



## Case Report

## Type 2 Mayer-Rokitansky-Kusner-Hauser syndrome

Manju Agarwal<sup>1</sup>, Somya Mathur<sup>1,\*</sup><sup>1</sup>Dept. of Obstetrics and Gynaecology, S.H.K.B.M. Hospital and Jhalawar Medical College, Jhalawar, Rajasthan, India

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## ABSTRACT

A 28-year-old married nulligravida presented with primary amenorrhea. There was no significant family history of similar complaints. Her secondary sexual characters were well developed with hypoplastic vagina. Ultrasonography of abdomen and pelvis revealed the absence of uterus and the right kidney. The patient had normal 46, XX Karyotype.

She was diagnosed as a case of Type 2 MRKH after diagnostic laparoscopy. MRKH is very commonly associated with renal agenesis. It had a huge psychological impact in the women and her family. The patient was advised vaginoplasty, and surrogacy or child adoption as an option.

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## 1. Introduction

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a condition with incidence of about 1 in 4500 female births, characterized by aplastic uterus and the upper part of vagina, but with presence of normal secondary sexual characteristics. The karyotype is 46, XX. It is the second most common cause of primary amenorrhea after gonadal dysgenesis. This disorder can be broadly categorized into 2 types, type I – isolated, in which there is congenital aplasia of the uterus. MRKH Type 2 is uterine aplasia, with renal, skeletal and/or hearing defects. Cardiac abnormalities are very rare. It is known as, Genital Renal Ear Syndrome, if the middle ear is also affected.<sup>1</sup> Urinary tract is the most commonly affected, in which, unilateral renal agenesis, ectopic kidney, horse-shoe kidney are seen. MRKH has a wide range of etiology.

The development of the female reproductive tract is from the Mullerian ducts, the fallopian tubes, uterus, cervix, and upper part of vagina are derived from it, any disturbance in the development of the Mullerian structures can lead to congenital defects in the female genital tract, which have variable expression and severity.<sup>2</sup>

\* Corresponding author.

E-mail address: [mathursomya@gmail.com](mailto:mathursomya@gmail.com) (S. Mathur).

## 2. History and Classification

Mayer, a German anatomist in 1829, first to described a patient with vaginal agenesis and a rudimentary uterus, which he called 'uterus bipartitus'. In the next few centuries, Rokitansky (1838), Kuster (1910) and Hauser (1961) further described the syndrome, and so it was called Mayer-Rokitansky-Kusner-Hauser Syndrome.<sup>3</sup>

It is classified as:

1. Typical MRKH: Aplastic uterus and vagina with normal Ovaries, fallopian tubes and renal system.
2. Atypical MRKH: Aplastic uterus and vagina associated with malformations of ovaries, fallopian tubes, and urinary system.

## 2.1. Etiopathogenesis

MRKH syndrome has a sporadic occurrence, familial cases have been seen which suggest that in some patients MRKH maybe an inherited disorder.

It is inherited as an autosomal dominant syndrome, and has incomplete penetration and variable expressivity.<sup>2</sup>

### 3. Clinical Report

A 28-year old nulligravida married patient presented in the OPD of SHKBM, Jhalawar with the complain of spotting per vaginum. On taking her history, she had primary amenorrhea with dyspareunia, and there was no spotting per vaginum. On examination, her developmental milestones and secondary sexual characters were normal. She was admitted for further evaluation. No significant medical or surgical history was present. The menarche was seen at around 11 years and the pubic and axillary hair developed at 14 years of age. Both her sisters had attained menarche at the age of 12 years.

Her height was 155 cm and on examination had normal appearing external genitalia and breasts were well developed.

On per abdominal examination- abdomen was soft, with no palpable mass.

On local examination- the external genitalia appeared normal, with normal distribution of pubic hair.

On per speculum examination-the entire length of the speculum could not be introduced, as the vaginal length was very short.

On per vaginal examination- The vaginal orifice was 3-4 cm in length, with a blind ending.

On karyotyping, she was 46XX.

Ultrasonography showed a single kidney with absence of uterus and bilateral normal ovaries.

On CT scan of abdomen and pelvis, a single kidney was identified, bilateral ovaries were seen, uterus could not be visualised.

She was further evaluated by doing diagnostic laparoscopy in which it was found that, there was uterine aplasia with normal sized bilateral ovaries.

She was diagnosed as a case of Type 2 MRKH syndrome.

### 4. Discussion

MRKH syndrome is subdivided into two types: type I or isolated MRKH and Type II which is associated with other abnormalities like renal agenesis, skeletal deformities. MRKH syndrome is the second most common cause of primary amenorrhea. The women presents with primary amenorrhea with normal development of the secondary sexual characteristics and normal external genitalia, the ovaries are developed normal as seen on imaging, and karyotype is 46, XX, without visible chromosomal anomaly.

In our case the patient reported as having primary amenorrhea, she had a blind vagina, with normal secondary sexual characters and genotype 46 XX. She had aplasia of uterus with single kidney and bilaterally normal ovaries. After investigating, she was diagnosed as having MRKH Type 2 syndrome.

Upper urinary tract malformations (40%), like, unilateral renal agenesis (23–28%), ectopia of one or both kidneys

(17%), horse shoe shaped kidneys, renal hypoplasia (4%), and hydronephrosis are with MRKH syndrome. Spinal abnormalities are seen in 30-40% of the cases. 10-25% have associated auditory defects, cardiac anomalies are seen rarely.<sup>4</sup>

The levels of FSH, LH, estradiol in the serum are usually normal.

When physical examination findings reveal hypoplastic vagina, the differential diagnosis includes MRKH and androgen insensitivity syndrome(AIS), the karyotype in AIS is 46 XY with normal levels of serum testosterone and decreased or absent pubic and axillary hair, and presence of intra-abdominal testicles.<sup>5</sup>

### 5. Conclusion

In females suspected to have MRKH, additional renal and skeletal abnormalities should be detected at an early stage. Patient should be counseled to decrease the psychological trauma. She should be offered the surgical procedure for vaginal reconstruction so that she can have a normal sexual life. She can be given the option of surrogacy or adoption of a child.

Examination of the newborn female infant to determine vaginal patency is a simple procedure that is usually not done. Although corrective operations should be deferred until maturity, early detection of the vaginal malformation is helpful for the patient and her family for later correction of anomaly.<sup>6</sup>

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### 7. Conflict of Interest

None.

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**Author biography**

**Manju Agarwal** Senior Professor and Unit Head

**Somya Mathur** P.G. Resident

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