CASE REPORT

MAYER-ROKITANSKY-KUSTER-HAUSER SYNDROME II, A RARE CASE REPORT FROM PESHAWAR, PAKISTAN

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ABSTRACT

Objectives: Mayer-Rokitansky-Kuster-Hauser Syndrome (MRKHS) is a rare congenital disorder with an incidence of 1 in 5000 females. It is characterized by uterovaginal aplasia with normal secondary sexual characteristics and genetic karyotype 46XX. The exact etiology of MRKH syndrome is not known. The diagnosis of MRKH mainly depends on imaging studies. Transabdominal ultrasonography is the first line of investigation but abdominopelvic MRI gives more precise and clear information than the prior. The MRI even had been done before but the Ultrasound previously done was not conclusive so I repeated the ultrasound which showed abnormality in the uterus and kidneys. The differential diagnosis included congenital vaginal agenesis, low transverse vaginal septum, androgen insensitivity, and imperforate hymen. This case presents that MRKH syndrome II can occur with normal endocrine function and secondary sexual characteristics. Surgical correction by creating a neovagina is a good treatment method for young females for sexual intercourse in the future.

Keywords: MRKHSII, Ectopic Kidney, Mayer-Rokitansky-Kuster-Hauser syndrome

This article may be cited as: Ullah I, Hussan J, Mayer-Rokitansky-Kuster-Hauser Syndrome II, A Rare Case Report From Peshawar, Pakistan. PMSRJ 2023 May-August;1(2):74-75

INTRODUCTION

Mayer-Rokitansky-Kuster-Hauser-Syndrome (MRKHS) or Mullerian dysgenesis is a rare congenital disorder with an incidence of 1 in 5000 females. MRKHS is characterized by uterovaginal aplasia with normal secondary sexual characteristics and genetic karyotype 46XX[1]. There are two types: Type 1 having only uterovaginal agenesis and type 2 having uterovaginal agenesis with anomalies in the fallopian tube, kidney, spine, heart, and other organs, amenorrhea, and painful sexual intercourse. [1] The exact etiology of MRKH syndrome is not known. Previously, drugs like diethylstilbestrol (DES) and thalidomide were said to have teratogenic causes for MRKH syndrome. [2] Counseling of the patient and neo-vagina creation for sexual intercourse is the mainstay for the management. [3] Here, I report a case of type 2 MRKHS with an ectopic kidney.

CASE REPORT

A 20-year-old virginal female presented for evaluation of primary amenorrhea and weakness despite having normal secondary sexual characteristics. Her two sisters had menarche at 12 and 13 years, respectively. There was no history of amenorrhea in the first and second-degree relatives. Her mother did not confirm exposure to any medication or maternal illness during pregnancy. Other parts of the history were noncontributory. She was from middle socioeconomic family and consumed a mixed diet. She had no past medical history and had not undergone surgery. General physical examination findings were normal. Her body weight was 65 kg, her height was 152cm with a BMI of 28.1 kg/m^2. All other vital signs were stable. Systemic examination was unremarkable with normal heart sounds. Skeletal examination was normal. A breast examination revealed Tanner's stage 5 for both breasts, which is typical for her age. Her genitalia examination revealed normal labia majora, labia minora, normal pubic hair development, and short vagina. Blood investigation revealed hemoglobin of 12.2 gm%, total leukocyte count (TLC) of 10,400/mm^3, platelets of 260,000/mm^3, RBS of 98 and creatinine 1.1. Hormonal levels of luteinizing hormone (LH), follicle-stimulating hormone (FSH), Prolactin, progesterone, and testosterone levels were all normal.

The hepatitis profile was normal. Magnetic resonance imaging (MRI) of the pelvis revealed kidneys located in the pelvis mainly on the right side. Ultrasonography reveals ectopic kidneys with very small uterus. Following this, she was diagnosed with MRKH II syndrome with an ectopic kidney. She was given psychological counseling and vaginoplasty for the future if desirable.

DISCUSSION

Mayer-Rokitansky-Kuster-Hauser syndrome (Mullerian agenesis) is a spectrum of congenital anomalies with no known exact cause however mutation in the WNT4 gene is the cause of Mullerian aplasia and hyperandrogenic.[4]

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Date Received: 17/01/2023
Date Revised: 03/06/2024
Date Accepted: 04/06/2024

There is utero-vaginal agenesis in women with normal ovaries and secondary sexual characteristics.[1] The uterus may be rudimentary as bilateral and non-cannulated muscular buds with normal fallopian tubes and normal ovaries with normal endocrine functions. [5]

It has an autosomal dominant mode of inheritance. It is caused by the abnormal development of Mullerian ducts and organs like kidneys also develop from the same embryonic tissue due to which they are involved. My patient presented with Mullerian agenesis and ectopic kidney which means MRKH type 2 (MURCS) association. MRKH type 2 may involve the upper urinary tract, the skeleton, and the conductive system; and cardiac defects are rarely seen. [1] The diagnosis of MRKH mainly depends on imaging studies. Transabdominal ultrasonography is the first line of investigation but abdominopelvic MRI gives more precise and clear information than the prior. The differential diagnosis includes congenital vaginal agenesis, low transverse vaginal septum, androgen insensitivity, and imperforate hymen. [6] Patient counseling is the first step done before any treatment which was done in my case as well regarding stress management, future childbearing, and vaginal treatment modalities for the future. Treatment involves both non-surgical creation of neo-vagina as well as surgical creation of neo-vagina. In non-surgical methods, Franck's dilator method is done in which vaginal dilators (Hegar candles) are placed on the perineal dimple for at least 20 minutes a day to increase the length and diameter of the vagina. This process has a success rate varying from 78% to 92% and is a first-line therapy as it is a non-invasive method and often successful. Surgical methods include the Abbe-McIndoe operation, sigmoidal Colpoplasty, and the Vecchietti operation [6].

CONCLUSION

In summary, I presented a very rare case of our region of type 2 MRKH syndrome with renal anomaly and normal external genitalia but short vagina and cervix. This case presents that MRKH syndrome can occur with normal endocrine function and secondary sexual characteristics. Surgical correction by creating a neovagina is a good treatment method for young females for sexual intercourse. Further studies are required to understand the different aspects of MRKHS.Written informed consent was obtained from the patient's brother for publication of this case report with maintenance of confidentiality and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

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CONFLICT OF INTEREST: Authors declare no conflict of interest

GRANT SUPPORT AND FINANCIAL DISCLOSURE: NIL

AUTHOR'S CONTRIBUTION

Following authors have made substantial contributions to the manuscript as under

Ullah I: Concept, planning, study design, study conduction, critical review, analysis, manuscript writing.

Hussan J: Critical review, discussion, interpretation, manuscript writing, study conduction.

Authors agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.