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**Case Report**

# Mayer-Rokitansky-Küster-Hauser syndrome type II: a case report and literature review

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## ABSTRACT

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a genetic disease consisting of absent uterus and upper part of vagina, and is seen in women with a normal karyotype (46, XX). It is one of the rare diseases where the estimated prevalence is 1:5000. Most patients present with primary amenorrhea at a young age, with or without bone or kidney abnormalities. The associated psychological aspects of this disease often enforce the need for a prompt diagnosis and treatment. The preferred method for preoperative evaluation of these patients is magnetic resonance imaging (MRI).

**Keywords:** MRKH syndrome, Primary amenorrhea, MRI

## INTRODUCTION

Müllerian agenesis, commonly known as Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome, is a controversial disease affecting women with normal karyotype. It is characterized by poor development or absent uterus and upper part of vagina (46, XX). Most patients have normal external genitalia, early breast development and normal hair growth, and primary amenorrhea during puberty. It is the second most common cause of primary amenorrhea after ovarian failure. It can often be associated with abnormalities affecting the bones and kidneys.[1]

## CASE REPORT

A 17-year-old female presented to the gynaecology department with primary amenorrhea. There was no history of any significant systemic illness or surgery in the past. Vitals were within normal limits. Per abdomen examination revealed soft and non-tender abdomen. Gynaecological examination revealed normal external genitalia, secondary sex characteristics, and hair growth, with short blind ending vagina. Biochemical parameters viz., follicle-stimulating hormone (FSH) and luteinizing hormone (LH) were within normal limits. She was referred for a pelvic magnetic resonance imaging (MRI) with the provisional diagnosis of utero-vaginal agenesis. MRI pelvis revealed aplastic uterus with non-visualisation of upper 2/3$^{rd}$ of vagina (Figure 1). Bilateral ovaries were normal (Figure 2). She was further evaluated for extragenital abnormalities using NCCT whole spine which revealed absent left kidney (Figure 3). No vertebral anomalies were found. Based on the MRI findings, a diagnosis of MRKH type II was made.

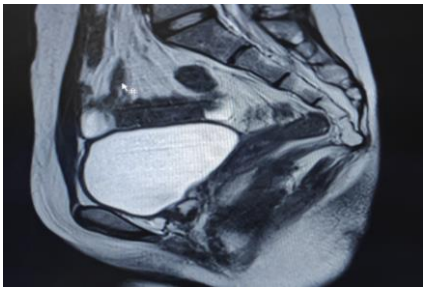

**Figure 1: Aplastic uterus with non-visualization of upper 2/3rd of vagina.**

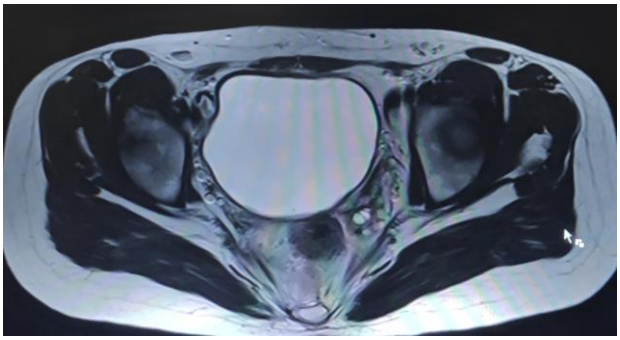

**Figure 2: Axial MRI pelvis (T2 weighted sequence) showing normal bilateral ovaries.**

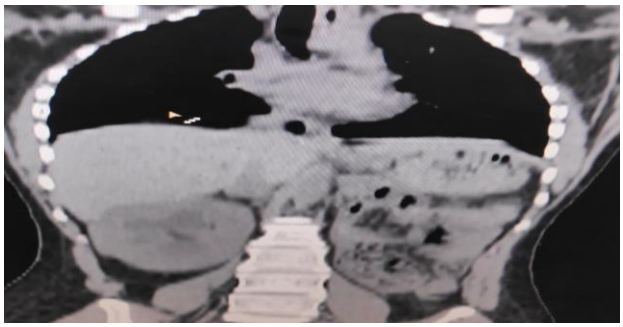

**Figure 3: NCCT spine coronal section showing absent left kidney.**

## DISCUSSION

It is generally thought that 1 in 5,000 female babies has MRKH syndrome.[2] However, there are currently only two population studies on the incidence of MRKH syndrome.

**Table 1: Population based studies regarding prevalence of MRKH syndrome.**

| Study | Cohort | Period | Place of study | Prevalence |
|---|---|---|---|---|
| **Aittomäki et al[2]** | 161 patients | 1978-1993 | Finland | 1 in 4961 newborn girls |
| **Herlin et al[3]** | 138 patients | 1974-1996 | Denmark | 1 in 4982 live female births |

### *Classification*

MRKH syndrome is classified into two groups. Type I (isolated) with no extragenital abnormalities and type II (may have MURCS association) with extragenital abnormalities.[4]

### *Embryology*

MRKH syndrome is caused by complete underdevelopment of the paramesonephric ducts (which begin to form around the 5th-6th weeks of pregnancy) that form the uterus and bladder.

### *Etiopathogenesis*

Multifactorial ranging from somatic event to autosomal dominant pattern.[5,6]

### *Genetic factors*

The 17q12 locus which includes LHX*1* and *HNF1B*, 16p.11.2 locus which includes *TBX6* gene.[4] Diagnostic workup of MRKH syndrome consists of- clinical history: primary amenorrhea, dyspareunia, abdominal pain; gynecological examination: Normal height, secondary sex characteristics, and hair growth, normal external genitalia, blind-ending vagina on palpation; ultrasound (transvaginal/transabdominal): absence of uterus or vaginal canal, two functional ovaries, kidney abnormalities; evaluation of other associated malformations (such as EOS scan, ENT assessment and echocardiography); pelvic MRI: the gold standard for diagnosis and early evaluation; biochemical markers: FSH, LH, estradiol supposed to be of normal range; and chromosome analysis: 46, XX.[4]

### *Associated malformations*

Associated malformations are given in Table 2.[4]

### *Psychosexual impact of the disease*

Especially in developing countries like India, this disease causes profound psychological effect on the individual. Due to traditional Indian culture, people with this condition often struggle with intense anxiety about sex and reproduction and may experience social stigma. Patients should be advised to seek good mental health services that can help them overcome their fear of social rejection. Various tests such as the feminine functioning scale (FSFI) and the feminine depression scale-revised (FSDS-R) are used after neovaginal treatment. Ernst et al investigated the factors that motivate and encourage disclosure, how the diagnosis affects patients' minds, and how the diagnosis affects their lives.[7] The World Health Organization calls for a positive attitude in this regard that reflects personal fitness and overall health including mental fitness.[8] Patients should be encouraged to seek help, confide in people they can trust.

### *Management*

Management of MRKH syndrome is shown in Table 3.[4]

### *Differential diagnosis*

Imperforate hymen or transverse vaginal septum- can be confirmed by vaginal exam and on imaging which will reveal collection in the cervix which is T1 moderate to high signal and T2 hyperintense (hematocolpos).

**Table 2: Malformations associated with MRKH syndrome.**

| Variables | Malformations |
|---|---|
| **Gynecological (48-95%)** | Uterine remnant, endometriosis, Herlyn-Werner-Wunderlich syndrome, leiomyoma, adenomyosis, inguinal Müllerian duct hernias |
| **Ovarian (5-10%)** | Unilateral agenesis, ectopic ovary, polycystic ovary, streak ovary, and tumors |
| **Renal (most common extragenital manifestation) (30-40%)** | Unilateral renal agenesis, pelvic kidney, duplex kidney, and horseshoe kidney |
| **Skeletal (10-40%)** | Scoliotic deformity, Klippel-Feil anomaly, hemivertebra, rib agenesis |
| **Cardiac (<5%)** | Pulmonary valvular stenosis, atrial septal defects |
| **ENT (<5%)** | Sensorineural and conductive hearing loss (such as atresia of external meatus, ankylosis of the stapes) |
| **Others** | VACTERL |

**Table 3: Management of MRKH syndrome.[4]**

| Variables | Management |
|---|---|
| **Dilation therapy (first line)** | Self-dilation (Frank's method-progressive dilation with the help of dilators for 10–30 min one to three times a day); dilation by intercourse (d'Alberton's method) |
| **Vaginoplasty** | McIndoe vaginoplasty (split-skin graft covering a mold placed in the dissected pouch between the rectum and bladder); Baldwin vaginoplasty (bowel graft); Davydov vaginoplasty (peritoneal graft)<br>Williams vulvavaginoplasty (labia majora flaps) ; laparoscopic Vecchietti vaginoplasty (subperitoneal threads are fastened to a mold in the vagina to create a surgical traction device that is positioned on the anterior abdominal wall) |
| **Uterine transplantation for infertility** | Uterine transplantation for the treatment of infertility is still in its early stages. It is usually performed through a midline incision below the umbilicus. During dissection, the anus and bladder are separated from the vagina. The external iliac vein and artery are then separated from the surrounding tissue and cleaned to make room for anastomosis. The round ligament, cardinal ligament, and sacral ligament are all tied with fixed sutures. After that, the uterus is flushed and placed in the pelvis The internal iliac segment of the uterine artery of the graft is connected to the external artery of the recipient during end-to-side anastomosis. After reperfusion, the uterus is fixed to the ligaments, the vagina is opened and vaginal anastomosis is performed. |
| **Prospects in the future** | Robotic assisted minimally invasive surgery |

## CONCLUSION

The spectrum of MRKH syndrome is extensive, necessitating a multispecialty approach to diagnosis and treatment. For a rare entity like this, MRI is a great modality to use as it helps rule out any other related illnesses. The disease's significance in a developing nation like India lies in the fact that care is often delayed because of the psychosocial effects of the illness and social stigma. The clinician's job should be not just to treat the illness but also to help the patient cope with it, by giving them the right advice. Nonetheless, the various advancements in this discipline over the years have been crucial in enhancing patient care.

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