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Mayer Rokitansky Küster Hauser Syndrome as a Cause of Infertility and Primary Amenorrhoea: A Case Report

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Abstract

Mayer Rokitansky Küster Hauser (MRKH) syndrome is a rare congenital disorder that occurs during foetal development. MRKH is characterised by agenesis of the uterus and part of the vagina. Its exact aetiology is still unknown due to the complexity of the genetic pathways involved in the embryogenetic development of the Müllerian ducts. Although these women have a female phenotype and functional ovaries, they do not have menstrual cycles. We report the rare case of a 23-year-old married woman who consulted us for primary amenorrhoea and desire for pregnancy. On assessment and investigation, it was confirmed that the patient had no uterus and a poorly developed vagina. However, she had normal functioning ovaries, and no abnormalities in other organs were detected. A multidisciplinary team provided advice on the nature of the anomaly, its implications for sex life, infertility and possible child adoption alternatives.

Keywords

Rokitansky Syndrome, Müllerian Ducts, Uterine Agenesis

1. Introduction

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a congenital malformation in phenotypically female subjects with 46 XX karyotypes. It has an autosomal dominant mode of inheritance [1] and is characterised by agenesis of the uterus and vagina with normal ovaries. It is linked to the failure of the Müllerian ducts to migrate to the urogenital sinus during embryonic life, resulting in the absence of the uterus and incomplete development of the vagina. It is divided into two types: type I is characterised by utero-vaginal aplasia, while type II is also associated with extra-genital anomalies, most often renal (30% - 40%), skeletal, auricular and cardiac. First-degree relatives of MRKH patients seem to have a 1% - 5% risk of congenital uterine anomalies as in most multifactorial disorders. The majority of the studies of familial cases suggest an autosomal dominant inheritance pattern limited to the female sex, implying that the genetic defect is typically inherited by the father.

At puberty, a woman with MRKH syndrome develops normal secondary sexual characteristics such as breast growth, axillary and pubic hair and primary amenorrhoea [2]-[4].

It is a syndrome that is not frequently diagnosed early, making a clear prevalence difficult. However, it is estimated that one in every 4500 to 5000 women has MRKH syndrome [2]. With regard to fertility in MRKH individuals, surrogacy remains the option; however, uterine transplantation has recently been introduced [4].

MRKH syndrome, which is little known in our context and is usually diagnosed late, poses a management problem. We therefore report this case because of its rarity, with the aim of discussing the diagnosis and management of this syndrome.

2. Observation of the Case

The case that we report here is made with the informed consent of the patient after the approval of the Direction of the hospital and the collective of the teachers-researchers of the same hospital.

Female patient, 23 years old, housewife, primary level, medersa school, 7th child in her sibling group, with one brother and five older sisters, all married, two of whom have never conceived a pregnancy. She has been married for four months and lives with her partner with regular sexual relations. She has consulted a doctor for primary amenorrhoea and a desire to become pregnant. She was fully vaccinated as a child.

Her mother would have been monitored during her pregnancy and would have benefited from various antenatal prophylaxes.

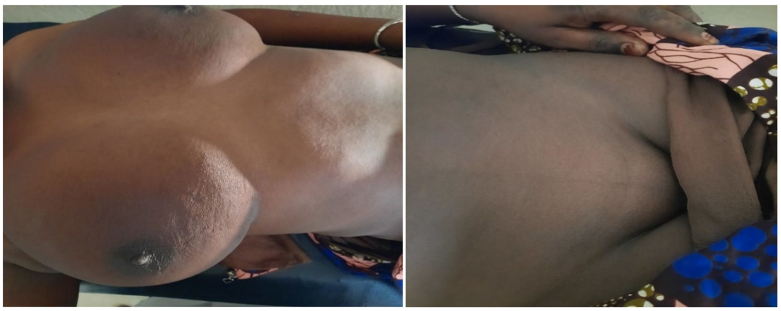
She had undergone ritual excision as a child. Her medical history was unremarkable and she had no history of surgery. She had never menstruated or experienced haematuria.

Physical examination revealed a female figure, height 1.73 m, weight 90 kg, blood pressure 110/60 mmhg, temperature 36.7˚C, pulse 96 beats/min. The lymph

nodes were free.

She had a feminine voice, and when unclothed she had a feminine morphotype.

Secondary sexual characteristics were marked by normal breast development and normal but shaved pubic hair. The appearance of the breasts and pubis is illustrated in Figure 1.



(a) (b)

Figure 1. (a) View of the breasts; (b) View of the pubis.

Examination of the external genitalia revealed a type II excision and a vulva with normal labia majora. Speculum examination revealed a blind vagina about 10 cm deep with macroscopically healthy walls. There was no perception of a uterus on vaginal and rectal touch.

Pelvic ultrasound and abdominopelvic CT scan without and with injection of contrast medium showed complete agenesis of the uterus in the upper third of the vagina.

Normal ovaries and kidneys. All these results, illustrated in Figure 2, led to the diagnosis of Mayer-Rokitansky-Kuster-Hauser syndrome.

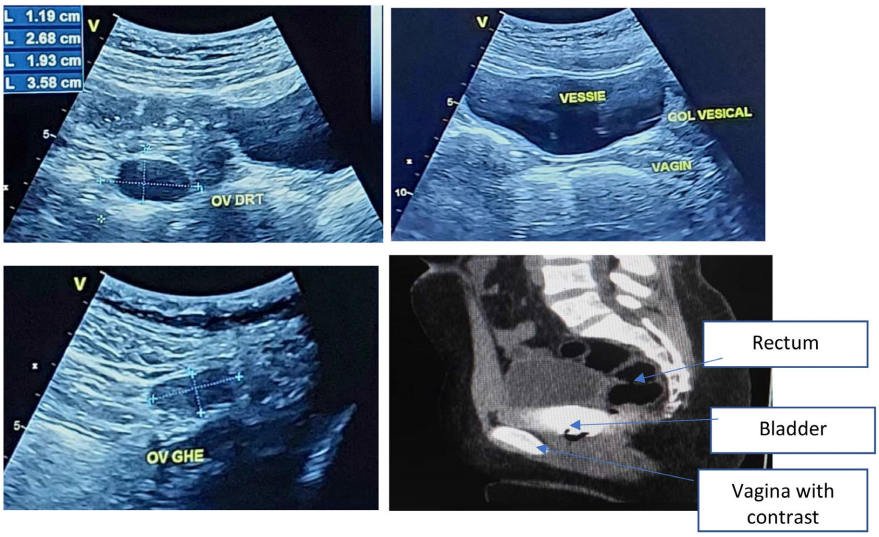


Figure 2. Pelvic ultrasound and abdominopelvic CT image.

The results of the hormone test are summarised in Table 1: FSH, LH, prolactin and estradiol are within the normal range for an adult woman.

Table 1. Blood test results.

Balance sheet	Results	Reference values
FSH	4.84 mIU/ml	3.9 - 24 mUI/ml
LH	14.97 mUI/ml	2.0 - 80 mUI/ml
Prolactinemia	281.49 µUI/ml	66.0 - 490.0 UI/ml
Estradiol	36.68 pg/ml	15 - 251 pg/ml
Testosterone	7.365 ng/ml	<1.00 ng/ml

The treatment included a psychological component, which consisted of informing the patient of her inability to conceive or menstruate due to the absence of a uterus, and the possible alternatives of surrogacy. On the other hand, she can lead a normal sexual life, given the normal size of her vagina. The karyotype is not feasible in our context; genotypic confirmation could not be obtained.

3. Discussion

Mayer Rokitansky Küster Hauser (MRKH) syndrome is a rare congenital disorder characterised by the absence of a uterus and vagina in a phenotypically female patient with a 46 XX karyotype [1].

Patients with MRKH syndrome usually present during adolescence with primary amenorrhoea after normal puberty and the development of secondary sexual characteristics [3]. Two clinical forms have been described: MRKH type I which corresponds to isolated uterovaginal agenesis and MRKH type II which is characterised by incomplete agenesis and/or associated with other congenital malformations. In our patient, this was MRKH type I, as imaging revealed no associated malformations. She was 23 years old with secondary female sexual characteristics and had consulted for primary amenorrhoea and a desire for pregnancy.

The case reported by Joshi *et al.* [1] was the same age as ours, 23 years, but the reason for consultation was a crisis of abnormal behaviour with recurrent episodes of dissociative convulsions most of the time.

Cases were diagnosed in adolescence at 17 years in Morocco [5] and 18 years in Tanzania [2] respectively, who had consulted for amenorrhoea but with well-developed secondary sexual characteristics.

U. Ray *et al.* [6]; Shimizu H *et al.* [7] reported cases of later diagnosis at 25 years of age, one a married woman complaining of primary amenorrhoea and painful intercourse and the other a single woman with a vaginal anomaly, all with normal secondary sexual characteristics.

The diagnosis of MRKH syndrome is based on pelvic ultrasound and MRI, which may be supplemented by a hormonal work-up. In our case, pelvic ultrasound and abdominopelvic MRI revealed complete agenesis of the uterus in the upper third of the vagina (**Figure 2**) and the hormonal profile confirmed normal ovarian function and the gonadotropic axis (**Table 1**). The karyotype could not be performed in our context because of the unavailability of this test in most

laboratories in our country, and also because of its very high cost.

As in our case, in most of the literature [2] [5] [6] [8], the confirmatory diagnosis of MRKH syndrome is based on hormonal assessment and imaging.

Because of the interference with sexual life and the lack of possibility of pregnancy, Rokitansky syndrome can have significant psychological repercussions for these patients. These women may have difficulty managing their intimacy and talking to their partners about it [5] [9]. In the case we report, the patient had difficulty accepting her illness and was unable to inform her partner. This is why, as soon as the diagnosis was confirmed, the patient benefited from psychological care thanks to which she did not develop any signs of mental depression.

Surgical and non-surgical treatments only improve the quality of the woman's sex life. The standard non-surgical treatment for Rokitansky syndrome is vaginal dilatation, known as Frank's method, which involves creating a neovagina using different-sized dilators that gradually dilate the vagina [5]. We did not have a vaginoplasty because she was a married woman who claimed to have a normal sex life. With regard to her desire to become pregnant, she was informed of the possible alternatives of adopting a child, as conception was impossible due to the absence of a uterus.

However, uterine transplantation, which is a recent and rapidly expanding field of research, may currently be considered invalidated and supervised clinical research programs.

This type of transplantation is subject to the risk of graft failure, mainly due to thrombosis of the anastomoses in around 20% of cases, risks associated with immunosuppression, and maternal pregnancy risks. In France, out of 34 pregnancies described in the literature that resulted in the birth of healthy children, only 4 were free of complications [10].

4. Conclusion

Mayer-Rokitansky-Küster-Hauser syndrome is a disease that can have significant psychological repercussions in patients with an impaired quality of life, but above all poor sexual esteem. It is a psychosocial problem that requires good psychological support, genetic counseling, which has become important in recent years with the introduction of surrogacy and, more recently, uterine transplantation.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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