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Original Research Article

Evaluation and management of Mullerian duct anomalies

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ABSTRACT

Background: Development of the female genital tract is a complex embryogenesis including cell differentiation, migration, fusion, and canalization. Mullerian duct anomalies (MDA) are structural anomalies which occur due to defective embryogenesis of the Mullerian ducts, resulting in abnormalities of internal reproductive organs and ambiguity of the external genitalia. Other causes include deficiencies in steroidogenesis, signalling defects of WNT4 gene and TP63, receptor defects and genetic abnormalities.
Methods: A prospective observational study was performed at our institute to observe the congenital anomalies of the female genital tract in our population and their clinical implication.
Results: Most common anomaly observed was septate uterus followed by bicornuate uterus. Gynaecological complication: most common presenting symptom was primary amenorrhea followed by cyclical abdominal pain. Obstetric complication: women with Mullerian duct anomalies have a higher incidence of preterm labour, repeated first trimester spontaneous abortions, foetal malpresentations, and intrauterine foetal growth restriction. Hysterosalpingography (HSG) and ultrasonography (USG) are the primary tools to detect genital tract anomalies, Magnetic resonance imaging (MRI) being gold standard for detecting Mullerian duct anomalies.
Conclusions: Mullerian duct anomalies can present with various obstetric and gynaecological complications. MRI is superior to other diagnostic modalities (HSG or USG) in establishing an accurate diagnosis and deciding further management options. Surgical approach used for correction of these anomalies is specific to the type of malformation and may vary in a specific group.

Keywords: Congenital anomalies, Mullerian duct anomalies, Hysterosalpingography, Ultrasonography, Magnetic resonance imaging

INTRODUCTION

The Mullerian ducts are the primordial anlage of the female reproductive tract. They differentiate to form the fallopian tubes, uterus, cervix, and upper one third of vagina.[1]

Müllerian anomalies occur as a congenital malformation of the Müllerian ducts during embryogenesis. The Müllerian ducts are also referred to as paramesonephric ducts, referring to ducts next to (para) the mesonephric (Wolffian) duct during foetal development. Paramesonephric ducts are paired ducts derived from the embryo, and for females develop into the uterus, uterine tubes, cervix and upper two-thirds of the vagina.[2]

Development of the female genital tract is a complex process dependent upon a series of events involving cellular differentiation, migration, fusion and canalization. Failure of any one of these processes results in different types of anomalies of the vagina and uterus. Two paired Mullerian ducts ultimately develop into: fallopian tubes, uterus, cervix, and the upper two thirds of the vagina.[3]

Present study includes malformation that affect the development and morphology of fallopian tube, uterus, vagina and vulva, with or without associated ovarian, urinary, skeletal or other organ malformation and it excludes the abnormality of sexual determination (involving chromosomal alteration, male histocompatibility antigen, sex determining region of y chromosome and testis-determining factor gene or the gonads) and sexual differentiation (by abnormal steroidogenesis or pseudo-hermaphroditism).[4]

Aims and objectives

Aims and objectives of the study were to observe congenital Mullerian duct anomalies (MDA) of female genital tract in our study population; to study clinical pictures associated with MDA; to study various diagnostic modalities for MDA; and to study various surgical techniques tailored to specific mullerian anomalies. To study the reproductive outcomes in women with MDA.

METHODS

A prospective observational study was done at SVP IMSR NHLMMC, Ahmedabad to observe the prevalence of Mullerian duct anomalies in our study population. The study period was from December 2022 to December 2024. A total of 52 cases were studied. They were divided into two groups. Group A included 21 gynaecological cases and group B included 31 obstetric cases.

Women who presented with various complaints (primary infertility, primary amenorrhea, repeated spontaneous abortions) or for operative intervention in already diagnosed cases of MDA or who presented with incidental diagnosis of MDA during caesarean section, diagnostic hysteroscopy or laparoscopy/laparotomy were included. Reproductive outcomes were also correlated.

Statistical analysis

Data analysis is done by using Microsoft excel or statistical package for the social sciences (SPSS) version 23.

RESULTS

Majority of the cases with Mullerian anomalies of female genital tract were detected in reproductive age group, which is between 21-30 years while patients with gynaecological problems were detected during adolescent age which is 11-19 years. Most common anomaly observed Was septate uterus 25% (n=13/52) followed by arcuate uterus 21% (n=11/52). 5.76% (n=3/52) cases had associated renal and axial skeleton developmental anomalies.

Due to close embryological relation with development of urinary tract and reproductive organs, renal tract anomalies are likely to be associated with Mullerian anomalies. Out of 3 cases of MRKH syndrome, one case had a single ectopic kidney. One case was documented with isolated skeletal anomaly (cervical vertebral fusion). Unilateral kidney was found in the case of unicornuate uterus without horn

Table 1: Age wise distribution of the patients.

Age (years)	Group A (gynec cases)	Group B (obstetrics cases)	Total patients, N (%)
11-15	6	0	6 (11.5)
16-20	7	3	10 (19.2)
21-25	6	14	20 (38.5)
26-30	2	10	12 (23.1)
>30	0	4	4 (7.7)

Table 2: Different associated anomalies of female genital tract.

Associated anomalies	Mullerian anomaly	No of patients' in present study
Absent kidney	Uni- cornuate uterus without horn, MRKH syndrome	2
Skeletal anomaly	MRKH syndrome	1

Table 3: Presenting symptoms.

Symptoms	N (%)
Group A	
Primary amenorrhea	10 (47.6)
Cyclical abdominal pain	5 (23.8)
Infertility	6 (28.57)
Group B	
Preterm labour	17 (54.83)
Repeated first trimester spontaneous abortion	5 (16.12)
Foetal mal-presentation	8 (28.80)

In gynaecological group, most common presenting symptom of obstructive MDA was primary amenorrhea 47.6% (n=10/21) followed by cyclical abdominal pain 23.8% (n=5/21). Infertility was noted in 28.57% (n=6/21). In the Obstetric group, women with MDA had a higher incidence of preterm labour 54.83% (n=17/31), repeated first trimester spontaneous abortions 16.12% (n=5/31) and foetal mal-presentations 25.80% (n=8/31). Breech presentation was the commonest malpresentation-19.2%. It was found in all cases of uni-cornuate uterus without horn. While incidence of transverse lie was 11.5%, more commonly observed in the arcuate uterus.

Mullerian duct anomalies will be primarily illustrated using HSG, 2D -USG. In a study USG is the primary modality of diagnosis in 100% of cases of mullerian anomalies. In present study, laparoscopy was carried out in 3 cases to confirm USG diagnosis. Two were the cases

of secondary infertility and diagnosis was laparoscopy, one as bicornuate uterus and other as uni-cornuate uterus without rudimentary horn. Hystero-laparoscopy was carried out in 6 cases of septate uterus to confirm ultrasound diagnosis and for hysteroscopic resection under laparoscopic guidance.

In doubtful or complex cases, MRI should be performed, particularly for the assessment of the cervix and vagina. In present study MRI was performed in 4 cases of MRKH syndrome, 1 case of cervical atresia and 2 cases of unilateral vaginal obstruction with uterine anomalies.

Table 4: Various modalities of diagnosis.

Parameters	Group A (no. of cases)	Group B (no. of cases)
HSG	8	0
USG	21	1
MRI	7	0
Hysteroscopy	6	0
Laparoscopy	3	0

Table 5: Accidental diagnosis during surgery (group B).

Variables	LSCS (no. of cases)	Normal labor (no. of cases)	Laparotomy (no. of cases)
Bicornuate uterus	3	0	0
Arcuate uterus	11	0	0
Septate uterus	2	0	0
Didelphic uterus	2	0	0
Uni-cornuate uterus with horn	0	0	1
Bicornuate uterus without horn	3	0	0

The patients having bicornuate uterus (57.1%) and septate uterus (38.5%) were accidentally diagnosed during LSCS and MRI that did not require surgical correction of anomaly. HSG 15% (n=8/52) and USG 42% (n=22/52) are the primary tools to detect Mullerian duct anomalies, MRI 13.4% (n=7/52) being gold standard for detecting Mullerian duct anomalies. Surgical correction was needed in a total of 38.46% cases (n=20/52) to improve reproductive outcomes (Table 6).

A total of 20 cases needed surgical correction to improve reproductive outcomes in present study. Utero-vaginal anomalies detected during LSCS, MRI and normal labour did not require any surgical correction.

In present study, 23.8% (5/21) cases were conceived, one after surgical correction and 4 naturally. Beginning of normal menstruation occurred in 3 cases of imperforate hymen, 1 case of transverse vaginal septum and 1 case of cervical atresia after surgical intervention, without any follow up complications.

Table 6: Surgical correction for group A and B.

Type of surgery	No. of cases
Group A	
Vaginal septum excision	1
Unilateral transverse vaginal septum excision	2
Uterine septum resection	6
Vaginoplasty	4
Utero-vaginal anastomosis for cervical atresia	1
Cruciate shapes incision for imperforate hymen	3
Total	17
Group B	
Excision of rudimentary horn	1
Division of isolates longitudinal vaginal septum	1
Excision of transverse vaginal septum	1
Total	3

Table 7: Various malpresentation in group B with MDA.

Variables	Breech (no. of cases)	Transverse (no. of cases)	Vertex (no. of cases)
Bicornuate uterus	1	0	3
Arcuate uterus	1	2	8
Septate uterus	1	1	2
Didelphic uterus	0	0	2
Unicornuate uterus without horn	2	0	1
Isolated longitudinal vaginal septum	0	0	1

Table 8: Follow up outcome in group A cases.

Variables	No. of cases
Conception after surgical correction	1
Conception without surgical correction	4
Menstruation after surgical intervention	5

DISCUSSION

The incidence in the present study was calculated by taking into account outdoor cases with gynaecological complaints and indoor cases and registered antepartum cases. The

present study shows prevalence of MDA 0.19% at our tertiary care centre.

Table 9: Incidence of congenital anomalies of female reproductive tract.

Different study	Incidence (%)
Byrne et al (2000)	0.4
Chan et al (2011)	0.06-35
Grimbizis et al (2012)	4-7
Pedro et al (2016)	3-6
Present study	0.19

In present study, maximum numbers of cases with Mullerian anomalies of female reproductive tract (group B) were detected in reproductive age group with mean maternal age of 21.8 years which is comparable to Hua et alal study, who reported mean maternal age of patients 29.3 years.[5]

In present study, septate uterus is the commonest uterine anomaly (25%) followed by arcuate uterus (21%) similar to Raj et al study.[6]

Out of 13 cases of septate uterus, 7 were diagnosed during pregnancy. Remaining 6 were presented with infertility, which were surgically corrected. USG and HSG are the primary tools to detect genital tract anomalies, 3D USG and MRI being the gold standard for detecting MDA. Minto et al also mentioned USG as the initial diagnostic modality for all patients with MDA and MRI has now replaced the previous second line diagnostic modalities-hysteroscopy and laparoscopy.[7]

Patients with uterine anomalies have significantly higher rates of malpresentation (30.7%), which is comparable with the study of Zhang et al 93%, in which the incidence of malpresentation was 38.8%.[8]

Patients with congenital utero-vaginal malformation had significantly higher rates of preterm delivery (30.7%) in present study. the study conducted by Zhang et al93 and Raj et al 79% reported preterm delivery rate was 19.8% and 22.22% respectively.[6,8]

Twenty-six patients of group B were delivered either vaginally or by caesarean section. Out of them 69.2% (18/26) had cephalic presentation including one twin pregnancy with 1st foetus cephalic, 19.2% (5/26) had breech presentation and 11.5 % (3/26) had transverse lie which is comparable to Hua et al 80% study in which breech was observed in 23.6% cases.9

In present study, 23.8% (5/21) cases conceived from group A which was comparable to the study by Nishida et al where 36.4% cases (4/11) conceived after surgical intervention.[10]

Limitations

The present study included women attending only SVP hospital Ahmedabad and representing only one geographical region. The present study was done at tertiary care center where all the facilities for ultrasonography and obstetrician and gynaecologists were available which might not be available at remote areas and at rural areas.

CONCLUSION

Utero-vaginal anomalies are a morphologically diverse group of developmental disorders that involve the internal female reproductive tract.

Mullerian anomalies are fairly prevalent and present with various gynecological and obstetric complications. A universally acceptable classification system remains elusive, and the ASRM classification should be used until further research proves the merit of the ESHRE/ESGE classifications system. USG and HSG are used as the primary tools of diagnosis. MRI is the gold standard diagnostic modalities. The surgical approach for correction of utero-vaginal anomalies is specific to the type of malformation and very in specific group. For most surgical procedures, the critical test of the procedure's value is the patients' post-operative ability to have healthy sexual relations and reproductive outcome.

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