

# Mayer - Rokitansky - Kuster - Hauser Syndrome: Two Cases Of A Rare Non Hereditary Disorder In Siblings

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## Abstract

Mayer-Rokitansky-Kuster-Hauser (M.R.K.H.) syndrome is a rare disorder often characterized by congenital absence of the uterus and vaginal. It may be associated with anomalies of the kidneys ranging from Ectopic to congenital absence, and also bony anomalies.

Two cases of this rare disorder occurring in siblings, which, to the best of our knowledge has not been reported before are hereby discussed.

The evaluation by laparoscopy, Ultrasound, Intravenous Urography, Computerized Tomography and classification of these two cases are also discussed.

## INTRODUCTION

Congenital absence of the uterus and vagina, Mayer-Rokitansky-Kuster Hauser (MRKH) syndrome is a rare disorder. The prevalence has been reported as 1 in 4000 -5000 female births.

Two cases of this rare, non-hereditary disorder presenting in siblings are hereby discussed.

## CASE REPORT (A)

EB is a 30year old spinster who presented at the GOPD with primary amenorrhoea. The family history revealed delayed menarche of the mother which was at age 21. She (the mother) however subsequently proceeded to have an uneventful reproductive period; the married elder sibling, age 32 years also has primary infertility and primary amenorrhoea.

General physical examination was essentially normal.

The Central nervous, Cardio-pulmonary and abdominal examination were also normal.

Vaginal examination revealed normal labia majora with Type I female genital mutilation. Normal labia minora, external urethra meatus, vestibule with shallow (2.5cm deep) vagina ; ending blindly.

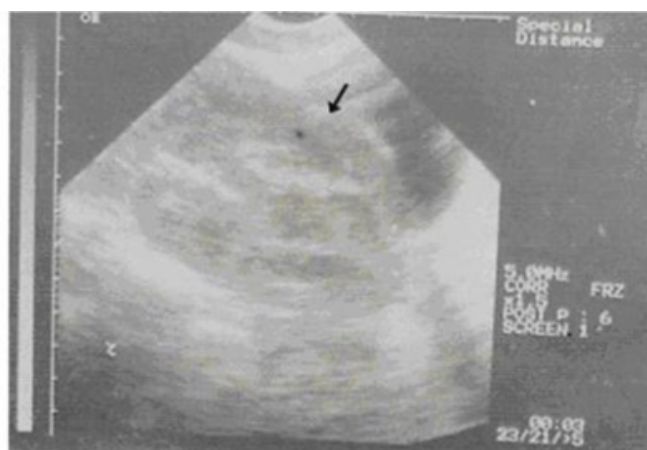
Bimanual pelvic examination revealed a palpable mass of

about 8 x 6cm in the suprapubic region.

Abdomino-pelvic ultrasound [Fig 1] revealed A solitary ectopic kidney which measures 10.2cm x 4.6cm in Longitudinal x Anterio-posterior dimension was seen superior to the urinary bladder. The uterus and ovaries were absent.

## Figure 1

Figure 1: Pelvic ultrasound showing an ectopic pelvic kidney indenting on the urinary bladder superiorly.



A diagnosis of congenital absence of the uterus with Ectopic pelvic kidney was made.

Intravenous urography [Fig 2] revealed partial sacralization

of the fifth lumbar vertebra; renal pelvicalyceal system was demonstrated in the right aspect of the pelvis, superior to the urinary bladder. The calyces appeared splayed with a short ureter which showed normal caliber and smooth outlines.

**Figure 2**

Figure 2: Intravenous urography showing pevicaecal system of the ectopic kidney.



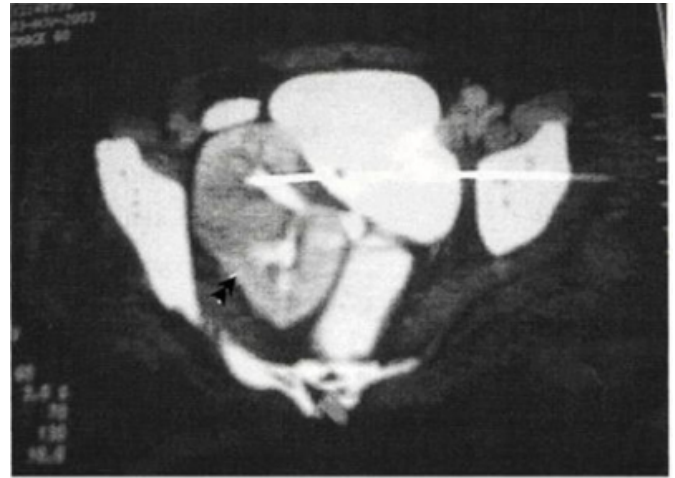
No renal outline or excretion was seen in the lumbar regions.

The diagnosis of an ectopic pelvic kidney was confirmed.

Enhanced Computerized Tomography scan [Fig 3] showed similar findings and also confirmed absence of the uterus.

**Figure 3**

Figure 3: Enhanced axial CT (with rectal contrast) of the pelvis showing ectopic kidney; contrast filled urinary bladder but no uterus.



The Karyotype result came back to be 46 XX; laparoscopy was not done to avoid injury to the pelvic kidney. The final diagnosis was Mayer – Rokitansky – Kuster – Hauser Syndrome.

She was counseled, and had vaginal dilatation (frank technique)

**CASE REPORT (B)**

OO is also a 32 year old teacher, an elder sister to EB, who presented Primary infertility after 4yrs of marriage and primary amenorrhoea.

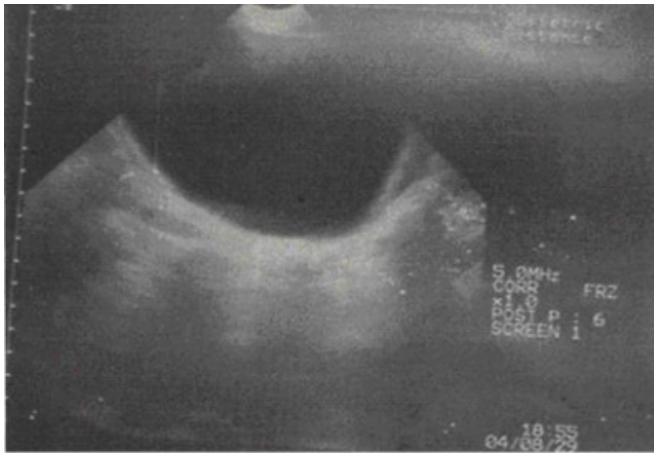
There was history of delayed menarche in the mother and her only sister, age 30 has primary amenorrhoea.

The significant findings were on vaginal examination which showed a blindly ended vagina with a normal depth (8 x6cm). The clitoris, labia majora and minora and the vestibule were normal.

A clinical diagnosis of primary amenorrhoea was made. Abdominopelvic ultrasound [Fig 4] revealed normal liver, spleen and both kidneys. However, no uterine tissue was seen in the pelvis. The ovaries too were not visualized. A diagnosis of congenital absence of the uterus was made.

**Figure 4**

Figure 4: Pelvic ultrasound longitudinal showing the urinary bladder and absent uterus.

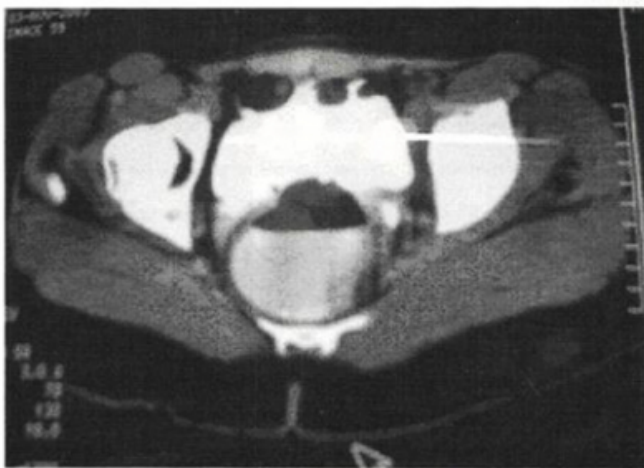


Intravenous urography showed spinal bifida occulta of the sacral vertebrae, the kidneys were normal.

Computerized Tomography scan [Fig 5] revealed similar findings (absence of uterus; with normal kidneys).

**Figure 5**

Figure 5: Enhanced axial CT of the pelvis showing contrast in the urinary bladder (and rectal contrast) but no uterus.



The karyotype result also came out to be 46XX and Laparoscopy demonstrated ovoid pearly white structures (in keeping with the ovaries) bilaterally. The fallopian tubes were also demonstrated but no uterus nor uterine bud was seen.

A diagnosis of Mayer- Rokitansky - Kuster-Hauser syndrome was made.

She was counseled with her husband and offered the option

of assisted conception: surrogate motherhood or adoption.

## DISCUSSION

Agenesis of the uterus and vaginal i.e Mayer -Rokitansky - Kuster - Hauser syndrome (MRKH) represents a dysplasia of the mullerian duct resulting in an absence of the uterus and the middle and upper thirds of vagina<sub>3</sub>. Although the uterus is not present, rudimentary uterine anlagen (structure) are often present. The tubes and ovaries are often normal.

In about 6-10% cases, endometrial tissue or even variable development of the uterus with haematometra may be present, resulting in cyclic abdominal pain<sub>1</sub>.

Associated congenital anomalies of the upper urinary tract are reported in about 30-40% of all cases<sub>2</sub> and these may vary from renal agenesis to pelvic kidney as seen in case A.

Two different types of MRKH syndrome have been described

The typical form/type of the syndrome is characterized by the absent or rudimentary vagina; absent uterus ,only symmetric uterine remnants(the muscular buds (uterine) , both fallopian tubes and normal ovaries are present<sub>1</sub>.

In the atypical variants, there is asymmetry of the uterine remnants: aplasia of one or both muscular bud; and when both are present, one may be larger than the other with associated hypoplasia or aplasia of one or both fallopian tubes. The atypical variant may also show cystic ovaries or anomalies in form or position of the ovaries<sub>1</sub>.

Discrimination between the type A (typical) and type B (atypical) is important because associated renal and ovarian abnormalities occur only in type B.

The second presented case is an example of the typical form because of the presence of normal ovaries and fallopian tubes with no urinary tract anomaly.

The first case presentation is likely to be the atypical form though, laparoscopy was not done due to the risk of injury to the pelvic kidney.

The association between renal and genital anomalies in MRKH syndrome is said to be due to the faulty gonadal differentiation which consequently produces mullerian inhibiting factor and induces mullerian ducts regression. Depending on the onsets of production of mullerian inhibiting factor, and the asymmetry in production by the

two gonads, the development of the mullerian ducts would stop at various stages<sub>1</sub>.

The karyotype of affected patient is usually 46XX<sub>4</sub>. It is however not clear whether this condition is autosomal recessive disorder or single gene defect. Both cases presented have 46XX karyotype.

Growth and development of these patients are usually normal as seen in both cases presented. This is due to normal ovarian function. The secondary sexual characteristics are often well developed though patients with TYPE B MRKH syndrome may have a slight form of female pseudohermaphroditism. None of the two cases have evidence of female pseudohermaphroditism.

The external genitalia are normal in appearance although the vagina may sometimes be a shallow pouch of three cm in depth or is absent. Normal external genitalia was found in both patients ; the vagina was shallow in the first case, but of normal depth, though ended blindly in case 2, possibly because of regular coital dilatation.

Review of the patients' medical history and a simple gynecologic examination usually suggest MRKH syndrome; most of the patients present with primary amenorrhea with or without primary infertility. Case I and Case II presented with amenorrhoea and primary infertility respectively.

Sonography has been recommended for diagnosis of these abnormalities<sub>3,5</sub>. It is useful to confirm uterine agenesis or in some cases, endometrial tissue with uterine bud or even a unicornuate uterus with haematometra may be seen.

Sonography will however not differentiate between TYPE A & TYPE B and may not be able to detect small remnants of endometrial tissue in the muscular bud<sub>5</sub>.

However, sonography will provide similar information to excretory urography on the anomalies of upper urinary tracts.

Preliminary films (of excretory urography) may show anomalies of bony structures which are seen in about 5% of cases of MRKH syndrome<sub>3</sub>. These include sacralization of the 5<sup>th</sup> lumbar vertebra and lumbarization of the 1<sup>st</sup> sacral vertebra.

Both cases have congenital bony anomaly: sacralization of the fifth lumbar and sacral spina bifida in Case I and II respectively.

Laparoscopy is useful in detecting the remnant endometrial tissue and assessing the fallopian-tube thereby classifying the syndrome into TYPE A or B.

However, Magnetic Resonance Imaging also has a role in demonstrating small amounts of endometrial tissue in patients with cyclical abdominal pain<sub>6,7</sub>. It is still very not certain whether MRI will be able to differentiate the two forms accurately<sub>6,7</sub>.

Management option include intensive psychological counselling followed by vaginoplasty - the Franks technique of vaginal dilatation, the Ingram passive dilatation technique or the MCIndoe Reed Split - thickness graft technique can be used.

The first patient had Frank's technique of vaginal dilatation because of the shallow vagina (2.5 cm in depth) while the second patient that needed no dilatation is being prepared for assisted conception. Both patients were lost to follow up.

## SUMMARY

Although, MRKH syndrome is rare, two siblings presenting with primary Amenorrhea with and without Primary Infertility have been discussed. The first patient had associated renal anomaly while the second had normal renal anatomy.

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