual removal of placenta, both were parous women. Another patient was referred to BSMMU for hysteroscopic resection.

Didelphys uterus arises when midline fusion of the müllerian ducts is arrested, either completely or incompletely. Approximately 11% of uterine malformations are didelphys uterus.³⁶ The complete form is characterized by 2 hemiuteri, 2 endocervical canals with

cervices fused at the lower uterine segment. Each hemiuteri is associated with one fallopian tube. Ovarian malposition may also be present.³⁷ The vagina may be single or double, with duplication a frequent component. A complete longitudinal vaginal septum occurs in 75% of these anomalies, although vaginal septa can also coexist with other Müllerian duct anomalies.^{38,39} Didelphys uterus was found 3 cases. One case was presented with fibroid, hysterectomy was performed. Another case was diagnosed during caesarean section no treatment was given.^{3rd} patient was advised to become pregnant because she was a history of one missed abortion.

Congenital elongation of cervix was found 2 women. Amputation was done. Both were conceived. Longitudinal vaginal septum was found 3 cases. Two of them were excised. Another patient needed no treatment.

Agenesis of the uterine cervix, is rare and usually occurs in association with complete or partial vaginal agenesis.⁴⁰ Additional anomalies include cervical atresia as well as defects involving the length, width, and/or size of the cervix.⁴¹ Isolated cervical defects are also rare.⁷ In this study one patient with cervical atresia was identified. Recanalization of cervix was not possible, so hysterectomy was done.

Table III: Treatment Summaries of congenital anomaly of genital tract (n-69)

Treatment Summaries	Number of patient	Percentage %
Treatment given at Dinajpur	27	39.13
Treatment not needed	32	46.37
Treatment refused	2	2.89
Patient referred	2	2.69
Observation	6	9.68
Total	69	100%

Conclusion

Müllerian anomalies are a morphologically diverse group of developmental disorders that involve the internal female reproductive tract. Establishing an accurate diagnosis is essential for planning treatment and management strategies. The surgical approach for correction of Müllerian duct anomalies is specific to the type of malformation and may vary in a specific group. For most surgical procedures, the critical test of the procedure's value is the patient's postoperative ability to have healthy sexual relations and achieve successful reproductive outcomes. Therefore, their management must also be individual, taking anatomical and clinical characteristics into consideration, as well as the patient's wishes.

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