

The Mayer-Rokitansky-Küster-Hauser (MRKH) Syndrome Associated with Anorectal Malformation – Rectovaginal Fistula; A Rare Combination: Case Report

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Abstract: *The Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is characterized by either isolated uterovaginal agenesis or associated with other organ anomalies in genetic women with normal development of secondary sexual characteristics. It affects at least 1 out of 4500 Women. We report an uncommonly seen unique sub type of MRKH syndrome, co-existing with imperforate anus and rectovaginal fistula (RVF) in a 13 year old adolescent girl presented with passage of stool through introitus. We have discussed in brief the management including vaginal reconstructive surgery and detailed counseling regarding the potentials of coitus and fertility issues.*

Keywords: Anorectal malformation, Mullarian agenesis, Mayer-Rokitansky- Küster -Hauser syndrome, Rectovaginal fistula, Vaginal agenesis.

1. Case Report

A 13 year old adolescent female from rural area of under developed Hyderabad Karnataka came with history of passage of stool through introitus since birth and not attained menarche. Further history revealed that she was delivered vaginally at home with an uneventful postnatal course. External examination showed normal female phenotype like normal breast development, body hair distribution, including pubic and axillary hair (Tanner stage IV), average height and weight, and normal arm span. Perianal examination showed well estrogenized vulva, labia minora, labia majora and clitoris.

Further examination revealed normal urethral opening and an imperforate anus. She was passing stools through vagina and a rectovaginal fistula was noticed (Figure 1). A tentative diagnosis of anorectal malformation with RVF was made and we proceeded with imaging studies for further evaluation.

An ultrasonography of abdomen and pelvis confirmed normal in size and appearance of renal system; ovaries and fallopian tubes bilaterally, absence of uterus and vagina, suggestive of MRKH syndrome. Further genitourinary contrast study through introitus showed that rectum was continuous with so called vagina suggestive of rectovaginal fistula (Figure 2). Skeletal and auditory anomalies were ruled out by skeletal survey and hearing test. Karyotyping, routine blood and urine profile were within normal range.

These findings suggest the rare co-existence of MRKH syndrome; in association with RVF. This patient was referred to pediatric surgery department, where a three staged surgical procedures was performed. Initially, the diverting transverse loop colostomy was done. This was

followed by posterior sagittal anorectoplasty (PSARP) at six weeks, where in the distal most part of rectovaginal fistula was transected at the level of peritoneal reflection and left as blind ending functional neovagina. The proximal bowel was pulled through at the proposed neo-anal site (Figure 3). Two months later, colostomy was closed completely. Toilet training was taught to the patient before discharge.

The patient and her family were counseled before and throughout the treatment procedure. The need of assisted fertility techniques including gestational surrogacy to create a family of genetic offspring in future was explained. Patient was also advised for regular follow up to curtail psychological distress and quality of life outcomes. On follow up, patient was doing well with good perineal cosmesis and regular passage of stools with satisfactory continence.

2. Discussion

Mayer-Rokitansky-Küster-Hauser syndrome is a müllerian-duct anomaly causing primary amenorrhea, reported in 1 out of 4500 female births [1]. It is characterized by congenital aplasia of the uterus and upper vagina in a young female with normal karyotype (46, XX), intact ovaries and fallopian tubes along with correctly timed pubarche, thelarche [2]. With recent studies expression and/or functional defects of one or several HOX genes is established and appear to be transmitted as an autosomal dominant trait with incomplete penetrance and variable expressivity [2][3]. It is subdivided in to two types: Type I (isolated) or Rokitansky sequence (OMIM 277000) and type II or Müllerian Renal Cervico-thoracic Somite (MURCS) association (OMIM 601076) [4][5].

Detection of isolated anorectal malformation is usually made due to passage of meconium through vagina in neonatal period whereas in missed cases diagnosis delayed until adolescence, where primary amenorrhea brings the patients to the clinicians. The use of ultrasound, intravenous urography and magnetic resonance imaging provide information about the degree of abnormality and associated anomalies.

Treatment includes creation of a neovagina to allow sexual intercourse. Recently intestinal vaginoplasty is gaining popularity due to reduced chances of stenosis and dyspareunia associated with traditional methods. Graft harvested from intestinal segment maintains its vascularity via mesentery and mucus secreted by bowel provides lubrication [6].

In such cases, interdisciplinary approach is required to provide custom-made care, because of significant diversity in anatomic variants and their respective long-term sexual and reproductive outcomes. Assisted fertility techniques/surrogacy enable women to have genetic offspring. It is essential for the patients and their families to attend counseling, pre-intra -postoperatively.

On extensive retrospective review of literature, incidence of imperforate anus with rectovestibular / rectovaginal fistula is found to be rare and has been reported to be less than 1% in large series of patients [7]. Scanty reported cases are associated with rectovestibular fistula rather rectovaginal fistula. In the case we have reported, MRKH with anorectal malformation - rectovaginal fistula is a rare unique combination.

Shu Wang (2010) reviewed 133 cases of MRKH in 10-year span; three cases of uterovaginal agenesis concomitant with rectovestibular fistula and imperforate anus were reported [8].

Mahajan JK (2009) described a MRKH syndrome with H-type anovestibular fistula and cloacal malformation presented in early infancy. He also mentioned that anorectal malformations are uncommonly associated with MRKH syndrome [9].

Shalika Jayaswal, Nitin Dhende, SB Mane reported similar case in 3 month old baby passing of stools from vagina on straining. Though the vaginoplasty was done in single stage, child had recession of neo anal opening which was further managed by reanoplasty and sigmoid colostomy [10].

In the present case patient was unaware of existence of anomaly. Though her parents noticed that she was passing stools through vagina, due to superstitious belief and social fear, they did not consult any doctor. Thus major congenital anomaly being detected by physical examination at birth was missed. This case also indicates the significance of newborn examination and institutional delivery. Despite of all, there was no history of repeated febrile episode suggestive of urinary tract infection.

In conclusion, though the MRKH syndrome association is rare and may masquerade, combination with anorectal

malformation - rectovaginal fistula is extremely rare. In every case of anorectal malformation imaging studies of mullarian structures are mandatory. Recently with vaginal reconstructive surgery, cosmetic and continence aspects are promising.

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Figure 1: Mrkh Patient Before Surgival Correction



Figure 2: Genitourinary Dye Study Showing Only Bowel System, No Genital Structures



Figure 3: Neoanus Created