

Bilateral Vesicoureteral Reflux in Crossed Unfused Pelvic Kidneys in Mayer-Rokitansky-Kuster-Hauser (MRKH) Syndrome

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Abstract

Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is characterized by hypoplasia or agenesis of uterus and upper one third of vagina with normal secondary sexual characters and normal karyotype 46XX. The incidence of MRKH is 1 in 4000-5000. We report a rare case of Mayer-Rokitansky-Kuster-Hauser (MRKH) with VUR and crossed unfused malrotated right pelvic kidney with left ectopic pelvic malrotated kidney. Other anomalies in this patient were fused cervical vertebra causing short neck, hearing loss, short vagina with agenesis of uterus and upper third of vagina. Patient was operated for bilateral VUR and started on vaginal dilatation.

Keywords: Mayer-Rokitansky-Kuster-Hauser (MRKH); VUR; Pelvic Kidney

Introduction

Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is a rare disorder characterized by anomalies of genitourinary (unilateral agenesis, ectopia of kidneys or horseshoe kidney), skeletal (Klippel-Feil anomaly; fused vertebrae, mainly cervical; scoliosis), cardiac, system. The main feature of MRKH is the absence of uterus and upper third of the vagina [1]. The incidence of congenital absence of the vagina is 1 per 4000 - 5000 female births. MRKH syndrome is a congenital disorder that is present at birth but may remain undiagnosed until adolescence or early adulthood. The diagnosis can be delayed due to the late presentation because of social taboos present in some conservative societies, lack of awareness among healthcare providers and atypical organ involvement [2]. We present a rare case of MRKH with uncommon anomalies not reported before.

Case Presentation

A 22 years female presented with recurrent urinary tract infections, deranged renal function tests, primary amenorrhoea. On blood investigation Haemoglobin 11.5 g/dl, TLC 5500/mm³, urea 32, creatinine 1.9 mg/dl. Ultrasound was suggestive of bilateral pelvic kidneys with the absence of uterus and upper one-third of the vagina. Computed tomography with urography was suggestive of an absent uterus and upper one-third of the vagina. The urinary system anomalies consisted of crossed right kidney to the left part of the pelvis, lying anterior to the left kidney. The right kidney 8 x 3.7 cm and left kidney 6.8 x 3 cm with the orthotopic insertion of both ureters. Both kidneys were small and corticomedullary differentiation was lost. The Renal artery arising from the common iliac artery and rein vein draining

into the left common iliac vein. The ureter of the right kidney crossing to the right side behind the urinary bladder to drain into the posterior wall of the urinary bladder (Figure 1a-1f).

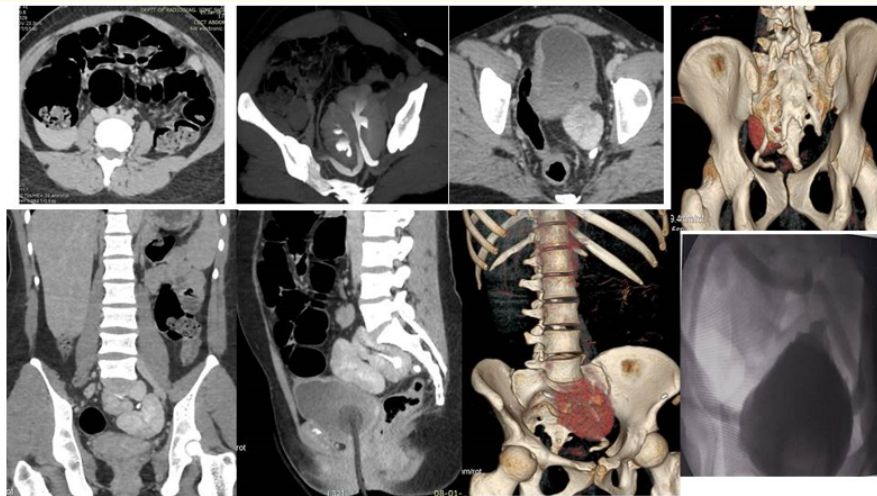


Figure 1: a: Bilateral empty renal fossa. b-f: CT urography and 3 D reconstruction showing both kidneys lying in pelvis posterior to the bladder, sigmoid colon with mal-rotated renal pelvises, coronal section showing both kidneys are facing each other over the upper part of the sacrum.

Hormonal profile, LH, FSH, Prolactin was normal. Karyotype was 46XX. On examination Patient was having a short neck; low hairline, short stature (148 cm), weight 46 kg. The patient was having a low hairline, a fusion of cervical vertebrae, short neck, and restriction of neck motion typical of Klippel-feil abnormality (Figure 2a-2c).

On genital examination small blind-ending pouch (vagina) was present (Figure 2d).

The other anomalies partial sacral agenesis and absence of right sacral foramina appearing as scimitar with bifid L5 spinous process (Figure 2e and 2f).

Micturating cystourethrogram was done suggestive of Grade III reflux in left kidney and grade IV reflux in right kidney. On micturition phase posterior urethra was normal and post-void residual volume was minimal (Figure 3a).

Cystoscopy showed bilateral effluxing ureteric orifices in orthotopic position (Figure 3b).

DMSA scan showed both kidneys in the left pelvis with an irregular cortical outline. Patient's family history was normal except brother having a short neck (fused cervical vertebrae). The patient was initially suspected to have secondary Vesicourethral reflux, however, ultrasound, micturating, and urodynamic study ruled out secondary VUR. The patient was counselled about the disease and opted for vaginal dilatation and bilateral ureteric reimplantation.

Pfannenstiel incision was given. Bilateral kidneys were located in left side of the pelvis, anterior to sacral promontory. Bilateral ureters were reimplanted by Modified Leisch Gregoire technique (Figure 3c and 3d).

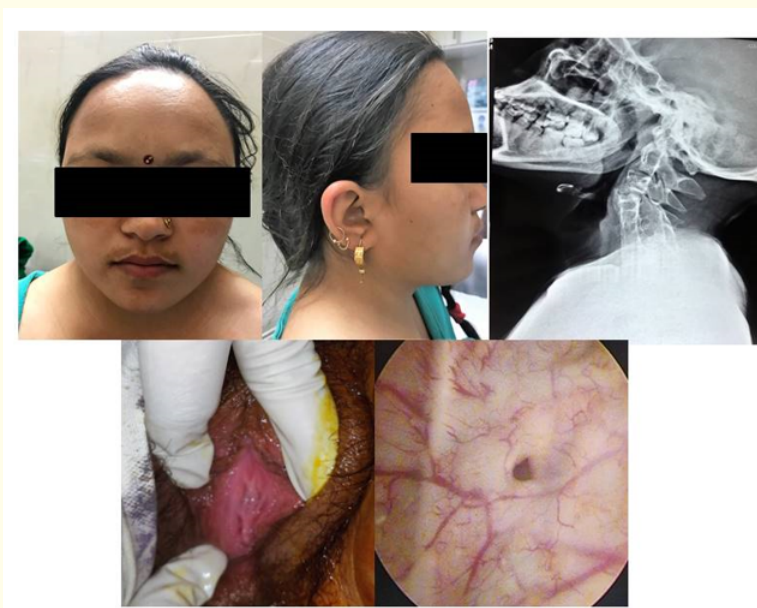


Figure 2: a-b: MRKH patient with short neck. c: X-ray neck show fused vertebra. d: Genital examination shows shallow vaginal pouch. e: Bilateral hydronephrosis on CT urography with ectopic kidneys. f: Partial sacral agenesis.

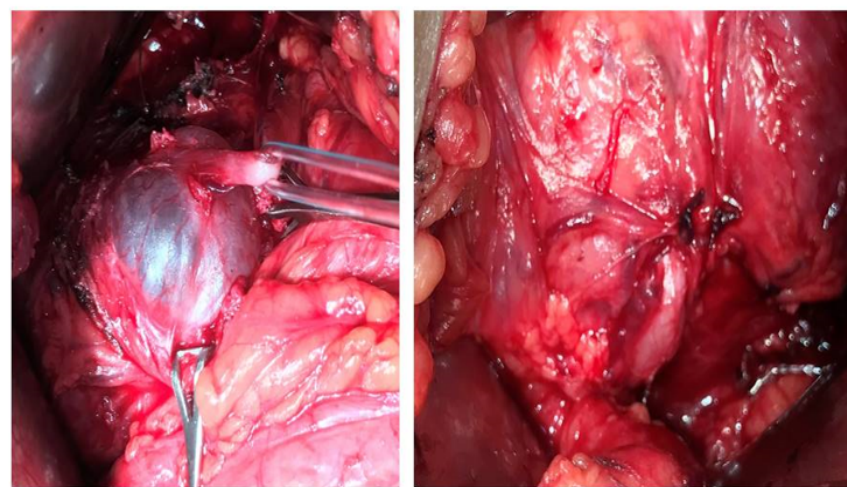


Figure 3: a: MCU showing bilateral vesicoureteral reflux. b: Refluxing ureteric orifice. c, d: Bilateral ureteric re-implantation by modified Leisch-Gregoire technique was done. Ureter dissected up to the ureterovesical junction with creating of an intravesical tunnel.

Postoperatively patient was kept on follow up for CKD and counselled regarding vaginal dilatation. Patient with MRKH was followed at 6 weeks with renal function tests, self-education on vaginal dilatation, counselling regarding fertility and planning for further interventions for genital anomalies. The patient creatinine stabilized on 1.9 mg% and micturating cystourethrogram was normal.

Discussion

Mayer-Rokitansky-Küster-Hauser syndrome is hypothesized to be a result of autosomal dominant inheritance with incomplete penetrance and variable expressivity. Previously MRKH was thought to be a sporadic syndrome, however familial clustering in some patients suggests genetic cause as etiology. MRKH is divided into 2 types according to the involvement of extragenital organs.

Type I (typical) MRKH syndrome is characterized by variable underdevelopment of the vagina and uterus. Type II (atypical) MRKH also incorporates extragenital/extramüllerian malformations, including vertebral, cardiac, urologic (upper tract), and otologic anomalies [1,2].

MURCS (müllerian duct aplasia, renal dysplasia and cervical somite anomalies) syndrome: uterovaginal aplasia or hypoplasia with malformation in the skeletal system and or the heart, muscular weakness and renal malformation In a meta-analysis of 521 cases of MRKH syndrome, Oppelt, *et al.* observed that 64% showed a typical form, 24% were atypical and 12% were of MURCS syndrome.

MRKH present with primary amenorrhoea, cyclic abdominal pain, and primary infertility in young women. In addition, the patient may present with difficulty in intercourse, hearing loss. These patients have a normal secondary sexual characteristic with XX karyotype. In a 25-patient series reported, the frequency of anomalies mentioned included scoliosis (20%), unilateral renal agenesis (28%), non-vertebral skeletal anomalies (16%), and cardiac anomalies (16%) [3].

Male synonym of MRKH syndrome is ARCS (Azoospermia, Renal anomalies, Cervicothoracic Spine dysplasia). Patient brother was having cervical abnormalities so the association with MRKH gene is not ruled out. This also suggests a genetic and familial association [4,5].

The management of these patients involves psychological support, Vecchietti procedure, McIndoe procedure, Sigmoid vaginoplasty, Williams vulvovaginoplasty, offering artificial fertility techniques.

Our patient presented with deranged RFT on evaluation found to have crossed unfused chronically scarred kidneys, with Bilateral VUR and CKD. The patient was operated for bilateral VUR and started on vaginal dilatation. On follow up patient renal function stabilized and the patient continued vaginal dilatation.

Conclusion

We report bilateral vesicoureteral refluxing in ectopic pelvic Kidneys in an MRKH type II patient. The crossed un-fused kidneys reported in an MRKH patient has not been reported before. This patient highlights urological abnormalities and management in MRKH syndrome.

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