



## Case Report

### MAYER-ROKITANSKY-KUSTER-HAUSER (MRKH) SYNDROME – A CASE REPORT

MK ISLAM<sup>1</sup>, MS ALAM<sup>2</sup>, A HANIF<sup>3</sup>, MK HASAN<sup>4</sup>, M RAHMAN<sup>5</sup>

#### Abstract:

We report a case of Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome in a 25 year old married lady. Though it is a congenital abnormality, she presented as primary infertility after 2 year of her marriage and was referred to us from a Gynaecologist. She had absent vagina, rudimentary uterus with no cervix. Her ovaries were severely hypoplastic. The anus was placed anteriorly and opened into the vulva. In spite of absence of her vagina, the lady somehow maintained her married life by doing intercourse through the anteriorly placed rectum. The vagina was made from the lower end of existing rectum which opened into the vulva. The proximal end of the rectum and left colon were pulled through the pelvis and opened into the perineum. It improved her quality of life.

#### Introduction:

Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is characterized by Mullerian duct structures agenesis, in genetically, phenotypically and developmentally normal (46XX) females. Though it is a congenital abnormality but often diagnosed after puberty as a case of primary amenorrhoea or infertility. However, the etiology of MRKH syndrome still remains unclear. For a long time the syndrome has been considered as a sporadic anomaly, but increasing number of familial cases now support the hypothesis of a genetic cause. In familial cases, the syndrome appears to be transmitted as an autosomal dominant

trait with incomplete penetrance and variable expressivity. This suggests the involvement of either mutations in a major developmental gene or a limited chromosomal imbalance. Treatment of vaginal aplasia, which consists in creation of a neovagina, can be offered to allow sexual intercourse. The question of fertility is not solved. As psychological distress is very important in young women with MRKH syndrome, it is essential for the patients and their families to attend counseling before and throughout treatment. However, it is a very good example where the Pediatric surgeons have to operate on a grown up person to improve the quality of life.

#### Case Report:

A 25 year old married garment worker was admitted to the Gynaecology department with infertility. Upon further interview, she confirmed that she has been presenting primary amenorrhea along with the infertility. Examination revealed that there was no anus in the perineum with a normal looking vulva having urethral opening and vaginal opening though which stool comes out. So, apparently the lady was diagnosed as a case of Imperforate anus with Recto-vaginal fistula. Subsequently, the patient was referred to the Pediatric Surgery department for correction of ARM.

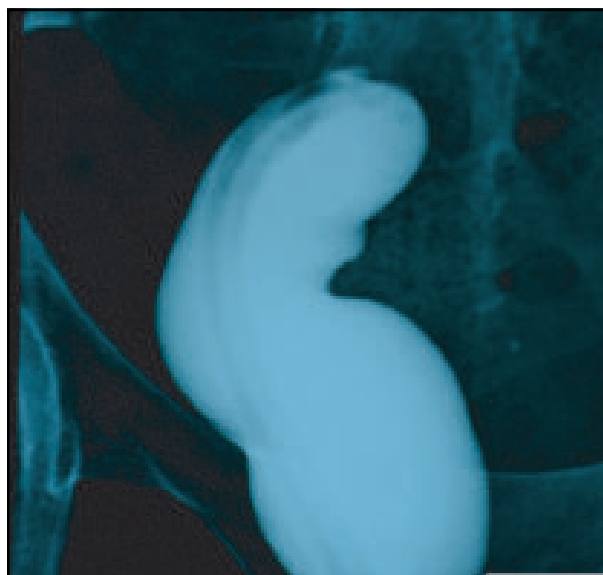
Thorough physical examination revealed a normal female phenotype, normal labia majora, minora and well developed clitoris. The cervix was not visualized on speculum examination and the suspected vaginal orifice was the rectum opening into the vulva as fecal matter was revealed from it. Surprisingly the girl maintained her 2 year married life through that opening and her husband complains of getting occasional fecal odor after copulation. The sacrum was complete and rest of the spine was normal. Bladder was not distended or expressible. Neurological examination was within normal limits.

1. Prof. Md. Kabirul Islam, Consultant, Pediatric Surgery Square Hospital Ltd.
2. Dr. Md. Shah Alam, Assoc. Professor, Dept. of Pediatric Surgery, Dhaka Medical College Hospital, Dhaka
3. Dr. Abdul Hanif, Assistant Professor, Dept. of Pediatric Surgery, Dhaka Medical College Hospital, Dhaka
4. Dr. Md. Kamrul Hasan, Assistant Registrar, Dept. of Pediatric Surgery, Dhaka Medical College Hospital, Dhaka
5. Dr. Mizanur Rahman, M.S. (Final part) student. BSMMU.

**Correspondence to:** Prof. Md. Kabirul Islam, Consultant, Pediatric Surgery, Square Hospital Ltd, 18 F, West Panthapath, Dhaka-1205, E. mail: kabirul.islam.bd@gmail.com

Her haemogram was found normal. Urine showed absence of any infection. Blood urea and serum creatinine values were also normal. Transabdominal ultrasound scan of the abdomen and pelvis confirmed rudimentary uterus with absence of cervix and vagina. Both the ovaries were situated in the pelvis which were hypoplastic. There was no anomaly in the urinary tract detected. The karyotyping showed 46XX but her hormonal levels were not measured. Considering these findings this patient was identified as a case of Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome.

As the rectum opened within the vulva, we decided to convert the lower end of rectum as the vagina. So, the lower end of the rectum was divided through a laparotomy at the level of about 25 cm proximal to the lower end and closed in layers, which gave the appearance of vagina. The proximal part of the rectum and colon were pulled through the levator muscle complex behind the uterus and opened into the site of anus in the perineum. As there was no cervix and the uterus was rudimentary, connection between the new vagina and the uterus was made. The lady was recovered uneventfully after 7 days post operatively. Regular anal dilatation with Hager's dilator was advised. After 3 months, she was found to have a good vaginal opening in the introitus (Figure – 1). But she had moderate amount of fecal incontinence due to anal stenosis. However, the incontinence was improved by systematic regular anal dilatation.



**Fig.-1:** Contrast X-ray of new vagina made from lower end of rectum.

### Discussion:

Mayer-Rokitansky-Kuster-Hauser(MRKH) Syndrome results from a failure of the mullerian ducts to reach the urogenital sinus due to a disorder involving the ureterovaginal canal or the vaginal plate. It appears to be a sporadic polygenic multifactorial disorder occurring between 4th and 12th weeks of gestation<sup>1</sup> as no specific gene could be identified yet. In 1829, Mayer first reported a case of stillborn female with multiple congenital anomalies; vaginal agenesis was one of them. But he could not explain the detail of the disease. In 1838 Rokitansky and in 1910 Kuster further described this clinical entity and recognized the presence of a rudimentary uterus with normal ovaries and normal external genitalia. Finally in 1961, Hauser and his coworkers described the frequent association of renal and skeletal anomalies. Thus, the combination of names as Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is used to describe this disorder.

The incidence is usually 1 in 4000 to 5000 live female births<sup>2</sup>. The patients are typically 46, XX female with normal secondary sex characters that commonly present at puberty with amenorrhoea as in our case. Uterine development in these patients may vary from normal to the more characteristic rudimentary bicornuate structure without a lumen. Sometimes they present with cramping lower abdominal pain due to rudimentary functioning endometrium. Like in our case, the vagina is completely absent in 75% cases and in 25% have a very short vaginal pouch(less than 2.5 cm.). Most patients have both fallopian tubes and ovaries. However, unilateral and bilateral gonadal agenesis is also observed (70,100). They are also associated with anomalies of urinary tracts and skeletal system<sup>3</sup>. But in our patient no such associated abnormality was noted.

Most of the diagnosis is made by the symptom of amenorrhoea at the time of puberty, which includes detection of vaginal agenesis. In our patient, amenorrhoea did not get much importance and the girl got married. She used to maintain her sex life by intercourse through the rectum as it was anteriorly placed within the vulva. Her major complain was infertility which has made the case interesting. However, in infants and older patients, a complete physical examination including vaginoscopy and cystoscopy, CT scan, MRI and laparoscopy are the best means of making a diagnosis<sup>4</sup>.

The treatment MRKH syndrome depends upon the anatomy of the individual patients. Functional, reproductive, and psychological issues must be carefully evaluated and addressed, taking into account both the physical and intellectual maturity of the patients<sup>5,6</sup>. The goal of therapy is to provide adequate sexual function and deal with the psychological impact that the patient has no uterus or vagina. Fertility is possible and is an option that should be offered to the patients because both the ovaries are usually normal and successful in vitro fertilization with surrogate pregnancy has been achieved<sup>7,8</sup>.

However, psychological studies have shown that infertility rather than expected difficulty with intercourse was the most problematic issue for MRKH syndrome patients to deal with<sup>5,9</sup>. Proper perioperative counseling of the patient and family members is essential. The timing of the procedure is also important. If the uterus is present and the vaginal remnant is too short to reach the perineum, construction of a vagina that will communicate with the uterus should be undertaken before the onset of menstruation<sup>10</sup>. If only the distal portion of vagina is absent, posterior or lateral skin flap can be mobilized and rotated into the proximal portion of the vagina to provide an introitus. This however, is difficult to perform in most cases, and long term result varies. If no uterus is present, construction of a vagina should be undertaken just before the initiation of sexual activity (Ravitch). Ideally neovagina should be located appropriately, be of adequate dimensions, be lined by elastic tissue, neither be constantly moist nor malodorous, be hairless, and be sensitive at least at the introitus<sup>11</sup>. The ideal procedure has been elusive, and there are differences of opinion among various authorities concerning the techniques<sup>5,12,13</sup>. A number of operative procedures have been advocated for creation of the neovagina, including insertion of a skin-covered vaginal mold (Abbe-McIndoe procedure), various fasciocutaneous and myocutaneous flaps vulvovaginal flap, labial flaps using tissue expanders, full thickness skin grafts with vacuum-assisted wound closure and use of amnion and peritoneum<sup>14-24</sup>. There are some recent reports of laparoscopic approach to fashion a neovagina with peritoneum<sup>25</sup>. However, bowel vaginoplasty using the distal part of colon, caecum or small gut have gained popularity. However, with the help of laparoscope, these procedures have become much easier to perform with less complication. In our

case as the uterus was found rudimentary and as the rectum opened into the vulva, we planned not to disturb the anatomy of the vulva. So, after laparotomy, the lower part of rectum was conserved and the proximal part was mobilized and pulled through the levator ani muscle complex and was placed into the perineum at the site of anal canal. So, there was anus in the posterior and the large gut into the vulva looked like a vagina. Due to very rudimentary uterus, there was no secretion from it. So, hopefully it will not cause any problem.

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