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VAGINAL AGENESIS (MAYER-ROKITANSKY-KUSTER-HAUSER SYNDROME) PRESENTING WITH EXERTIONAL DYSPNOEA, FATIGUE AND INTERMITTENT PALPITATIONS

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ABSTRACT

Introduction: Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome represents a spectrum of Müllerian duct anomalies. MRKH type I involves isolated agenesis of the uterus and vagina, while MRKH type II involves extra-genital defects most commonly affecting the skeletal, renal, cardiovascular and auditory systems with functional ovaries along with agenesis of the uterus and vagina.

Case description: A 19-year-old female presented with exertional dyspnoea, fatigue and intermittent palpitations for the last 2 years. Clinical evaluation revealed a classic machinery murmur at the left second intercostal space, suspecting the diagnosis of patent ductus arteriosus (PDA), which was confirmed by two-dimensional echocardiography. She also presented with primary amenorrhea, for which an ultrasound of the abdomen and pelvis was done, which showed a rudimentary uterus and absence of the upper third of the vagina with normal ovaries. Hence, a diagnosis of MRKH syndrome type II was made. A cardiac computed tomography scan revealed a persistent left superior vena cava (PLSVC), which is a rare association in an MRKH patient. There was also the presence of an extrarenal pelvis seen on a kidney, ureter, bladder (KUB) X-ray.

Conclusion: This case highlights the rare association of MRKH syndrome type II with PDA, PLSVC, and the extrarenal pelvis, emphasising the need for comprehensive multisystem evaluation. These patients need effective counselling regarding sexual well-being and appropriate management of extra genital manifestations.

KEYWORDS

MRKH type II, patent ductus arteriosus, persistent left superior vena cava, Müllerian agenesis, extrarenal pelvis

LEARNING POINTS

- Mayer-Rokitansky-Küster-Hauser (MRKH) Type II is a multisystem congenital syndrome that requires comprehensive evaluation. A multidisciplinary approach is essential, involving gynaecology, cardiology, nephrology and radiology.
- Rare cardiovascular anomalies can coexist with MRKH type II, like patent ductus arteriosus and persistent left superior vena cava (PLSVC). PLSVC may be asymptomatic in a patient, but it is important to know because it can make certain procedures more challenging, like pacemaker insertion, intra-cardiac device implantation, left-sided central line placement and cardiac surgeries. A card should be given to these patients in case they need such procedures.

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- An extra-renal pelvis is a benign congenital condition which does not need treatment. Generally, it is asymptomatic, but sometimes it increases the risk of urinary tract infection (UTI) due to slowed urine flow, so such patients should be monitored for UTI and have renal function tests regularly.

INTRODUCTION

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a congenital anomaly in which there is aplasia or hypoplasia of the uterus and upper two-thirds of the vaginal canal in a female who is karyotypically 46, XX[1]. MRKH syndrome is divided into two types. Type 1 is an isolated MRKH syndrome in which only the development of the uterus and upper 2/3rd of the vagina is arrested due to a defect in the Müllerian duct, whereas in type 2 MRKH syndrome, along with these uterovaginal defects, there is renal dysplasia, cervicothoracic somite anomalies, hearing defects and, rarely, cardiac defects[1].

This complex developmental disorder occurs in approximately 1 in 4,500 to 5,000 female live births[2]. Urological alterations associated with MRKH type 2 are unilateral renal agenesis, horseshoe kidney, or renal ectopia and occur in 40% to 60% of affected patients[3]. Extrarenal pelvis is a benign structural anatomical variation that is less commonly associated with this syndrome but predisposes patients to urinary stasis and recurrent urinary tract infections (UTIs)[3]. Congenital cardiovascular anomalies are reported in less than 4% of MRKH cases, and it is generally confined to defects like atrial septal defects, tetralogy of Fallot, or valvular stenosis[1]. Patent ductus arteriosus (PDA) combined with a persistent left superior vena cava (PLSVC) existing together is a very unusual finding within the clinical spectrum of MRKH type 2[4].

CASE DESCRIPTION

A 19-year-old female patient born out of a non-consanguineous marriage presented to the medicine outpatient department with chief complaints of exertional dyspnoea, fatigue and intermittent palpitations on and off for the last 2 years. She also complained of burning micturition for the past 2 weeks and was admitted to the medicine ward. She reported that the dyspnoea was slightly exacerbated by climbing three flights of stairs but denied any syncope, orthopnoea, paroxysmal nocturnal dyspnoea or dyspnoea at rest, so her cardiovascular symptoms were categorised as New York Heart Association class I. The palpitations were intermittent and used to get relieved on their own. It was significant that the patient had reported that she had not attained menarche to date, that is, primary amenorrhea. Her family and past medical histories were unremarkable, with no known exposure to teratogens during her mother's pregnancy or any maternal illness.

On admission, her pulse was 110 bpm, blood pressure was 110/80 mmHg, oxygen saturation was 98% on room air, and respiratory rate was 14 breaths/minute. Cyanosis, clubbing and oedema were absent. The head-to-toe examination revealed normal female external genitalia (Tanner stage 5) with appropriately developed secondary sexual characteristics. On cardiovascular system examination, a grade III/IV continuous "machinery" murmur was heard that was loudest at the left second intercostal space. Mild suprapubic tenderness was noted on per abdomen examination. She did not have a short neck or low posterior hairline, and no skeletal deformities were noted. Her ear, nose, and throat examination were normal.

Her routine lab investigations, including complete blood count, renal function test, liver function test, and serum electrolytes, were normal except for mild leucocytosis

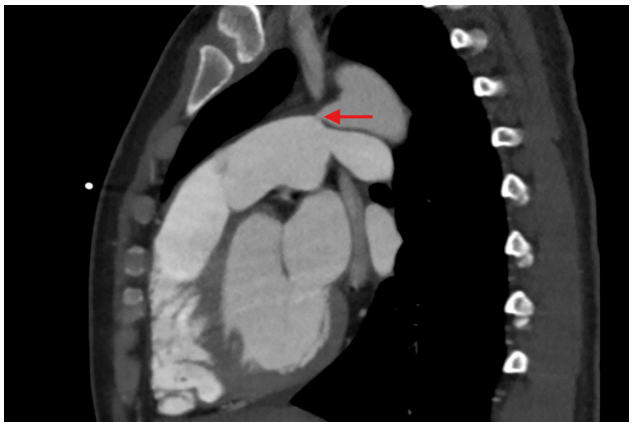


Figure 1. *Cardiac computed tomography scan, sagittal section showing the 11 mm patent ductus arteriosus (arrow).*



Figure 2. *Cardiac computed tomography scan, coronal section showing persistent left superior vena cava (arrow).*

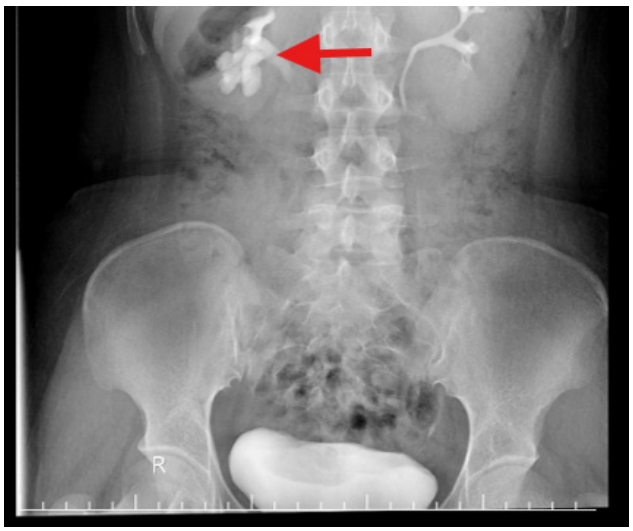


Figure 3. Kidney, ureter, bladder X-ray showing an extra-renal pelvis on the right side (arrow).

(12000/µl). The urine routine showed abundant pus cells, bacilli 2+, and the urine culture identified *Enterococcus faecalis* colonies (>10^5 CFU/ml), confirming a UTI. The electrocardiogram showed sinus rhythm, left ventricular hypertrophy with non-specific ST and T wave changes noted in leads I, aVL, V1-V3. Cardiac enzymes (creatine kinase-MB and troponin I) were sent, which were within normal limits. A chest X-ray, posterior-anterior, view showed normal cardio-thoracic ratio with right pulmonary artery dilatation. Due to the clinical suspicion of PDA, the patient underwent two-dimensional echocardiography (2D Echo), which revealed a large, 11 mm PDA (Krichenko Type A) with a high-velocity left-to-right shunt >2:1, systolic gradient/diastolic gradient- 93/55 mmHg and mild pulmonary hypertension. A cardiac computed tomography scan was performed, which confirmed the 11mm PDA *(Fig. 1)* and also revealed PLSVC *(Fig. 2)*. Her KUB X-ray *(Fig. 3)* and ultrasound (US) of the abdomen and pelvis showed a right extrarenal pelvis with associated mild hydronephrosis. It also revealed a rudimentary uterus and absence of the upper third of the vagina with well-developed normal ovaries *(Fig. 4)*. Her hormone profile-follicle- stimulating hormone-7.03 mIU/Ml, luteinizing hormone-8.01 mIU/Ml and estrogen- 27 pg/ml were all within normal limits. Our limitation was that we could not do genetic testing.

She was started on a course of injectable ampicillin 1 g administered intravenously three times a day for 14 days for UTI management. A repeat urine culture was done after treatment, which reported no growth. The patient was started on enalapril (2.5 mg) and furosemide (20 mg) once a day to manage volume overload and reduce the risk of heart failure progression. Following stabilisation, an elective PDA device closure was performed by the cardiology team using a 12 × 14 mm HeartR™ PDA Occluder (LifeTech Scientific, Shenzhen, China). The patient was referred to the gynaecologist because of MRKH syndrome. She was advised about vaginal dilatation, vaginoplasty or uterus transplant.

Post PDA closure a chest X-ray, posterior-anterior view, showed the PDA device in situ, and 2D- Echo revealed no residual shunt across the PDA device *(Fig. 5)*. The patient was counselled regarding the need for infective endocarditis prophylaxis if she undergoes any dental procedures in the next 6 months and to follow up with the cardiologist regularly.

DISCUSSION

Our patient was initially diagnosed as having PDA clinically as well as on 2D Echo. Since she also had primary amenorrhea, she was investigated further. US showed a rudimentary uterus and absence of the upper third of the vagina with the presence of bilateral, normal, ovaries; hence she was diagnosed with MRKH syndrome type 2. She also had PLSVC and a right-sided extrarenal pelvis.

In type 1 MRKH syndrome, there is a developmental failure during the 4th-5th week of embryogenesis, particularly

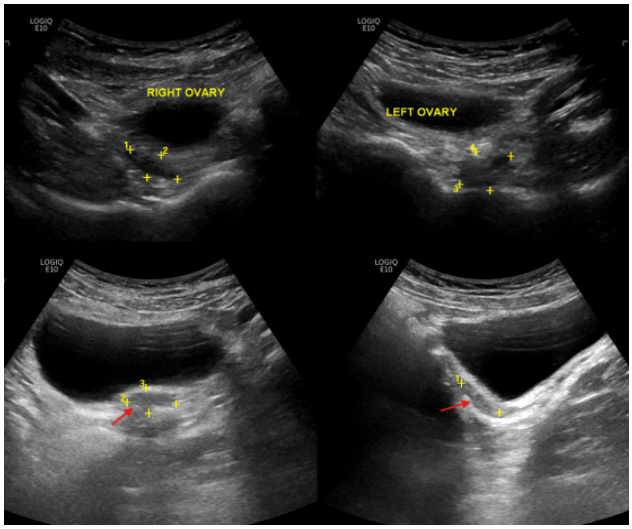


Figure 4. Pelvic ultrasound showing radiological evidence of Müllerian agenesis, rudimentary uterus, and absent vagina with normal ovaries.

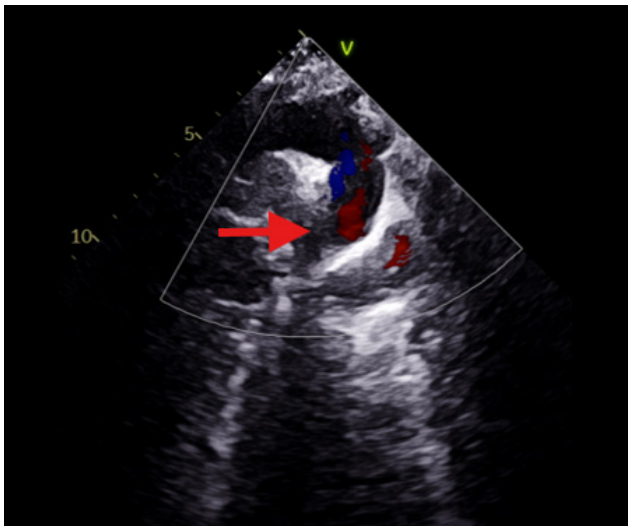


Figure 5. Two-dimensional echocardiography showing patent ductus arteriosus closure device in-situ (arrow) and no residual shunt across the device.

affecting the intermediate mesoderm that specifically gives rise to the Müllerian ducts. The failure of these Müllerian ducts to grow results in the absence of a uterus and vagina in these females. In type 2 MRKH syndrome, along with the absence of a uterus and vagina, there are gross renal deformities. This is because the Wolffian duct guides the growth of the Müllerian duct. When the entire mesoderm is affected, it results in impaired kidney development and Müllerian development.

Also, since the lateral plate mesoderm is affected, it results in coexisting cardiovascular shunts and anomalies. PLSVC is seen in approximately 0.61% of the general population[5]. In MRKH syndrome, to the best of our knowledge only one case of PLSVC has been reported, which reflects the rare presentation of this congenital anomaly in such patients[4]. The ductus arteriosus is a small vessel connecting the pulmonary artery with the aorta and transports oxygenated blood from the right side of the heart to the aorta. Its persistence even after birth is known as PDA. Over the years, if a PDA does not close it results in the development of a right-to-left shunt and irreversible pulmonary artery hypertension, which can be life-threatening[6]. PDA closure was done on time in our patient before the development of irreversible pulmonary hypertension.

The presence of primary amenorrhea in a young female account for various differential diagnoses such as complete androgen insensitivity syndrome (CAIS), Imperforate hymen/tranverse vaginal septum. CAIS patients are phenotypically females but are karyotypically 44, XY. The presence of testes in such patients results in the production of high levels of testosterone, which differentiates this condition from MRKH syndrome, in which there are functional ovaries and low levels of testosterone[7]. Patients with imperforate hymen or transverse vaginal septum have normal development of uterus and ovaries[8].

Counselling for sexual well-being is of paramount importance in these patients. They can be informed about surrogacy as an option for motherhood[9]. This case has added to the existing knowledge of MRKH type 2.

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