Society for Fetal Urology
Fall 2023 Case Reports

FROM THE GUEST EDITOR
Evalynn Vasquez, MD
Children’s Hospital Los Angeles

The Society of Fetal Urology meeting was held in conjunction with the SPU Fall Congress in Houston. As is tradition, interesting case presentations were selected and presented by medical students, urology residents, and pediatric urology fellows. This year’s cases highlighted the wide variety of congenital diagnoses that can present both prenatally and postnatally and was moderated by Duong D. Tu, MD from University of Minnesota, Masonic Children’s Hospital and Yvonne Y. Chan from University of Texas Southwestern Medical Center. Awards were given based on the collective strength of the written abstract, oral presentation, and the teaching opportunities of the case. The first-place award was given to Catalina Sanchez from the Hospital of Sick Children in Toronto for the succinct description and surgical management of bladder embryonal rhabdomyosarcoma. The authors provided an exceptionally written abstract, illustrations, and radiologic images depicting the rare perinatal presentation of two patients, a female with obstructive uropathy due to the tumor and a male with concomitant posterior urethral valves. Adding to the complexity of this case, the male patient may not have been prenatally diagnosed without the obstructive uropathy from the posterior urethral valves. The second-place award was given to Maya Overland from Children’s Hospital of Philadelphia, who described a patient with Prune-Belly Syndrome with Urethral Hypoplasia. The case was managed with vesicoamniotic shunts, then vesicostomy, then the P.A.D.U.A procedure. This case highlighted how one patient can present with several urologic diagnoses and the role a pediatric urologist has in the ongoing urologic treatment of the complex patients. The third-place prize went to Megan M. Roedel from the University of Wisconsin with the abstract entitled Challenges in Managing Complex Genitourinary Anomalies in a Neonatal Patient: Emphasizing the Role of Parental Engagement. This case underscored the important role a Pediatric Urologist can play in a Fetal-Maternal Care Center during a prenatal consultation and how the inability to receive this consultation can lead to the detriment of the child, as well as the pregnant mother.

Ashley Sims, BSN, CCRN, C-NPT from Texas Children’s Hospital Fetal Center Clinical Nurse Manager shared her perspective both as a medical care provider and as a patient. She described her experience as a Nurse Care partner when she became pregnant and needed prenatal consultations from Nephrology and Urology. She described how this changed her perspective as a provider because she could better empathize with her patients. She saw firsthand how much speaking with specialists during her pregnancy eased her anxiety. They were able to answer all of her questions and come up with a plan after delivery. She also realized that as providers, we see certain diagnoses as “minor” and non-life threatening. She warned against downplaying the magnitude of any diagnosis because pregnant mothers are feeling many emotions, including grieving the loss of a “normal” pregnancy.

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Lastly, a panel of experts engaged in an enlightening discussion and sharing of opinions about the management of ureteroceles and cecoureteroceles. Christina Ching, MD from Nationwide Children’s Hospital, Chris Cooper, MD from University of Iowa Stead Family Children’s Hospital, Rafael Gonsalbez, MD from Nicklaus Children’s Hospital, and Erin McNamara, MD-MPH from Boston Children’s Hospital reviewed three cases which brought to light the variation in presentation and management. They discussed different pathways for diagnostic work-up and what could be learned based on the nuanced findings on diagnostic tests, such as the VCUG. The discussion for surgical management of patients with ureteroceles and cecoureteroceles spanned puncturing, reimplants, ureteroureterostomies, cutaneous ureterostomies, vesicostomies, hemi-nephrectomies, and bladder neck reconstruction. Chris Austin, MD, summarized the consensus of this subject with this quote: “Ureteroceles are the most malignant benign etiology that Pediatric Urologists treat.” Many attendees commented on how helpful this panel discussion was in terms of the high-yield learning on a difficult subject. If you would like to learn more, Hans Pohl, MD recently published a review in *Urology Clinics* titled Embryology, Treatment, and Outcomes of Ureteroceles in Children.

Complete Sagittal Bladder Duplication in a Male

Callum Lavoie, Andy Y. Chang
Department of Surgery, Division of Urology, Children’s Hospital of Los Angeles

**Introduction**

Bladder duplication is a rare anomaly with roughly only 70 reported cases in the English literature. It can be associated with duplication anomalies of the urethra, genitalia, and GI tract with complete duplication being distinguished by each distinct bladder having its own ureter and urethra.

Embryologically, the bladder derives from the urogenital sinus which is an endoderm structure, and the trigone derives from the mesonephric ducts which is a mesoderm structure. Normally, at 5-6 weeks of fetal development, the cloaca is divided into the ventral urogenital sinus and the dorsal anorectal canal by the urorectal septum. Prior to the formation of the urorectal septum, the mesonephric (Wolffian) ducts fuse with the cloaca and the ureteral bud separates and ultimately forms the ureteral tunnel within the detrusor.2

Duplication events are theorized to be secondary to abnormal budding of the urogenital sinus during fetal development, with some specific theories being published including that by Abrahamson and colleagues which theorizes supernumerary cloacal septum indenting the epithelial wall of the bladder causing it to split, or excessive constriction between urogenital and vesicourethral portions of the ventral cloaca.1

**Case Report**

Our patient (E.N.) was born at term with an omphalocele and no other known anomalies. This was repaired uneventfully on day 2 of life. Post-operatively, routine abdominal US identified 2 indeterminate fluid-filled structures in the pelvis and an MRI was then performed which confirmed a duplicated bladder (Figure 1). A VCUG was performed which showed no communication between the two bladders, grade 4 right VUR when the right bladder was catheterized, and no VUR on the left side with filling of the left bladder.

The patient remained stable on prophylactic antibiotics until 16 months of age when he developed right pyelonephritis. After this was treated, he was taken to the operating room at 17 months of age for cystoscopy and PIC cystograms of both bladders. In the OR we identified a urethral duplication beginning in the membranous urethra in a Type IIb-III Y-configuration with a branch going into each bladder (Figure 2).4 The bladders had a normal appearance bilaterally with an orthotopic left ureteric orifice with no reflux, and a laterally displaced right ureteric orifice with no reflux exhibited at this time.

**Figure 1:** Axial and Coronal MRI images illustrating complete sagittal bladder duplication.
Complete Sagittal Bladder Duplication (continued from previous page)

Conclusion:
Bladder duplication is an extremely rare congenital anomaly with variable management strategies depending on the associated findings. In the absence of adverse symptoms, infection, renal dysfunction, obstruction, or reflux, conservative management with close follow-up is advised. Our patient, E.N., continues to be doing well at this time at an age of 21 months with no further complications and off prophylactic antibiotics with plans for continued monitoring but no aggressive intervention. It would be very interesting and informative to perform video urodynamics at an older age to investigate detrusor and sphincteric function of each independent bladder.

References

Figure 2: Cystoscopic view of urethral duplication beginning at the bulbomembranous junction and extending into two independent bladders

A Rare Case of Bladder Outlet Obstruction in a Female Fetus with Misleading Prenatal Imaging

Emily Clennon, MD, MPH, Sarah Hecht, MD, Julie Cheng, MD, & J. Christopher Austin, MD
Division of Pediatric Urology, Doernbecher Children’s Hospital, Oregon Health & Sciences University

Background
Cloacal anomalies are a spectrum of congenital malformations of the urinary, genital, and intestinal tracts. Though cloacal anomalies are often first detected on prenatal ultrasound, fetal MRI is often performed in these cases and is considered more accurate in establishing the diagnosis and anatomy. Classic MRI findings in cloacal anomalies include intestinal obstruction and dilation, pelvic cystic structures, and urologic and spinal abnormalities.1,3

Case
A 28-year-old G3P1 female presented at 22 weeks gestation after prenatal ultrasound revealed severe bilateral hydronephrosis, a pelvic cystic structure, female external genitalia, and a single perineal orifice suspicious for cloacal anomaly. Non-invasive prenatal testing indicated a 46XX fetus. Fetal MRI at 27 weeks gestation demonstrated a normal spine, normal colon and rectum with meconium extending to the perineum, and a structure communicating with the perineum suggesting a urogenital sinus abnormality rather than a cloaca (Figure 1).

Figure 1. Fetal MRI at 27 weeks demonstrating dilated cystic structure in anterior pelvis and meconium signal extending to perineum.

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Bladder Outlet Obstruction (continued from previous page)

She was born at 37 weeks after induction of labor for new oligohydramnios. Examination revealed dysmorphic genitalia without palpable gonads, fused labioscrotal folds, a perineal orifice, and urine draining from the end of a phallic structure with no visible meatus (Figure 2). Postnatal ultrasound showed UTD P3 bilateral hydronephrosis, a dilated proximal urethra, hydrocolpos, and the distal rectum ending 7 mm from the perineal skin.

Exam under anesthesia, retrograde urethrogram, vaginoscopy, cutaneous vesicostomy, and diverting loop colostomy were performed on the first day of life. The urethra opened at the tip of the glans clitoris and was atretic, precluding catheterization. A 1 cm perineal common channel located anterior to the expected location of the anus led to a confluence of the single vagina and rectum. VCUG via vesicostomy demonstrated bilateral grade V vesicoureteral reflux without visualization of the urethra or vagina.

Cystoscopy, vaginoscopy, and cystogram at 4 months of life demonstrated 1cm of normal proximal urethra prior to atresia and laterally displaced, gaping ureteral orifices. Spinal MRI revealed a fibrolipoma of the filum terminale, and she underwent spinal cord detethering at 10 months old. At age 14 months, she underwent posterior sagittal anorectovaginoplasty with vaginoplasty and labioplasty. The urethra was re-evaluated at that time and found to be too small for progressive dilation.

Her clinical course has been complicated by many urinary tract infections and stage IV chronic kidney disease with concomitant feeding difficulties and gastric tube dependence. Further urologic management has been deferred until it is clear whether early renal transplantation is indicated but will include urodynamics and likely either urethroplasty or appendicovesicostomy.

Conclusion

Cloacal anomalies are challenging to diagnose prenatally with only approximately 6% of cases being diagnosed on screening ultrasound. The most common findings associated with cloacas on ultrasound are pelvic cystic mass (40%) and hydronephrosis (36%), and the combinations of intestinal obstruction of bowel calcifications and either urologic or abnormalities are highly suggestive of cloaca. In cases where cloaca is suspected, MRI is typically performed as it is widely regarded as superior to US in delineating anatomy of these cases. Rohrer et al. found that MRI had a detection rate of 50% in a series of anorectal malformation cases compared to US’s 31%. MRI findings in cloaca have been described in many series and are consistent with those demonstrated on ultrasound.

Posterior cloacal anomalies are malformations in which the urethra and vagina are fused and form a urogenital sinus that opens in or immediately anterior to the rectum. Posterior cloacal anomalies and variants thereof – as in this case – can be particularly difficult to diagnose prenatally, as they often do not have the classic imaging findings associated with cloacal malformation. This patient did not have intestinal dilation, hydrocolpos, or spinal abnormalities on any prenatal imaging. Meconium was visualized at the level of the perineum on imaging not because the rectum was normally developed but because meconium was draining into the common channel. This case demonstrates the limitations of prenatal MRI in the diagnosis of posterior cloacal variants. A high index of suspicion for these diagnoses should be maintained if suggested on other imaging modalities.

References

Seeing Double: A Case of Bladder and Gastrointestinal Duplications

Deepansh Dalela, MD, Alexandra M. C. Carolan, MD, and Micah A. Jacobs, MD
University of Texas Southwestern Medical Center, Children’s Health, Dallas, TX

Review of Case

A male infant was born at 36 weeks to a 29-year old G3P2 mother. Prenatal imaging was concerning for esophageal atresia with possible trachea-esophageal fistula, polyhydramnios and possible ureterocele. The kidneys appeared normal. Due to ongoing feeding intolerance on day of life 27, abdominal ultrasonography was performed to rule out malrotation of the gut. This revealed a 3.5 cm pelvic cyst adjacent to the bladder. On physical exam, there was a coronal hypospadias, but the rest of the genital exam was unremarkable with no accessory perineal openings. In order to further characterize the cystic structure, magnetic resonance imaging of the abdomen and pelvis was performed, demonstrating bladder duplication in the sagittal plane (Figure 1), bilaterally morphologically normal kidneys, and a urethra from each bladder that appeared to converge at the level of the prostate.

The baby continued to have persistent feeding difficulties and was taken to the operating room by Pediatric Surgery at around 8 weeks of life for gastrostomy tube placement. Intraoperative findings included intestinal malrotation, along with several gastrointestinal duplications—gallbladder, terminal ileum, right and left colon, and rectum, with one rectal limb leading to the anus and another blind ending limb. He underwent a side-to-side ileal anastomoses and end ileostomy with mucus fistula.

At 6 months of age, greenish-brown feculent discharge was noted from the urethra. Retrograde urethrogram did not demonstrate entero-urethral fistula, with the urethral catheter preferentially entering the left bladder. On cystoscopy in the operating room, there was normal urethra up to the proximal bulbar urethra where it split (Figure 2), no discernible verumontanum, and no fistula with the gastrointestinal tract.

Following a prolonged NICU stay, the baby was seen in clinic at 9 months of age and was growing well with enough wet diapers, no reported urinary tract infections, and no further reports of feculent discharge from the urethra. Plans for hypospadias repair were discussed should the patient require general anesthesia for any other intervention, and other urological interventions will be considered if voiding dysfunction, urinary incontinence, or urinary tract infections arise.

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Bladder and Gastrointestinal Duplications (continued from previous page)

Discussion:

Bladder duplication is a very rare congenital anomaly, with case reports and small series in literature. Anatomically, bladder duplication is described based on Abrahamson 1961 classification: complete bladder duplication (with duplication of bladder and urethra), incomplete bladder duplication (with a single urethra), complete sagittal septum and incomplete sagittal or frontal septum. Typically, bladder duplication occurs in the sagittal plane, as noted in our patient. While the etiology is largely unknown, it is commonly associated with varying degrees of urethral duplication, as defined by the Effman classification, and rectourethral/urethrovaginal fistulae. Specifically, our case could be described as a variant of either complete sagittal septum or complete bladder duplication, with a urethra from each bladder uniting distally (Effman Type 2b urethral duplication) at the level of proximal bulbar/bulbo-membranous urethra. Additionally, as seen in our patient, lower gastrointestinal duplications have been reported in up to nearly 1/3rd of cases. While there was a concern for rectourethral fistula at one point, focused evaluation did not reveal this in our patient.

While bladder duplication can be diagnosed antenatally, the more common presentation is postnatal, either symptomatic or incidental diagnosis on imaging. Clinical presentation for bladder duplication varies and is predicated on the specific anatomy. For patients with urethral duplication, persistent incontinence or inability to toilet train can be the presenting symptoms. Contrast enhanced imaging, such as voiding cystourethrogram, retrograde urethrogram and magnetic resonance urogram can aid in diagnosis. In cases of poorly draining bladders, hydronephrosis, urolithiasis and/or recurrent urinary tract infection puts the ipsilateral renal moiety at risk. This may necessitate temporizing vesicostomy, cystoscopic bladder septostomy, or resection of the non-functional bladder and urethral reimplant into the functioning one. Isolated bladder duplication without upper tract obstruction often can be managed conservatively, while those associated with higher degrees of urethral duplication, urinary fistula or incontinence may need surgical correction. Lastly, more complex associated anomalies (such as vaginal duplication or bladder/cloacal extrophy), may require a staged reconstruction of the bladder, with or without bladder augment and bladder outlet procedures.

References

Early Delivery In A Patient With Antenatally Detected Phimosis, Urethral Dilation, Thickened Bladder Wall, Progressive Bilateral Hydroureteronephrosis, And Renal Dysplasia

William Fox, University of Illinois College of Medicine Rockford
Patrick H. McKenna MD, FACS, FAAP, Director of GU Reconstructive Center Mercyhealth, Professor of Surgery University of Illinois College of Medicine Rockford

Introduction

Prune belly syndrome is diagnosed based on the presence of a triad of patient findings: abnormal abdominal wall musculature, undescended testes, and urinary tract abnormalities. The cause of this disease has yet to be definitively proven, however it may be due to obstructive, embryologic, or genetic abnormalities. Patient outcomes range from stillbirth to living capable adult; however, this varies depending on disease severity. Surgical management should be tailored to specific patients as there is not an accepted definitive operation for all prune belly patients. In this case we present a patient who was picked up in the second trimester, however adequate amniotic fluid was maintained throughout the pregnancy. Antenatal intervention is seldom indicated but in situations with worsening hydronephrosis, parenchymal thinning, and cystic changes, early delivery is a safe option for antenatal intervention.

Case

The patient underwent an ultrasound at 20 weeks gestation which identified bilateral upper tract dilation and bladder wall thickening (Figure 1). Amniocentesis confirmed a normal male, with no chromosomal anomalies.
somal abnormalities. Subsequent ultrasounds showed a dilated bladder with a probable keyhole sign, bilateral dilated ureters, bilateral hydronephrosis and a cystic mass seen on the perineum. Ultrasounds suggested severe phimosis with urethral dilation and worsening bilateral hydroureteronephrosis with bilateral dysplastic and cystic changes in the left kidney. The patient was referred to a tertiary center that performs antenatal interventions. At the center, it was noted he had cardiac abnormalities, however they did not recommend early intervention. They believed it was less likely posterior urethral valves but commented on the megalourethra secondary to meatal stenosis. The center felt that there would be a high likelihood of renal failure at birth. At 32 weeks, a left pulmonary effusion was noted. As the patient was followed to 34 weeks, hydronephrosis worsened and renal parenchymal echogenicity increased with cystic changes. Parents elected for an early delivery at 35 weeks.

On delivery, the patient had a variant of prune belly syndrome. Initial creatinine 0.9mg/dL rising to 1.4mg/dL over 48 hours despite having an indwelling catheter. Patient had good urine output, and was monitored with an indwelling catheter for 8 days without creatinine change. Workup identified severe phimosis with a dilated urethra, no palpable testis, thickened bladder wall, and bilateral refluxing obstructing megaureters (Figure 2). The parents considered multiple options, and elected to undergo bilateral end cutaneous ureterostomies. At the time of surgery, both ureters had distal adynamic segments, with ureters proximal to this segment massively dilated. Patient had a megalourethra with severe phimosis, and a distal urethral fistula. Postoperatively, creatinine lowered to 0.5mg/dL with resolution of bilateral hydroureteronephrosis.

Discussion

This is a rare case of severe urethral obstruction and bilateral refluxing obstructing megaureter resulting in a prune belly variant. Antenatal intervention is seldom indicated, however this represents a case where the best form of antenatal intervention was early delivery. Likewise, upper tract diversion is seldom indicated, however in this rare variant upper tract diversion provided the best drainage.

Citations

From Prune Belly Syndrome (PBS) to Megacystis, Microcolon, Intestinal Hypoperistalsis Syndrome (MMIHS): A Familial Link with Diverging Outcomes

Allison Grant, Soo Jeong Kim, Christina P. Carpenter, Belinda Li  
New York Presbyterian – Morgan Stanley Children’s Hospital, New York, NY

Introduction

Megacystis microcolon intestinal hypoperistalsis syndrome (MMIHS), or Berdon syndrome, is a congenital disorder of visceral smooth muscle function characterized by an enlarged bladder, microcolon, and decreased intestinal motility. The syndrome is most often diagnosed prenatally and has a potentially poor prognosis. First reported by Berdon and colleagues in 1976, MMIHS has been reported in the literature in roughly 450 cases and has gained traction in the field of genetics. Herein we report a unique case of MMIHS with favorable prognosis and a surprising familial link.

Case report

The patient is a 12-month-old girl with third trimester ultrasound (US) findings of fetal megacystis (Figure 1). Specifically, US identified a distended (5.5x4cm), irregularly shaped bladder, without bladder wall hypertrophy. No other abnormalities were identified, and amniotic fluid index was normal. Given her sex, the parents were extensively counseled on the differential diagnoses of MMIHS or cloacal anomaly, discussing their rarity and potentially poor prognosis. Notably, the patient’s father was diagnosed with prune belly syndrome (PBS) in childhood. He has an ileostomy and incontinent urostomy after undergoing 50 reconstructive surgeries.

After discussing the potential diagnoses, the patient’s mother underwent amniocentesis with whole genome sequencing. A pathogenic variant in the ACTG2 gene was identified, with autosomal dominant inheritance from Dad. This not only confirmed our patient’s diagnosis of MMIHS, but also suggested that the father’s PBS may be better characterized as being part of the MMIHS spectrum, which would explain some of his intestinal complications.

The patient was delivered at 38 weeks and 4 days via c-section for breech presentation. Upon delivery, a nasogastric (NG) tube was immediately placed for gastrointestinal tract decompression in the setting of potential microcolon and intestinal hypoperistalsis. Renal bladder US (Figure 2) and abdominal X-ray obtained on day of life one showed large bladder capacity (62cc) with normal kidneys, and normal bowel gas pattern, respectively. She voided spontaneously, with mild to moderate post void residual (PVR), so was initiated on clean intermittent catheterization (CIC) every six hours. Favorably, she demonstrated spontaneous bowel function, so NG tube was removed. The patient tolerated oral feeding, passed all newborn milestones, and was discharged on CIC.

Video urodynamics (UDS) at 2 months of life was performed and found large bladder capacity (>60cc), normal compliance, normal voiding detrusor pressure, and a low to moderate PVR. She was transitioned to CIC every 12 hours and eventually weaned off catheterization altogether with US confirmation of complete bladder emptying. Since birth, the patient has had appropriate oral intake, weight gain, and bowel function. She continues to thrive and is currently doing well.

Discussion

Our case, including reclassifying Dad’s PBS as part of the MMIHS spectrum, highlights the autosomal dominant inheritance of the ACTG2 gene, the resulting phenotypic spectrum of smooth muscle disease, and range of prognoses. Gene sequencing links ACTG2 gene mutations to a spectrum of smooth muscle diseases affecting the bladder and GI systems, known as ACTG2 visceral myopathy. MMIHS and PBS both fall on this spectrum of visceral myopathies caused by ACTG2 (continued on next page)
gene mutation\(^3\). The mutation is noted to have complete penetrance and variable expressivity.

A case series of 53 families with visceral myopathies found that 26 of the families had a de novo or inherited ACTG2 mutation. Of the ACTG2 related visceral myopathies, 92\% (24/26) had megacystis and 62\% (16/26) had microcolon. The researchers determined that within the cohort, 75-90\% of the MMIHS specific disease features (i.e., megacystis and microcolon) could be attributed to ACTG2 mutations. Notably, only one of the cases had prune belly syndrome and this was not associated with ACTG2. This paper highlights that the ACTG2 gene mutation is more closely associated with the MMIHS diagnosis/phenotype than it is with PBS\(^4\).

While there may be diagnostic uncertainty when it comes to phenotypically similar visceral myopathies, in the setting of ACTG2 gene mutation, a diagnosis of MMIHS is more likely than a diagnosis of PBS. This highlights the importance of genetic testing in the diagnostic workup of visceral myopathies as well as the need for further research into the genetics of PBS. With this information, we can better differentiate and diagnose the visceral myopathies both pre and postnatally.

References
Which Key for Which Hole? Unlocking the Mystery of Fraternal Twins with Posterior Urethral Valves

Kevin Hanna, Patricio C. Gargollo, Candace F. Granberg
Department of Urology, Mayo Clinic

Introduction

Posterior Urethral Valves (PUV) can be suspected prenatally and present with varying phenotypes. The incidence of PUV is rare in siblings. We present dichorionic, diamniotic twins with discordant prenatal ultrasounds, both found to have PUV.

Case Description

The twins were first noted to have hydronephrosis at 23 weeks’ gestation: Twin A had 5mm right renal pelvis dilation, and Twin B had bilateral renal pelvis dilation (1.2cm left, 2cm right) and keyhole sign concerning for PUV (Figure 1). On subsequent studies, both babies had alternating, undulating levels of bilateral hydronephrosis, at times raising confusion of which baby was which, and who had the keyhole sign. Notably, throughout pregnancy, amniotic fluid levels were normal. Delivery was at 37 weeks via planned C-section, and both made excellent wet diapers initially. Postnatal ultrasound at 48 hours revealed Twin A with bilateral SFU grade 4 hydroureteronephrosis, while Twin B had SFU grade 4 right, SFU grade 1 left hydronephrosis. Both were placed on antibiotics pending voiding cystourethroscopy (VCUG). Twin A had unsuccessful catheter placement for VCUG, while Twin B’s VCUG demonstrated significant proximal urethral dilation concerning for PUV (Figure 2). Both babies underwent cystoscopy in the first week of life, where both had evidence of PUV, ablated via cold knife (Figures 3 and 4). At last follow-up at 6 months of age, both had improved hydronephrosis and normal creatinine.

Discussion

Posterior Urethral Valves (PUV) pose a complex urological challenge with a wide range of phenotypes. PUV is estimated to affect 1 per 10,000 live births. While the precise cause of PUV remains elusive, several hypotheses, including genetic factors, environmental influences, and abnormal fetal development, have been proposed. Familial posterior valves have been reported in both twin and non-twin siblings, as well as instances of discordance in monozygotic twins, suggesting potentially both a genetic component as well as random mutation.

The clinical presentation of PUV exhibits wide variation, ranging from mild cases with minimal symptoms to severe phenotypes with life-threatening complications. Early urinary tract obstruction can lead to prenatal issues such as hydronephrosis, oligohydramnios, and most severely, pulmonary hypoplasia. Postnatally, affected infants may display symptoms including urinary tract infections, failure to thrive, and chronic kidney disease. Timely recognition is crucial for intervention and improved outcomes, as anywhere between 20 and 50% of boys with PUV will progress to ESRD in their lifetime.

Accurate diagnosis is paramount in managing PUV. While prenatal ultrasonography has revolutionized the antenatal detection of urinary tract anomalies, a definitive postnatal diagnosis often necessitates a combination of imaging modalities, including VCUG and renal ultrasonography. Ultimately, PUV is most accurately diagnosed via direct visualization on cystoscopy.

Figure 1: Twin B’s 23-week ultrasound demonstrating a “keyhole sign” highly suspicious for posterior urethral valves.
Effectively managing PUV requires a multidisciplinary team involving pediatric urologists, nephrologists, and neonatologists. Primary treatment involves relieving urethral obstruction through endoscopic ablation of the valves, though long-term sequelae such as neurogenic bladder leading to urinary tract infections and incontinence, chronic kidney disease, and surgical complications secondary to intervention including urethral injury and stricture can occur. As such, children with PUV require long-term follow-up.

PUV presents a multifaceted challenge in pediatric urology, demanding a comprehensive understanding of their etiology, clinical presentations, diagnosis, and management. Furthermore, we demonstrate the importance of maintaining a high degree of clinical suspicion for PUV especially in twins where the potential for missed urethral obstruction could have devastating long-term consequences.

References
Congenital Anterior Urethral Diverticulum Without Associated Anterior Urethral Valve in an Infant

Adele Raymo, University of Miami Miller School of Medicine / Aden Swayne, FIU Herbert Wertheim College of Medicine
Ruben Blachman, Jackson Memorial Hospital / Mariana F. Jucá Moscardi / Daniel M. Tennenbaum, Nicklaus Children’s Hospital
Luciana Lerendegui / Dr. Alireza Alam, Jackson Memorial Hospital

Introduction

Anterior urethra diverticulum is a rare condition, which can be congenital or acquired, with the latter being more common [1,2,3]. Congenital anterior urethral diverticulum (CAUD) is characterized by the formation of a saccular diverticulum in the anterior urethra [4]. The etiology of CAUD remains unclear, but it is postulated to be associated with incomplete development of the corpus spongiosum [5]. Here we report a case of CAUD in a five-week-old infant, without concomitant anterior urethral valve.

Case History

A five-week-old was referred to the pediatric urology clinic for genital evaluation. On physical examination the patient was found to have a ventral bulge located at the distal third of the penis. The meatus was normal in caliber and located orthotopically. The phallus was also normal in size and testicles were located within the scrotum. The patient presented intermittent dribbling of urine at the time of voiding, secondary to dilation of the diverticulum and urine accumulation, often requiring his parents to manually press the dilated urethra to fully empty the cavity after voiding. The patient reported no history of urinary tract infections (UTIs), and renal bladder sonogram at four-weeks of age was unremarkable. Management options were discussed with parents, and it was decided to perform a ventral cutaneous urethrostomy as the first step of a staged repair to optimize urinary drainage, decrease the risk of UTIs, and decrease operative time.

At six weeks of age the patient was taken to the operating room. A retrograde urethrogram showed a saccular dilation of the anterior urethra. A cystoscopy was performed and confirmed the presence of an anterior urethral diverticulum located 2-3 cm from the meatus and with no associated anterior urethral valve (Figure 1a). The proximal urethra was of normal appearance, the ureteral orifices were in orthotopic position and no trabeculation was observed. Then, ventral cutaneous urethrostomy was performed, at the proximal portion of the diverticulum a 1 cm longitudinal incision was made, and the urethra was then marsupialized (Figure 1b). The 6-French Foley catheter was placed into the bladder with no issues and removed on postoperative day five, after which the patient voided spontaneously. On the two months follow-up, he was voiding spontaneously and had no UTIs (Figure 2). The patient will likely undergo surgical correction at one year using an approach similar to that of a hypospadias treatment.

Discussion

Here we reported a rare case of a large CUAD without an associated anterior urethra valve that was managed with ventral cutaneous urethrostomy during infancy. The decision to operate at such an early age was due to urine outlet obstruction and increased risk of UTI secondary to the stasis of urine within the diverticulum. We decided to correct the CAUD with a staged approach as extended time under general anesthesia is not recommended at this age. After initial management with a ventral cutaneous urethrostomy, this patient can be further managed with reconstruction at a later age.

In conclusion, surgeons should be aware that if intervention of CAUD is needed during infancy, a staged reconstruction starting with a ventral cutaneous urethrostomy is a feasible management option.

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Congenital Anterior Urethral Diverticulum (continued from previous page)

Key Messages
1. Congenital Anterior Urethral Diverticulum (CAUD) without an associated anterior urethral valve is a rare anomaly in infants, characterized by the formation of a sac-like pouch in the anterior urethra, causing urine accumulation and obstructed urine flow.
2. Ventral cutaneous urethrostomy is an effective initial management strategy for large CAUD cases in infants without an accompanying anterior urethral valve. It helps improve urinary drainage, reduces the risk of urinary tract infections (UTIs), and can be followed by planned reconstruction at a later age.

References

Figure 2. Ventral cutaneous urethrostomy at 14 weeks after surgery remains open.
Introduction

We present the case of an infant female who was prenatally diagnosed with right upper pole hydronephrosis mimicking an obstructed duplex system. Antenatally the patient was found not to have a duplex system, but instead, right upper pole infundibular stenosis and massive hydrocalyx. This case report will detail the importance of certain circumstances in which more involved follow up studies may be important when prenatal hydronephrosis is diagnosed.

Case

Prenatally, our patient was found to have right upper pole hydronephrosis on the mother’s anatomy scan and was subsequently diagnosed as duplex collecting system presumed secondary to ectopic ureter. Postnatally, UTD P3 upper pole hydronephrosis was noted (Figure 1). After the child developed a febrile urinary tract infection, MAG3 was performed and confirmed the diagnosis of a duplex system without upper pole megaureter. VCUG was also performed showing no reflux. Worsening hydronephrosis on follow up ultrasound prompted cystovaginoscopy with retrograde pyelogram at 10 months of age. Intraoperatively, no upper pole ureteral orifice was identified, and retrograde pyelogram failed to reveal a calyceal diverticulum.

With the working diagnosis of ectopic ureteral insertion we planned for ureteroureterostomy; however, pelvic exploration revealed no upper pole ureter. Post-operatively a CT urogram showed filling but no drainage from the hydronephrotic calyx (Figure 2a). A delayed radiograph revealed contrast indicating a small connection to the renal pelvis (Figure 2b). At this point, a severely stenotic infundibulum with massive hydrocalyx was hypothesized as the diagnosis. Our patient then underwent ureteroscopy with laser incision through the stenotic infundibulum with ureteral stent placement for one month. At her three month postoperative visit, renal ultrasound showed improvement in the hydrocalyx and MAG3 renogram showed preservation of function with good drainage.

Discussion

Hydrocalyx secondary to infundibular stenosis can imitate alternative pathologies including renal cysts and duplex collecting systems. Our case was ultimately diagnosed after a surgical exploration with absence of a duplex ureter. Infundibular stenosis, although rare, should be considered for children of all ages with hydrocalyx without associated hydroureter. The absence of hydroureter in patient’s diagnosed with obstructed upper pole moiety should prompt additional workup to rule out congenital infundibular stenosis.

Figure 2a: CTU demonstrating functional right upper pole with contrast filling a hydrocalyx without obvious drainage and no duplex ureter.

Figure 2b: KUB 6 hours after CTU showing persistent contrast filling the right superior pole hydrocalyx. KUB 12 hours after CTU showing clearance of contrast.
The Case of A Congenital Female Urethral Diverticulum
Alexandra R. Siegal, MD1, Fernando A. Ferrer, MD2, Neha R. Malhotra, MD2

1 Icahn School of Medicine at Mount Sinai, Department of Urology, New York, NY, USA
2 Mount Sinai Kravis Children’s Hospital, Department of Pediatric Urology, New York, NY, USA

We present a case of a neonatal female with a congenital urethral diverticulum. At birth urology was consulted for a 10mm midline interlabial bulge (Figure 1). A normal hymenal flap with a posterior midline opening was also noted on physical exam. A 3.5 Fr umbilical catheter was able to be placed into the urethra, superior to the midline bulge, without lateral displacement. The child had normal voiding. Ultrasound demonstrated a thin-walled, hypoechoic, avascular mass, a mildly distended vagina with fluid, and normal kidneys/bladder (Figure 2A-B).

The differential diagnosis included paraurethral (Skene’s gland) cyst, imperforate hymen, urethral diverticulum or prolapse, prolapsed or ectopic ureterocele, paravaginal (Bartholin gland) cyst, or an inclusion cyst. The mass was thought to be mostly consistent with either a paraurethral cyst given the imaging findings, or an imperforate hymen given the position of a midline urethra superior to a vaginal bulge.

At 3 months of age, she was seen for follow-up. The bulge was noted to still be present and similar in size. Consistent with prior exam it was midline and just posterior to an orthotopic, patent urethra. The urethra and vagina were able to be catheterized with an 8 Fr feeding tube. At this point, there was less concern for an imperforate hymen. She was voiding well and had no abdominal pain or distention. A plan was made to repeat an exam and pelvic ultrasound at 6 months of age. Ultrasound at 6 months confirmed the presence of the cyst, which slightly decreased in size. The rest of the ultrasound revealed an age-appropriate uterus and ovaries (Figure 2C).

To better delineate the anatomy, a discussion was had to undergo further diagnostic testing with either an MRI or exam under anesthesia. The mother opted for an exam under anesthesia, which still noted the presence of a periurethral mass. On cystoscopy there was no diverticulum or os noted, and there was no fluid released into the urethra upon manipulation. A single cervix was noted on vaginoscopy. Paraurethral cyst remained at the top of the differential; given their

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Congenital Female Urethral Diverticulum (continued from previous page)

natural history of likely regression, the decision was made to observe. The child was evaluated at 2 years of age with no resolution of the mass. After another goals of care conversation, the mother opted for marsupialization of presumed paraurethral cyst due to her preference to avoid continued surveillance. The patient was taken to the operating room. Upon examination of the removed specimen, a small opening was noted at its base. A catheter was passed into the urethra and was visualized on the vaginal side. The urethra was subsequently everted and the opening of what we now know to be a urethral diverticulum was seen. Uneventful urethral diverticulectomy and closure was completed. The catheter was left for 12 days post-operatively and the patient voided without issue. At 2-month follow-up the patient was still voiding well. The incision line was well-healed with no evidence of fistula. There is a plan for follow up at the time of toilet training.

Female urethral diverticula are rare and usually presents in the 3rd to 5th decade with nonspecific lower urinary tract symptoms.[1] The most accepted theory for urethral diverticulum formation involves periurethral glands. Occlusion of these glands causes their secretions to be retained. This can lead to infection within the obstructed gland, ultimately resulting in an abscess which may subsequently rupture into the urethral lumen forming the diverticulum.[2] There is also case report evidence for congenital origin; these theories include remnants of an ectopic ureterocele or the result of an abortive attempt at formation of a second urethra.[3] In conclusion, the presented case illustrates the value of a broad differential and limitations in diagnostic tests and imaging.

References
Case Presentation

A newborn male was born at 36 weeks with massive abdominal distension, normal abdominal musculature and genitalia. Severe bilateral hydroureteronephrosis was found prenatally. A Foley catheter was placed with little urine return. Ultrasound demonstrated multiple large cystic collections in both upper and right lower quadrants with right mild and left moderate hydronephrosis (Figure 1).

The bladder was displaced dramatically to the left without VUR and a normal urethra on VCUG. Despite appropriate bladder drainage, the child’s creatinine continued to rise. Repeat ultrasound after several days of bladder decompression showed no change in the large cystic structures. MRI of the abdomen and pelvis demonstrated large separate bilateral cystic fluid collections thought to be urinomas (Figure 2).

He was taken to the operating room. His bladder was significantly distorted due to a large, right posterolateral mass, which prevented retrograde urography. Percutaneous 8.5 Fr drains were placed in the bilateral cystic collections. Postoperatively, the right drain consistently produced urine, while the left drain output stopped several days after placement. Further abdominal imaging with antegrade contrast injection of the drains did not clearly delineate that the drains were in the urinary tract. However, there was a significant reduction in both cystic masses following percutaneous drainage. Nuclear renography demonstrated a differential function on the right of 82% with the left at 18% and little excretion of the radiotracer from either kidney. Because of concerns for right renal obstruction and presumed urine leak from the right collecting system, a right percutaneous nephrostomy was placed. Antegrade contrast injection through the right (continued on next page)
nephrostomy tube did not communicate with the right percutaneous drain. Due to the continuous urine output from the right abdominal drain, the child was taken back to the operating room. Retrograde pyelography was again unsuccessful. Through a right inguinal incision, a right distal end ureterostomy was performed for his right UVJ obstruction. A left inguinal exploration was made with plans to create a left cutaneous ureterostomy but the left ureter was not found. A left flank incision was made to expose his left kidney and proximal ureter. This revealed a massively dilated and decompressed left ureter. Antegrade contrast injection through a ureterotomy demonstrated a tortuous and massively dilated left ureter which crossed the midline and filled the right lateral paracolic space. The right percutaneous drain was found in the mid left ureter. Because of the poor renal function, a nephroureterectomy was performed. The child did well following surgery. The right ureterostomy was taken down with reimplantation at 9 months of age. He is clinically stable with a serum creatinine of 0.57 mg/dL at 2 years of age.

Discussion

Congenital giant megaureter (CGM) is a rare anomaly and has been defined as dilation 10 times greater than normal ureteral diameter in the presence of normal bladder function. It is distinct from the more commonly encountered congenital megaureter (diameter $\geq 7$ mm) based on ureteral dilation. However, CGM and megaureters likely share a common etiology, distal obstruction from either an adynamic segment or focal obstruction.

CGM was first described by Chatterjee in 1964 in a 6-year-old boy with progressive abdominal distension due to a massive left ureter occupying the left hemiabdomen. Huang reported a 21-patient CGM case series with an age range of 2 months to 8 years. All patients required nephroureterectomy. Prenatal manifestation of CGM, such as in our case, is rare. Massive ureteral dilation, distortion of intra-abdominal contents, and unusual anatomic findings seen with CGM can make an accurate radiologic diagnosis elusive. In spite of multiple and varied preoperative abdominal imaging studies, we made the diagnosis of CGM only at the time of surgical exploration.

The surgical treatment of CGM largely depends on the renal function of the affected kidney and the extent of CGM dilation. In patients with extensive ureteral dilation or poor renal function such as in our case, a nephroureterectomy is the preferred surgical approach.

Conclusion

We report a rare case of neonatal CGM, which was a diagnostic dilemma despite prenatal and extensive postnatal imaging and evaluation. CGM should be considered in newborns with large cystic abdominal masses, particularly those with confusing clinical and radiologic findings.

References

The Mystery of the Fluctuating Scrotal Cyst: A Rare Case of Congenital Posterior Urethroperineal Fistula

Sikai Song, Ala’a Farkouh, Joshua Pearce, David A. Chamberlin, Minh-Hang Chau, Catherine J. Chen, Joshua D. Chamberlin
Department of Urology, Loma Linda University Children’s Hospital

Introduction
Congenital posterior urethroperineal fistula is a rare congenital anomaly with only a few reported cases in the literature. We present a unique case of a congenital posterior urethroperineal fistula with an associated scrotal cyst successfully treated with excision.

Case Report
A 21-month-old male presented with a fluctuating, intermittently draining scrotal fluid collection noted at 4 months of age. Physical exam showed Tanner stage I male genitalia, bilateral descended testes, and a 2 cm midline scrotal fluid collection that was non-tender, non-erythematous (Figure 1A). Clear fluid was expressed through a pinpoint opening at the base of the midline scrotum. Scrotal ultrasound confirmed a 2.2 cm midline scrotal cyst independent of the testes (Figure 1B). VCUG demonstrated a fistulous tract originating from the prostatic urethra draining into the midline scrotal cyst (Figure 1C-D).

The patient subsequently underwent cystoscopy and excision of the urethroperineal fistula. Cystoscopy confirmed a patent urethra without stricture and normal-appearing bladder with orthotopic bilateral ureteral orifices. A 0.018" hydrophilic wire passed externally retrograde into the fistulous tract confirmed the proximal origin at the right lateral aspect of the verumontanum. The scrotal cyst and fistulous tract were excised below the corpus spongiosum and external urethral sphincter (Figure 2). The fistula was suture ligated with 4-0 absorbable monofilament suture. Pathology revealed squamous cell epithelium, consistent with a fistula. Post-operatively, the patient recovered well with complete resolution of the scrotal cyst and drainage at 6 months.

Discussion
Congenital posterior urethroperineal fistula (CUPF) is rare with only 25 reported cases in the literature.1 The embryologic origin is unclear, but the age of presentation ranges from infancy into adulthood. The most common presentation is leakage of urine from the perineum during voiding.2 The diagnosis of CUPF is confirmed with a voiding cystourethrogram/retrograde urethrogram and cystourethroscopy, demonstrating a dominant dorsal urethra with a hypoplastic ventral fistulous tract.3,4 All reported cases of CUPF have been successfully treated with partial or complete surgical excision of the ventral fistula without surgery of the dorsal urethra. To our knowledge, this is the first reported case of CUPF with an associated scrotal cyst.

Urethral duplication anomalies have similar clinical presentations to CUPF, but surgical correction requires a different approach. The key to diagnosis between urethral duplication and CUPF requires the identification of the functional urethra, the underdeveloped urethral channel, and their respective orientation. In 1976, Effmann et al. proposed the commonly utilized urethral duplication classification.5 Per this classification system, the Effmann Type II A2, Y-duplication subtype most closely resembles CUPF. However, the Effmann Type II A2, Y-duplication subtype consists of a hypospadic urethral duplication where the principal functional urethra courses ventrally, terminating in the perineum or anus, and the accessory urethral channel terminates dorsally along the course of the phallus. In CUPF, the principal functional urethra courses dorsally along the course of the phallus to the normal urethral meatus, while the accessory urethral fistula terminates ventrally in the perineum. For the hypospadic urethral duplica-

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tion type, reconstructive surgery is required to mobilize the functional ventral urethra and relocate the meatus. Meanwhile, CUPF is cured with excision of the ventral accessory urethra alone.

This clinical case is most consistent with a urethroperineal fistula, with the addition of a scrotal cyst. Histopathology is also consistent with a urethroperineal fistula. Treatment was successful with proper diagnosis and surgical excision of the cyst and fistulous tract.

Conclusion

To our knowledge, this is the first reported case of a congenital urethroperineal fistula (CUPF) with an associated midline scrotal fluid collection, successfully excised in a male infant. Successful manage-

References


Figure 2. (A) Excision of the scrotal cyst and fistulous tract. (B) Length of tract was 7 cm.