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Mayer-Rokitansky-Kuster-Hauser Syndrome: Clinical & Surgical Management

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Abstract

Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is a rare pathological condition characterized by congenital aplasia of the uterus and upper part (2/3rd) vagina in women with normal secondary sexual characteristics, normal external genitalia, functional ovaries & karyotype (46, XX).(1)Herein, we report the case of 24yr old housewife who presented to us with primary amenorrhea and inability to achieve penetrative sexual intercourse. She had vaginal atresia and absent uterus. Vaginal reconstruction was done by McIndoe operation of vaginoplasty using split-thickness skin graft. We saw an immediate & satisfactory outcome in our patient who is sexually active now.

Keywords : Mayer- Rokitansky – Kuster - Hauser Syndrome, Case report, surgical management

Introduction

Mayer-Rokitansky-Kuster-Hauser Syndrome is a rare anomaly characterized by congenital aplasia of the uterus and vagina in women with normal secondary sexual characteristics development & karyotype (46, XX) (1). Incidence of MRKH is estimated as 1 in 4000-5000 female births & is a common cause of primary amenorrhea (2). The isolated uterovaginal aplasia is known as type 1 MRKH which is less common than type 2 MRKH known as MURCS (Mullerian, Renal, Cervical Somite) wherein females along with uterovaginal agenesis have associated renal (unilateral agenesis, ectopia of kidneys/horseshoe kidney), skeletal & vertebral (Klippel-Feil anomaly; fused vertebrae, scoliosis) or hearing defects.(3,4)Our case fits in type 1 MRKH Syndrome. The diagnosis is made during adolescence when a girl with well developed secondary sexual characteristics presents with primary amenorrhoea with the vagina reduced more or less to a dimple.(5,6) Diagnostic Imaging includes USG & MRI, Laparoscopy being a gold standard.(1) Treatment should be offered when patients are emotionally mature & are

ready to commence sexual activity. Treatment can be either surgical or non-surgical & treatment should always be individualized as per the patient's needs & interest. As our patient was already married & psychologically mature enough to understand the nature of anomaly & wanted a satisfactory sexual intercourse with her husband, we offered her treatment in the form of creating a functional neovagina with an adequate diameter & length to accommodate sexual intercourse & she was also counselled regarding her fertility issues.

Case Report

A24yr old housewife along with her husband presented to the OPD with complaints of primary amenorrhea; Pain and resistance during attempted intercourse with inability to achieve penetrative sexual intercourse. There was no history of cyclical lower abdominal pain or urinary symptoms. There was no family history of similar illness & no significant past medical history.

The larche was attained at 12yr and adrenarche at 13yr of age. Her secondary sexual characteristics were well developed with no delay. Physical examination revealed a young female patient with normal secondary sexual characteristics with well-developed breasts (Tanner stage 4), axillary hair & female pubic hair distribution being normal (Tanner stage-5). On Local examination external genitalia (labia majora &minora), external urethral meatus& anus were found to be normal.Per vaginal & bimanual examination revealed a blind vaginal pouch admitting 1/3rd of little finger (about 3cm)& absence of uterus. Routine investigations were done and were found to be within normal limits. An abdominal ultrasound scan showed normal ovaries and absent uterus. The kidneys & other intra-abdominal organs were normal.

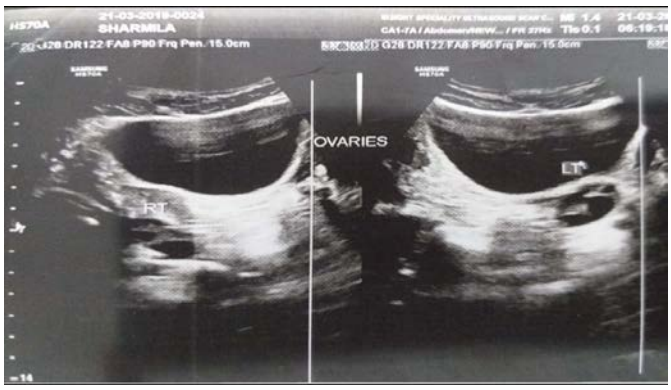

Figure 1: Ultrasonography scan of patient showing absence of uterus and normal looking ovaries

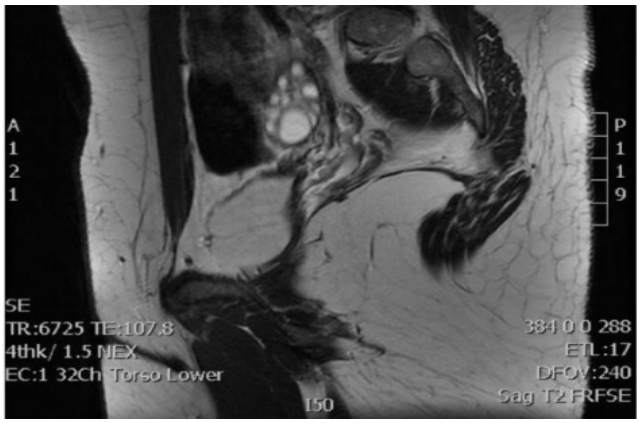

Figure 2

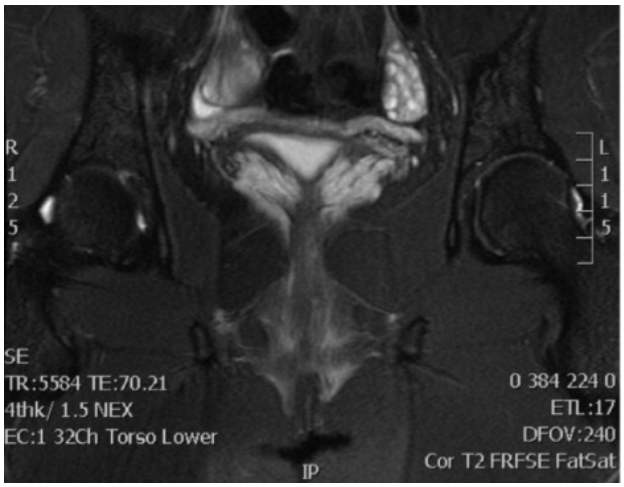

Figure 3

Figure 2 & Figure 3: Magnetic Resonance Image showing absence of uterus.

MRI also showed absent uterus & everything else within normal limits. Based on these clinical & radiological findings diagnosis of MRKH Type-1

Syndrome was made. Our patient was counselled on the nature of anomaly and was discussed about the treatment options available counseling regarding the lack of innate reproductive potential &prospect of child bearing was also done.

Patient opted for surgical method of treatment. She was offered treatment in the form of creating a neovagina. She was taken up for McIndoe technique of vaginoplasty under spinal anaesthesia.

The procedure consisted of 2 important steps-first being creating a new space between the rectum and the vagina. The patient was placed in dorsal lithotomy position and catheterized to define the anterior urethra bladder boundary. A transverse incision was made at the apex of vaginal dimple. Dissection was initiated in the fibroconnective tissue on either side of the midline creating a space between the bladder and rectum. Dissection was continued upto 4cm depth and 1 finger width & once the space created seemed to be adequate, the intervening median raphe was excised. Injury to peritoneum was avoided and hemostasis was ensured.

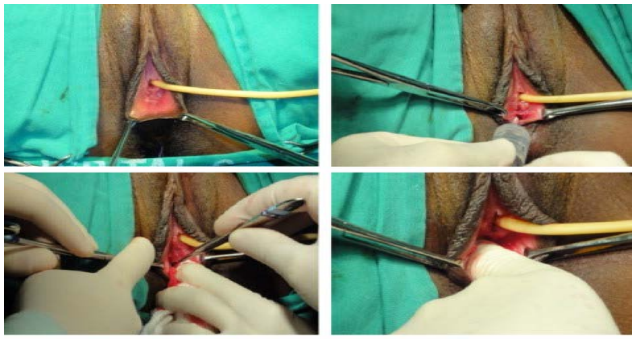

Figure 4: (a,b,c,d)in clockwise direction-
(a)-Pre-operative picture showing blind ended vagina.
(b,c,d)- Intra-operative picture showing the dissection to create neo-vagina

The second part being prosthesis assembly and placing the graft prosthesis assembly in the neovagina.

The prosthesis was made with 10 x 10 cm sponge which was sterilised rolled over another sponge until a width of 3cm was obtained. 2 condoms were placed over this mould and tied at the lower end. Split thickness skin graft of -0.018 inch thick, 8-9 cm wide, 16-20 cm in length was obtained from the back of upper thigh using a derma tone. The graft was washed in saline and was secured around the mould using 3-0 catgut.

Figure 5: Prosthesis made with sponge measuring 10cm in length & 3cm width.

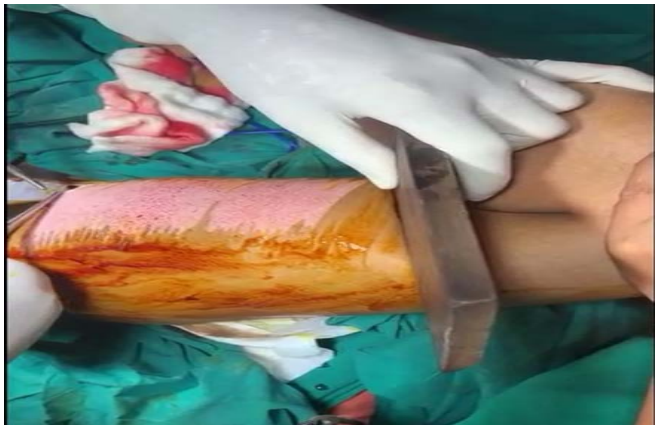

Figure 6: Split thickness skin graft of -0.018 inch thick, 8-9 cm wide, 16-20 cm in length was obtained from the back of upper thigh using a derma tone.

Figure 7

Figure 8

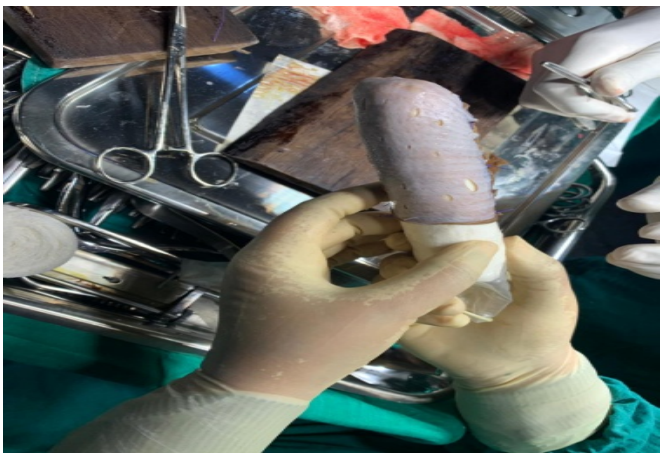

Figure 9

Figure 7, 8 &9 : Graft being secured around the mould using 3-0 catgut.

After ensuring hemostasis, the graft covered mould was placed in neovagina. The labia minora was now sutured to keep the Mould in position.

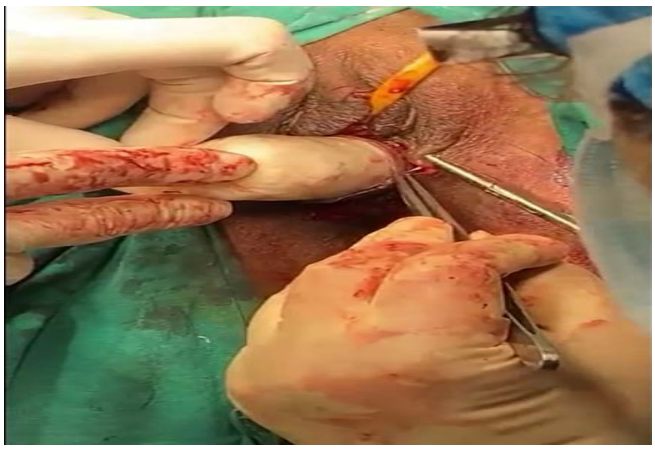

Figure 10

Figure 11

Figure 10 & 11 - The labia minora sutured toGraft prosthesis assembly(mould) in the neovagina to secure its position.

Post-operatively, the patient was kept on intravenous and oral fluids, antibiotics & a low residue diet for 72 hours. After 1 week, the labial sutures were removed and the mould was gently withdrawn with minimal traction. The neovagina was irrigated with povidone-iodine and saline. On examination, it was found that graft had taken up well. Further, similar mould was prepared with sponge of same diameter and width, covered with condom & was changed at 48 hour interval thrice. During 2nd week, patient was educated & trained regarding how to use an acrylic mould by herself. She was taught how to irrigate vagina and clean

the acrylic mould. She was instructed to apply it, continuously for 6 weeks except at the time of urination & defecation&further next 12 months to be applied only during night. The couple was advised that coitus is possible once soreness is passed & healing is complete. She was instructed to follow up every week till 1 month& thereafter monthly follow up.Currently patient is having her monthly follow-up at our OPD & is also undergoing counselling addressing her fertility issues.

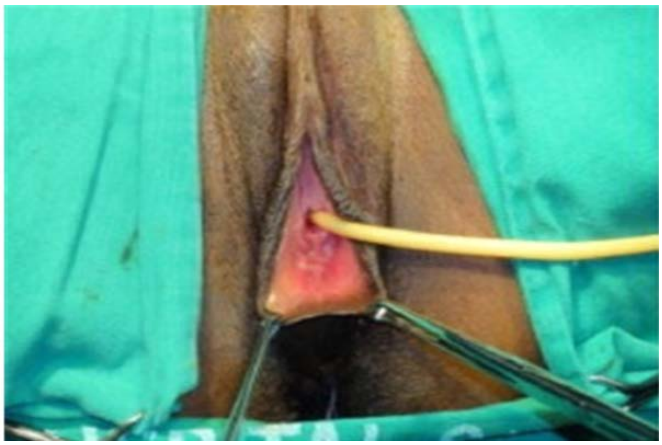

Figure 12

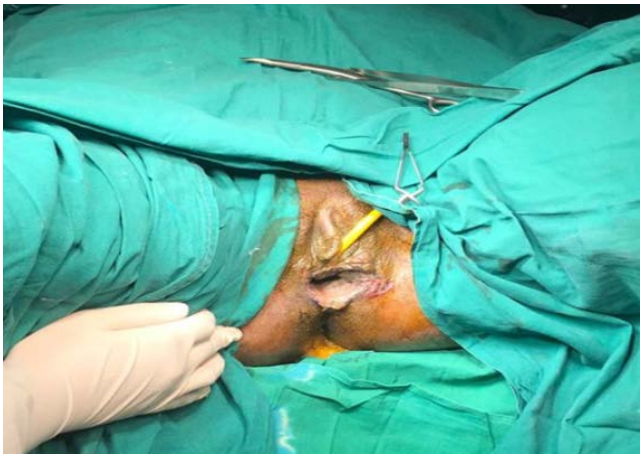

Figure 13

Figure 12 & 13: Comparing Pre-op & post-op image

Figure 13- 10th Post-operative day.

Discussion

MRKH was described by physiologist Mayer (1829), Rokitansky(1938), Kuster (1910) and gynecologist Hauser (1961);It is 2nd most common cause of primary amenorrhea with incidence being 1 per 4000-10000 females. It is a Sporadic anomaly /Autosomal Dominant Inheritance with variable expressivity. It occurs due to interrupted embryonic development of the paired mullerian ducts between the 4th and 12th week of gestation. Patients with this syndrome have normal 46XX karyotype, normal ovarian function, normal external genitalia, partial or incomplete absence of vagina & an absent or hypoplastic uterus with non-canalized tubes.According to American Fertility Society classification there are 3 forms of MRKH Syndrome:-

TYPICAL Form (47%)-Fallopian tubes, ovaries and renal system are normal .

ATYPICAL Form (21%)-Malformation of the ovary or renal systems.

MURCS (Mullerian duct aplasia, renal aplasia, cervico-thoracic somite dysplasia -32%)Associated with skeletal / heart malformations, muscular weakness &renal malformations.

USG showing absence of uterus & fallopian tube with normal ovaries, aids in diagnosis of MRKH. MRI**,** CT**,** IV Pare more helpful in detecting other co-existing abnormalities. Laparoscopy is the Gold standard Investigation. Management of vaginal agenesis in the MRKH syndrome remains controversial. The choice of procedure & patient age at reconstruction depends upon individual anatomy, fertility potential, psychosocial & social factors.[(2)]Previously – Timing of surgery was considered to be just before marriage, but it lead to difficulties with marriage until vagina is healed completely.Hence recently it is done on patients from 17to20 years of age who are emotionally mature enough to understand the anomaly and intellectually

reliable enough to manage post op requirements for care.

The goals of long term treatments are to create a functional neo-vaginal canal with an adequate diameter & length, appropriate axial direction to accommodate sexual intercourse and to address the issue of fertility.[7,8]There are 2 main types of procedures; the first one consists of the creation of a new cavity which can be done by non-surgical or surgical method. The most commonly used non-surgical procedure is the Frank's dilatation method, which involves Series of graduated dilators (hegar candles)applied first by clinician & then by patient herself to dilate vaginal space. Dilators are placed on the perineal dimple for at least 20min a day. Creates a functional vagina within 3-6months. The other non-surgical method is Ingram's technique. Vaginal dilators have few complications as there are no anesthetic or surgical risks, but these are time consuming, tedious, causes discomfort & requires motivation[7].

The second part is vaginal replacement with a pre-exisiting canal lined with a mucous membrane(a segment of bowel) or like in our case using split-thickness skin-graft for vaginoplasty. Sometime amnion is also used as a graft. Our patient being already married was an ideal candidate for this surgery. The Abbe Mcindoe procedure was adopted by us to create the neovagina. Three important principles of McIndoe include- Dissection of adequate space between bladder & rectum followed by Inlaying of a split thickness skin graft& then prolonged dilatation during the contractile phase of healing. Various alternatives like amnion[9], peritoneum, minora labia grafting or synthetic materials [10]have been used. Other surgery techniques includes Williams vaginoplasty, which involves suturing the labia majora into a perineal pouch, but the vagina created is external, short & unsatisfactory for penetrative intercourse; it is no longer practiced. Laparoscopic Vecchietti procedure aims to create a neovagina by using an acrylic olive mould that is placed against the vaginal dimple. The olive is attached to a traction device mounted on the abdomen with laparoscopically placed sub peritoneal sutures. Then, traction is applied to the olive to produce 1.0-1.5 cm of invagination per day, creating a neovagina in approximately 7-10 days.

No consensus has been reached regarding the ideal method for creating a functional vagina. Our patient treated surgically with McIndoe Vaginoplasty had uneventful postoperative recovery and was also satisfied with immediate outcome. At present, the most common operation is McIndoe vaginoplasty. Though non invasive laparoscopic surgery seems promising, additional data is required for it to gain general acceptance.[11]

In conclusion,MRKH is a rare anomaly but with devastating effects on women's reproductive health and sexual life.Non-operative approach is recommended as a first-line therapy; No consensus regarding best surgery option, approach being most often based on surgeon's experience. Long term follow up will be needed to assess the long-term outcome, with regard to complications, sexual satisfaction of patients & spouses, their adaptation & resolution of fertility issues.

List of Abbreviations –

OPD- Out Patient Department

CT- Computed Tomography

MRI- Magnetic Resonance Imaging

USG- Ultrasonography.

IVP- Intravenous Pyelogram.

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