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Case Report

A case of Mayer-Rokitansky-Kuster- Hauser syndrome type I

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ABSTRACT

Rokitansky-Kuster-Hauser (MRKH) syndrome is a rare congenital disorder characterised by absence of uterus and vagina. Ovaries and fallopian tubes are normal and functional. Its incidence is 1 in 4500-5000 female. Patients present with primary amenorrhea with normal external genitalia and development of secondary sexual characteristics, and 46XX karyotype. It is usually diagnosed by MRI, CT or transabdominal USG, with MRI being the gold standard.

1. Introduction

Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is a rare congenital disorder characterised by absence of uterus and vagina. Ovaries and fallopian tubes are normal and functional.[1] Its incidence is 1 in 4500-5000 female.[2] Patients present with primary amenorrhea with normal external genitalia and development of secondary sexual characteristics, and 46XX karyotype. It can present as an isolated uterovaginal aplasia or may present with renal agenesis, urinary anomalies, skeletal abnormalities and very rarely, pulmonary hypoplasia.[1] It is usually diagnosed by MRI, CT or transabdominal USG, with MRI being the gold standard.[3]

2. Case Report

A 24-year old nulligravida married female presented with chief complain of dyspareunia. She had a history of primary amenorrhea and had been married for 5 months. Her height was 163 cm and she weighed 45kg. On examination, she had normal development of secondary sexual characteristics along with hirsuitism. She had hair over her upper lip, on her chin, sternal area and over lower abdomen. Her hirsuitism score was calculated to 7. Auscultation showed normal heart sound with S1 and S2 present and breath sounds. On per vaginal examination, a vaginal opening with a short non-patent vaginal canal of about 1cm was appreciated. Hormonal profile showed elevated DHEA and normal levels of other hormones. USG abdomen revealed an absence of uterus, with presence of bilateral normal ovaries and kidneys. Chest X-ray showed normal lungs with cardiac shadow. ECG reports were unremarkable with normal left ventricular size and function.

The patient opted to undergo Davydov's vaginoplasty. Consent was taken and the surgery was performed in the 3rd week after initial visit. The patient was put under general anaesthesia and approached laproscopically through the abdomen. Dissection was carried out posterior to vagina to create rectovaginal space. Similarly dissection was carried out anteriorly and the vesicovaginal fold was opened. The

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bladder was pushed down. Peritoneal fold was dissected lateral to infundibulopelvic ligament. Incision was made on the most prominent part of vaginal mucosa. A sponge on cotton swab was placed to mark the bridging tissue between vagina and peritoneum. The peritoneal folds created were sutured to the vagina. A vaginal glass mold was inserted and was kept inserted post-operatively.

The patient was discharged in 5 days after she was taught how to self insert the mould along with necessary precautions to take. Patient was to follow up after 1, 6, and 12 months. On one month follow up, the patient had no complains and has been regularly inserting the mould as instructed.

3. Discussion

Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is a rare congenital disorder characterised by absence of uterus and vagina. Ovaries and fallopian tubes are normal and functional.[1] Its incidence is 1 in 4500-5000 female.[2] MRKH is caused due to the agenesis or aplasia of paramesonephric duct during the 5-6th gestational week which leads to absence of uterus and vagina.[3]

MRKH is broadly classified into 2 groups - type I and type II. In MRKH type I, patients present with isolated uterovaginal aplasia, as is with the patient discussed above. MRKH type II presents with Mullerian duct aplasia, renal agenesis and cervicothoracic somite dysplasia also known as MURCS.[1] MRKH type II is a syndrome with incomplete uterovaginal aplasia along with other malformations including renal and urinary anomalies, skeletal abnormalities, hearing defects and cardiac anomalies.[4] Rarely, pulmonary hypoplasia has been observed in about 10% of the patients.[1]

The exact cause of MRKH is unknown. Majority of familial studies suggest an autosomal dominant inheritance pattern limited to female sex. The defect is generally inherited by the father which many present as Wolffian duct hypoplasia or agenesis, renal anomalies, hearing impairment and skeletal deformities.[5] There have been some studies showing genetic association to copy number variations (CNV) located in the in 17q12 and 16p11.2 chromosomal regions.[3] Deletions in 16p11.2 have also been associated with congenital uterine anomalies, autism and neurological disorders in which the TBX6 gene maybe responsible. LHX1 found in 17q12 has found to be associated with MRKH type II and unilateral renal agenesis.[5] Whereas the HNF1B gene in the same locus has been associated with renal and uterine abnormalities.[3,6] Deficiency of WNT4 gene in 1p31-1p35 has also been associated with MRKH presenting with hypoandrogenism.[7]

Patients present with primary amenorrhea and dyspareunia. The external genitalia and development of secondary sexual characteristics is normal. The patient has 46XX karyotype. It may present as an isolated uterovaginal aplasia or may present with renal agenesis, urinary anomalies, skeletal abnormalities and very rarely, pulmonary hypoplasia.[1] It is generally diagnosed around the ages of 16-19.[3] Some patients may also present with hirsuitism due to hyperandrogenism. This has been associated with WNT4 gene deficiency.[7]

Diagnosis is by MRI, CT or transabdominal USG, with MRI being the gold standard.[3] Differential diagnosis includes imperforate hymen, transverse vagina, septum, Turner's syndrome, complete androgen insensitivity syndrome and congenital adrenal hyperplasia.[2,3]

Diagnosis of MRKH has a profound imapct on the pyschology and psychosexual aspect of the patient such as identity issues, sexuality, low self esteem, depression and anxiety. Hence, counselling and support is required prior to starting any treatment or interventions.[2,3]

The simplest choice of treatment giving good sexual result should be considered for the patient.[8] Non-surgical procedure, that is the Frank and Ingram method is preferred over surgical methods. Here, dilators are inserted into vaginal cavity to stretch the vaginal walls. The size of the dilators is gradually increased to invaginate the mucosa. Surgical methods include McIndoe procedure, William's vaginoplasty, Sigmoid vaginoplasty, Vechietti vaginoplasty and Davydov's vaginoplasty. Out of these, the latter 2 are laproscopic procedures.[2]

The patient discussed above opted for surgical correction by Davydov's Vaginoplasty wherein the space between the urethra, bladder and rectum is dissected followed by peritoneal mobilisation. The patient's own peritoneum is used as a lining for neovagina along with insertion of a glass mould for a month.[2] This technique is beneficial as it is less painful, has less bleeding, shorter stay and quick recovery.[2] The complications for this surgical method are injury to the bladder, rectum, and/ or ureter, peritonitis, vesicovaginal fistula formation, and lower urinary tract symptoms.[9]

4. Conclusion

MRKH is a rare complex syndrome affecting females physically, psychologically and sexually. Hence, the treatment requires a multidisciplinary approach. Patient should be counselled and provided appropriate treatment options. Davydov's vaginoplasty is one of the effective surgical treatment option. Adoption and surrogacy can be considered for infertility. Uterine transplant may also be considered.

5. Source of Funding

None.

6. Conflict of Interest

None.

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