CERVICO-VAGINAL AGENESIS WITH A UNICORNUATE UTERUS: A CASE REPORT AND LITERATURE REVIEW

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ABSTRACT

Introduction: Mayer-Rokitansky-Kuster-Hauser syndrome (MRKH-S) is a rare condition that results from defects in the organogenesis and morphogenesis of the internal female organs. MRKH-S occurs sporadically, and the exact incidence is mostly unknown. Furthermore, MRKH-S poses a diagnostic and treatment challenge.

Objective: To describe a case of MRKH-S and a literature review on the clinical aspects of this rare condition.

Case: A 12-year-old African female presented with cyclical lower abdominal pain not having attained menarche. A pelvic ultrasound scan was done, which revealed haematometra. At laparoscopy, cervical and upper vaginal agenesis with haematosalpinx were demonstrated. Vaginoplasty and a hysterectomy were done, and subsequent follow up was made.

Conclusion: MRKH-S occurs rarely and sporadically. The clinical diagnosis of MRKH-S may suffice for obstructive lesions, in which imaging tests may be requested. A multidisciplinary approach is essential to manage MRKH-S cases effectively.

Keywords: Cervico-vaginal agenesis, Hysterectomy, Mullerian agenesis

INTRODUCTION

Mayer - Rokitansky - Kuster - Hauser syndrome (MRKH-S) is a rare condition affecting 1 in 4000 – 10,000 women (1). Synonyms attributed to MRKH-S include; Mullerian agenesis, Mullerian aplasia, and vaginal agenesis (2). Mullerian agenesis defects arise from disorders in organogenesis and morphogenesis of the internal female reproductive organs from the Mullerian ducts with vaginal anomalies associated with uterine anomalies (3). However, vaginal anomalies can occur in isolation due to the failure of the fusion of the Mullerian ducts and urogenital sinus (4).

Anomalies due to Mullerian agenesis are varied and affect different levels of the female genital tract; hence various classification systems exist. The two most used classifications are the American Society of Reproductive Medicine (ASRM) and the European Society of Human Reproduction and Embryology (ESHRE) (5,6). The main distinction between the two classification systems is that the ESHRE classification considers vaginal malformations, while the ASRM classification does not (3). The diagnosis of Mullerian anomalies is based on clinical presentation and diagnostic tests and may include diagnostic laparoscopy (7-9). Managing MRKH-S cases is complex and requires a multidisciplinary approach, involving psychological counselling, surgical treatment, and reproductive specialist follow-ups (8,10,11). MRKH-S is a rare and complex anomaly that may pose a clinical dilemma to gynaecologists.

CASE PRESNTATION

A 12-year-old African female presented with a four-month history of cyclical monthly lower abdominal pain. However, she had not attained menarche. She had been managed with oral analgesics, which partially resolved the pain. Also, she had been on Depot Provera injection; however, her symptoms did not resolve.

A clinical examination of the abdomen revealed a soft, non-tender abdomen with no pelvic masses. The pelvic ultrasound was done, which showed a normal size uterus, with an endometrial fluid collection of 5 cubic centimetres and normal ovaries, a diagnosis of hematometra was made. Magnetic resonance imaging (MRI), showed an elliptically shaped uterus (solitary cornua) that deviated to the left side with normal myometrial and endometrial differentiation. MRI also revealed a non-communicating horn next to the solitary horn with endometrial and myometrial differentiation, and there was no connection between the upper vagina and the uterus, with the non-communicating segment's length between the two being 2.5cm. It further revealed an average vagina with a length of 4.5cm with no septum (Figure 1).

The patient received counselling on her diagnosis and hence was planned for further counselling as her management continued. The patient was scheduled for examination under anesthesia and laparoscopy for further diagnostic workup. Pelvic examination revealed a blind-ending vaginal vault of about 5 cm in length with no discernible cervix and a unicornuate blind-ending uterus with left haematosalpinx (Figure 2). A right blind-ending 'cord like' structure was probably an atretic tube with a right cystic ovary (Figure 3). The lower uterus thinned out to a blind end with no discernible cervix. A diagnosis of a unicornuate uterus with cervical and partial upper vaginal agenesis was made.

Parental consent was obtained, and the patient was booked for vaginoplasty. Dissection at the cranial end of the blind-ending vagina to separate it from the bladder anteriorly and the rectum posteriorly, maintaining a plane towards the non-connected uterus's direction was done. Two 'stay suture' stitches were placed at the lateral aspects of the vagina's cranial end to keep the dissected area from healing back together. An attempt to connect the vagina's apex and the uterus's lower segment through a vaginal approach was unsuccessful. A pack was left in the vaginal cranial end to prevent re-closure of the dissected apex. Together with her parents, the patient was debriefed after the surgery about her diagnosis, and they were informed on the need for further staged surgery and probable prolonged subsequent follow-up.

Following extensive consultations, cervico-vaginoplasty or a hysterectomy with vaginoplasty options were considered. Since the atretic cervicovaginal portion's length would have difficulty approximating the uterus to the vagina during surgery, a hysterectomy with subsequent follow-up for vaginal dilatation was opted instead of uterine conservation surgery after weighing both procedures' benefits and risks. Subsequent healing issues with a high risk of anastomotic breakdown due to repair under tension was also considered. Moreover, it was thought that the size of the unicornuate uterus might not have offered adequate reproductive function regarding carrying a pregnancy to term (12). The patient was later admitted, and a hysterectomy was performed (Figure 4). The patient subsequently recovered well from surgery and is currently on the serial use of vaginal dilators to facilitate vaginal expansion.

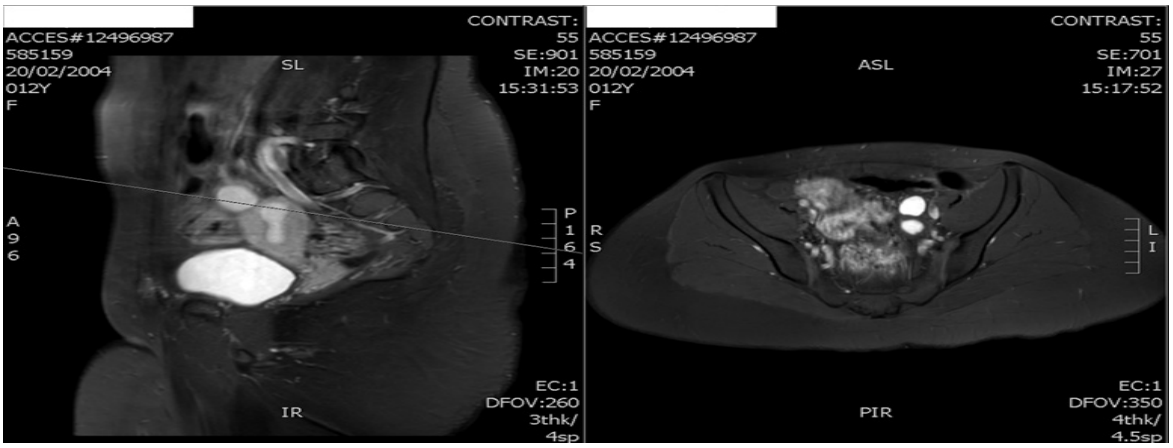

Figure 1. MRI images showing an elliptically shaped uterus (solitary cornu) deviated to the left side with normal myometrial and endometrial differentiation (A). Non-communicating horn is next to it also with endometrial and myometrial communication (B). The absence of connection between the upper vagina and the uterus – length of the non-communicating segment is 2.5cm (C). Normal vagina with a length of 4.5cm with no septum (D).

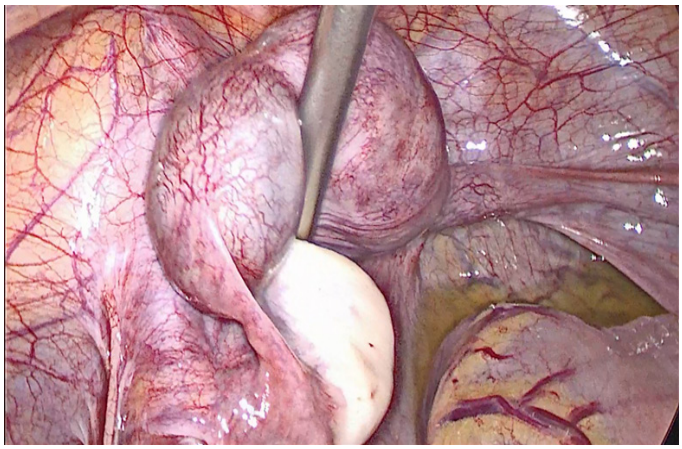

Figure 2. Unicornuate blind-ending uterus with proximally distended left tube with haematosalpinx. The left ovary has a normal appearance.

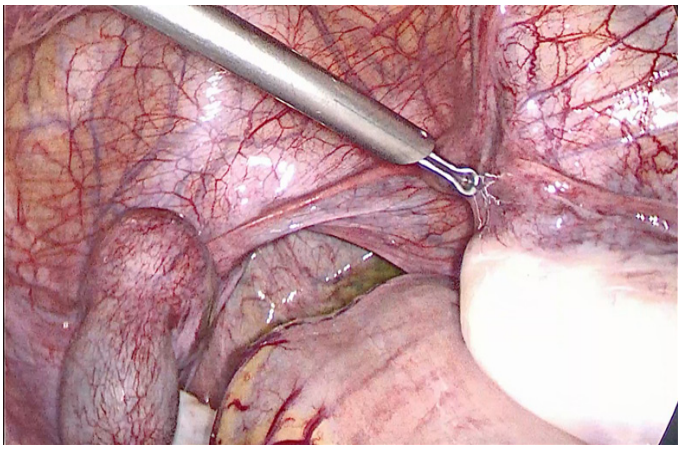

Figure 3. A right blind-ending cord-like structure with a right cystic ovary.

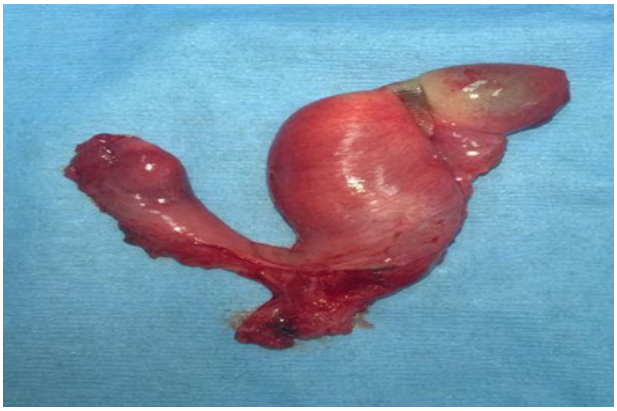

Figure 4. Post-hysterectomy specimen of the unicornuate uterus.

DISCUSSION

Mayer - Rokitansky - Kuster - Hauser syndrome (MRKH-S) is a rare condition, and it is essential to document such a case for posterity and offer insight into the presentation and management of this condition. The exact incidence of MRKH-S is unknown, although it can vary from as low as 4 in 1000 in the general female population to 80 in 1000 among women with recurrent miscarriages (12,13). MRKH-S occurs sporadically, and no predisposing or risk factors have been identified. Molecular studies have so far failed to identify alterations and polymorphic changes in the Mullerian embryonic controller genes (14). The female genital tract's internal parts develop from the Mullerian ducts, bilateral embryonic structures that undergo morphogenesis, migration, and vertical and horizontal fusion to result in the fallopian tubes, uterine corpus, cervix, and upper-vagina (1,4). Consequently, any defect in this complex process can result in agenesis or underdevelopment of the fallopian tubes, uterus, or vagina (4), and can broadly be described as the MRKH-S, which can, as a result, have many variations.

The American Society of Reproductive Medicine (ASRM) and the European Society of Human Reproduction and Embryology (ESHRE) classification systems are the two commonly used in the classification of MRKH-S (5,6). The ASRM classification, which is older, is the most widely used and divides uterine anomalies into seven major groups (5). However, this system does not include vaginal anomalies and certain combined anomalies. The ESHRE classification, which includes malformations of the uterus, cervix, and vagina, is newer and maybe increasingly used (6). The current case could be classified as type I and II as per the ASRM classification and Class U6 as per the ESHRE classification (5,6).

Clinical presentation of MRKH-S varies, with the typical age of presentation being at adolescence (12), since most MRKH-S defects affect the internal female reproductive tract; hence, the external genitalia is typical, with no reason for concern from early childhood (13). Patients with obstructive lesions, including non-communicating horns, cervico-vaginal atresia, vaginal septum, vaginal atresia, and imperforate hymen, would present with delayed onset features (12). Furthermore, these patients would have delayed onset of menses, with cyclical pain at the time of menarche, which progressively worsens (12). However, patients who do not have obstructive defects, uni- or bicornuate

uterus, an arcuate or septate uterus, uterus didelphys, or longitudinal vaginal septum, are usually diagnosed incidentally later in life when undergoing investigation for recurrent miscarriages, delayed conception, and during vaginal or caesarean delivery (13). The case presented was of an obstructive lesion of cervico-vaginal atresia hence presented with cyclical pain and delayed menarche.

Mullerian agenesis is diagnosed based on clinical investigation and radiological workup (2). In patients with obstructive lesions, as in this case, clinical diagnosis may suffice. In obstructive lesions affecting the vagina, a pelvic exam may point out the diagnosis (12). Cases with imperforate hymen are easily diagnosed, while those with higher up lesions, as in this reported case, may have obstructive symptoms with no apparent cause (15,16). Imaging studies are required to demonstrate MRKH-S. A pelvic ultrasound is usually the first imaging study in obstructive lesions (17). The scan may reveal a haematocolpos like in our patient but rarely will it provide additional information on the extent of lesions. Magnetic Resonance Imaging (MRI) is the gold standard of imaging MRKH-S lesions (18). An MRI may also identify additional malformations associated with MRKH-S like renal anomalies and hence may be useful in wholesome planning of the patient's management (12,18).

The management of patients with MRKH-S requires a multidisciplinary team and is often complex and prolonged (11,16). Patients require psychological counselling following the diagnosis of MRKH-S due to the implications it will have on their sexual and reproductive life (19). Surgery forms the mainstay of the management of most cases of MRKH-S (2,13). It may vary from simple procedures such as cruciate incision for an imperforate hymen to more complex and staged surgery, such as creating a neovagina (2, 16). Further counselling after interventional surgery is recommended to ensure compliance with dilators and subsequent follow up if needed (2,19). Uterine and cervical congenital malformations are treated by either creating an endometrium-vaginal fistula using rubber or polythene tubes or re-implanting the uterine corpus the vagina (20,21). Re-implantation has been demonstrated to have favourable clinical outcomes, including term pregnancy and near-normal menstrual and sexual function (22). This involves laparotomy with resection of the atretic cervix and upper vagina (if present), re-implantation of the uterine corpus to the vagina, and finally, placement of a Foley's catheter or a stent in the uterus coming down to the vagina (16,23). Re-implantation may also be performed laparoscopically (24).

Postoperative follow-up in MRKH-S involves regular irrigation of the created tract through the stent or catheter using antibiotics and anti-inflammatory drugs for the subsequent month (16). Afterwards, the catheter is removed, and the patient is subsequently followed up for complications (23,24). Good outcomes have been reported, with even spontaneous conception and term pregnancies documented (25). In some patients, a hysterectomy may serve the patient best depending on the uterus's size and the state of the fallopian tubes. Also, risks of recurrent pelvic infections and the overall risk of endometriosis may outweigh the benefit of conserving the uterus (12).

CONCLUSION

Mayer-Rokitansky-Kuster-Hauser syndrome is a rare complex condition with sporadic occurrence; hence, the exact incidence is unknown. The presentation of MRKH-S and the age at diagnosis depends on whether the associated malformation is obstructive or non-obstructive. MRKH-S is managed in a multidisciplinary approach, and favourable clinical outcomes, including term pregnancies, have been reported.

Conflict of Interest: The authors have no conflicts of interest to declare.

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