FROM THE EDITOR
Douglas Storm, MD

I have the pleasure and privilege of introducing the 2023 Spring Society of Fetal Urology case presentation edition of Dialogues in Pediatric Urology. The SFU has a long and constructive relationship with DPU. This relationship is seamless, as both are focused on sharing experiences and information in a digestible and conversational format.

Case presentations related to fetal or perinatal urology have been a long-standing tradition with the Society for Fetal Urology. These sessions have been an important moment for many students, residents, and fellows - to have the opportunity to present interesting cases in an amicable environment. These SFU case conferences are invaluable, as the discussions that occur with each case allows the presenters and audience members the opportunity to reap clinical and academic benefits from these conversations. I for one come away from each SFU case conference with a new fund of knowledge that has been invaluable in the fetal and perinatal care I provide to children with urologic abnormalities.

This edition includes outstanding case presentations from the 2023 Spring SFU meeting, which was held in Chicago, Illinois, in conjunction with the SPU-AUA meeting. My sincere thanks to the SFU executive board members who helped moderate and judge the evening case session.

I strongly encourage you to read the cases contained within this edition of DPU and I hope that you find them as interesting and educational as I did. Thank you again to the authors for their intelligent and thoughtful contributions. I truly believe that these cases will be of interest to all pediatric urologists.

Breaking the Rules: A Duplicated Collecting System with an Obstructed Lower Pole Moiety
Nicole Ackerman, Guanqun Li, Anthony J. Tracey, Matthew Mason, Jeffrey Villanueva
Department of Pediatric Urology, SUNY Upstate Medical University, Syracuse, NY

Background
A completely duplicated collecting system with an obstructed lower pole moiety is a rare anomaly with few cases reported in the literature. The Weigert Myer rule traditionally predicts the draining pattern of duplex ureters with the upper pole moiety draining inferomedially in an ectopic location, resulting in obstruction. We discuss a female with a history of a left duplicated collecting system with an obstructed lower pole moiety.

(continued on next page)
Case Presentation

We present a female with a history of a left duplicated collecting system with an obstructed lower pole moiety. Left hydronephrosis and oligohydramnios (AFI: 2.84cm) were initially identified on her prenatal ultrasound at 35 weeks (Figure 1). Therefore, she was delivered at 35 weeks by emergent C-section. Postnatal ultrasound on her first day of life confirmed moderate left lower pole hydroureteronephrosis. Differential diagnosis at that time included vesicoureteral reflux, ectopic ureter, and ureterocele. However, on her third day of life, a voiding cystourethrogram was performed which had no reflux. A diuretic renal scan at 3 months of age revealed left lower pole moiety obstruction with a split function of 52/47% for right/left kidneys. The lower pole moiety contributed 33% of the left sided function (Figure 2). A magnetic resonance urogram also identified left lower pole hydroureteronephrosis with a distended, tortuous ureter measuring up to 19mm. There was no obvious ectopic insertion of the lower pole ureter. Clinically, she remained free of febrile urinary tract infections on continuous antibiotic prophylaxis. However, she had progressively increased lower pole hydroureteronephrosis with an APRPD of 4.18cm from 1.77cm two years earlier. Thus, a left robotic uretero-ureterostomy was performed when the child was 18 months of age. Cystoscopically, we identified the left lower pole ureter inserting laterally and proximally – a break in the Weigert Myer rule. At this point, we performed an end-to-side anastomosis of the left lower pole moiety into the upper pole ureter. She was discharged on post-op day one after her catheter was removed. She was briefly taken back to the operating room four weeks post-operatively to remove the left ureteral stent. Currently at the age of two, she has been doing well, with no breakthrough urinary tract infections. Radiographically, post operative ultrasound revealed markedly improved lower pole hydronephrosis and ureteral dilation.

Conclusions

A duplicated collecting system with an obstructed lower pole moiety is a rare anomaly. Lower pole hydroureter typically occurs due to vesicoureteral reflux. Throughout the literature, three similar case reports of lower pole moiety obstruction have been demonstrated. However, this case represents the only one where the lower pole obstructed moiety remained functional. We therefore recommend that postnatal evaluation including renal scan, VCUG, and MRU can be helpful in identifying the need for surgical intervention in patients with a duplicated collecting system.

Figure 1: Prenatal ultrasound at 35 weeks demonstrating left hydronephrosis and oligohydramnios (AFI: 2.84cm).

References

Case Presentation

A 28-week prenatal ultrasound raised concern for bladder outlet obstruction in a male fetus. Fetal magnetic resonance imaging showed a “keyhole sign” with bladder and posterior urethral dilation, consistent with a diagnosis of posterior urethral valves (PUV). It also demonstrated preserved amniotic fluid, SFU grade 3 right hydronephrosis, left-sided urinoma, and urinary ascites (Figure 1). Lung volumes appeared adequate.

The mother entered spontaneous labor at 37 weeks. However, progress stalled, and the fetus required percutaneous paracentesis to be delivered vaginally. The bladder was drained with a feeding tube. On day of life 1, the patient’s creatinine was 1.6 mg/dL. Ultrasound showed a left-sided urinoma and SFU grade 3 right hydronephrosis (Figure 2, C); the left kidney was not identified. Due to respiratory distress and abdominal distention, a percutaneous drain was placed into the urinoma. After stabilization, a voiding cystourethrogram (VCUG) confirmed PUV. VCUG also showed bilateral vesicoureteral reflux (VUR), grade 5 on the left and reflux into a dilated ureter on the right (Figure 2, A and B).

The urinoma drain output was initially 150-450 mL per day and trended down to about 60 mL per day. Due to the complexity of his condition, the patient underwent vesicostomy at 2 weeks for maximal urinary drainage. The urinoma drain output continued to decline, and it was removed after a clamping trial. His creatinine nadired at 0.2 mg/dL at 6 weeks. Interestingly, after urinoma drain and vesicostomy, renal ultrasound identified the left kidney, which was not seen at all on prior ultrasounds (Figure 2, D).

Ultrasound at 6 months showed decompressed kidneys bilaterally with improved corticomedullary differentiation (Figure 2, E and F). Dimercaptosuccinic acid (DMSA) renal scan showed the left-sided function to be much better than expected given initial imaging, with differential function of 60% left and 40% right. There was some left sided scarring (Figure 3, G). His creatinine remained stable around 0.3 mg/dL.

At 10 months, the patient underwent valve ablation and vesicostomy closure. At that time, there was no discernible lumen at the location of the valves which likely fused after his prolonged catheterization as an infant. Currently, the patient is doing well at 14 months of age. He remains on antibiotic prophylaxis, and the status of his VUR will be reassessed in the future.

Discussion

A variety of potential pop-off mechanisms may present in patients with posterior urethral valves, including urinoma and urinary ascites, as seen in this case; unilateral vesicoureteral reflux dysplasia (VURD); or large bladder diverticula. Studies on whether the presence of these pop-off mechanisms is protective of renal or bladder function have been conflicting. A report in 1988 described the protective effect of pop-off mechanisms on preservation of renal function1.

(continued on next page)
Posterior Urethral Valves (continued from previous page)

Subsequent studies have been conflicting, with some studies supporting the assertion that pop off mechanisms protect kidney function\(^2\), while others have not detected differences in renal or bladder function\(^3\).

While many studies have analyzed all pop-off mechanisms together or focused on VURD, Lundar and colleagues looked specifically at urinoma and urinary ascites by comparing 12 patients with urinary extravasation to 48 patients without\(^2\). They found a statistically significant lower prevalence of chronic kidney disease stages 2-5 in those with urinary extravasation (8%, 1/12 patients with urinary extravasation vs 42%, 20/48 patients without urinary extravasation; \(p=0.03\)), with a median follow up of 5 years.

The case reported here demonstrates the use of a urinoma drain and vesicostomy for maximal decompression, with preservation of the left kidney which initially appeared significantly compromised. It provides reminders that given the heterogeneity in presentation, outcomes, and evidence, treatment for patients with PUV and urinary extravasation must be individualized and that renal outcomes are challenging to predict.

References


Figure 2. Postnatal imaging. A and B: VCUH with VUR and PUV. C: Right renal ultrasound at 72 hours of life. The left kidney could not be identified. D: Renal ultrasound 3 days after vesicostomy creation, with left kidney identified. E: 6-month renal ultrasound, right. F: 6-month renal ultrasound, left. G: DMSA renal scan at 9 months.
Horseshoe Kidney with Retrocaval Ureter and Ureteropelvic Junction Obstruction

Ala’a Farkouh, Matthew Buell, Kai Wen Cheng, Ruby Kuang, Tida Thaipejr, David A. Chamberlin, Joshua D. Chamberlin*
Department of Urology, Loma Linda University Children’s Hospital

Case Presentation

The patient is a male infant diagnosed with bilateral prenatal hydronephrosis, born full-term via Caesarean section. Postnatal ultrasound (US) confirmed bilateral SFU grade 4 hydronephrosis and voiding cystourethrogram showed no reflux or posterior urethral valves. Continuous antibiotic prophylaxis was initiated. At one-month of age, he developed a breakthrough febrile UTI, which was treated with culture-directed oral antibiotics.

At 10 weeks, a repeat US revealed worsened bilateral hydronephrosis and the right kidney lying parallel to the lower pole of the left kidney, suspicious for horseshoe kidney (Figure 1). At 4 months of age, a diuretic renogram revealed a split renal function of 10% on the right with obstruction, with normal serum creatinine. Physical exam revealed a distended abdomen with a palpable right kidney.

Cystoscopy showed orthotopic bilateral ureteral orifices without stenosis or duplication. Right retrograde pyelogram (RPG) demonstrated a medially deviated right distal ureter with complete obstruction at the level of the midureter, with inability to pass a guidewire. Left RPG showed a normal caliber laterally deviated left ureter to a moderately hydronephrotic malrotated kidney. MR urogram revealed a dysmorphic, transversely oriented right kidney in horseshoe configuration extending past the midline with marked calyceal distension and parenchymal thinning. The dilated right collecting system caused mass effect on the adjacent proximal left ureter, leading to moderate left-sided hydronephrosis.

Based on these findings, a right nephrostomy tube was placed with progressive urine output and a follow-up US demonstrated complete decompression of the right kidney. Due to subsequent tube displacement, recurrent infections, and right split function of 10%, a laparoscopic right nephrectomy was planned at 8 months of age.

Astonishingly, intraoperative findings identified a right retrocaval ureter with the majority of the right renal pelvis also behind the vena cava (Figure 2). Given this finding, and the continued urine output from the right kidney after nephrostomy placement, a dismembered pyeloplasty with repair of the retrocaval ureter was performed instead of a nephrectomy. The nephrostomy tube was removed, and a ureteral stent was inserted with subsequent uncomplicated removal 5 weeks postoperatively.

Four months after surgery, a repeat US revealed normal right moiety of the horseshoe kidney without pelvicalyceal dilation and improvement of left-sided hydronephrosis. Patient had resolution of previous abdominal distension with no further urinary tract infections.

Discussion

Horseshoe kidney and retrocaval ureter are two distinct congenital anomalies. Horseshoe kidney is the most common kidney fusion defect, while retrocaval ureter is a vascular anomaly that results from failure of obliteration of the subcardinal vein in the fetal circulation.

Horseshoe kidney and retrocaval ureter are two distinct congenital anomalies. Horseshoe kidney is the most common kidney fusion defect, while retrocaval ureter is a vascular anomaly that results from failure of obliteration of the subcardinal vein in the fetal circulation.

Figure 1: Ultrasound at 10-weeks of age demonstrating bilateral hydronephrosis and suggestive of horseshoe kidney. (A) Right kidney sagittal view. (B) Left kidney compressed by the hydronephrotic right kidney.

(continued on next page)
been reported at an average of 1-2 cases per decade. To our knowledge, we present the first case of horseshoe kidney with retrocaval ureter and symptomatic presentation necessitating surgical intervention in infancy, as well as the first reported case successfully managed by minimally invasive surgery.

Most cases reported this condition in young adults or middle-aged patients, presenting with symptoms of flank pain, urosepsis, urolithiasis, or hydronephrosis. In most reported cases, preoperative detection of both anomalies together was challenging and was confirmed intraoperatively. In the latest report by Shen et al., the ureter was only visualized when CT scan was performed immediately after injection of retrograde contrast, but not with CT urography².

In our case, RPG and MRI delineated the underlying abnormal anatomy, which resulted in symptomatic bilateral hydronephrosis. The presence of a retrocaval ureter was only seen intraoperatively, leading to a change in the planned surgery from nephrectomy to pyeloplasty and transposition of the retrocaval ureter. Given the advances in minimally invasive surgery, conversion to open surgery was not necessary. This is consistent with the reported successes of laparoscopic pyeloplasty in children with horseshoe kidneys and laparoscopic ureteropyeloplasty in adults with retrocaval ureter³⁴.

Although rare, pediatric urologists should keep in mind the possibility of concurrent anomalies. Especially in the presence of horseshoe kidneys, as they are more likely to have associated IVC anomalies, including retrocaval ureter, left IVC, or double IVCs⁵.

**Conclusion**

This is the first reported case of retrocaval ureter with horseshoe kidney and associated ureteropelvic junction obstruction that was repaired laparoscopically in an infant.

**References**


---

**Figure 2**: Intraoperative picture of horseshoe kidney and retrocaval ureter with illustration: A) Right renal pelvis, B) Inferior vena cava, C) Dilated right renal pelvis.
Obstructed Bilateral Upper Pole Ectopic Ureters:
To Divert or Not to Divert?

Kristen Meier, MD,1,2 Margret Bock, MD,3 Mariana Meyers, MD,4 Vijaya Vemulakonda MD, JD1,2
1 Department of Surgery, Division of Urology, University of Colorado School of Medicine, Aurora, CO, USA.
2 Pediatric Urology Research Enterprise, Department of Pediatric Urology, Children’s Hospital Colorado, Aurora, Colorado, USA.
3 Department of Pediatrics, Division of Pediatric Nephrology, Children’s Hospital Colorado, Aurora, USA.
4 Department of Radiology, Children’s Hospital Colorado, University of Colorado School of Medicine, Aurora, CO, USA.

Introduction
Upper pole hydronephrosis secondary to bilateral ureteral ectopia is a rare cause of lower urinary tract obstruction in the neonatal population. Deciding whether and when to perform urinary diversion is needed due to concerns about the effects of obstruction on long-term renal function. Here we describe such a case successfully managed with early diversion.

Case
A 39w0d female was born via c-section at a tertiary care facility. The mother was followed throughout the pregnancy by Maternal Fetal Medicine (MFM) for her advanced maternal age. Prenatal ultrasound at 18 weeks showed bilateral SFU grade 2 hydronephrosis and echogenic structures in bladder concerning for ureteroceles. The fetus was noted to have normal amniotic fluid levels. Fetal MRI demonstrated bilateral duplex collecting systems with SFU grade 4 upper pole hydronephrosis and suspected bilateral ureteroceles (Figure 1). The fetus was monitored closely throughout the remainder of the pregnancy and was delivered at 39 weeks. The infant voided spontaneously after birth and was started on amoxicillin prophylaxis. A renal ultrasound and voiding cystourethrogram (VCUG) obtained on DOL#2 demonstrated bilateral duplex systems with SFU grade 4 hydronephrosis of the upper poles with filling defects concerning for bilateral ureteroceles and no reflux. She was unable to void after VCUG and creatinine was noted to be rising despite foley drainage. She subsequently underwent cystoscopy for planned transurethral incision of the ureteroceles on DOL#3. Bilateral ectopic ureters were noted at the bladder neck without ureteroceles. MR Urogram was obtained which confirmed these findings (Figure 2). She was discharged home with a catheter in place and close follow up in the Congenital Anomalies of the Kidney and Urinary Tract (CAKUT) clinic. With the foley, creatinine nadired at 0.50 and she was noted to have persistent significant hydronephrosis on follow up. Based on this, she was offered bilateral end cutaneous ureterostomies of her upper pole segments which were completed at 3 weeks of age. By POD#1 her Cr had improved to 0.37 and

(continued on next page)
she was discharged home without a catheter. On follow up, creatinine nadired at 0.23 with improvement of the upper and lower pole renal dilation. Ureterostomy takedown with bilateral upper-to-lower-pole ureteroureterostomies was performed at 18 months of age. She remains stable after reconstruction.

**Discussion**

This case highlights the difficulty of diagnosis and decision-making in infants with ureteral anomalies. In challenging cases such as this, ultrasound and VCUG alone may not provide definitive diagnosis; additional imaging with MR urography can be very helpful in delineating difficult anatomy.1

The decision of whether and when to perform urinary diversion is also a difficult consideration in this population. On one hand, performing early diversion means multiple procedures and anesthetics for very young patients. On the other hand, longstanding obstruction in immature kidneys can have significant effects on long-term renal function.

Adding to the complexity is how difficult it is to accurately measure renal function in the neonatal and infant period, knowing that prematurity, maternal renal function in the first few days of life, and lack of reference ranges for newer tests like cystatin C all make interpreting renal function tests in newborns and infants a challenge.2 Nevertheless, post-natal renal maturation is most pronounced within the first 18 months of life; because of this, optimizing low-pressure drainage during this period is key and studies generally recommend a low threshold to intervene along with early involvement of nephrology for these discussions.

Percutaneous nephrostomies, while a reasonable temporary option, are less successful as a long-term solution, requiring frequent exchanges in the infant period. More commonly loop or end cutaneous ureterostomies are chosen based on provider preference, whether there is a single system versus duplex system and ultimate plan to perform reimplant versus ureteroureterostomy. In this case, we chose to perform upper pole end ureterostomies with planned upper-to-lower pole ureteroureterostomy given the absence of lower pole reflux and the potential benefit of maintaining bladder function due to urine cycling from the lower pole segments.

**Conclusions**

Here we present a case of bilateral duplex systems with ectopic ureters successfully managed with cutaneous ureterostomies with subsequent conversion to ureteroureterostomies. This case demonstrates the importance of maximizing drainage during the early neonatal period to optimize renal function.

**References**


Uroepsis and bladder outlet obstruction (BOO) with bilateral hydronephrosis secondary to large ureterocele is a rare anomaly with few reported cases in literature.

Case Presentation

We present a female with a right ureterocele resulting in bladder outlet obstruction. She was identified to have severe right hydroureteronephrosis with ureterocele measuring 1.1x0.7 cm on prenatal ultrasound at 22 weeks. Urology was not involved in her prenatal care. Patient was born at 37 weeks and post-natal ultrasound revealed a duplicated right system with severe hydronephrosis of both moieties, and a large right ureterocele measuring 2.9x2.7x2.1; no left hydronephrosis was noted (Figure 1). She also had a VCUG performed, which showed a filling defect corresponding to the ureterocele, no vesicoureteral reflux, and complete bladder emptying. Patient was discharged home on day two of life with prophylactic Keflex and plans for outpatient follow up in one month. However, patient presented to the emergency department three weeks later due to fevers and urinary retention. Labs revealed leukopenia and severe thrombocytopenia. Ultrasound showed enlargement of ureterocele to 3.5x3.0x2.5 cm with worsening right hydronephrosis and new left hydronephrosis (Figure 2). Foley was placed at time of presentation and patient was initiated on IV meropenem for Enterobacter bacteremia. She was taken to the operating room for ureterocele puncture after one week of antibiotics and platelet normalization. Intra-operatively, a large right sided ureterocele was immediately identified and puncture was performed with Bugbee electrocautery. Urinary drainage was noted with puncture and on re-inspection, the ureterocele appeared decompressed. The following day, the patient had a successful trial of void and was discharged home after completing her antibiotic course based on Infectious Disease recommendations. Four weeks post operatively, patient was seen in clinic and ultrasound showed improvement in right hydronephrosis and no left hydronephrosis or visualization of ureterocele within the bladder.

Conclusions

BOO with sepsis is a rare sequela of an enlarging ureterocele, which may present sometime after birth. This can even be the case when immediate post-natal work-up does not demonstrate impaired bladder emptying. We describe how ureterocele puncture may be an effective method in decompressing the ureterocele, a delicate procedure in a neonate.

References

True and False Ureters and a Vanishing Ureterocele Among VACTERL Anomalies Identified Prenatally

George W. Moran, Jane T. Kurtzman, Soo Jeong Kim, Belinda Li, Christina P. Carpenter
Department of Urology, New York-Presbyterian Morgan Stanley Children’s Hospital, New York, NY

A singleton male fetus was found on prenatal ultrasound at 26 weeks’ gestation to have anomalies suggestive of VACTERL (vertebral defects, anal atresia, cardiac defects, tracheo-esophageal fistula, renal anomalies, and limb abnormalities) association, including non-visualized anorectal structures, a horseshoe kidney, right duplicated collecting system, bilateral hydronephrosis, right hydroureter, and a ureterocele.

The baby was born at 36 weeks by caesarian section scheduled for vasa previa. His exam was notable for imperforate anus without cutaneous fistula. Following colostomy creation DOL0 (Day of Life) and tracheo-esophageal fistula ligation (DOL1), a DOL3 ultrasound confirmed a horseshoe kidney with a severely hydronephrotic right moiety with ureteral dilatation, and mild left hydronephrosis (Figure 1). Antibiotic prophylaxis was initiated at this time. A Foley catheter remained in place following a traumatic catheterization preceding his first surgery. VCUG on DOL6 demonstrated grade V VUR into a very dilated lower pole moiety of a duplicated right system and what appeared to be low grade VUR into an ectopic left ureter with low insertion, presumably explaining the mild left sided hydronephrosis (Figure 2a). Despite the prenatal finding of a ureterocele, none was visualized on immediate postnatal imaging.

Following Foley removal, the patient voided without issue. He soon started clean intermittent catheterization, however, due to his dilated upper tracts and new concern for tethered cord on spinal MRI. In the ensuing months, he developed UTIs despite antibiotic prophylaxis, and ultrasounds demonstrated worsening right hydronephrosis. Due to concern for obstruction of the right upper pole renal moiety, he underwent a MAG-3 renal scan at 3.5 months. This demonstrated 40% right/60% left split function and high-grade obstruction on the right. Repeat ultrasound now showed a ureterocele at the right trigone, so he underwent transurethral incision of what turned out to be a cecoureterocele intermittently obstructing the outlet of his severely trabeculated bladder. Post-operatively, the right-sided hydronephrosis improved.

At thirteen months, he underwent cystoscopy, cystogram, and distal colostogram. Contrast injected into the mucous fistula promptly filled the urinary tract through a fistula entering at the prostatic ure-
VACTERL Anomalies (continued from previous page)

...thra (Figure 2b). This clarified that what was formerly thought to be a refluxing left ureter was in fact a rectourethral fistula. Cystoscopy and retrograde pyelograms clarified that he had two very dilated right ureters emptying at the ureterocele defect, and a PIC cystogram confirmed a non-refluxing left ureteral orifice. At fourteen months, he underwent successful posterior sagittal anorectoplasty and ligation of his rectourethral fistula.

The patient remained healthy though the following year but began having breakthrough UTIs at age two and was found to have worsening hydronephrosis of both right renal moieties. Video urodynamics confirmed VUR up both right ureters, additionally demonstrating detrusor overactivity and detrusor external sphincter dyssynergia. While compliance could not be assessed given the early grade V VUR, bladder capacity was estimated to be 50mL. Predicting that a bladder this small might prohibit common sheath ureteral reimplantation, we performed a right upper-to-lower pole ureteroureterostomy, reimplantation of the lower pole ureter, and excision of right ureterocele tissue. A retrograde pyelogram six weeks after surgery showed good drainage from both right systems through the lower pole ureter, and an ultrasound two months post-op showed significant improvement in the dilatation of the right kidney, with stable mild left hydronephrosis. Antibiotic prophylaxis was discontinued and he has remained infection-free.

As this case demonstrates, rectourethral fistulas can be mistaken as ectopic ureters, particularly in patients in whom one expects to find urinary tract anomalies. It is important to have a high index of suspicion for rectourethral fistulas in boys with imperforate anus. This is the most common anorectal malformation in males, while imperforate anus without fistula is rare, only about 5% in a large retrospective series.1 Performing a high pressure distal colostogram and placing a Foley catheter preoperatively are critical to identifying a fistula and avoiding injury to the urinary tract at the time of anorectoplasty.2

Furthermore, ureteroceles should be suspected in patients with renal duplication anomalies, even if they are not detected on preliminary imaging. On ultrasound, a ureterocele might collapse in an over-distended bladder. In the under-distended bladder, it may fill the entire bladder, giving the impression of a full bladder with no ureterocele. Similarly, on VCUG, dense contrast filling the bladder and urethra may obscure a ureterocele.3 Recognition of ureteroceles is crucial, as they are often treatable causes of upper tract obstruction.

References
Spontaneous Resolution of Congenital Megalourethra

Alexandra R. Siegal, MD1, Neha R. Malhotra, MD2, Jeffrey A. Stock, 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 the case of a male with congenital megalourethra, found on the antenatal sonogram of a 33-year-old African American female with a past medical history of asthma, endometriosis, GERD, interstitial cystitis, anxiety, and depression.

At the mother’s 20-week sonogram, a 2x2x6 mm cystic dilation was noted at the distal end of the penis (eccentric to the left and ventral) (Figure 1). Multiple instances of bladder cycling were noted without upper tract dilation seen. Amniotic fluid volume, the remaining fetal anatomy, and genetic testing appeared normal.

Differential diagnosis comprised of congenital megalourethra, either the scaphoid or fusiform type. The more common and less severe scaphoid type is characterized by the absence of corpus spongiosum, whereas the more rare and severe fusiform type is marked by the maldevelopment of both the corpus spongiosum and cavernosa. The fusiform type is often associated with long-term sexual and voiding dysfunction. It has been described that as the urethra “balloons” with voiding, an obstructive process may occur causing retrograde bladder and kidney anomalies. Additional differential diagnoses include urethral anomalies such as anterior urethral valves, urethral atresia, webs, or a diverticulum, or other findings such as a mass in the abdominal wall or an umbilical cord loop.1 Surgical treatment of congenital megalourethra ranges from one- or two-stage reduction urethroplasty for the scaphoid type to major phallic reconstruction for the fusiform type.2

A multicenter cohort and systematic review of the literature reported that 24% of cases of congenital megalourethra end in medical termination.3 In this case presentation, the mother became severely depressed after learning about the fetal diagnosis of congenital megalourethra; she was admitted for suicidal ideation and also considered pregnancy termination. Based on state law, the mother had 4 weeks from the time of the fetal diagnosis to decide whether she was to continue her pregnancy.

Two weeks later, a reevaluation of the fetal genitalia was conducted to identify the corpora to rule out the more severe fusiform type; at this time the cystic anomaly appeared to be resolving (Figure 2). Given the normal remaining urinary tract, the mother elected to continue her pregnancy. Fortunately, at 26-weeks gestation, there was involution of the cystic anomaly; the resolution was confirmed on her subsequent sonograms at 31-weeks and onward.

The boy was ultimately born via cesarian section at 37w4d with normal external genitalia. To ensure the megalourethra had completely resolved, a RBUS and VCUG were obtained at 22 days; these studies revealed no hydronephrosis, no reflux, and a normal urethra.

Congenital megalourethra is a very rare anomaly, diagnosed by the presence of a blind cystic structure between the fetal legs or arising from the perineum. To date, there have only been case series and one systematic review describing this diagnosis and its outcomes.

(continued on next page)
anomaly, diagnosed by the presence of a blind cystic structure between the fetal legs or arising from the perineum. To date, there have only been case series and one systematic review describing this diagnosis and its outcomes. It is important to note that only half of congenital megalourethra cases are associated with bilateral hydronephrosis (58%) and megacystis (52.0%). When these features are absent, as seen in this fetus, there is a decreased chance of renal, bladder, and sexual impairment. Furthermore, there is a 10% spontaneous resolution rate of congenital megalourethra; however, only five of these instances have been reported. This case illustrates the value of repeat sonograms and the necessity of appropriate patient counseling regarding prognosis to produce the best outcomes.

References

Figure 2: Megalourethra noted on 22-week prenatal sonogram
Prenatally Diagnosed Echogenic Cystic Kidneys and Right Uretero-pelvic Junction Obstruction in a Patient with a Family History of HNF1B Mutation

Introduction

Hepatocyte nuclear factor 1 beta (HNF1B) is a transcription factor that is essential for the development of kidneys. HNF1B mutations are characterized by autosomal dominant inheritance with variable expressivity leading to a spectrum of clinicopathologic manifestations. Mutations in HNF1B can lead to congenital anomalies of the kidney and urinary tract including hyperechogenic kidneys, multicystic kidney disease, and urinary tract dilation. Here we present the case of prenatally diagnosed bilateral enlarged echogenic kidneys and significant right hydronephrosis in a female fetus with a family history of HNF1B mutation.

Case Presentation

Initial prenatal evaluation of a 36-year-old G3P2 female at 26 weeks of gestation revealed a female fetus with bilateral enlarged echogenic kidneys with cysts and severe right hydronephrosis with anterior pelvic diameter (APD) which had increased from 4 mm to 21 mm at 18 and 26 weeks of gestation, respectively. No ureteral dilation or bladder abnormalities were noted. Amniotic fluid index was within normal range. The mother revealed that she and her husband have no history of renal disease, but she reported that her living older son was diagnosed with mild chronic kidney disease and tested positive for HNF1B mutation. Mother’s own genetic testing was negative. Last fetal ultrasound at 33 weeks of gestation showed worsening right hydronephrosis with APD of 38 mm (see Figure 1).

A healthy baby girl was delivered at 37 weeks and 3 days via Caesarean section. Her creatinine nadired at 0.48 mg/dL (elevated). Postnatal evaluation of the infant with ultrasound and voiding cystourethrogram (VCUG) revealed persistent severe right hydronephrosis suggestive of uretero-pelvic junction (UPJ) obstruction without vesicoureteral reflux. Given the fetal presentation and family history, HNF1B mutation and highlights the importance of detailed prenatal and postnatal evaluation to provide a timely intervention and necessary longitudinal follow up with nephrology is recommended.

Discussion

HNF1B is a transcription factor that plays a crucial role in early development of the kidneys. The first pathogenic variant of HNF1B was reported in 1997 and was shown to be inherited in an autosomal dominant pattern (1), although de novo mutations can occur. Since then, more than 230 different variants have been observed revealing a multifaceted syndrome with heterogenous genetic, pathologic, and clinical profiles. Mutations in HNF1B account for up to 10% of congenital abnormalities of the kidney and urinary tract and can lead to ESRD in 13-15% of patients (3). Prenatal phenotypes observed in patients harboring a mutation in HNF1B include bilateral hyperechogenic kidneys with normal or moderately increased kidney size, unilateral or bilateral multicystic kidney disease, unilateral or bilateral renal agenesis, and urinary tract dilation (4,5). Despite autosomal dominant inheritance there is a wide clinical variability among patients harboring the mutation making the prognosis of renal function decline extremely challenging.

Our patient’s presentation with right UPJ obstruction in addition to enlarged, echogenic kidneys with cysts deviates slightly from an expected presentation and confirms previously reported variability in disease phenotype (4). In this case, additional evaluation with a functional renal scan allowed for diagnosis of an obstruction which can be surgically corrected before irreversible changes in renal function occur. It is important to remember, however that renal dysplasia associated with HNF1B mutation is a chronic condition for which a life-long follow up with nephrology is recommended.

Conclusion

This case illustrates both medical renal and obstructive manifestations of prenatal presentation of an infant in the setting of HNF1B mutation and highlights the importance of detailed prenatal and postnatal evaluation to provide a timely intervention and necessary longitudinal follow up to avoid future renal complications.

References


Figure 1. Fetal sonography at 33 weeks of gestation showing severe right hydronephrosis with APD 38 mm.