African Journal of Urology (2017) **23**, 154–159

African Journal of Urology

Official journal of the Pan African Urological Surgeon's Association
web page of the journal

www.ees.elsevier.com/afju
www.sciencedirect.com

Urology
Short communication

Ileum neovaginoplasty for Mayer–Rokitansky–Küster–Hauser: Review and case series

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Received 26 February 2016; received in revised form 14 July 2016; accepted 21 September 2016
Available online 21 February 2017

KEYWORDS
Vaginal agenesis;
Mayer–Rokitansky–Küster–Hauser syndrome;
Ileum segment neovagina

Abstract
Objective: To review treatment modalities of Mayer–Rokitansky–Küster–Hauser syndrome, and to present further evidence on the successful use of ileum segment as an additional procedure for the creation of a neovagina.
Methods: Five women presented with primary amenorrhea, normal secondary female sexual characteristics, normal external anatomy, shortening of the vagina, with only vaginal dimples. Abdominal ultrasound scans suggested the absence of uterus. Both ovaries were present with normal kidneys. Karyotyping confirmed XX genotype. Pelvi-abdominal MRI confirmed the diagnoses. All patients were started on nonsurgical treatment, in the form of graduated dilators, as a first-line approach. This was not acceptable to the patients. Decisions were made to resort to ileum vaginoplasty.
Results: The mean surgical time was 5 h. Three patients sustained intra-operative bladder injury. All patients reported watery vaginal discharge. Four patients were followed up for 5 years, and were satisfied with their results. This was confirmed by clinical examination. The fifth patient failed to attend for immediate postoperative care. The vagina was 6–7 cm long with introital stricture.
Conclusion: Women with Mayer–Rokitansky–Küster–Hauser syndrome who need vaginal reconstruction have the surgical option of a vascularized free ileum graft that provides a durable, lubricated neovagina without the scarring of skin grafts.

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Peer review under responsibility of Pan African Urological Surgeons' Association.

http://dx.doi.org/10.1016/j.afju.2016.09.003

Introduction

Mayer–Rokitansky–Küster–Hauser (MRKH) syndrome is a rare syndrome that affects 1 in 4000–5000 women in the general population [1]. It is characterized by congenital aplasia of the uterus and the upper two-thirds portion of the vagina in women who have normal ovarian function and normal external genitalia, with normal secondary sexual characteristics during puberty and primary amenorrhea [1].

Development of the female genital tract is a complex process dependent upon a series of events involving cellular differentiation, migration, fusion, and canalization. Failure of any one of these processes results in a congenital anomaly [1].

The female genital tract is derived from the Müllerian ducts, urogenital sinus, and vaginal plate. The two Müllerian ducts are initially composed of solid tissues and lie side by side. Subsequently, internal canalization of each duct produces two channels divided by a septum that is reabsorbed in a cephalad direction by 20 weeks. The cranial portions develop into the fimbria and fallopian tubes, while the caudal, fused portions form the uterus and upper vagina [1].

The sinovaginal bulbs are two solid evaginations originating in the urogenital sinus at the distal aspect of the Müllerian tubercle. The sinovaginal bulbs proliferate into the caudal end of the uterovaginal canal to become a solid vaginal plate then degeneration of the central cells of this vaginal plate occurs in a cephalad direction. Canalization is complete by 20 weeks [1].

Approximately 50% of MRKH syndrome patients have urologic anomalies, such as unilateral renal agenesis, pelvic or horseshoe kidneys, or irregularities of the collecting system, and 45% have skeletal anomalies involving the spine, ribs, and extremities. Other less common anomalies include congenital heart lesions, abnormalities of the hand, deafness, cleft palate, and inguinal or femoral hernias [2].

The signs and symptoms of MRKH syndrome vary greatly. In most cases, the uterus and/or the vagina are aplastic; in other rare cases, there may be atresia of the upper portion of the vagina and an underdeveloped or rudimentary uterus. In some cases, the Fallopian tubes may be affected. The initial symptom of MRKH syndrome is primary amenorrhea. On physical examination, the external genitalia are normal. A vaginal dimple or small pouch with a hymenal fringe is usually present, as the vaginal pouch and hymen are both derived from the urogenital sinus [2].

The cause of MRKH syndrome remains largely unknown. Initially, the syndrome was thought to occur sporadically due to non-genetic factors such as gestational diabetes or exposure to teratogens, but no link between an environmental cause and MRKH syndrome has ever been established [3].

Some case studies suggest that MRKH syndrome is a genetic disorder that is inherited as an autosomal dominant trait with incomplete penetrance and variable expressivity. Polygenic multifactorial inheritance has also been proposed as a cause. Seven deletions of chromosomal segments have been identified in chromosomes 1, 4, 8, 10, 16, 17 and 22, and 1 duplication on the X chromosome. The candidate genes HNF1B (formerly TCF2), LHX1, TBX6, ITIH5 and SHOX are currently under investigation [4].

It has been noted that some males may exhibit absence or underdevelopment of the Wolffian duct, thus affecting vas deferens, with oligo- or azoospermia, kidney abnormalities, spinal malformations and hearing impairment. Rare cases in both males and females in the same family suggest a shared genetic origin [5].

Some disorders may be similar to those of MRKH syndrome, where comparisons are useful in the differential diagnosis. A mutation of the WTN4 gene causes a rare genetic syndrome that affects females. It is characterized by the absence of the uterus, short or stenosed vagina, abnormally high levels of androgens, with acne and hirsutism, and normal female secondary sexual characteristics during puberty, but with primary amenorrhea. The gonads may be of ovitesticular types that produce both male and female sex hormones [6].

Other rare syndromes include complete androgen insensitivity syndrome, Winter syndrome, McKusick–Kaufman syndrome, Frasier syndrome, Goldenhar syndrome, VACTERL association, and Turner's syndrome [6].

Other disorders may be associated with MRKH syndrome secondary characteristics, and are not necessary for a differential diagnosis, include Klippel–Feil syndrome, Sprengel deformity and DiGeorge syndrome [6].

The diagnosis of MRKH syndrome is based upon detailed history, clinical evaluation and transabdominal ultrasonography, complemented by magnetic resonance imaging. Karyotyping may be performed to rule out other conditions. Once MRKH syndrome is diagnosed, a search must be undertaken for renal, skeletal, hearing and cardiac abnormalities. Normal levels of follicle stimulating hormone, luteinizing hormone, 17ß-oestradiol and androgens will confirm the integrity of ovarian function.

The treatment of MRKH depends upon the patient's age at diagnosis. Gynecologists, pediatricians, urologists, orthopedic surgeons, plastic surgeons, physiotherapists and psychological support may be needed for a comprehensive approach to treatment.

For the treatment of vaginal aplasia, medical and surgical care is necessary for creating a neovagina when the patient is emotionally and sexually mature.

Nonsurgical techniques are considered. The first-line approach is the Frank and Ingram technique through the application of self-administered progressive pressure to the perineum using vaginal dilators [7].

The surgical modalities include a variety of techniques, with no consensus as to which vaginoplasty technique is best. The most common surgical procedure used for vaginal reconstruction is the McIndoe procedure, where a split-thickness skin graft from the thigh or buttocks is placed in a pocket between the urethra and rectum that is created by blunt dissection. A cylindrical stent is left in situ. The edges of the graft are sutured to the cut edges of the introitus. The labia majora are sutured together to hold in the mold. A Foley catheter is inserted. Both catheters and stent are removed after one week; the neovagina is irrigated [8].