

MAYER-ROKITANSKY-KUSTER-HAUSER SYNDROME - A RARE CASE OF UTERINE AND BILATERAL OVARIAN AGENESIS

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ABSTRACT

BACKGROUND

Meyer-Rokitansky-Kuster-Hauser syndrome belongs to Class 1 Mullerian duct anomalies characterised by uterine anomalies including agenesis, hypoplasia or duplication.

CASE REPORT

We are presenting a case of a 19-year-old female patient with primary amenorrhoea and absence of bilateral breasts. Ultrasound and MRI pelvis were done.

CONCLUSION

MRI is an excellent modality to demonstrate the uterine, ovarian and renal anomalies that may be associated with Meyer-Rokitansky-Kuster-Hauser syndrome.

KEYWORDS

Mullerian Ducts, Primary Amenorrhoea, Dyspareunia.

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BACKGROUND

Mayer-Rokitansky-Kuster-Hauser syndrome is a malformation of the female genital tract due to interrupted embryonic development of the Mullerian (paramesonephric) ducts leading to hypoplasia of the uterus and the upper two-thirds of the vagina.⁽¹⁾ Incidence is 1 in 5000 female live births.⁽²⁾ Patients have a normal female karyotype, external genitalia, secondary sexual development and normal ovaries and fallopian tubes. There may be associated renal and skeletal anomalies.⁽³⁾

CASE REPORT

A 19-year-old female patient with a history of primary amenorrhoea was referred to the Department of Radiology for an ultrasound examination of abdomen and pelvis. On physical examination, bilateral breasts were found to be absent. (Tanner Scale 1) and absent of axillary and pubic hair. Ultrasound abdomen and pelvis revealed absent uterus. Colour Doppler was used to identify the internal iliac arteries and thus to locate the ovaries. Ovaries were found to be absent bilaterally. Both the kidneys were normal in size and location [Figure 1a & b].

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MRI abdomen and pelvis was performed, which confirmed the findings of ultrasound. Uterus and ovaries were absent [Figure 2a, b, c]. There were no vertebral anomalies. Both the kidneys were normal in location, size and contour [Figure 3].



Figure 1a

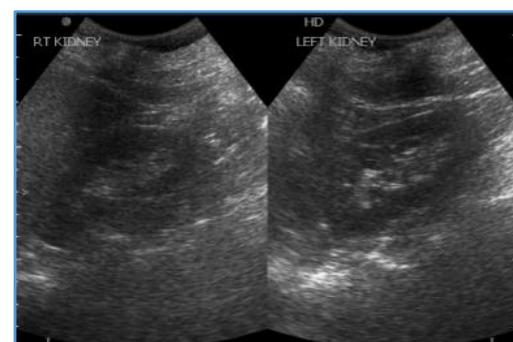


Figure 1b

Figure 1a & b. Ultrasound Images revealing Absent Uterus and Ovaries. Bilateral Kidneys were Normal in Position, Size and Contour



Figure 2a



Figure 2b

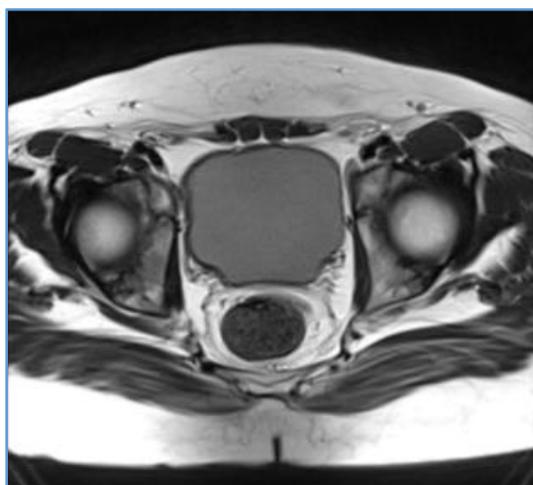


Figure 2c

Figure 2a, b & c. T1W1 Coronal, T2WI Axial and Sagittal Images showing Bladder and Bowel Loops (Arrow) in the Pelvis. Uterus and Ovaries are Absent

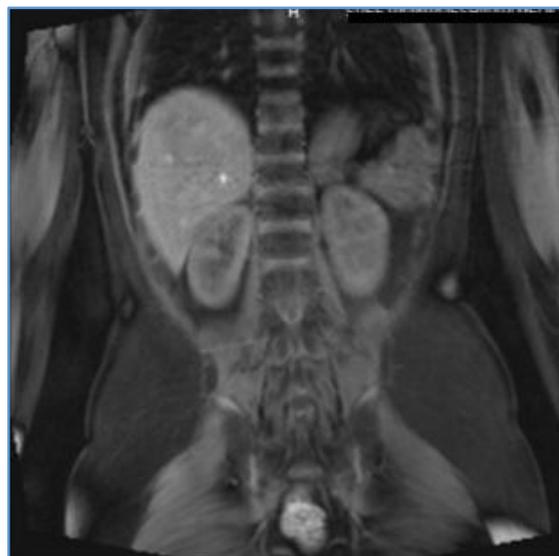


Figure 3. Both the Kidneys and Visualised Vertebrae are Normal

DISCUSSION

Mayer-Rokitansky-Kuster-Hauser syndrome is named after August Franz Joseph Karl Mayer, Carl Freiherr von Rokitansky, Hermann Küster, and G. A. Hauser and is an important aetiological factor in about 15% of cases of primary amenorrhoea. It has a reported incidence of one in 5000 female live births.

Fallopian tubes, uterus, cervix and upper three fourth of vagina develop from Mullerian ducts between 8th to 12th gestational week. Developmental defect occurring at this stage leads to agenesis of Mullerian structures.⁽³⁾

The development of kidneys, ureter and bladder occurs concomitantly at 6th - 12th week. Hence, renal anomalies such as renal agenesis, ectopic kidney, fused kidney, renal hypoplasia and horseshoe kidney are seen in 30% - 40% of patients of Mayer-Rokitansky-Kuster-Hauser syndrome (MRKH). Vertebral abnormalities are also found in about 10% of patients.

There are two different form of this Syndrome

- The typical form (type A) of this syndrome is characterised by the congenital absence of the uterus and upper vagina with normal ovaries and fallopian tubes.
- The atypical form (type B) of the syndrome includes associated abnormalities of the ovaries and fallopian tubes and renal anomalies.⁽¹⁾

The patient may present with primary amenorrhoea and cyclic abdominal pain. If the ovarian function is normal then the patient attains puberty, but does not attain menarche. Although, the ovaries function normally, the fallopian tubes may be closed and the uterus is often anomalous. The patient may present with inability to have intercourse. The degree of vaginal aplasia can vary from complete absence to a blind pouch. The more shallow the canal, the greater the likelihood of the patient having dyspareunia.

Although, the diagnosis of MRKHS can be easily established clinically, the confirmation of the diagnosis and detection of associated abnormalities is possible by laparoscopy, imaging and karyotyping.⁽¹⁾ Ultrasound should be the first choice in evaluating MRKHS and can provide information about associated renal malformation.

However, it is an operator-dependent imaging modality that may generate conflicting results when performed several times.⁽⁴⁾ In addition, US may not always allow for the recognition of Müllerian buds and ovaries in extrapelvic location, which is an essential information for defining the best surgical treatment.⁽⁵⁾ Thus, differentiation between type A and type B may not always be possible using US. Although, Computed Tomography may give information about congenital anomalies, it is not routinely performed due to the risk of ionising radiation.⁽⁶⁾

Magnetic Resonance Imaging with its multiplanar capability, undoubtedly provides detailed pelvic anatomy and has high accuracy in demonstrating the cavitation of the uterus, presence of a cervix and extrapelvic location of the ovaries.⁽³⁾ Magnetic Resonance Urography has high accuracy in demonstrating the kidneys, ureters and urinary bladder.

Plain radiographs should be done to exclude vertebral anomalies.

Laparoscopy is useful in confirmation and classification of MRKH and helps in planning the definitive reconstructive surgery.⁽⁷⁾

CONCLUSION

MRKH syndrome is rare with an incidence of 1 in 5000 live births. We chose to present this case as absent ovaries and breasts with normal kidneys and no associated vertebral anomalies makes the condition rarer still.

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