

## Atypical Mayer-Rokitansky-Kuster-Hauser (MRKH) Syndrome with Unilateral Renal Agenesis: A Case Report with Review of Literature

Rahul Dev<sup>1\*</sup>, Gitanjali Khorwal<sup>2</sup>, Vibhu Goel<sup>1</sup>, Mohit Tayal<sup>1</sup>, Nandolia Khanak Kumar Kantilal<sup>1</sup>, Udit Chauhan<sup>1</sup>, Sandeep Kumar<sup>1</sup>, Venkata Subbaih A<sup>1</sup>, Navojit Chatterjee<sup>1</sup>, Smily Sharma<sup>1</sup>, Sumit Kumar<sup>1</sup>, Navneet Kumar<sup>1</sup>, Sudhir Saxena<sup>1</sup>

<sup>1</sup>Department of Radiodiagnosis and Imaging, All India Institute of Medical Sciences, Rishikesh, Uttarakhand, India

<sup>2</sup>Department of Anatomy, All India Institute of Medical Sciences, Rishikesh, Uttarakhand, India

### \*Corresponding author

Rahul Dev

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**Abstract:** MRKH syndrome is a rare congenital anomaly characterised by uterine and vaginal hypoplasia. Normal external physical appearance precludes diagnosis until puberty with primary amenorrhoea being presenting symptom in majority. The ovarian function and karyotype are normal. MRI is the imaging modality of choice for evaluation of uterus, ovaries and vagina. The syndrome is classified into two types with type I being confined to reproductive system and type II having additional anomalies most frequent being of urinary system. We report a case of 22 year old patient presenting with primary amenorrhoea with absent uterus and right kidney as seen on ultrasonography with MRI confirming the same findings.

**Keywords:** Atypical, congenital, mullerian, mri, primary amenorrhoea, renal agenesis.

### INTRODUCTION

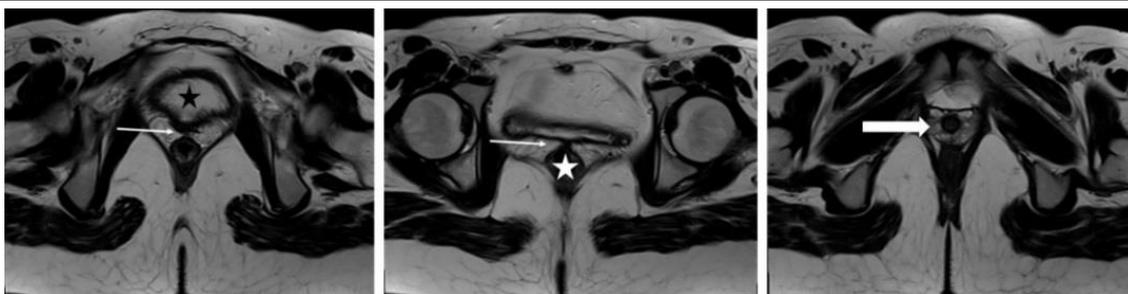
Mayer-Rokitansky-Kuster-Hauser syndrome or MRKH syndrome is a rare mullerian anomaly characterized by varying degree of hypoplasia of uterus and vagina. As the patient is phenotypically normal, it is difficult to diagnose this condition before puberty when patient invariably presents with complaints of primary amenorrhoea.

The role of imaging is to confirm the same as well as to evaluate for anomalies of other organ systems mainly excretory, musculoskeletal and respiratory system with MRI playing a pivotal role supplemented by ultrasonography and computed tomography.

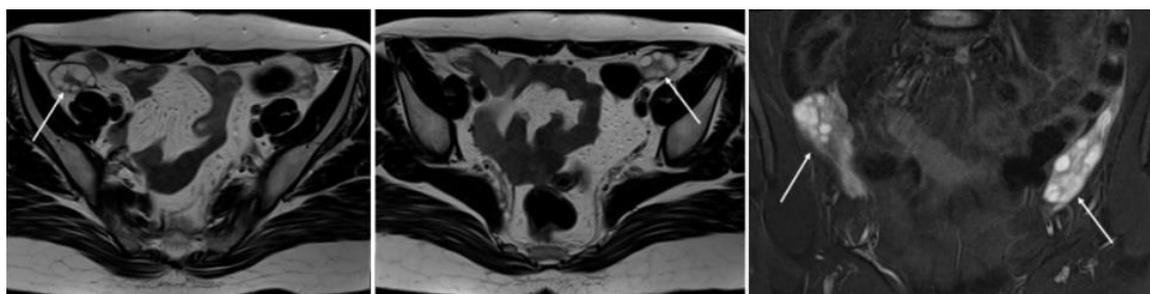
### CASE REPORT

A 22 year old girl was referred from gynaecology outpatient clinic for MRI examination on account of primary amenorrhoea. The patient has no significant past or family history with systemic examination and laboratory parameters within normal limits. The patient already underwent ultrasonography of pelvis revealing non visualisation of uterus and right kidney, however both ovaries appeared normal. The patient went MRI examination after taking written consent and ensuring no conditions precluding the scan. Axial and sagittal T2 weighted fast spin echo sequences

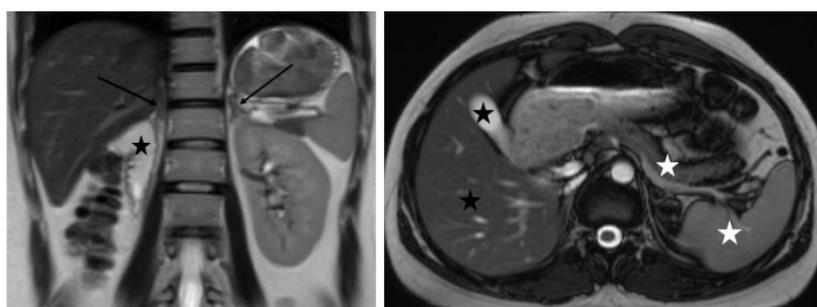
reveal absence of uterus and cervix with preserved vaginal stump (Fig-1). T2 weighted fast spin echo sequences with and without fat suppression also showed normal appearing both ovaries with multiple hyperintense subcentimetric follicles (Fig-2). Coronal and axial T2 weighted sequence shows absence of right kidney with normal appearing left kidney, adrenals and upper abdominal organs including liver, gall bladder, spleen and pancreas. Sagittal and coronal T2 weighted sequence show normal appearing dorsolumbar spine, pelvis including both hips and sacroiliac joints underview.



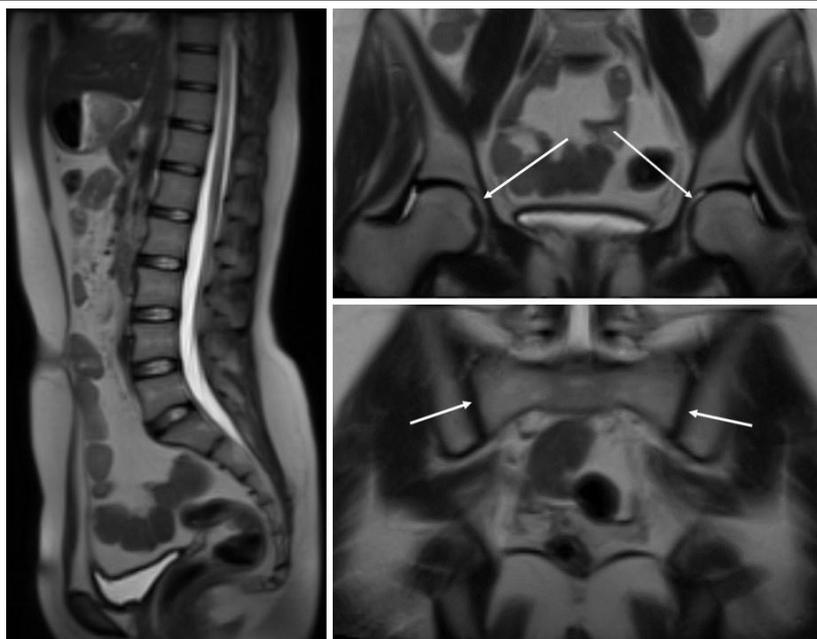
**Fig-1:** Axial T2 weighted images show absence of uterus in expected location (thin white arrows) between urinary bladder anteriorly (black asterisk) and rectum posteriorly (white asterisk). The vaginal stump was however visualised in lower pelvic section (thick white arrow)



**Fig-2:** Axial T2 images show normal appearing ovaries showing multiple hyperintense follicles (thin white arrows) with same seen in coronal T2 fat saturated image (thin white arrows)



**Fig-3:** Coronal T2 weighted image shows normal appearing both adrenal gland (both thin black arrows), absent right kidney with duodenum (black asterisk) occupying right renal fossa. The left kidney appears normal. Axial T2 weighted image shows normal appearing liver, gallbladder (both black asterisk), spleen and pancreas (both white asterisk)



**Fig-4: Coronal T2 weighted image shows normal appearing lumbar and visualised dorsal spine. Coronal T2 weighted image shows normal appearing both hip and sacroiliac joints (both thin white arrows)**

#### DISCUSSION

MRKH syndrome is a congenital syndrome and one of the common causes of primary amenorrhoea. The incidence of this condition is 1:400 live female births majority of cases being sporadic [1]. The etiology of this condition is poorly understood postulated being polygenic and multifactorial. Mullerian ducts are the precursors of female reproductive system organs with uterus, cervix, upper half of vagina and fallopian tubes originating from it. As per the American fertility society classification of mullerian duct anomalies MRKH syndrome is classified as type I, being most commonly occurring anomaly with its etiology being arrested development of mullerian duct around 7<sup>th</sup> week of intrauterine gestation [2]. The karyotype of these patients is 46XX [3]. Primary amenorrhoea is the presenting symptom in vast majority of cases where uterine tissue is completely absent. On physical examination the external genitalia appear normal, normal secondary sexual features and normal ovarian hormonal profile implying intact ovarian function. There are two subtypes of MRKH syndrome type I and II also known as typical and atypical forms differentiation among them being important as associated extragenital anomalies are seen in latter only. The typical form or type I affects only internal genitalia seen as uterine aplasia whereas the atypical form or type II has additional anomalies, kidney being most commonly implicated seen as unilateral agenesis, renal ectopia or fusion anomalies like horseshoe kidney. There can also be involvement of skeletal system in the form of vertebral anomalies, hearing defects secondary to middle/inner ear anomalies along with increased predisposition for ovarian malignancy [3]. This type is more common than type I and also known as Mullerian renal cervical somite or MURCS. In our case there was

uterine aplasia along with unilateral right renal agenesis and no musculoskeletal or pulmonary abnormality with no underlying ovarian mass lesion. Karyotyping, Imaging and or laparoscopy are used for diagnosis and further evaluation of this condition before embarking upon definite treatment. Among imaging ultrasonography is used as first line of investigation having advantage being easily available and low on cost providing straightforward information regarding status of uterus and ovaries and simultaneous assessment of kidneys in same sitting [6,7]. However on the downside ultrasonography being operator dependant the results may be inconsistent and conflicting especially in cases with ectopic location of pelvic organs. This information is critical for surgical management. One such case of bilateral uncomplicated ovarian hernia was seen in which both ovaries were seen lying within inguinal canal investigated primarily with ultrasonography and colour doppler followed by MRI evaluation [8]. Cross sectional imaging is next in line with computed tomography being faster and relatively expensive, not routinely performed due to use of ionizing radiation and iodinated contrast administration. Few specific conditions in which computed tomography finds utility is evaluation of ovarian neoplasms in reference to detection of calcification or small amount of fat, detection of cervical vertebral fusion anomalies [3,7,9]. Another rare association of pulmonary hypoplasia with MRKH syndrome is described showing ipsilateral mediastinal shift [10]. MRI is the best imaging modality for non-invasive evaluation of MRKH syndrome. MRI is advantageous in being non-invasive, devoid of ionizing radiation and iodinated contrast administration and at the same time having superior contrast resolution of soft tissue structures with multiplanar capability [11]. The key role of MRI in such cases is answering whether

uterus is absent or present and when present is hypoplastic and/or rudimentary [7,12]. MRI clearly demonstrates uterine morphology seen as absence of uterus and upper two-third of vagina by virtue of having common embryology with associated primary amenorrhoea. In a minority of cases where rudimentary uterine bud if present and functional the patients present with clinical symptoms of cyclical abdominal pain and lower abdominal distension. The MRI spectrum in MRKH syndrome ranging from three layered morphology to eccentric nodule to no trilaminar appearance. The features suggestive of functional endometrial tissue include presence of blood products or features of adenomyosis which not necessarily signify reproductive potential [12]. In all cases presence of T2 hyperintense follicles signify functional ovarian tissue. Another value area of MRI is in evaluation for possibly ovarian neoplasm of varied histology known to be associated in cases of MRKH syndrome. However, the downside of MRI is its inability to differentiate between polycystic ovarian disease and multilocular ovarian neoplasm [3]. MRI was postulated to be 100% sensitive and specific on laparoscopy for the diagnosis of MRKH syndrome [4]. MRI was also recommended as first line of investigation in MRKH syndrome by American College of Obstetricians and Gynaecologists [5]. Other than evaluation of MRKH syndrome including subtyping another important application of MRI is to rule out testicular feminization syndrome also known as androgen insensitivity syndrome. The latter shows absence of both uterus and ovaries with presence of rudimentary ectopic testis [6]. There are no standard set protocols for imaging in a case of MRKH syndrome however few principles are required to be taken under consideration beforehand. The sequences are required to be acquired in all three planes for proper evaluation. Sagittal and axial T2 weighted fast spin echo sequences both with and without fat suppression are used to evaluate degree of uterine hypoplasia. Small FOV T2 weighted sagittal fast spin echo sequence is also used for evaluation of uterine zonal anatomy. Axial T2 weighted fast spin echo sequence is used for vaginal atresia. Coronal T1 or T2 weighted fast spin echo sequence with large field of view is used for evaluation of associated renal anomalies if present. Axial and coronal T2 weighted fast spin echo sequences are of particular importance in localisation of ovaries [1]. T1 weighted pre and postcontrast fast spin echo sequences along with diffusion weighted sequence are useful when pelvic mass is suspected [3].

## CONCLUSION

MRKH syndrome is a rare congenital anomaly diagnosed invariably at or after puberty with presenting complaints of primary amenorrhoea. Imaging plays a key role in its diagnosis, subtyping, evaluation of associated systemic anomalies as well as management. Ultrasonography is used as first imaging modality with colour doppler in selected cases. Computed tomography

is useful in selected cases when pulmonary or skeletal anomalies are in question. MRI is the imaging modality of choice depicting morphology of genital and extragenital organ system with greater contrast resolution and exclusion of underlying ovarian neoplasm.

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