CASE SERIES

NATIONAL JOURNAL NJMR MEDICAL RESEARCH

Mayer-Rokitansky-Küster-Hauser Syndrome: A Case Series

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DOI:
10.55489/njmr.160120261223

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Date of Submission: 20/09/2025
Date of Acceptance: 25/11/2025
Date of Publication: 01/01/2026

Funding Support:
None Declare

Conflict of Interest:
The authors have declared that no conflict of interests exists.

How to cite this article:
Bhugra P, Duhan N, Malik R, Verma M, Karmakar O, Chowdhary N. Mayer-Rokitansky-Küster-Hauser Syndrome: A Case Series. Natl J Med Res 2026;16(01):33-38. DOI: 10.55489/njmr.160120261223

ABSTRACT

Mayer-Rokitansky-Küster-Hauser Syndrome (MRKH syndrome), also referred to as müllerian dysgenesis, is a rare congenital disease that causes utero-vaginal hypoplasia or aplasia with normal secondary sexual characteristics and a karyotype 46, XX. This retrospective study included five MRKH syndrome patients. The mean age at presentation was 20.2 years. All five patients (100%) reported with nonattainment of menarche; among them, two patients (40%) experienced dyspareunia, while one patient (20%) reported cyclic abdominal pain. Clinical assessment and diagnostic investigations confirmed the presence of MRKH syndrome. The patients and their families were counselled regarding anatomical abnormalities and available fertility options. Vaginoplasty, aimed at creating a neo-vagina, was a key component in the management approach. The neo-vagina was successfully constructed in four out of five patients (80%), whereas in one patient (20%), the procedure was discontinued due to a rectal injury. One patient (20%) also underwent additional surgery involving removal of the left-sided rudimentary uterine horn and preservation of the functional endometrium on the right side. This preserved endometrium was anastomosed with the neo-vagina to facilitate menstrual outflow, potentially maintaining the patient's reproductive capability.

Keywords: Mayer-Rokitansky-Küster-Hauser syndrome (MRKH syndrome), Mullerian dysgenesis/aplasia, Primordial uterus, Primary amenorrhea, Vaginoplasty

INTRODUCTION

One in 4,500 female newborns has Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome, a rare reproductive system disorder.[1] Aplasia or hypoplasia of the uterus, fallopian tubes, and upper vagina characterize this syndrome. MRKH syndrome has two types: 1 and 2. Only the reproductive system is affected in type 1, also known as isolated Müllerian aplasia or Rokitansky sequence. Type 2 syndrome also known as Mullerian duct aplasia-renal agenesis-cervicothoracic somite dysplasia (MURCS association) comprises uterovaginal hypoplasia or aplasia, renal, skeletal, and cardiac abnormalities.[2]

The etiology of MRKH syndrome is undetermined. Mutations in the WNT4 gene, high androgen levels, and diethylstilbestrol and thalidomide exposure are suggested causes. [3-5] The WNT4 gene is essential for the control of ovarian androgen production as well as the development of the Müllerian ducts. Failed Mullerian duct formation and virilization are associated with a particular mutation (L12P) in exon 1 of the WNT4 gene, which results in increased expression of androgen synthesis-related enzymes such as 3β-hydroxysteroid dehydrogenase and 17α-hydroxylase.[3]

Patients with MRKH syndrome are typically diagnosed

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Publisher: Medsci Publications [www.medscipublications.com] ISSN: 2249 4995 Official website: www.njmr.in

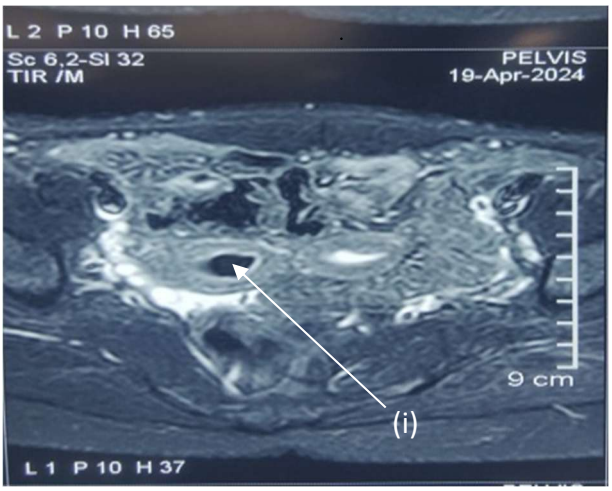
during adolescence or early adulthood when they seek medical consultation for primary amenorrhoea at a gynaecology and obstetrics clinic.[6] Because their ovaries are functioning, these individuals typically have normal secondary sexual characteristics even when they do not menstruate.[7] Given the rarity of this condition, both diagnosis and surgical management of MRKH syndrome are infrequently encountered. This report presents a case series involving five patients diagnosed and managed for MRKH syndrome at a tertiary care centre, highlighting the importance of early identification and treatment strategies.

CASE SERIES ANALYSIS

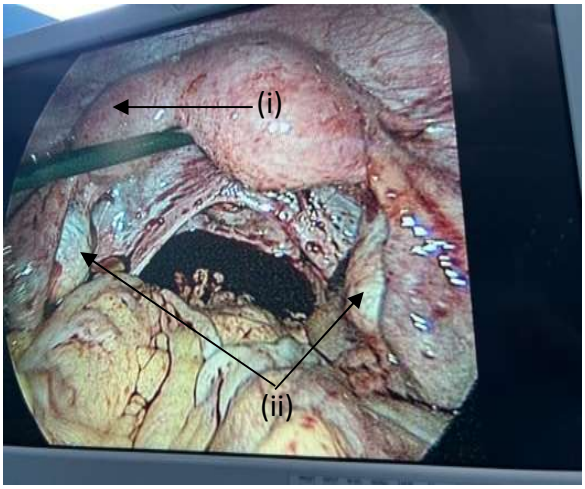
Case 1

A 14-year-old girl visited our outpatient department (OPD) with primary concerns of absent menarche and experiencing cyclic lower abdominal pain occurring every 35-40 days over the past four months. She measured 5'4" in height and exhibited a broad neck. Her axillary and pubic hair development corresponded to Tanner stage 3. Abdominal examination revealed no palpable mass. The local examination showed a normally located urethral opening, the vaginal opening, and clitoris were absent [Fig1(a)]. The clinical diagnosis of MRKH syndrome was made, and investigations were obtained to confirm the diagnosis. Abdomino-pelvic ultrasound revealed right unicornuate uterus, for detailed findings MRI was obtained which was reported as – right unicornuate uterus of size 56 X 35 X 24 mm containing hypodensities suggestive of blood products, small hypoplastic cervix was present on the unicornuate uterus and a blood-filled horn was arising from left side of the lower uterine segment of right unicornuate uterus [Fig 1(b)]. The karyotyping revealed 46, XX chromosomes. Laparotomy was performed, left non-communicating horn arising from the right unicornuate uterus, along with left fallopian tube, was excised [Fig1(c) and (d)].

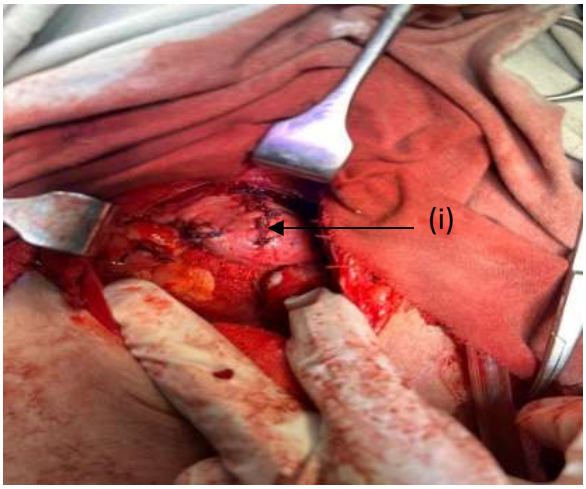
(a)



(b)(i) Left hypoplastic uterine cavity filled with blood



(C) (i) Left non-communicating rudimentary horn
(ii) Normal Ovaries



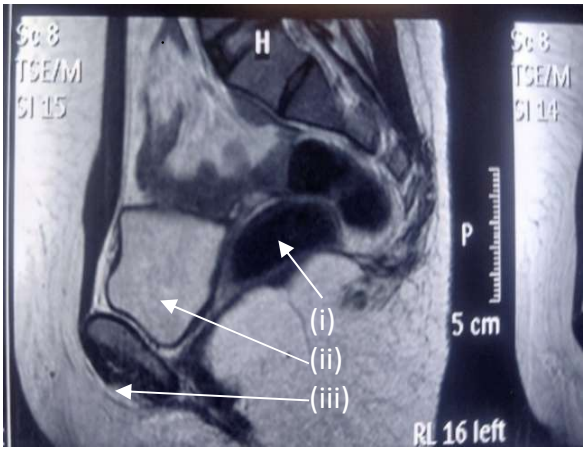
(d) (i) Right rudimentary uterine cavity

Figure 1: MRKH Syndrome diagnosed in a 14- year-old girl showing (a) absent vaginal opening (b) two hypoplastic uterine cavities; left filled with blood on MRI scan-axial section T2 weighted image (c) Laparoscopic view showing two hypoplastic uterine bodies connected with each other and normal ovaries (d) operative view of the same patient showing right uterine body; it was connected with newly created vagina

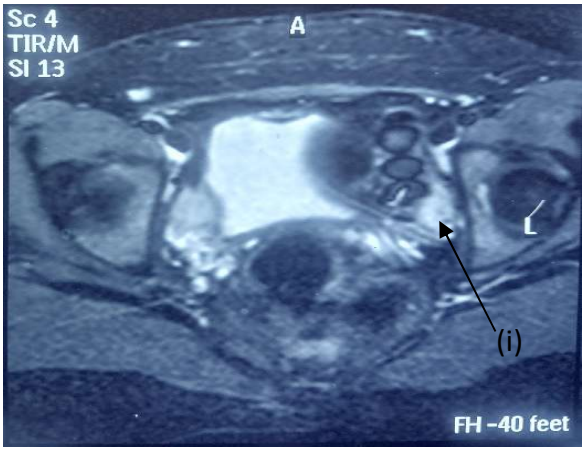
A neo-vagina was created between external urethral opening anteriorly and rectum posteriorly. An opening was created at the lower end of uterine body. It was connected with the neo-vagina. A T-tube was placed in the uterine cavity. Its lower end was secured in the neo-vagina to prevent cervical stenosis. A vaginal mould was placed within the neovagina, which has been lined using split-thickness skin graft harvested from anterior aspect of thigh. Patient has been under regular follow-up for the past year and has been menstruating consistently.

Case 2

A 22 y/o unmarried female reported to OPD with the primary complaint of absent menstruation. She did not report any history of cyclic abdominal pain, vasomotor symptoms, delayed puberty, or a family history of delayed menarche. On examination, her height, weight, and body mass index were within normal range. Development of breasts, pubic, and axillary hair was found to be normal. On local examination blind vaginal pouch of approximately 1 cm length was detected. The abdomino-pelvic ultrasound showed aplasia of uterus and fallopian tubes; the ovaries and kidneys were normal in position and echotexture. The MRI imaging showed two small 22×20×20 mm-sized uterine tissues in both adnexa abutting the medial side of ovaries [Fig2 (a) and (b)]. Serum levels of luteinizing hormone, follicle-stimulating hormone, and prolactin were all within normal limits, and the serum testosterone concentration corresponded to the normal female range. The karyotyping showed 46, XX chromosomes. Counselling of the family and patient was done regarding menstruation, childbirth, and sex life. Amnion vaginoplasty was performed successfully. Postoperatively, she was advised to use a mould [Fig (c) and (d)] for repeated dilation of the neo-vagina. The patient is on regular follow-up with 4 inches vaginal length.



(a) (i) Rectum (ii) Urinary Bladder (iii) Pubic Symphysis



(b) (i) Normal Ovary

(c)

(d)

Figure 2 (a) Coronal section T2 weighed image shows absent uterus, cervix and vagina (b) Axial image of the same patient show bilateral normal ovaries (c) Blind vagina before vaginoplasty (d) Longitudinally fenestrated cylindrical hard mould covered with amnion

Case 3

A 20-year-old female presented to OPD with the primary complaint of absent menarche. She reported no history of cyclic abdominal pain, vasomotor symptoms, or a familial pattern of delayed menarche. On clinical examination, her height, weight, and body mass index were all within normal limits. The secondary sexual characteristics were well developed. There was no evidence of virilisation, hirsutism, or webbed neck. On local examination, the external genitalia were normally developed, but the vaginal introitus was absent. Abdominopelvic ultrasound revealed absence of uterus as well as fallopian tubes, with no detectable abnormalities in the ovaries or kidneys. Serum concentration of luteinising hormone, prolactin, follicle-

stimulating hormone, moreover testosterone was all found to be within normal physiological limits. The karyotyping showed 46, XX chromosomes. Counselling of the family and patient was done, and amnion vaginoplasty was performed. Postoperatively, with repeated use of mould, she has vaginal length of approximately 3.5 inches.

Case 4

An 18-year-old girl reported to the OPD with the chief complaint of dyspareunia. On further history taking she admitted to have non-attainment of menarche. On examination her height was 5′ 2″ Her breast, axillary and pubic hair examination suggested Tanner stage 4. On local examination the external genitalia appeared normal for the age but the vaginal introitus was absent. On ultrasound uterus and fallopian tubes were absent, ovaries were normal in size and echo-texture. Serum levels of luteinising hormone, follicle stimulating hormone and prolactin were normal. The karyotyping was done for confirming the diagnosis of MRKH syndrome. After explaining the fact that she will continue to have amenorrhea even after the surgery and she can have baby with the help of artificial reproductive techniques and surrogacy, amnion vaginoplasty was performed. A hard vaginal mould was provided for dilation of the neo-vagina in the post-operative period. On routine follow-up 3 months after surgery, she had a vagina of approximately 3 inches.

Case 5

A 27-year-old woman, an already diagnosed case of MRKH syndrome reported to the OPD for vaginoplasty. She was advised vaginoplasty before her marriage when she consulted for absence of menstruation. Presently, she had history of dyspareunia. On examination her height was 5′ 3″. The breasts, axillary & pubic hair of the patient were normally developed. On abdominal examination no lump was palpable. The local examination revealed that the external genitalia were normal for the age but the vaginal pouch was blind, approximately less than one cm in length. On per rectal examination no mass was palpable anteriorly which suggested absent uterus and cervix. Her ultrasound showed rudimentary uterine bulb, absent cervix and vagina; the ovaries and the kidneys were normally placed and normal in echo-texture. The MRI confirmed above findings. The patient was prepared for McIndoe vaginoplasty after explaining that it will serve only coital function. The median raphe between the urethra and rectum was extremely thin in this patient, this predisposed to rectal injury at median raphe in spite of careful blunt dissection. The rectal injury was repaired in two layers and further surgery was abandoned. The patient was advised to follow up after 3 months but unfortunately, she did not turn up.

Clinical details of all above patients are summarized in table 1.

DISCUSSION

The initial clinical indication of MRKH syndrome is typically primary amenorrhoea in individuals who exhibit a normal female phenotype, possess a 46, XX karyotype, have functional ovaries, and show no signs of androgen excess.[1] MRKH syndrome must be distinguished from other conditions that also present along primary amenorrhoea and normal secondary sexual development, such as transverse vaginal septum, imperforate hymen, polycystic ovarian syndrome (PCOS), androgen insensitivity syndrome, and female intersex disorders.[8]

Once MRKH syndrome is clinically suspected, diagnostic evaluation is necessary to identify abnormalities of the reproductive tract. Abdominal and pelvic ultrasonography is generally the first-line imaging technique, though magnetic resonance imaging (MRI) of abdomen and pelvis offers more comprehensive visualization of uterine anomalies.[9] When findings from ultrasonography and MRI are inconclusive, laparoscopy may be employed to aid further assessment. A definitive diagnosis of MRKH syndrome is established by correlating imaging and laparoscopic findings with confirmation of a 46, XX karyotype.[1]

In patients with MURCS association, imaging such as ultrasonography and MRI may also reveal renal tract anomalies in approximately 40% of cases, including unilateral renal agenesis, hypoplastic kidneys, or horseshoe-shaped kidneys. Additionally, skeletal abnormalities-such as scoliosis, spina bifida, and rib malformation-are observed in 30-40% of these patients, while cardiac anomalies like ventricular or atrial septal defects occur less frequently.[10]

Patients diagnosed with MRKH syndrome frequently experience significant psychological distress and anxiety after discovering the anatomical absence or malformation of uterus and vagina. Therefore, initial step in management involves comprehensive counselling of both the patient and their family, focusing on future possibilities for pregnancy and sexual function before initiating any form of treatment.[11] One treatment goal for most of the affected women is creation of a functional vagina.[12] This may be accomplished conservatively or surgically(vaginoplasty). The surgical treatment is indicated especially if the vaginal length is less than 1 cm. The vaginoplasty can be performed by various methods; difference between these procedures is in the tissues used to line the vaginal canal, such as using split thickness skin graft (McIndoe procedure), amnion, oxidised cellulose, using the sigmoid colon (Davydov's procedure), and Vecchietti's procedure, where traction of the vaginal vestibular mucosa is performed.[13] In our case series, the amnion was used in 3 patients to line the vaginal canal (amnion vaginoplasty). Amnion causes epithelisation of the vagina and avoids painful scars over the buttocks and thighs in postoperative period, which can occur when split split-thickness skin graft is used.[13] McIndoe vaginoplasty was successfully performed in 1 patient, while in another patient, this procedure was abandoned due to rectal injury.

Table 1: Salient features of patients with MRKH syndrome

Criteria studied	Case 1	Case 2	Case 3	Case 4	Case 5
Age (years)	14 yrs	22 yrs	20 yrs	18 yrs	27yrs
Symptoms					
a) failure to attain menarche	Yes	Yes	Yes	Yes	Yes
b) cyclical abdominal pain	Yes	_	_	_	_
c) Dyspareunia	_	_	_	Yes	Yes
Signs					
a) Vagina	Absent	Blind vaginal pouch of approx. one cm	Absent	Absent	Blind vaginal pouch of less than 1 cm
b) Rest of the external genitalia and secondary sexual characteristics	Developed except absent clitoris	Developed	Developed	Developed	Developed
Imaging					
a) Ultrasound	Right unicornuate uterus with normal ovaries	Absent uterus and fallopian tubes, normally located ovaries	Absent uterus and vagina, normal ovaries present	Absent uterus and its appendages but normal ovaries present	rudimentary uterine bulb present along with normal ovaries
b) MRI	Right unicornuate uterus of 56X35X24mm having hypoplastic cervix, another left hypoplastic blood filled horn arising from lower part of right horn	Two small 22X20X20 mm uterine tissues present in both adnexa abutting the medial side of ovaries		_	Rudimentary uterine bulb of 18X20x24 mm present, absent cervix and vagina
Surgical procedures					
a) Vaginoplasty	McIndoe vaginoplasty	Amnion vaginoplasty	Amnion vaginoplasty	Amnion vaginoplasty	Procedure abandoned due to rectal injury during vaginoplasty
b) Excision of the rudimentary horn and preservation of functional primordium	yes	-	-	-	-

In 2 to 7 percent of women with MRKH syndrome active endometrium develops in the hypoplastic uterus, and patients typically present with cyclic abdominal pain.[14] One of the patients in our case series presented with cyclic abdominal pain, which suggested the functional endometrium within the primordial uterus. The abdominal and pelvic magnetic resonance imaging (MRI) in this patient showed a) right unicornuate uterus of size 5.6x3.5x2.4cm, containing blood products, b) small cervix was present on right unicornuate uterus, and c) left noncommunicating horn arising from lower uterine segment. In this patient, we preserved the functional endometrium on the right side, excised the rudimentary horn on left side, and used split-thickness skin graft for vaginoplasty. The surgical procedure relieved the obstruction of the uterine cavity, allowing the patient to resume normal, unobstructed menstrual flow.

Pregnancy in patients with MRKH syndrome may be achieved by a) preservation of functional endometrium, b) oocyte retrieval, fertilization, and gestational surrogacy, and c) uterine transplantation. [12,15] Uterine transplantation is a newer treatment modality in which the uterus from a compatible donor (usually mother or sister) is implanted in patients with müllerian agenesis. The woman is put on immunosuppression throughout pregnancy to avoid rejection. Uterine transplantation has resulted in successful live births.[13]

CONCLUSION

MRKH syndrome is a rare disease, but it must be suspected in a patient presenting with primary amenorrhoea. The clinical examination, ultrasound & MRI of the pelvis & abdomen, hormonal studies, and karyotyping help in confirmation of the diagnosis of MRKH syndrome and detection of reproductive tract anomalies. Parental and patient counselling is crucial when it comes to managing reproductive tract abnormalities and future pregnancy choices. The most common surgical procedure in the affected women is creation of a sexually viable vagina. If functional endometrium is present, its preservation and anastomosis with neovagina will ensure menstruation and may preserve fertility potential.

Individual Author's Contribution: PB contributed to study conception, design, data collection, analysis, and

manuscript preparation. **ND** participated only in manuscript writing, while **RM** contributed to data analysis. **MV** assisted in data collection and manuscript preparation. **OK** and **NC** supported the study through data collection activities only, without involvement in other research processes.

Availability of data: The data that support the findings of this study are available from the corresponding author on reasonable request.

Declaration of Non-use of generative AI Tools: This article was prepared without the use of generative AI tools for content creation, analysis, or data generation. All findings and interpretations are based solely on the authors' independent work and expertise.

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