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Case Report

A rare case of Mayer-Rokitansky Küster-Hauser syndrome with ectopic kidney

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ABSTRACT

Mayer-Rokitansky Küster-Hauser (MRKH) syndrome is a congenital anomaly of female reproductive system and is the second most common cause of primary amenorrhea. Utero-vaginal agenesis also may be associated with different other congenital anomalies and mostly involve the renal and skeletal system. Karyotyping and gonadal development are normal. So, patient's anthropometric measurement, secondary sexual characteristics and hormonal profile are usually normal. Transabdominal sonography and MRI can confirm the absence of uterus, cervix and upper part of vagina, as well as can identify another associated congenital anomaly. We report the case of 25-year-old women presented with primary amenorrhea and normally developed secondary sexual characteristics with normal external genitalia. Transabdominal sonography and MRI revealed agenesis of uterus, normal ovary and ectopic right kidney. MRKH syndrome with ectopic kidney is a rare form of mullerian agenesis.

Keywords: Mayer-Rokitansky Küster-Hauser syndrome, Ectopic kidney, Primary amenorrhea

INTRODUCTION

Mayer-Rokitansky Küster-Hauser (MRKH) syndrome is a rare congenital anomaly of 46, XX female characterized by utero-vaginal agenesis.[1] Exact underlying cause is still unknown but WNT4 genetic mutation, familial predisposition and discordance in monozygotic twin are the probable cause.[5] During embryogenesis interrupted Mullerian duct differentiation leads to agenesis of uterus and vagina but ovarian and external genitalia development is normal as they developed from different embryonic source.[2] So, patient typically present with primary amenorrhea with normal height and secondary sexual characteristics. Depending on the associated congenital anomaly with utero-vaginal agenesis it is classified into three types.[3,4] Type three (MURCS) is more severe form where Müllerian duct aplasia is associated with renal aplasia, cervicothoracic somite dysplasia, renal, skeletal, and cardiac malformations, and hearing impairment. Renal and skeletal anomalies are more commonly associated with this syndrome.[6,7] Chromosomal analysis and hormone profile is mostly normal.[9] Transabdominal ultrasonography and pelvic MRI can identify the genital organs and malformation as well as confirm the diagnosis.[10] Management involves multiple discipline to improve the quality of life, vaginal reconstruction but fertility issue is still not satisfactory other than assisted reproductive technology.[11]

CASE REPORT

A 25-year-old women presented to Endocrinology department of a tertiary level hospital in Bangladesh for evaluation of primary amenorrhea. She had normal pubertal development with thelarche at the age of 10 year and pubarche at the age of 11. Thereafter pubertal progression was normal but she never felt any cyclical lower abdominal pain. Her mother's natal and postnatal period was unremarkable and she was born by full term vaginal delivery. No other family member had such type

of illness or any congenital anomaly. Her medical, surgical and drug history was also unremarkable. Physical examination revealed normal body habitus with height 160 cm, weight 56 kg, arm span 159 cm. She had phenotypically normal female genitalia and her vitals were normal. Sexual maturation scale showed Tanner stage- B5 P4. Speculum examination was not done. Her routine laboratory tests including complete blood count and hormone profile were within normal limit (Table 1). She had normal female karyotype of 46 XX.

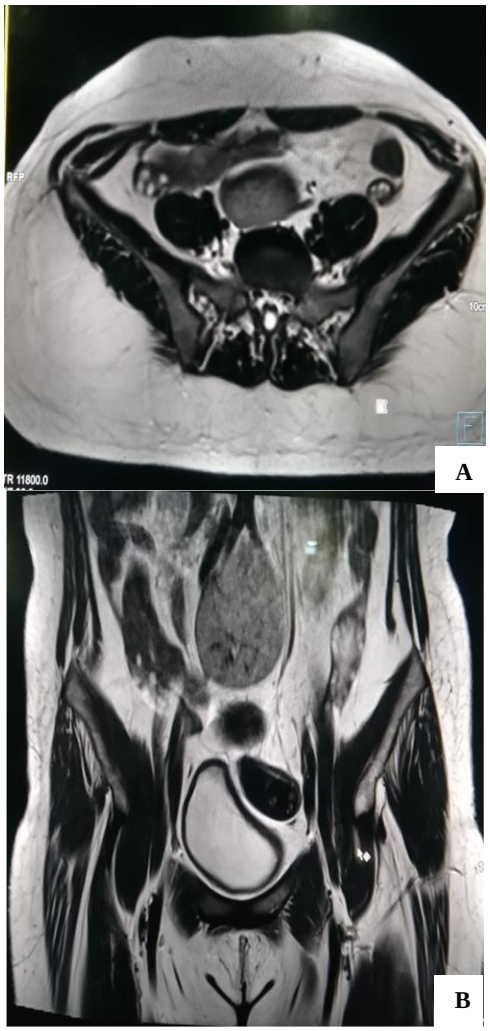

Figure 1: MRI of pelvis T1-sequence without contrast shows; blue arrow- ectopic right kidney, green arrow-ovary. A) Axial view. B) Coronal view.

Transabdominal sonography could not identify uterus and ovaries in pelvic cavity rather right kidney was located in the right side of pelvic cavity and left kidney in normal position. Subsequently, MRI of pelvis with and without contrast was done which revealed vaginal canal was visible but uterus was not visualized, two ovaries were seen at the level of L5-S1 (right ovary measuring 2.4×1.1 cm and left ovary measuring about 2.2×1.1 cm), right kidney was noted at prevertebral region of lower abdomen (measuring about 8.7×3.2 cm) at the level of L4-L5 and left kidney was on normal position. After IV contrast (Gd-DTPA) administration, no abnormal enhancement was noted (Figure 1). Based on the above findings we diagnosed the case as type-2 MRKH syndrome and planned for multidisciplinary approach for her management involving Urology department for vaginal reconstruction, Psychiatry for psychological support and we also explained her about her future fertility.

Table 1: Hormone profile of the patient.

Hormones	Patient's value	Reference value
Serum estradiol, pg/ml	430	21.0-251.0
Serum luteinizing hormone (LH), mIU/ml	3.59	1.10-11.60
Serum follicle stimulating hormone (FSH), mIU/ml	1.68	2.50-10.20
Serum prolactin, ng/ml	6.06	2.80-29.20
Serum total testosterone, ng/ml	0.62	0.54-0.71
Serum thyroid stimulating hormone (TSH), µIU/ml	1.86	0.35-5.50
Serum anti-mullarian hormone (AMH), ng/ml	4.59	1.50-4.0

DISCUSSION

Mayer-Rokitansky Küster-Hauser (MRKH) syndrome is an uncommon congenital anomaly of unknown etiology where there is utero-vaginal agenesis in a 46 XX karyotype female with normal secondary sexual characteristics. It is the second most common cause of primary amenorrhea and the prevalence is 1 in 5000 females. Though exact cause is unknown, discordance in monozygotic twin, WNT4 gene mutation and familial occurrence are the probable genetic and nongenetic basis of disease occurrence.[5] We could not perform genetic analysis for our patient owing to unavailability of local facility and financial constraint. In our case there was no familial occurrence as well.

During fourth to twelfth weeks of gestation Wolffian duct regress normally but due to interruption of Mullerian duct development fallopian tube, uterus and upper two-third of vagina become hypoplastic or aplastic. Ovarian development is normal as because ovary developed from different source.[2] So, patients typically present with primary amenorrhea, difficulty in sexual intercourse and sometimes cyclical abdominal pain. As ovarian development and function is normal anthropometric measurement, secondary sexual characteristics and external genitalia including labia majora, labia minora and lower part of the vagina appear normal.[3,4]

MRKH is classified into three types: in type I/typical MRKH there is isolated uterovaginal agenesis, in type II/atypical MRKH renal and ovarian malformations, and in the third type there is Müllerian duct aplasia, renal aplasia, and cervicothoracic somite dysplasia (MURCS)

association with renal, skeletal, and cardiac malformations, and hearing impairment.[6] Index case was type-2 MRKH syndrome, as patient had uterovaginal agenesis associated with renal malformation (ectopic right kidney). Most common congenital anomalies involve skeletal system and renal system including ectopic kidney, renal agenesis, renal malformation, duplex kidney etc. Some may present with ectopic ovaries.[6,7] Clinical information, Transabdominal sonography, MRI imaging help to stablished the diagnosis. Chromosomal analysis is normal in 97.1% cases and hormonal profile is normal in 93.8% cases.[9] It is differentiated from androgen insensitivity syndrome by the presence of pubic and axillary hair, normal testosterone level and 46, XX karyotype. Ultrasonography (USG) is usually the first line investigation to identify the pelvic organs but sometimes report may be inconclusive. So, MRI is the choice of investigation to identify the pelvic organs and other malformation to confirm the diagnosis.[10]

In the index case uterus and ovary could not be visualized by USG but both ovaries and right kidney found in ectopic position. Because of the absence of menstruation and future fertility issue, diagnosis of MRKH syndrome affect markedly the psychological aspect and quality of life. Management should be interdisciplinary approach, involving psychiatrist for psychological counselling, surgical intervention for vaginal reconstruction and assisted reproductive technology for those what to have children. Intervention for ectopic kidney usually not required if there is no associated renal complication like pelvic ureteric junction (PUJ) obstruction.[11,12]

CONCLUSION

Patients of MRKH syndrome present typically with primary amenorrhea with normal secondary sexual characteristic. It is associated with different congenital anomaly of uterus, vagina skeletal and renal system. So, patients who present with primary amenorrhea should search for another associated congenital anomaly. Multidisciplinary treatment approach can improve the conjugal life and quality of life.

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