

Case Report of Mayer-Rokitansky Kuster-Hauser Syndrome

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Abstract: 18yrs old female was brought by her mother with chief complaint of primary amenorrhea. USG was done. Uterus and cervix were not visualized. Right ovary was found in right inguinal region and left ovary in left inguinal region and left iliac fossa. Absence of left kidney was noted with normal right kidney. Later MRI was done and it revealed absence of uterus, cervix & vagina. Other findings were similar to that of USG. All the above features are suggestive of Mullerian duct anomaly-likely Mayer-Rokitansky-Küster-Hauser syndrome.

1. Introduction

Mayer-Rokitansky-Küster-Hauser syndrome (MRKH), is a malformation of the female genital tract, characterized by vaginal atresia and a variety of uterine anomalies. Patients have a normal karyotype and normally developed secondary sexual characters. Associated renal and skeletal abnormalities may be present. It is one of the causes of primary amenorrhea and affects at least 1 of 4500 women.

2. Case Report

A 18 year old female presented to gynaecologist with complaints of suspected primary amenorrhea. Patient did not have any history of cyclical abdominal pain or menstrual bleeding. She had well developed secondary sexual characters.

On sonographic examination of abdomen and pelvis, uterus and cervix were not found. Left kidney was not visualized in left renal fossa and to the extent abdomen and pelvis was visualized. Right ovary was noted in right inguinal region and left ovary in left inguinal region. Multiple small follicles seen in both ovaries.

Serial axial, coronal and sagittal T1WI, T2WI and SPAIR images revealed complete absence of uterus, cervix, vagina, left kidney and ureters. Right ovary was noted in right inguinal canal and left ovary in left inguinal and iliac fossa region. Vaginal stripe was not demonstrable. Scoliosis of lower thoracic spine with curvature towards left.

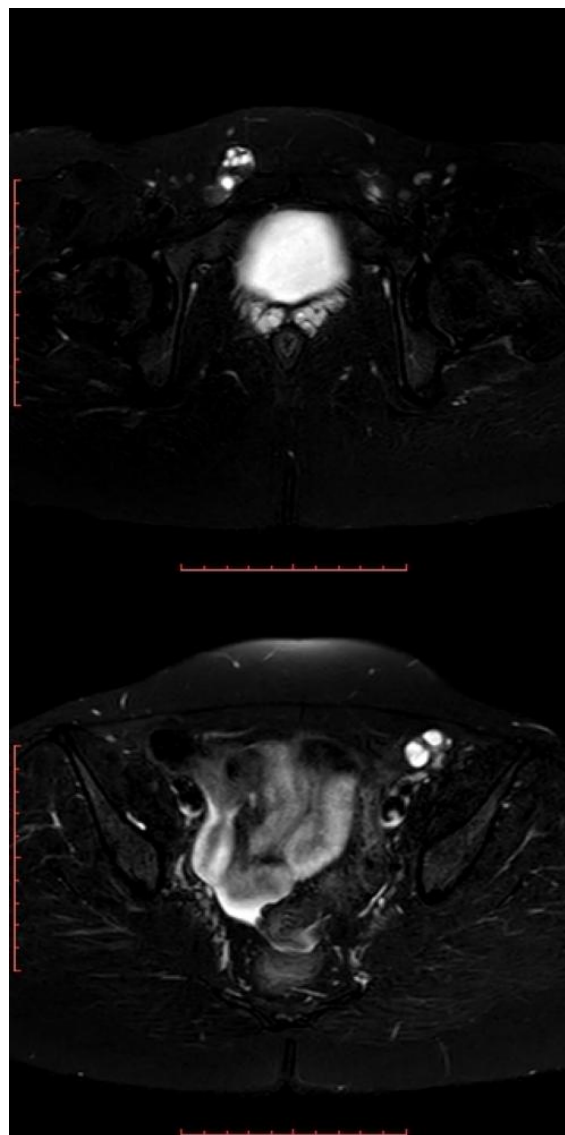


Figure A and B: SPAIR axial images right ovary in right inguinal canal and left ovary in left iliac fossa and left inguinal region.



Figure C: Sagittal T2 images reveal rectum and anal canal immediately posterior to urinary bladder and urethra

Figure D: Axial T2 image at the level of hip joints show rectum immediately posterior to lower urinary bladder with no evidence of vagina in between

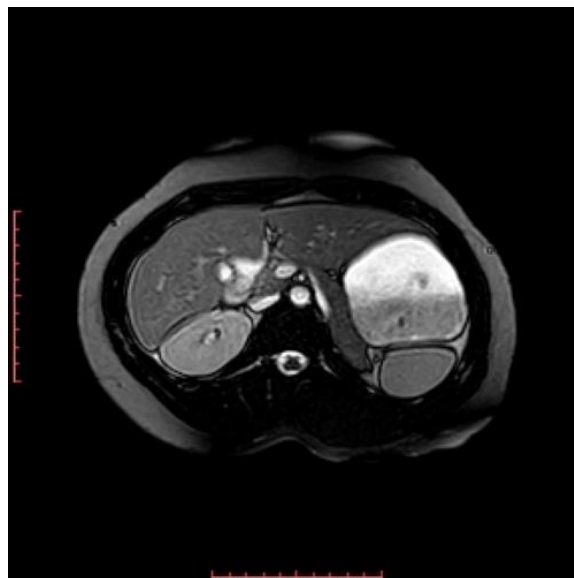


Figure E: Coronal SPAIR images show absence of left kidney in left renal fossa. Associated scoliosis of dorsolumbar spine noted. **Figure E:** Gradient axial images show reveal absence of left kidney in left renal fossa

3. Discussion

Fallopian tubes, uterus, cervix and upper three-fourth of vagina develop from paraesonephric (Mullerian) ducts between 8th-12th gestation weeks. Developmental defect at this stage leads to agenesis of Mullerian structures.

Renal anomalies are seen in 30 - 40 % of patients of Mayer Rokitansky Kuster Hauser syndrome (MRKH). 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 associated with Mullerian agenesis.

Vertebral abnormalities are also found in 10 % of patients. Other rare associations are cardiac anomalies and anorectal malformations.

Patient always present with primary amenorrhea. Patient usually undergoes normal puberty as ovaries functions are always preserved. Other symptoms that may be present are cyclic abdominal pain, inability to have intercourse. Degree of vaginal aplasia may vary from complete absence to blind pouch. Shallow the vaginal canal, more likelihood of the patient to have dyspareunia. Some patients may present with voiding abnormalities .

MRKH syndrome	Associated malformations
Typical	Tubes, ovaries, and renal system generated and developed
Atypical	Malformations in the ovary or renal system
MURCS	Malformations in the skeleton and/or heart; muscular weakness, renal malformations

Classification of the Mayer–Rokitansky–Kuester–Hauser (MRKH) syndrome according to Schmid-Tannwald and Hauser (1977) and Duncan *et al.* (1979)

MURCS = Müllerian aplasia, renal aplasia, and cervicothoracic somite dysplasia (association).

References

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