Mayer-Rokitansky-Küster-Hauser syndrome associated with a urogenital sinus anomaly in a 4-year-old: report of a case

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Abstract

The Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a rare entity characterized by congenital aplasia of the vagina and uterus in the presence of normal ovarian function, in conjunction with a 46 XX karyotype. This condition is mostly signalled by primary amenorrhoea around the season of puberty. We report on its diagnosis in a 4-year-old child presenting with urinary incontinence. Also of interest in our index patient is the presence of a pure urogenital sinus anomaly. This extremely rare association has not been previously diagnosed in childhood.

Introduction

The Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is defined by congenital aplasia of the uterus and upper part (2/3) of the vagina and is associated with normal secondary sexual characteristics along with a 46 XX karyotype.[1,2] This was first described by Mayer (1829), then Rokitansky (1838) as an abnormality of the development of the uterine ducts which resulted in agenesis of the uterus and vagina.[3,4] The urological associations were recognized by Küster (1910) and later, Hauser (1961) distinguished MRKH syndrome from testicular feminization.[3]

The MRKH syndrome is also known as Mullerian Aplasia (MA) or Genital Renal Ear Syndrome (GRES) with a reported incidence of approximately 1 in 4000 - 5000 female births.[1,2,4]

Despite the duration of its evolution spanning almost two centuries, the exact aetiology of the MRKH syndrome stills remains unclear.[1,5]

Here, we describe the MRKH syndrome along with a rare association being diagnosed in a child.

Case report

A 4-year-old female presented with a history of total urinary incontinence. This symptom had been present since birth and was not associated with burning on micturition or

haematuria. There was an absence of any daytime variation. There was no history of any previous urogenital trauma or surgery.

On general paediatric history (including family history) no abnormalities were detected.

On examination of the back, a presacral dimple was noted. The rest of the neurological examination was normal.

On inspection of the external genitalia, a normal clitoris and labia was observed. The urethral meatus was difficult to visualize.

Abdominal ultrasound study revealed a large (9.3 x 4.2 cm) left kidney. The right kidney was not demonstrated.

Figure 1.

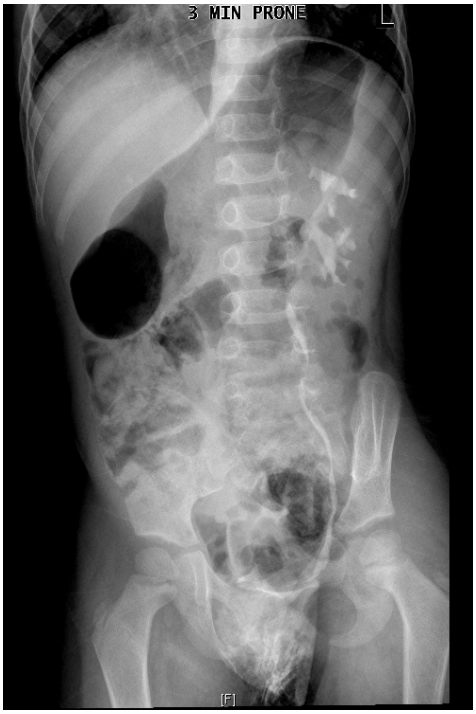

Figure 1. The IVP study demonstrating a solitary left kidney and contrast leakage confirming the communication between the bladder and vagina. Spina bifida occulta can also be observed.

The left kidney handled and excreted contrast well during the intravenous pyelogram (IVP) study (figure 1). The right renal system was not visualized. Spina bifida occulta was noted. The IVP also revealed a normal bladder positioning with a communication between the bladder and vagina. During the study, contrast was seen draining through the vagina.

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Figure 2.

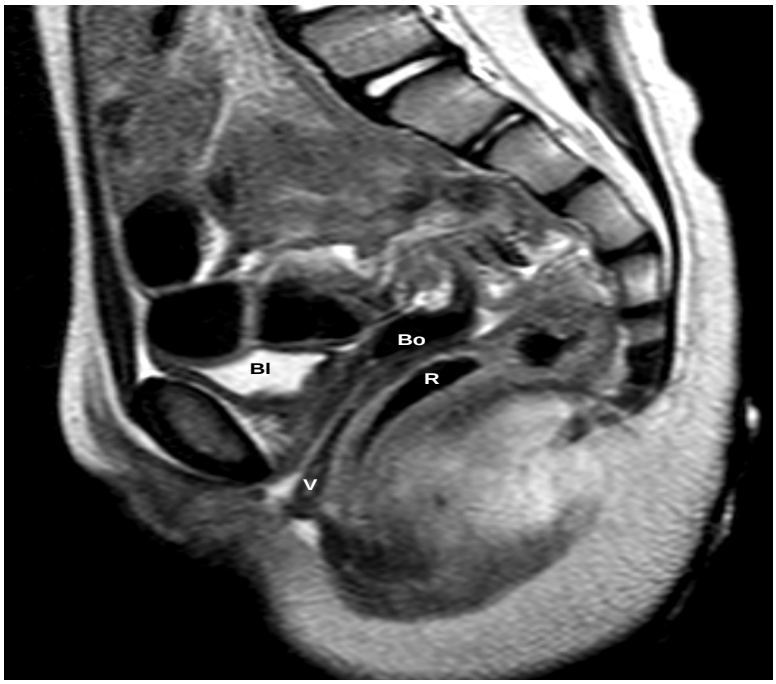

Figure 2. MRI pelvis (T2-weighted) mid-sagittal view illustrating the presence of bowel loops filling the anatomical location of the absent “uterus”. (Bo: bowel, Bl: bladder, V: vagina, R: rectum)

Magnetic resonance imaging (MRI) of the abdomen and pelvis (figure 2) illustrated both uterine and upper vaginal aplasia. The common channel (figure 3) arising from the confluence of the distal bladder and vagina was also visualised.

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Figure 3.

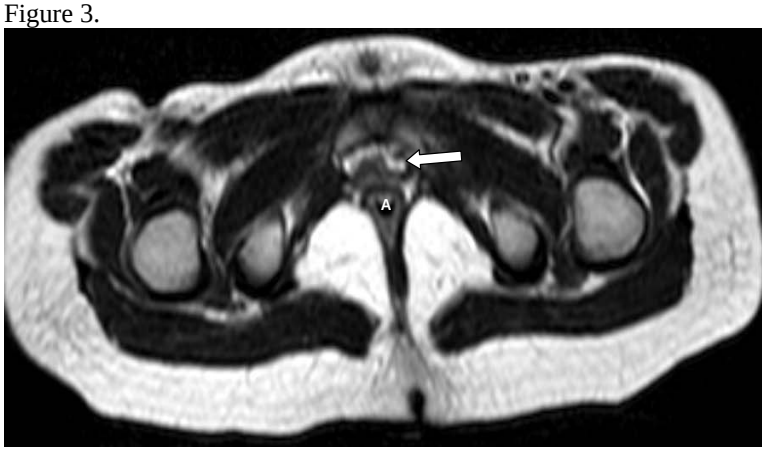

Figure 3. MRI pelvis (T2-weighted), axial view of the common channel (arrow), seen anterior to the anal canal, produced as a result of the urogenital sinus anomaly.
(A: anal canal)

Karyotyping was consistent with a 46 XX pattern. Urea and electrolyte levels were normal. The hormonal profile (including; follicle stimulating hormone, luteinizing hormone, oestradiol and progesterone) values were well within the normal range for age.

At the examination under anaesthesia (EUA) and genitogram, the clitoris was normal in size and location. The hymen was not present and a shallow vaginal recess with a depth of 0.5 cm was noted. The urethral meatus was absent. Due to the absence of the anterior vaginal wall and the posterior urethra, a common channel between the bladder and vagina was observed. Cystoscopy revealed a solitary left ureteral orifice. The anus was well located and patent.

In view of the uterine and vaginal aplasia, hormone profile findings, karyotype results and right renal agenesis, a diagnosis of type II MRKH syndrome with an associated urogenital sinus anomaly was established.

After extensive counselling, the family opted for a urinary diversion via a vesicostomy at this stage, with urethral and vaginal reconstruction at a later setting. Follow-up psychological review is also to be scheduled at regular intervals.

Discussion

A diagnosis of MRKH syndrome made at an early (pre-pubertal) age is an extremely rare entity.[6] This association with a urogenital sinus anomaly is also an extremely rare occurrence and was first described in 2007, in a 24-year-old who presented with primary infertility.[4] Here we narrate the first reported case (to the best of our knowledge) of this association being diagnosed in childhood.

The most common presenting symptom in MRKH syndrome is primary amenorrhoea.[3,4] Patients may also present with cyclical abdominal pain, primary infertility and dyspareunia.[1,3,4] In view of the above, the mean age for diagnosis is estimated at 17 years.[3,6] The external genitalia in the MRKH syndrome is usually normal since the development of the lower third has a different embryological origin to that of the upper segment (2/3) of the vagina.[5]

MRKH syndrome is divided into two distinct types:

- type I, also called typical or Rokitansky sequence
- type II, also called atypical or MURCS (Mullerian duct aplasia, Renal dysplasia and Cervical Somite abnormalities) association.[1,2,5]

The type II MRKH syndrome tends to occur more frequently than the type I and has been shown to have associations with cardiac defects, conductive hearing loss and skeletal abnormalities.[1,5] Upper tract anomalies including renal agenesis, pelvic kidney, renal hypoplasia and horseshoe kidney have been reported to occur in up to 60% of patients with type II MRKH syndrome.[1-5]

Among the differential diagnosis to consider is that of Winter syndrome which may also consist of renal agenesis and vaginal developmental abnormalities.[5] The vaginal anomaly in Winter syndrome is that of distal third atresia, while the MRKH syndrome affects the proximal segment, resulting in vaginal aplasia.[5]

Diagnostic imaging modalities in MRKH syndrome include transabdominal ultrasonography and MRI.[1,4] Laparoscopy can also be used, however MRI is more precise and less invasive.[4]

A holistic approach to the management of MRKH syndrome should address the surgical, psychological, reproductive, social and sexual issues.[1,3,4] The management options for infertility include adoption and in vitro fertilization with a surrogate pregnancy.[3,5]

The neovagina can be created via a surgical and nonsurgical approach.[1] The nonsurgical approach is based on gradual dilatation and is only applied if the vaginal recess is between 2-4 cm deep.[1,3] The surgical options include a sigmoidal colpoplasty, Abbe-McIndoe and Vecchietti operations.[1,5]

Early diagnosis allows for better psychological preparation prior to the dawn of puberty. Management options for vaginal reconstruction can also be discussed and contemplated well before the commencement of sexual intercourse.

Since MRKH syndrome is known to have a common association with upper urinary tract anomalies and can be associated with a urogenital sinus anomaly, its diagnosis and comprehensive management should be familiar to the consulting urologist.

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