CASE REPORT

Mullerian Agenesis with Primary Amenorrhea: A Case Report of a Normal Phenotypic Female

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A B S T R A C T

Meyer–Rokitansky–Kuster–Hauser syndrome is a rare congenital abnormality occurring sporadically in females. It is the most common cause of primary amenorrhea. Affected persons usually appear normal on physical examination. This is a case of a 25-year-old woman who presented with primary amenorrhea and, subsequently, had diagnostic laparoscopy to confirm the diagnosis of Mullerian agenesis.

Key words: Laparoscopy, Mayer-Rokitansky-Kuster-Hauser syndrome, Mullerian agenesis, primary amenorrhea

ملخص البحث:

تعتبر متلازمة Meyer–Rokitansky–Kuster–Hauser (MRKH) من الحالات النادرة التي تحدث بشكل متقطع لدى النساء. وهي من أكثر أسباب انقطاع الطمث الأولى. تبدو النساء المصابات بهذه المتلازمة طبيعيات عند الفحص السريري. يعرض الباحثون حالة لسيدة في الخامسة والعشرين من العمر تعاني من انقطاع طمث أولي. تم تشخيص الحالة بواسطة تنظير البطن التشخيصي على أنها ناتجة عن عدم تخلق مولر (Mullerian agenesis).

INTRODUCTION

The findings of a blind-ending vagina, primary amenorrhea and absent uterus are rare occurrences in clinical practice. It occurs in about 1 in 5000 live female births.[1,2] This is eponymously referred to as Meyer–Rokitansky–Kuster–Hauser (MRKH) syndrome and is the most common cause of primary amenorrhea.[3] The condition poses a serious challenge to the attending gynecologist because it is not amenable to conventional forms of assisted reproductive techniques. The challenge to the patient concerned is multifaceted. This includes the distress of discovering that some organs are absent and the difficulty/inability to conceive and have penetrative sexual intercourse.

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We present a report of a 25-year-old woman seen at our unit.

CASE REPORT

A nonsexually active 25-year-old patient presented at the Assisted Reproduction Technology Unit of the University of Ilorin Teaching Hospital, Kwara State, Nigeria, due to primary amenorrhea. She had started developing secondary sexual characteristics at about 14 years of age; the sequence of development could not be recalled. The general physical examination revealed a young woman, 1.46 m tall and weighing 42 kg with a body mass index of 19.7 kg/m^2. Her thumbs were hypoplastic, and she

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had a short neck. Her breasts were well developed at Tanner Stage V. She had normal axillary and pubic hair distribution. The gynecological examination revealed grossly normal vulva; however, the vagina was blind-ending with only the urethral opening being patent. The transabdominal ultrasonography performed did not reveal a uterus. She subsequently had diagnostic laparoscopy and the uterus was not visualized; however, grossly normal fallopian tubes and ovaries were seen bilaterally with evidence of follicular activity [Figure 1]. Karyotyping and hormone assay were also carried out, the results of which were normal.

DISCUSSION

The constellation of features described above points to MRKH syndrome.[1] The typical presentation in this condition is primary amenorrhea. Some women may present with cyclical abdominal pain, and gynecological examination may reveal absent or rudimentary vagina.[2] MRKH syndrome is a form of Mullerian abnormality also known as Mullerian aplasia. It is caused by embryonic growth failure resulting in agenesis or underdevelopment of the vagina or uterus or both.[3] The ovaries are of a different embryologic origin and they are normal in structure and function; thus, patients with this syndrome usually appear normal on physical examination, with normal height and secondary sexual characteristics. The labia majora, labia minora, clitoris, hymen and distal portion of the vagina are usually present because this portion is of a different embryonic origin. The karyotype and hormone profile are also normal,[4] as seen in this case.

There are two subtypes of MRKH: the typical and the atypical forms.[5] The typical form is characterized by laparoscopic/laparotomy findings of Mullerian remnants and normal fallopian tubes. The atypical form shows asymmetric hypoplasia of one or two buds, possible dysplasia of the fallopian tubes with one or more of the anomalies, such as unilateral or bilateral renal agenesis, ectopic kidneys or horseshoe kidneys in 40–60% of cases. Other abnormalities include cervicothoracic (asymmetric, fused or wedged vertebrae, scoliosis and Klippel–Feil anomaly), hearing defects and varying degrees of digital anomalies. The most severe form of the atypical form is referred to as Mullerian renal cervical somite association.[5,6]

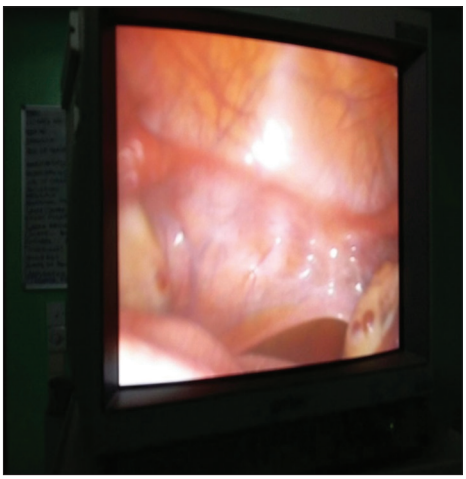

Figure 1: Operative findings: Absent uterus with blind ended vagina, grossly normal fallopian tubes and ovaries with evidence of follicular activities.

The diagnosis is confirmed mainly with imaging modalities of ultrasonography and magnetic resonance imaging. These help to definitively characterize the anatomy. The preferred ultrasonography is the three-dimensional mode. Laparoscopy is considered when the earlier mentioned modalities have not yielded adequate information or in the treatment of rudimentary uterine horns.[7] Our patient had a laparoscopy because it was readily available and accessible in our center.[8]

Karyotyping is also needed in establishing the diagnosis of MRKH syndrome as it helps in differentiating it from the other clinical conditions that appear similar in appearances such as androgen insensitivity syndrome and 17α-hydroxylase syndrome.[9] However, the presence of hypoplastic thumbs and a short neck strengthened the diagnosis of MRKH syndrome.[5]

The management of this condition involves the exclusion of other clinical malformations that will hinder the well-being of this patient. The treatment is multidisciplinary and involves surgical and nonsurgical treatment options including the creation of a neovagina to have a normal sex life. Vestiges of the uterus can be removed to avoid the development of endometriosis.[1] The timing of the surgical or nonsurgical creation of the neovagina should be planned for when the woman is emotionally mature and expresses the desire for correction.

The nonsurgical creation of the neovagina requires patients to manually place successive dilators on the vaginal dimple for 30 min to 2 h/day. In highly motivated patients, a functional vagina will be created over several months. However, following successful dilation, intermittent dilation therapy will be required in those that are not sexually active.[9] Surgical creation of a neovagina is an option for patients who prefer the surgical correction or those who were unsuccessful with

dilators. Surgical correction just like the nonsurgical options requires postoperative dilation or regular sexual intercourse to maintain adequate vaginal length and diameter.[9]

Surgery aims to create a vaginal canal in the correct axis of adequate size and secretory capacity to allow intercourse. A procedure commonly done involves dissection of space between the rectum and the bladder, placement of a mold into the space covered with a split-thickness skin graft. After healing, serial dilation is done to prevent skin graft contracture. A neovagina can also be created laparoscopically. Other forms of grafts that can be used include buccal mucosa, bowel mucosa and amnion.[9]

Routine gynecologic care is expedient in women undergoing therapy to optimize and maximize the care. In Africa, where a high premium is placed on having biological children, assisted reproduction with surrogate mothers can be attempted, especially in the presence of functional ovaries. The other available option is adoption.

CONCLUSION

MRKH syndrome is a rare anomaly of the Mullerian duct. Despite the clinical management options available, the distress of having such a condition is better managed with support from psychologists, counselors and a strong social and family support group.

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Conflicts of interest

There are no conflicts of interest.

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