

A Case of Mayer-Rokitansky-Kuster-Hauser (MRKH) Syndrome Type B With Vesico-Vaginal Fistula

Dr. Ebenezer E. Dadson^{1*}, Dr. (Mrs.) Augustina Badu Peprah¹, Dr. Edward Dassah², Dr. Kwasi Ankomah¹, Dr. Obed Otoo¹, Dr. Kwasi Twum¹, Dr. Akosua Nsiah Achiaamah¹, Dr. Anna Adjei Yeboah¹, Dr. Esi de-Graff Johnson¹, Dr. Adu Tutu Amankwa¹ and Dr. Achiaa Afreh¹

¹Staff of The Radiology Directorate, Komfo Anokye Teaching Hospital, Kumasi, Ghana.

²Directorate of Obstetrics and Gynecology, Komfo Anokye Teaching Hospital and School of Public Health, Kwame Nkrumah University of Science and Technology.

Citation: Dadson EE, Peprah AB, Dassah E, et al. A Case of Mayer-Rokitansky-Kuster-Hauser (MRKH) Syndrome Type B With Vesico-Vaginal Fistula. *Gynecol Reprod Health*. 2022; 6(2): 1-3.

Keywords

Mayer Rokitansky-Kuster-Hauser, Vesico-Vaginal Fistula, Amenorrhea.

Introduction

The Mayer Rokitansky-Kuster-Hauser (MRKH) syndrome is caused by embryologic underdevelopment of the Müllerian duct, with resultant agenesis or atresia of the vagina, uterus or both. It is characterized by congenital aplasia of the uterus and the upper two thirds of the vagina [1-4].

MRKH syndrome constitutes the second most common cause of primary amenorrhea, after gonadal dysgenesis [5]. The incidence is 1 in 5000 [2-5].

MRKH syndrome has been divided into two types: type A (typical form) is an isolated anomaly also known as the Rokitansky sequence. Type B (atypical form) is characterized by asymmetric uterine buds or abnormally developed fallopian tubes. This atypical form is associated with anomalies that involve other systems - renal, cardiac, otologic, and skeletal systems. MRKH syndrome in association with vesico-vaginal fistula and a solitary ectopic (pelvic) kidney has not been reported in literature.

We present the history, clinical and imaging findings of a 28 year-old female with primary amenorrhea who was referred for further diagnostic evaluation. Further diagnostic evaluation demonstrated MRKH syndrome type B with a vesicovaginal fistula.

Clinical Presentation and Findings

A 28 year-old female with primary amenorrhea was referred from a district hospital after ultrasound scan detected the uterus was absent. She was not married and had had previous surgery for

*Correspondence:

Dr. Ebenezer Ekow Dadson, Radiologist, KATH, Ghana.

Received: 04 Mar 2022; **Accepted:** 06 Apr 2022; **Published:** 10 Apr 2022

imperforate hymen. Her height was 1.6m. There was no skeletal deformity or hearing impairment. Her breasts, pubic hair and external genitalia appeared normal.

Imaging

The upper part of the vagina and the uterus were absent. The outlined lower vagina measured about 2.8cm long. Subsequent filling of the vagina with approximately 20ml of gel failed to distend it. However, evidence of the instilled gel was seen within the urinary bladder as serpentine hyperintensities on T2W images. There is evidence of gas within the antero-superior aspect of the urinary bladder implying some degree of communication between the urinary bladder and the external environment.

Both ovaries were present but enlarged. The right kidney was absent. The left kidney measured 10.2 x 7.9cm and is noted within the left side of the pelvis with normal signal characteristics.

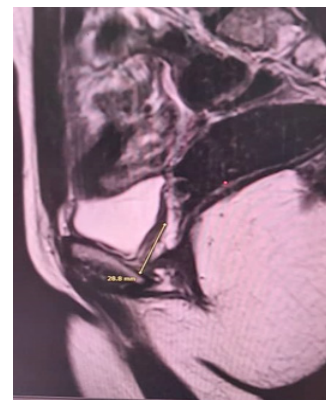


Figure 1: Sagittal T2W (right) images of the pelvis showing agenesis of the upper vagina and the uterus.

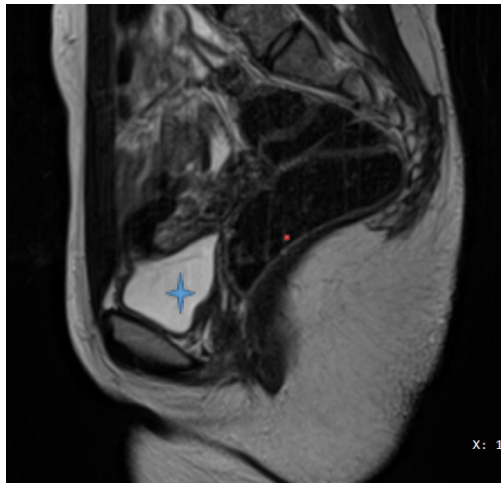


Figure 2: Sagittal T2W image of the pelvis showing the urinary bladder (+) before the introduction of gel into the vagina.

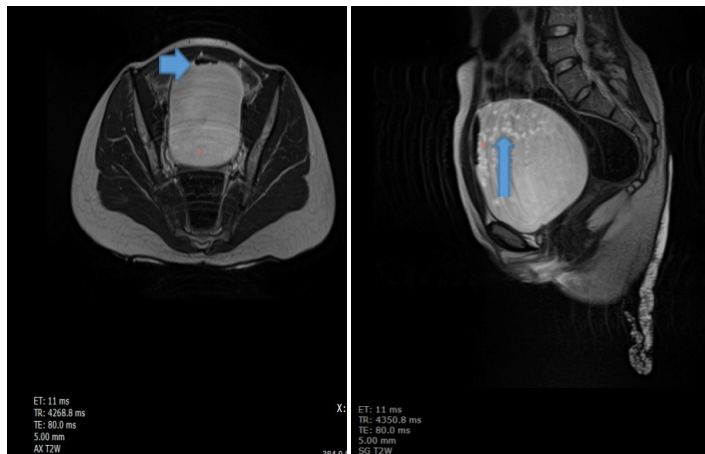


Figure 3: Sagittal T2W (left) and axial T2W (right) images of the pelvis showing serpentine hyperintensities (→) within the urinary bladder after gel application. The vaginal failed to distend. A signal void area within the antero-superior aspect of the urinary bladder consistent with gas (↑).

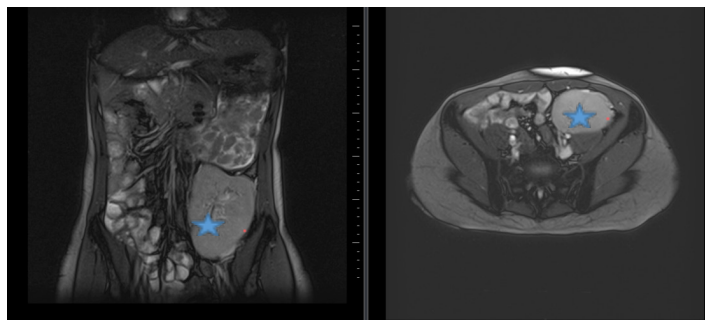


Figure 4: Coronal SSFP (left) and axial SSFP (right) images of the abdomen showing a solitary ectopic (pelvic) left kidney (★) with normal signal characteristics.

Discussion

Mayer–Rokitansky–Kuster–Hauser (MRKH) syndrome is a class I Mullerian anomaly characterized by congenital absence of the

upper two-thirds of the vagina and an absent or rudimentary uterus in women who have normal development of secondary sexual characteristics and a 46, XX karyotype. It results from agenesis or hypoplasia of the Mullerian (paramesonephric) ducts. Associated abnormalities of the kidneys and other organ systems are often seen [3,7]. In this case the patient had single kidney on the left in an ectopic position (pelvis).

Etiologically, this syndrome may be caused by the lack of development of the Mullerian ducts between the fifth and the sixth weeks of gestation. To explain this condition, it has been suggested that in patients with MRKH syndrome, there is a very strong hypersecretion of Mullerian-inhibiting factor (MIF), which would provoke the lack of development of the Mullerian ducts from primitive structures (as what normally occurs in male phenotype). These alterations are commonly associated with renal agenesis or ectopia. Specific mutations of several genes such as WT1, PAX2, HOXA7-HOXA13, PBX1, and WNT4 involved in the earliest stages of embryonic development could play a key role in the etiopathogenesis of this syndrome [3].

The diagnosis of MRKH syndrome is associated with profound psycho-social issues ranging from low self-esteem to infertility. As such early recognition by way of diagnosis is a crucial step in the multidisciplinary management of such cases.

Complete androgen insensitivity syndrome (CAIS, also called Morrison syndrome) is an important differential (absent gonads virilization) [6].

CAIS is an androgen receptor defect disorder associated with vaginal and uterine agenesis in women with a 46,XY karyotype [7].

Individuals with CAIS are phenotypic females with normal breast development, minimal pubic and axillary hair. Although the external genitalia are within normal limits, the vagina is typically absent or rudimentary and the uterus absent. Gonads found in the labia majora, inguinal ring, or intra-abdominally, were variably noted on physical exam [8].

Associated with profound psycho-social issues ranging from low self-esteem to infertility. Early recognition by way of diagnosis is a crucial step in the multidisciplinary management of such cases

Conclusion

In patients presenting with primary amenorrhea and congenital absence of the uterus, a high index of suspicion for MRKH should be maintained.

Further radiological evaluation may detect associated anomalies of the other systems, which will be essential for optimal management of the patient, as was demonstrated in our case.

Chromosomal analysis (Karyotypic studies) is essential to rule out CAIS which is the main differential.

References

1. Elmezughi K, Panicker M, Ekpebegh C. Mayer-Rokitansky-Küster-Hauser syndrome presenting with primary amenorrhea and chronic kidney disease: a case report. *PAMJ Clin Med.* 2021; 5.
2. Morcel K, Camborieux L, Guerrier D. Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome. *Orphanet J Rare Dis.* 2007; 2: 13.
3. Pizzo A, Laganà AS, Sturlese E, et al. Mayer-Rokitansky-Küster-Hauser Syndrome: Embryology, Genetics and Clinical and Surgical Treatment. *ISRN Obstet Gynecol.* 2013; e628717.
4. Tiwari C, Shah H, Waghmare M, et al. Mayer-Rokitansky-Küster-Hauser syndrome associated with rectovestibular fistula. *Turk J Obstet Gynecol.* 2017; 14: 70-73.
5. Kłosowicz E, Komenda J, Zmaczyński A, et al. Patient with Mayer-Rokitansky-Küster-Hauser syndrome (MRKH) who Underwent Laparoscopic Davydov-Moore vaginoplasty. *Gynecol Obstet Case Rep.* 2020; 6.
6. Ghirardini G, Popp LW. The Mayer-von Rokitansky-Küster-Hauser syndrome (uterus bipartitus solidus rudimentarius cum vagina solida): the development of gynecology through the history of a name. *Clin Exp Obstet Gynecol.* 1995; 22: 86-91.
7. Oakes MB, Eyvazzadeh AD, Quint E, et al. Complete androgen insensitivity syndrome-a review. *J Pediatr Adolesc Gynecol.* 2008; 21: 305-310.
8. Morris JM. The syndrome of feminization in male pseudohermaphrodites. *Am J Obstet Gynecol.* 1953; 65: 1192.