CASE REPORT

Mayer-Rokitansky-Kuster-Hauser Syndrome: Surgical Management of Two Cases

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ABSTRACT

The Mayer-Rokitansky-Kuster-Hauser (MRKH) Syndrome is a rare anomaly characterized by congenital aplasia of the uterus and vagina in women showing normal development of secondary sexual characters and normal 44 XX karyotype. We report our experience in the management of two patients with congenital absence of the vagina due to the MRKH syndrome. The first case was a 24-year-old student, who presented with primary amenorrhea, uterovaginal agenesis, right pelvi-ureteric junction obstruction, and left renal agenesis. The second patient was a 24-year-old housewife, who presented with primary amenorrhea and inability to achieve penetrative sexual intercourse. She had vaginal atresia and a grossly hypoplastic uterus. Both had successful sigmoid colovaginoplasty and are sexually active. Vaginal reconstruction using the sigmoid colon saw an immediate and satisfactory outcome in both patients

Key words: Mayer-Rokitansky-Kuster-Hauser, syndrome, surgical management

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INTRODUCTION

The Mayer- Rokitansky-Kuster-Hauser (MRKH) Syndrome is a rare anomaly characterized by congenital aplasia of the uterus and vagina in women showing normal development of secondary sexual characters and normal 44 XX karyotype.[1] The uterine and vaginal agenesis in these karyotypic females may be accompanied by urogenital, skeletal or dental defects and other dysmorphias.[2-6] It is a rare disorder occurring in about 1 in 4000 to 5000 female births and is a common cause of primary amenorrhea.[7]

The diagnosis is often made during adolescence in the context of primary amenorrhea with normal puberty.[8] The cause of this syndrome is still not completely understood, but the frequent association of other malformations involving the kidneys, skeleton, and ears suggests the involvement of major developmental genes, such as those of the homeobox (HOX) family. These genes are known to play a crucial role during embryogenesis, particularly in the axial skeleton, the hindbrain, as well as, during urogenital differentiation.[9]

Treatment of the MRKH syndrome consists of creating a neovagina, which can be offered to patients when they are emotionally mature and ready to commence sexual activity. Treatment may either be surgical or non-surgical, but the chosen method needs to be tailored to the individual needs, motivation of the patients, and the options available.[1,10]

We report our experience in the management of two patients with congenital absence of the vagina due to the MRKH syndrome.

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CASE REPORTS

Case 1

FAA was a 24-year-old student, referred to our unit with complaints of primary amenorrhea. There was no history of associated periodic lower abdominal pain or urinary symptoms. She was said to have had delayed development of secondary sexual characters compared to her siblings, but no history of sexual exposure. The patient had no significant past medical history and no history of a similar problem in the family.

Examination showed a young female patient with normal secondary sex characters and well-developed breasts (Tanner stage 4). She had normal axillary and pubic hair distribution and had no palpable abdominal masses. Vaginal examination showed normal vulva and urethral meatus, but a short (3 cm) blind ending vagina [Figure 1].

An abdominal ultrasound scan showed an enlarged right kidney, with a moderate hydrocalicosis and hydroureter. The left kidney was not visualized. The uterus was not demonstrated. Intravenous urography showed a low

lying right hydronephrotic kidney and pelvi-ureteric junction (PUJ) obstruction. Computerized tomography (CT) confirmed the absence of a uterus, vagina, and left kidney, as well as, the presence of a right kidney and PUJ obstruction [Figures 2 and 3]. Serum electrolytes and creatinine levels were within normal limits.

The patient had initial Anderson-Hynes dismembered pyeloplasty, with an uneventful postoperative recovery.

The patient was counseled on the nature of the anomaly and the options available as well as the prospects of child bearing. Six months later, she had sigmoid colovaginoplasty [Figures 4-6]. She was subsequently discharged and later got married. Postoperative follow up visit at one year and subsequent telephone communication indicated normal and satisfactory sexual intercourse with her spouse.

Case 2

MM was a 24-year-old housewife who presented with complaints of primary amenorrhea, and inability to achieve penetrative sexual intercourse. There was no history of cyclical lower abdominal pain or urinary symptoms.

She had delay in the development of secondary sexual characters compared to her siblings. There was no family history of a similar illness; there was no significant past medical history. Physical examination revealed a young female patient with normal secondary sex characters; well-

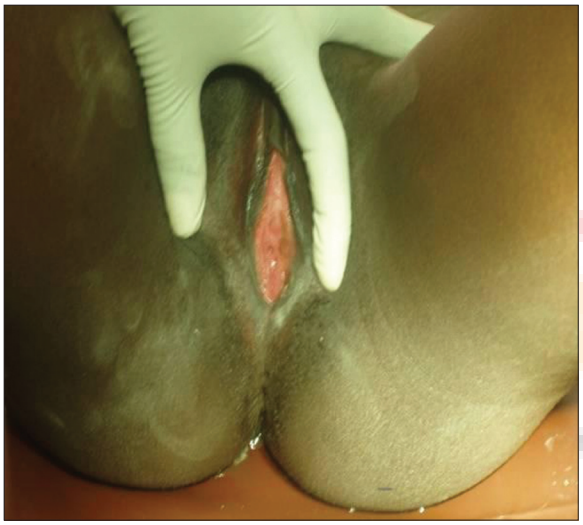

Figure 1: Preoperative picture of Case no.1 showing blind ended vagina

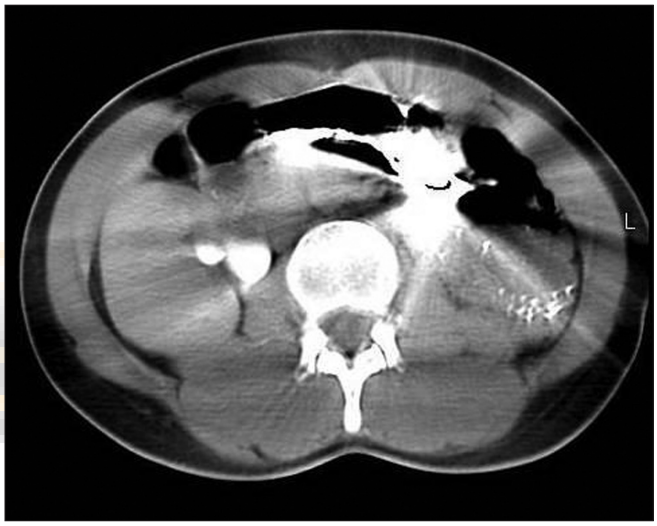

Figure 2: Computerized tomography scan of the first patient showing absence of left kidney, and PUJ obstruction of right kidney

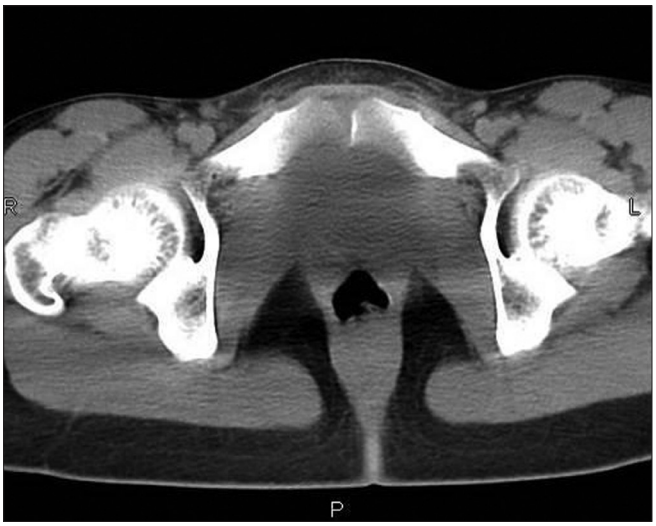

Figure 3: Computerized tomography scan of the first patient showing absence of uterus

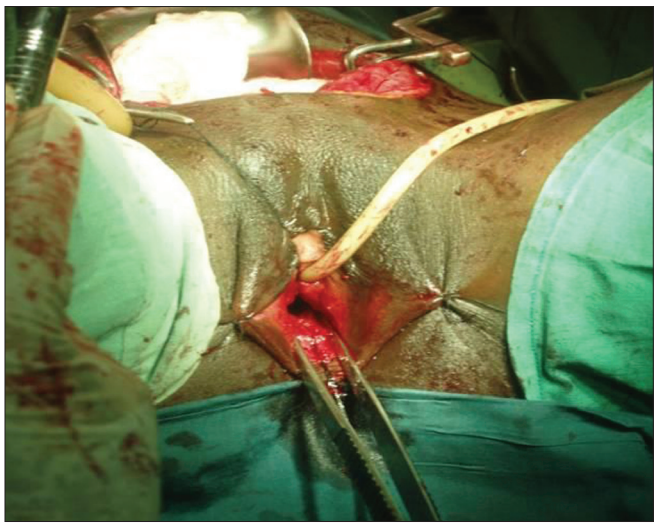

Figure 4: Intraoperative picture showing the prepared channel and distal vaginal segment ready for anastomosis with the colonic segment, to create a neovagina

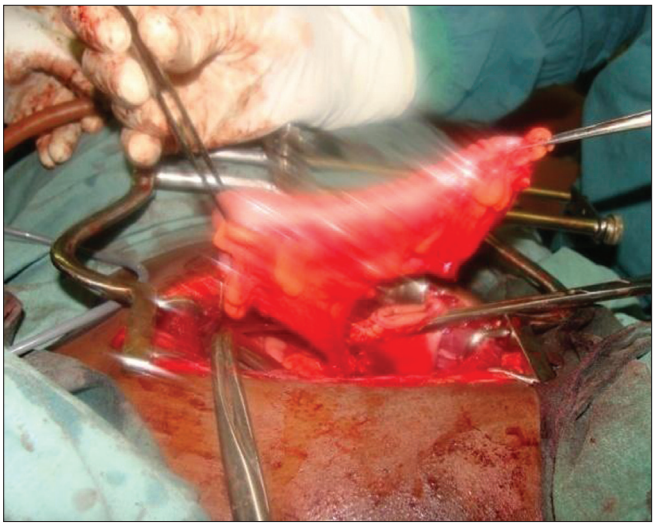

Figure 5: Colonic segment prepared for vaginal reconstruction

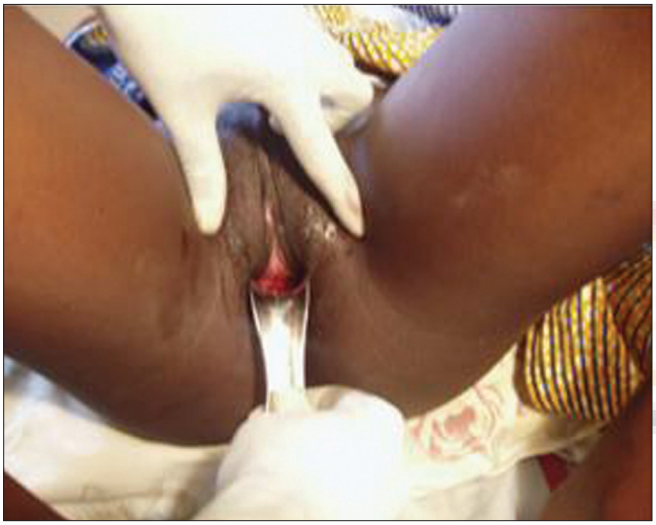

Figure 6: Postoperative speculum examination of the first patient showing adequacy of the neovagina

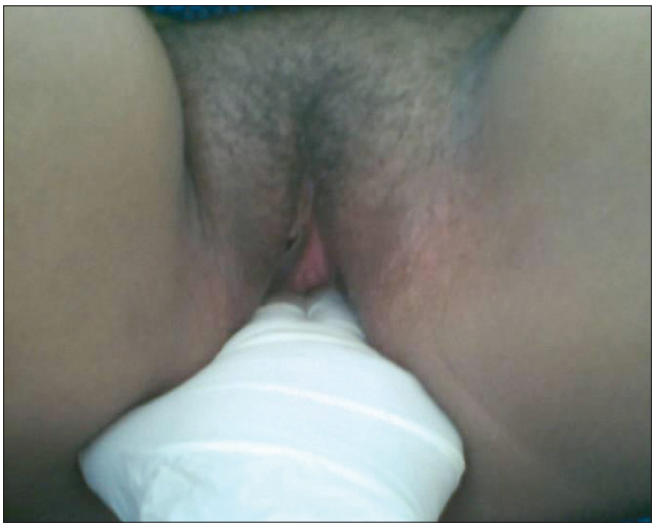

Figure 7: Postoperative picture of Case no 2, accommodating two fingers

developed breasts (Tanner stage 4), normal axillary hair and female pubic hair distribution; there were no palpable groin or intra-abdominal masses. Vaginal examination showed normal vulva and urethral meatus, but a blind ending vagina (about 3cm).

An abdominal ultrasound showed a rudimentary uterus and follicular cyst. The kidneys and other intra-abdominal organs were normal. Laparoscopy showed a hypoplastic uterus and fallopian tubes. Serum creatinine, urea, and electrolytes were normal.

The patient received three courses of estrogen and progesterone, to assess the functional state of the uterus, but there was no evidence of increase in the uterine size. She was counseled on the nature of the anomaly and the treatment options available, as well as, the prospect of child bearing. She had excision of the grossly hypoplastic uterus and successful construction of a colovagina [Figure 7]. The lady was discharged on the fifteenth postoperative day, and counseled to resume coitus three months after surgery. Subsequent follow-up indicated normal and satisfactory sexual intercourse with her husband.

DISCUSSION

The MRKH syndrome is a rare, but widely discussed developmental failure of a part of, or the whole Mullerian duct. Patients with this syndrome have a 46XX karyotype, normal female external genitalia, normal ovarian function, partial or incomplete absence of the vagina, and an absent or hypoplastic uterus with bilateral non-canalized tubes. While most patients present with the typical absence of the uterus and vagina, few present with the atypical form, where, in addition they have an asymmetric uterine remnant and abnormalities of the fallopian tubes.[6] These patients suffer severe distortion of the body image, anxiety, depression, interpersonal sensitivity, and face a lot of psychological distress at diagnosis.[2] In our environment, where gender role and identity are very crucial, these patients and families face difficult times.

Diagnosis is frequently made clinically, but often confirmed either radiologically or laparoscopically, in patients whose hormonal and karyotypic investigations for primary amenorrhea are normal. The use of ultrsonography, intravenous urography, computerized tomography, and magnetic resonance imaging provide information about the degree of abnormality and associated renal anomalies.[2,11-13]

Management of vaginal agenesis in the MRKH syndrome remains controversial. The choice of procedure and patient

age at reconstruction depend upon individual anatomy, fertility potential, and psychological and social factors.[2] The ideal timing for intervention is at or after adolescence, when the woman has reached physical and psychological maturity. In the past, vaginal reconstruction procedures were performed on infants and pre-pubertal girls and this required inevitable surgical revision in adolescence before sexual activity. Deferring treatment allows the woman herself to be involved in the decision making and also increases compliance with the adjuvant dilatation therapy that may be required.[14]

The goals of long-term treatments are to create a functional neo-vaginal canal with an adequate diameter and length, appropriate axial direction, and normal secretion/lubrication to accommodate sexual intercourse and to address the issue of fertility.[15,16]

There are two main types of procedures; the first consists of the creation of a new cavity and can be done surgically or non-surgically. The second is vaginal replacement with the pre-existing canal lined with a mucous membrane (a segment of bowel).[1] The most commonly used non-surgical procedure is the Frank's dilatation method, which involves the application first by the clinician and then by the patient of vaginal dilators, of progressively increasing length and diameter, and also Ingram's technique and its modifications.[14-17]

Vaginal dilators have few complications as there are no anesthetic or surgical risks, but it is time consuming, causes the patient discomfort, and requires good patient motivation.[14]

Surgical treatment of the MRKH syndrome is achieved by vaginal reconstruction, which includes; Williams vaginoplasty, which involves suturing the labia majora into a perineal pouch, but the vagina created is external, short, and unsatisfactory for penetrative intercourse; this procedure is no longer practiced. The Vecchietti procedure consists of increasing the vaginal size by gradually applying traction to the vaginal wall. Finally, the neo-vagina can be created within the rectovesical space and lined by different tissues such as skin (McIndo-Reed), peritoneum (Davydov), and intestine.[14]

Reconstructing the vagina using intestinal segments creates an aesthetically pleasing vagina, does not require moulds, dilatation or lubrication, and in children, the neo-vagina grows with the child with less risk of stenosis.[18]

The sigmoid colon has certain advantages, such as, a thick wall, large diameter, does not traumatize easily, has adequate mucosal secretion, which although adequate for lubrication is not excessive or irritating, and does not require regular dilatation after the postoperative period.[8,19]

All our patients were treated surgically by colovaginoplasty using the sigmoid colon. Both had an uneventful postoperative recovery and were satisfied with the immediate outcome.

In conclusion, the MRKH syndrome is a rare anomaly. Various options of surgical treatment have been practiced by many surgeons. Vaginal reconstruction using the sigmoid colon has given a good, immediate outcome in this report. Long-term follow-up will be needed to assess the long-term outcome, with regard to complications, sexual satisfaction of patients and spouses, as well as, their adaptation and resolution of fertility issues.

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