Mayer-Rokitansky-Küster-Hauser Syndrome with Imperforate Anus: A Rare Association and an Innovative Surgical Management

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Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is one of the rare disorder of Mullerian agenesis leading to non-development of uterus and vagina. Its association with anorectal malformation is rare. We report a case of MRKH syndrome with recto-vestibular in a female child. The child had undergone a sigmoid loop colostomy in the neonatal period. On clinical examination of the perineum, a fistula was present in the vestibule just below the urethral opening, but no vaginal canal was found. It was managed with an innovative surgical technique, preserving the fistula and lower rectum to function as vagina.

Keywords: Mayer-Rokitansky-Küster-Hauser syndrome, Mullerian dysgenesis, Vaginal agenesis, Vestibular fistula

INTRODUCTION

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a rare disorder that affects women, 1 in 5000 live births.¹ It is characterized by the dysgenesis of Mullerian ducts leading to failure of the uterus and the vagina to develop properly in women who have normal ovarian function and normal external genitalia. Women with this disorder develop normal secondary sexual characteristics during puberty (e.g., breast development and pubic hair), but do not have a menstrual cycle (primary amenorrhea). MRHK syndrome can be divided into two types, Type 1 which is an isolated anomaly while Type 2 is associated with other organ system anomalies, mainly renal, skeletal and cardiac. The clinical appearance is of two openings in the vestibule, which are urethral and the rectovestibular fistular openings and this can be confused with imperforate anus without a fistula.² We present a case of 1-year-old female child, who presented with a vestibular fistula and absent uterus and vagina along with left renal agenesis.

A 1-year-old female child was referred to us as a case of rectovestibular fistula for definitive management. The child had undergone a sigmoid loop colostomy in the neonatal period. On clinical examination of the perineum, a fistula was present in the vestibule just below the urethral opening, but no vaginal canal was found. Ultrasound also could not locate the uterus and vagina, however, both ovaries were present normally. Left kidney could not be located. A magnetic resonance imaging scan was advised to confirm the findings, which also revealed the absence of uterus and vagina along with left kidney. Both ovaries were found. No skeletal and cardiac malformations were found on X-rays and echocardiography. A distal cologram was done to delineate the length of distal segment of colon which was found to be normal. A karyotyping revealed normal 46XX. The child was taken up for surgery with a plan to preserve the fistula and lower rectum to act as substitute for vagina. An abdomino-perineal approach was used. On laparotomy, no uterus found, while both ovaries were present attached to a cord-like structure from the lateral pelvic walls and were found to be elongated in shape without any evidence of fallopian tubes. Left kidney could not be found. The distal colon was mobilized until just below the peritoneal reflection and divided. The distal rectal stump was closed in two layers. A retorectal tunnel was developed by blunt dissection. Proximal colon was mobilized adequately to reach the proposed anal site without tension. The

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sphincteric complex was identified with the help of a muscle stimulator, and a tunnel was developed through it and gradually dilated by progressive Hegar’s dilators. The colon was pulled through the sphincteric complex and anoplasty was done using vicryl 4-0 interrupted sutures. Biopsy was taken from both the gonads, and abdomen was closed. The child made an uneventful recovery. The gonadal biopsy came out to be normal (Figures 1-4).

DISCUSSION

Association of the imperforate anus with MRKH syndrome is very rare, with <25 cases reported so far in world literature. In a series of 1007, cases of imperforate anus, Pena has described only 8 cases with uterovaginal or vaginal agenesis. Gross reported only 2 cases of vaginal agenesis in a series of 507 cases. Ein and Stephens published in 1971 their experience with two patients also operated on with an abdomino-perineal approach, preserving the lower rectum as a neo-vagina. The identification of this syndrome is very important before a definitive pull through is attempted. External appearance of these patients is like a typical girl with either a rectovestibular fistula or imperforate anus without fistula. There are two ways of surgical management of these cases. The first is the traditional method using posterior sagittal anorectoplasty (PSARP) or anterior sagittal anorectoplasty (ASARP) to mobilize the fistula and doing a pull through. However, in these case, a some form of vaginoplasty will be required at a later date. We used the method of leaving the distal most rectum and vestibular fistula to become the neovagina, and the proximal colon was pulled through the sphincteric complex to create the neoanus.

Preservation of fistula and lower rectum can be done only if a diagnosis of absent vagina is made pre-operatively, because once the fistula is mobilized as in traditional PASRP or ASARP, it cannot be used as vaginal replacement. The advantage of the procedure we have described is that it is relatively simple to perform and at the same sitting both vagina and anus can be created. There is no chance of damaging any neural innervation which can happen if attempts are made to mobilize the fistula from urethra as there is no vagina in between. Pena has described a similar technique, but they have used a posterior sagittal approach for doing it. This has advantage of avoiding a laparotomy. But if a uterus is present with absence of a part or complete absence of the vagina, then an abdomino perineal approach might offer a better exposure to allow anastomosis of the rectal pouch to uterus or upper vagina, to achieve continuity of reproductive system. Second, a laparotomy also allows inspecting the ovaries and taking biopsies from them which

![Figure 1: Ligated distal rectal stump just below the peritoneal reflection](image1)

![Figure 2: Right ovary without any fallopian tube](image2)

![Figure 3: Left ovary without any fallopian tube](image3)

![Figure 4: Final appearance after pull through of proximal colon and the undisturbed vestibular fistula](image4)
cannot be done via PSARP. Nowadays with the use of laparoscopy, this procedure can be done laparoscopically too. Pena reported in their case series of two cases which had become old enough, where the neovagina created with this method was functional sexual activity and was capable of menstruation. 

**CONCLUSION**

The unique combination of a vestibular fistula in a case of anorectal malformation with vaginal agenesis, offers this novel method of reconstruction, which is easy to reproduce with minimal dissection and opportunity to correct both the problems at the same sitting, and thus, avoiding a subsequently more elaborate and difficult vaginal replacement. We have found this technique to be very useful, and we recommend its use more widely for this particular anomaly.

**REFERENCES**


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