DOI: https://doi.org/10.53350/pjmhs20221611200

CASE REPORT

A Rare Case of Mullerian Agenesis (Mayer-Rokitansky-Kuster-Hauser Syndrome)

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SUMMARY

A 21 years old girl presented in Gyne OPD of Lady Aitchison Hospital Lahore with complaint of primary amenorrhea. There was no complaint of cyclical dysmenorrhea, urinary retention or feeling of abdomino pelvic mass. Her mother had menarche at 14 years of age. Her height was 5 feet and one inch. Her secondary sexual characteristics were of normal (female) and well developed, external genitalia were of female with rudimentary vagina.Ultrasound showed normal size ovaries and hypoplasticuterus and rudimentary vagina.Hormones (FSH, LH, Prolactin) were normal. HerKaryotype was 46XX.On the basis of these findings, diagnosis of Mullerianagenesis (Maye-Rokitansky-Kuster Hauser Syndrome) was made. Family was counselled about the case. They were informed the patient is unable to menstruate and reproduce. However, for sexual function, it is possible to create a new vagina bynon-surgical method using Hegar dilator or surgical by vaginoplasty.

INTRODUCTION

Mayer-Rokitansky-Kuster-Hauser Syndrome (MRKHS) is a very rare congenital malformation of mullareanducts. Its incidence is 1 in 5000 females[1]. It has two types; type 1 is characterized by uterovaginal aplasia with normal secondary sexual characters and karyotype 46XX. In type 2 MRKHS there is absence of uterus and upper 2/3 of vagina along with anomalies of other systems like renal, skeletal, hearing and cardiac[2]. It is associated with both physical and psychological complications for the patient. It is a big challenge to cope with for the family and treating obstetrician and surgeon. The etiology of this syndrome is not known although studies show its association with inutero exposure of Diethyl stilboestrole[2].

The diagnosis of MRKHS is confirmed by ultrasound and MRI[3]. The treatment options are both non-surgical and surgical along with counselling of patient and family. Innon-surgical treatment, vaginal dilator is used regularly to create a new vagina suitable for sexual intercourse. The success rate of vaginal dilators is reported to be up to 95% in some studies[4]. Ifnon-surgical treatment fails, there is option of surgical treatment to create a new vagina by vaginoplasty. The options of vaginal tissue engineering and gene therapy are under research.[5]Thereare options of ovum retrieval from ovary and surrogacy or adoption to have a child.

CASE REPORT

A 21 years old unmarried girl with primary amenorrhea presented in outpatient department of Obstetrics and Gynecology Unit-4 Lady Aitchison Hospital in October 2022.She had no complaint of cyclical dysmenorrhea, urinary retention or feeling of abdomino pelvic mass.Her mother had menarche at 14 years of age. Her height was 5 feet and one inch. Her secondary sexual characteristics were normal (female). Her external genitalia were of female. Ultrasound and MRI showed normal size ovaries and hypo plasticuterus. There was no sign of uterine bleeding with blockage of genital tract leading to hematocolpose, hematometra and hematocolpose clinically or on ultrasound and MRI. Her karyotype was that of a normal female (XX). On the basis of these findings, diagnosis of Mullarean agenesis (Maye-Rokitansky-Kuster Hauser Syndrome) was made. Family was counselled that the girl is unable to menstruate and reproduce. However for sexual function, it is possible to create a new vagina by either using Hegar's dilators or by vaginoplasty (if required) before marriage.

Received on 11-06-2022
Accepted on 28-10-2022

DISCUSSION

In this case report, a case of 21 years old girl with MRKHS is presented. She presented with symptom of primary amenorrhea. Hersecondary characters including breast development, pubic axillary hair development and growth spurt were within normal age limit of puberty. In her family history, her mother had menarche at 14 years of age. Our initial differential diagnosis included genital tract anomalies like imperforate hymen, vaginal septum, MRKHS, and androgen insensitivity syndrome. Clinically there were no signs of obstruction like cyclical abdomino pelvic pain, feeling of any abdominal mass or urinary retention. This also ruled out presence of a functionally active endometrium capable of monthly bleeding.Her secondary characters were normally developed indicating normally functioning ovaries and adrenals.Her karyotype was that of normal female i.e. 46XX.

Our case was that of Type-1 MRKHS[7] as we did not found anomaly of any other system like renal, skeletal, hearing and cardiac found in type-2 MRKHS[8]. Our diagnosis was clinical and confirmed by ultrasound and MRI of pelvis. These two modalities are reported to be the gold standard for diagnosis of MRKHS[9]. On MRI, there was hypo plastic uterus (0.7x2.6x1, 4 cm) as shown in Figure 2. The pelvic ultrasound also showed a hypoplastic uterus and normal sized ovaries with growing follicles as shown in Fig. 3. Her Hormonal report (FSH, LH, Prolactin) was also within normal range.

So, based on clinical, ultrasound, MRI, hormonal and karyotyping reports, a diagnosis of Mayer-Rokitansky-Kuster-Hauser Syndrome was made. The family was informed about the diagnosis and counselled regarding psychological and physical challenges and management options which included non-surgical and surgical. The non-surgical option is regular use of vaginal dilator to create or enlarge rudimentary vagina[10,11]. Family opted for non-surgical option as first line treatment. They were counselled for follow up visits to monitor the success of treatment and option of creating a new vagina in case non-surgical treatment is not successful. The parents were informed that girl will not be able to menstruate and reproduce, but there are options of surrogacy and adoption in future. There is role of genetic analysis and gene replacement therapy for this poorly understood anomaly of female genital tract in future[12].

Figure 1: MRKH Syndrome[6]

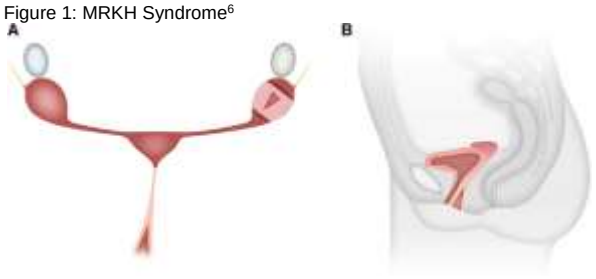
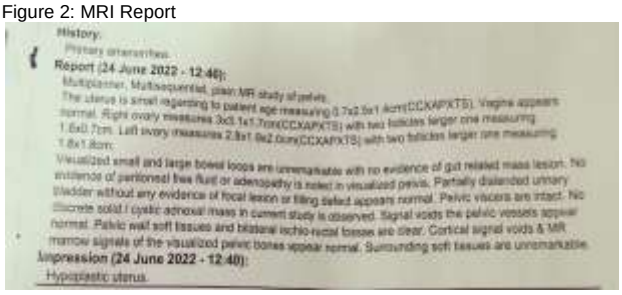


Figure 2: MRI Report

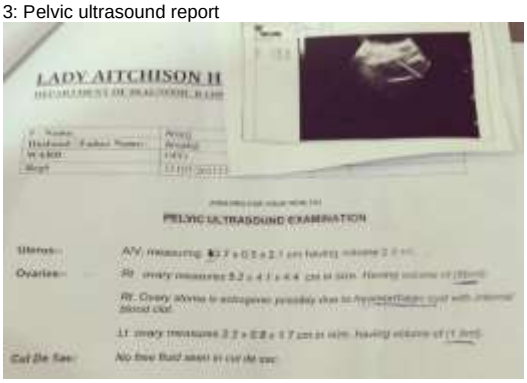

History:

Report (24 June 2022 - 12:46):

Impression (24 June 2022 - 12:46):

Hypoplastic uterus

Figure 3: Pelvic ultrasound report



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