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ORIGINAL ARTICLE

Management of Fertility and Sexual Function in Cases of Mayer-Rokitansky-Kuster-Hauser: An Overview

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ABSTRACT

Background: Mayer-Rokitansky-Kuster-Hauser (MRKH) is one of the causes of primary amenorrhea. It is characterized by the absence of uterus and upper two-third vagina. Apart from being infertile, these patients may have psychological problems and associated anomalies, which need to be addressed.

Objective: To address the management of fertility and sexual function in these patients.

Materials and methods: This has been a retrospective study conducted in the Department of Obstetrics and Gynecology, at a tertiary health care center in Ahmedabad from April 2012 to December 2015. Patients who were diagnosed with uterovaginal agenesis on sonography as a workup for primary amenorrhea were included. Other Müllerian anomalies were excluded.

Results: There were seven patients with MRKH, out of which three patients had Müllerian agenesis with no other associated anomaly, two patients had associated renal anomaly, and one patient had anal anomaly and malrotated right single kidney with absent uterus (cloaca). Six patients underwent McIndoe vaginoplasty and one underwent laparoscopic vaginoplasty. None of them had graft rejection; however, one patient had rectovaginal fistula, which was presented on 10th postoperative day. On follow-up, adequate vaginal length was seen in patients who underwent McIndoe vaginoplasty as compared with laparoscopic vaginoplasty.

Conclusion: Though these patients do not have a uterus and vagina, there is still hope that they can have good sexual life and can have their own genetic offspring.

Keywords: Cloaca, Fistula, Mayer-Rokitansky-Kuster-Hauser, Renal anomalies, Vaginoplasty.

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INTRODUCTION

Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is a rare anomaly, which is characterized by the absence of uterus and upper two-third of vagina, with normal secondary sexual characters and normal karyotype (46,XX). The MRKH syndrome can be of two types: Type I (isolated) or Rokitansky sequence, and type II or Müllerian duct aplasia, renal dysplasia, and cervical somite anomalies (MURCS) association.[1] Incomplete aplasia with or without other associated malformations are type II MRKH syndrome, which are more common.

The incidence of MRKH is 1 in 4,500 women.[2] It is considered to be sporadic in occurrence. However, in familial cases, the proposed pathology is the mutations in developmental gene (like *HOX* genes) or a limited chromosomal deletion, which may transmit as an autosomal dominant trait with an incomplete degree of penetrance and variable expressivity.[3] Apart from genital tract anomalies, there can be associated renal, skeletal, or cardiac anomalies. Although anorectal anomalies are rare to be found in patients with MRKH, rectovestibular fistula and cloacal malformations with MRKH have been described.[4]

MATERIALS AND METHODS

This is a retrospective study conducted in the Department of Obstetrics and Gynecology, at a tertiary health care center in Ahmedabad.

Inclusion Criteria

Patients who were diagnosed with uterovaginal agenesis on sonography as a workup for primary amenorrhea were included.

Methods

Secondary sexual characters were examined and Tanner staging was done. Karyotyping was done, following

which these patients underwent magnetic resonance imaging (MRI) abdomen and pelvis, echo, and X-ray spine to rule out other associated anomalies like renal anomalies (absent kidney, small kidney, horseshoe kidney), cardiac anomaly (patent ductus arteriosus), and skeletal deformities (Klippel–Feil anomaly, fused vertebrae, scoliosis). After ruling out the above-mentioned anomalies, plan for vaginoplasty was made as all these patients were married and were to start their sexual life. For fertility management, one patient underwent *in vitro* fertilization (IVF) with controlled ovulation hyperstimulation followed by transabdominal ovum pickup and surrogacy.

Following vaginoplasty, all patients were educated about change and care of vaginal mold. All these patients were followed monthly until 6 months postsurgery.

Exclusion Criteria

Other Müllerian anomalies were excluded.

RESULTS

There were seven patients with MRKH, with mean age of patients being 21.85 years. All these patients were married. These patients presented in the outpatient department (OPD) with chief complaints of primary amenorrhea. There were no complaints of abdominal pain. Secondary sexual characters of all these patients were in accordance to Tanner stage 4. Their karyotyping was also normal, 46,XX.

Out of the seven patients, three patients had Müllerian agenesis with no other associated anomaly, two patients had associated renal anomaly, and one patient had anal anomaly and malrotated right single kidney with absent uterus (cloaca). Among renal anomalies, one patient had one small kidney and one had single right kidney (Table 1).

Patient with cloaca had a history of colostomy for fecal diversion being done on 2nd day of life, followed by definitive repair and colostomy closure at 6 years of age. However, at that time, no vagina was created due to risk of vaginal stenosis, only urethral and anal opening reconstruction was done. Later on, at age of 21 years, she was present in OPD with chief complaints of primary amenorrhea. There were no urological or gastro complaints.

Out of seven patients, six patients underwent vaginoplasty by McIndoe method and one underwent laparoscopic vaginoplasty. Vaginoplasty was done with McIndoe's method using split-thickness skin graft taken from left thigh (Fig. 1). In one patient, laparoscopic peritoneal pull-through technique was done for neovagina creation. None of them had any intraoperative or immediate postoperative complications.

Table 1: Patients with MRKH and their management

Sl. no.		*Case 1*	*Case 2*	*Case 3*	*Case 4*	*Case 5*	*Case 6*	*Case 7*
1	Age	26 years	20 years	25 years	19 years	22 years	20 years	21 years
2	Education	Middle school education	Primary education	Illiterate	Illiterate	Primary education	Primary education	Middle school education
3	Marital status	Married	Married	Married	Married	Married	Married	Married
4	Duration of marriage	1 year	2 years	3 years	6 months	1.5 years	15 months	1 year
5	Uterus + vagina	Hypoplastic uterus + absent vagina	Absent uterus + absent vagina	Hypoplastic uterus + absent vagina	Absent uterus + absent vagina	Absent uterus + absent vagina	Absent uterus + absent vagina	Absent uterus + absent vagina
6	Ovaries	Present	Present	Present	Present at pelvic brim	Present	Present at pelvic brim	Present at pelvic brim
7	Associated anomaly	None	Single kidney	One small kidney	None	None	None	Cloaca with malrotated right kidney
8	Type of vaginoplasty done	McIndoe	McIndoe	McIndoe	Laparoscopic	McIndoe	McIndoe	McIndoe
9	Intraoperative complications	None	None	None	None	None	None	None
10	Postoperative complications	None	None	Rectovaginal fistula	None	None	None	None
11	Vaginal after 1 length months	8 cm	7.5 cm	7 cm	6 cm	8 cm	8 cm	7.8 cm
	After 6 months	7 cm	7 cm	6 cm	4 cm	7 cm	7 cm	7 cm
12	Postsurgery difficulty in coitus	None	None	None	Yes	None	None	None

Fig. 1: Vaginal mold being covered with split-thickness skin graft

Fig. 2: Vaginoscopic view of healthy graft

On 8th postoperative day, all patients underwent hysteroscopic examination of neovagina to evaluate graft (Fig. 2). Their vaginal mold was changed and patients were taught how to change their mold every day. There was no graft rejection in any of these patients. However, one patient developed rectovaginal fistula, which was diagnosed on the 10th postoperative day. Patient was present with complaints of stool incontinence since 2 days. There was no history of stool incontinence before surgery. On examination, small rectovaginal fistula was found 3 cm away from anal opening, which healed spontaneously.

For fertility management, one patient underwent IVF with controlled ovarian hyperstimulation, followed by transabdominal ovum pickup and surrogacy. Ovarian follicular aspiration was done by transabdominal ultrasound-guided percutaneous route with 2 to 5 MHz transabdominal probe through lower quadrant of the abdominal wall using aspiration needle (24-cm long, 17-gauge aspiration needle). Aspiration needle was briskly thrust through the abdominal wall directly into the ovary and oocytes were retrieved (Fig. 3).

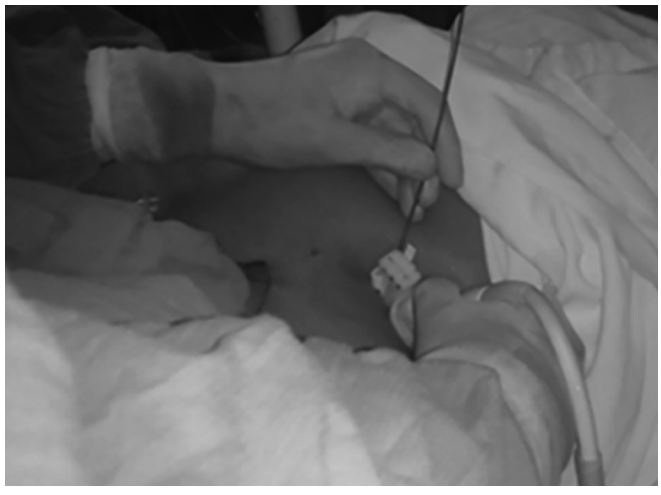

Fig. 3: Transabdominal ovum pickup done in a patient with MRKH

DISCUSSION

The MRKH is a result of developmental abnormality, i.e., aplasia of Müllerian duct, which are paired ducts that present along the lateral sides of the urogenital ridge and terminate at the sinus tubercle in the primitive urogenital sinus.[5] In females, they form the uterine tubes, uterus, cervix, and the upper one-third of the vagina, while, in males, they disappear due to the presence of antiMüllerian hormone.[6] It is the total absence of Müllerian duct development that will lead to aplasia of uterus and vagina, while the partial development which is more common will lead to tubal and partial uterine development and complete absence of upper two-third of vagina. In our study, two patients had hypoplastic uterus and the rest had uterine aplasia. Author also[7] reported the first case of vaginal agenesis in the 16th century. The term Müllerian aplasia was first coined by an American College of Obstetricians and Gynecologists Committee.[8]

In most of the cases, fallopian tubes and ovaries are normal, although ovaries can be present high up at the level of pelvic brim.[9] However, ovaries in these patients function normally. In our study, three patients had ovaries at the level of pelvic brim. Renal anomaly can be seen in 15 to 40% of cases, while skeletal anomalies can be found in 12 to 50% of cases.[10] Other anomalies like hearing defects, cardiac anomalies, anorectal anaomalies, and other anomalies can also be found. Among the mentioned associated anomalies, anorectal anomalies are rarest, especially cloaca. The incidence of cloaca is 1:50,000 live births,[11] out of which 30% of girls may have associated Müllerian anomaly.[12] One of our patients also had cloaca with MRKH, for which she underwent corrective surgery for urethral and anal opening in childhood, and vaginoplasty in adulthood.

Clinically, these patients are present with primary amenorrhea with well-developed secondary sexual characters. Occasionally, a short vaginal dimple can be seen, located 1 to 2 cm above the hymenal ring. Ultrasonography can diagnose this syndrome, but MRI is more helpful as it can diagnose other anomalies too.[13] The mainstay of treatment for MRKH is vaginoplasty, but before surgery, it is important to rule out coassociated anomalies. It is necessary to do intravenous pyelography before surgery to rule out urinary tract abnormalities. The optimum time to perform surgery is when patient is sexually active and highly motivated to use a vaginal prosthesis for several months after surgery, else surgery will fail leading to vaginal stenosis.[14] These patients also require psychological counseling and support as inability to have coitus can lead to low self-confidence and depression.[15]

Many different methods of neovagina creation have been described in the literature. The main principle is to create a space between the bladder and the rectum. Earlier passive and dilators were used for creating space. This has been replaced by surgical creation of neovagina. Various materials have used for creating neovagina. In 1892, Sneguireff[16] used distal part of rectum to create vagina, which got abandoned because of need of colostomy and contracture. Baldwin[17] and Ruge[18] used sigmoid for creating neovagina; however, these methods are not preferred due to the need for bowel surgery.

It was Abbe[19] in 1898 who first used a rubber pouch covered with a skin graft to create a neovagina. However, this method was popularized by Banister and McIndoe[20] in 1938. Until date, the McIndoe vaginoplasty using split-thickness skin graft is the most preferred method as it has a high rate of success and patient satisfaction. The main disadvantage of the McIndoe method is risk of contraction and prolonged period of postoperative dilatation, which can be overcome by using full-thickness skin graft. Patients' satisfaction following vaginoplasty ranges from 80 to 100%.[21]

With recent advances in laparoscopy, laparoscopic peritoneal pull technique is gaining popularity due to shorter operating time and reduced postoperative morbidity. The main drawback of laparoscopic vaginoplasty is reduction of the vaginal length by 30 to 50% over a period of 1 year.[22]

Postoperative complications like infection, hemorrhage, and graft failure can occur. Serious complications like enterocele and postoperative fistula (4% risk) have also been reported.[23] One of our patients also developed fistula, which healed spontaneously. Contracture of the graft and the development of excess granulation tissue can also occur.[23]

Postoperatively, patient is advised bed rest for 1 week and is prescribed antibiotics and a low-residue diet. After 1 week, the labial sutures are removed, and vaginal mold is removed with the patient under mild sedation. The neovagina is irrigated with warm saline solution and carefully inspected to determine whether the graft is healthy or not, following which a new form covered with a sterile sheath is reinserted. Patient is counseled regarding vaginal mold replacement. The mold is to be worn continuously for 6 weeks and is removed only for urination and defecation. Low-pressure douches with warm water are to be done daily and mold should be cleaned with a povidone–iodine solution, covered with a fresh condom, lubricated, and reintroduced into the neovagina. After 6 weeks, a silicone mold can be used at night for the next 12 months. In most of the cases, the vagina is functional 6 to 10 weeks postsurgery.[23]

In special cases like cloacal with MRKH, fecal diversion and urinary diversion are done first, followed by definitive repair later on.[11] In some cases, vaginoplasty can be combined with definitive repair, but vaginoplasty at an early age is associated with risk for vaginal stenosis, requiring redo surgery.

As far as fertility is considered, these patients can undergo IVF surrogacy. Ovaries in these patients can be high up at the level of pelvic brim, making transabdominal ovum pickup a more feasible approach alternative to laproscopy. A study by Barton et al[24] demonstrated that number of mature oocytes retrieved by laparoscopy and transabdominal approach was comparable. There was no increase in damage to oocytes noticed with transabdominal route.

CONCLUSION

The MRKH syndrome is a spectrum of congenital abnormalities of the vagina and the uterus with varying anatomical presentation in a phenotypically and genotypically normal female. Management includes psychological support and the creation of a neovagina for sexual function. Pshychosexual counselor has a vital role to play. Advances in laparoscopy have widened the horizon. As this condition is relatively rare, centralizing the care of such women in centers of expertise may improve patient care and help with future research into etiology and management.

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