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CASE REPORT

A Case of Persistent Urogenital Sinus

Pitfalls and challenges in diagnosis

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تقرير عن حالة مصابة بالجيب البولي المستمر

المزالق والتحديات في التشخيص

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ABSTRACT: Persistent urogenital sinus (PUGS) is a rare anomaly whereby the urinary and genital tracts fail to separate during embryonic development. We report a three-year-old female child who was referred to the Sabah Women & Children Hospital, Sabah, Malaysia, in 2016 with a pelvic mass. She had been born prematurely at 36 gestational weeks via spontaneous vaginal delivery in 2013 and initially misdiagnosed with neurogenic bladder dysfunction. The external genitalia appeared normal and an initial sonogram and repeat micturating cystourethrograms did not indicate any urogenital anomalies. She therefore underwent clean intermittent catheterisation. Three years later, the diagnosis was corrected following the investigation of a persistent cystic mass posterior to the bladder. At this time, a clinical examination of the perineum showed a single opening into the *introitus*. Magnetic resonance imaging of the pelvis revealed gross hydrocolpos and a genitogram confirmed a diagnosis of PUGS, for which the patient underwent surgical separation of the urinary and genital tracts.

Keywords: Urogenital Abnormalities; Congenital Defects; Clean Intermittent Catheterization; Hydrocolpos; Cystography; Case Report; Malaysia.

الملخص: الجيب البولي المستمر هو شذوذ نادر حيث تفشل المسالك البولية والتناسلية للفصل خلال التطور الجنيني. هذا تقرير حالة عن طفلة تبلغ من العمر ثلاث سنوات تم إحالتها إلى جامعة المستشفى كيبانجسان ماليزيا، كوالالمبور، ماليزيا، في عام 2016 لوجود ورم في منطقة الحوض. كانت الطفلة قد ولدت قبل الأوان في 36 أسابيع الحمل عن طريق الولادة المهبلية العفوية في عام 2013 وتم تشخيصها خطأ في البداية غلى أنها حالة خلل وظيفي عصبي في المثانة. بالكشف الظاهري وجدت الأعضاء التناسلية الخارجية طبيعية، ولم يظهر السونار أو اعادة الفحص بالأشعه علي المثانة، ومجرى البول أثناء التبول وجود أي شذوذ بولي أو تناسلي. لذلك تم علاجها بعمل قسطرة متقطعة نظيفة للبول. بعد ثلاث سنوات، تم تصحيح التشخيص أثناء فحص الطفلة لوجود ورم كيسي مستمر في منطقة الحوض خلف المثانة البولية. في هذا الوقت، أظهر الفحص السريري للعجان وجود فتحة واحدة في مقدمة. كشف التصوير بالرنين المغناطيسي للحوض عن وجود تجمع مائي (موه) في المهبل وأظهرت الأشعة على الأعضاء التناسبية وجود الجيب البولي المستمر، وخضعت الطفلة لعملية فصل جراحي للمسالك البولية والتناسلية.

الكلمات المفتاحية: تشوهات الجهاز البولي التناسلي؛ العيوب الخلقية؛ قسطرة نظيفة متقطعة؛ موه المهبل؛ تصوير المثانة؛ تقرير حالة؛ ماليزيا.

A persistent urogenital sinus (PUGS) occurs as a result of the arrested migration of the Müllerian ducts from the Müller tubercle to the vestibule.[1] The incidence of this rare congenital anomaly is estimated to be six in every 100,000 female births.[2] While PUGS can present as an isolated anomaly, it has also been associated with other diseases, including congenital adrenal hyperplasia or McKusick-Kaufman syndrome.[3–5] The presence of PUGS along with digit anomalies, *retinitis pigmentosa*, obesity, learning disabilities and male hypogenitalism are phenotypical characteristics of Bardet-Biedl syndrome.[6] In addition, PUGS may also be seen in hand-foot-genital syndrome, in which affected males have hypospadias and females have both Müllerian and limb abnormalities.[7]

Case Report

A three-year-old female child was referred to the paediatric surgical clinic of the Sabah Women & Children Hospital, Sabah, Malaysia, in 2016. She had been born prematurely at 36 gestational weeks via spontaneous vaginal delivery in 2013. At 35 gestational weeks, an antenatal ultrasound had shown a distended bladder with a thickened wall. At birth, the neonate had a distended abdomen and appeared tachypnoeic with intermittent grunting sounds. Consequently, she underwent bladder catheterisation during which 100 mL of urine was drained, which resulted in an improvement of her symptoms. However, she subsequently developed urinary retention with incomplete voiding after the catheter was removed. Initial blood

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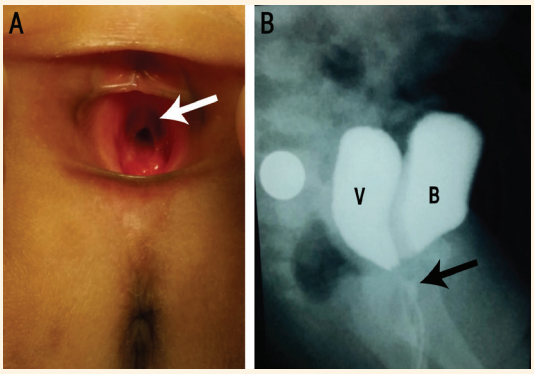

Figure 1: A: Clinical photograph of the perineal region of a three-year-old female showing a single opening at the *introitus* (arrow). **B:** Genitogram of a three-year-old female showing a common channel (arrow) bifurcating anteriorly into the urinary bladder and posteriorly into the vagina.
V = vagina; B = bladder.

investigations showed renal impairment with urea levels of 6.3 mmol/L and creatinine levels of 161 mmol/L. She was diagnosed with neurogenic bladder dysfunction and underwent clean intermittent catheterisation (CIC) every six hours, with approximately 30–40 mL of residual urine being drained each time.

Seven days after birth, an ultrasound of the kidneys, ureter and bladder showed bilateral mild hydronephrosis with no evidence of hydroureter. The bladder appeared normal. At three weeks of age, a micturating cystourethrogram (MCUG) did not reveal any vesicoureteral reflux and the contours of the bladder seemed normal. At two months, a technetium-99m mercaptoacetyltriglycine scan indicated a differential function of 46% and 54% for the left and right kidneys, respectively, with no evidence of a urinary outflow obstruction. At three months, a magnetic resonance imaging (MRI) scan of the spine appeared normal. The patient attended regular follow-up appointments at the paediatric outpatient clinic due

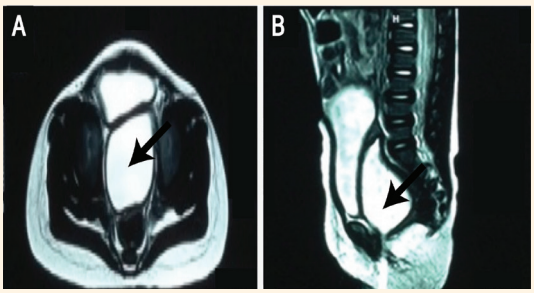

Figure 2: T2-weighted magnetic resonance imaging scans of the pelvis of a three-year-old female in the (**A**) axial and (**B**) sagittal views showing a large well-defined thin-walled cystic lesion (arrow) posterior to the urinary bladder, suggestive of hydrocolpos.

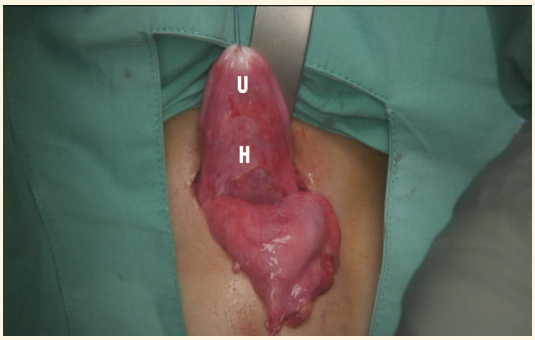

Figure 3: Intraoperative photograph of the perineal region of a three-year-old female undergoing corrective surgery for a persistent urogenital sinus showing hydrocolpos and a normal uterus.
U = uterus; H = hydrocolpos.

to her presumed neurogenic bladder dysfunction. Her mother continued to ensure that CIC was performed every six hours. The patient appeared to be well and thriving during this period without any history of urinary tract infections. However, serial ultrasounds of the kidneys, ureters and bladder at seven, 19 and 31 months of age indicated a persistent cystic structure posterior to the bladder.

In 2016, the patient was referred to the paediatric surgical clinic at the age of three years for an investigation of the persistent cystic mass. At this time, a clinical examination of the perineum revealed a single opening into the *introitus*, with a normal anus [Figure 1A]. A cystic mass was felt anteriorly upon rectal examination. She was also found to have bilateral upper and lower limb postaxial polydactyly. A repeat MCUG did not show any abnormalities; however, a genitogram revealed a PUGS with a common channel measuring 2 cm in length [Figure 1B]. An MRI of the pelvis showed a large cystic mass posterior to the bladder suggestive of gross hydrocolpos [Figure 2]. Based on the clinical and radiological findings of a PUGS in combination with the bilateral limb postaxial polydactyly, the child was diagnosed with McKusick-Kauffman syndrome.

The decision was subsequently made to surgically separate the child's urinary and genital tracts. Intraoperatively, the vagina appeared grossly distended, although the uterus was normal [Figure 3]. The vagina was surgically detached from the urogenital sinus and a tube vaginoplasty was performed, created from an inverted U-flap taken from the posterior vaginal wall. The common channel was preserved as the urethra. Postoperatively, the patient recovered well. At the time of writing, she demonstrated normal bladder emptying without requiring any further CIC.

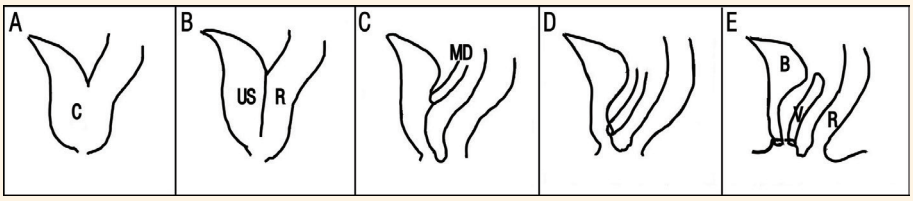

Figure 4: Diagram showing the normal embryological development of the female genitourinary system. **A:** At four gestational weeks, the urogenital sinus and rectum enter a common cavity known as the *cloaca*. **B:** At six gestational weeks, the urorectal septum grows in a caudal direction, separating the *cloaca* into a ventral urogenital sinus and a dorsal rectum. **C:** At eight gestational weeks, a pair of Müllerian ducts develop lateral to the mesonephric ducts, crossing ventromedially to fuse in the midline and join the urogenital sinus to form an elevation known as the Müllerian tubercle. **D:** Between the eighth and twelfth gestational weeks, the Müllerian ducts migrate caudally from the Müllerian tubercle to the vestibule. **E:** From the twelfth gestational weeks onwards, the development of the genitourinary system is complete.

C = cloaca; US = urogenital sinus; R = rectum; MD = Müllerian ducts; B = bladder; V = vagina.

Discussion

During embryonic development, the urogenital sinus forms as a common channel into which both the urinary and genital tracts enter before subsequently being separated later on by the migration of the Müllerian ducts [Figure 4]. The arrested migration of the Müllerian ducts at an early stage of development results in a long urogenital sinus with a short vagina and a high urethral opening, while arrested migration at a late stage results in a short urogenital sinus with a vaginal vestibule of almost normal length and a low urethral opening.[1] Consequently, PUGS can be classified into high and low anomalies according to the length of the common channel (>3 cm or <3 cm, respectively).[8] In general, short urogenital sinuses are more common, as observed in the current case.[8] Furthermore, PUGS patients can be divided into those with confluence above or below the neck of the bladder.[9] An additional classification can be made based on the distance of the vaginal confluence from the bladder neck and perineal *meatus*; this is often crucial for both clinical management and surgical reconstructive purposes.[10]

A diagnosis of PUGS is challenging due to the complexity and rarity of this condition. As demonstrated by the present case, the presence of a urogenital tract anomaly can be easily overlooked by unsuspecting practitioners when inspecting essentially normal-looking external genitalia with typical labial folds and a normally positioned anus. However, a high index of suspicion for this anomaly is necessary, especially among newborns with grossly distended abdomens and lower urinary tract symptoms for whom an antenatal ultrasound reveals a pelvic cystic mass.[1] Among infants with PUGS, the presence of such a mass occurs due to the storage of urine in the vagina (i.e. urocolpos) when the common channel is partially blocked, causing the urine to flow up into the vaginal *lumen*. The pathophysiology of this phenomenon can be explained by the location of the vaginal confluence above the bladder sphincter, the formation of a cusp in the sinus by the posterior lip of the hymen and the contraction of the *puborectalis* muscle sling.[1] As observed in the current patient, massive urocolpos may be mistaken for a distended bladder in certain cases. Hence, a thorough clinical examination of the perineum is crucial as the finding of a single opening in the *introitus* is pathognomonic of a PUGS diagnosis.

Ultrasonography is the diagnostic modality of choice in the detection of a PUGS because of the ubiquitous and minimally invasive nature of this imaging technique. The presence of a cystic lesion with internal *debris* posterior to the fetal bladder—which can be detected by sonogram as early as the second trimester of pregnancy—is suggestive of a PUGS.[11] A postnatal ultrasound with similar findings may also help to confirm the diagnosis. In the present case, a postnatal ultrasound when the patient was seven days old did not show any abnormalities; however, this might be due to the fact that the urocolpos had been drained as a result of CIC having been performed prior to the ultrasound. Structural anomalies of the ureter, bladder and urethra may also be revealed using a MCUG, which is a dynamic fluoroscopic method.[12] During the micturating phase, a lower urinary tract abnormality can be demonstrated after the removal of the catheter if the contrast medium flows into the urethra.

However, performing an MCUG on a child can be challenging. Younger patients may need to be coaxed into following instructions and bladder catheterisation can be difficult among patients with abnormal urinary tracts.[13] Among female patients with PUGS, there is also a risk of accidentally catheterising the vagina instead of the bladder. Furthermore, the child may refuse to void their bladder once the catheter has been removed.[13] The outcome of the procedure is therefore

dependent on the experience of the operator. In the current case, both MCUGs failed to demonstrate a PUGS; this was most likely because the urocolpos failed to drain spontaneously after the catheter was removed.

In certain cases, a distended vagina may be mistaken for the bladder on a MCUG, since the two organs are difficult to distinguish. In such instances, an ascending genitogram may constitute a better imaging modality since it can clearly delineate the length of the common channel and the level of vaginal confluence.[14] However, it is important to place the tip of the catheter just above the perineal opening as there is a risk of either the bladder or vagina alone being opacified if the catheter penetrates too deeply. In addition, an ascending genitogram may help the surgeon to choose a suitable approach and method of surgical repair.[14] Besides conventional imaging methods, the use of MRI has gained popularity in the paediatric field over the last decade due to advances in resolution, reduced motion artefacts, volumetric imaging and imaging time.[15] An MRI scan of the pelvis can effectively depict the relationship between urogenital structures, the pelvic muscle complex and any associated spinal or renal abnormalities.[16] Additionally, a combination of multidetector computed tomography, MRI and fluoroscopy of the urogenital tract can help to clearly illustrate the anatomy of associated structures in a three-dimensional reconstruction.[17–19] However, although the advantages of this technique are undeniable, the risks of increased radiation exposure and prolonged general anaesthesia should not be overlooked. Moreover, a cystovaginoscopy assessment prior to surgery is still needed to definitely delineate the anatomy of the genitourinary tract.[20,21]

As a result of various conditions such as PUGS, the vagina can distend and reach a potentially massive size, causing gross abdominal distension and respiratory distress if it splints the diaphragm.[22] In such cases, management consists of drainage and decompression by catheterising the common channel or, if catheterisation fails, performing an ultrasound-guided percutaneous tube vaginostomy, an open vaginostomy/vesicostomy or a ureterostomy/nephrostomy.[9,21,23,24] In the event of a rupture, a laparotomy, peritoneal drainage and open vaginostomy are temporary solutions prior to definitive surgical correction. For example, urocolpos drainage may prevent urinary tract obstructions, infections, pyocolpos or, in severe cases, rupture and peritonitis.[23,24] Fortunately, CIC was effective in draining the hydrocolpos for the current patient, thus avoiding other more invasive procedures such as a vaginostomy or vesicostomy which can cause scarring and adhesions, potentially complicating subsequent surgeries.[20]

Conclusion

A PUGS is a very rare developmental anomaly which can be easily misdiagnosed as neurogenic bladder dysfunction or overlooked entirely, especially if the external genitalia appear normal. As such, a thorough examination of the perineal orifices of a newborn is necessary to rule out complex urogenital abnormalities, particularly among those who present with a pelvic mass and urinary symptoms. False-negative ultrasound and MCUG findings may lead to a delay in diagnosis; however, genitograms and MRI modalities are useful to clearly visualise the anatomy of the genitourinary tract.

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