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MRKH Syndrome and A Better Tomorrow

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ABSTRACT

Vaginal agenesis may present as a single defect in development or associated with other anomalies. It is usually associated with Mayer-Rokitansky-Kuster-Hauser (MRKH) and Androgen Insensitivity Syndromes (AIS). The incidence is about 1 in 4,000-5,000 live female births. The principal aim of treating the congenital absence of vagina is to solve problems of sexual life and reconstruction of vagina.

AIM AND OBJECTIVES

AIM

Treating the congenital absence of vagina to solve problems of sexual life and reconstruction of vagina.

OBJECTIVES

To assess the operative time, blood loss, post-operative pain of surgery.

To reconstruct vagina anatomically to establish the patency and calibre of vagina.

METHOD

By surgical intervention

DISCUSSION

A case of MRKH unmarried patient aged 28 years who presented with primary amenorrhea at our institution.

-Workup for primary amenorrhea was done which was suggestive of MRKH syndrome.

-Surgical intervention was planned accordingly

-An artificial mould coated with a fresh amnion graft was created using a 10cc syringe, coated with foam sponge and covered with a condom.

-Fresh amnion graft was harvested from a patient and triple washed with normal saline and antibiotics.

CONCLUSION

An adequate length vagina was created and normal sexual activity initiated.

Keywords: Vaginal agenes; MRKH Syndrome; Uterus

INTRODUCTION

Mullerian agenesis or Mayer-Rokitansky-Kuster-Hauser syndrome has an incidence of 1 in every 4,000 – 10,000 females. It results from embryologic failure of development of mullerian duct, which leads to agenesis or hypoplasia of uterus and vagina. Patients normally present with primary amenorrhea but have normal secondary sexual characteristics and are genetically female. They have an absent or short blind ending vagina. The ovaries are normal in structure and function because they have a different embryological source. There is an association with other congenital anomalies.

MRKH syndrome is classified as type I (isolated uterovaginal aplasia) or type II (associated with extragenital manifestations). Extragenital anomalies typically include renal, skeletal, ear or cardiac malformations. The etiology of MRKH syndrome still remains elusive. However the use of various genomic techniques has allowed the identification of recurrent genetic abnormalities in some patients.

The psychosexual impact of having MRKH syndrome should not be underestimated. The clinical care foremost involves thorough counseling and support apart from creating a neovagina for satisfactory intercourse. Options include non surgical and surgical methods. The external genitalia appear normal and the patients typically have a normal reproductive endocrine function and reach puberty showing normal signs of thelarche and pubarche. Patients typically present with primary amenorrhea during adolescence. During the last decade several advances have been made in MRKH syndrome, uterus transplantation as the first available fertility treatment. Furthermore, it is important to be aware of potential cultural aspects and their influence on reactions to the diagnosis in patients and their families and peers.

CASE PRESENTATION

A 18 year old Unmarried Female presented to the OPD with history of primary amenorrhea. No H/O cyclical abdominal pain, abdominal distention, trauma to head, headache, vomiting, seizures, visual disturbances, abnormal weight loss, excessive physical activity, heat or cold intolerance, fatigue , lethargy, discharge from nipple, excessive hair growth, voice change, weight gain, hot flushes, vaginal dryness, decreased libido, young girl moderately built and well nourished. On examination her secondary sexual characteristics were normal. Breast and pubic hair – Tanner 5. Systemic examination was normal. On Local examination, vagina was not developed. Seen as a blind pouch of approximately 1.5 cm. On per rectal examination no mass was palpable.

Hormonal evaluation was done. Serum TSH , FSH and prolactin were within normal limits. On Ultrasound two uterine horns were appreciated and seen communicating with lower uterine body. Cervix and Vagina cannot be seen. Ovaries were normal. MRI showed rudimentary uterine horns without endometrium or thickened fallopian tubes .Cervix and vagina not visualized. Karyotyping was done and was found to be 46 XX.

On the basis of history and clinical examination a provisional diagnosis of MRKH Type-1 was made which was confirmed by the investigations. Surgery was chosen as the line of treatment. Modified McIndoe's

Vaginoplasty was done. Amnion graft was harvested from a placenta of a Caesarean section patient and triple washed with antibiotics and normal saline. Transverse incision was made in the vaginal dimple to create a pocket between urinary bladder and rectum. After serial vaginal dilatation, amniotic membrane graft was taken and placed on the upper and lower vaginal wall. A soft vaginal mould made up of a syringe coated with the amnion graft which was sewn over the external mucosa using vicryl no 2-0. On the 7th postoperative day, the mould was removed. Time of surgery was approximately 45 minutes and blood loss was less than 30 ml. A new mould was prepared, using a syringe coated with foam covered with condom and was advised to place it in the vagina for 24 hours for six weeks and only at night in the next six weeks, except at the time of defecation and micturition. Vaginal length was 10 cm. Pain score - At time of surgery was 3, during post operative dilation was 6, at 3 month follow up was 2. After 7 days, the graft was well taken up and there was no pressure necrosis or ulceration.(Figure 1-8)

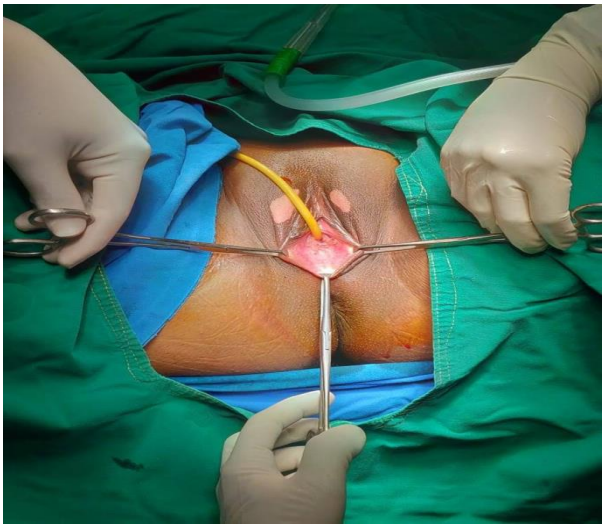

Figure 1: Vaginal dimple.

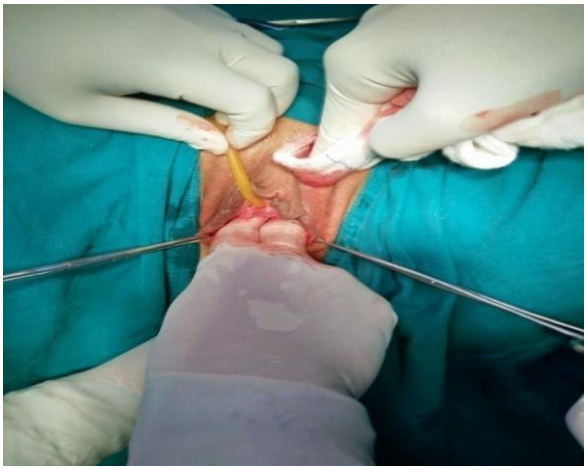

Figure 2: Creating a pocket between urinary bladder and rectum.

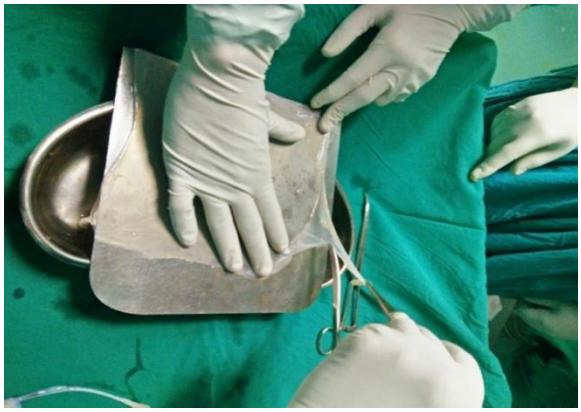

Figure 3: Modified Mc Indoe's Vaginoplasty using Amnion Graft.

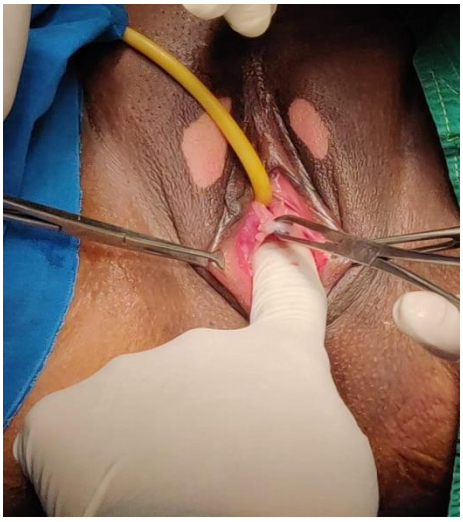

Figure 4: Serial dilatation.

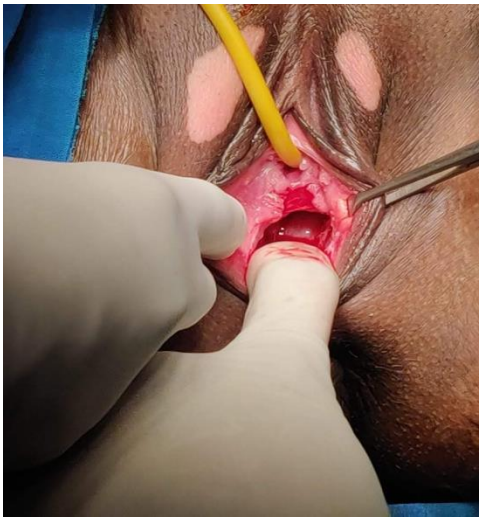

Figure 5: Serial dilatation with fingers.

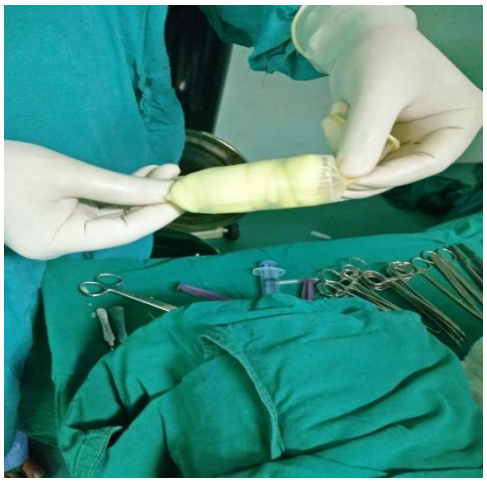

Figure 6: A soft vaginal mould was created using a syringe.

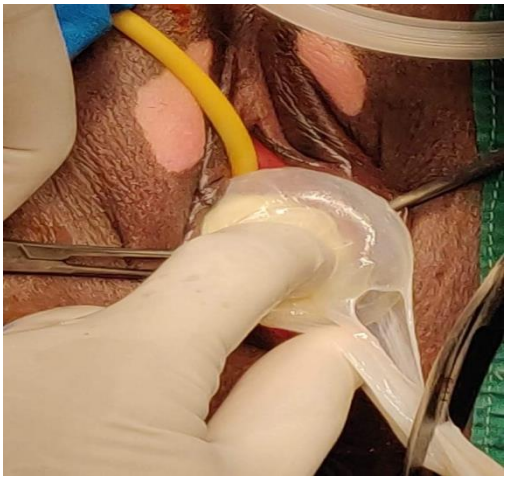

Figure 7: A soft vaginal mould was sewn over external mucosa.

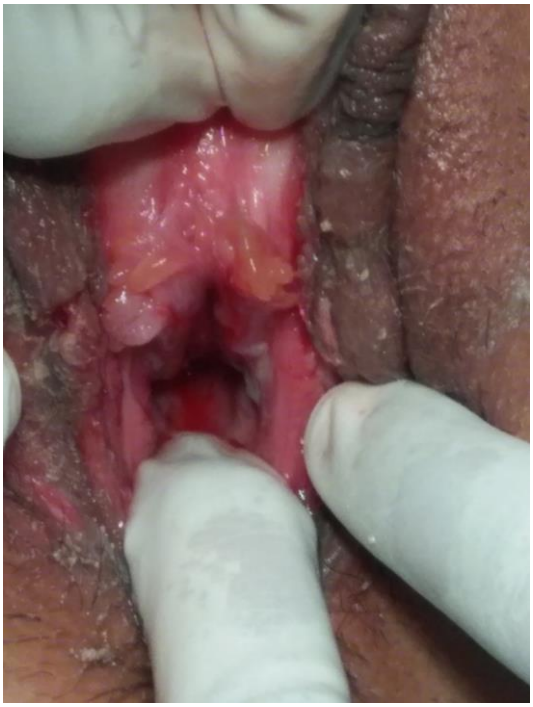

Figure 8: Success story of amnion taken up as mucosa.

DISCUSSION

MRKH syndrome, also referred to as Mullerian aplasia, is a congenital disorder characterized by agenesis of the uterus and upper part of the vagina in females with normal a normal female karyotype. The fallopian tubes, uterus, cervix and upper two-thirds of the vagina originates from the paramesonephric (Mullerian) ducts. The lower part of the vagina origins from the urogenital sinus. The caudal part of the two mullerian ducts fuses to form the uterus, cervix and upper vagina. Whereas the upper parts of the paramesonephric ducts form the two oviducts. MRKH syndrome is caused by either complete agenesis or aplasia of the paramesonephric ducts to form the uterus and upper vagina.

The etiology of MRKH syndrome still remains unclear. Patients with MRKH syndrome typically present during adolescence with absent menstrual periods following normal puberty. Physical examination is carried out which may include examination of external and internal genitalia. Transperineal or transabdominal ultrasound is performed revealing absence of uterus and presence of ovaries. MRI is considered gold standard method of diagnosis. Chromosomal analysis is often performed to confirm normal female karyotype. Other relevant tests include FSH, LH, androgens and estradiol which are generally normal.

Continuous research in the genetics of MRKH syndrome is imperative to provide better knowledge of the pathogenesis and improve the patient care and counselling. Correction of vaginal agenesis in MRKH syndrome with creation of a functional neovagina has been a hallmark in the treatment. The increasing availability of uterus transplantation as fertility treatment or In Vitro Fertilisation (IVF) using a gestational carrier will allow more patients to achieve biological motherhood in the future.

CONCLUSION

Nonsurgical methods are still the first line of choice, however patient compliance is the biggest problem. Modifications of McIndoe's procedure using Split thickness skin graft and amnion graft are still excellent techniques to be used in patients not suited for a major surgery. Amnion graft being the easiest to obtain is still preferred in the Indian scenario. The care of patients with MRKH syndrome is complex and requires a patient – centered multidisciplinary approach. Management includes addressing all-together gynaecological, sexual, psychological and infertility issues. Continuous research efforts are pivotal in order to expand the current knowledge and improve future care.

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