CASE REPORT

Mayer-Rokitansky-Kuster-Hauser Syndrome, Type 2 presenting with End Stage Kidney Disease: A rare Occurrence

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Abstract

Background: The association unilateral renal agenesis, renal malformation, kidney disease in residual kidney, uterine agenesis, vaginal atresia and skeletal malformations is a rare occurrence being reported in between 1 in 5000 to 1 in 20,000 live births. It is also known as Mayer-Rokitansky-Kuster-Hauser (MRKH) Syndrome and is as a result of Mullerian duct abnormalities. It is a rare case and is associated with anomalies of the urinary tract, ovaries, kidneys. cervix and vagina. While it has been recognized worldwide, it is rare and this is the first time it is being reported in this environment presenting this late with End Stage Kidney Disease necessitating hemodialysis.

Method: The patient's history, physical examination findings and investigations were carefully evaluated. A diagnosis of End stage kidney disease thought to be as a result of the repeated Urinary Tract Infections and hypertension was made. The patient was re-evaluated in detail and the diagnosis of MRKH syndrome was made.

Conclusion: There is need for clinicians to recognize the associations between primary amenorrhea, the presence of secondary sexual characteristics, recurrent Urinary Tract Infection, skeletal muscle abnormalities as a part of the MRKH syndrome. This is important so that close follow up will be dome early to prevent or delay the onset of end stage kidney failure, as well as to bring together a multi-specialist team to manage the medical, renal, psychological and gynecologic issues that are associated with the syndrome.

Key Words: Mullerman duct abnormalities, Mayer-Rokitansky-Kuster-Hauser Syndrome, Renal Agenesis, Uterine Agenesis, Recurrent Pyelonephritis, End Stage Kidney Disease.

Highland Med Res J 2022;22(1):59-62

Introduction

Mayer-Rokitansky-Kuster-Hauser Syndrome (MRKH) is a relatively rare condition occurring in 1 in 4000-20000 live births. It is characterized by the congenital absence of the uterus and upper two-thirds of the vagina.

The problem is as a result of Mullerian agenesis caused by embryologic underdevelopment of the mullerian duct, with resultant agenesis or atresia of the vagina, uterus or both, and sometimes with associated renal defects. Patients with such are usually identified when they are evaluated for primary amenorrhea with otherwise typical growth and pubertal development. MRKH syndrome usually presents with primary amenorrhea due to the absent uterus, referred to as Type 1.[1,2]

The second presentation involves additional renal and other manifestations, usually a solitary kidney either located in the normal position or in the pelvis. Patients are at risk for repeated urinary tract infections (UTIs), pyelonephritis which could be recurrent, renal stones and eventually loss of kidney tissue leading to chronic kidney disease (Type 2).[3,4] There are sometimes also skeletal cardiac and ear abnormalities associated with Type 2.[4]

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Case Report

We report this case of a 48-year-old female who presented with recurrent UTIs and pyelonephritis necessitating antibiotic therapy and several admissions. The patient also had a history of primary amenorrhea and a congenital deformity of a smaller right leg and only two toes on the right foot.

She had been seen at a private hospital, prior to presenting at the University of Benin Teaching Hospital (UBTH) where she had surgery twice, in 2002 and 2008 to remove kidney stones. She was subsequently sent to the nephrology clinic on account of elevated blood pressure and a single ectopic kidney. She was commenced on snit-hypertensives, amlodipine, lisinopryl and antibiotics guided by microscopy culture and sensitivity reports on account of the UTI.

It was also discovered during her follow up, that she had unilateral renal agenesis and the solitary right kidney was ectopically placed in the pelvic region (Figures 1a and 1b).

She had been regular on follow up clinic from 2014 to 2022 and had developed progressive proteinuria and a progressive deterioration in renal function probably as a consequence of the repeated episodes of pyelonephritis.

At her initial presentation in 2014, there was no proteinuria and electrolyte, urea and creatinine results were within normal reference values.

Despite other clinical findings, the managing

physician did not note that the repeated urinary tract infections, hypertension, unilateral ectopically placed kidney and a history of pronounced limp due to a smaller right leg with oligodactyly may have been part of a MRKH syndrome (Figure 2). She was also seen by the Urologists who wanted to put in a stent but the patient did not have the financial wherewithal to do the surgery

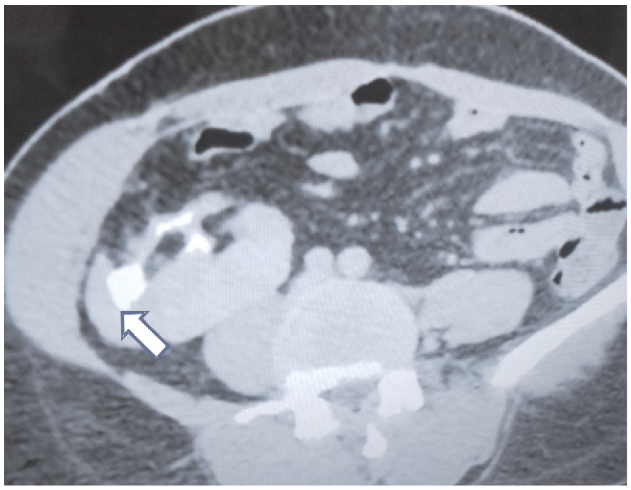

Figure 1a: Right mal-rotated ectopically placed kidney

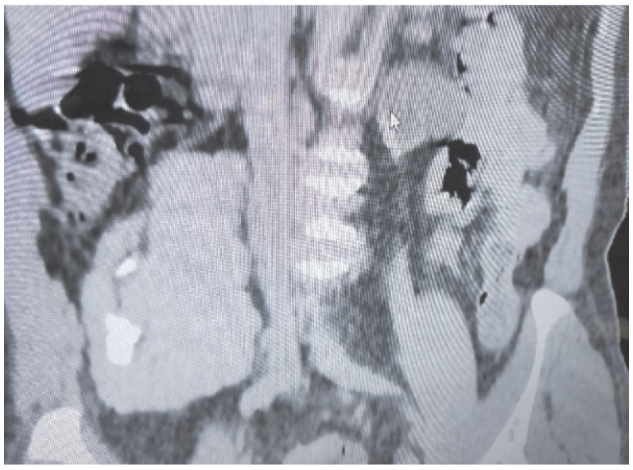

Figure 1b: Unilateral ectopic kidney

She had presented with worsening ill health, increasing weakness, vomiting and colicky abdominal pains which had become more marked in the two weeks prior to the incident presentation.

On examination at presentation, patient was markedly ill looking. There was moderate pallor, patient was in moderate respiratory distress and had bilateral peripheral pitting pedal edema. Of note was that the right leg was smaller than the left, and had oligodactyly of the right foot (patient had only 2 toes on the right leg.)

Abdominal examination showed epigastric tenderness, soft tender hepatomegaly and a central abdominal mass.

Limb examination showed a hypoplastic right leg with only 2 toes which gave her a limping gait while walking. Neurological examination, apart from the limping gait, showed that patient had asterixis. Gynecological examination done previously revealed normal secondary sexual characteristics with normal external genitalia. However, patient had a blind ending vagina.

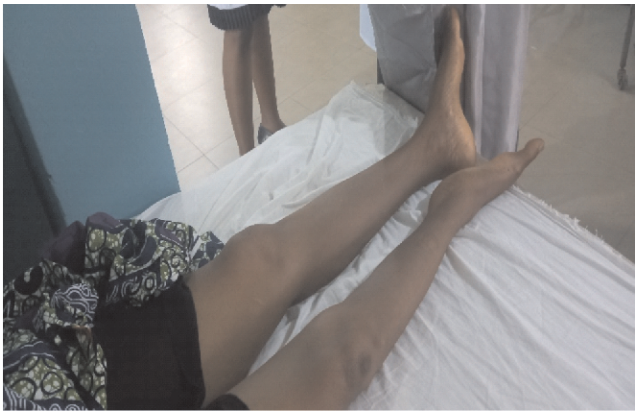

Figure 2: Hypoplastic right leg and normal left leg

Chest examinations showed tachypnea and bilateral basal crepitations. Blood pressure was elevated at 186/110 mmHg with 1st and 2nd heart sounds and a mildly enhanced aortic component of the second heart sound. There were no added heart sounds. Abdominal examination showed a soft tender abdomen 4cm below the right costal margin. Tenderness was marked in both the epigastric and suprapubic regions of the abdomen with a tender mass in the lower abdomen. Vaginal examination showed normal labia majora and minora with a shallow blind ending vaginal canal. (Figure 3).

A clinical diagnosis of chronic kidney disease with possible end stage kidney disease with uraemic gastritis to rule out UTI and pyelonephritis was made.

Patient had to be dialysed on account of the uraemic symptoms and the electrolytes urea and creatinine results which were markedly deranged.

The ultrasound results and previous CT demonstrated a unilateral malrotated ectopically placed right kidney, left renal agenesis, uterine agenesis and other features.

The reduced size of the right leg and the oligodactyly were noted.

The diagnosis of Mayer-Rokitansky-Kuster-Hauser Syndrome, Type II, with chronic kidney disease in end stage kidney disease, secondary to chronic pyelonephritis and also possible hypertensive nephrosclerosis was made.

She is currently on maintenance hemodialysis from home and is also on follow up in the Nephrology clinic and is clinically stable.

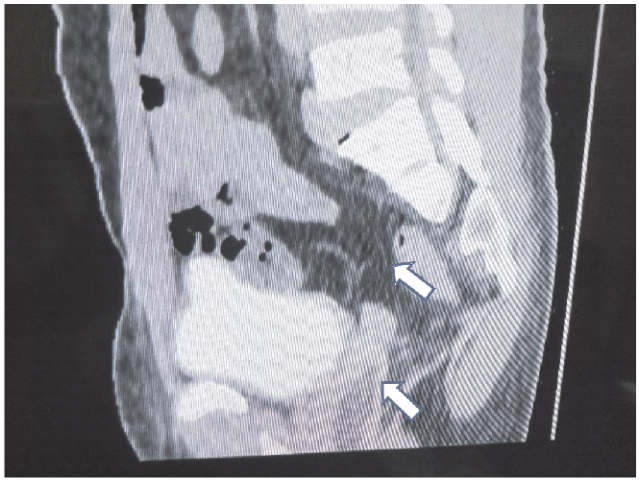

Figure 3: Blind ending vagina and uterine agenesis

She is also being referred to the gynaecology clinic to enable the gynecologists co-manage her.

Discussion

This patient had a history of recurrent UTIs that made her present to the University of Benin Teaching Hospital and was subsequently diagnosed to have Mayer-Rokitansky-Kuster-Hauser syndrome as a result of the clinical presentation and investigative findings.

Mayer-Rokitansky-Kuster-Hauser syndrome is a rare congenital disorder, said to affect about 1-4,500 to 1-5,900 live female births.[1,2] It was first reported by a Bonn anatomist and gynaecologist Mayer in 1829, and was seen over the next 130 years before it was given the name-Mayer-Rokitansky-Kuster-Hauser Syndrome, with respect to others who reported similar diagnosis of malformation.[3] It is a rare disorder characterized by the failure of the uterus and the vagina to develop properly in women who have normal ovarian function amid oftentimes normal external genitalia.[1] It is characterized by the absence of the upper two thirds of the vagina, accompanied by various types of uterine anomalies. It could be isolated or accompanied by other congenital malformations.[4]

It is subdivided mainly into type 1, which mainly presents with amenorrhea as the initial complaints and type 2 which is associated with disorders of the other organs and systems especially the kidneys and the skeletal system.[4]

Type 1 Mayer Rokitansky Kuster Hauser syndrome is characterized by a failure of the uterus and the vagina to develop properly. It could be an aplasia, where the uterus and the vagina are absent or atresia, where there may be uterus buds only with a poorly developed vagina.

The type 1 of MRKH syndrome is sometimes referred to as Mullerian aplasia. The severity the clinical presentation may vary according to the level of penetrance.

When the disorders of the type 1 described above occurs in additional physical findings, it is classified as MRKH type 2 or Mullerian duct aplasia, Renal dysplasia and cervical somite abnormalities [MURCS][5]

The affected woman with MRKH syndrome type 2 may exhibit absence of the kidney, unilateral renal agenesis, underdeveloped hypoplastic kidneys and improper positioning within the body of one or both kidneys [renal ectopia].[6] This patient had unilateral renal agenesis, and ectopic right kidney. It has also been found out that there could also be a single ectopically placed malrotated kidney.[7] Abnormalities of the extremities may include absence of a portion of 1 or more fingers or toes.

Renal abnormalities can cause growth deficiency, kidney stones and increased susceptibility to urinary tract infections and abnormal accumulation of urine within the kidney due to obstruction leading to hydronephrosis. Our index patient had all the above clinical features hydronephrosis, recurrent urinary tract infections and recurrent pyelonephritis. This tends to predispose the patient to chronic kidney disease from the chronic interstitial nephritis leading to end stage kidney disease as happened in this patient.[8,9,10] In addition, the patient had a hypoplastic right leg and foot as well as extrodactyly.[11,12,13]

Chronic pyelonephritis was most likely induced by the renal abnormality causing obstructive symptoms and stasis in the urinary tract and nephrolithiasis for which the patient had 2 previous surgeries by a urologist in a private hospital.

The frequency of renal abnormalities in MRKH is about 30 to 40%. The most frequent anomaly is the presence of a solitary kidney either located normally or in the pelvis as we had in this incident patient.[6,14] The major anatomic kidney abnormalities are a risk factor for chronic pyelonephritis induced by recurrent kidney infection. The scarring caused by chronic pyelonephritis leads to loss of renal tissue and renal function which may progress to end stage renal disease, as occurred in this incident patient.[8,13,15]

The issues raised were important because in the years of her follow up, all the symptoms and clinical features were not initially recognized as Mayer-Rokitansky-Kuster-Hauser Syndrome because the condition is rare and could end up in chronic kidney disease and end stage kidney disease.

It is important that due to the rarity of this condition, nephrologists should be alert for the existence of an association between urinary tract abnormalities, solitary kidney, skeletal anomalies and MRKH syndrome (Type II). There is need for clinicians and nephrologists to extensively assess cases such as these to possibly detect

them at an earlier stage.[13] This diagnosis was totally missed for a duration of over 7 years, the diagnosis being made for the first time at the age of 48 years. This patient would need a multi-specialist care programme involving the physicians (nephrologists and cardiologists) gynecologists, surgeons and mental health physicians to attend to her psychological needs.

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