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Mayer-Rokitansky-Kuster-Hauser Syndrome in an 18-year-old Female: The Radiodiagnostic Perspective

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Abstract:

Müllerian duct anomalies are a group of congenital uterine disorders that arise from an arrest in development, incomplete fusion or incomplete resorption of the mesonephric ducts. They are usually asymptomatic but diagnosed incidentally. An 18-year-old girl presented at the gynaecology clinic with a history of primary amenorrhea and failure to develop secondary sexual characteristics. She is the only daughter of a widow. According to the mother, her pregnancy and birth histories were normal, and delivery was by spontaneous vaginal delivery. The patient had satisfactory developmental milestones and had completed secondary school at the time of presentation. She is of average physical stature with rudimentary breasts (tanner Stage 2), lacking axillary and pubic hairs. A pelvic examination revealed a 1.5 cm blind-ending vaginal pouch. A radiologic evaluation was done using ultrasonography and magnetic resonance imaging showed the absence of the uterus and the ovaries. The patient and the mother were counselled on management options, including the future fertility options. The mother vehemently rejected the option of surgery and has not been seen since after that.

Keywords:

Absence of the uterus, Mayer-Rokitansky-Küster-Hauser syndrome, mullerian agenesis, Port Harcourt, primary amenorrhoea

Introduction

Müllerian duct anomalies (MDAs) are a group of congenital uterine disorders that arise from an arrest in development, incomplete fusion or incomplete resorption of the mesonephric ducts.[1] They are uncommon, usually asymptomatic and are diagnosed incidentally at delivery or during a routine gynaecologic examination. Less commonly, MDAs can cause infertility, endometriosis, recurrent miscarriages and symptoms arising from an obstructed reproductive tract.[1]

Mayer-Rokitansky-Küster-Hauser syndrome (MRKH), also known as müllerian agenesis, is a congenital anomaly characterised by vaginal agenesis associated with, in most cases, a spectrum of other genitourinary tract abnormalities. MRKH syndrome belongs to Class I mullerian duct anomalies.[2] MRKH is a specific type of mullerian duct malformation characterised by congenital absence or hypoplasia of the uterus and upper two-thirds of the vagina in both phenotypically and karyotypically normal females with functional ovaries.[3]

It is the second most common cause of primary amenorrhoea.[4] A case of MRKH syndrome was reported in a 30-year-old female in the tropics of Northern Nigeria.[5] Mullerian agenesis occurs in approximately 1 in 5000 live births.[6] Due to the paucity of data in our environment, it is imperative to report this case of mullerian agenesis in an 18-year-old female. The report will also open a research window for mullerian agenesis and other uterine disorders in our environment.

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Case Report

Miss LOC is an 18-year-old girl who presented with a history of primary amenorrhoea and failure to develop secondary sex characteristics. She is the only daughter of a widow. According to the mother, her pregnancy and birth histories were normal, and delivery was by spontaneous vaginal delivery. The mother was an elderly primigravida. The patient had satisfactory developmental milestones and had completed secondary school at the time of presentation.

There was no associated history of difficulty with urination, cyclical pelvic or abdominal discomfort or lower abdominal pains. The patient was not sexually active; therefore, apareunia was not elicited. There were no features of hyperandrogenism such as excessive hair growth, male pattern hoarseness of voice or baldness.

On physical examination, she had an average physical presentation and stature with a height of 150 cm and weighed 42 kg. She was not pale, afebrile, anicteric and had no facial acne, hirsutism or striae. Her pulse rate, respiratory rate and blood pressure were 80 beats/min, 16 breaths/min and 100/60 mmHg, respectively. The first and second heart sounds were heard without murmurs. The breasts were rudimentary (tanner Stage 2), and she lacked axillary and pubic hairs. There were no features suggestive of facial dysmorphism, webbed digits or skeletal anomalies. Abdominal examination revealed a full and soft abdomen. No mass or swelling was palpated. The clitoris, pudendal cleft and external urethral meatus appeared normal. A pelvic examination revealed a 1.5 cm blind-ending vaginal pouch. On digital rectal examination, there was a good anal sphincteric tone with no palpable uterus or cervix. A clinical diagnosis of congenital müllerian agenesis was made. Serum electrolytes, as well as hormonal assays, were within normal limits.

A radiologic evaluation was done using ultrasonography and magnetic resonance imaging (MRI). Abdominal ultrasonographic examination demonstrated normal kidneys, liver, spleen and other intra-abdominal organs. Greyscale transabdominal longitudinal and transverse images of the pelvic ultrasound scan showed a full urinary bladder only; the uterus and the ovaries were absent [Figures 1 and 2]. MRI of the pelvis with better soft-tissue resolution also revealed an absence of the uterus, as shown in Figures 3-8.

Figure 3 is a T2-weighted parasagittal image of the pelvis showing the hyperintense full urinary bladder, signal void pubis symphysis inferoanterior to the urinary bladder and the rectum posteriorly. The uterus was not visualised. Figure 4 shows

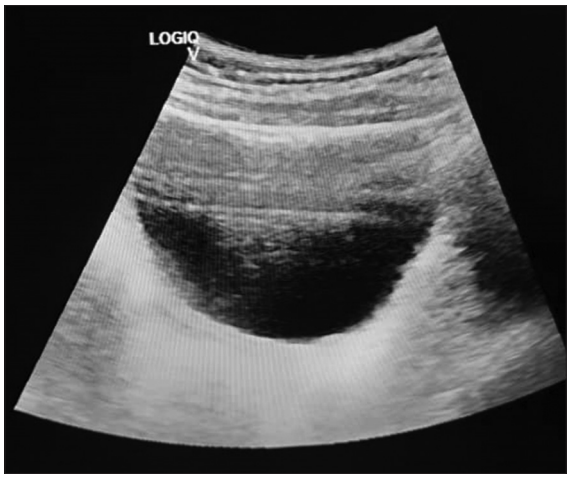

Figure 1: Greyscale transabdominal ultrasound longitudinal image of the pelvis showing a full urinary bladder while the uterus and adnexa, which are supposed to be posteroinferior to the urinary bladder, are not demonstrated

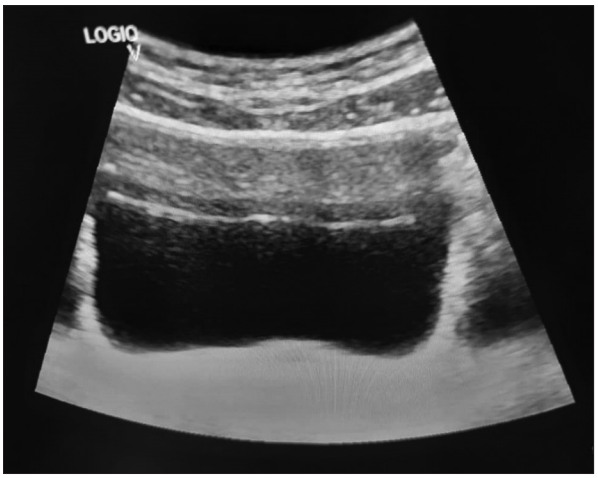

Figure 2: Greyscale transabdominal ultrasound transverse image of the pelvis showing a full urinary bladder while the uterus, which is supposed to be posterior to the urinary bladder, is not demonstrated

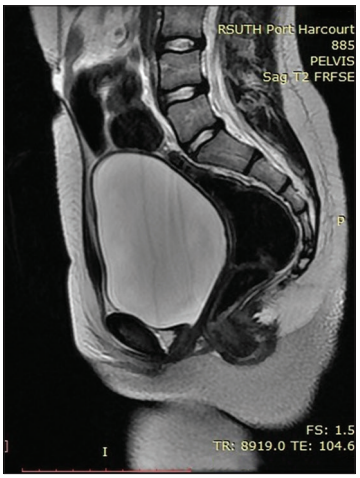

Figure 3: T2-weighted parasagittal magnetic resonance image of the pelvis showing the hyperintense full urinary bladder, signal void pubis symphysis, which is inferoanterior to the urinary bladder and the rectum posteriorly. The uterus and adnexa are not visualised

a T1-weighted parasagittal image of the pelvis showing the hypointense full urinary bladder, iso- to hyperintense pubis symphysis located inferoanterior to the urinary bladder. The rectum and sacrum are located posteriorly, while the uterus was not demonstrated. T1-weighted axial section image of the pelvis also demonstrated the hypointense full urinary bladder, hyperintense ischia laterally, with the rectum and its contents visualised inferiorly. The uterus was not visualised [Figure 5]. Similar findings were seen in other planes and sequences in Figures 6-8. A radiologic diagnosis of MRKH syndrome in an 18-year-old female was made. The patient was counselled on different management options including the options for fertility in the future. The mother vehemently rejected the option of surgical treatment, and both of them have not been seen in the clinic since then.

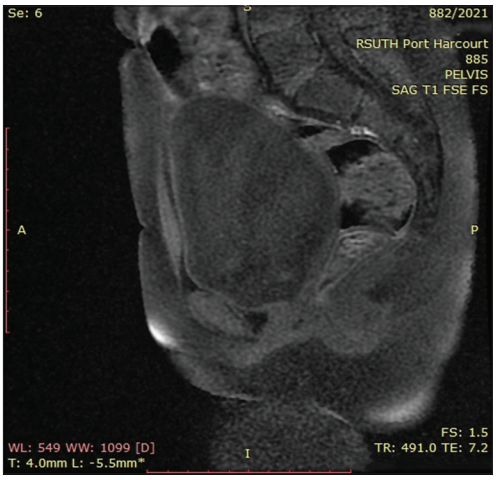

Figure 4: T1-weighted parasagittal magnetic resonance image of the pelvis showing the intermediate signal intense full urinary bladder, iso- to hyperintense pubis symphysis located inferoanterior to the urinary bladder. The rectum and sacrum are located more posteriorly. The uterus and adnexa are not visualised

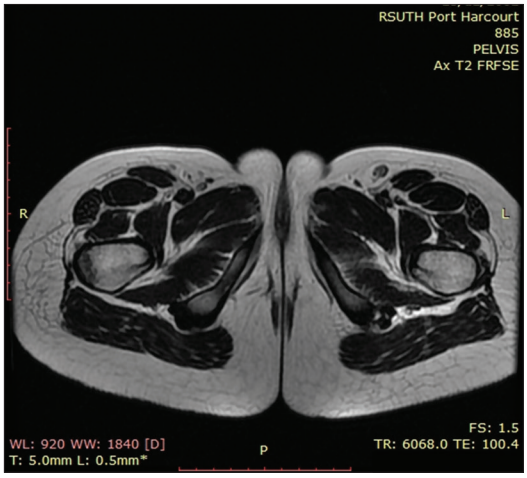

Figure 6: T1-weighted axial magnetic resonance image of the pelvis showing the head of femurs, labium majorum and anal cleft. The uterus is not seen. One of the axial series

Discussion

Background

MRKH syndrome dates back to the Hippocratic era (460 B. C.–377 B. C.).[7] However, it was first described by Mayer in 1829 and then by Rokitansky in 1838. Hauser and Schreiner, in 1961, described the distinguishing features of MRKH syndrome from androgen insensitivity syndrome.[8] It is a congenital disorder of the female characterised by aplasia of the uterus and the upper two-third of the vagina in females with typical secondary sexual characteristics and XX karyotype.[9] Turner's syndrome is the most common cause of primary amenorrhoea, followed by müllerian agenesis, which has an incidence of 1:5000 live female births.[6,10,11] Although some cases are familial, the majority of the cases are sporadic.[12]

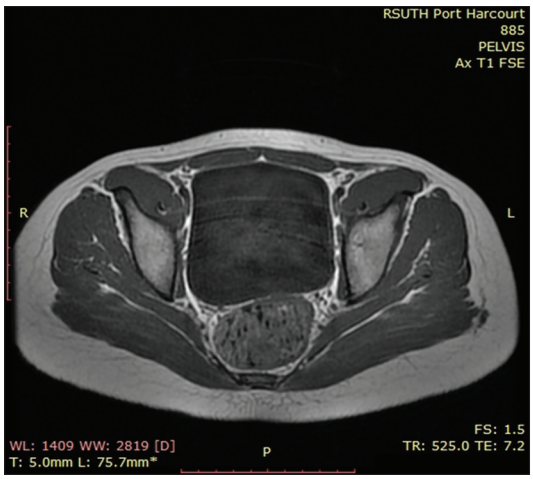

Figure 5: T1-weighted axial magnetic resonance image of the pelvis showing the hypointense full urinary bladder, hyperintense ischia laterally, while the rectum and its content as visualised inferiorly below the urinary bladder. The uterus is not visualised

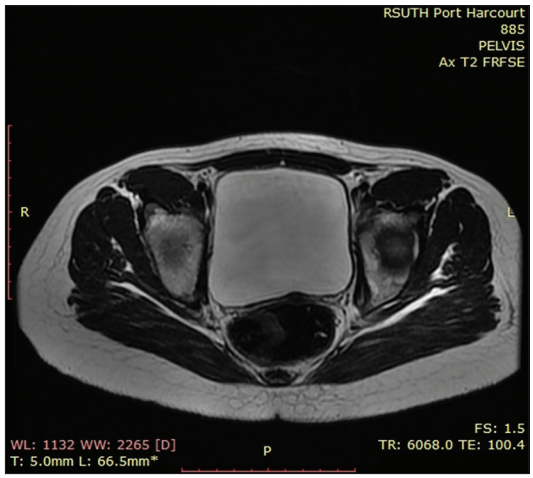

Figure 7: T2-weighted axial magnetic resonance image of the pelvis showing the hyperintense full urinary bladder, pelvic muscles and rectum with its content is visualised inferiorly. The uterus, fallopian tubes and ovaries are not visualised at their supposed locations

MRKH syndrome is subdivided into two types. Type I is the Rokitansky sequence, an isolated disorder characterised by an isolated absence of the proximal two-thirds of the vagina and the uterus; Type II is the MURCS syndrome associated with müllerian duct aplasia, renal dysplasia and cervicothoracic somite anomalies.[9]

Epidemiology

The prevalence of mullerian agenesis is estimated to be about 1 in 5000 live female births,[6,10] with the average age at diagnosis ranging from 10 to 18 years.[13] Congenital absence of the vagina occurs in 0.001%–0.025% of the population.[14]

Aetiology

The aetiology of MRKH syndrome is said to be due to a wide range of malformations and defects encountered during embryogenesis[12] following the activation of mutation of the antimullerian hormone or by a mutation against the antimullerian hormone receptor, which eventually results in defective embryogenesis.[12,15] The WNT4 gene has also been implicated in the aetiology of the atypical type of müllerian agenesis.[7] The gene, which is located on the short arm (p) of chromosome 1, promotes female sex development and represses that of males. Thus, the defective genetic activity of the gene results in defective embryogenesis.

Clinical presentation

They are uncommon, asymptomatic and diagnosed incidentally at the time of delivery or during a routine gynaecologic examination. Primary amenorrhoea is one of the most familiar presentations as with our index patient.

The vagina may be shortened with associated difficult, painful intercourse, while others may present fertility-related issues.[15]

Diagnosis

The diagnostic features encompass clinical history, findings on physical examination, genetic analysis and radiologic evaluation.[16] Radiologic imaging modalities have played a pivotal role in the diagnosis of mullerian agenesis and other urogenital malformations. Pelvic ultrasonography, hysterosalpingography, computed tomography and MRI will show the absence of the uterus, ovaries or any other associated abnormality.

Pelvic MRI is highly effective in demonstrating the normal anatomy of the uterus, as seen in Figure 9.[17] Image[17] shows the zonal anatomy of the uterus with the endometrium hyperintense, hypointense junctional zone and the intermediate signal intensity myometrium,[17] which are not demonstrated in Figures 3-8.

In the diagnosis of the index case, ultrasonography was first done as shown in Figures 1 and 2, which was later complemented by MRI [Figures 3-8] which also showed the absence of the uterus.

Treatment

Surgical approaches to create a functional vaginal is standard, while uterine transplant has been performed in some people with MRKH. However, uterine transplant surgery is still in its experimental stage.[7,13,18]

Psychotherapy and counselling concerning lifestyle modifications have been incorporated into the treatment

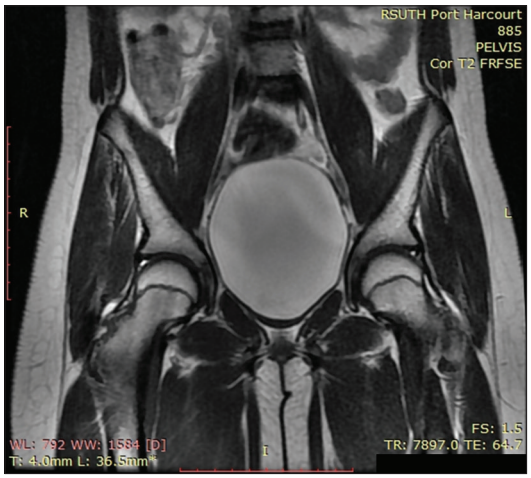

Figure 8: T2-weighted coronal magnetic resonance image of the pelvis showing the hyperintense full urinary bladder and hyperintense femoral epiphysis laterally while the uterus, fallopian tubes and ovaries are not visualised at their supposed locations

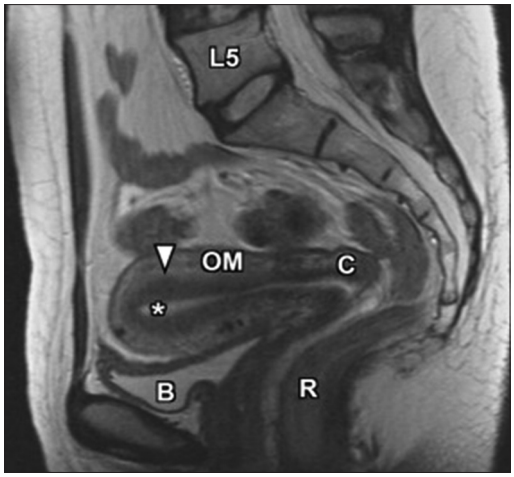

Figure 9: Sagittal T2-weighted images of the female pelvis showing the zonal anatomy of the uterus. Hyperintense endometrium (asterisk), hypointense junctional zone (arrowhead) and intermediate signal intensity of the myometrium. B, bladder; C, cervix; L5, L5 vertebral level; O.M., outer myometrium; R, rectum; Image is adapted from (Normal and Variant Pelvic Anatomy on MRI. https://radiologykey.com/normal-and-variant-pelvic-anatomy-on-mri/. Last accessed on 2021 Apr 1)[16]

options due to the enormous psychological effect of the disorder.[18]

Summary

Miss LOC is an 18-year-old female who presented with a history of primary amenorrhoea and the absence of secondary sexual characteristics. Radiologic investigations showed the absence of the uterus and ovaries in both pelvic ultrasonography and MRI. The patient and her mother were counselled on treatment options which they declined.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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