ORIGINAL RESEARCH PAPER

Radio-Diagnosis

KEY WORDS: Case Report, Cloacal Malformations, Hydrosalpinx, Pelvic Inflammatory Disease, Persistent Urogenital Sinus.

PERSISTENT UROGENITAL SINUS – A RARE CASE REPORT

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ABSTRACT

Introduction: Urogenital disorders are one of the most common congenital anomalies presenting in infants and children. Persistent urogenital sinus is a condition where, there is failure of separation between urinary and genital system. This condition is associated with many other congenital anomalies and syndromes like spinal defects, ano-rectal and uterine malformations. There are multiple presentations known for this condition, depending on the age of presentation. **Case History:** We report a case of a 25 year old woman referred to our hospital with pelvic inflammatory disease. A clinical examination of the perineum revealed a single Genito-urinary meatus between the labia. The Patient had associated skin disease all over the body which was proved histopathologically as kindlers syndrome (epidermolysis bullosa) with associated oesophageal webs. There are no other associated renal, bladder, rectal and anal anomalies. Radiological evaluation through Micturating cystourethrogram (MCUG), Genitogram, Ultrasound, pelvic CT and MRI revealed low confluence persistent urogenital sinus which was confirmed through cystoscopy. There is a rare presentation of persistent urogenital sinus in a young female of reproductive age. **Conclusion:** This case emphasizes the need for radiologists, gynaecologists, urologists and other physicians to be aware of congenital urogenital anomalies when a patient presents with abnormal external genito-urinary openings, primary amenorrhea, pelvic cystic mass and ambiguous genitalia.

INTRODUCTION

Persistent urogenital sinus (PUGS) is a rare clinical entity characterised by the failure of urinary and genital tracts to separate during embryonic development. The current case presented to us with complaints suggestive of pelvic inflammatory disease with bilateral hydrosalpinx. Most possible theories for developing PID in a case of PUGS include retrograde passage of urine into the vaginal canal and uterus. The formation of urinomas, hydrometro-colpos and pelvic cysts are other frequent complications of the persistent urogenital sinus.

We report rare case of persistent urogenital sinus in an adult to conclude that proper radiological evaluation of PUGS and pelvic anatomy is needed, as it plays a crucial part in surgical planning. The imaging modalities which include ultrasonography, VCUG, MRI, could be misinterpreted if not correctly performed.

Case History

A 20-year-old woman presented with chief complaints of pain in lower abdomen since one-month duration, and passing of menstrual blood in urine while micturating.

Lower abdominal pain was dull-aching, intermittent, non-radiating and was subsiding on medication. Pain was not associated with menstrual cycles. Patient's bowel and bladder habits were regular. Patient is unmarried and attained menarche at the age of 15 yrs. Menstrual history recorded regular cycles with normal flow and duration (5/28days).

Patient's past history revealed diagnostic esophagoscopy and two surgeries for esophageal webs performed in 2017 and 2019. Patient has no siblings and there is no family history of urological/genital abnormalities (figure 1).

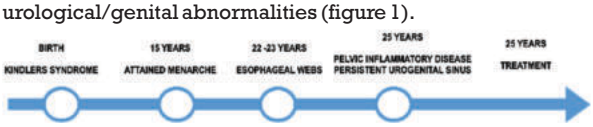

Figure 1 shows timeline of events in the patient's history

Clinical Examination

On local examination of the perineum, pubic hair is sparse and labia majora and minora are underdeveloped. There is a single orifice in the midline (which is presumed as the external urethral meatus) with absence of vaginal orifice (figure2).

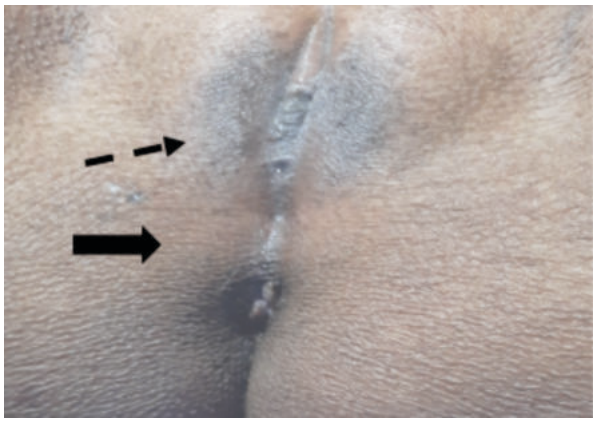

Figure 2: Image of perineum, showing underdeveloped labia majora, minora, single urogenital orifice (dotted arrow) and an anal opening (block arrow).

Anal orifice is located 3cm below the urethral orifice. General examination revealed Tanner stage 3 breasts and axillary hair. Patient's skin was dry with multiple hypo and hyper pigmented patches which have been present since the fourth day of birth. Patient was thus diagnosed with Kindlers syndrome (figure 3).

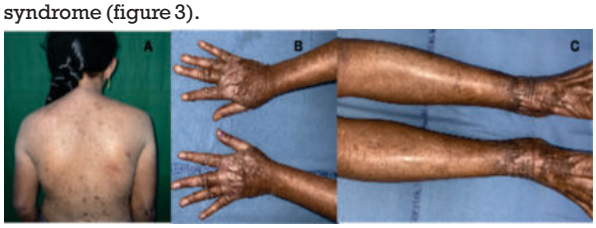

Figure 3A, B, C: Skin images of 20 yr. old woman showing dry skin with multiple mixed hyper and hypo pigmented patches.

Patient was referred to the radiology department for VCUG with the diagnosis of anomalous outflow tract of urethra and vagina with PID.

Diagnostic Assessment

VCUG was done under fluoroscopy and spot films were taken in full bladder and during voiding phases. During micturition two separate contrast filled tracts were noted from the urethra, one extending proximally and superiorly into the bladder neck, and the other tract extending more postero-superiorly. Bladder appeared normal in capacity with no evidence of wall irregularities (figure4).

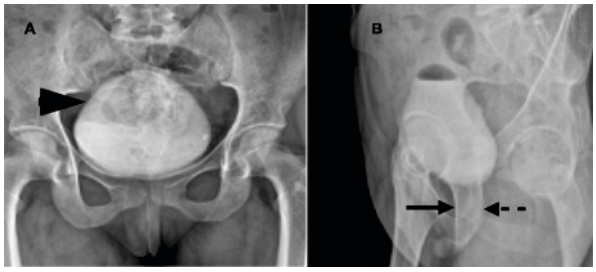

Figure 4A and 4B: VCUG spot films of full bladder(A) and micturition (B) showing normal bladder (arrowhead) and two urinary streams one opening into the bladder neck(arrow) and other opening more posteriorly into vagina (open arrow). There is mild overlapping of bones over the urine stream.

CT cystogram was done in the same setting as VCUG which revealed contrast in the bladder as well as contrast in both urethra and vagina figure (5 A and B).

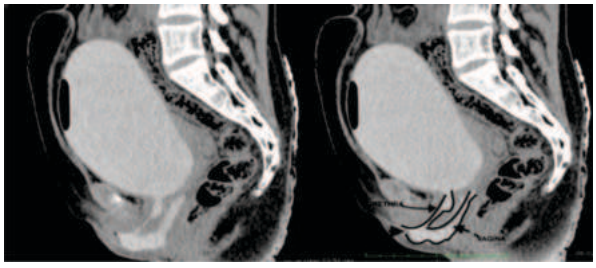

Figure (5 A and B): Sagittal section of CT cystogram showing posterior contrast filled vaginal tract joining the anterior urethral tract forming a common channel.

A common urogenital channel measured approximately 3-4 cm in length and the urogenital septum measured approximately 9 mm in thickness (figure 6).

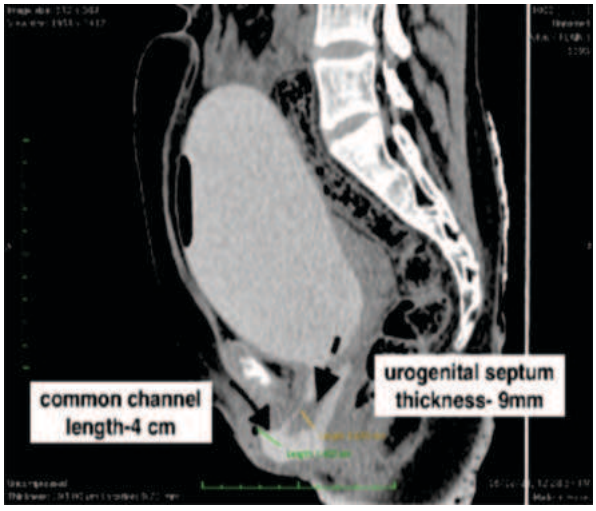

Figure 6: sagittal section of ct cystogram during voiding phase showing contrast filled anterior urethral and posterior vaginal tracts. Length of Common channel was 3-4 cm and thickness of urogenital septum was 9-10mm.

Contrast MRI of pelvis revealed normal sized uterus with intense post contrast enhancement. Moderate to gross bilateral hydrosalpinx and fat stranding in the pelvis suggested pelvic inflammatory disease. (figure 7 A and B).

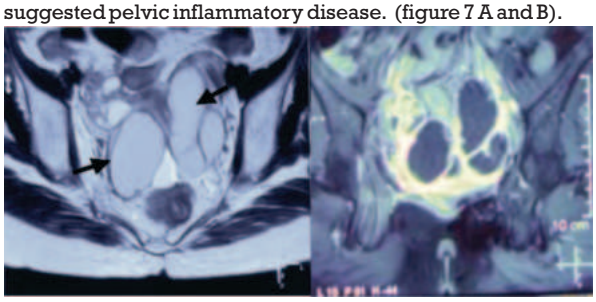

Figure 7A: Axial section of T2W MRI pelvis shows dilated, oblong, convoluted right (arrow) and left (arrow head) fallopian tubes. 7b: coronal section of post contrast T1W image shows intense enhancement of pelvic structures and fat stranding.

Uterus and vaginal tract appeared normal in length and structure (figure 8).

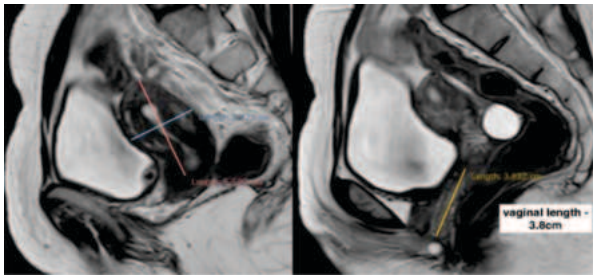

Figure 8A and 8B: sagittal T2W pelvis showing normal sized bladder, uterus and vagina.

Bilateral ovaries showed few simple cysts. Right ovary also revealed a cyst with haemorrhagic component. (Figure 9)

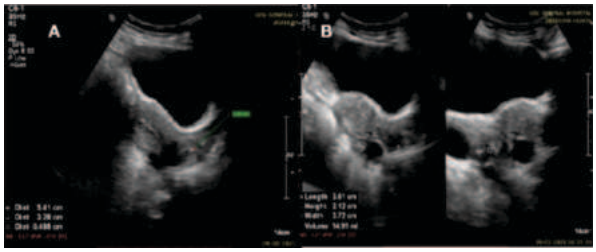

Figure 9: Sagittal(A) and axial sections (B) of ultrasound pelvis showing normal uterine endometrium and simple cysts in ovaries (B).

On cystoscopic evaluation, urethral and vaginal openings were seen after passing through a common channel of length 3-4 cm from the external orifice. The same cystoscope was passed through the urethral opening anteriorly and vagina posteriorly. Cervix gripping was felt during cystoscopy and the cystoscope was advanced into the endometrial cavity through the vagina, thus confirming persistent urogenital sinus. Urogenital septum separating the urethra and vagina measured 9-10 mm thick (figure10).

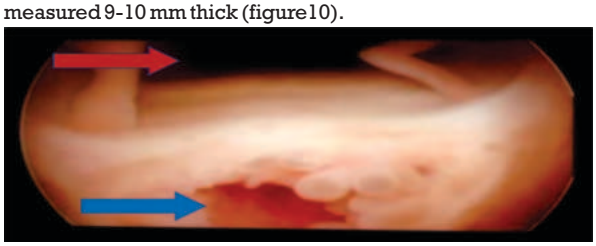

Figure 10: Cystoscopy image showing anterior urethral opening (red arrow) and posterior vaginal opening (blue arrow) after passing through the common channel.

Patient blood investigations, renal function tests and complete urine analysis were normal, and patient was referred to gynaecology and urology for further treatment

DISCUSSION

Persistent urogenital sinus (PUGS) is a congenital abnormality characterized by an abnormal communication between the urethra and vagina, with an incidence of 0.6/10,000 female births [5](Valentini et al., 2016).

Embryology Of CLOACA

The genital and lower urinary tract is embryologically derived from a common entity known as the urogenital sinus. The urogenital sinus is the ventral partition of the cloaca, dorsal partition of which results in the lower alimentary tract. This partition occurs around the 6th week of intrauterine life, by the development of a urorectal septum. The failure of division of the cloaca by the urorectal septum leads to a spectrum of anorectal malformations[3](Berrocal et al., 2002).

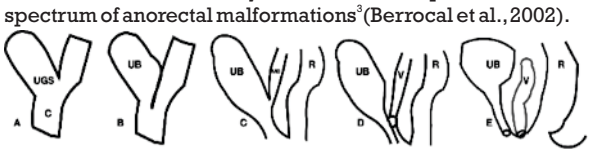

Figure 11: Development of female genitourinary system. A: At 4 weeks GA, the urogenital sinus and rectum form a common channel, the cloaca. B: At 6 weeks of GA, the urorectal septum develops in caudal direction, separating the cloaca into a ventral urogenital sinus and a dorsal rectum. C: At 8 weeks, 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 8th and 12th 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; UGS = urogenital sinus; R = rectum; MD = Müllerian ducts; UB = bladder; V = vagina.)

The migration of the Mullerian ducts results in the separation of the urogenital sinus into the renal and the genital system components. The early arrest of migration of the Müllerian ducts results in a long urogenital sinus(common channel) with a short vagina and a high urethral opening, while late arrested migration results in a short urogenital sinus with a vaginal tract of almost normal length and a low urethral opening. Our case corresponds to arrested migration of Mullerian ducts in a late stage with short urogenital sinus.

Classification

PUGS can be classified into high confluence and low confluence anomalies based on the length of the common channel, whether it is >3 cm or <3 cm.

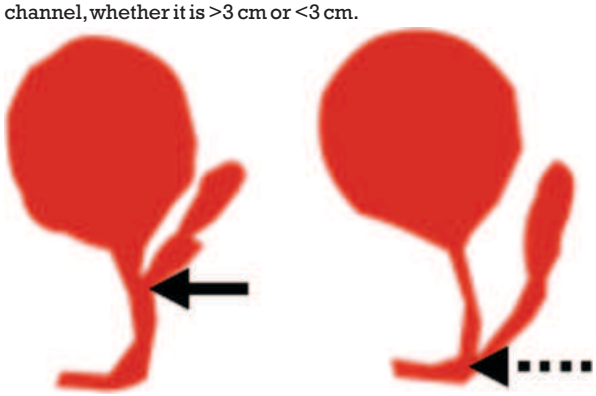

Figure12: Illustration showing high (arrow) and low confluence (dotted arrow) urogenital sinus.

Studies which were recorded in the West have attributed the condition not only to the abnormal foetal development, but also to conditions such disordered sexual differentiation (DSD). Among these, the most common is congenital adrenal hyperplasia (CAH). The severity of virilization in females with CAH is reflected in the degree of clitoral hypertrophy and the length of the tubular urethra. When clitoral hypertrophy is mild, the vagina and urethra are usually present as separate structures for most of their length with only a short distal common channel (urogenital sinus). [2]Thomas, 2020.

The literature available on PUGS till date also shows that short urogenital sinuses are more common, and the current case also records similar observations.

Syndromic Associations

PUGS could manifest as a singular entity or as part of a syndrome (McKusick-Kaufman Syndrome). McKusick-Kaufman Syndrome (SMK) is an autosomal recessive multiple malformation syndrome characterized by hydrometrocolpos, congenital heart disease and polydactyly, few of the cases showing PUGS. PUGS along with digit anomalies, retinitis pigmentosa, obesity, learning disabilities and male hypogenitalism are features of Bardet-Biedl syndrome.

However our case was associated with kindler syndrome, presenting with skin abnormalities and esophageal webs.

Other conditions associated with persistent urogenital sinus are uterine anomalies, vaginal atresia or duplication, hydronephrosis, renal agenesis, imperforate anus, polycystic kidneys, oesophageal atresia and sacral hypoplasia[4,13,14](Tan et al., 2017a, 2017b, Taori et al., 2010.), which are absent in our case.

Contrary to the presentation of the current case, PUGS most commonly presents as a pelvic mass, which may be due to a distended bladder, hydrometrocolpos, or both. Neonatal presentation with hydrocolpos should draw attention to the nature of the condition, as it demands early recognition and intervention in order to prevent detrimental compressive symptoms on adjacent structures (dilatation of upper urinary tract with urinary retention, compression of the inferior vena cava, or elevation of the diaphragm). In few cases, severe hydrometrocolpos may result in the passage of fluid from uterus to abdominal cavity through the tubes, causing hemoperitoneum and subsequent peritoneal irritation [11]Simonetti et al., 2018)

Imaging In Pugs

MCUG is the first diagnostic modality of choice for evaluating PUGS. However sometimes distended vagina may be mistaken as the bladder on VCUG. Sometimes VCUG is difficult to perform in neonates. In such scenarios, an ascending genitogram may constitute a better imaging modality as it can clearly demonstrate the length of the common channel and the level of vaginal confluence. It also helps the surgeon to choose a suitable approach for surgical repair. In foetuses and neonates, however ultrasound gives much valuable information about the anatomy and associated conditions. Besides conventional imaging methods, the use of MRI pelvis has gained popularity as it can clearly depict the relationship between urogenital structures, the pelvic muscle complex and any associated spinal or renal abnormalities. Additionally, a combination of computed tomography of pelvis, MRI and fluoroscopy of the urogenital tract can help to illustrate the anatomy of associated structures in 3D plane, as we have done in this case. 'Written informed patient consent for publication has been obtained.'

Outcome

Clinical management and surgical correction and reconstruction of this embryonal condition is based on the distance of the vaginal confluence from the bladder neck and perineal meatus [4] Tan et al., 2017a, 2017b. High-confluence variants are associated with a greater incidence of co-existing

spinal anomalies and have poorer outcomes from reconstructive surgery than low confluence variants. Less severe forms of urogenital sinus are usually corrected by use of a perineal-based skin flap incorporated into an incision in the posterior vaginal wall.

Take Home Message

Anomalies of the urogenital tract are amongst the most common organ system anomalies found in the foetus or neonate. A complete understanding of the embryologic development of the urinary and genital tract is very useful in the evaluation and treatment of a child or adult with a congenital genito-urinary malformation [3]

Teaching Points

1. Cloacal malformations are congenital anomalies in which the urinary, genital, and gastrointestinal tracts all drain through a common perineal opening. It occurs due to the failure of the uro-rectal septum to join the cloacal membrane during the 4th to 6th week of GA. This results in the formation of ano-rectal malformations and single orifice in the perineum of phenotypic female draining urine, genital secretions and faeces
2. Persistent urogenital sinus occurs due to the failure of Müllerian duct migration caudally from the Müllerian tubercle to the vestibule during 6-12 weeks of GA. this results in two perineal openings one draining urine, genital secretions and other draining faeces/meconium in phenotypic females.
3. It is necessary for treating physicians and surgeons to be aware of the embryology of cloaca and imaging features of urogenital anomalies to accurately diagnose the condition along with any syndromic associations and treat them.

Abbreviations

CAH - Congenital Adrenal Hyperplasia
CT - Computed Tomography
DSD - Disordered sexual differentiation
GA - Gestational age
MRI - Magnetic Resonance Imaging
PID - Pelvic Inflammatory Disease
PUGS - Persistent Urogenital Sinus
SMK - McKusick-Kaufman Syndrome
USG - Ultrasonography
VCUG - Voiding Cysto-Urethrogram

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