A rare case of Mayer–Rokitansky–Kuster–Hauser (MRKH) syndrome with solitary ectopic pelvic kidney and uretropelvic junction (UPJ) obstruction

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Introduction

The Mayer–Rokitansky–Kuster–Hauser (MRKH) syndrome results from failure of the paramesonephric (Müllerian) duct formation and occurs at a ratio of 1:4500–5000 female newborn. These patients have normal secondary sexual characteristics, external genitalia, sexual hormonal profile, and karyotype. There are two subtypes of MRKH syndrome: type A, having isolated uterine aplasia without associated anomalies, and type B, which is associated with other gynecological anomalies (abnormal fallopian tubes) and non-gynecological anomalies. The renal involvement is the most common, which occurs in 40% of patients. We report a rare case of MRKH with solitary ectopic pelvic kidney and uretropelvic junction (UPJ) obstruction with acute kidney injury and post obstructive diuresis.

Case report

This is a 23 year-old female patient known case of Mayer–Rokitansky–Kuster–Hauser (MRKH) syndrome presented to the emergency department with recurrent vomiting and lower abdominal pain for 5 days, associated with constipation. The patient received multiple oral analgesia before she presented to our hospital. The patient denied any similar episode previously. She had of primary amenorrhea and was evaluated by a gynecologist for the primary amenorrhea when she was 17-year-old. At that time she was examined and evaluated by the hormonal profile, MRI and Chromosomal analysis. The diagnosis was established in 2013 by the gynecologist with a diagnostic laparoscopy which revealed the absence of the uterus and the presence of both ovaries and a pelvic kidney.

The patient was vitally stable afebrile. The abdominal examination revealed lower abdominal tenderness with palpable mass. The genit examination revealed normal external genitalia. The laboratory investigations showed marked elevation in the creatinine 522 μmol/L and BUN 14.1 mmol/L. U/S abdomen and pelvis was done and revealed absence of uterus with solitary pelvic kidney. There was severe hydronephrosis with preserved parenchyma and normal cortico-medullary differentiation (Fig. 1). The CT scan of the abdomen showed solitary left pelvic kidney with severe hydronephrosis. There was neither ureteral dilation nor renal or ureteric stones (Fig. 2).

The patient was admitted as a case of uretropelvic junction obstruction of a solitary pelvic kidney with acute kidney injury. She underwent an emergency cystoscopy with left retrograde pyelography, which revealed absence of the right ureteric orifice, normal left ureteric orifice shape and position and severely hydronephrotic left pelvic kidney with high insertion of the ureter, UPJ obstruction and normal caliber of the ureter (Fig. 3). There was failure of drainage after 10 minutes of contrast injection, so a double J stent was inserted.

After the operation, the patient passed 500 ml of urine in the first hour and another 500 ml in the second hour, so she was diagnosed as a case of post-obstructive diuresis. Fluid replacement was started, and the patient was monitored closely. The Creatinine improved over the next day and normalized in the third post-operative day. The patient was screened for other skeletal and cardiac anomalies, which were normal. The karyotype was done for the patient which was normal 46 XX. The patient was managed as an emergency case of UPJO and acute kidney injury.
injury with the insertion of the ureteric stent. She is planned for open pyeloplasty for a definitive management after a few weeks.

Discussion

The MRKH syndrome results from failure of the Müllerian duct development and results in lack of uterine formation, with or without other systemic anomalies. The syndrome was named after those who described the syndrome in the 19th and 20th centuries, namely Mayer, Rokitansky, Kuster, and Hauser and Schreiner. It is categorized as type A (typical) and type B (atypical) subtypes. While type A is characterized by isolated absence of both the uterus and upper two-third of the vagina, type B has been reported to be associated with renal (40%), skeletal (12%), cardiac and/or ear anomalies. Type B is also called MURCS association (Müllerian, Renal and Cervical Somite). Patients with MRKH syndrome have normal external genitalia, normal secondary sexual characteristics, normal hormonal profile and normal karyotype.

For type B patients, the renal anomalies varied between abnormal number (agenesis), position (ectopia), and/or shape (horseshoe kidney). Most of MRKH syndrome cases are sporadic; however, some familial cases were reported with autosomal dominant inheritance mode, with both variable expressivity and incomplete penetrance.

To our knowledge, only one case was reported for MRKH with a solitary ectopic pelvic kidney and UPJ obstruction and was managed with open pyelovesicostomy, and another case was reported with bilateral ectopic pelvic kidneys and bilateral UPJ obstruction with highly inserted ureters that were managed with bilateral dismembered

Fig. 1. Ultrasound of the abdomen and pelvis showing empty left (a) and right (b) renal beds with ectopic pelvic kidney with severe hydronephrosis (c).

Fig. 2. CT scan images showing a severely hydroureteric solitary ectopic pelvic left kidney in a axial (a) and a coronal (b) views.
pyeloplasty and ureteral switch.3,4

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**Appendix A. Supplementary data**

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Fig. 3. Intravenous pyelography showing a normal ureter caliber (a) with high insertion of the ureter (b) and a severely hydronephrotic pelvic kidney with immediate and delayed contrast injection (c and d).