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# Mayer-Rokitansky-Küster-Hauser (MRKH) Syndrome; Case Report and Literature Review

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***Abstract:***
***Introduction:*** *Mayer-Rokitansky-Küster-Hauser (MRKH) Syndrome is a rare congenital absence of uterus and the upper two-thirds of the vagina in women who have normal development of secondary sexual characteristics and normal 46 XX karyotyping.*
***Case report:*** *We here report on a 17 years old girl with type I MRKH syndrome.*
***Discussion:*** *Laboratory studies should include, Chromosomal analysis to exclude karyotypic abnormalities of the X chromosome and androgen insensitivity syndrome (AIS). Normal levels of luteinizing hormone (LH) and follicle-stimulating hormone (FSH) indicate appropriate ovarian function. On the other hand, Ultrasound and MRI plays a pivotal role in highlighting the exact anatomical details.*
***Conclusion:*** *absence of normal menstruation in young normal looking adolescent female should raise suspicion of MRKH syndrome. Management should be tailored to the individual needs and motivation of the patient.*
***Keywords:*** *MRKH Syndrome, Paramesonephric (Mullerian) ducts, Mesonephric (Wolffian) ducts, Aplasia of the uterus, Genital renal ear syndrome, Mullerian agenesis, Mullerian aplasia, Mullerian dysgenesis, Rokitansky syndrome.*

## I. Introduction

Female genital system usually develops during fetal life in her mother's uterus. At age 6 weeks of intrauterine life, both male and female genital organs are similar [1, 2]. They consist of two sets of paired ducts: the paramesonephric (Mullerian) ducts and the mesonephric (Wolffian) ducts. In the absence of the Y chromosome the mesonephric duct start to degenerate, while, paramesonephric ducts develop bi-directionally along the lateral aspects of the gonads. The proximal segments of the ducts remain unfused and open into the peritoneal cavity and they form the fallopian tubes [1, 3]. On the other hand, the distal segments, progress caudomedially and join each other. Added to that, contacting the posterior aspect of the pelvic urethra. These distal segments give rise to the uterus and upper 4/5th of the vagina [1].

At 9 weeks, the paramesonephric ducts fuse at their inferior margin forming the single lumen of the uterovaginal canal [4, 5]. After that, the septum between the two tubes regresses. There are two theories for the regression of the septum, a classical unidirectional regression theory (caudal to cranial regression of the uterovaginal canal), and, an alternative bidirectional theory where the regression of the septum proceeds simultaneously in both the cranial and the caudal directions [4-6]. By week 12, the uterus exhibits its normally developed configuration [5, 6].

Both the urinary and genital systems arise from a common ridge of mesoderm along the dorsal body wall, and both systems rely on normal development of the mesonephric system [1, 3, 5, 6].

The Mayer-Rokitansky-Küster-Hauser Syndrome is defined as congenital aplasia of the uterus as well as the upper two-thirds of the vagina in women who have normal development of secondary sexual characteristics and normal 46 XX karyotyping [3, 5]. It is also known as congenital absence of the uterus and vagina (CAUV), genital renal ear syndrome (GRES), Mullerian agenesis, Mullerian aplasia, Mullerian dysgenesis and Rokitansky syndrome. It is thought to be an autosomal dominant syndrome with the incidence of 1 out of 4500 women. Type I MRKH syndrome is usually isolated type while type II is associated with renal, vertebral, and, to a lesser extent, auditory and cardiac defects [2, 3, 5, 7].

The usual presentation is young females present with normal female type body with the normal functioning ovaries. The external genitalia do not show any abnormality as per the anatomical lay out. The patients usually have normal thelarche and adrenarche. This requires complete work up as it may overlap other similar types of syndromes [2, 4-6]. We here report on a case of type I MRKH syndrome.

## II. Case Report

A 17 years old Saudi girl presented with a history of primary amenorrhea. She did not complain of any gastrointestinal or urinary symptoms. There is no family history of same problem or any other endocrine

diseases. She had normal growth parameter (normal height). Vital signs within normal. She had normal secondary sexual characteristics and normal looking external gentalia.

On investigation, the hormonal profile was normal, Chromosomal analysis 46XX. While ultrasound of the pelvis showed normal bilateral ovaries with query absent uterus. On MRI, there was Right-sided ectopic pelvic kidney, uterus cannot be visualized (??Rudimentary), very small-sized bilateral ovaries seen and no other abnormality seen Figure 1.

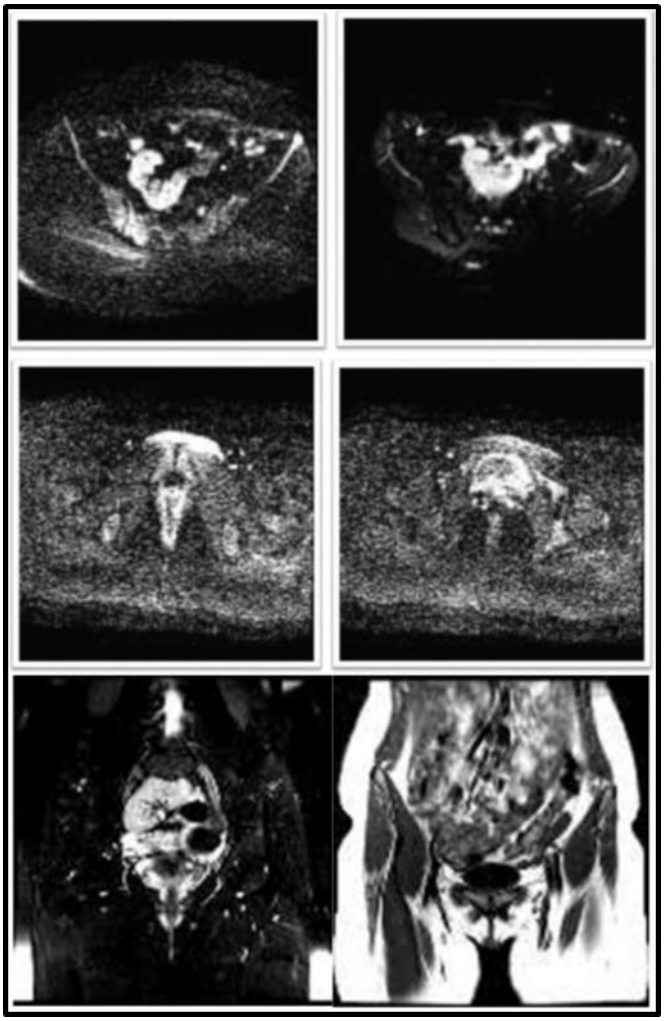

**Figure 1**; Patient MRI

## III. Discussion

There are two subtypes of MRKH syndrome, the typical (type I or isolated) and the atypical form (type II). The frequency of type II being much greater than type I [3, 4, 7]. The typical form is characterized by laparoscopic or laparotomic findings of symmetric muscular buds (the Müllerian remnants) and normal Fallopian tubes; this is referred to as Rokitansky sequence, where only the caudal part of the Müllerian duct (upper vagina and uterus) is affected. On the other hand, atypical form shows, in addition to what was mentioned above, asymmetric hypoplasia of one or two buds, with or without dysplasia of the Fallopian tubes. It may be associated with renal alterations (40%): renal agenesis, ectopic kidney, horseshoe kidney. Vertebral alterations: scoliosis, spinal fusions, malformations of the ribs 3-5. Facial malformations and malformations of the distal portion of the extremities. Auditory alterations: deafness, stapedial ankylosis, adysplasia of the pinna. Heart malformations: aorto-pulmonary malformations, atrial septal defect, fault in pulmonary valve, Tetralogy of Fallot [2, 3, 7].

Patient usually undergoes puberty with normal thelarche and adrenarche, however, menses do not begin (i.e., primary amenorrhea). Patients may report cyclic abdominal pain due to cyclic endometrial shedding without a patent drainage pathway. Because ovarian function is normal, patients experience all bodily changes associated with menstruation. There is usually Infertility, Difficulty with intercourse, Voiding difficulties, urinary incontinence, or recurrent UTIs and/ or vertebral anomalies (most commonly scoliosis). The degree of

vaginal aplasia can vary from complete absence to a blind pouch. The shallower the canal, the greater the likelihood of the patient having dyspareunia or inability to have intercourse [5, 6].

On examination, normal secondary female sexual characteristics are present after puberty, height is normal, Speculum examination of the vagina may be impossible or difficult because of the degree of vaginal agenesis. In contrast, vulva, labia majora, labia minora and clitoris are normal [4, 6, 7].

Laboratory studies should include, Chromosomal analysis to exclude karyotypic abnormalities of the X chromosome and androgen insensitivity syndrome (AIS), since, individuals with complete AIS have female external genitalia but a 46XY karyotype. Circulating levels of luteinizing hormone (LH) and follicle-stimulating hormone (FSH), which are normal in MRKH syndrome, indicating appropriate ovarian function. Added to that, Testosterone levels are in the normal female range [2, 3]. Radiologically, ultrasound scan [8] is the first test that must be requested for the confirmation of the syndrome. Abdominal and pelvic US does not only serve to evaluate the absence of uterus, but also for the renal study. Alternatively, MRI must be used when ultrasound findings are incomplete or inconclusive. Rarely in case of diagnostic doubts, it is possible to resort to diagnostic laparoscopy, which will be able to evaluate the uterine aplasia or hypoplasia, the presence of uterine rudimentary horns and normal ovaries placed in the high part of the pelvis [4, 5, 7].

Differential diagnosis list is long and include Gonadal dysgenesis, Androgen insensitivity syndrome, Agenesis of vagina and uterus, vaginal transverse septum, and Imperforated hymen. Treatment is usually delayed until the patient is ready to start sexual activity. The management depends upon the type and underlying abnormality. It may be either surgical or non-surgical but the chosen method needs to be tailored to the individual needs and motivation of the patient and the options available. Non-surgical option in some cases is Franck's technique or perineal dilatation. The surgical techniques include various options like McIndoe technique, William vaginoplasty, Rotational flap procedure, Intestinal neovagina and Vacchietti technique. In addition, it is important to manage psychological symptoms in women with Müllerian agenesis. This is because a young woman who discovers that she has a congenital malformation involving her reproductive organs may develop extreme anxiety about her feminity and physical image [2, 3, 7].

## IV. Conclusion

When there is complaint of not having started normal menstruation in spite of being young normal looking adolescent female with normal secondary sexual characters, this is enough to raise suspicion. Radiological studies including MRI and US plays a pivotal role in highlighting the exact anatomical details to decide about the management. The management is either surgical or non-surgical and should be tailored to the individual needs and motivation of the patient and the options available. It is still important to manage psychological symptoms in these women.

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