CASE REPORT

Mayer-Rokitansky-Kuster-Hauser syndrome in a young female: diagnosis and treatment: a case report

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Abstract

Mayer-Rokitansky-Kuster-Hauser Syndrome is a rare condition in which Müllerian system does not develop and ends up with rudimentary upper vagina and the uterus. As compared to normal physiology of the ovaries and puberty, the patients present with primary amenorrhoea a key clinical symptom. However, the exact aetiology of the disease is still unknown. A few reports considered environmental and epigenetic changes, hormonal imbalance, and cellular receptor abnormalities as possible risk factors associated with the disease.

This case was reported at the Department of Family Medicine, The Indus Hospital, Karachi. A 24-year-old woman, married for eight months, presented with primary amenorrhoea and painful intercourse. Upon detailed clinical evaluation and relevant radiological and diagnostic investigation, an assessment, of Mayer-Rokitansky syndrome was made.

Keywords: Primary Amenorrhoea, Mullerian agenesis, Mayer-Rokitansky-Kuster- Hauser Syndrome.

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Introduction

A genetic condition, Mayer-Rokitansky-Kuster-Hauser Syndrome presents rarely in patients where they have normal development of secondary sexual characteristics. However, an aberrant paramesonephric duct formation results in primary amenorrhoea, hypoplasia of the cervix uterus and two-thirds of the vaginal portion. The patient seems normal phenotypically and genotypically, having equal number of chromosomes (46,XX).[1-2] Majority of the Mayer-Rokitansky-Kuster syndrome patients did not present with any hormonal defect. The chromosomal karyotyping was also normal. Some researchers consider the formation of abnormal paramesonephric duct as a developmental genetic disorder.

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Mayer-Rokitansky-Kuster is classified further into two types: i) Type-A Mayer-Rokitansky-Kuster syndrome and, ii) Type-B Mayer-Rokitansky-Kuster syndrome.[3-5] Type-A Mayer-Rokitansky-Kuster syndrome cases represent isolated uterine aplasia whereas Type-B Mayer-Rokitansky-Kuster syndrome cases represent both gynaecological and non-gynaecological complexities. The major gynaecological complexities include development of aberrant fallopian tubes and renal dysfunction. About 40% cases of the Type-B Mayer-Rokitansky-Kuster syndrome represent non-gynaecological complexities.[3, 6]

The patient faces multiple challenges starting from the clinical investigations of the affected organs, penetrative sexual intercourse, and inability to conceive. The psychological impact of the Mayer-Rokitansky-Kuster syndrome include patient's fear of being identified, sexuality, and infertility. Mayer-Rokitansky-Kuster syndrome is diagnosed commonly during adolescence, which is considered a sensitive phase of physical and emotional development. However, considering cultural aspects, religious norms and ethical values, it is essential to treat Mayer-Rokitansky-Kuster syndrome patients.

The current case report aims to critically assess the physiological, clinical and diagnostic characteristics of Mayer-Rokitansky-Kuster syndrome identified at family medicine department, The Indus Hospital Karachi, Sindh, Pakistan.

Case Report

A 24-year-old woman, who was married for eight months, first visited the outpatient clinic at the Department of Family Medicine in June 2021. There was normal body hair distribution and breast development. Similarly, upon clinical investigations clitoris, labia majora and labia minora were noted to be normal. However, the vagina was 2cm short ending with a closed pouch. She presented with primary amenorrhoea, and painful and unsuccessful intercourse. This was very disturbing for the patient. Upon examination, the woman was 154cm tall and weighed 40kg. The physical signs of puberty including developed breasts, presence of axillary and pubic hair were present. There was a blind ending of the vagina, however, no severe skeletal muscle-related anomalies were observed.

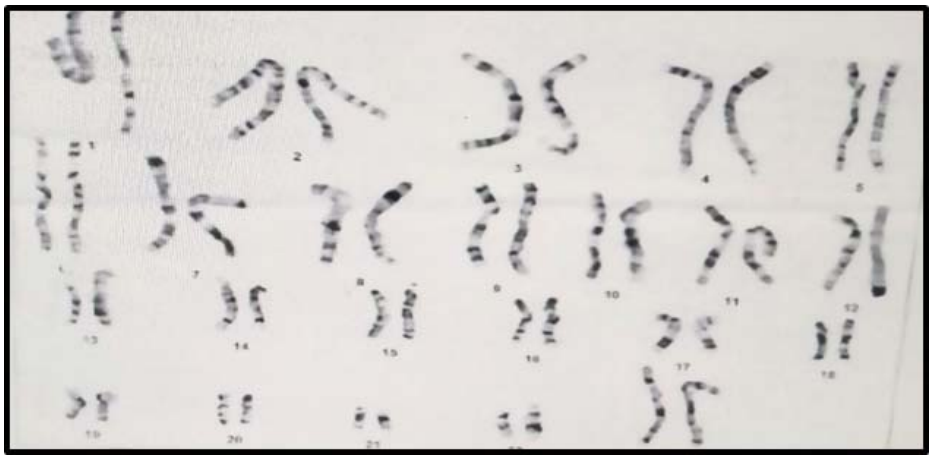

Figure: Karyotyping of the patient.

Upon detailed radiological examination by using Magnetic Resonance Imaging, 2/3rd of the upper vaginal canal could not be seen, and only 1/3rd of the lower vaginal canal was observed. The fallopian tubes were normal. The findings in this patient were consistent with Type-A Mayer-Rokitansky-Kuster syndrome. Further, cytogenetic analysis was also done to achieve karyotyping. Normal karyotyping was observed (46, XX) as shown in Figure 1.1. Further, diagnostic examination was extended for laparoscopy and she was managed by performing vaginoplasty under anaesthesia.

A detailed counselling regarding the outcomes of the condition were discussed and the need to maintain a good quality of life and stability in relationship was emphasised.

The vulva was normal, vagina was short (3cm), with normal ovaries and fallopian tubes, whereas the pelvic sidewalls were displaced. About 6cm vaginoplasty was applied and further assessed by laparoscopy and digital examination. The post-operative vaginal length was observed to be 4cm.

This case report was approved by the Head of Department of Family Medicine, The Indus Hospital, Karachi. The patient's informed consent was also taken prior to writing this paper as per standard protocols of protecting human research participants.

Discussion

The paramesonephric duct further matures into fallopian tubes, uterus, cervix, and upper two third of the vagina as well. However, the lower 1/3 portion is derived from the urogenital sinus. Recent research reports acknowledge paramesonephric duct inhibiting factor also known as anti-Mullerian inhibiting hormone.[7-9] The imbalance in this hormone is potentially associated with abnormal maturation of paramesonephric duct, which ultimately results in abnormal fallopian tubes, uterus cervix, and vaginal duplication related complexities.[10]

In addition to the physical complexities, there are many psychosocial challenges associated with these problems.[11-12] The current report determines that a woman presenting with normal secondary sexual characteristics with primary amenorrhoea and unsuccessful intercourse-related symptoms may be a possible case of Mayer-Rokitansky-Kuster syndrome. The age for developing secondary sexual characteristics starts from 12 years as consistent with this report.[13] The patient was evaluated upon physiological and psychosocial aspects and the diagnosis was established by MRI and laparoscopy and, hence, could be managed appropriately. This is a superior technique over the biochemical testing to confirm the diagnosis of Mayer-Rokitansky-Kuster syndrome.

Conclusion

The complications associated with Mayer-Rokitansky-Kuster syndrome are not limited to vaginal agenesis. It ends up with infertility and psychological distress as well. A comprehensive management approach is required to treat Mayer-Rokitansky-Kuster syndrome by engaging psychologists, family medicine experts and family support group.

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Conflict of Interest: None to declare.

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