DOI: http://dx.doi.org/10.18203/2320-1770.ijrcog20200879

Original Research Article

Study of mullerian anomalies over 5 years in a tertiary care centre

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Received: 20 December 2019 Revised: 20 January 2020 Accepted: 24 January 2020

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ABSTRACT

Background: Mullerian anomalies-developmental anomalies of the mullerian system might involve the uterus, cervix, fallopian tubes and vagina of which anomalies of the uterus are the most common. They are often regarded as a treatable form of infertility and have a prevalence of 0.5% in the general population. Though most of them remain asymptomatic, they contribute a good proportion of recurrent pregnancy losses, obstetric complications and infertility. Their timely diagnosis, management and evaluation of associated anomalies proves to be necessary.

Methods: A prospective observational study was undertaken at KIMS, Hubli from 2014 to 2018. The study involved those women who were admitted in the department of obstetrics and gynecology at KIMS, Hubli for various reasons and were detected to have a mullerian anomaly. Their mode of presentation, method of detection, associated anomalies were analysed and statistical conclusions drawn from the same.

Results: During the study period of 5 years, a total of 85 cases of mullerian anomalies were detected amounting to an incidence of 0.15%. 35% of them were asymptomatic, infertility (24%) being the next common mode of presentation. Septate/sub-septate uterus was found to have the poorest obstetric outcome. Mullerian anomalies were seen in combination in 14% of the cases and had an association with other anomalies like renal anomalies in 10% of the cases. Surgical correction was done for most of the cases of vaginal septum and septate/sub-septate uterus.

Conclusions: Mullerian anomalies are often regarded as an uncommon but treatable form of infertility. Their timely detection, treatment and watchful obstetric management is in need.

Keywords: Hystero-laparoscopy, Infertility, Mullerian anomalies, Septate

INTRODUCTION

Congenital anomalies of the female reproductive tract may involve the uterus, cervix, fallopian tubes or vagina. Uterine anomalies are the most common mullerian anomalies but the true incidence is not known as many women are asymptomatic and sensitive imaging modalities have only recently become available.^{1,2} Most cases are diagnosed during evaluation for obstetric or gynaecological problems but in the absence of symptoms, most anomalies remain undiagnosed. Reported population prevalence rates have varied between 0.06% and 38%.³ As nearly 47% of women with uterine defects

have successful fertility and pregnancy, the true incidence of congenital mullerian defects is significantly understated. Mullerian anomalies are known to be associated with renal anomalies in 30-50% of cases and defects include renal agenesis, severe renal hypoplasia, ectopic or duplicate ureters.³

Clinical presentation of mullerian anomalies

Though many women with mullerian anomalies are asymptomatic, several gynaecologic signs and symptoms are associated with specific anomalies. Women may present with cyclic or non-cyclic pelvic pain and dysmenorrhea suggestive of an obstructive anomaly, retrograde menstruation and endometriosis.¹ Primary amenorrhea with pelvic, vaginal or back pain or a pelvic mass is concerning for a transverse vaginal septum or an imperforate hyemen. Mullerian agenesis presents with amenorrhea. Abnormal uterine bleeding may occur with a septate uterus, a partial or microperforate obstruction or a longitudinal septum. Of all mullerian anomalies, those involving the uterus are most commonly associated with poor obstetric outcomes. Uterine anomalies may be associated with diminished cavity size, inadequate musculature, impaired ability to distend, abnormal myometrial and cervical function, inadequate vascularity abnormal endometrial development.^{2,5} These and abnormalities of space, vascularity and associated local defects contribute to increased rates of recurrent pregnancy loss, preterm delivery, malpresentation, IUGR.^{1,6} An increased rate of caesarean section is associated with higher rates of malpresentation and vaginal anomalies like longitudinal vaginal septum (Figure 1).



Figure 1: Classification of mullerian anomalies.⁴

Evaluation of mullerian anomalies

HSG is commonly used to assess patency of tubes and can provide further information about the contour of the endometrial cavity but the external uterine contour is poorly defined by HSG.⁷ Transabdominal, transvaginal or transperineal USG effectively evaluates the internal and external uterine contour. MRI is considered the gold standard technique for diagnosing mullerian anomalies and is both sensitive and specific.⁸ Historically, laparoscopy and hysteroscopy were the gold standard for evaluating the contour of the uterine fundus and assigning the classification to a uterine anomaly.⁹

Surgical correction

Surgery for mullerian anomalies is often indicated for women with pelvic pain, endometriosis, obstructive anomalies like vaginal septum, poor obstetric outcomes like RPL, infertility. Prior to performing surgery, it is important to exclude extrauterine factors which may cause pregnancy loss.^{1,2}

METHODS

The present study was conducted among the women admitted in the department of obstetrics and gynaecology at KIMS, Hubli for various complaints with a mullerian anomaly detected during the present stay at the hospital between January 2014 to December 2018 - a period of 5 years.

Inclusion criteria

• All women admitted in the department of obstetrics and gynaecology at KIMS, Hubli with a newly detected Mullerian anomaly between January 2014 and December 2018.

Exclusion criteria

- Those with previously diagnosed Mullerian anomalies
- Women who have already undergone surgical correction of the anomaly
- Those who do not give consent to take part in the study.

Present study is a prospective observational study. Considering inclusion and exclusion criteria, selected patients were counselled regarding the study. After obtaining valid consent, a pre-designed proforma was used. Detailed clinical history regarding the presenting complaints, obstetric history, menstrual history was taken. General physical examination, systemic and necessary gynaecological examination was done. Other investigations were performed with relevance to their presenting complaints. The mode of detection of the anomaly was noted i.e., through clinical examination, imaging, hysterolaparoscopy and so on. Surgical correction feasible in our set up were done wherever necessary. However, there was no follow-up of the treated cases.

Statistical analysis

All the collected data variables were entered into an exel sheet and after appropriate data filtration, the data sheet was transferred to and analysed using SPSS software version 22. Appropriate descriptive statistics were used to describe the data.

RESULTS

The study period of 5 years between 2014 and 2018, a total of 85 cases of mullerian anomalies were detected among patients who were admitted in the department of obstetrics and gynaecology at KIMS, Hubli for various reasons. These numbers were 14,14,16,22 and 19 in the years 2014, 2015, 2016, 2018 and 2019 respectively. The incidence was found to be 0.15% over 5 years (Table 1).

Year >	2014	2015	2016	2017	2018	Total
No of cases of mullerian anomalies detected at KIMS Hubli	14	14	16	22	19	85
% with respect to the total no of cases admitted	0.13	0.13	0.15	0.19	0.18	0.15

Table 1: Incidence of mullerian anomalies at KIMS, Hubli.

Table 2: Age at diagnosis of mullerian anomalies.

Age at diagnosis	2014	%	2015	%	2016	%	2017	%	2018	%	Total	%
< 15 years	1	7.1	1	7.1	0	0	1	4.5	1	5.2	4	4.7
15-20 years	2	14.2	1	7.1	4	25	5	22.7	2	10.4	14	16.4
21-25 years	8	57.1	5	35.5	9	56.2	10	45.4	11	57.8	43	50.5
26-30 years	2	14.2	3	21.3	2	12.5	3	13.6	4	21	14	16.4
31-35 years	0	0	2	14.2	1	6.2	0	0	0	0	3	3.5
> 35 years	1	7.1	2	14.2	0	0	3	13.6	1	5.2	7	8.2

Table 3: Presentation of mullerian anomalies.

Presentation	2014	%	2015	%	2016	%	2017	%	2018	%	Total	%
Incidental finding	5	35.7	2	14.2	5	31.2	9	40.9	9	47.3	30	35.2
Infertility	1	7.1	4	28.5	7	43.7	5	22.7	3	15.7	20	23.5
Recurrent pregnancy loss	3	21.3	2	14.2	1	6.2	1	4.5	4	21	12	14.1
Hematometra/hematocolpos	2	14.2	1	7.1	3	18.7	1	4.5	1	5.2	8	9.4
Primary amenorrhea alone	3	21.3	4	28.5	0	0	4	18.1	2	10.4	13	15.2
Others	0	0	1	7.1	0	0	1	4.5	0	0	2	2.3

The women of various age groups who were admitted at KIMS, Hubli maximum detection rate of mullerian anomalies was noted in the age group 21-25 years in all years (50%). Very few cases were detected in women of extreme age groups (Table 2).

A large proportion (35%) of women with mullerian anomalies were asymptomatic and the anomaly was only an incidental finding. The next common mode of presentation was infertility (24%). This pattern was observed in the years 2014, 2017 and 2018. However, infertility was the commonest mode of presentation in 2016 constituting 44% and the commonest modes of presentation were infertility and primary amenorrhea in the year 2015 each amounting to 29%. Other modes of presentation were recurrent pregnancy loss, primary amenorrhea, hematometra and hematocolpos. only 2 cases had presentations other than the above mentioned like abnormal uterine bleeding (Table 2).

Type of anomaly	2014	%	2015	%	2016	%	2017	%	2018	%	Total	%
Uterine hypoplasia/ aplasia	3	21.4	4	28.5	0	0	2	9	2	10.5	11	12.9
Vaginal septum (horizontal)	2	14.2	1	7.1	1	6.2	1	4.5	0	0	5	5.8
Vaginal septum (vertical)	0	0	1	7.1	0	0	1	4.5	0	0	2	2.3
Vaginal agenesis	0	0	0	0	1	6.2	1	4.5	0	0	2	2.3
Unicornuate uterus	0	0	1	7.1	1	6.2	1	4.5	3	15.7	6	7
Uterus didelphys	0	0	0	0	2	12.4	1	4.5	4	21	7	8.2
Bicornuate uterus	4	28.5	2	14.2	5	31.2	6	27.2	4	21	21	24.7
Septate/subseptate uterus	4	28.5	5	35.7	5	31.2	7	31.8	6	31.5	27	31.7
Arcuate uterus	1	7.1	0	0	1	6.2	2	9	0	0	4	4.7

Table 4: Type of mullerian anomaly.

An average over 5 years, the commonest mullerian anomaly was septate/sub-septate uterus (32%). This was followed by bicornuate uterus (25%). Considering the years separately, septate/ subseptate uterus remained the commonest anomaly in 2015, 2017 and 2018. However,

septate/subseptate uterus and bicornuate uterus were detected in equal numbers in the years 2014 and 2016. (Table 4). Important tools for the detection of mullerian anomalies were clinical examination, ultrasonography, hysterosalpingography and hysterolaparoscopy. All cases of vaginal septum were identified by local examination and most cases of uterine aplasia/ hypoplastic uterus were picked up by USG. A large number of anomalies were detected by hysterolaparoscopy the commonest being septate/ subseptate uterus (Table 5).

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Type of anomaly	Detected by clinical examination	HSG	Picked by USG	Diagnosed by hystero laparoscopy	Incidental finding on laparotomy (most commonly for LSCS)
Hypoplastic uterus	0	3	7	1	0
Unicornuate uterus	0	2	1	1	2
Uterus didelphys	1	2	1	2	1
Bicornuate uterus	0	3	4	3	11
Septate/subseptate uterus	0	4	0	16	7
Arcuate uterus	0	0	0	1	3
Vaginal septum	9	0	0	0	0
Total	10	14	13	24	24

Table 5: Mode of detection of various anomalies.

Table 6: Surgical correction.

Type of anomaly	No of cases for which surgical correction was done (over 5 years)
Hypoplastic uterus	0
Unicornuate uterus	0
Uterus didelphys	0
Bicornuate uterus	1
Septate/subseptate uterus	16
Arcuate uterus	0
Vaginal septum	11

Over a period of 5 years, surgical correction was done for 16 cases of septate/ sub-septate uterus (septal resection), 11 cases of vaginal septum (vaginoplasty) and 1 case of bicornuate uterus (metroplasty). Septal resection in cases of septate/sub-septate uterus diagnosed by means of hysteroscopy were done in the same sitting (Table 6).

All cases of hypoplastic uterus presented as primary amenorrhea. Unicornuate uterus was an incidental finding in most of the cases. Uterus didelphys was an incidental finding in in 43% of the cases, 43% of the cases presented with infertility (Table 7).

Bicornuate uterus was an incidental finding in 67% of the cases. The most common incidentally detected mullerian anomaly was bicornuate uterus. Patients with septate and sub-septate uterus most commonly presented as infertility (52%). Septate/sub-septate was found to be the most common mullerian anomaly leading to infertility as well as recurrent pregnancy loss.

Table 7:	Type of	anomaly	versus	presentation.
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Presentation → Type of anomaly	Incidental finding	Infertility	Recurrent pregnancy loss	Primary amenorrhea	Hematometra /hematocolpos	Others
Hypoplastic uterus	0	0	0	11	0	0
Unicornuate uterus	5	0	1	0	0	0
Uterus didelphys	3	3	1	0	0	0
Bicornuate uterus	14	2	3	1	1	0
Septate /subseptate uterus	5	14	7	0	0	1
Arcuate uterus	3	1	0	0	0	0
Vaginal septum	0	0	0	1	7	1

Arcuate uterus was an incidental finding in most (75%) of the cases.

A total 78% of the cases with vaginal septum presented with hematometra/hematocolpos and only 2 cases of

vaginal septum had other modes of presentation like primary amenorrhea and cyclic abdominal pain.

Though mullerian anomalies were found to cause infertility and recurrent pregnancy losses in a large percentage of cases, obstetric complications were noted in only 14% of the women with mullerian anomalies the commonest complication being malpresentation. Malpresentations (most commonly breech) were noted commonly in patients with bicornuate uterus. The presence of vaginal septum led to caesarean sections in 2 cases, preterm labour was noted in 2 patients and only 1 patient presented with dystocia (septate uterus) (Table 8).

Mullerian anomalies were seen in combination in 14% of the cases. Most of them were a combination of a uterine anomaly with vaginal septum. The commonest associations were uterus didelphys with vaginal septum (3.5%) and bicornuate uterus with vaginal septum (3.5%) (Table 9).

Table 8: Obstetric complications.

Obstetric complications→	Malpresentation	Preterm labour	Dystocia	Primary indication for LSCS
Hypoplastic uterus	0	0	0	0
Unicornuate uterus	1	1	0	0
Uterus didelphys	0	0	0	0
Bicornuate uterus	4	0	0	0
Arcuate uterus	1	0	0	0
Septate/subseptate uterus	3	1	1	0
Vaginal septum	0	0	0	2

Table 9: Associated mullerian anomalies.

Associated mullerian anomalies	No. of cases	%
Vaginal septum + uterus didelphys	3	3.5
Vaginal septum + unicornuate uterus	1	1.1
Vaginal septum + bicornuate uterus	3	3.5
Vaginal septum + septate uterus	1	1.1
Vaginal septum + arcuate uterus	1	1.1
Bicornuate uterus with cervical stenosis	1	1.1
Bicornuate uterus with malformed tubes	2	2.3
Total	12	14.1

Table 10: Associated anomalies in other systems of the body.

Other associations	Absent/ ectopic kidney	Displaced or patulous urethral opening	Absent or malformed ovaries	Anal stenosis
Hypoplastic uterus	1	0	0	0
Unicornuate uterus	1	0	0	0
Uterus didelphys	1	1	0	0
Bicornuate uterus	1	0	2	1
Septate/subseptate uterus	0	0	0	0
Arcuate uterus	0	0	0	0

Nearly 10% Mullerian anomalies were found to be associated with other anomalies, the commonest being renal anomalies found in around 5% of the cases. The other anomalies that were noted were malformed ovaries, displaced urethral opening and anal stenosis (Table 10).

DISCUSSION

This study was conducted to analyze the incidence and presentations of various anomalies of the uterus as, a high

degree of suspicion is required to diagnose the same and also the associated anomalies.

Among the cases admitted in the department of obstetrics and gynaecology at KIMS, Hubli between the years 2014-18 (a period of 5 years), 85 of them were diagnosed to have mullerian anomalies giving an incidence of 0.15%. This is similar to the incidence of 1 in 200 to 1 in 600 i.e., 0.1 to 0.5% quoted by Rock JA et al.¹⁰ The commonest age group at which the mullerian anomalies were detected was 21 to 25 years in this study. the median age at diagnosis was 26 in a study conducted by Gurtcheff SE et al.¹¹ This depends on the median age at which the women plan to conceive and seek medical help for the same in a particular population.

The mullerian anomaly was only an incidental finding in 35% of the women in this study, this being very much similar to the results of the study conducted by Gurtcheff et al, wherein the percentage of incidentally detected mullerian anomalies was 32%.¹¹ However, the next common mode of presentation in our study was infertility in contrast to recurrent pregnancy losses and obstetric complications in the study conducted by Gurtcheff SE et al.

About 23% of the women in whom mullerian anomalies were detected presented with infertility in comparison to the study conducted by Golan A et al wherein the incidence of unexplained infertility in women with congenital anomalies was found to be 35%.¹²

Septate / sub-septate uterus was found to have the poorest obstetrical outcome. This goes as stated by Heinonen PK et al. Associated non-mullerian congenital anomalies were found in 10% of the cases. this is similar to the study conducted by Gurtcheff SE et al, wherein this association was noted in 8.5% of the cases. Though standard authors quote an association of renal anomalies with mullerian anomalies to be 30-50%, only 5% of the cases were detected to have renal anomalies in our study.

Though imaging modalities like 3D USG are the main stay of diagnosis of mullerian anomalies, diagnostic hysterolaparoscopy was a major diagnostic tool in our study especially for anomalies like septate/ sub-septate uterus and this proved beneficial as correction could be done in the same sitting. Since most of the mullerian anomalies are an incidental finding and have good obstetric outcomes, surgical correction seemed necessary only in a handful of patients majority being vaginoplasty for vaginal septum and septal resection in case of septate/sub-septate uterus.

CONCLUSION

Developmental anomalies of the mullerian system are often regarded as a treatable form of infertility. Though most of the mullerian anomalies are asymptomatic and have good obstetric outcomes, they become responsible for pregnancy losses, infertility and obstetric complications in a considerable number of cases. A high degree of suspicion is required for the timely diagnosis and treatment of mullerian anomalies and their watchful obstetric management to prevent complications. It is also necessary to keep an eye on the possible associated anomalies and syndromes.

Funding: No funding sources Conflict of interest: None declared Ethical approval: The study was approved by the Institutional Ethics Committee

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Cite this article as: Antaratani RC, Kumar S, Raju V. Study of mullerian anomalies over 5 years in a tertiary care centre. Int J Reprod Contracept Obstet Gynecol 2020;9:1083-8.