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Case Report **Obstetrics & Gynaecology**

A Case of Mayer-Rokitansky-Küster-Hauser Syndrome: Case Report of a 22-Year-Old Undergraduate with Primary Amenorrhoea

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Abstract

Amenorrhea in a woman of reproductive age can be sporadic, temporary or permanent. It can result from lesion affecting the hypothalamic-pituitary-ovarian axis, uterus and vagina. A woman is said to have primary amenorrhoea if she fails to attain menarche at the age of fifteen or at thirteen years without evidence of secondary sexual characteristics. The causes of primary amenorrhoea are broadly classified into constitutional delay, anatomical causes such as Mayer-Rokitansky-Küster-Hauser (MRKH) or genetic causes (gonadal dysgenesis) and other physiological causes including isolated deficiency of gonadotropin releasing hormone. MRKH (Mullerian agenesis) is a rare syndromic disorder that usually occurs when there is agenesis or hypoplasia of the paramesonephric ducts (the precursor for the development of the uterus, fallopian tubes and upper two-third of the vagina). We report on a case of a 22-year-old undergraduate with primary amenorrhoea and subsequent diagnosis of MRKH.

Keywords: MRKH, Primary Amenorrhoea, Mullerian agenesis, Nigeria.

INTRODUCTION

Mayer–Rokitansky–Küster–Hauser (MRKH) syndrome is a very rare congenital defect that affects the reproductive system in females [1]. This malformation results in agenesis of the uterus, cervix and upper two-thirds of the vagina secondary to failure of migration of paramesonephric or mullerian ducts to the urogenital sinus. In women of reproductive age, MRKH is a common cause of primary amenorrhea (second only after Turner's syndrome) [2]. It occurs in women of normal karyotype and secondary sexual characteristics [1, 2]. Evidence has shown that MRKH occurs in 1 in 4,500 to 5,000 of biologically assigned female gender at birth [1].

Women with MRKH are usually asymptomatic until puberty when they present with absence menses despite normal secondary sexual characteristics. The severity of the clinical presentation depends on if it is type 1 MRKH which is associated with uterine agenesis or type 2 with other associated anomalies and varied manifestation [3-5].

Although, medical history of absent menses and physical examination finding of normal sexual development may be suggestive of Mayer-Rokitansky-Küster-hauser syndrome, a combination of these with normal hormone profile, genetic testing (karyotype), pelvic ultrasound scan and Magnetic resonance imaging plays a vital role in making the diagnosis [1, 6, 7].

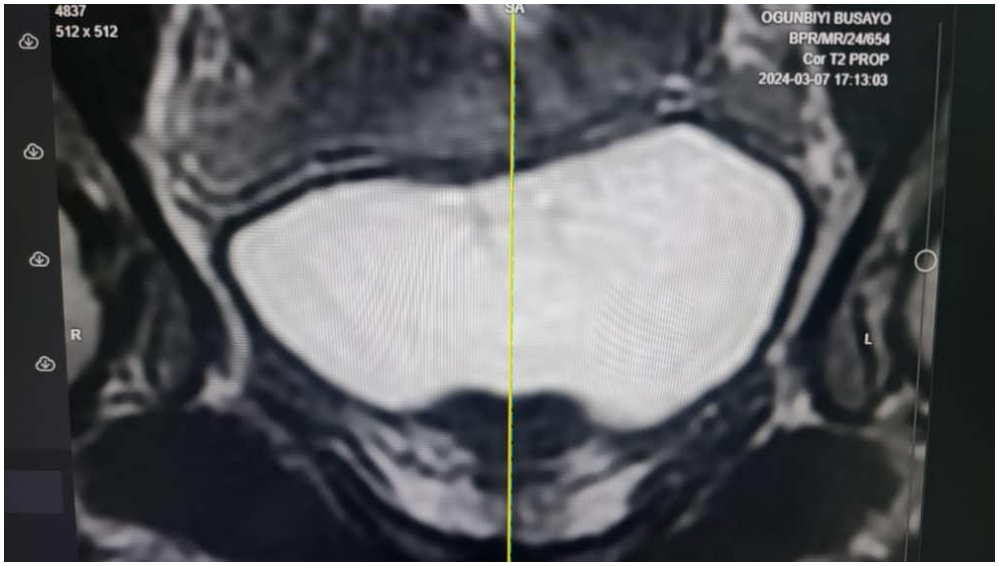
In addition, treatment of MRKH depends on the clinical presentation and the desire of the woman. It ranges from psychological counselling to emotional support, surgery for creation of a neovagina and possible referral for assisted conception (surrogacy) as need arises [1, 7].

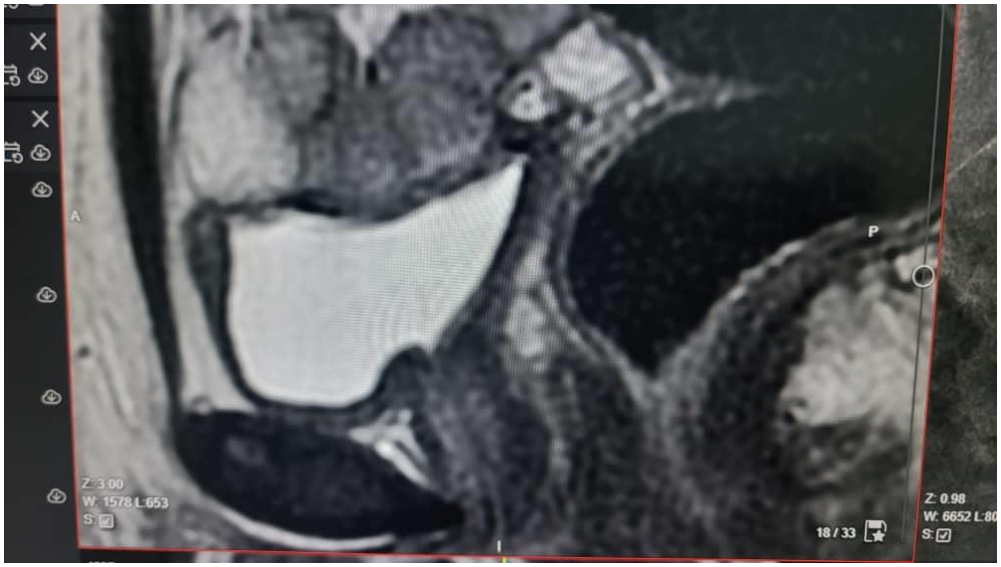
CASE

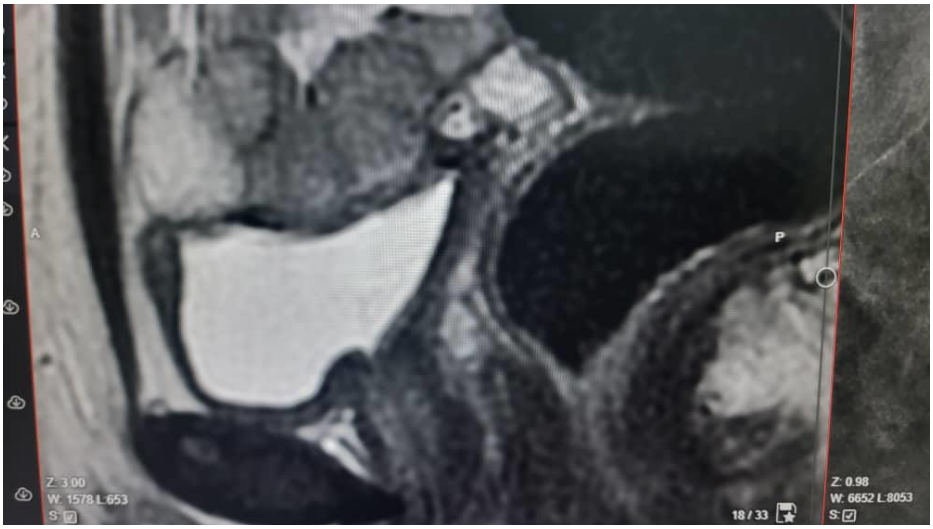
A 22-year-old single undergraduate came to our gynaecology outpatient clinic on referral from a private hospital with the chief complaints of primary amenorrhea. She had normal secondary sexual characteristics, (she attained thelarche at 13 years, pubarche subsequently). She presented to a private hospital previously where pelvic ultrasonography was done, the result of which showed hypoplastic uterus with normal sized ovaries bilaterally. She has no known chronic medical condition. She has no history of previous surgeries. Her mother and sisters had normal development and puberty. She does not consume alcoholic beverages and does not smoke or take other recreational drugs.

Her general physical examination findings were normal. She weighed 68kg, height was 156 centimeters, and all other vital signs were, heart rate was 91 beats per minute, Blood pressure was 121/56mmHg, SP02 98%. Her breast examination revealed Tanner stage 5 for both breasts, which is normal for her age. Her pelvic examination revealed normal labia majora, labia minora, normal pubic hair distribution, and external urethral meatus.

Routine blood investigation done for her at presentation showed hemoglobin 13.5 gm%, total white blood cell count of 10,000/mm3, platelets of 200,000/mm3 and peripheral blood karyotype 46, XX. She did a hormone profile which showed normal luteinizing hormone (LH), follicle stimulating hormone (FSH), prolactin, progesterone, estradiol, and testosterone levels. Her thyroid function test was also normal. Abdominopelvic Magnetic resonance imaging (MRI) revealed agenesis of uterus, cervix and upper two thirds of vagina, normal ovaries, bladder, urethra and rectum, no ascites or pelvic adenopathy. A diagnosis of Mayer Rokitansky Kuster Häuser Syndrome (MRKH) was made. She was counseled on the subsequent management and outcome. She is being worked up for vaginoplasty.







DISCUSSION

We have presented a case of a 22-year-old undergraduate with primary amenorrhoea secondary to MRKH. MRKH is a syndrome with varied manifestation that can affect genitourinary systems [4]. The exact cause of this spectrum of anomalies is not known, however a mutation in WNT4 gene has been suggested [3]. It is an autosomal dominant anomaly resulting from abnormal development of paramesonephric duct. Based on the findings in recent literature, MRKH can be divided into two subtypes [2-4]. Type 1, which is commonly associated with anomalies in reproductive organs (uterus and vagina) while type 2 affects other organ systems such as kidneys, spine, ears [1-3]. Type 2 MRKH closely mirrors MURCS association as cervicothoracic somite anomalies, unilateral renal agenesis and conductive deafness are seen in both [1].

Just as in the index case, most women with MRKH present with primary amenorrhoea with normal growth and pubertal development (normal secondary sexual characteristics) [2, 5]. The presence of normal secondary sexual characteristics may be attributed to the fact both ovaries are not affected in MRKH.

Several imaging techniques play a significant role in the diagnosis of MRKH. Three Dimensional transvaginal or abdominopelvic ultrasound is the initial imaging of choice. However, abdominopelvic abdomino-pelvic Magnetic Resonant Imaging gives a better tissue resolution and is the diagnosis of choice [5-7]. Our patient had a transvaginal ultrasound prior to presentation which was suggestive of absent uterus. MRI was carried out in our facility which led to the diagnosis. Her hormone profile expectedly was normal.

Just as important, the most important step in the effective management of paramesonephric anomalies starts with making appropriate diagnosis, presence or absence of other congenital anomalies, and consideration of the psychological and emotional impact on the patient. The treatment ranges from psychological counselling, emotional support, non-surgical vaginal dilatation, vaginoplasty, uterine transplant and guidance on the options available for conception via assisted reproductive techniques.

CONCLUSION

Informed Consent: Our patient agreed for her anonymized data to be published so as to advance medical knowledge.

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