Section: Radiodiagnosis

Case Series

MAGNETIC RESONANCE IMAGING IN MAYER-ROKITANSKY-KUSTER-HAUSER SYNDROME: A CASE SERIES

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ABSTRACT

The Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is characterized by congenital aplasia of the uterus and the upper part (2/3) of the vagina in women due to failure of formation or failure of fusion of the mullerian ducts. The first sign of MRKH syndrome is a primary amenorrhea in young women presenting otherwise with normal development of secondary sexual characteristics and normal external genitalia, with normal and functional ovaries, and karyotype 46, XX without visible chromosomal anomaly. The phenotypic manifestations of MRKH syndrome overlap with various other syndromes or associations and thus require accurate delineation. For a long time, the syndrome has been considered as a sporadic anomaly, but increasing number of familial cases now support the hypothesis of a genetic cause. In familial cases, the syndrome appears to be transmitted as an autosomal dominant trait with incomplete penetrance and variable expressivity. The reported prevalence of the condition is 1 in 5000 females and it is the second most common cause of primary amenorrhea after gonadal dysgenesis.[1] .First described by the German anatomist and physiologist Mayer in 1829, reported by Rokitansky (1838), Küster (1910), and Hauser (1961), later named MRKH syndrome.[2] Ultrasound is the first imaging investigation in cases of suspected mullerian anomalies while MRI is the key imaging modality of choice due to its capabilities of demonstrating the female genital tract remarkably well and ability to provide the details of intra-uterine anatomy, external fundal contour and its ability of imaging of entire female pelvis into multiple imaging planes. Also, it provides additional information about the other associated anomalies particularly those of urinary system and helps in differentiating other causes of amenorrhea. As psychological distress is very important in young women with MRKH, it is essential for the patients and their families to attend counselling before and throughout treatment.

INTRODUCTION

The Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is characterized by congenital aplasia of the uterus and the upper part (2/3) of the vagina in women due to failure of formation or failure of fusion of the mullerian ducts. The reported prevalence of the condition is 1 in 5000 females and it is the second most common cause of primary amenorrhea after gonadal dysgenesis.[1] First described by the German anatomist and physiologist Mayer in 1829, reported by Rokitansky (1838), Küster (1910), and Hauser (1961), later named MRKH syndrome.[2] MRKH syndrome (class I Mullerian duct anomaly) is a rare congenital disorder characterised by uterine and cervical aplasia/hypoplasia with normally functioning ovaries in genotypical normal female (46XX) due to failed/interrupted development of the mullerian duct.[3,4] Patients usually present with normal secondary sexual characteristics and primary amenorrhea.[5] The diagnosis of MRKH has grave implications on the psychological and reproductive outcomes of the patient. MRKH can be divided into type 1 and type 2. MRKH type 1 is also known as isolated Mullerian aplasia or Rokitansky sequence. This type has symptoms that vary from one individual to another. In many cases, the uterus and vagina are underdeveloped (aplasia), and in more severe cases there may be atresia of the upper portion of the vagina and underdeveloped uterus.[6] The fallopian tubes can also be affected by their normal function. In type 2 it is also called Mullerian duct aplasia, Renal dysplasia, and Cervical Somite anomalies or also known as the MURCS association. Typical symptoms of type 2 MRKH are developmental disorders of the kidneys and

malformations in the bones, especially in the vertebrae. Malformations of the heart and hearing loss can also occur but are very rare.[7]

MATERIALS AND METHODS

Patients presenting with history of primary amenorrhea who came for MRI in the department of Radio-diagnosis, JNIMS, Imphal after gynecological examination referral from Gunae OPD were taken for the study. Patients were imaged using Phillips Achieva 3 Tesla MR system and following imaging sequences were performed T1 AXIAL, CORONAL, SAGITTAL; T2 AXIAL, CORONAL & FAT SUPPRESSED T2 WEIGHTED MULTIPLANAR IMAGING of pelvis and T2 HASTE CORONAL AND AXIAL of abdomen. MR images were obtained for all patients using a pelvic phased-array coil. All individuals were examined in supine position. MR images were obtained from the aortic bifurcation to the symphysis pubis. Transabdominal ultrasound (Phillips) was done for each patients following MRI and detailed clinical history was taken. Further, patients were asked to perform the laboratory work-up and called upon along with hormonal/laboratory test reports to collect the MRI reports.

Imaging analysis was done based on:

1. Presence or absence of uterus, cervix vagina with description of morphology and imaging characteristics
2. Presence or absence of midline or paramedian triangular soft tissue structure
3. Presence of absence of uterine buds, laterality, location, evidence of cavitation
4. Presence or absence of fibrous bandlike structure extending between the uterine buds (if present)
5. Presence or absence of ovaries, location and morphology
6. Associated non gynaecological anomalies.

RESULTS

Case 1: An 18-year-old girl, c/o primary amenorrhea

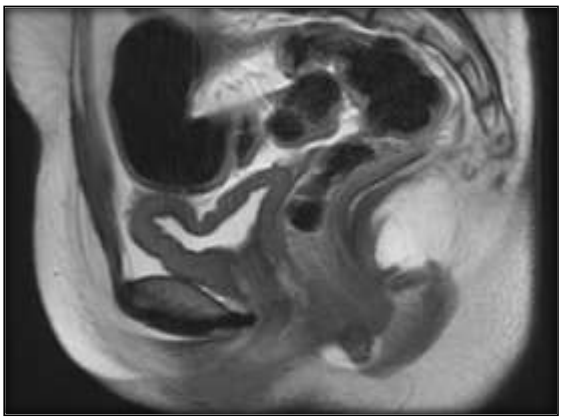

Figure 1: MRI images showing absence of uterus and upper part of vagina suggestive of complete agenesis of uterus and 2/3rd of vagina however both ovaries were normal

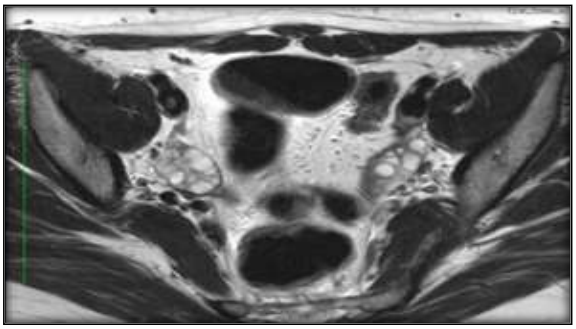

Figure 2: MRI images showing absence of uterus and upper part of vagina suggestive of complete agenesis of uterus and 2/3rd of vagina however both ovaries were normal

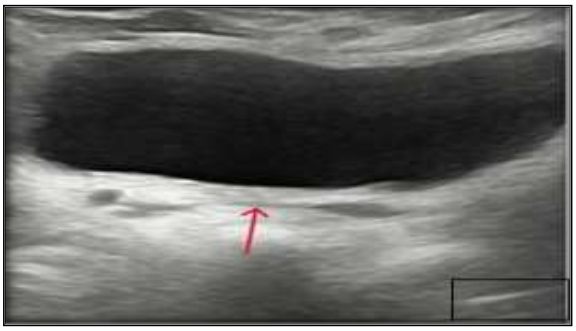

Figure 3: shows Ultrasound images of absent uterus.

Based on the MRI findings, a diagnosis of Myer-Rokitansky-Kuster-Hauser Syndrome was made.

Case 2: A 20-year-old girl, c/o primary amenorrhea

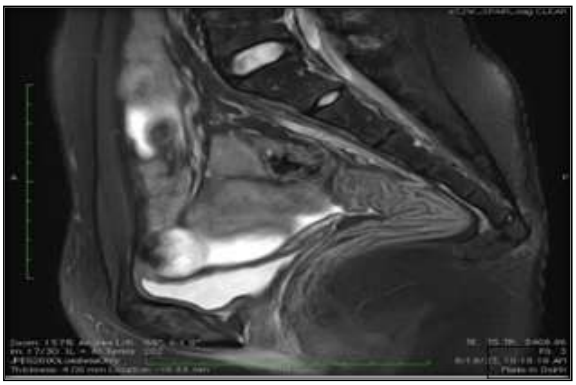

Figure 4: T2 weighted sagittal image of pelvis shows absence of normal morphology of uterus and cervix.

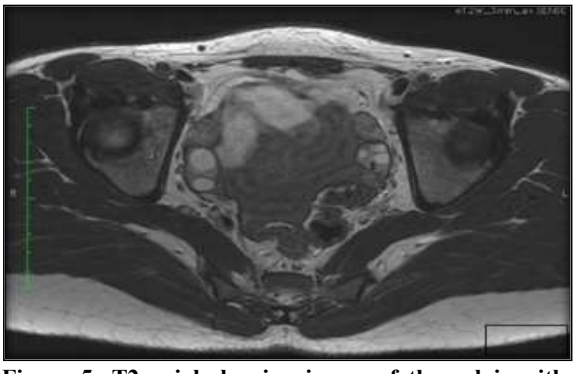

Figure 5: T2 axial showing image of the pelvis with normal location of bilateral ovaries

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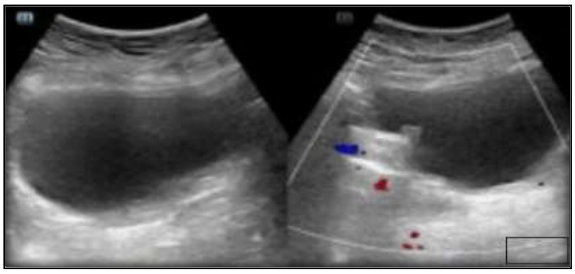

Figure 6: Ultrasound Color Doppler showing no evidence of uterus

Case 3: A 21-year-old girl, c/o primary amenorrhea

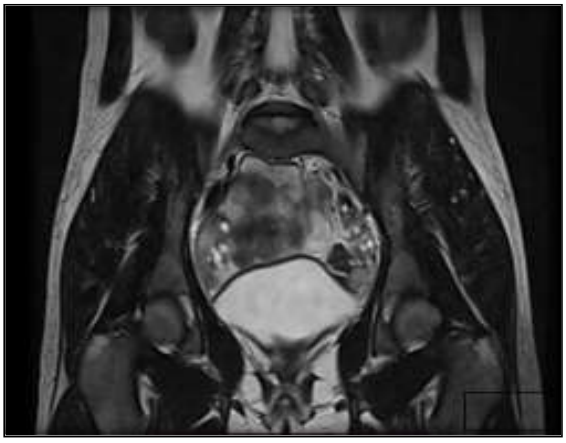

Figure 7: MRI images of absent uterus and upper part of vagina

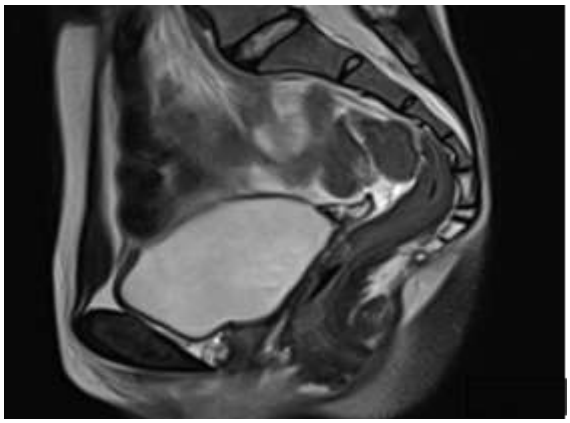

Figure 8: MRI images of absent uterus and upper part of vagina

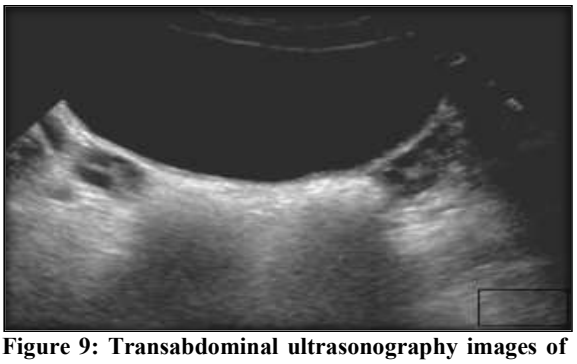

Figure 9: Transabdominal ultrasonography images of the same patient which shows normal ovaries and presences of inferior third of vagina. The image shows absence of the uterus.

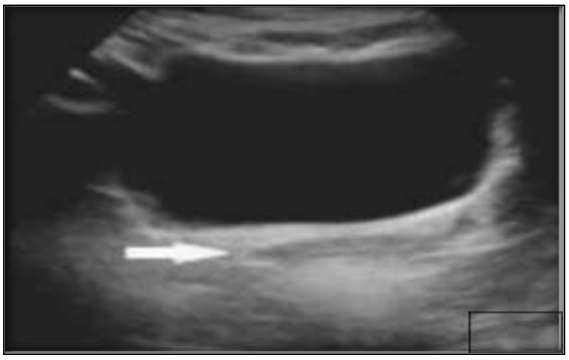

Figure 10: Transabdominal ultrasonography images of the same patient which shows normal ovaries and presences of inferior third of vagina. The image shows absence of the uterus.

Case 4: A 16-year-old girl present with primary amenorrhea

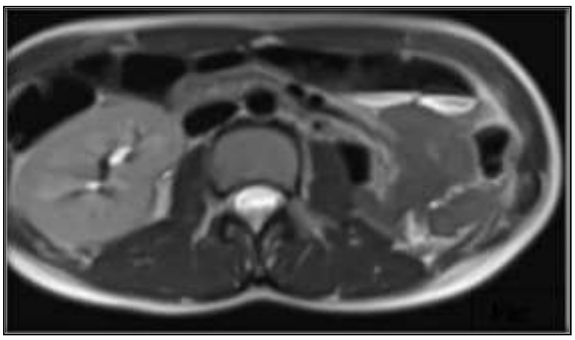

Figure 11: Axial T2 MRI showing absent left kidney.

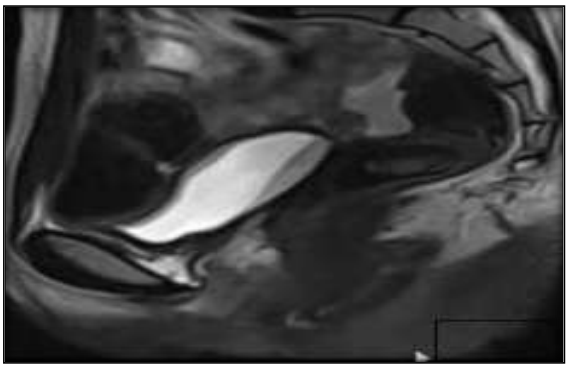

Figure 12: Sagittal T2 MRI showing lower 1/3 of vagina with absent uterus at uterine fossa.

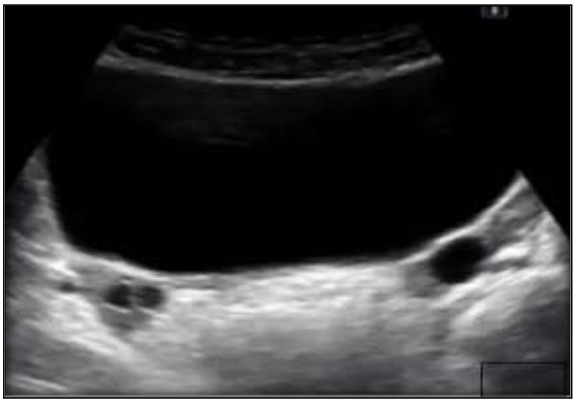

Figure 13: showing ultrasound images of normal bilateral ovaries with follicles

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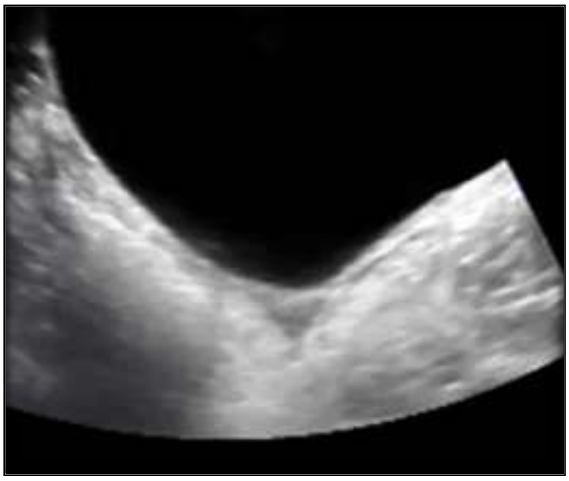


Figure 14: showing rudimentary uterus in the uterine fossa where failure of normal uterine development is seen.

Summary findings:

- Unremarkable medical, surgical and social history
- No history of congenital anomalies in three cases. One had absent left kidney. Unremarkable obstetric history
- No hormone therapy or radiation. Average height and weight
- Normal secondary sexual characteristics. Trans-abdominal USG – absent uterus. Normal bilateral kidneys in three cases. No urinary tract anomalies in the three cases.
- No skeletal malformation
- Serum FSH and LH levels were within normal limits
- Based on the findings, all the above three cases were diagnosed as Type 1 MRKH syndrome and only one was graded as MRKH Type 2.

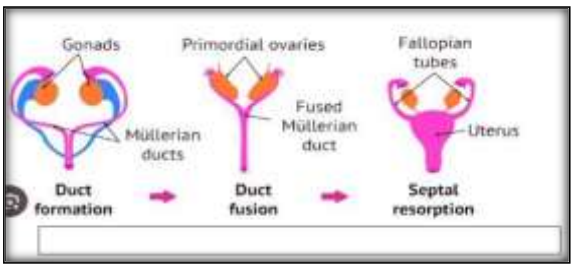
Table 1: Characteristic, description and case study of the patients

Sl. No.	Characteristics	Case 1	Case 2	Case 3	Case 4
1	Uterus, cervix & upper part of vagina	Absent	Absent	Absent	Rudimentary uterus
2	Ovaries, location	Normal location & morphology	Normal morphology	Normal in location	Normal with follicles
3	Associated non-gynecological anomalies	None	None	None	Absent left kidney
4	Diagnosis	MRKH Type 1	MRKH Type 1	MRKH Type 1	MRKH Type 2

DISCUSSION

Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is a rare congenital disorder characterized by partial or complete agenesis of the uterus and upper two-thirds of the vagina, while the external genitalia, ovarian function, and secondary sexual characteristics remain intact. MRKH syndrome is typically diagnosed in adolescence, when patients present with primary amenorrhea, as in the four cases described in this series. The diagnosis of MRKH syndrome is typically made through imaging studies. Pelvic ultrasonography is usually the first modality to reveal the absence of the uterus and upper vagina, as well as, we can scan for kidneys morphology and anomalies though it may not provide sufficient detail regarding the full extent of the malformation. MRI is the gold standard in confirming the diagnosis, as it provides information about the uterus and vaginal anatomy and can assess for associated renal and skeletal anomalies.

In our series, all the patients were diagnosed with MRKH through a combination of pelvic ultrasound and MRI. Which revealed the characteristic findings. Primary amenorrhea affects about 5% of the female population. It is diagnosed in girls with no pubertal development at an average age of 14 years and slightly later in those with normal development of secondary sexual characteristics at around the age of 16 years.[8] MRKH syndrome (Type 1 Mullerian duct anomalies) is the most common cause of primary amenorrhea, even though it is a rare disorder. The MRKH syndrome has a significant influence on both fertility and psychological health of women. Hence, it is essential to diagnose and accurately visualize the anatomical detail to allow clinical and psychological inputs to patients. Surgery is necessary for restoration of normal sexual function. Even reproduction may be possible if assisted reproductive techniques are performed.[9] Urological anomalies associated with Type II MRKH syndrome are renal ectopia, horse-shoe kidney and rarely renal agenesis. Varying degrees of musculoskeletal anomalies have been noted ranging from vertebral segmentation anomalies, scoliosis to abnormalities of radius, carpals, phalanges and femoral capital epiphyses. Ovarian cancers,[10,11] and cardiac malformations,[12] have been reported with Type II MRKH syndrome. The Mullerian (paramesonephric) ducts form the uterus, cervix, upper two-third of vagina and fallopian tubes.[14]



The Mullerian duct remnants described are a triangular, midline soft tissue structure lying above the dome of the urinary bladder and fibrous bands extending from this structure to the rudimentary uterine horns.[15]

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The levels of FSH and LH are normal with no sign of androgen excess which can be differentiate from androgen insensitive syndrome.[16] The MRKH syndrome should be differentiated from androgen insensitivity syndrome, and isolated vaginal hypoplasia or atresia. In androgen insensitive syndrome, end organ resistance to androgen resulting in virilisation of external genitalia resulting in female phenotype of baby with development of female secondary sexual characteristics and genotypically male (46XY) with undescended testes which MRI can differentiate from MRKH syndrome.[17] USG may not always detect the uterine buds or ovaries even in ectopic location, it can falsely detect rectovesical quadrangular structure as hypoplastic uterus. Information about rudimentary buds is essential before surgical treatment.[18] MRI detects absence of uterus and ovaries with presence of rudimentary ectopic testis in androgen insensitive syndrome.[19] Multiplanar capability of MRI can be exploited effectively to detect and characterize various anomalies in such patients for example the uterine aplasia is best detected on sagittal plane, whereas the vaginal atresia can be better diagnosed on axial plane.[20] Sometimes the rudimentary uterine buds may show cavitation which may be the reason for cyclical pain and cause endometriosis, which on identification can be surgically removed. The presence of cavitation is evidenced as central zone of high signal intensity, a middle layer of low signal intensity and an outer zone of intermediate signal intensity.[21]

CONCLUSION

We report three rare cases of type 1 MRKH syndrome who presented with primary amenorrhea. The classic features of utero-vaginal agenesis and bilateral normal ovaries were well shown. No associated renal or musculoskeletal anomalies were present. In one case we got absent left kidney with tiny rudimentary uterus with normal ovaries. MRI is now considered the imaging modality of choice, because of its ability to accurately identify female genital tract malformations along with associated renal and skeletal anomalies. The MRI findings in conjunction with the physical examination were sufficient for diagnosis. In our study, we report three cases of type 1 Mullerian duct anomalies while we could detect one case of type 2 Mullerian duct anomaly suggested that type 1 mullerian duct anomaly is more common than type 2.

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