CASE REPORT

MAYER ROKITANSKY KUSTER HAUSER SYNDROME WITH UROGENITAL SINUS ANOMALY

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ABSTRACT

Mayer Rokitansky Kuster Hauser (MRKH) syndrome is a rare disorder, characterized by the congenital absence of uterus and associated renal tract anomalies. The case presented with primary amenorrhea and primary infertility, despite development of normal female secondary sexual characteristics. CT scan revealed absent uterus, a solitary left sided pelvic kidney and a vesicovaginal communication that, on cystoscopy, revealed urogenital sinus anomaly manifesting as a common channel formed due to absent anterior wall of vagina and posterior wall of urethra. The urogenital sinus anomaly in MRKH syndrome has not been reported earlier.

KEY WORDS: *Mayer-Rokitanski-Küster-Hauser syndrome. Renal agenesis. Pelvic kidney. Urogenital sinus anomaly.*

INTRODUCTION

Mayer-Rokitanski-Küster-Hauser (MRKH) syndrome, characterized by the mullerian duct anomaly is a rare disorder. The prevalence has been reported as one in 4000 - 5000 female births. Congenital absence of upper vagina, uterus and cervix is the main feature of the disease which, in addition, is often found associated with renal tract anomalies.[1] These malformations are the source of intense concern for both the patient and her family due to their repercussions on reproductive health as well as psychological and sexual impact. Initial diagnosis of these anomalies is made usually during adolescence.[2] Patients usually present with normal pubertal development, primary amenorrhea and cyclic abdominal pain. Other concerns that prompt medical attention include infertility or inability to have intercourse. The syndrome was described as the agenesis of the uterus and vagina due to abnormal development of the müllerian ducts; uterine and vaginal agenesis were reported by Rokitansky, while Mayer described some vaginal duplications. In 1910, Küster recognized urologic associations of this syndrome , such as renal ectopy or agenesis. It has also been suggested that this syndrome is due to a deficiency of the estrogen receptors, which may inhibit further development of the embryonic mullerian duct.[3]

CASE REPORT

A 24 years old married lady presented in gynaecology out-patient department for the treatment of primary infertility with history of primary amenorrhea and leakage of urine from vagina since birth. There was no history of dyspareunia. There was no family history of congenital urogenital disorders. On examination, her clitoris was normal looking but external urethral meatus was rudimentary, small and located much below the clitoris. Her pubic and axillary hair were normal. Her breasts were well-developed and there was no galactorrhoea. On per vaginal examination, her vagina was short in length and communication with urinary bladder was felt in the upper anterior wall of vagina as two fingers could easily be inserted into bladder through vesico-vaginal communication. Her uterus and cervix could not be palpated. Her renal function tests were normal and her hormonal profile revealed normal estrogen, progesterone, luteinizing hormone, and follicle-stimulating hormone values. Her ultrasound revealed absent kidneys in both renal fossae with a solitary left sided pelvic kidney. Her contrast enhanced computed tomographic scan (CT) revealed a solitary left sided normally functioning pelvic kidney surrounded by bowel loops (Figure 1). The inferior portion of pelvic kidney was resting against sacrum and iliac bone and superolateral portion was abutting the urinary bladder. No hydronephrosis or hydroureter was seen. Ureter of the pelvic kidney was inserting in the urinary bladder on the left side.Uterus and cervix were absent and there location was

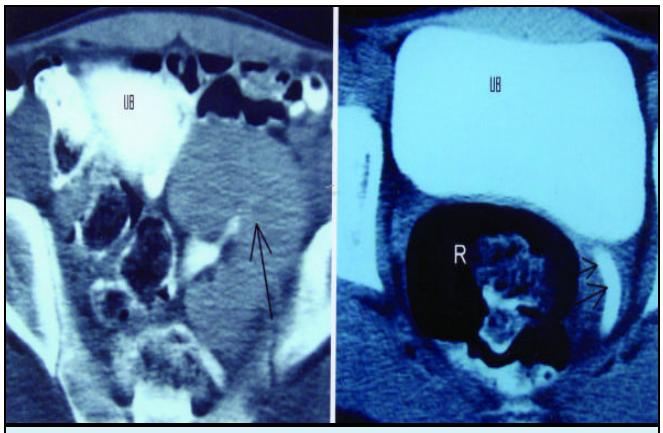

Figure 1: CT scan pelvis. A solitary normally functioning pelvic kidney (single arrow). No uterus seen.Notice rectum (R) occupying the expected location of the uterus posterior to urinary bladder (UB). Lt ureter (double arrows) opening into the urinary bladder.

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occupied by the rectum.CT scan also revealed direct communication between bladder and vagina (Figure 2). Contrast in the vagina was seen in the lower scans of the pelvis as well. No skeletal or spinal malformation was seen and the case was referred to Armed Forces Institute of Urology.

At the AFIU, her cystoscopy was performed. It revealed urogenital sinus anomaly as posterior wall of the urethra and

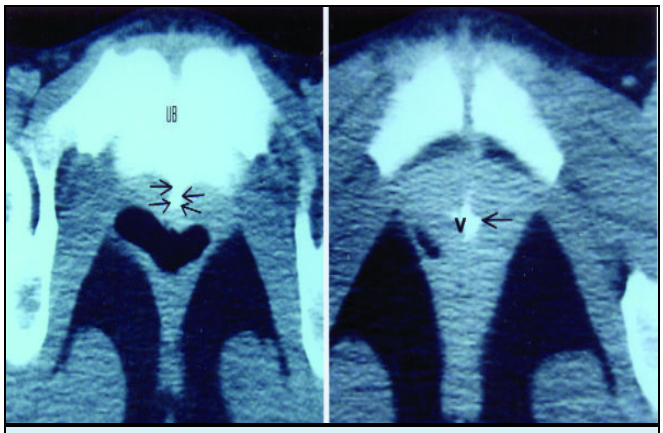

Figure 2: CT scan pelvis. Urinary bladder is draining into the common channel formed due to urogenital sinus anomaly (double arrows). Contrast can be seen in the common channel (v) (single arrow).

anterior wall of vagina were absent thus, forming a common channel. Urinary bladder was opening into this common channel. Bladder neck was also absent. Chromosome analysis of the patient revealed normal 46XX karyotype thus differentiating it from testicular feminization syndrome. Her laparoscopy revealed normal fallopian tubes and ovaries situated on the pelvic sidewalls but absent uterus and cervix. The patient was informed of her anomaly in detail and was offered surgical repair of the vesico-vaginal communication with formation of a separate drainage pathway for the urinary bladder. The patient, however, refused undergoing any treatment once she knew that she could not conceive even after surgical correction and was discharged on request.

Discussion

MRKH syndrome is characterized by congenital absence of mullerian duct structures. The renal abnormalities in MRKH syndrome include unilateral renal agenesis, pelvic kidney, renal hypoplasia and horseshoe kidney.[4] It can be rarely associated with skeletal and spinal malformations. Congenital scoliosis may also be seen in MRKH syndrome. These patients typically present with primary amenorrhea and cyclic abdominal pain. Other clinical presentations include inability to have intercourse, infertility, and voiding difficulties or recurrent urinary tract infections secondary to renal anomalies. In some patients with this syndrome, only a shallow vaginal pouch is present. The more shallow the canal, the greater the likelihood of the patient having dyspareunia.These patients require surgical correction including dilation of the vaginal pouch or grafting a vaginal canal. Normal sexual functioning can be attained after surgical reconstruction, although conception cannot occur.

Patankar et al have described a case of rectovestibular fistula in Mayer-Rokitansky syndrome with symmetrical mullerian structure agenesis and renal anomalies.[5] Pittock et al. described 25 patients of MRKH syndrome between 1975 and 2002, with different renal, skeletal and cardiac defects.[6] Every diagnosed patient of MRKH must undergo thorough investigations for associated anomalies. Women with mullerian agenesis show normal 46 XX karyotype and functional ovaries.[7] Laparoscopy is useful in confirmation of MRKH and helps in planning the definitive reconstructive surgery. Another important issue of concern is counseling of parents. Enormous efforts are taken to counsel the parents about the complex anatomical problem, associated anomalies, the method of reconstruction and the risks and problems involved. They are made aware that there will not be any menstruation and conception and that the procedure is mainly for sexual activity.

Psycho-social rehabilitation of a child, having this complex problem, should be done. Laparoscopy is often used in the diagnostic process or to remove remnants of uterus. Magnetic resonance imaging is more precise than laparoscopy and sonography in defining the anatomical characteristics of MRKH syndrome, and is less invasive and expensive than laparoscopy.[8] Detailed history, through clinical examination and minimal investigations are required to diagnose cases of primary amenorrhoea.[9] Genetic link for MRKH syndrome has not been demonstrated and female relatives of the patient apparently have no increased risk.[10]

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