CASE REPORT

ANORECTAL MALFORMATION AND RENAL DEFECT ASSOCIATED WITH MAYER-ROKITANSKY-KUSTER-HAUSER SYNDROME, MRI FEATURES: A RARE CASE REPORT

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ABSTRACT

Mayer-rokitansky-kuster-hauser syndrome is a rare congenital syndrome occurring in approximately 1out of 4500 to 1 out of 5000 women.[4,7] There is a defect in the embryonic development of para mesonephric ducts leading to aplasia or hypoplasia of the uterus and proximal 2/3 of vagina, despite normal hormonal status and female karyotype.[3,4] It is divided into three types:

Type 1 an isolated deformity of the uterus and cervix

Type 2 is associated with renal and ovarian disorders (atypical form)

Type 3 shows anomalies including anal atresia, tracheoesophageal fistula, renal, cardiac and vertebral deformities.[1,3,4,7] Typical MRI findings of this disorder can be helpful in the diagnosis of this syndrome. Therefore, we have presented here a case of an 8-year-old who was diagnosed with Mayer-rokitansky-kuster-hauser syndrome and her MRI pelvis findings along with other investigations.

Keywords: Mayer-Rokitansky-Küster-Hauser syndrome, uterine hypoplasia, congenital disorder, MRI findings.

Introduction

Mayer-rokitansky-kuster-hauser syndrome (MRKH) being the second most common cause of primary amenorrhea is a rare congenital disorder affecting the development of Mullerian duct during embryogenesis.[1-9] German anatomist Mayer was the first person to detect this abnormality back in 1829. Later Rokitansky and Kuster also contributed to the diagnosis of MRKH by identifying various diagnostic features.[2] Patients have a normal hormonal status, karyotype (46, XX) and normal secondary sexual characteristics.[1,2] The disease is present in approximately 1 in 4500 births.[1-9]

The etiology remains unidentified. It was previously considered a sporadic disorder, associated with diethylstilbestrol (DES) and thalidomide use during pregnancy,[1] however recently genetic involvement has been reported.[2] The most striking finding is the agenesis or aplasia of female reproductive organs especially uterus and upper third of vagina.[1,2,3,4] Many cases have reported presence of normal ovaries or physiologically normal ectopic ovaries.[2,5] Based on organ involvement there are three types, type 1 has isolated involvement of uterus and vagina. Type 2 has renal and ovarian involvement whereas type 3 has other associated malformations, renal vascular and skeletal along with mullerian agenesis.[1,2,3] The associated anomalies result in presentation of the disease earlier, MR imaging is however important in the diagnosis of such patients to identify the anatomical details and intern help in further management of the patients. Unfortunately, not enough work has been done to support MRI findings in such patients despite it being accepted as a useful modality.[5]

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Surgical intervention and assisted conception can help the patients take a step towards normal life, thus helping with the impact of disease on fertility and mental health of the patient.[3] Here we have discussed a case of Mayer-Rokitansky-Küster-Hauser syndrome type 2 in an 8-year-old girl who presented to us with the complaint of urinary incontinence since birth and history of PSARP.

Case Report

According to the mother she was born full term through SVD.She did not pass meconium for 3 days after birth and also develop vomiting's. Her initial workup was done in ER and she was diagnosed with a common cloaca, for which emergency colostomy was done initially. Later on she was referred to the radiology department for further workup with IVU and ultrasound abdomen. On intravenous pyelography there was an ectopic left pelvic kidney and ureter (ureteric opening below the neck of urinary bladder) with hydronephrosis and hydroureter, there is non visualization of right kidney on IVU. Ultrasound shows congenitally absent right kidney and ectopic left pelvic kidney. Uterus is also not visualized and MRI was recommended for genital tract evaluation.

MRI pelvis of the patient was done which showed complete absence of uterus, vagina and cervix. Bilateral ovaries were present and were normal. Right kidney was absent, left kidney was ectopic and was lying in the left iliac fossa. It was malrotated antero-medially with gross hydronephrosis and hydroureter. Pull through surgery was noted involving the urinary bladder and anal canal with central coning. Findings are suggestive of MRKH Type 2.

Her cystoscopy and ureteric re-implantation with DJ stent placement were performed later on. Patient underwent uneventful recovery. Her findings and implications regarding the disease were explained to the family.

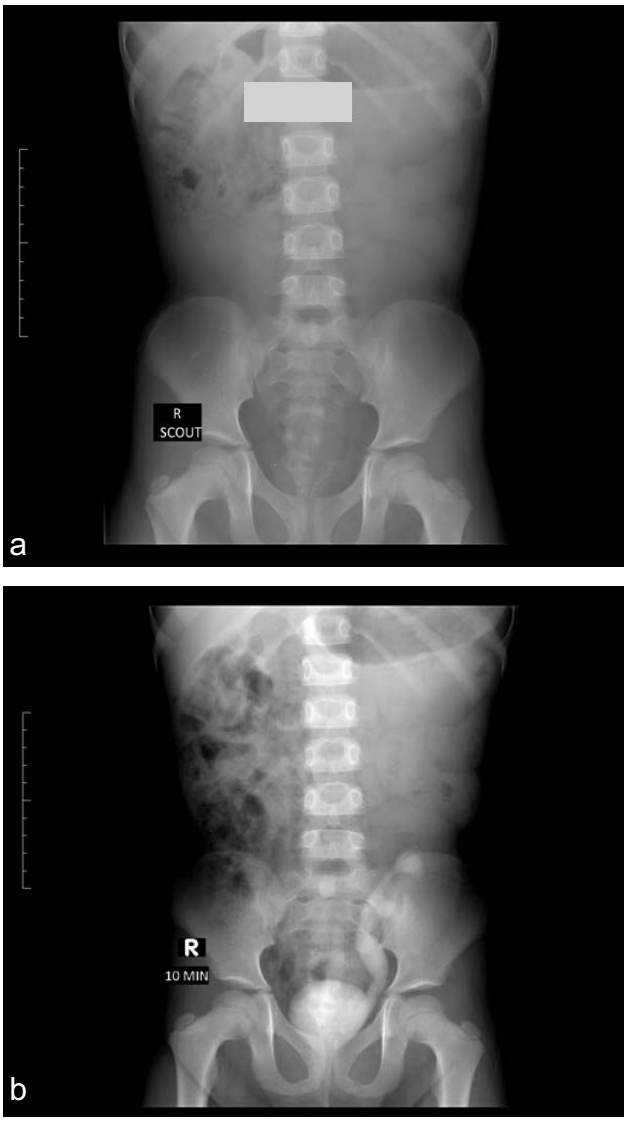
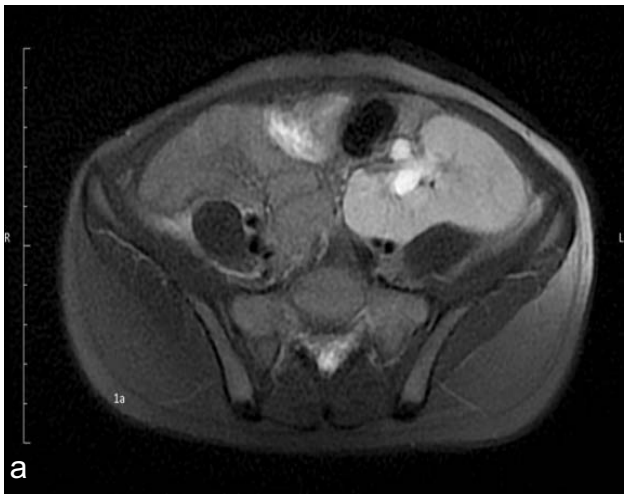
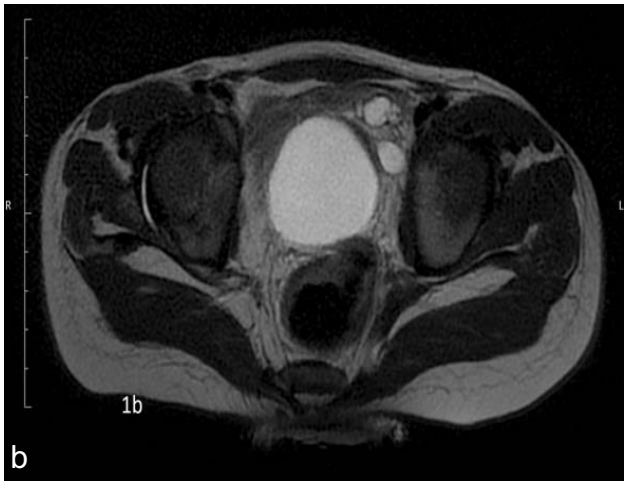


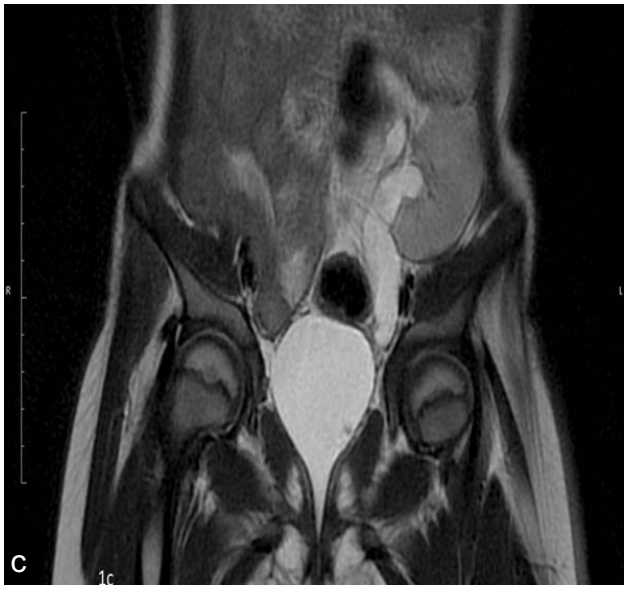
Figure 1: an IVU of the patient showing **A.** scout film of the patient, **B.** showing post contrast images with ectopic left kidney lying in the left pelvis with hydronephrosis and hydroureter. There is non visualization of the right kidney.

Discussion

Mayer-Rokitansky-Küster-Hauser syndrome is a rare congenital developmental defect, interruption in the Müllerian duct development causing agenesis of the female reproductive organs.[1-3] Being a very rare entity, it is present in only 1 in 4500 births.[1,2,3,4] Some researchers have shown a genetic involvement[2] while others believe in a sporadic origin of the disease, some studies have shown an association with die-thylstil bestrol (DES) and thalidomide use during







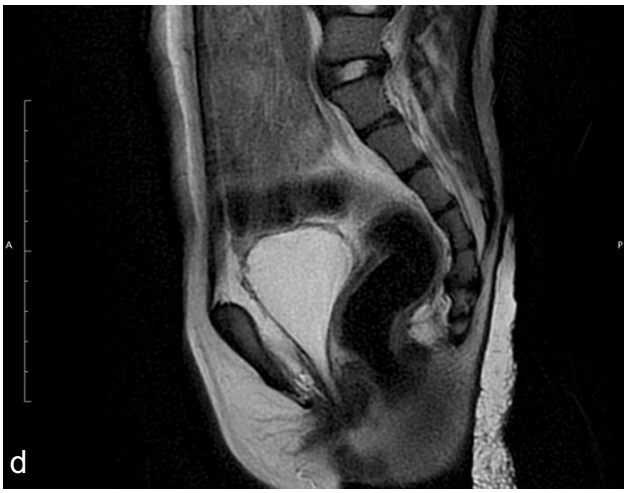


Figure 2: MRI images showing A & B Axial Cuts, C Coronal cuts & D Sagittal: **A** shows left kidney in left iliac fossa with hydronephrosis right kidney is not visualized. **B** shows absent uterine cavity and normal ovaries. **C** shows absence of right kidney and ectopic hydronephrotic left kidney. Left ureter also dilated. **D** sagittal section of the same patient showing complete absence of uterus, cervix and vagina.

pregnancy.[1] However, there is no clear association as yet. The key findingof MRKH are absence or aplasia of uterus and vagina, normal female karyotype, normal ovaries and external genitalia.[3] MRKH is divided into 3 basic types MRKH type 1 is an isolated defect involving the female reproductive organs only, including the uterus and vagina. Type two and three are the least common and have associated renal and ovarian anomalies and renal, vascular and skeletal anomalies respectively.[1,3]

Clinically, the patients with MRKH present most commonly with primary amenorrhea, however, in type 2 and 3 there is earlier presentation with renal, skeletal (backache due to scoliosis or limb deformities) and GI symptoms (inability to pass stools due to anal atresia).[1]

The first imaging investigation that should be carried out is ultrasonography which will demonstrate the absence of or rudimentary uterus. However, the presence of normal ovaries. The ovaries may be ectopic in some cases. The associated renal anomalies can also be appreciated on an ultrasound. The investigation of choice however remains MR imaging. Which can clearly demonstrate the major findings, in case of any confusion on an ultrasound examination.[2,5,9]

Typical MRI findings in such cases are the absence of uterus, and upper third of vagina. With the presence

of normal ovaries. In type 2 and 3 there will also be absence of or ectopic kidney, scoliosis and esophageal anomalies. The MRI findings can then be correlated with the surgical findings.[1]

Conclusion

To sum up MRKH is a rare congenital anomaly that may present with symptoms other than amenorrhea due to its extra uterine involvement. Hence, it should be kept as a differential in female infants presenting with renal, esophageal, skeletal or cardiac anomalies. MR imaging in such cases can be of use to rule out the uterine and extra uterine involvement.

Conflict of Interest: None to declare.

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