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ULTRASOUND DIAGNOSIS OF MAYER-ROKITANSKY-KÜSTER-HAUSER SYNDROME IN A RESOURCE-LIMITED SETTING: A CASE REPORT FROM NIGERIA

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ABSTRACT

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a congenital Müllerian anomaly and an important cause of primary amenorrhoea. We report a 27-year-old Nigerian woman with normal secondary sexual characteristics. Transabdominal pelvic ultrasound showed complete uterine agenesis, with normal bilateral ovaries and kidneys, consistent with type I MRKH syndrome. This case demonstrates that ultrasound can provide a reliable, accessible and affordable diagnosis in resource-limited settings where magnetic resonance imaging is unavailable or unaffordable, enabling timely counselling, psychological support and multidisciplinary management.

Keywords: MRKH syndrome, uterine agenesis, ultrasound, primary amenorrhoea, Nigeria

Introduction

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a congenital Müllerian duct anomaly defined by agenesis or hypoplasia of the uterus and upper two-thirds of the vagina in phenotypically normal females with a 46,XX karyotype and preserved ovarian function. [1,2] It accounts for approximately 10-15% of primary amenorrhoea cases, with an estimated prevalence of 1 in 4,500-5,000 female births.[3].

Embryologically, MRKH results from failure of Müllerian duct development between the 6th and 12th weeks of gestation, while the ovaries develop normally from the gonadal ridge.[4,5] The syndrome is classified as type I (isolated uterovaginal agenesis) or type II (associated with renal, skeletal, cardiac or auditory anomalies).[1,6].

Affected girls undergo normal puberty, including breast development and pubic hair growth, but menstruation does not occur, resulting in primary amenorrhoea. Because the anomaly is congenital, it often goes unnoticed until adolescence.[1] Ultrasonography is the first-line imaging modality because of its accessibility and cost-effectiveness, especially in low-resource settings. MRI provides superior anatomical detail and is considered the gold standard, [7,8] but it is frequently unavailable in rural and semi-urban Nigeria.

This report describes a case of MRKH syndrome diagnosed using ultrasound in Nigeria and emphasises the important role of radiographers in resource-constrained environments.

Case Presentation

A 27-year-old unmarried Nigerian woman with no previous pregnancies presented to the radiology department with a lifelong history of primary amenorrhoea. She had never menstruated. She reported normal development of secondary sexual characteristics: breast development began at approximately 12 years of age, and pubic and axillary hair developed at an age-appropriate time. There was no history of cyclical pelvic pain, previous surgery or chronic illness. She reported normal libido and mild dyspareunia. No family history of a similar condition was reported.

On physical examination, she was a phenotypically normal female with Tanner stage V breast development and normal external genitalia. Pelvic examination revealed a shallow vaginal dimple approximately 2-3 cm deep, with no identifiable vaginal canal beyond this

point. No cervix was palpable on rectoabdominal examination, and no abdominal or pelvic masses were palpable. On further enquiry, she reported having attended several primary healthcare centres on multiple occasions, but no imaging or hormonal investigation had been performed to establish a diagnosis. Hormonal assays performed at an external laboratory showed normal follicular-phase levels of follicle-stimulating hormone (FSH), luteinising hormone (LH) and oestradiol, indicating intact ovarian function. Based on the clinical presentation and normal hormone levels, a Müllerian anomaly was strongly suspected. The differential diagnoses included androgen insensitivity syndrome (AIS) and gonadal dysgenesis.

Imaging Findings

Transabdominal pelvic ultrasound was performed using a 3.5-5 MHz curvilinear transducer with adequate bladder filling. The uterus was not visualized despite optimal scanning conditions and assessment in multiple imaging planes (Figure I). Both ovaries were identified and appeared normal in size, morphology and follicular activity (Figures II and III). No adnexal mass or pelvic free fluid was identified. A limited abdominal survey was performed during the same ultrasound examination to screen for renal abnormalities. Both kidneys were visualised in their normal anatomical positions, with no evidence of agenesis, ectopia or hydronephrosis. Renal ultrasound is recommended in all suspected cases of MRKH syndrome because 15-40% of patients have associated urological abnormalities, most commonly unilateral renal agenesis or a pelvic kidney. The absence of renal anomalies in this patient supported classification as type I MRKH syndrome. The findings were consistent with uterine agenesis and were suggestive of type I MRKH syndrome.

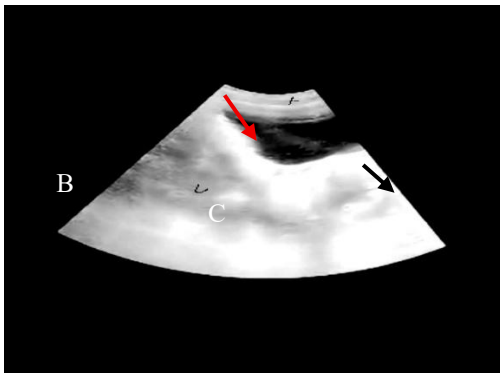

Figure I. Sagittal ultrasound image showing the normally distended urinary bladder (A), loops of bowel posterior to the bladder (B), and the empty region where the uterine body and cervix would normally be visualised (C), consistent with uterine agenesis in type I MRKH syndrome.

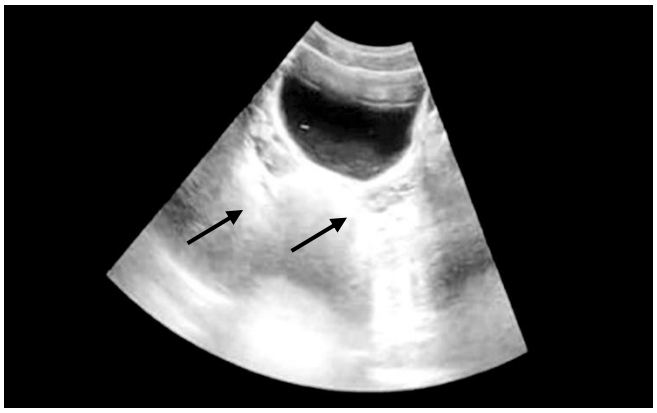

Figure II (transverse view): Arrows indicates the right ovary with normal morphology and several small follicles.

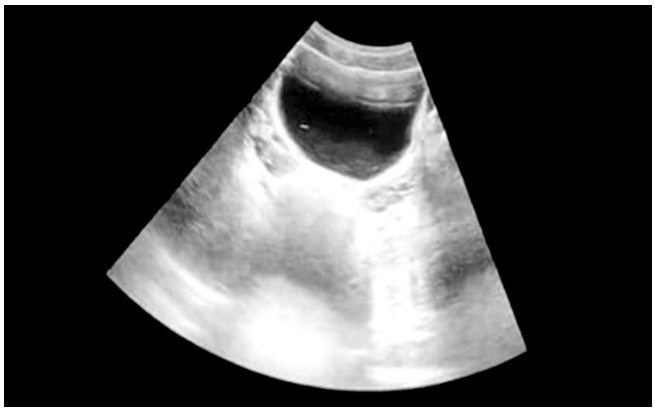

Figure III (transverse view): Arrow indicates the left ovary, also normal in size and appearance.

Following the ultrasound diagnosis, the patient was referred to the gynaecology and psychology departments. She received detailed counselling about MRKH syndrome, including its non-progressive nature and the absence of an increased risk of malignancy. Vaginal dilatation therapy was offered as the first-line, non-surgical treatment for vaginal agenesis; she accepted and was trained in its use. At the 6-month follow-up, she reported satisfactory vaginal length for intercourse and good emotional adaptation. Fertility options, including in vitro fertilisation with gestational surrogacy, were discussed but not pursued. She expressed interest in future uterine transplantation should it become accessible. She remains under annual psychosocial follow-up.

Discussion

MRKH syndrome is a well-recognised cause of primary amenorrhoea, yet diagnosis is often delayed in Nigeria because of limited access to imaging and low

awareness among primary healthcare providers.[9] This case demonstrates how timely ultrasound may prevent years of unnecessary investigation and psychological distress. This case is notable because it is one of the few documented Nigerian cases in which ultrasound served as the definitive diagnostic tool rather than as a screening examination before MRI. In northern Nigeria, Yakasai et al.[4] reported a 22-year-old woman with type II MRKH syndrome confirmed by MRI and associated unilateral renal agenesis. In contrast, the present patient had no associated renal anomalies, consistent with type I MRKH syndrome. Similarly, Behr et al.[10] described a 19-year-old patient who was initially misdiagnosed with an imperforate hymen, leading to an unnecessary examination under anaesthesia. This case reinforces the value of early ultrasound assessment in adolescents and young adults presenting with primary amenorrhoea and normal secondary sexual characteristics, particularly in low-resource settings where invasive procedures should be avoided.

Although not a primary case report, a comprehensive review by Ukoaka et al.[11] highlighted that menstrual irregularities, including primary amenorrhoea, are under-investigated among Nigerian women because of poverty, low health literacy and limited access to imaging equipment. The present case provides a practical example of how a single, low-cost ultrasound examination resolved a decade of diagnostic uncertainty.

MRI was not performed because of cost and logistical constraints. However, ultrasound provided a confident diagnosis of uterine agenesis with normal ovaries, and no immediate surgical intervention was planned because vaginal dilatation was the first-line treatment. MRI would therefore not have changed the immediate management plan. American College of Obstetricians and Gynecologists guidelines state that, in typical type I MRKH syndrome with clear ultrasound findings, MRI is confirmatory but not mandatory.[6]

Chromosomal analysis was not performed, which is a significant limitation of this report. Definitive exclusion of complete androgen insensitivity syndrome (CAIS) requires karyotyping; individuals with CAIS have a 46,XY karyotype and may present with primary amenorrhoea, vaginal hypoplasia and normal female external genitalia. However, the clinical findings in this patient—normal pubic and axillary hair, absence of inguinal masses, and normal ovaries on ultrasound—were more consistent with MRKH syndrome than with CAIS. In CAIS, testes are typically present, often within the abdomen or inguinal canals, and pubic and axillary hair is sparse or absent because of androgen insensitivity. Nevertheless, karyotyping remains the gold standard for excluding CAIS and should be performed whenever feasible in similar cases.[7]

Management of MRKH syndrome requires a multidisciplinary approach involving gynaecological care, psychological support and reproductive counselling. Vaginal dilatation is the first-line treatment for vaginal agenesis; surgical vaginoplasty, such as the McIndoe procedure, is reserved for patients for whom dilatation is unsuccessful or unacceptable.[12] Although affected individuals cannot carry a pregnancy, in vitro fertilisation with gestational surrogacy is an established option, and uterine transplantation is an emerging treatment.[13].

Conclusion

This case demonstrates that transabdominal pelvic ultrasound is a reliable, accessible and affordable tool for diagnosing MRKH syndrome in resource-limited settings where MRI is not feasible. Early detection by radiographers enables timely referral, appropriate counselling and improved quality of life for affected women. Clinicians practising in similar environments should consider MRKH syndrome in any young woman presenting with primary amenorrhoea and normal secondary sexual characteristics and use ultrasound as the first-line imaging modality.

Declarations

Ethical approval: Ethical approval was obtained from the appropriate institutional review board. The approval number was unavailable because the case report was conducted under the institution's general ethical guidelines for case reports.

Patient consent: Informed consent was obtained from the patient for publication of this case report and the accompanying images.

Conflict of interest: The authors declare no conflicts of interest.

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