ORIGINAL ARTICLE

MAYER-ROKITANSKY-KÜSTER-HAUSER SYNDROME: MR MANIFESTATIONS OF TYPICAL AND ATYPICAL CASES

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Background: The Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a group of congenital malformations arising due to agenesis or arrest of the growth of paired Mullerian ducts leading to absence of uterus, fallopian tubes and upper part of vagina. MRI has an unmatched role in diagnosing MRKH syndrome. The aim of the present study is to elaborate MRI manifestations of typical and atypical MRKH syndrome and its common and rare associations. **Methods:** It was a cross sectional analysis of 19 cases in the Radiology Department of a public sector tertiary health care hospital of Rawalpindi over a period of one year. All patients clinically diagnosed as MRKH syndrome were included in the study. MRI pelvis of the study patients was performed and evaluated by two experienced radiologists. **Results:** The mean age of study patients was 22.2 years. Out of 19 study patients, 16 (84%) had type I while 3 (16%) had type II MRKH syndrome. None of the patients had normally cited uterus of normal morphology. Bilateral ovaries with follicular activity were identified in all patients. A well-defined leiomyoma was identified arising from the left uterine bud in one of these patients. All three patients with MRKH II syndrome had ectopic pelvic kidneys. One of the patients showed a neoplastic mass lesion arising from ectopic left kidney. **Conclusion:** MRI is an excellent imaging modality for accurate diagnosis and evaluation of other system anomalies in MRKH syndrome. Rudimentary uterine buds are commonly seen in these patients and may have functioning endometrium. Ovaries are of normal appearance but frequently ectopic.

Keywords: Magnetic resonance imaging; Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome; Mullerian defects; Primary amenorrhea

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INTRODUCTION

The Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a group of congenital malformations arising due to agenesis or arrest of the growth of paired Mullerian ducts leading to absence of uterus as well as fallopian tubes along with upper part of vagina.[1] Lower 1/3rd of vagina and the hymen that do not develop from Mullerian ducts but arise from urogenital sinus are spared and present.[2] It is also called as Müllerian agenesis. Ovaries which do not develop from Mullerian ducts are also spared.[3] Secondary sexual characteristics in these females are normal due to presence of normal functioning ovaries and normal karyotype, i.e., 46XX.[4] This disorder affects one in 4500 women.[5]

Mayer-Rokitansky-Küster-Hauser is of two types; (type I) that occurs in isolation and (type II or MURCS association) which is associated with renal, vertebral, auditory and cardiac anomalies. The main presenting complaint is primary amenorrhea but with normal secondary sexual characters. The exact cause of the anomaly is not known. Previously it has been considered sporadic; however, with the increase in familial cases, it is believed that genetic factors have a major role.

Since Magnetic Resonance Imaging (MRI) excellent imaging modality for visualization of female genital system due to good soft tissue differentiation, multiplanar multi sequential capabilities and lack of ionizing radiation. MRI has capability of identifying absence of normal uterus, vestigial uterine buds, status of ovaries and vagina and small uterine bud remnant. MRI therefore have unmatched role in diagnoses of MRKH syndrome. The aim of the present study is to elaborate MRI manifestations of typical and atypical MRKH syndrome and its common and rare associations.

MATERIAL AND METHODS

It was cross sectional analysis of 19 cases in the Radiology Department of a public sector tertiary health care hospital of Rawalpindi over a period of one year, i.e., 01-01-2019 to 31-12-2019. Patients presenting with complaints of primary amenorrhea and clinically diagnosed as MRKH syndrome on the basis of: Ultrasound evidence of an absent uterus, Normal secondary sex characters, and Normal Follicular Stimulating Hormone (FSH) and Luteinizing Hormone (LH) levels. The study was approved by ethical review board of the institute. After obtaining informed written consent, a questionnaire was administered to the study participants at the time of scan to inquire about brief clinical history and examination findings. Patients were examined for secondary sexual characteristics (axillary

hair and breast development) by the trained investigators before the start of examination. MRI pelvis was done using 1.5 Tesla MRI GE machine. Fast spin echo (FSE) axial T1WI and axial coronal and sagittal T2WI images of pelvis was obtained followed by fat suppressed T1WI coronal and axial images of pelvis. Moreover, axial and coronal T2WI of abdomen were also obtained to examine the kidneys. Two radiologists evaluated each MRI and made note for: Absence/Presence of normally cited uterus, upper third and lower two third of vagina. Rudimentary uterine buds where seen were assessed for their morphology and volumes. Status of bilateral ovaries with their volumes and additional anomalies if any were recorded. In the case of disagreement, the definitive decision was made with consensus. The data was analysed using SPSS 21.0.

RESULTS

During the study period a total of 19 patients of MRKH fulfilling the inclusion criteria were studied. The mean age of study patients was 22.2 years. Out of 19 study patients, 16(84%) had type I while 3(16%) had type II MRKH syndrome. None of the patients had normally cited uterus of normal morphology. (Figure-1)

Vestigial uterine buds were identified in 17 patients (89.4%) and were bilateral in 16 patients. Identified uterine buds were invariably seen in close relation to the ovaries and always seen in caudal relationship to the ovaries. A low signal triangular shaped fibrotic remnant is seen in midline in 13 patients (68.4%) while low signal convergence bands identified in 12 patients (63.1%). Mean volume of the right uterine bud was 2.1ml and volume of left uterine bud was 1.9 ml. A well-defined leiomyoma was identified arising from the left uterine bud in one of these patients. Only in one patient a well differentiated endometrium identified in right uterine bud.

Bilateral ovaries with follicular activity were identified in all the study patients. Mean volume of the right ovary was 4.2ml and of the left ovary was 4.5 ml. In one of the study patients, left ovary along with left uterine bud were seen ectopically cited in the left inguinal ring.

All three patients with MRKH II syndrome had ectopic pelvic kidneys. One of the patients had agenesis of the contralateral kidney and ectopic left kidney showed a neoplastic mass lesion

Table-1: MRI findings (quantitative) in patients of MRKH syndrome

Age (yrs)	Lower third of vagina	Triangular cord sign	Convergence Band	Right uterine bud Volume (ml)	Left uterine bud Volume (ml)	Right ovary Volume (ml)	Left ovary Volume (ml)	Associated/ additional findings	Type of MRKH
30	Present	+	+	0.4	0.9	6	5.8	Left ectopic pelvic kidney	II
36	Present	+	+	3.5	2.0	6.5	6.1	-	I
17	Present	+	-	2.9	1.26	3.5	4.5	Three layered right uterine bud	I
21	Present	-	-	3.5	-	4.1	5.1	-	I
25	Present	-	+	0.6	2	6.0	4.6	Ectopic left uterine bud and left ovary seen at inguinal ring. Right renal agenesis. Left ectopic pelvic kidney with neoplasm, ascites and lymph nodes	II
16	Present	+	+	2.0	3.5	3.2	2.1	-	I
15	Present	+	+	2.5	2.1	2.8	3.5	-	I
22	Present	+	-	-	-	3.5	4.0	-	I
17	Present	+	+	2.9	2.5	3.8	4.5	-	I
26	Present	-	-	0.8	1.25	3.5	5.2	Ectopic pelvic right kidney	II
18	Present	+	+	3.5	0.5	6.5	5.4	-	I
35	Present	+	+	2.5	*	4.0	3.2	*Leiomyoma in the left uterine bud	I
20	Present	+	+	4.5	3.4	3.1	3.8	-	I
23	Present	+	+	0.9	1.2	4.8	5.6	-	I
16	Present	+	+	2.4	2.3	4.1	4.1	-	I
17	Present	-	-	0.8	1.6	2.8	3.9	-	I
24	Present	-	+	3.8	2.8	3.5	4.8	-	I
19	Present	+	-	-	-	3.8	4.2	-	I
29	Present	-	-	2.5	1.9	4.8	5.6	-	I

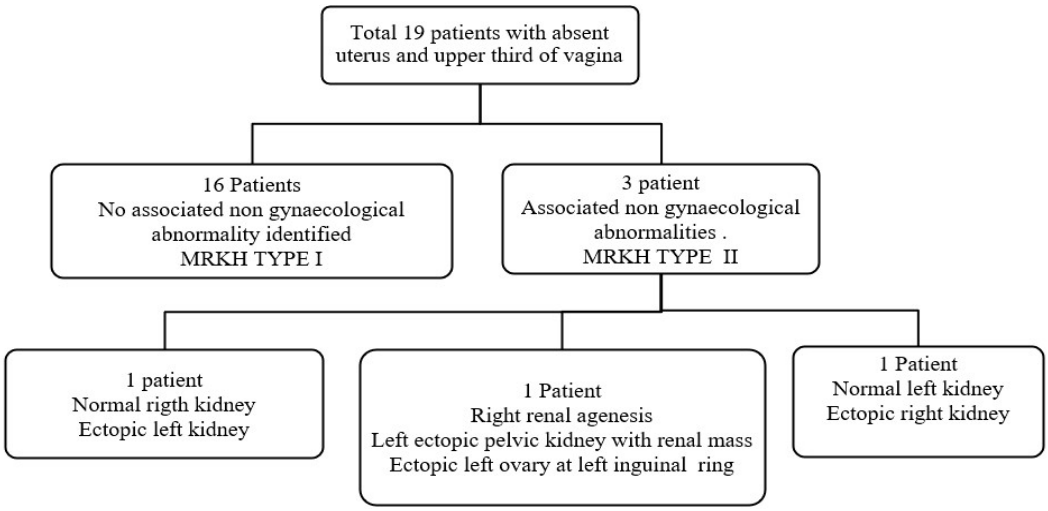

Figure-1: Flow chart showing salient MRI findings of study patients

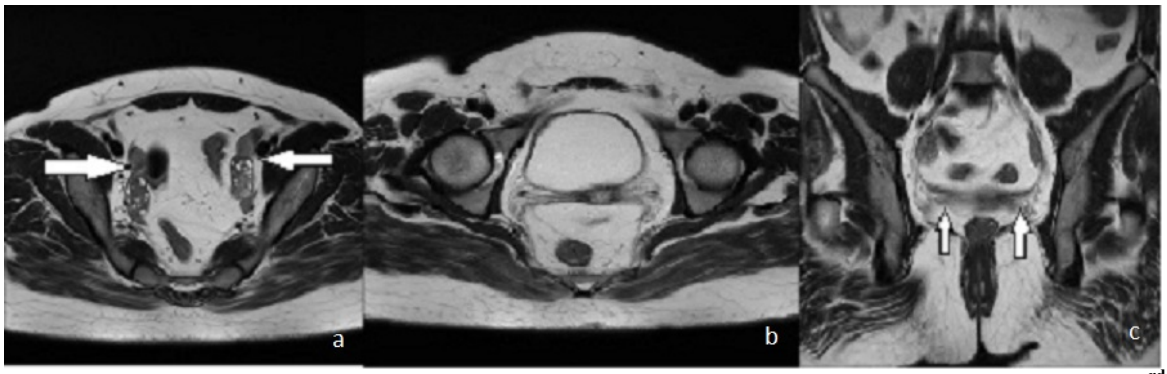

Figure-2: 36 years old female patient with MRKH I (a, b) axial T2WI show absent uterus and upper 2/3rd of vagina with normal sized ovaries showing follicular activity. Arrows show bilateral rudimentary uterine buds closely related to the ovaries. (c) Arrow shows low signal convergence band.

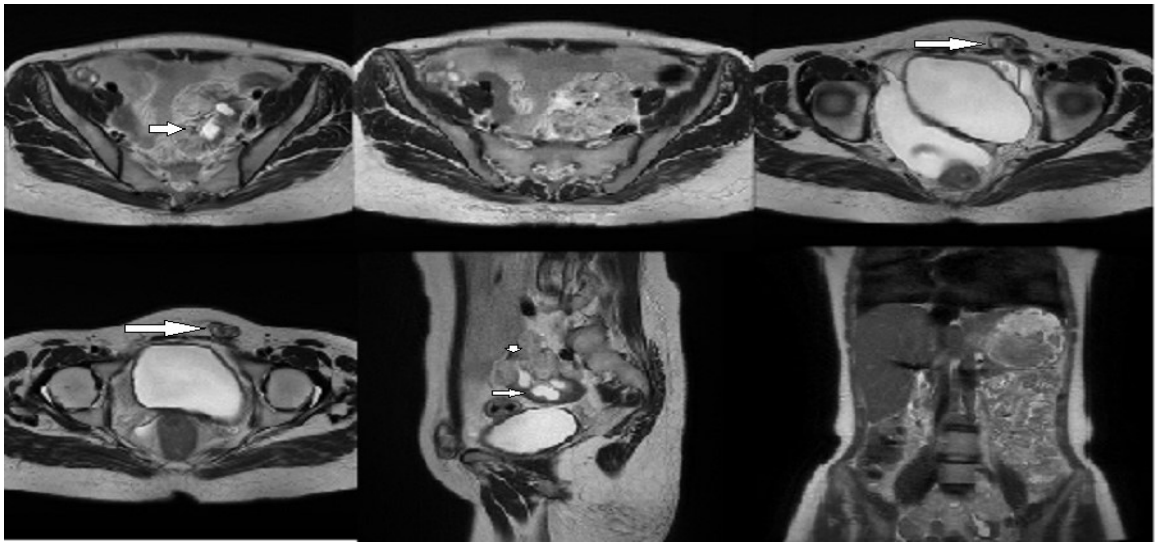

Figure-3: Axial, sagittal and coronal images of 25years old patient with MRKH II. In addition of absence of uterus, left ovary and rudimentary left uterine bud are ectopically seen herniating in the inguinal canal (long arrows). Right ovary is normally cited in pelvis. Empty renal fossa seen in the coronal image with ectopic left pelvic kidney (small arrows) showing abnormal signal neoplastic lesion along with pelvic ascites (arrowhead)

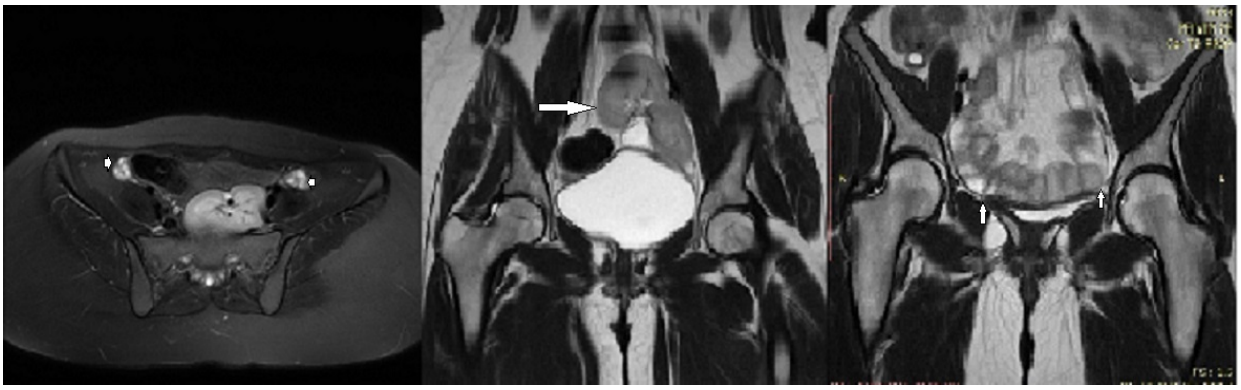

Figure-4: Bilateral ovaries with follicular activity (arrowheads) and ectopic left kidney (long arrow). Uterus and upper vagina are absent with coronal image showing convergence bands (small arrows).

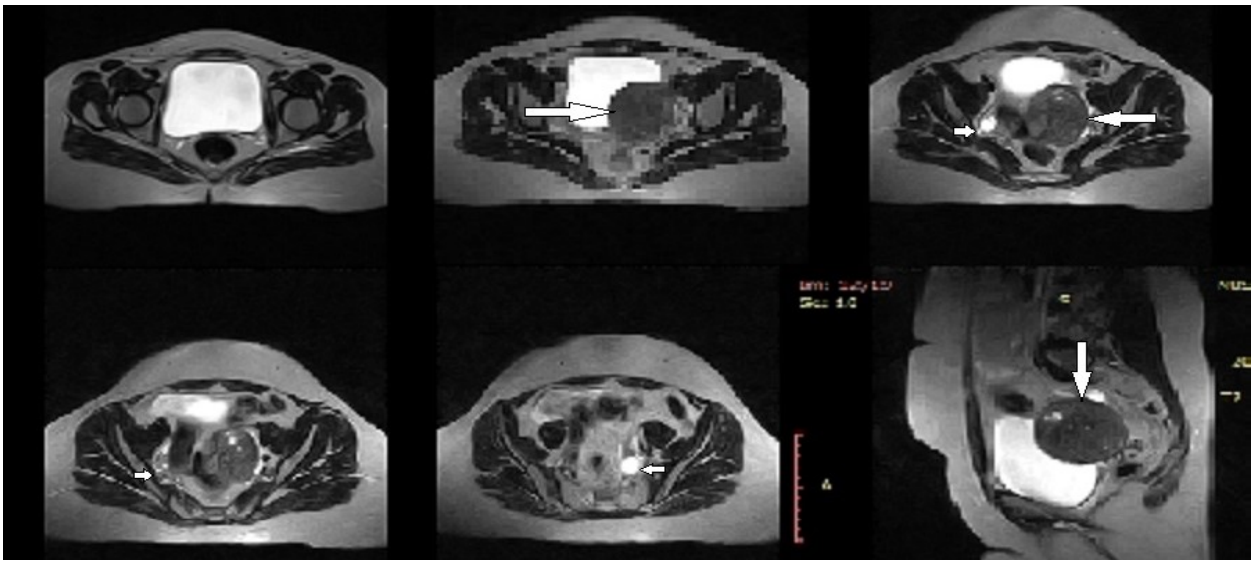

Figure-5: MRI of 35 years female with MRKH syndrome shows a well-defined lobulated low signal lesion with internal areas of high signal on T2WI arising from rudimentary left uterine bud representing a leiomyoma (long arrows). Both ovaries are separately identified with follicular activity (small arrows).

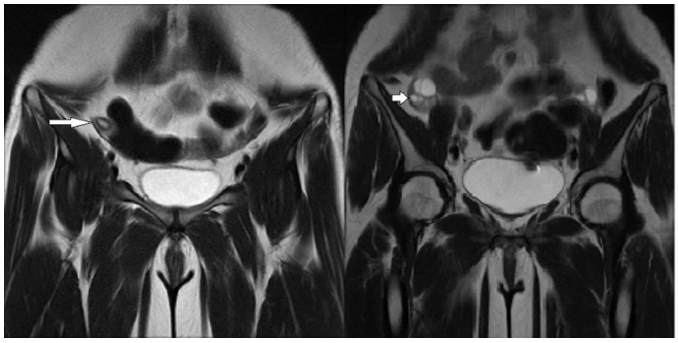

Figure-6: Patient with MRKH presented with pain lower abdomen showed a three layered right rudimentary uterine bud (long arrow) closely abutting the inferior aspect of the right ovary (small arrow).

DISCUSSION

MRKH syndrome is the second commonest cause of primary amenorrhea and accounts for 15% of the patients with primary amenorrhea.[5,6] In the embryonic life after the descent of ovaries into the pelvis Paramesonephric ducts (Mullerian ducts) fuse to form Fallopian tubes, uterus and upper 2/3rd of vagina. An arrest in this process or agenesis of Paramesonephric ducts results in MRKH syndrome

while absence of midline fusion of the two paramesonephric ducts results in congenital malformations of uterus and spectrum ranges from uterus didelphys to arcuate uterus. Lower 1/3rd of the vagina having its embryological origin from urogenital sinus may be seen with its complete development in MRKH syndrome or may be atretic/hypoplastic.

Obstructive vaginal developmental anomalies like imperforate hymen, vaginal atresia, transverse and longitudinal vaginal septum are the clinical differentials of MRKH syndrome. Ultrasound as first line of investigation which is cost effective and non-invasive is sufficient to differentiate the two categories of malformation. In isolated vaginal atresia, there is variable presence of upper 2/3rd of vagina, however uterus and ovaries appear normal. In imperforate hymen and transverse vaginal septum, uterus and upper vaginal are normal in appearance.[7]

MRI is the investigation of choice for MRKH syndrome as it is free of ionizing radiation and its ability to provide high soft tissue resolution and clear view of anatomy in multiple planes is unmatched with other radiological investigations. MRI is also imperative in evaluation of renal and anorectal anomalies. Uterus and the upper 2/3rd of vagina are not demonstrated while the lower 1/3rd of vagina which is of different embryonic origin is always observed in MRKH syndrome. Hematometra can be seen to a varying degree in about 6–10% of patients with MRKH syndrome due to the occurrence of endometrial glands leading to cyclic abdominal pain.

Type II MRKH syndrome also knowns as MURCS are associated with non-gynaecological abnormalities including renal and skeletal anomalies. Renal anomalies being the commonest due to closely related embryonic development of two systems from Mesonephric and Paramesonephric ducts. These renal anomalies range from ectopic kidneys, crossed fused ectopia, horseshoe kidney, unilateral agenesis and urachal remnant.[8–10] In our study all three patients with type II MRKH syndrome had associated renal anomalies. Two patients had unilateral ectopic kidney with normally cited contralateral kidney.

One of our patients had left renal agenesis and right ectopic pelvic kidney. Ectopic pelvic kidney showed abnormal signal neoplastic mass replacing part of the renal parenchyma. Patient also had pelvic ascites and deranged renal function. Percutaneous biopsy of the mass showed Renal cell carcinoma. The same patient had ectopic left ovary in the left inguinal canal and MR showed left ovary along with left uterine bud herniating into the inguinal canal. Patient was being prepared for renal transplant but unfortunately died before the procedure secondary to renal failure. Our literature review has revealed some cases of ovarian tumour in patients of MRKH but renal neoplasm in solitary kidney in patient with MRKH has not been previously reported.

In the most recent publications on MRKH syndrome in patients presenting with pelvic masses showed presence of leiomyoma in the uterine remnant.[11–16] One of our patients similarly showed well defined rounded leiomyoma in the left uterine remnant. The patient presented with lower abdominal discomfort.

Strength of the article is that a rare disease is studied but a more robust sample size would have been better. However, we believe that it will add to existing body of knowledge in a meaningful way.

CONCLUSION

MRI is an excellent imaging modality for accurate diagnosis and evaluation of other system anomalies in MRKH syndrome. Rudimentary uterine buds are commonly seen in these patients and may have functioning endometrium. Ovaries are of normal appearance but frequently ectopic.

Conflict of interest: The authors declare that there is no conflict of interest

AUTHORS' CONTRIBUTION

NK: Conception and design of the work; data acquisition, data analysis, interpretation of data, writing the manuscript. MK: Conception and design of the work; data acquisition, data analysis, drafting the manuscript and literature review. MMA: Conception and design of the work; data acquisition, interpretation of data, literature review. RR: Conception and design of the work; data acquisition, data analysis, interpretation of data, revising and writing the manuscript.

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