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# A UNIQUE CASE REPORT OF MRKH SYNDROME

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**Abstract**
Mullerian duct malformation occurs in various forms and each anomaly is distinctive. Cervical atresia or agenesis(AFS class 2B) is a rare anomaly and is associated with absence of a portion or whole of vagina. These patients present with primary amenorrhea and well developed secondary sexual characters and cyclical abdominal pain. A comprehensive pre operative evaluation is essential for patients with functioning uterus with congenital absence of cervix and vagina. Hysterectomy is a definitive initial procedure.
A 16 year old girl came to Saims opd on 1/9/2020 with complain of cyclical abdominal pain since 6 months and leakage of urine pervaginal since 2 months. With similar complaints she went to private hospital 4 months back where USG was done suggestive of hematometra of 8X4.7X3.6cm.Vaginal dilatation with drainage of hematometra by abdominovaginal route was attempted but was not successful. Post operatively she had leakage of urine and was referred to higher center.
O/E Patient was stable with well developed secondary sexual characters. P/A 5x8cm. firm mass palpable in right iliac fossa. L/E a blind vaginal pouch of 1x1.5cm was seen with catheter in situ. On cystoscopy large VVF was seen at bladder neck ,2cm.proximal to vaginal orifice and on MRI 14x5.6x5cm distended uterus was seen with atretic cervix and vagina along with absent right kidney and dysplastic wedge shaped vertebra in lumbo sacral area consistent with type 2 MRKH syndrome. Patient taken in OT on 18/9/2020 for hysterectomy and VVF repair .Abdomen was opened 14x5x5.6cm size uterus seen .Hysterectomy done.1.5cm. vaginal length left. 2x1 cm . VVF seen at bladder neck and repaired. Postoperative SPC along with urethral catheter and perivesical drain placed. Patient discharged on day 5 with SPC and urethral catheter in situ.
Cervical agenesis hinders outflow tract for menstruation affects reproduction and represents therapeutic dilemma .Hysterectomy is eventual treatment which eliminates suffering from cryptomennrohea , sepsis, endometriosis and multiple operations. Recent advances for restoration or reconstruction of cervical canal is yet to be explored.
**Keywords:** type 2 MRKH syndrome

## Introduction

• Mullerian duct malformation occurs in various forms and each anomaly is distinctive.

• Mullerian ducts differentiate, gets fused and canalised to form fallopian tubes, uterus, cervix and upper vagina. Lower vagina is formed by urogenital sinus(1).

• This progresses cranially to caudally. Arrest at any level leads to “localised gynatresia”.

• Cervical atresia or agenesis (AFS Class 2B) is a rare anomaly and is associated with absence of a portion or whole of vagina.

• Patients with congenital absence of cervix faces a diagnostic challenge.

• Incidence of cervical agenesis is 0.01%, and represents 3% of all anomalies.

• 4.8% of cervical agenesis is associated with functioning uterus.

• Usually presents with primary amenorrhoea and well developed secondary sexual characters with cyclical abdominal pain (2).

• A comprehensive pre-operative evaluation of patients with suspected mullerian duct malformation is essential.

• After complete evaluation the physician should allay the anxiety of patient by giving her accurate description of reproductive implication or the obstetrics consequences of her utero-vaginal anomaly(3).

**Two basic categories of cervical anomalies**:

1.Cervical aplasia -Lacks a uterine cervix and lower uterine segment narrows to terminate in a peritoneal sleeve well above the normal communication with vaginal apex

2. Cervical dysgenesis - Fibrous band, Intact cervical body with obstruction of cervical os, Stricture of mid portion of cervix, Fragmentation of cervix.

• When both cervix and vagina are absent with a functioning uterine corpus it is difficult to obtain a

satisfactory fistulous tract through which menstruation can occur.

• Attempts have been made to make a passage through fibrous tissue & place a stent to keep the tract open. But the endocervical gland couldn't be developed leading to closure of tract by concentric fibrous tissue.

• Endometriosis can develop along tract due to retrograde menstruation.

• Recurrent and severe pelvic infection is common.

• For patients with functioning corpus with congenital absence of cervix and vagina, hysterectomy is done as initial procedure with vaginoplasty by using split thickness graft.

• A hysterectomy eliminates much needed suffering from associated problems such as cryptomenorrhea, sepsis, endometriosis and multiple operations(4).

**Case Report** • A 16 year old girl came to SAIMS OPD on 01/09/2020 with complaint of cyclical abdominal pain since 6 months along with leakage of urine per vaginum since 2 months.

• 4 months ago with similar complaint of primary amenorrhoea and cyclical abdominal pain, she went to private hospital

• USG whole abdomen and pelvis suggested of bulky uterus (9.4* 5.5* 3cm) with cystic distension in endometrial cavity with numerous fine echoes (hematometra/hydrometra) 8* 4.7* 3.6cm, 71ml (?imperforate hymen/septa) along with right renal agenesis.

• Vaginoplasty with drainage of hematometra was done on 22/05/2020. Patient had no relief of symptoms.

• Post vaginoplasty USG revealed 5.8* 4.9cm collection in lower uterine cavity and cervical canal.

• Vaginal dilatation attempted on 20/07/2020 for hematometra drainage by abdomino-vaginal route.

• Post dilatation patient had complaint of urine leakage per vaginum, VVF was diagnosed.

• Patient was then referred to SAIMS.

**Examination**

Upon Examination:

• The general condition of patient is fair, she is well responsive and well oriented to time, place and person.

• Thin built (height-152cm; weight-39kgs; BMI-16.9)

• Secondary sexual characters were well developed

• Bilateral breasts well developed with Tanner stage 4

• Pubic and axillary hair well developed (stage 4)

• Vitally stable.

• Per abdomen: soft/Non tender, NO GTR, No distension, 10cm transverse scar seen over supra-pubic region. A 5*8cm hard lump palpable in Right Iliac fossa and hypogastrium, lower margin not reached.

• Local examination: vulva, labia minora, labia majora and clitoris were normal looking and well estrogenised.

• Per vaginum: vagina appears blind with leakage of urine on coughing/straining. On Digital examination vagina appears to be 1*1.5cm.

**Investigation**

• B positive, Serology-NR

• Hb/TLC/Plt- 13.6/8200/4.26

• Pt/Inr-14/1.0

• Urea/creat-16/0.56

• Urine RM- occ P/c; 1-2 E/c

• Urine C/S showed growth of pseudomonas aeruginosa

• Rest all blood investigations were normal

• Chest X-ray normal Special Investigations

• Cystoscopy 1. Oedema at bladder neck and Right uretric orifice not visualized, left appeared normal. 2. Large VVF seen just below bladder neck, 2cms proximal to vaginal orifice.

• MDCT Urogram Right renal fossa empty Collection of 8.2 * 6.4 * 6.4cm (167cc) in endometrial cavity (hematometra) MRI whole abdomen and Pelvis

• Large oblong structure noted in pelvis region between bladder & rectum. The superior extent of this structure is upto right iliac fossa. This structure contains T1 hyperintense & T2 isointense contents-suggestive of distended uterus with endometrial collections.

• Distended uterus is 14*5.6*5cm.

• Normal cervical anatomy is not visualized ?atretic.

• Atretic small vagina.

• Defect between lower bladder neck and small vagina (?site of fistula).

• Bilateral ovaries normal, with normal follicles.

• Distended endometrial cavity with large uterus

• Absent right kidney and dysplastic wedge shaped vertebrae in lumbosacral region of spine consistent with spectrum of Type 2 MRKH Syndrome

**Intra operative**

• DJ stent placed in left ureter with ureteroscope.

• Pfannesteil incision given to open abdomen. • Oblong distend uterus of size 14*5*5.6cm seen behind the bladder.

• Left side round ligament absent

• Bilateral tubes and ovaries normal.

• A small opening made in fundus of uterus to drain hematometra.

• Hysterectomy done

• Anterior cystostomy done by longitudinal incision.

• 2*1cm defect present at bladder neck, using 11 no blade fistula tract margin freshened with a plane created between urinary bladder and vagina

• Vaginal wall closed with vicryl 2-0 & bladder closed with vicryl 2-0.

• 20 Fr foley's placed in suprapubic catheter along with 14 Fr. foley's in urethra.

• Anterior cystostomy closed in 2 layers with vicryl 2-0

• Abdomen closed after placing peri-vesical drain.

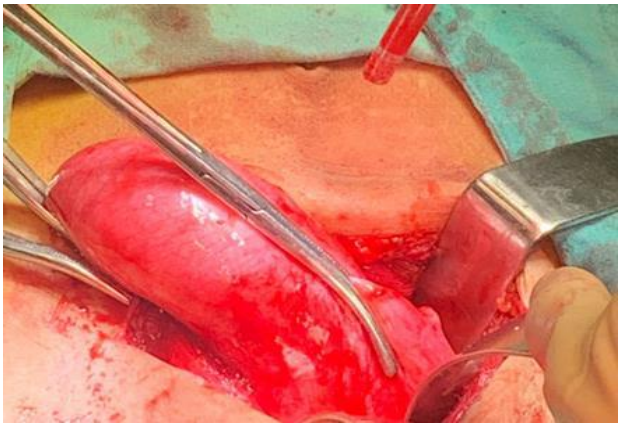

**Figure 1: Distended uterus**

## Discussion

• The MRKH syndrome is rare sporadic anomaly.

• Congenital absence of upper vagina and uterus is the prime feature of the disease and is often associated with unilateral renal agenesis or a dysplasia and skeletal malformations (MURCS association). (5)

• Also called, GRES [genital (G), renal (R), ear (E), skeletal (S)] syndrome.(6)

• This disorder can either be isolated comprising of CAUV (Congenital Absence of the Uterus and Vagina), MA (Müllerian Aplasia), or associated with renal, skeletal and/or hearing defects.(7)

• Cardiac and digital anomalies are rare. Patients with associated anomalies are classified as MURCS association (Mullerian duct aplasia, Renal Dysplasia and Cervical Somite anomalies), indicated as MRKH type II syndrome, or GRES (Genital Renal Ear Syndrome) if middle ear is also affected.(8)

• MRKH is thus of two types 1. Type 1 or Rokitansky 2. Type 2 or MURCS (GRES)

• This patient presented as a case of MRKH syndrome associated with MURCS. (9)

• The treatment of MRKH syndrome patient is complex requiring a multidisciplinary approach.

• Careful dialogue with patient addressing all together gynecological, sexual, psychological and infertility issues is a must.

## Conclusion

• The uterine cervix provides an outflow tract for menstruation

• Cervical agenesis hinders this function, affects reproduction and represents therapeutic dilemma

• Hysterectomy is eventual treatment for cervical agenesis.

• Recent advances led to conservative surgical treatment for restoration of cervical function

• Main problem with restoration or reconstruction lies with development of endometriosis and pelvic infection.

• Safer and simpler therapeutic option, which will restore normal outflow tract and fertility preservation is yet to be explored

## References


1. J Negat Results Biomed 2006 Jan 27;5:1. doi: 10.1186/1477-5751-5-1.
2. Clin dysmorphol 1994 July;3(3): 192-9
3. Niger postgrad Med J 2010 March; 17(1): 64-6
4. Morcel K,Camborieux L, Guerrier D: Mayer-Rokitansky-KusterHauser (MRKH) syndrome. Orphanet J Rare Dis. 2007, 2: 13- 10.1186/1750-1172-2-13
5. DC Dutta's Textbook of Gynecology.
6. ACOG Committee Opinion No. 728 Mullerian agenesis: diagnosis, management, and treatment. Obstet. Gynecol. 2018;131(1):e35–e42.
7. Londra L., Chuong F.S., Kolp L. Mayer-Rokitansky-Kuster-Hauser syndrome: a review. Int. J. Women's Health. 2015;7:865–870.
8. Sultan C, Biason-Lauber A, Philibert P. MayerRokitansky-Kuster-Hauser syndrome; recent Clinical and genetic findings. Gynecol Endocrinol. 2009;25:8-11
9. Sharma BB, Sharma S, Singh Y, Balesa J (2015) Mayer-Rokitansky-KusterHauser Type I Syndrome – A Case Report. Indian J Case Reports 1: 8-10.