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Original Article

Radiology Section

Magnetic Resonance Imaging in Mayer-Rokitansky-Kuster-Hauser Syndrome: A Retrospective Study

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ABSTRACT

Introduction: Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is a congenital anomaly in female genital tract owing to irregular embryonic development of para-mesonephric ducts and thus leads to uterine and proximal vagina aplasia or hypoplasia. MRKH syndrome has devastating effects for fertility and sexual intercourse in young women. After diagnosis by the imaging features of Magnetic Resonance Imaging (MRI) surgery may allow patients to have sexual function with possible attainment of reproduction after assisted reproduction technique or surrogacy.

Aim: To analyse the MRI findings in females suspected of MRKH syndrome in a primary amenorrhea workup.

Materials and Methods: A retrospective study comprising of 11 patients of MRKH syndrome presented in a tertiary care centre from May-2013 to April-2019 was evaluated in a 1.5 Tesla MRI scanner. The diagnostic confirmation of MRKH syndrome was made on the basis of the following features: i) presence or absence of uterine buds; ii) fibrous band like structures connecting bilateral uterine buds; and iii) Midline soft tissue at uterine region. The data was entered in the excel sheet and results were expressed in terms of frequency and percentages.

Results: MRI revealed small vestigial uterus in the form of uterine buds in five patients (45.4%) and rudimentary streaky uterus in six patients (54.54%). The presence of the endometrium with prominent cavitation could be detected only in four patients (36.3%), while the remaining seven patients (63.6%) showed no cavitation. The uterine buds were connected with fibrous band-like structures in three patients (27.2%) and in the remaining 8 (72.7%) patients the uterine buds were located laterally without any apparent connection. The midline soft tissue posteriosuperior to the urinary bladder dome is seen in three cases (27.2%). Bilateral ovaries were normal in size and morphology in all cases. In good number of cases the uterine buds were closely associated with ovaries. Most of them were located in the iliac fossa. All patients displayed only lower 1/3rd of the vagina with the absence of upper 2/3rd vagina. Associated malformations were found in four cases, which were related to renal in 9.09% case, vertebral in 27.2% cases and congenital vesicovaginal fistula in 9.09% case. In brief Type I MRKH is seen in seven cases (63.6%) and Type II MRKH is seen in four cases (36.3%).

Conclusion: The diagnosis of MRKH syndrome is made based on clinical findings, but radiological evaluation is also essential for the confirmation. 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.

Keywords: MRKH, Mullerian, Primary amenorrhoea, Remnant

INTRODUCTION

MRKH syndrome is a congenital mullerian anomaly characterised by malformation in the female genital tract due to absence or reduced embryonic development of the para-mesonephric ducts which leads to uterine and proximal vagina aplasia/hypoplasia. In most of the cases the cause is unknown, though it results from combined genetic and environmental factors [1]. It affects approximately 1 in 4500 live births [2]. Both ovaries are functionally normal: thus, patients usually present with primary amenorrhea during adolescence with normal pubertal development and secondary sexual characteristics. Skeletal abnormalities also can co-exist in about 10% of the patients.

MRKH syndrome has devastating effects for fertility and sexual intercourse in young women. The diagnosis must be reached quickly to initiate clinical and psychologic treatment. For the restoration of normal sexual function, surgery is necessary. In appropriate cases Currently assisted reproductive techniques are performed for reproduction. Prior to surgery or assisted reproduction technique, thorough evaluation of the anatomy of uterus, ovaries, and vagina are essential for best surgical outcome [2].

The role of MRI in these patients is to depict the pelvic anatomy and to identify abnormally developed or positioned gonads. MRI is a non-invasive technique for accurately assessing the pelvic organs anatomy, if any associated spinal anomalies, renal, and osseous structures. T1W, T2 weighted MR images provide excellent zonal anatomy of the uterus i.e., endometrium, junctional zone and myometrial anatomy [3,4]. The present study was conducted with an aim to analyse the MRI findings in females of MRKH syndrome in a primary amenorrhea group.

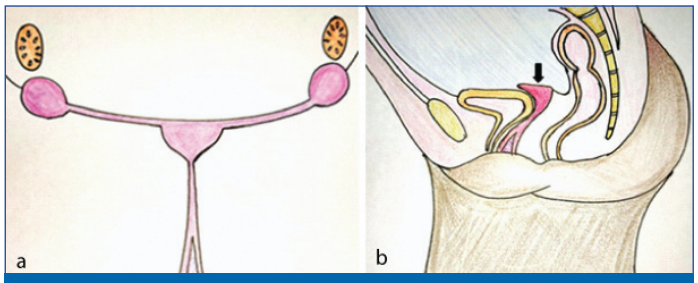
MATERIALS AND METHODS

A retrospective study was conducted from May 2017 to April 2019 in a tertiary care centre, in which 11 cases of MRKH syndrome from May-2013 to April-2019 were analysed. This study was approved by institution ethical committee via no. 508/19. Retrospective evaluation of the medical records of patients was done and females with primary amenorrhea and clinical suspicion of uterine anomalies on outpatient basis were included while MR images with poor quality were excluded from the study.

MR images were obtained for all patients with 1.5T (PHILLIPS ACHIEVA) 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. After a localiser scan, initially upper abdomen screening was done with unenhanced T1-weighted transverse fast spin-echo sequence. Subsequently, unenhanced T2-weighted fast spin echo sequences was imagined in three planes, and a T1-weighted transverse fast spin echo sequences of pelvis were acquired. Coronal T2-weighted image

(T2WI) with repetition time (TR) of 6,100 ms, echo time (TE) of 90 ms; T2-weighted image SPAIRin the sagittal plane (TR: 5,000ms, TE: 80ms); T2WI (TR: 4,700 ms,TE: 100ms) in the transverse plane; T2-weighted image SPAIR in the transverseplane (TR: 6,500 ms, TE: 80 ms); sagittal T2WI TSE (TR: 3,500 ms, TE: 90 ms), sagittal T2W STIR with long TE (TR: 3,600 ms, TE: 80 ms), T1WI SPAIR Axial (TR: 1,200 ms, TE: 7 ms), BTFE (TR: 2,800 ms, TE: 1.4sec), slice thickness of 3-5 mm, Field Of View (FOV): 250-350 mm. flip angle- 90^0 interslice gap: -1 to 1 mm.

Two radiologists with more than 15 years of experience in imaging patients with congenital mullerian anomalies working in consensus analysed MR images for the following features i) presence or absence of uterine buds; ii) fibrous band-like structures connecting bilateral uterine buds; and iii) Midline soft tissue at uterine region. Differentiation of the uterus into one, two, or three layers (myometrium, junctional zone, and endometrium) was analysed, and any signs of intraluminal blood was noticed. The presence of the distal vagina was noticed. The anatomical schematic drawings of typical mullerian remnants are depicted in [Table/Fig-1].



[Table/Fig-1]: Anatomical diagram representing typical mullerian remnants: a) Bilateral uterine buds are connected by a fibrous band like structure which meet at midline triangular soft tissue. Ovaries abutting uterine buds also shown; b) Sagittal image showing triangular soft tissue (arrow) lying posteriosuperior to urinary bladder. This soft tissue is continuous with the vagina showing atrophy in upper 2/3rd.

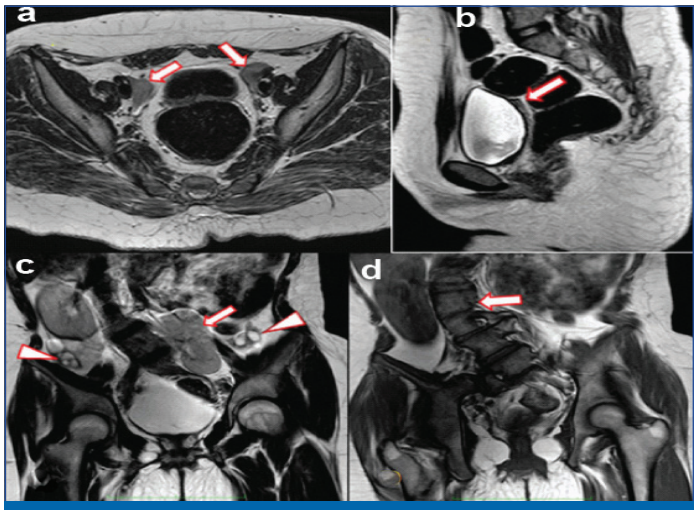
STATISTICAL ANALYSIS

All the statistical analysis were calculated using Statistical Package for the Social Science (SPSS) version 16.0 SPSS Inc., Chicago, USA). The data was expressed in terms of frequencies and percentages.

RESULTS

The mean age of the study patients were 17.09 years (age range: 10-25 y).

MRI revealed small vestigial uterus in the form of uterine buds in five patients (45.4%) and rudimentary streaky uterus in six patients (54.54%). The presence of the endometrium with prominent cavitation could be detected only in four patients (36.3%), while the remaining seven patients (63.6%) showed no cavitation. The uterine buds were connected with fibrous band-like structures in three patients (27.2%) and in the remaining 8 (72.7%) patients the uterine buds were located laterally without any apparent connection. The central soft tissue posteriosuperior to the urinary bladder dome was seen in three cases (27.2%). Bilateral ovaries were normal in size and morphology in all cases. Most of them were located in the iliac fossa. All patients displayed only lower 1/3rd of the vagina with the absence of upper 2/3rd vagina. Associated malformations were found in four cases, which were related to renal in 1 (9.09%) case, vertebral in 3 (27.2%) cases (same patient has vertebral anomoly and renal malformation i.e., case 7 as seen in [Table/Fig-2]) and congenital vesicovaginal fistula in 1 (9.09%) case. In brief, Type I MRKH was seen in seven cases (63.6%) and Type II MRKH was seen in four cases (36.3%) [Table/Fig-2]. The few representative cases are described in [Table/Fig-3-5].



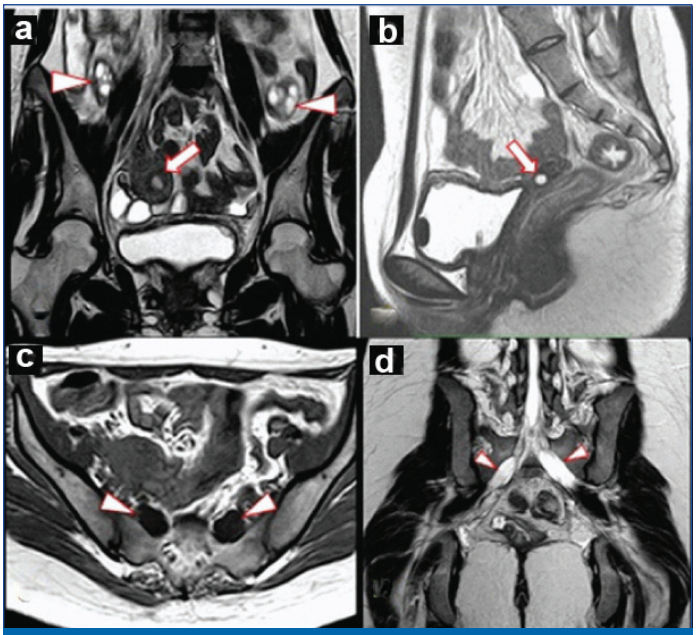
[Table/Fig-3]: A representative case of MRKH syndrome in 25-year-old woman: a) On the axial T1-weighted image, bilateral hypoplastic uterine buds (thick arrows) are found on each side of lateral pelvic walls; b) Sagittal T2-weighted image showing, fibrous strand (thick arrow) found posteriosuperior to urinary bladder, which is the usual location for the uterus and vagina; c) Coronal T2-weighted image showing ectopic left kidney (thick arrow). Bilateral ovaries (arrow heads) are also foun d in the iliac fossa and appear normal; d) Coronal T2-weighted image showing severe scoliosis with possible vertebral segmentation anomalies at lumbosacral region (thick arrow).

DISCUSSION

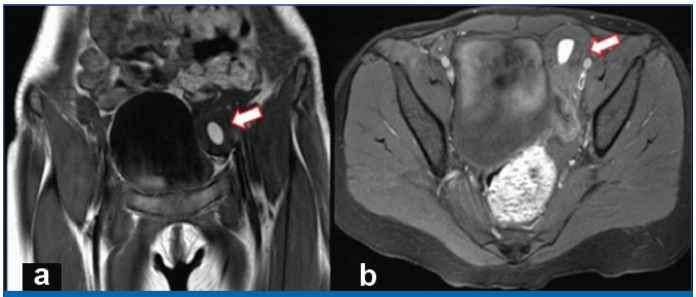
In present study all the patients had rudimentary uterus. The location of the rudimentary uteri of all patients was lateral to the pelvis, with the inferior caudal margin tending medially toward the midline and uteri were not conjoined. MRI revealed small vestigial uterus in the form of uterine buds in five patients (45.4%) and rudimentary streaky uterus in six patients (54.54%). The findings of the present study is shown in [Table/Fig-6] [3,5-8].

S. No	Age in years	Uterus buds/ hypoplastic	Endometrial cavity	Ovaries	Fibrous connection b/w mullerian rudiments	Presence of paramedian soft tissue above urinary bladder dome	Secondary defects	Type
1	18	Buds	Present	Normal	Present	Present	Transitional lumbosacral vertebra. Dural ectasia.	II
2	18	Buds	Absent	Normal	Present	Absent	---	I
3	20	Hypoplastic	Absent	Normal	Absent	Present	Haemangioma	II
4	12	Hypoplastic	Absent	Normal	Absent	Present	Congenital vesicovaginal fistula.	II
5	16	Buds	Absent	Normal	Present	Absent	---	I
6	18	Buds	Absent	Normal	Absent	Absent	----	I
7	25	Hypoplastic	Absent	Normal	Absent	Absent	Severe scoliosis with possible vertebral segmentation anomalies at lumbosacral region. Ectopic left kidney	II
8	10	Buds	Present	Normal	Absent	Absent	----	I
9	10	Hypoplastic	Present	Normal	Absent	Absent	----	I
10	18	Hypoplastic	Present	Normal	Absent	Absent	----	I
11	23	Hypoplastic	Absent	Normal	Absent	Absent	----	I

[Table/Fig-2]: Table depicting the findings of present study.



[Table/Fig-4]: a) Coronal T2-Weighted image, showing right paramedian soft tissue represents uterine bud with persistent small endometrial rudimentary cavity (thick arrow). Bilateral ovaries (arrow heads) are also found in the pelvic cavity and appear normal; b) Sagittal T2-weighted image showing, fibrous tissue (thick arrow) at the usual location of uterine region with small cyst within it; c,d) shows Axial T1-weighted image and Coronal T2-weighted image showing bilateral dural ectasia of sacral nerve roots. (arrow heads).



[Table/Fig-5]: A representative case of MRKH syndrome in 10-year-old girl. a,b) Coronal and axial T1 weighted images showing hypoplastic uterine bud with hematometrain left uterine bud (thick arrows).

Boruah DK et al., reported that type-II MRKH syndrome is associated with ovarian cancers and cardiac malformations [3]. Hence, differentiation between MRKH syndrome and androgen insensitivity syndrome are essential for treatment planning of such patients. In present study we didn't find any ovarian and cardiac anomalies.

Oppelt P et al., has reviewed 53 cases of MRKH syndrome and divided into three subtypes: typical, atypical and MURCS association. A total of 521 cases were included of which 64% were typical, 24% atypical and only 12% MURCS. The most common type of malformation was of renal system [9].

The work by Hall-Craggs MA et al., proved that rudimentary uteri were common in MRKH syndrome [10]. The rudimentary uteri can be relatively large and have a functioning endometrium, which may be complicated with pelvic pain.

Limitation(s)

Apart from the intrinsic limits of any retrospective study, few other limitations of present study include:

1. The sample size was small to generalise present study findings and thus study including larger population would be required.
2. At present no patient underwent any surgical corrective procedures, so pathological correlation of the findings could not be done.

CONCLUSION(S)

MRI is considered as the imaging modality of choice, because of its ability to accurately identify female genital tract malformations along with associated renal and skeletal anomalies. Following MRI diagnosis, surgery may allow patients to have sexual function with possible attainment of reproduction after assisted reproduction technique or surrogacy.

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Author name	Number of cases	Age/ mean age (years)	Location of Rudimentary Uterine bud	T2 hypointense converging band in between the uterine bud (n)	Endometrial differentiations in uterine bud	Location of ovaries (in numbers)	Identified lower 1/3rd vagina (in numbers of patient)	Associated non-gynaecological anomalies
Present study	11	17.09 (age range 10-25 years)	Uterine buds-5 Hypoplastic-6	Present-3 (27.2%)	Yes-4 (36.3%)	-	-	Renal in 1 (9.09%), vertebral in 3 (27.2%) and congenital vesicovaginal fistula in 1 (9.09%)
Boruah DK et al., [3]	9	17.8	17 located in pelvic cavity and 1 located in left inguinal canal.	7 (77.8%) patients	17 uterine buds shows only 1 layer and 1 uterine bud shows more than 2 layers		9 (100%)	Ectopic kidney-1, scoliosis of lumbar spine-2, CRS-V with intrasacral meningocoele and hemi-vertebra-1,
Giusti S et al., [5]	1	15	Hypoplastic blind ended uterus located right iliac fossa	-	More than 2 layers	Pelvic cavity-2	1	-
Reinhold C et al., [6]	12	-	Uterine agenesis -9, unicornuate hypoplastic uterus-1, atresia of lower uterine segment-1, small fibrous remnant-1	-	-	Pelvic cavity-24. One left ovary shows endometrium	Complete vaginal agenesis-5, Agenesis of proximal 2/3rd-3, Fibrous remnant -2, Agenesis of proximal 1/3rd-2	-
Yoo R-E et al., [7]	15	23.7	Pelvic cavity -30	15 (100%)	only 1 layer- 28 uterine buds, more than 2 layers-2	Pelvis-30 (100%)	14 (93%)	Renal=2 (13.3%) Vertebral=4 (26.7%)
Kara T et al., [8]	16	19.4	Uterine aplasia -5 (31.3%), Uterine hypoplasia-11 (68.8%)	-	-	Pelvic cavity-21 Not detected-10, agenesis-1	16 (100%)	Renal=4 Vertebral=2

[Table/Fig-6]: Table comparing the findings of present study and other case studies [3,5-8].

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