

CASE REPORT

CONGENITAL ABSENCE OF UTERUS

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ABSTRACT

These are two case reports of females with primary amenorrhea, well developed secondary sexual characteristics and congenital absence of uterus. The incidence of Mayer-Rokitansky-Kuester-Hauser syndrome was not clearly established, but studies indicate a variation of 1/4,000 and 1/5,000 live births of the female sex. The syndrome is characterized by aplasia of the Müllerian duct (uterus and upper two-thirds of the vagina) on a person who has karyotype 46, XX with female phenotype characteristic of primary amenorrhea in adolescence. Treatment is usually delayed until the patient is ready to begin sexual activity.

KEY WORDS: Rokitansky syndrome; Mullerian duct; Karyotype; Primary amenorrhea.

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INTRODUCTION

Anatomical abnormalities of the genital tract account for approximately 1% of cases of amenorrhoea. Congenital malformations of female genitalia are often a challenge for doctors, requiring a great knowledge of embryonic development of the genital tract due to the wide variety of possible diagnoses. In girls with breast development but absent uterus, Mayer-Rokitansky-Kuester-Hauser syndrome is considered as common diagnosis. This syndrome is due to an early development failure of the Mullerian system. Affected girls have a normal XX karyotype, normal ovaries and secondary sexual characteristics. The vagina is absent or hypoplastic.¹

Mayer-Rokitansky-Kuester-Hauser syndrome (MRKH syndrome) is named after its most famous discoverer Baron Karl von Rokitansky (Czechoslovakia, 1804–1878), a physician and professor at the University of Vienna. In 1829 and in 1838, Mayer-Rokitansky described a syndrome that includes agenesis of the uterus and vagina, while Kuester then observed a correlation with urological defects. For this reason, this condition is also known as MRKH syndrome.^{2,3}

CASE REPORT

Case 1: A 17 years old adolescent, with phenotype

of a female, pathan by caste was brought to the Department of Gynaecology and Obstetrics, Mufti Mehmood Teaching Hospital (MMTH), D.I.Khan on June 03, 2019. She presented with off and on abdominal pain associated with burning micturition for four weeks. The mother told that the girl had not attained menarche so far. There was no history of any other significant illnesses since her childhood. She had four older and one younger sister's. None of them had any reproductive tract problems and had attained menarche at the ages of 11 and 12 years. Her parents reported an unremarkable family history and denied congenital anomalies among family members. On examination her height was 5 feet 2 inch while her weight was 60 kg. Her physical exam was notable for Tanner stage IV pubic and axillary hair and Tanner stage V breast development. Her abdomen was soft and non-tender while pelvic examination revealed normal external female genitalia with estrogenized tissues, and a gentle digital examination demonstrated a short 1cm blind ending vaginal pouch. No inguinal or pelvic masses were palpable. Her pelvic ultrasound revealed absent uterus even on full bladder, bilateral ovaries were visualized with normal morphology measuring 4.7 x 2.4 cm right ovary and 2.3 x 1.2 cm left ovary. The urinary bladder, kidneys, rectum, and spine were normal in appearance. Impression was congenital absence of uterus.

Case 2: A 25 years old lady was brought to Department of Gynaecology and Obstetrics, Mufti Mehmood Teaching Hospital (MMTH), on February 14, 2019. The lady was married for 7 years and her husband was abroad. She gave history of primary amenorrhoea and painful intercourse. On examination her height 156

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cm and weight were 65 kg while secondary sexual characters were fully developed corresponding to Tanner IV classification of pubic, axillary hair and well developed breasts. Her abdomen was soft non tender while pelvic examination demonstrated normal female genitalia and a very short blind ended vagina measuring hardly 2cm. Her transvaginal ultrasound demonstrated congenitally absent uterus, both the ovaries were normal with maturing follicles, right ovary measuring 3.6 x 3.4 cm while left ovary measuring 2.8 x 2.2 cm. Rest of the pelvic and abdominal viscera were normal.

Her karyotyping was performed on peripheral blood confirming 46 XX karyotype of normal female.

DISCUSSION

The incidence of Mayer Rokitansky-Küster-Hauser syndrome is not clearly established, but studies indicate a variation of 1/4,000 and 1/5,000 live births of the female sex. The etiology of MRKH unfortunately, remains unclear. The majority of cases appear to be sporadic; however, rising accounts of familial cases with an autosomal dominant mode of inheritance are noted in the literature.^{4,1,6}

The syndrome is characterized by aplasia of the Müllerian duct (uterus and upper two-thirds of the vagina) on a person who has karyotype 46, XX with female phenotype characteristic of primary amenorrhea in adolescence. The lower third of the vagina, the ovaries and external genitalia do not usually have alterations present as distal vagina is embryologically derived from the urogenital sinus.^{5,1,6}

Approximately 40-60% of patients have renal disorders such as unilateral agenesis, horseshoe kidney, ectopic or bilateral utero pelvic obstruction. In addition, 20% had bone changes, thoracocervical asymmetry, spinal fusion, and scoliosis.^{6,7}

Once the diagnosis is certain, management can be divided into two phases, first is devoted to the psychological counseling of the patient and second involves the correction of vaginal anatomy. Some patients may present with already attempted intercourse and this may be in fact entirely satisfactory and therefore no attention needs to be paid to the anatomical side of their management. However it is important that all patients with MRKS needs to be assessed carefully so that appropriate therapy can be instigated at the correct time. A full psychological assessment must be carried out before any treatment is commenced or success will be extremely limited.⁸

Treatment is usually delayed until the patient is ready to begin sexual activity because these patients manifest problems that are devastating and profound. They have feelings of fear and confusion around their sexual orientation and may express feelings of rejection and isolation. The help of a skilled psychologist in managing these patients and a multi-disciplinary

approach means that the outcome will be successful in a holistic way, not merely an anatomical success.⁹

The creation of vagina should always be attempted by non surgical method as the treatment of first choice by using graduated glass dilators for 8 to 10 weeks. With failed non-surgical technique, angioplasty will need to be considered.⁹

The first patient was given all pertinent information regarding MRKH syndrome as well as treatment options. After everything was explained to the patient and her family, it was decided that due to her age, she would continue regular follow ups at the hospital until which time she starts having sexual activity while In our second patient, angioplasty as well as child adoption was suggested by us.

CONCLUSION

The diagnosis and treatment of MRKH syndrome can be difficult. Considering the varying presentation of MRKH and numerous treatment options, it is important that each case be individualized. Any discussion about the cultural, linguistic, financial or psychological issues that ultimately lead the families to decline further evaluation and treatment for such girl needs to be considered. Any study of the realistic barriers to diagnosis and treatment can make a great contribution to the medical community, especially in third world countries.

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CONFLICT OF INTEREST
Authors declare no conflict of interest.
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