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An Unusual Presentation of Mayer-Rokitansky-Kuster-Hauser Syndrome with Rectovaginal Fistula

Authors
Dr Dalia Rafat[1], Dr Imam Bano[2], Dr Shaheen[3]
[1]Assistant Professor, Deptt. of Obstetrics & Gynaecology, J N Medical College & Hospital, Faculty of Medicine, Aligarh Muslim University, Aligarh, UP, India-202002
[2]Professor, Deptt. of Obstetrics & Gynaecology, J N Medical College & Hospital, Faculty of Medicine, Aligarh Muslim University, Aligarh, UP, India-202002
[3]Associate Professor, Deptt. of Obstetrics & Gynaecology, J N Medical College & Hospital, Faculty of Medicine, Aligarh Muslim University, Aligarh, UP, India-202002
Corresponding Author
Dr Dalia Rafat
Assistant Professor, Deptt. of Obstetrics & Gynaecology, J N Medical College & Hospital, Faculty of Medicine, Aligarh Muslim University, Aligarh, UP, India-202002
Email: *drdaliarafat.16@gmail.com,*

Abstract

Mayer-Rokitansky-Kuster-Hauser Syndrome is a rare congenital anomaly characterised by mullerian duct agenesis resulting in hypoplastic or absent uterus and vaginal aplasia or hypoplasia with normal ovaries. Physical growth, development and secondary sexual characters as well as the external genitalia are also normal. These patients most commonly presents with primary amenorrhea and difficulty or inability of sexual intercourse. Here we report a case of 24 year old woman with Müllerian agenesis and rectovaginal fistula following coital injury who has presented to us for secondary amenorrhea and infertility.

Keywords: *Mayer Rokitansky Kuster Hauser syndrome , Rectovaginal fistula, Coital injury*

Introduction

Female genital tract anomalies comprises a unique subset of congenital malformations. During embryogenesis, the complete formation of the genital tract is dependent on proper development of mullerian ducts and sinovaginal bulb followed by their fusion and septal resorption [1]. The American Society of Reproductive Medicine has classified Müllerian anomalies into seven categories: (1) hypoplasia or agenesis; (2) unicornuate uterus; (3) didelphic uterus; (4) bicornuate uterus; (5) septate uterus; (6) arcuate

uterus; and (7) Tshaped uterus from diethylstilbestrol (DES) exposure [2]. Mayer-Rokitansky-Kuster-Hauser syndrome (MRKHS) comes under class 1 and is characterized by mullerian duct structures agenesis resulting in partial or complete absence of the uterus with an absent or hypoplastic

vagina. Mayer first described this syndrome in 1829 as abnormal development of the Müllerian ducts resulting in various vaginal anomalies [3]. Further Rokitansky described uterine and vaginal agenesis in 1838. Later in 1910, Kuster recognized urological as well as skeletal associations [4] and in 1961, Hauser distinguished MRKHS from testicular feminization [5]. These patients most commonly presents with primary amenorrhea and difficulty or inability of sexual intercourse. Here we report a case of 24 year old woman with Müllerian agenesis and rectovaginal fistula following coital injury who has presented to us for secondary amenorrhea and infertility.

Case Summary

A 24-year old woman presented to the gynaecology clinic with complaint of infertility and secondary amenorrhea. According to her she developed menarche at the age of 18 years and her first episode of vaginal bleeding was immediately after having sexual intercourse with her husband, which was her third or fourth coital experience with him. Her initial sexual relationships were associated with dyspareunia and after one act she developed severe pain and profuse vaginal bleeding. It was managed with hot water and some home remedies and subsided spontaneously after 4 to 5 days. She considered it to be a welcome sign. After this episode she continued to have on and off spotting for few initial months but since then she did not have any vaginal bleeding or discharge. On clinical examination she was found to have well developed secondary sexual characteristics and no abnormality was detected on abdominal examination. Vaginal examination revealed normal vulva and urethral meatus, but a short (3 cm) blind ended vagina [Figure 1] and we were appalled to find a communication between the vagina and rectum. The vaginal defect was about 2 cm and extending up to rectum [Figure 2]. The anal sphincter was intact and normal. After discovering the rectovaginal fistula we cross questioned the patient again for any leakage of flatus and feces per vaginam to which patient agreed and told that since onset of her vaginal bleeding she has developed this leakage which was quite troublesome to start with but with time it has improved and now she passes stools per vaginum only during episodes of loose stools but patient has never consulted anywhere for it as she didn't wanted her husband to know about it. Ultrasonography (US) and magnetic resonance imaging (MRI) revealed the absence of uterus but ovaries were reported to be present. Her chromosomal study revealed normal karyotype of 46 XX. Based on these findings a diagnosis of Mayer-Rokitansky-Kuster-Hauser syndrome with rectovaginal fistula was made. Patient was told about her condition and her fertility potential and was counselled for repair but patient and her attendants didn't consented for it as there concern was only her infertility and not her morbidity.

Figure 1: Shows blind ended vagina

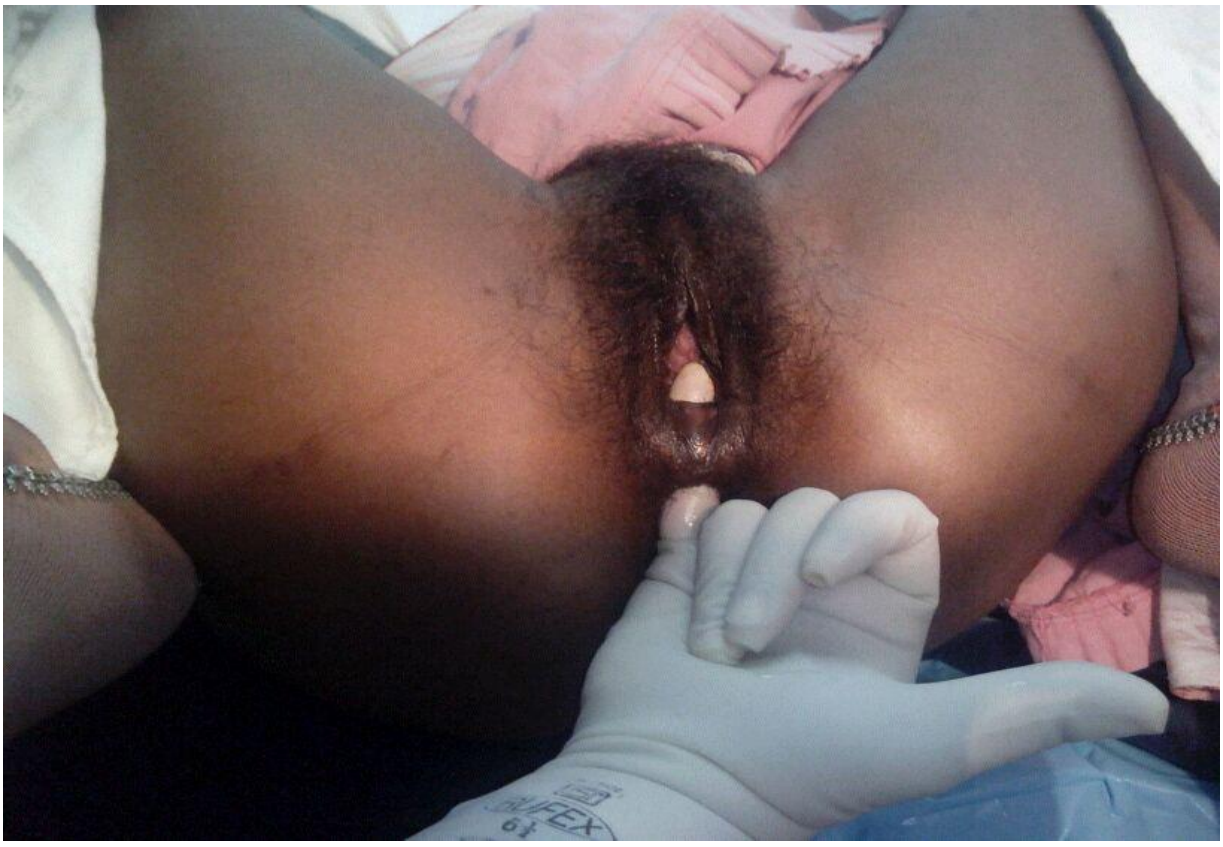

Figure 2: Shows vaginal defect extending upto rectum admitting finger

Discussion

Mayer-Rokitansky-Kuster-Hauser Syndrome is a rare congenital anomaly characterised by mullerian duct agenesis resulting in hypoplastic or absent uterus and vaginal aplasia or hypoplasia with normal ovaries. The prevalence of MRKHS has been reported as 1 in 5000 woman [6]. Its etiology is still not completely understood, and is believed to be polygenic and multifactorial. Patients with MRKHS have a normal 46 XX karyotype. These patients most commonly presents with primary amenorrhea and difficulty or inability of sexual intercourse due to vaginal aplasia/hypoplasia. On physical examination, secondary sexual characteristics are normal due to normal ovarian function, with no sign of androgen excess. Because of the normal external appearances of MRKHS females, the syndrome is difficult to diagnose until puberty. Though such patients may occasionally be diagnosed at birth or during childhood due to other health problems, the usual mean age of diagnosis is between 15 and 18 years [7], when patient with MRKHS presents with primary amenorrhea with well developed secondary sexual characteristics. US is usually the first method for the diagnosis and in the majority of patients, can lead to the correct diagnosis. MRI is the investigation of choice as it clearly delineates the uterovaginal anatomy and also add information about associated anomalies [8]. Treatment employs either creation of a neovagina via surgical and nonsurgical methods or vaginal replacement with a pre-existing canal lined with a mucous membrane like bowel [9]. The key factors for the success of treatment are patient cooperation and psychological maturity, and for these reasons treatment should begin when the patient has reached emotional maturity and is ready to start sexual activity [10]. Rectovaginal fistula (RVF) is an abnormal connection between the rectum and vagina and most cases are caused by either obstetric injuries or surgical complications. RVF due to coital injury is rare with only few cases reported in literature[10,11] and RVF in a patient of MRKHS is even rarer with probably only one recent report thus far reported in literature [10] and our case report being the second one. Our case is also unique because of the presentation of MRKHS as secondary amenorrhea and infertility and not as RVF.

Conclusion

Patients of MRKHS usually presents with primary amenorrhea and inability of sexual intercourse. But such patients can present with atypical symptoms like RVF and secondary amenorrhea (not in true sense). This case report also highlights the fact that despite of all the technological advances of the 21st century, we still encounter such patients like the one reported above who has considered her injury as welcome sign of her menarche and was having RVF for 6 years but never consulted anywhere for it and even after being diagnosed of the condition didn't agreed for its repair with her only concern being infertility. The only thing which explains such presentation is ignorance and social pressures. To the best of our knowledge our case is the second case of RVF in MRKHS, in literature so far and the first of its kind.

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