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Mayer-Rokitansky-Kuster-Hauser Syndrome with Unilateral Renal Agenesis

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Abstract: Mayer Rokitansky Kuster Hauser syndrome is a malformation of the female genital tract which occurs due to interruption in the embryonic development of the paramesonephric ducts characterized by agenesis of uterus and vagina in females with normal ovaries and fallopian tubes, normal secondary sexual characteristics and 46 XX karyotype. We reported a case of MRKHS In 18 years old female who presented with primary amenorrhea stressing the role and benefit of imaging in differential diagnosis.

Key words: Mayer Rokitansky Kuster Hauser Syndrome (MRKHS) · Primary Amenorrhea · Mullarian Duct Anomaly

INTRODUCTION

Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome was introduced by Meyer in year 1829 and by Rokitansky in year 1838. They described it as a syndrome which included agenesis of the uterus and vagina due to abnormal development of the Mullerian ducts. Rokitansky later reported uterine and vaginal agenesis and Meyer described various vaginal duplications as additional features. In 1910, Kuster recognized urologic associations, such as renal ectopy or agenesis, along with skeletal abnormalities [1].

Case: 18 years old female with history of primary amenorrhea was presented to department of radiology sir ganga ram hospital Lahore with request for ultrasound abdomen pelvis.

On interrogation she gave history of thelarche at age of 12 years and appearance of axillary hair at 13 years. There was no history of cyclic pain, any systemic ailment or of prolonged use of any drug. On examination there were no signs of stunted growth. Her secondary sexual characteristics appeared well developed including normally developed breasts and external genitalia.

Her ultrasound was performed with full bladder. A well-developed uterus could not be identified. Left ovary was visualized normally. However there was nonvisualization of right ovary. On ultrasound abdomen her right kidney was not seen at the anatomical or any ectopic site. Left kidney having normal size and echogenicity was seen at the anatomical site. Rest of abdominal viscera was unremarkable. On basis of these sonographic findings MRKHS was suspected and MRI pelvis was advised for further confirmation. MRI Pelvis axial and sagittal T1 and T2 Weighted images were performed which confirmed the above mentioned findings along with showing the presence of a blindly ending distal vagina on axial Images.

DISCUSSION

MRKHS is a rare congenital malformation of the female genital tract which occurs due to developmental arrest of mullerian ducts in the embryonic stage at about 7 weeks of fertilization [2, 3]. Mullarian ducts are primordia of female genital tract and they differentiate into uterus, cervix upper 2/3rd of vagina and fallopian tubes [2]. Interruption in development of Mullarian ducts thus results in absence or hypoplasia of these structures. It affects at least 1 out of 4500 women[3]. Its penetrance varies as does involvement of other systems [4].

MRKHS is mostly sporadic although few familial cases have also been reported [5]. It is the second most probable cause of primary amenorrhea (after gonadal dysgenesis) [5] and the diagnosis is usually considered when a patient who is having normal secondary sexual

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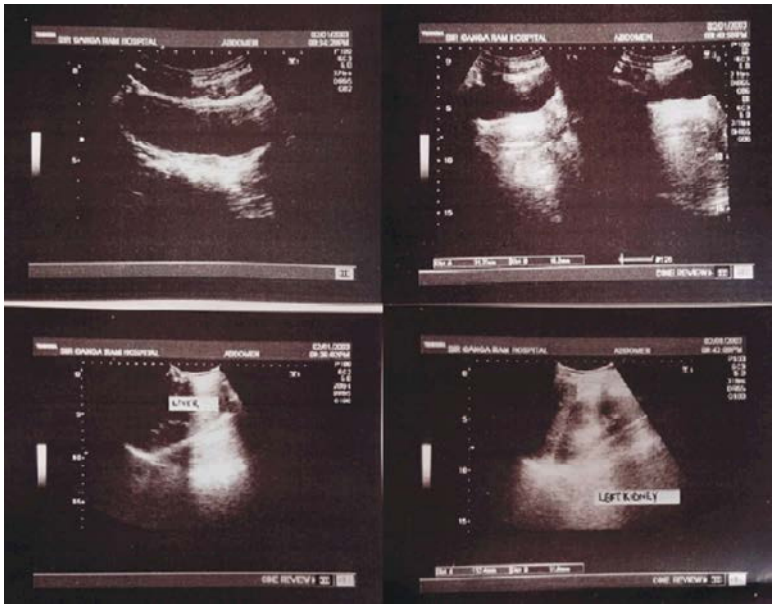

Fig. 1: (a) Transverse ultrasound image showing absence of uterus between bladder and rectum (b) Demonstrates normally visualized left ovary with non-visualization of ovarian tissue in right adnexal region (c)Demonstrates absent right kidney in hepatorenal fossa, (d) With normally visualized left kidney.

characteristics presents with history of primary amenorrhea and her primary radiological investigation are unable to demonstrate normally developed uterus. MRKHS has characteristic findings of:

- Absence or hypoplasia of uterus, cervix and upper third of vagina
- Normal secondary sexual characteristics including breasts, external genitalia and pubic and axillary hair growth
- Normally functioning ovaries
- Normal female phenotype 46XX

Thus supplemental tests including hormonal and chromosomal analysis in these patients will confirm normal levels of FSH and LH and normal 46 XX karyotype respectively [6].

MRKHS is classified into two broad categories according to the involvement of structures:

Type A: Typical, only characteristic features of MRKHS identified, no associated anomalies are seen in this type and

Type B: Atypical, In this type there are associated anomalies that may involve ovaries, fallopian tubes, kidneys (unilateral agenesis, horseshoe etc), bones usually vertebrae (scoliosis, vertebral fusion defects, klippel feil syndrome etc.) and heart.

This condition must be differentiated from androgen insensitivity syndrome in which patient is phenotypically male (46 XY) with virilization of his external genitalia causing female like appearance, by demonstrating presence of ectopic testis along with absence of uterus and ovaries[7]. Other differentials include isolated vaginal hypoplasia, gonadal dysgenesis and WTN syndrome

Ultrasound is usually the 1st line of investigation in workup for patients presenting with primary amenorrhea as it is easily available, inexpensive and non-invasive method which does not require any radiation exposure [7]. Ultrasound will help us confidently reveal absence of uterus between urinary bladder and rectum [4] and it may also be helpful in categorizing the subtype of MRKHS by identifying renal and ovarian anomalies. However ultrasound is an operator dependent technique and may require experienced personal [4]. In the case of ectopically located ovaries ultrasound may be unable to recognize ovaries[4]and may falsely classify it as type II MRKHS.

Although CT may provide useful information it is not routinely performed to avoid use of ionizing radiation in young female patients [2].

MRI is a noninvasive technique which has much superior contrast resolution for soft tissue structures and together with its multiplanar capability and lack of ionizing radiation [2], it has proven to be much more sensitive and specific than ultrasonography in diagnosing MRKHS. MRI has proven to be comparable to and much more cost

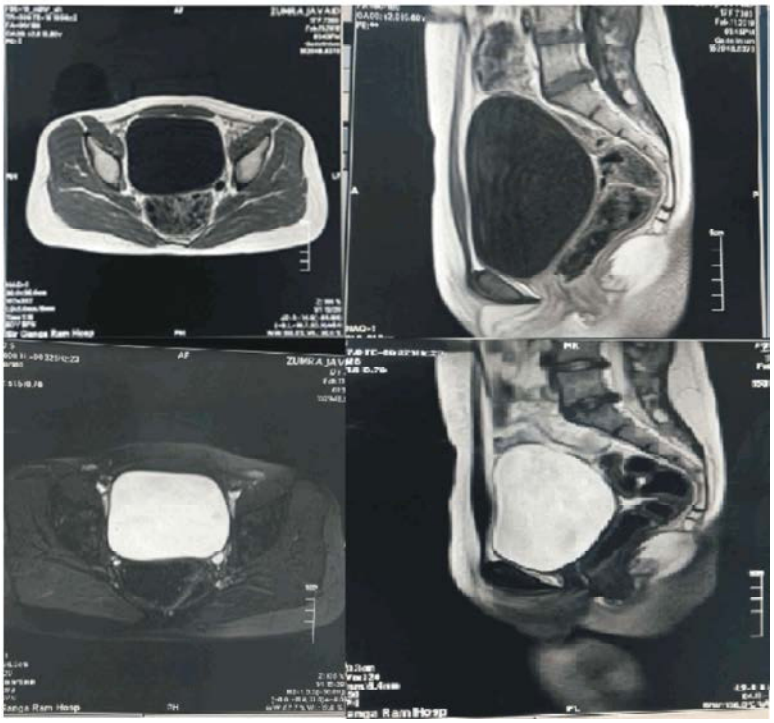

Fig. 2: (a),(b) Axial and sagittal T1WI (c)(d) Axial and sagittal T2WI Fat/Sat images showing absence of uterus between bladder and rectum.

effective than laparoscopy in diagnosing the condition [2]. MRI has now become the imaging modality of choice in the case where MRKHS is suspected as it provides us with the detailed anatomy and morphology of female internal genital organs [4] and can also identify associated renal, vertebral and even cardiac anomalies. Axial MRI images are best in demonstrating vaginal aplasia whereas absence of uterus is best confirmed on sagittal images [1, 4].

As it is an irreversible condition thus screening for associated renal and skeletal abnormalities and physiological counselling are the mainstay of its management. The aim of surgical management includes creation of a neovaginal canal having adequate length and width to allow intercourse [8].

Reproduction is also possible in these patients using assisted techniques, due to presence of functioning ovaries.

CONCLUSIONS

MRKHS is common cause of primary amenorrhea which can be radiologically identified using ultrasonography as primary mode of evaluation and MRI being usually used for confirmation or a problem solving tool in case of inconclusive sonographic findings. MRI has proven to be much more sensitive and specific in diagnosing this condition due to its superior soft tissue contrast resolution, multiplanar capability and no operator dependence. Although this condition is irreversible and thus physiologically straining however surgical creation of neovagina and assisted reproductive techniques are now playing important role in normalizing the reproductive life in these patients.

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