Mullerian Duct Anomaly - A Spectrum of Varied Clinical Manifestations

Poonam[1], Amit Kumar[2], Rajiv Ranjan Sinha[3]

ABSTRACT

Introduction: Multiple integrated steps are required for normal development of the female genital tract. A wide variety of malformations can occur when this system is disrupted either in form of non- development or defective fusion or failure of resorption. The purpose of this study was to review the spectrum of symptoms with which mullerian anomalies present so that timely intervention could be done to save sexual and reproductive life.

Material and methods: The study was done in the department of Obstetrics and Gynaecology of a tertiary care teaching hospital in Bihar. Only the admitted cases i.e,those who required surgical correction were taken into study.

Results: Majority of patients presented with severe abdominal pain(71.4%) followed by menstrual problems (46.4%), and urinary symptoms (35.7%) including vvf. 25% patients reported subfertility, 14.2% abdominal lump and 14.2% dyspareunia, 10.7%PID and 10.7% rectal pressure symptoms.

Majority had transverse vaginal septum (28.5%),followed by MRKHS and OHVIRA syndrome (14.2% each),

Conclusion: The variety of ways with which mullerian duct anomalies present hints that clinical suspicion should be there if early diagnosis is not to be missed.

Keywords: Mullerian Anomaly, MRKH Syndrome, OHVIRA Syndrome, Unicornuate Uterus.

INTRODUCTION

Müllerian duct anomalies occur during embryologic development. Multiple integrated steps are required for normal development of the female genital tract. A wide variety of malformations can occur when this system is disrupted either in form of non- development or defective fusion or failure of resorption. They range from uterine and vaginal agenesis to duplication of the uterus and vagina to minor uterine cavity abnormalities. Müllerian malformations are frequently associated with abnormalities of the renal and axial skeletal systems and are often recognized after the onset of puberty.[1] The actual incidence and prevalence of müllerian anomalies in the general population is not known. Most authors report incidences of 0.1-3.5%.[2,3,4] The prevalence of female genital tract anomalies is 4%–7% in general population and up to 8%–10% in women who have recurrent pregnancy loss.[5]

The primary purpose of this study was to review the spectrum of symptoms with which mullerian anomalies present so that early diagnosis and timely intervention could be done. This saves the sexual and reproductive life of patients as they usually present at an early age.The study period is from March 2016 to March 2019.

MATERIAL AND METHODS

The study was done in the department of Obstetrics and Gynaecology of a tertiary care teaching hospital in Bihar. Only the admitted cases i.e,those who required surgical correction were taken into study.

This was a descriptive review of three years of all cases of Mullerian duct anomalies that required surgical correction between March 2016- March 2019). Data were retrieved from hospital registration statistics, gynaecology admission register, case files and operation theatre records of women under study. They were searched for age and complaints at presentation, prior history of intervention,type of intervention done along with complications before and after surgery. The data thus extracted from the case files, which are preserved in the hospital, were entered into Microsoft excel spread sheets and analysed with the help of calculator.

RESULTS

Total number of indoor admissions (March2016-19) in the department was 2466.The number of cases operated in Gynaecology routine OT was 815. Out of which 28 cases were operated upon for mullerian duct anomalies during this period. So the percentage contribution of mullerian duct anomaly was 3.4% of the total operated cases and 1.1% of the total indoor admissions.

Table-1 Deals with different types of mullerian anomalies along with the age of presentation. Four patients (14.2%) in age range of 16-19 were admitted with Mayer Rokitansky Kuster Hauser syndrome. Ohvira syndrome was found in five patients(17.8%). Out of this, four patients(14.2%) were diagnosed to have uterus diadelphus with hemiobstructed vagina and right renal agenesis where as only one patient with OHVIRA had bicornuate uterus with right hemiobstructed

[1]Additional Professor, Department of Obstetrics and Gynaecology, Indira Gandhi Institute of Medical Sciences, Patna, [2]Associate Professor, Department. of Radiodiagnosis, Indira Gandhi Institute of Medical Sciences, Patna, [3]Associate Professor, Department of Anatomy, Indira Gandhi Institute of Medical Sciences, Patna, India

Corresponding author: Dr Poonam, Doctors Qr-D-6/1, IGIMS Campus, Sheikhpura, Patna-14, India

How to cite this article: Poonam, Amit Kumar, Rajiv Ranjan Sinha. Mullerian duct anomaly - a spectrum of varied clinical manifestations. International Journal of Contemporary Medical Research 2019;6(11):K10-K13.

DOI: http://dx.doi.org/10.21276/ijcmr.2019.6.11.23

Anomaly	Age	Number	Percent
Uterus diadelphus with obstructed hemivagina & ipsilateral renal agenesis (OHVIRA syndrome)	13-25	4	1 4.2%
Mayer Rokitansky kuster Hauser Syndrome	16-19	4	14.2%
Bicornuate uterus with obstructed hemivagina & ipsilateral renal agenesis	35	1	3.5%
Transverse vaginal septum	12-24	8	28.5%
Longitudinal vaginal septum	21	1	3.5%
Unicornuate uterus with non communicating rudimentary horn	16	1	3.5%
Imperforate hymen	16-17	2	7.1%
Microperforation	18-20	2	7.1%
Vaginal agenesis	15-17	3	10.7%
Cervical agenesis	15-26	2	7.1%
Total		28	

Table-1: Showing different anomalies along with age at presentation.

Complaints	Number	Percentage
Menstrual problems	13	46.4%
Abdominal lump	4	14.4%
Abdominal Pain	20	71.4%
Subfertility	7	25%
Urinary symptoms	10	35.7%
Rectal pressure	3	10.7%
Dyspareunia	4	14.2%
Pelvic Inflammatory Disease,Discharge	3	10.7%
Note: Some cases had more than one complaints.		

Table-2: Showing various complaints at presentation

Complications	N=	%
Urinary tract infection	8	28.5%
Febrile morbidity	4	14.2%
Injury to bladder	1	3.5%
Injury to rectum	1	3.5%

Table-3: Complications during and after surgery

vagina and ipsilateral renal agenesis.

Eight patients(28.5%) in the age range of 12-24 yrs. had transverse vaginal septum. Out of this four were in high and two each in mid and lower vagina.

One case(3.5%) each of septate uterus with longitudinal vaginal septum in a 21 year old married girl and unicornuate uterus with non communicating rudimentary horn in an 18 year old unmarried girl was seen. There were two cases (7.1%) each of imperforate hymen and microperforations in the age range of 16-20 years.

Cervical agenesis was seen in a 15 year old and a 26 year old girl.This constituted 7.1% of the total cases.

Table -2 shows the variety of ways with which mullerian anomalies present. Maximum number of patients 71.4% (n=20) presented with abdominal pain followed by menstrual symptoms seen in thirteen patients (46.4%). Ten patients (35.7%) had urinary symptoms followed by subfertility in seven cases (25%).Equal number(n=4) of patients were seen with abdominal lump and dyspareunia.

In some patients there were overlap of symptoms.

Table -3 Deals with the complications encountered either during surgery or in immediate the post operative period. UTI was seen in eight cases(28.5%), hematuria in one (3.5%) and post operative fever in 4 (14.2%)cases. All eight cases had anaemia prior to surgery. In one case bladder was opened accidentally and in another rectum opened inadvertently. Both cases had history of prior attempt to surgical correction elsewhere.

DISCUSSION

Congenital mullerian anomaly accounted for 3.4% of the total operated cases and 1.1% of the total indoor admissions. Patients with simple anomalies are mostly asymptomatic and are diagnosed incidentally during investigations for other gynaecological or obstetrical problem, mostly at puberty. Because of the wide variation in clinical presentations, müllerian duct anomalies may be difficult to diagnose. After an accurate diagnosis is made, many treatment options exist, and they are usually individualized to the specific müllerian anomaly. The most common müllerian duct defects involve the vagina and the uterus. These anomalies are the most easily corrected surgically.Abdominal pain (71.4%) was the most common presentation in this study,followed by obstructed menstruation(46.4%). Others included primary and secondary amenorrhea, subfertility, urinary retention, hematuria, increased frequency of micturition and dyspareunia.

Maximum number of cases (28.5%))had transverse vaginal septum.Four in the upper third of vagina and two each in mid and lower third of vaginal. This anomaly causes hematocolpos in patients with functioning endometrium. This is consistent with the findings of Rock JA and Blask AR et al.[7,8] Diagnosis in all cases were confirmed by Magnetic Resonance Imaging.

It is important to note that diagnosis of a uterine or vaginal anomaly by imaging before puberty can be challenging and misleading because of the small size of the prepubertal uterus, the lack of endometrial stimulation, and the lack of menstrual distention of the vagina.[6]

The most common symptom associated with obstruction is pain. In a patient who presents with pain, the presence of menstruation does not rule out obstruction[6], as was seen in patients with uterus diadelphus or bicornuate with hemiobstructed vagina and also in cases of microperforations. Pain may occur in a cyclic manner or may be continuous.

Surgery, which is the definitive treatment of uterovaginal obstructions, should only be performed by experienced

surgeons to avoid a repeat surgery. Otherwise, scarring of tissues and further distortion of anatomy could lead to urinary tract and bowel injuries. This problem was faced by us also.In this study, the two significant complications i.e, bladder and rectal injury occured in two different cases who had history of prior unsuccessful attempts to surgical correction. In both the cases, problem of severe, continuous pain persisted after the first surgery.

In the case where bladder wasaccidentally opened,resection of transverse vaginal septum (vertical fusion defect) was attempted about three months earlier.This persisted either due to incomplete resection of septum, with resulting reapproximation and distortion of anatomy or involvement of vaginal mucosa which could have led to scarring. Bladder was found to be projecting into the vaginal canal.Before starting the procedure, catheter could not be negotiated into the urethra.Probably, alignment of urethra with bladder was obliterated.

Surgical management includes completely excising the intervening septal tissue. As the risk of vaginal stenosis is high in such patients, surgical intervention should be followed postoperative dilatation.

Longitudinal vaginal septum in a septate uterus was seen in only one case. Septate uterus increases the risk of early pregnancy loss and hysteroscopic intervention is required to resect the septum in symptomatic patients. However, this patient presented with dyspareunia as the chief complaint.

An 18 yr old unmarried girl with unicornuate uterus and non communicating rudimentary horn presented with heart shaped suprapubic abdominal lump, and severe pain abdomen. The left side of lump was just palpable whereas the right horn was about the size of 14-16 weeks pregnant uterus.Her menarche was four months back with no mense since then. On the third day of admission she menstruated, reducing the size of lump drastically. The heart shape also disappeared but pain persisted. MRI revealed hematometra in non communicating rudimentary horn of unicornuate uterus which was resected. If MRI indicates no endometrium in a uterine horn, resection typically is not necessary.

If there is an active endometrium,resection of the uterine remnants to decrease pain, retrograde menstruation, and endometriosis is needed.[10] A unicornuate uterus accounts for 2.4 to 13% of all Müllerian anomalies.[5] In our series, it accounted for 3%.

There were two cases each of imperforate hymen and microperforations(Cribriform hymen) in the age range of 16-20 years.Here also pain was the chief complaint along with hypomenorrhea in the one with cribriform hymen.

There were five cases of OHVIRA syndrome(Obstructed hemivagina and ipsilateral renal anomaly) also known as Herlyn-Werner-Wunderlich **syndrome**. It is a rare Mullerian duct anomaly with uterus didelphys (or bicornuate uterus), unilateral obstructed hemivagina, and ipsilateral renal agenesis. Incomplete resorption of uterine or vaginal septa leads to rudimentary and obstructed uterine horns or obstructive hemivagina and ipsilateral renal anomaly. Surprisingly, in all cases right side was involved.

Patients with a history of a solitary or multicystic dysplastic kidney have a risk of an obstructed uterine horn or an ipsilateral obstructing vaginal septum with a patent contralateral vagina.[9] Most patients with obstructive hemivagina and ipsilateral renal anomaly have a diadelphic, bicornuate, or septate uterus.

Patients with this anomaly who present post menarche often report with severe dysmenorrhea pelvic pain and/or a mass and rarely, in later years, with primary infertility. Strong suspicion and knowledge of this anomaly is essential for a precise diagnosis.In one of the cases of OHVIRA syndromes, hemiobstruted vagina (6x5cm elongated mass in upper anterolateral wall of vagina) was mistakenly diagnosed as gartners duct cyst. Drainage of hematometrocolpos was done and longitudinal vaginal septum was resected.

Next commom presentation was that of menstrual abnormalities ranging from primary and secondary amenorrhea to hypomenorrhea, and severe dysmenorrhea. Four cases in the age range of 16-19yrs presented with primary amenorrhea. MRI revealed complete aplasia of mullerian ducts (Mayer-Rokitansky-Kuster-Hauser syndrome). Müllerian aplasia is an uncommon, but not a rare, anomaly.

Mayer-Rokitansky-Kuster-Hauser syndrome represents the failure of vaginal development associated with uterine and cervical agenesis or varying degrees of hypoplasia. Müllerian aplasia can be partial or complete. Partial müllerian aplasia is more rarely encountered and is characterized by a normal uterus and small vaginal pouch distal to the cervix. Complete müllerian aplasia (MRKH syndrome) is the most common variant encountered and it is characterized by congenital absence of the vagina and the uterus in 90-95% of cases. The fallopian tubes are normal, and the ovaries have normal endocrine function.[12]

The two cases with microperforations presented with hypomenorrhea, discharge per vaginum and pain lower abdomen (pelvic inflammatory disease).Ascending infection due to microperforation could have led to pelvic inflammatory disease.

Other associated symptoms included urinary frequency(n=3), dysuria, hematuria (n=1), and urinary retention(n=5). Urinary retention was seen in 28.3% of cases; although as high as 46% has been reported by Abu- Ghanem S et al[11] in hematometrocolpos caused by imperforate hymen.

One patient, aged 24 years old with high transverse vaginal septum had vesico vaginal fistula with cyclical hematuria but no urine leak.Actually hematocolpos found a way through upper vagina into bladder during menses. The transverse septum was resected but the small fistula was left as such anticipating spontaneous healing. As the enclosed pressure in upper vagina decreased after septal resection, fistula disappeard.

This patient conceived two months after treatment and delivered vaginally.Hematuria resolved completely.

Two cases were diagnosed to have cervical agenesis. Both presented with primary amenorrhea with severe cyclical abdominal pain.One amongst these had previous

unsuccessful attempt to correct the anomaly with resulting dense scarring at the introitus. The main objective of treatment of cervical agenesis was symptom relief.However to facilitate sexual function, vaginoplasty was also done apart from hysterectomy. But during the procedure rectum was accidently opened which was repaired after conversion to complete perineal tear.

If a patient undergoes a hysterectomy for cervical atresia, vaginal elongation for vaginal sexual activity typically can be accomplished by vaginal dilation alone without further surgical treatment. Dilator therapy should be deferred until the patient is ready.[10]

CONCLUSION

Because of the wide variation in clinical presentation, müllerian duct anomalies may be difficult to diagnose. Clinical suspicion should be there if early diagnosis is not to be missed.

After an accurate diagnosis is made, many treatment options exist, and they can be individualized according to the specific anomaly.

REFERENCES

1. Lawrence S Amesse et al.Mullerian Duct Anomalies. Medscape 2018,April 2.
2. Strassmann EO. Operations for double uterus and endometrial atresia. Clin Obstet Gynecol. 1961. 4:240.
3. Strassmann EO. Fertility and unification of double uterus. Fertil Steril. 1966;17:165-76.
4. Golan A, Langer R, Bukovsky I, Caspi E. Congenital anomalies of the mullerian system. Fertil Steril. 1989;51:747-55.
5. Acién P. Incidence of Müllerian defects in fertile and infertile women. Hum Reprod. 1997;12:1372-6.
6. Dorairajan G, Pegu B. Pessary Compared With Vaginal Progesterone for the Prevention of Preterm Birth in Women With Twin Pregnancies and Cervical Length Less Than 38 mm: A Randomized Controlled Trial. Obstet Gynecol. 2019;133:1283.
7. Blask AR, Sanders RC, Rock JA. Obstructed uterovaginal anomalies: Demonstration with sonography. Part II. Teenagers. Radiology 1991;179:84-8.
8. Rock JA. Anomalous development of the vagina. Semin Reprod Endocrinol 1986;4:13-31.
9. Smith NA, Laufer MR. Obstructed hemivagina and ipsilateral renal anomaly (OHVIRA) syndrome: management and follow-up. Fertil Steril 2007;87:918–22.
10. Müllerian agenesis: diagnosis, management, and treatment.ACOG Committee Opinion No. 728. American College of Obstetricians and Gynecologists. Obstet Gynecol 2018;131:e35–42.
11. Abu-Ghanem S, Novoa R, Kaneti J, Rosenberg E. Recurrent urinary retention due to imperforate hymen after hymenotomy failure: a rare case report and review of the literature.Urology 2011;78:180–2.
12. Baramki TA. Treatment of congenital anomalies in girls and women. J Reprod Med. 1984;29:376-84.

Source of Support: Nil; **Conflict of Interest:** None

Submitted: 11-10-2019; **Accepted:** 19-10-2019; **Published:** 23-11-2019