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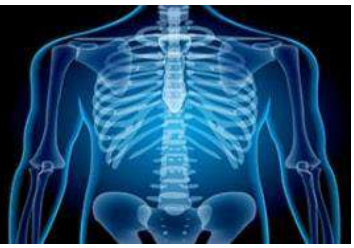

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Array of magnetic resonance imaging findings in Mayer Rokitansky Kuster Hauser syndrome

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Abstract
MRKH syndrome (class I mullerian duct anomaly) is a rare congenital disorder characterized by uterine and cervical aplasia/hypoplasia with normally functioning ovaries in genotypical normal female (46XX) due to failed/interrupted development of the mullerian duct [1,2]. Patients usually present with normal secondary sexual characteristics and primary amenorrhea. In our study we reviewed the array of magnetic resonance imaging findings in MRKH syndrome and the various MR imaging appearances.
A retrospective descriptive case series of six cases was done. Out of the six cases, one patient had imaging features consistent with Mayer-Rokitansky-Kuster- Hauser syndrome type II (vertebral column anomalies) and five patients with Mayer-Rokitansky-Kuster-Hauser syndrome type I. Four patients had mullerian duct remnants evident on imaging as either and or presence of rudimentary uterine horns with cavitation, mid line triangular soft tissue structure, fibrous band. One patient had ectopic location of left ovary.

Keywords: Mayr Rokitansky Kuster Hauser syndrome, mullerian duct, vertebral anomalies, secondary sexual charactersitics

Introduction
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 [1, 2]. Patients usually present with normal secondary sexual characteristics and primary amenorrhea [3]. The diagnosis of MRKH has grave implications on the psychological and reproductive outcomes of the patient. The management includes but is not limited to surgery and artificial reproductive techniques. The accurate diagnosis is essential for good clinical outcomes following treatment. Detailed description of spectrum of findings in MRKH syndrome has been documented in limited number of women. Identification of morphology and location of mullerian remnants, the presence of rudimentary uterine horns, ectopic location of ovary being reported as absent has lead to diagnostic dilemma, mismanagement of the patient and poor outcomes [4]. MRI is the imaging modality of choice to visualise and characterise uterus, vagina and ovaries. Therefore this study was done to document the various imaging findings in patients suspected of the rare disorder of MRKH syndrome to aid in management.

Materials and Methods
Patient population
MRKH syndrome is a rare disorder, hence retrospective descriptive case series was done. MR images of female patients presenting to the department of Radio-diagnosis, Bangalore medical college and research institute, Bangalore with clinical suspicion of MRKH syndrome were reviewed. Clinical suspicion was based on 1. Clinical history and gynaecological examination 2. Hormonal study 3. Transabdominal ultrasound
Detailed clinical history, physical examination and laboratory work up details were taken from hospital records.

Imaging technique

Patients were imaged using SIEMENS magneto avento 1.5 Tesla MR system and following imaging sequences were done T1 AXIAL, CORONAL, SAGITTAL; T2 AXIAL, CORONAL & FAT SUPPRESSED T2 WEIGHTED MULTIPLANAR IMAGING of pelvis and T2 HASTE CORONAL AND AXIAL of abdomen was performed.

Study period: June 2021 to August 2022

Imaging analysis

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 band like structure extending between the uterine buds if present
5. Presence or absence of ovaries, location and morphology
6. Associated non gynaecological anomalies

Case series

Case 1

A 17 year old female presented with complaints of primary amenorrhea associated with cyclical abdominal pain since 2 years. Patient had unremarkable medical history and was not on any medications. There was no history of congenital anomalies in the family or history of similar complaints in the family. There was no history of hormonal replacement/therapy in the mother during pregnancy.

On physical examination, vitals were stable, patient was of appropriate height and weight for chronological age. No abnormalities were detected on systemic examination.

Breast development: Tanner stage 5

Pubic hair: Tanner stage 5

Laboratory tests: Serum FSH and LH levels were within normal limits

Patient was referred to department of Radiodiagnosis, BMCRI for MRI of pelvis:

Preliminary ultrasound revealed non visualisation of uterus and left ovary. Right ovary was normal in morphology and location.

Findings

Absence of normal morphology of uterus and cervix.

No mid line triangular soft tissue structure noted.

A soft tissue structure noted anteromedial to the left external iliac vessels-likely remnant left uterine bud

A soft tissue structure noted anterior and superior to the right ovary. However no cavitation or intraluminal blood noted- likely remnant right uterine bud

A fibrous band noted extending from the right remnant uterine bud to the left remnant uterine bud.

Right ovary is normal in size and location.

Left ovary is normal in size and extra pelvic in location, located in the left iliac fossa overlying the iliacus and just lateral to the psoas muscle. A thickened and elongated round ligament noted extending from the left ovary to the left uterine bud.

Minimal free fluid noted in the pelvic cavity

Visualized extent of vertebral column appears normal.

On MRI screening of abdomen-Bilateral kidneys appear normal in size, location and signal characteristics.

Based on the findings a diagnosis of Mayer-Rokitansky-Kuster-Hauser syndrome type I was made.

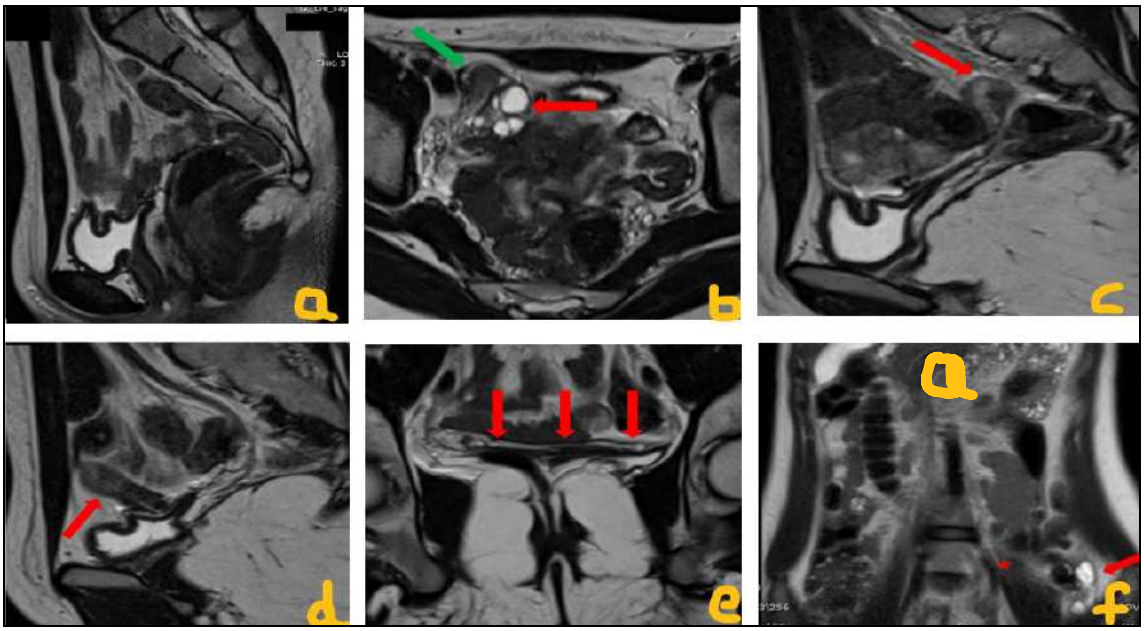

Fig 1: (a) T2 weighted sagittal image of pelvis shows absence of normal morphology of uterus and cervix. (b) T2 weighted axial image of the pelvis shows normal morphology and location of right ovary (red arrow) and uterine bud (green arrow) lateral to the right ovary, medial to the iliac vessels. (c) T2 weighted sagittal image of pelvis shows remnant right uterine bud (red arrow). No cavitation or blood products noted within. (d) T2 weighted sagittal image of the pelvis shows left remnant uterine bud (red arrow). (e) T2 weighted coronal image of the pelvis shows a fibrotic band connecting the two uterine buds (red arrow). (f) T2 weighted coronal image of the abdomen shows normal morphology of the left ovary (red arrow), located in the left iliac fossa lateral to the psoas muscle (red asterisk)

Case 2

An 18 year old female presented with complaints of primary amenorrhea. She had no significant past history, unremarkable medical history and was not on any medications. There was no family history of similar complaints or congenital anomalies. There was no history of hormonal replacement/therapy in the mother during pregnancy.

On physical examination, vitals were stable, patient was of appropriate height and weight for chronological age. No abnormalities were detected on systemic examination.

Breast development: Tanner stage 5

Pubic hair: Tanner stage 5

Laboratory tests: Serum FSH and LH levels were within normal limits

Patient was referred to department of Radiodiagnosis, BMCRI.

A preliminary ultrasound revealed bilateral ovaries in normal location and morphology with non visualisation of the uterus. MRI of pelvis was done.

Findings

Absence of normal morphology of uterus, cervix and upper 2/3rds of vagina

A midline triangular soft tissue structure noted poster superior to the urinary bladder

No remnant uterine buds noted bilaterally.

Bilateral ovaries normal in location and morphology.

On screening MRI abdomen: Both the kidneys are visualized and are normal in size, location and signal characteristics.

On CT screening

Wedge shaped lateral halves with smooth convex opposing margins at the site of sagittal cleft noted in T12, L3and L5 vertebrae - S/o Butterfly vertebrae

Congenital block vertebrae noted with fusion of vertebral body in the left lateral aspect and posterior elements of L2 and L3 vertebrae noted.

Sagittal cleft noted in the sacrum.

Based on the finding a diagnosis of Mayer-Rokitansky-Kuster-Hauser syndrome type II was made.

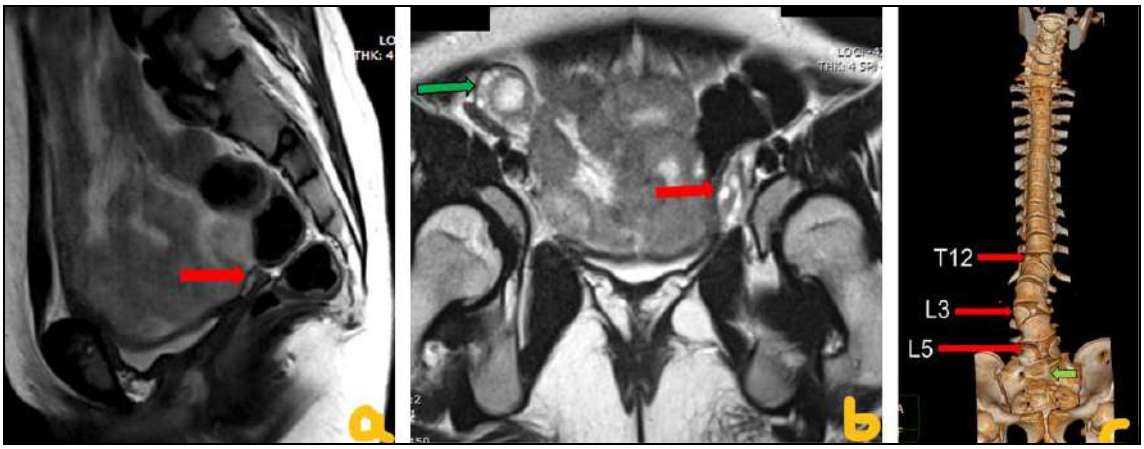

Fig 2: (a) T2 weighted sagittal image of the pelvis shows absence of normal morphology of normal uterus and cervix with a midline soft tissue structure (red arrow) noted in the rectovesical pouch superior to the urinary bladder. (b) T2 weighted sagittal image of pelvis shows bulky left ovary (red arrow) in normal location with remnant uterine bud (green arrow) with cavitation (central T2 hyper intense zone, T2 hypo intense middle zone and intermediate signal intensity of outer zone). (c) T2 hyperintense axial image of the pelvis shows bilateral bulky ovaries (red arrows) with multiple tiny follicles within. Rudimentary left uterine horn (green arrow) noted anterior to the left ovary.

Case 3

A 17 year old female patient presented complaints of primary ammenorrhea. Past medical history and family history was unremarkable. Patient was not on nay medication. There was no exposure to any hormone replacement drugs to the mother during pregnancy.

On examination, patient's vitals were stable, height and weight were normal for chronological age and systemic examination revealed no abnormality.

Breast development: Tanner stage 5

Pubic hair: Tanner stage 5

Patient was referred to department of Radiodiagnosis, BMCRI. A preliminary ultrasound revealed bilateral ovaries in normal location and morphology with non visualisation of the uterus. MRI of pelvis was done.

Findings

Absence of normal morphology of uterus and cervix between the bladder and rectum.

Lower vaginal canal appears normal.

A midline, triangular soft tissue structure noted posterior to dome of urinary bladder.

A unilateral left uterine bud noted anterior to the left ovary with central T2 hyperintese zone, T2 hypo intense middle layer and T2 intermediate signal outer zone-suggestive of cavitation

No fibrous band noted extending from the uterine bud.

Right ovary measures 3.5x5.0x3.2cm (28cc) and left ovary measures 3.4x3.3x2.5cm (14cc). Both ovaries are bulky in size with multiple peripherally arranged follicles measuring 2-9mm.

Visualised extent of the vertebral column appears normal.

On MRI screening of abdomen-Both the kidneys are normal in size, location and signal characteristics.

Based on the finding a diagnosis of Mayer-Rokitansky-Kuster-Hauser syndrome type I with polycystic ovaries was made.

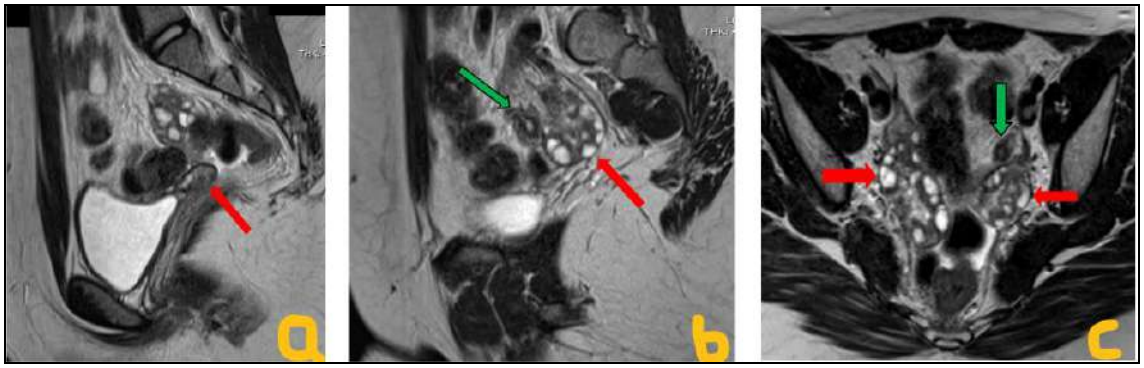

Fig 3: (a) T2 weighted sagittal image of the pelvis shows absence of normal morphology of normal uterus and cervix with a midline soft tissue structure (red arrow) noted in the rectovesical pouch superior to the urinary bladder. (b) T2 weighted sagittal image of pelvis shows bulky left ovary (red arrow) in normal location with remnant uterine bud (green arrow) with cavitation (central T2 hyper intense zone, T2 hypo intense middle zone and intermediate signal intensity of outer zone). (c) T2 hyperintense axial image of the pelvis shows bilateral bulky ovaries (red arrows) with multiple tiny follicles within. Rudimentary left uterine horn (green arrow) noted anterior to the left ovary.

Case 4

A 13 year old female presented with complaints of primary amenorrhea. She had no significant past history, unremarkable medical history and was not on any medications. There was no family history of similar complaints or congenital anomalies. There was no history of hormonal replacement therapy in the mother during pregnancy.

On physical examination, vitals were stable, patient was of appropriate height and weight for chronological age. No abnormalities were detected on systemic examination.

Breast development: Tanner stage 5

Pubic hair: Tanner stage 4

Laboratory tests: Serum FSH and LH levels were within normal limits

Patient was referred to department of Radiodiagnosis, BMCRI.

A preliminary ultrasound revealed bilateral ovaries in normal location and morphology with non visualisation of the uterus. MRI of pelvis was done.

Findings

Absence of normal morphology of uterus and cervix between the bladder and rectum. A midline, triangular soft tissue structure noted posterior to dome of urinary bladder.

Vaginal canal appears normal in morphology, measures~ 5.4cm in length.

A unilateral right uterine bud noted inferior to the right ovary. However there was no evidence of cavitation within the uterine bud.

No fibrous band noted extending from the uterine bud.

Bilateral ovaries were normal in location and morphology.

Visualised extent of vertebral column appears normal.

Minimal free fluid noted in the pelvic cavity.

On MRI screening of abdomen-Bilateral kidneys appear normal in size, location and signal characteristics.

Based on the findings a diagnosis of Mayer-Rokitansky-Kuster-Hauser syndrome type I was made.

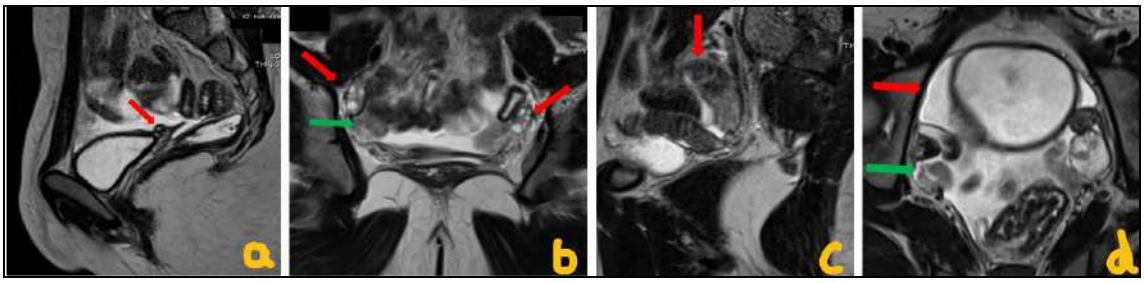

Fig 4: (a) T2 weighted mid sagittal image of the pelvis shows absence normal uterus and cervix with a midline soft tissue structure posterior to the urinary bladder (Red arrow). (b) T2 weighted coronal image shows normal location and morphology of bilateral ovaries(red arrows). The right uterine bud is noted inferior to the ovary. (green arrow). (c) T2 weighted axial image shows minimal T2 hyperintense fluid in the pelvic cavity(red arrow). T2 hyper intense uterine bud noted inferior to the right ovary (green arrow). No evidence of cavitation noted within the uterine bud. (d) T2 weighted sagittal image shows T2 hyper intense right uterine bud (red arrow)

Case 5

A 17 year old female presented with complaints of primary amenorrhea. Patient had no other significant complaints. Past medical history and family history was unremarkable. Patient was not on nay medication. There was exposure to any hormone replacement drugs to the mother during pregnancy.

On examination, patient's vitals were stable, height and weight were normal for chronological age and systemic examination revealed no abnormality.

Breast development: Tanner stage 5

Pubic hair: Tanner stage 5

Laboratory tests: Serum FSH and LH levels were within normal limits

Patient was referred to department of Radiodiagnosis, BMCRI.

A preliminary ultrasound revealed non visualisation of the uterus. Bilateral ovaries in normal location with a well defined cystic lesion with no separations or echogenic content within measuring~3.2x3.1cm in the right ovary. MRI of pelvis was done.

Findings

Absent morphology of normal uterus and cervix.
Lower 1/3rd of vagina visualized, measures~3.5cm in maximum length.
No triangular midline soft tissue structure noted.
Bilateral ovaries in normal in size and location.
A well defined T1 hypointense, T2 hyperintense lesion measuring~ 3.2x2.8x4.0cm noted in the right ovary-likely simple cyst.
Visualised extent of the vertebral column appears normal.
On MRI screening of abdomen-Both the kidneys are normal in size, location and signal characteristics.
Based on the finding a diagnosis of Mayer-Rokitansky-Kuster-Hauser syndrome type I was made.

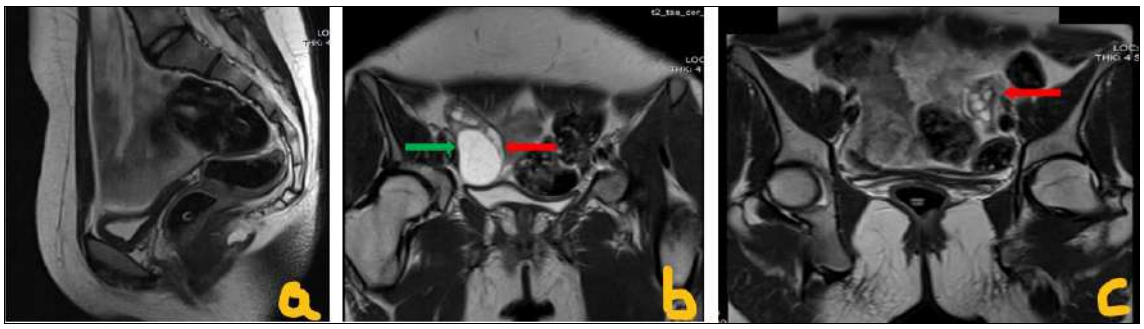

Fig 5: (a) T2 weighted axial sagittal image of pelvis shows absence of normal morphology of uterus and cervix. (b) T2 weighted coronal image of the pelvis shows normal location of right ovary (red arrow) with a T2 hyper intense cyst within (green arrow). (c) T2 weighted coronal image of pelvis shows normal location and morphology of left ovary (red arrow)

Case 6

A 23 year old female presented with complaints of primary amenorrhea. There was no other significant medical history. Patient was not on any medication. There was no history of similar complaints in the family or family history of other congenital anomalies. No history of hormonal exposure of the mother during pregnancy.
On physical examination, vitals were stable, height and weight were normal for the chronological age. Systemic examination revealed no abnormalities.

Breast development: Tanner stage 5

Pubic hair development: Tanner stage 5

Patient was referred to department of Radiodiagnosis, BMCRI.

A preliminary ultrasound revealed bilateral ovaries in normal location and morphology with non visualisation of the uterus. MRI of pelvis was done.

Findings

Absence of normal morphology of uterus, cervix and upper two thirds of vagina.
No midline triangular soft tissue structure noted.
No remnant uterine buds noted.
Both ovaries are normal in size and signal intensity with multiple tiny follicles noted within.
Visualised extent of the vertebral column appears normal.
On MRI screening of abdomen-Both the kidneys are normal in size, location and signal characteristics.
Based on the finding a diagnosis of Mayer-Rokitansky-Kuster-Hauser syndrome type I was made.

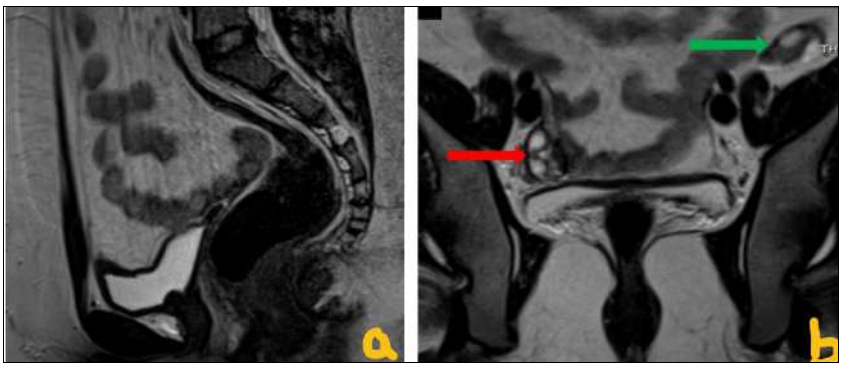

Fig 6: (a) T2 weighted sagittal image of the pelvis shows absence of normal morphology of the uterus and cervix. (b) T2 weighted coronal image of the pelvis shows normal location of the both the ovaries (red arrow-right ovary; green arrow: left ovary) with multiple tiny follicles within

Results and Discussion

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 [5].
MRKH syndrome, (class I mullerian duct anomalies) is the second most common cause of primary amenorrhea, even though it's a rare disorder with a reported incidence of 1 in 4000 to 5000 [5].

The female reproductive tract develops from a pair of Müllerian ducts. Uterus, cervix, fallopian tube and the upper two-thirds of the vagina are derived from the mullein duct. Failure or interrupted embryonic development of Müllerian duct leads to uterine and vaginal agenesis [2]. Ovaries, which are derived from primordial germ cells and gonadal ridge, are generally anatomically and functionally normal in patients with MRKH syndrome. MRKH syndrome is suspected in patients with normal development of secondary sexual characteristics with the absence of menarche and normal endocrine tests [3]. It comprises atresia of the upper 2/3rd of the vagina with either absent or rudimentary uterus. [5] Bilateral ovaries are normal in morphology and function but might be present in either the anatomical location or higher up in the pelvis or in the abdominal cavity. There may be associated renal, vertebral column and cloacal abnormalities [6].

The syndrome is broadly classified into type I with isolated utero-vaginal abnormalities and type II which is associated with non gynaecological abnormalities like renal, skeletal, cardiac abnormalities most common being renal anomalies, which may include ectopic pelvic kidneys, solitary ectopic kidney and renal agenesis. Vertebral anomalies could range from limbo sacral transition vertebra to scoliosis to sacral agenesis [6].

Ultrasound is generally the first modality for evaluating patients presenting with primary amenorrhea, but limitations being inability to perform transvaginal ultrasound in sexually inactive patients, difficulty identifying ectopic location of ovaries and misinterpreting rudimentary uterine horns or vestigial lamina as either pre pubertal uterus or hypo plastic uterus [6].

MRI is highly sensitive and specific for diagnosing MRKH syndrome. It also helps in providing important information regarding rudimentary Mullerian remnants and location of ovaries which helps in further management in line with assisted reproductive techniques [7].

The Mullerian duct remnants described in MRKH 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. [8] 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 [3, 7].

Ovaries are usually extra pelvic in location, commonly located at or above the pelvic brim, anterior or antero-lateral to the poas muscle, bilateral inguinal canals or lateral to the iliac vessels. A T2 coronal image with a large field of view helps in identifying ovaries located in extra pelvic locations, identification of which can aid in ova retrieval or prevent torsion [3].

Table 1: Characteristics, description and case study of MRKH syndrome

Sr. No	Characteristics	Description	Case 2	Case 3	Case 4	Case 5	Case 6
1	Uterus, Cervix and upper 2/3rd of vagina	Absent	Absent	Absent	Absent	Absent	Absent
2.	Mid line or paramedic soft tissue structure	Absent	Midline soft tissue triangular structure	Midline soft tissue triangular structure	Midline soft tissue triangular structure	Absent	Absent
3.	Uterine buds, laterality, location, evidence of cavitation	Bilateral remnant uterine buds with no cavitation	Absent	Left uterine bud with cavitation	Left uterine bud with cavitation	Absent	Absent
4.	Fibrous band like structure extending between the uterine buds	Present	Absent	None	None	Absent	Absent
5.	Ovaries, location and morphology	Right ovary normal in location and morphology. Left ovary extra pelvic in location	Normal in location and morphology	Normal in location with polycystic appearance	Normal in location with polycystic appearance	Normal in location and morphology	Normal in location and morphology
6.	Associated non gynaecological anomalies	None	Butterfly vertebrae and block vertebrae	None	None	None	None
	Diagnosis	MRKH type I	MRKH type II	MRKH type I	MRKH type I	MRKH type I	MRKH type I

Conclusions

Mullerian duct remnants are commonly present in MRKH syndrome. In addition, there is a high incidence of extra pelvic location of ovaries and presence of non gynaecological abnormalities. Evaluating the patient keeping in mind these associations of MRKH syndrome will aid in a comprehensive diagnosis and provide vital information for further management.

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Conflict of Interest

Not available

Financial Support

Not available

References

1. Folch M, Pigem I, Konje JC. Mullerian agenesis: Etiology, diagnosis and management. Obstetrical & Gynecological Survey. 2000;55(10):644-9.
2. Chandler TM, Machan LS, Cooperberg PL, Harris AC, Chang SD. Müllerian duct anomalies: From diagnosis to intervention. The British Journal of Radiology. 2009;82(984):1034-42.
3. Hall-Craggs MA, Williams CE, Pattison SH, Kirkham AP, Creighton SM. Mayer-Rokitansky-Kuster-Hauser Syndrome: Diagnosis with MR imaging. Radiology.

2013;269(3):787-92.
4. Yoo RE, Cho JY, Kim SY, Kim SH. Magnetic Resonance Evaluation of Müllerian remnants in Mayer-Rokitansky-Küster-Hauser syndrome. Korean Journal of Radiology. 2013;14(2):233.
5. Coco I, Fiaschetti V, Taglieri A, Gisone V, Simonetti G. Mayer-Rokitansky-Kuster-Hauser syndrome diagnosed by magnetic resonance imaging. Role of imaging to identify and evaluate the uncommon variation in development of the female genital tract. Journal of Radiology Case Reports; c2012, 6(4).
6. Behr SC, Courtier JL, Qayyum A. Imaging of müllerian duct anomalies. RadioGraphics; c2012, 32(6).
7. Pompili G, Munari A, Franceschelli G, Flor N, Meroni R, Frontino G, *et al*. Magnetic Resonance Imaging in the preoperative assessment of Mayer-Rokitansky-Kuster-Hauser syndrome. La radiologia medica. 2009;114(5):811-26.
8. Strübbe EH, Willemsen WN, Lemmens JA, Thijn CJ, Rolland R. Mayer-Rokitansky-Küster-Hauser Syndrome: Distinction between two forms based on excretory urographic, sonographic, and laparoscopic findings. American Journal of Roentgenology. 1993;160(2):331-4.

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