DOI: 10.7860/JCDR/2025/74783.21102

Case Report

Obstetrics and Gynaecology Section

Beyond Classification: A Unique Case of Mullerian Malformation Featuring a Normal Uterus with the Absence of Unilateral Ovary and Fallopian Tube

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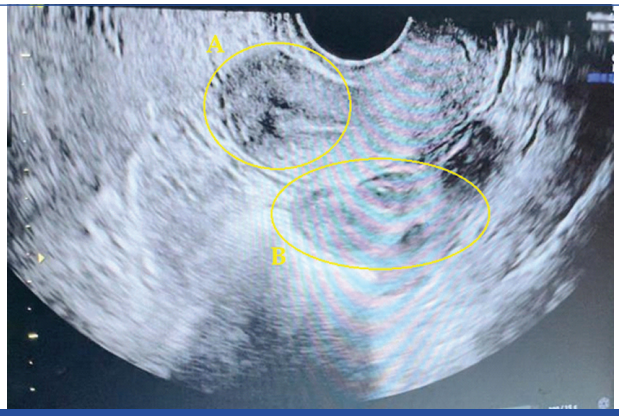
ABSTRACT

Müllerian malformations represent a range of genetic conditions impacting the female reproductive tract, often posing diagnostic and therapeutic challenges. According to the reports, these anomalies cause the Müllerian ducts to abruptly form during foetal development. Müllerian ducts are related to the normal formation of reproductive internal and exterior organs in the females. This is a case of a 29-year-old female with a major complaint of primary infertility, for the past 10 years. The patient was screened by ultrasonography and hormonal profile along with the husband's semen analysis. On further evaluation, a unilateral absence of the left fallopian tube and ovary was noted, with the presence of a normal uterus, which defied current classification systems for Müllerian anomalies. This case highlights the complexity and diagnostic dilemmas associated with Müllerian malformations, emphasising the importance of a multidisciplinary approach and personalised management strategies in such cases.

Keywords: Menstrual irregularities, Polycystic ovarian disease, Primary infertility, Psammoma bodies

CASE REPORT

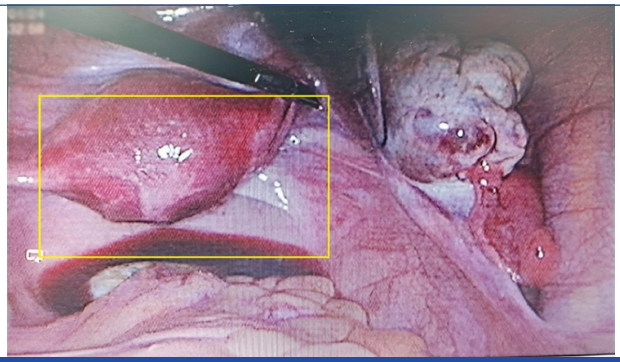
A 29-year-old nulligravida female, with a major complaint of primary infertility since 10 years, visited the out-patient department of our tertiary care facility for further management for conception. The patient had regular menstrual cycles. Medical history revealed the patient to have Irritable Bowel Syndrome (IBS) spanning five years and stable with conservative management. Patient had two failed attempts of Intrauterine Insemination (IUI) and the patient underwent an ovum pick-up procedure one year ago as part of Assisted Reproductive Technology (ART) interventions. Physical examination of the pelvic revealed a normal external appearance of the vagina, genitalia and the uterus. Subsequent radiological imaging by the ultrasound was suggestive of normal findings of the uterus and right ovary with absent left fallopian tube and ovary [Table/Fig-1]. The patients and her partners' baseline investigations for infertility were found to be normal, except for hysterosalpingography, which was suggestive of a normal uterus, with a blocked fallopian tube (right) while the left fallopian tube was undetectable. Her hormonal profile was normal as showed in [Table/Fig-2]. Considering these findings, the decision was made to proceed with diagnostic laparoscopy and hysteroscopy. Hysteroscopy findings demonstrated a structurally normal uterine cavity and endometrium, with unremarkable visualisation of both the right and left ostia and normal cervical canal. Subsequent laparoscopic exploration showed left ovary and its adjacent fallopian tube absence as shown in [Table/Fig-3].



[Table/Fig-1]: Radiographic image showing only right ovary and uterus and left ovary not visualised. A: Endometrium with endometrial thickness; B: Right ovary.

Parameters	Patient value	Normal range
Follicle-stimulating hormone (mIU/mL)	7	3-9
Luteinising hormone (mIU/mL)	4	2-10
Prolactin (ng/mL)	4	0-20
Oestradiol (pg/mL)	96	27-161
Thyroid stimulating hormone (mIU/mL)	0.4	0.2-4.7
Anti-mullerian hormone (ng/mL)	0.9	0.7-3.5

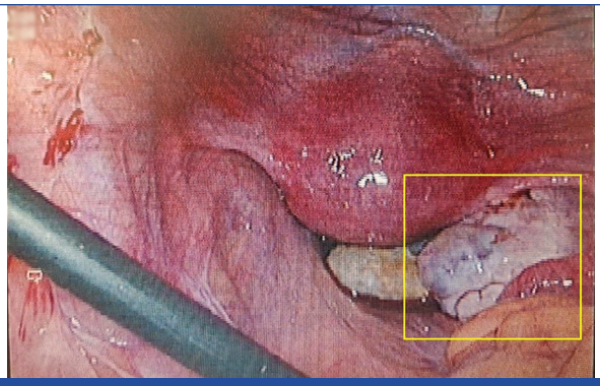
[Table/Fig-2]: Laboratory test profile of the patient.



[Table/Fig-3]: Figure depicting absent left ovary and left fallopian tube (Yellow marked area indicates the absence of the left ovary).

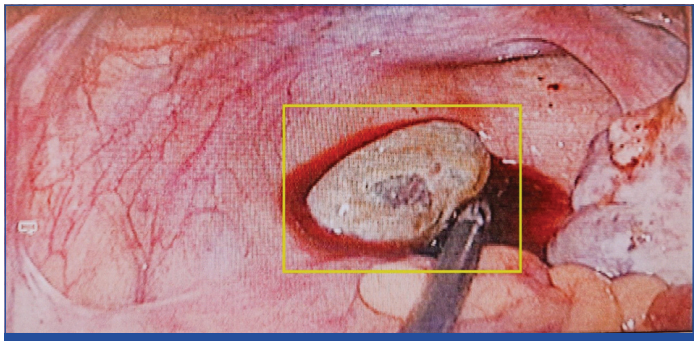
A bulky, polycystic appearance of the right ovary suggestive of Polycystic Ovary Syndrome (PCOS) as shown in [Table/Fig-4]. Polycystic Ovarian Disease (PCOD) drilling was carried out under laparoscopic guidance on the right ovary. Ovarian drilling was done bilaterally under laparoscopic guidance. A total of six punctures were made in each ovary with a total current of 150 joules. The surface was intermittently cooled by Hartmans solution.

The uterine surface appeared normal, with a normal-sized uterus and patent right fallopian tube demonstrated by methylene blue dye spillage. Left-side, a stony hard mass of size 3×3 cm was seen in the

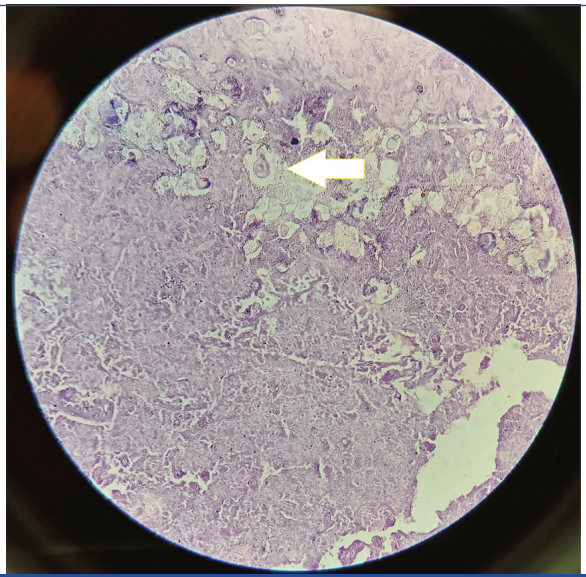


[Table/Fig-4]: Figure depicting bulky right ovary with yellow mark highlighting bulky right ovary.

pouch of Douglas adjacent to the right ovary, which was removed and sent for histopathology [Table/Fig-5]. Histopathology showed complete coagulation necrosis with extensive areas of calcification with psammoma bodies as shown in [Table/Fig-6].



[Table/Fig-5]: Figure depicting hard mass adjacent to the right ovary. (Yellow box highlighting the mass).



[Table/Fig-6]: Histopathological staining (Haematoxylin and eosin) at 40x magnification of the excised specimen showing Psammoma body.

Further exploration provided insights into the complex anatomy of the patient's reproductive tract, with a special focus on the absence of the left fallopian tube and left ovary, coupled with PCOS features on the right ovary. These findings further contributed to the diagnostic puzzle surrounding her infertility. Genetic analysis conducted on peripheral blood samples revealed a normal karyotype of 46, XX. Additionally, a subsequent intravenous pyelogram unveiled an intact urinary tract devoid of any congenital abnormalities. The final diagnosis was concluded as Müllerian unilateral agenesis. The patient has history of three failed IUI cycles, hence she was managed by ovum pickup and embryo transfer. The pregnancy was successful as evident by a positive HCG urine test on day 21 postimplantation.

DISCUSSION

Müllerian Duct Anomalies (MDA) are rare clinical presentations, usually defined as the absence of fallopian tubes with normal ovaries and uterus. It is typically seen in females with infertility issues. It is reported that 6.3% of the patients with infertility problems have uterine malformations [1]. Also, the incidence is not clearly defined as asymptomatic females without any fertility concerns are not screened for the same, so the incidence of MDA remains unclear. A classification of MDA by the American Society of Reproductive Medicine is commonly accepted. Nonetheless, there exist rare anomalies that do not fit within this classification scheme [1,2]. Ipsilateral absence of the ovary and the fallopian tube is a rare noted presentation with an approximate incidence of 1 in 11240 cases [3]. The exact causes behind the occurrence of ovarian and tubal ipsilateral absence remain elusive. Based on reported cases thus far, three potential explanations have been proposed: adnexal torsion, maldevelopment of the ovarian and tubal structures following ischaemic changes triggered by vascular events and abnormalities in the formation of the mesonephric and Müllerian system, apart from affecting one side entirely or localised to specific regions such as the genital ridge and caudal segment [3-6].

Ovarian and tubal agenesis can be attributed to two potential aetiopathogenic factors. The first hypothesis implicates non symptomatic torsion of one or both the adnexa, occurring in adulthood, childhood and foetal development [4]. Evidence supporting this theory includes past radiological screenings indicative of the presence of ovaries and tubes, as well as intraoperative findings of distinct structures within the abdominal cavity, later confirmed by histologic analysis as tubal and ovarian tissues. However, in present case, neither laparoscopic surgery nor preoperative ultrasound imaging were suggestive of the presence of separated ovary and fallopian tube tissue remnants. Unilateral congenital absence can also stem from abnormalities affecting the genital ridge and the Müllerian duct. Furthermore, abnormalities during the course of development, affecting the entire mesonephric duct system and the Müllerian duct on a side might be the cause [7]. Failure of the formation of the canals of the Müllerian might result in improper development of fallopian tube or its absence and might contribute towards developmental abnormality in the form of a unicornuate uterus. Gonad agenesis might result from an inadequate canalisation of one fallopian tube or a vascular event because of unknown paracrine and autocrine signaling processes. Attributed to any additional anatomical anomalies, the theory of congenital defects appears plausible [4]. However, due to the limited number of documented cases, this hypothesis cannot be definitively ruled out.

The existing classification system for MDA lacks comprehensiveness, often failing to encompass rare cases that do not align with its parameters. Oppelt P et al., proposed an alternative classification system known as the Vagina Cervix Uterus Adnexa-associated Malformation (VCUAM) classification, aiming to provide a more detailed description of complex genital anomalies [8]. VCUAM subdivides the internal and external female genitalia into distinct classes, allowing for individual grading of anomalies in each anatomical structure. These complex anomalies included rudimentary uterus, unilateral adnexal aplasia, uterine malformations rising due to improper Müllerian ducts fusion, cervical and vaginal atresia. Remarkably, no cases of adnexal aplasia concomitant with minor structural changes in the uterus were found in previously described abnormalities. According to the VCUAM classification, this case can be noted as V0C0U0A3aM0, indicating a rare manifestation of MDA.

This is a rare case that had an unusual MDA presentation opposed to the typical definition, which was diagnosed during exploring the reasons for primary infertility. Other cases of MDA are also found incidentally. One such case by Alsina JL and Khamvongsa P reported the findings during a caesarean section delivery [9]. Similarly,

Kansara M et al., reported a series of cases of Müllerian anomalies in young women with different clinical manifestations and aiming at making their correct diagnosis and appropriate management to improve the obstetric and gynaecological outcome [10]. There are differential clinical presentations, with a rare variant of MRKH syndrome presenting with primary amenorrhoea, had a hypoplastic non cavitated uterus in the middle with two rudimentary horns on either side [11]. This patient also, had reproductive complications with a long history of inability to conceive. She had an unusual combination of a normal uterus and ipsilateral absence of ovary and its adjacent fallopian tube. Hence, it is important to create awareness among health professionals to provide proper management to individuals. The significance of this case lies in its implications for clinical practice. Firstly, it emphasises the importance of MDA to be considered as a differential diagnosis in females with apparent fertility issues. Secondly, it draws attention to the limitations of existing classification systems in clinical practice. This case can be used as an example of adapting a flexible approach in the diagnosis of MDA, recognising that deviations from the norm may warrant individualised management strategies.

CONCLUSION(S)

This case highlights the diagnostic challenge associated with MDA, particularly in the context of infertility evaluation. The absence of ipsilateral ovary and fallopian tube with a normal uterus emphasises a significance of comprehensive diagnostic modalities and individualised management strategies. Despite the rarity of such cases, thorough evaluation and consideration of alternative classification systems might be crucial for accurate diagnosis and optimal patient care. Further research is required to elucidate its aetiology with improving the outcomes for patients with similar presentations of MDA.

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PLAGIARISM CHECKING METHODS: [Jain H et al.]
- Plagiarism X-checker: Aug 09, 2024
- Manual Googling: Feb 13, 2025
- iThenticate Software: Feb 15, 2025 (2%)

ETYMOLOGY: Author Origin

EMENDATIONS: 6

AUTHOR DECLARATION:
- Financial or Other Competing Interests: None
- Was informed consent obtained from the subjects involved in the study? Yes
- For any images presented appropriate consent has been obtained from the subjects. Yes

Date of Submission: **Aug 06, 2024**
Date of Peer Review: **Oct 10, 2024**
Date of Acceptance: **Feb 17, 2025**
Date of Publishing: **Jun 01, 2025**