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Mayer-Rokitansky-Küster-Hauser (MRKH) Type 2 Syndrome: A Case Report

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Abstract
The Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a congenital disorder characterized by aplasia of the uterus and the upper part of vagina in an XX individual with normal development of secondary sexual characteristics.The exact etiology of MRKH syndrome is not known.
Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is the second most common cause of primary amenorrhea, after gonadal dysgenesis.
To help establish the best criteria for early diagnosis and treatment options for a comprehensive therapeutic approach to MRKH patients, we report the case of a 24-year-old woman who presented with primary amenorrhea. With difficulty in coitus. Correct evaluation of these patients and proper management is mandatory.

Keywords: *Primary amenorrhea, mullerian duct anomalies, MRKH syndrome*

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I. Introduction

Mayer-Rokitansky-Kuster-Hauser Syndrome (MRKHS) or Mullerian dysgenesis is a rare congenital disorder with an incidence of 1 in 5000 females. MRKHS is characterized by uterovaginal aplasia with normal secondary sexual characteristics and genetic karyotype 46XX [1]. They are of two types: type 1 having only uterovaginal agenesis and type 2 having uterovaginal agenesis with anomalies in fallopian tube, kidney, spine, heart and other organ amenorrhea and painful sexual intercourse **[1].**

The reproductive abnormalities of MRKH syndrome are due to incomplete development of the Müllerian duct. This structure in the embryo develops into the uterus, fallopian tubes, cervix, and the upper part of the vagina. The cause of the abnormal development of the Müllerian duct in affected individuals is unknown. This mullerian duct abnormality is characterized by congenital aplasia of the uterus and the upper part (2/3) of vagina, in young women presenting otherwise with normal endocrine status. It may include absence or hypoplasia of the uterus and fallopian tubes. The patients present with normal secondary sexual characteristics, as the functional ovaries are present, but menstruation is absent **[2].**The exact etiology of MRKH syndrome is not known. Previously, drugs like diethylstilbestrol (DES) and thalidomide were said to have teratogenic causes for MRKH syndrome **[3]**

Here we present a case of a 24-year-old woman who presented with a complaint of primary amenorrhea and coital difficulties. After thorough history and clinical examination, pelvic magnetic resonance imaging (MRI) was performed, which revealed agenesis of uterus ,cervix and proximal 2/3rd of vagina ,along

with agenesis of Right kidney and the ovaries presented with good volume and without any abnormality. This is a rare form of Müllerian agenesis.

II. Case Report

We report the case of a 24 year old woman , nulliparous married female who presented in genecology opd with complaint of primary amenorrhea with difficulty with sexual intercourse in the form of pain and resistance. She had no complaints of cyclical abdominal pain.There was no history of any surgical or medical illness in childhood .and no family history for any reproductive tract anomaly in all blood relatives.

On examination her BP is 110/70mm hg,PR -80/min, vitally stable .And she had normal secondary sexual characteristics but blind ended vagina(as shown in Figure 1) .

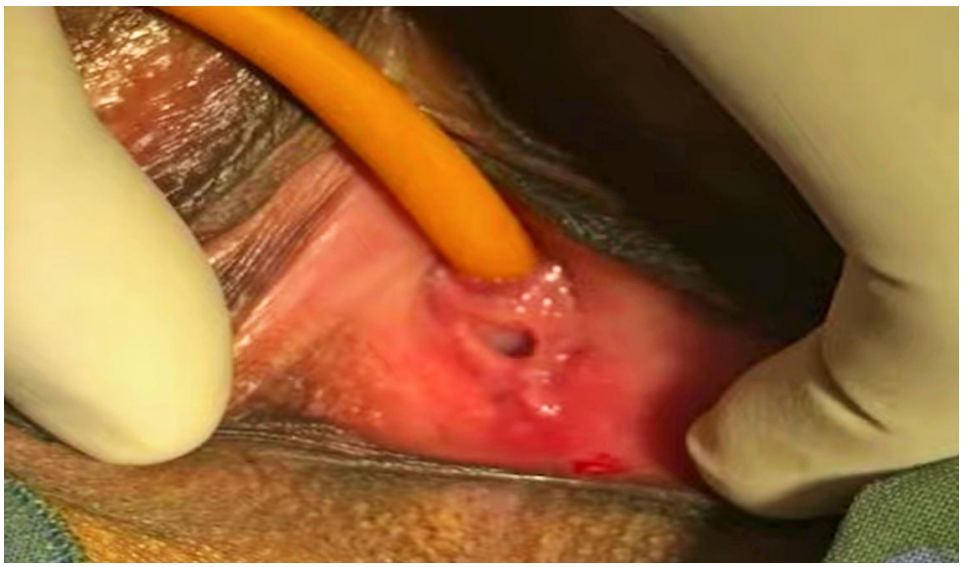

Fig 1 MRKH syndrome patient with blind ended vaginal opening

Her hormonal profile sent which was within normal limit and her karyotype was 46 XX.Transabdominal ultrasound did not demonstrate the presence of uterus ,there was difficulty in visualizing the ovaries.Therefore pelvic MRI examination was done .In MRI of pelvis revealed agenesis of uterus ,cervix and proximal 2/3rd of vagina ,along with agenesis of Right kidney and the ovaries presented with good volume and without any abnormality.(As shown in figure 2)

Genetic evaluation revealed karyotype 46 XX, thus determining the diagnosis of MRKH syndrome. A surgeon specialized in reconstructive surgery and a consultant psychologist was involved .The findings and implications regarding potential fertility and child bearing were explained to the patient.After initial non-operative treatment, the patient underwent vaginoplasty.

Split thickness skin graft was taken from anteromedial aspect of right thigh. Blunt dissection was done and space was created on either side of the vestibule. Intervening tissue were resected and neovagina was created. The mold was wrapped with the skin graft and it was inserted inside the neovagina. Per rectal examination was done in which no abnormality was detected.

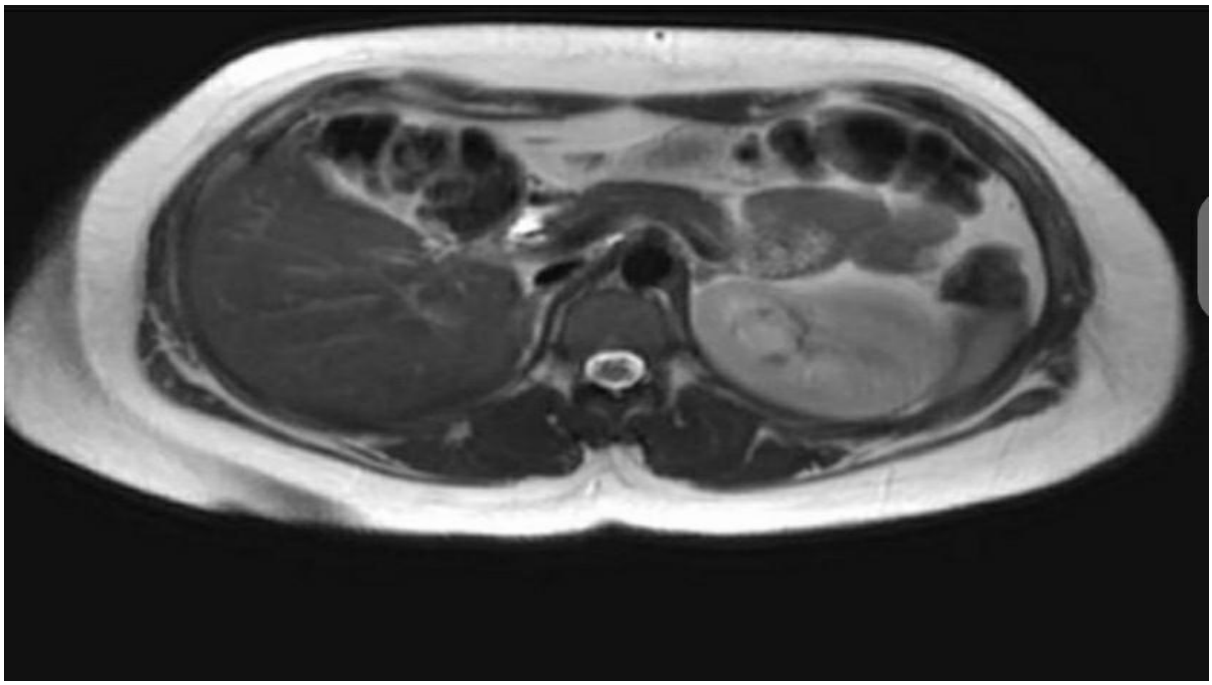

Fig 2 MRI pelvis shows complete absence of uterus and vagina with normal both ovaries

III. Discussion

MRKH syndrome is most frequently diagnosed in female adolescent patients with primary amenorrhea accompanied by the development of normal secondary sex characteristics in health facilities. Pelvic ultrasound is the main modality chosen to be performed in patients with these clinical symptoms. Furthermore, if it cannot be visualized properly, it will be done with an MRI examination **[4]**

Imaging studies such as ultrasonography and magnetic resonance imaging, in association with or without laparoscopy, are necessary to allow the determination of the anatomic characteristics of the syndrome. Ultrasonography is the initial method of choice. This method can demonstrate the absence of the uterus between the bladder and the rectum **[7]**

Misdiagnosis is also common where doctors make further observations and do not provide early therapy so that there is a delayed diagnosis. Women with MRKH syndrome have never had menstruation or pregnancy. Even though ovarian cell donors and in vitro fertilization can be carried out, the patient may still feel depressed. In patients with vaginal agenesis, surgery is also needed so that they can have sexual intercourse with partners **[5].**

Patients who want to have children should be encouraged to adopt, or the possibility of having biological children by means of assisted reproduction techniques should be suggested, considering that the presence of functional ovaries in these women allow the production of normal ovules **[6].**

Even with advanced management of this syndrome, its diagnosis causes significant psychological distress, affecting the patients' quality of life because of the absence of menstruation and impossibility of pregnancy. The distress caused by the diagnosis may be alleviated by surgical or non-surgical treatments, by the passage of time, by counselling, by family's support and by support groups **.[7]**

The differential diagnoses of the condition include androgen insensitivity, transverse vaginal septum and imperforate hymen. Androgen insensitivity syndrome is distinguished from MRKH syndrome by decreased pubic and axillary hair, a 46, XY karyotype, male testosterone level and presence of rudimentary testicles on magnetic resonance images **[8,9].**

IV. Conclusion

MRKH syndrome is one of the most common causes of primary amenorrhea and Ultrasonography is useful for diagnosing any associated renal anomalies. MRI is more precise than USG and less invasive and less expensive than laparoscopy, contributing significantly to treatment planning and patient management.

This case presents that MRKH type 2 syndrome can occur with normal endocrine function and secondary sexual characteristics. Surgical correction by creating a neovagina is a good treatment method in young females for sexual intercourse

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