Psychosexual and social consequences in a woman with undiagnosed Mayer-Rokitansky-Küster-Hauser syndrome

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Summary

Purpose: To report a woman with devastating psychosexual and social consequences as a result of undiagnosed Mayer-Rokitansky-Küster-Hauser syndrome (MRKH) syndrome. *Materials and Methods:* An 18-year-old woman was referred after being divorced for "absent vagina and uterus". On examination, secondary sexual characteristics were normal. Vaginal and rectal examinations revealed absent vagina and uterus. Investigations showed normal hormonal profile, 46 XX karyotype, and normal intravenous pyelography. Pelvic ultrasonography and magnetic resonance imaging (MRI) confirmed the absence of the uterus and presence bilateral ovaries. *Results:* A diagnosis of MRKH syndrome was made and she underwent successful modified laparoscopic Vecchietti operation for creation of a new vagina. Vaginal dilators were used after the surgery. Two years of follow up confirmed that vaginal length was about ten cm. *Conclusion:* The proper diagnosis, counseling, and prompt treatment of MRKH syndrome can prevent tragic consequences.

Key words: Intercourse; Mayer-Rokitansky-Küster-Hauser syndrome..

Introduction

Mayer-Rokitansky-Küster-Hauser syndrome (MRKH) is defined as the congenital absence of the vagina and of the uterus [1]. Anomalies of the urinary tract and the skeletal system are commonly associated with the syndrome. The exact incidence of the syndrome is unknown. It is estimated to be of 1:1,500 to1:4,000 infants [2]. The etiology remains unclear. It has been considered to be a multifactorial anomaly, but there is some evidence to suggest a genetic defect that is transmitted as an autosomal dominant trait with incomplete penetrance and variable expressivity [3]. The condition usually goes unperceived until puberty. Then the clinical presentation is usually primary amenorrhea, with a normal ovarian function, normal female karyotype, and secondary sexual characteristics. Anomalies of the urinary tract and the skeletal system are commonly associated with the syndrome. In the present case, the objective was to highlight an atypical presentation of MRKH syndrome.

Case Report

An 18-years old woman was referred to King Abdulaziz University Hospital after being divorced for "absent vagina and uterus." She was married for five months and sought medical advice for painful and difficult intercourse. She was told that there was congenital absence of the vagina and the uterus and therefore cannot have children. Consequently, she was divorced because of the sexual difficulties. She was seen four years ago for primary amenorrhea but was re-assured. On examination, her weight was 60 kg and height was 168 cm. Secondary sexual characteristics were normal. Vaginal and rectal examinations revealed absent vagina (Figure 1) and uterus. Investigations showed normal hormonal profile, 46 XX karyotype, and normal intravenous pyelography. Pelvic ultrasonography and magnetic resonance imaging (MRI) confirmed the absence of the uterus and presence bilateral ovaries. A diagnosis of MRKH syndrome was made and she underwent successful laparoscopic Vecchietti operation for creation of a new vagina [4]. Vaginal dilators were used after the surgery. Two- year of follow up confirmed that vaginal length was about ten cm and that the vaginal width easily accommodated two fingers.

Discussion

MRKH syndrome is the second most frequent cause of primary amenorrhea. Treatment of MRKH syndrome is controversial. Numerous non-surgical and surgical approaches exist in the literature. The aim is to create a neovagina of adequate size and physiologic condition to permit normal sexual intercourse. The American College of Obstetricians and Gynecologists recommends non-surgical vaginal dilatation method as a first-line approach [5]. Edmonds *et al.* in 2012, reported achieving successful vaginal length (de-

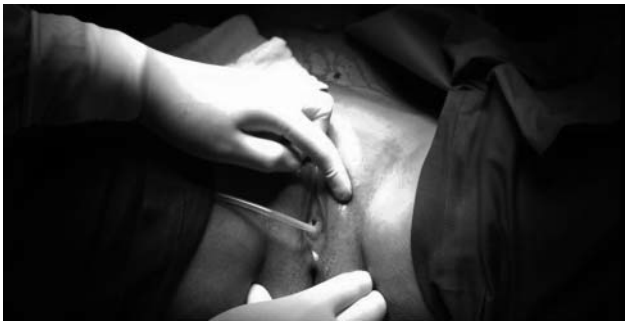

Figure 1. — Preoperative photography showing absent vagina.

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fined as greater than six cm in length and maximum width throughout the vagina and especially at the apex) and sexual function in 232 (94.9%) women out of 245 women using vaginal dilatation [6]. The surgical approaches include many vaginal (McIndoe, Sheare's, and modified Williams), laparoscopic (Vecchietti and Davydov), and open transabdominal procedures [7]. In the United States the most commonly used operation for the creation of a neo-vagina is the McIndoe, while in Europe the Vecchietti procedure is gaining popularity [8]. The advantage of the laparoscopic Vecchietti method is the creation of a longer neovagina (9.6 cm mean length) with a normal anatomy, histomorphology, and functionality in 47.5 minutes (mean operative time) and 4.8 days (mean traction days) [4]. Because vaginal malformations are rare and occur on a wide spectrum, it is frequently difficult to diagnose. This is illustrated in the present case. She was undiagnosed when she presented with primary amenorrhea. It is of paramount importance to understand the anatomy, preparedness, preference, and expectations of each individual case to offer optimal treatment. The aim is to maintain the woman's self-integrity and minimize the consequences of the diagnosis of MRKH syndrome. Creation of a neovagina may help in having sustained marital relationship. Unfortunately, as a result of the lack of the proper diagnosis and treatment, the present case was subjected to the devastating psychosexual and social consequences. Timely creation of a functional neovagina that is satisfactory for sexual intercourse is an essential part of the management of MRKH syndrome to minimize short and life-long consequences.

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