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Society for Fetal Urology
Spring and Fall 2022 Case Reports

FROM THE GUEST EDITOR

Elizabeth Malm-Buasti, MD

The Society of Fetal Urology continues to enjoy celebrating each other’s work with our in-person meetings, Spring and Fall 2022. We would like to thank everyone for making it to the meeting and for participating in the case presentations and main session. The engagement from the audience was top notch at both sessions. The case presenters did a stellar job with their intriguing case conundrums.

In this edition, you will find a synopsis from the presenters who were kind enough to submit their case reports from the Fall and Spring 2022 meetings. I would like to give them my personal gratitude for preparing their cases for submission to this edition of Dialogues in Pediatric Urology and their willingness to enrich us with their expertise and unique experiences. Dialogues in Pediatric Urology as an educational part of SFU will continue to thrive with such high engagement.

FROM THE EDITOR

Douglas Storm, MD

I would like to thank Elizabeth Malm-Buatsi for collecting case reports from the Spring and Fall 2022 Society of Fetal Urology meetings for inclusion in this edition of DPU. All these authors are to be congratulated on their interesting cases and I thank them for their insightful presentations that we all can learn from.

Urethral Atresia and Its Upstream Effects

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INTRODUCTION

Urethral atresia is a rare entity, with an estimated incidence of one in 100,000 boys.1,2 The pathophysiologic effects are similar to all other types of bladder outlet obstruction, such as the more common diagnosis, posterior urethral valves. Oligohydramnios due to low urine output can lead to pulmonary hypoplasia and renal dysplasia can occur as a primary nephropathic insult. Reflux nephropathy and primary bladder dysfunction can also lead to significant renal damage.

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Urethral Atresia and Its Upstream Effects  
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CASE

Our patient is a male with a prenatal diagnosis of partial bladder outlet obstruction, noted on his 20-week anatomy scan. He had normal amniotic fluid volumes and lung volumes, so no prenatal intervention was recommended. Fetal MRI demonstrated severe hydronephrosis with a distended bladder and posterior urethra (Figure 1).

He was born at term without pulmonary complications and with a working diagnosis of partially obstructing posterior urethral valves. He was noted to void only small amounts by day of life one, with creatinine of 1.58 and an elevated potassium. A renal bladder US was obtained showing severe bilateral hydronephrosis and a distended bladder.

Placement of a catheter was attempted, however after multiple attempts, a catheter was unable to pass into the urinary bladder. He was taken to the operating room for cystoscopy and a narrowing in the proximal bulbar urethra was noted. A wire passed, but dilation was not feasible due to the degree of atresia. A vesicostomy was performed and he subsequently did well, with appropriate urine output. Postoperatively, his workup continued with a VCUG, showing left grade V reflux, and confirmed anterior urethral atresia (Figure 2). An ultrasound on postoperative day three showed slightly decreasing hydronephrosis.

Two weeks postoperatively, another ultrasound was obtained, due to rising creatinine. This demonstrated worsening of bilateral hydronephrosis with left pyonephrosis (Figure 3). Bilateral nephrostomy tubes were placed, and his creatinine decreased appropriately. A MAG3 renal scan demonstrated 10% function in the left kidney and the right kidney showed no evidence of ureteral obstruction. Exam demonstrated prolapse of the vesicostomy with concern for poor drainage leading to the elevated creatinine.

To address these concerns, the vesicostomy was closed and a left loop ureterostomy was created, allowing for bladder cycling and emptying via the left refluxing ureter. The patient was readmitted multiple times with urosepsis thought to be due to inadequate drainage from the ureterostomy. This was managed with an indwelling catheter via the ureterostomy. At one year of age, the patient underwent conversion of the ureterostomy to a ureteral catheterizable channel and simple left nephrectomy. Urethroplasty was planned but not completed due to antegrade and retrograde urethrogram demonstrating a 3cm pan-bulbar stricture.

Currently the patient is doing well with clean intermittent catheterization through his ureteral Mitrofanoff channel with a creatinine of 0.42. The plan is to reassess his bladder and if suitable, reconsider a urethroplasty around puberty.

In summary, our patient is a now 22-month-old with a known 3cm segment of urethral atresia. He had associated high-grade left vesicoureteral reflux with associated reflux nephropathy. He had been managed with a vesicostomy and loop ureterostomy, both of which were inadequate for drainage. He is now doing well, catheterizing through a ureteral Mitrofanoff with the plan for urethral reconstruction if he demonstrates safe bladder compliance as he ages.

DISCUSSION:

Key points from this case are that urethral atresia can be a devastating cause of fetal lower urinary tract obstruction, even if partially obstructing. Utilizing multiple imaging modalities is key to diagnosing atresia. High grade reflux is common and both reflux nephropathy and renal dysplasia are causes of renal deterioration. Finally, patients and their families can expect multiple reconstructive surgeries to optimize outcomes and temporizing supravesical drainage may be required to achieve this.

References

INTRODUCTION
Renal anomalies are reported in up to 80% of children with VACTERL association; however, ectopic and blind ending ureters with concomitant ureteropelvic junction (UPJ) obstruction are exceedingly rare.1, 2

CASE
We present a case of a full-term girl with VACTERL and recurrent urosepsis. Post-natal work up revealed multiple anomalies consistent with VACTERL, including tethered cord, imperforate anus, bilateral superior vena cava, and limb anomalies. Renal bladder ultrasound (RBUS) and spinal MRI at birth indicated severe bilateral hydroureteronephrosis, mild parenchymal thinning, horseshoe kidney, and difficult to evaluate ureters (Figure 1a-b). VCUG showed no reflux, mild funneling of the urethra, a low postvoid residual, and sacral dysraphism (Figure 1c). Urodynamics (UDS) at 3 months of age suggested detrusor sphincter dyssynergia and a small (30 cc) bladder; clean intermittent catheterization and oxybutynin were initiated.

A MAG3 renal scan at 4 months showed 88% function on the left side; there was delayed drainage prior to Lasix administration - but with Lasix, the kidney cleared normally with no evidence of obstruction. This MAG3 scan was coupled with a RBUS that together showed no significant drainage and no hydronephrosis of the right side. After an extensive conversation with a pediatric radiologist, due to the horseshoe configuration and absence of dilation, it was determined that the MAG3 scan likely overread the right moiety, which was ultimately determined to be non-functional and likely non-recoverable. The decision was made to observe.

Between 5 and 7 months, she presented with multiple episodes of urosepsis with poor response to antibiotics. She had two breakthrough infections on trimethoprim/sulfamethoxazole prophylaxis which were treated with amoxicillin; she was switched to nitrofurantoin prophylaxis. However, she still had persistent fevers with vomiting and was subsequently treated with amoxicillin/clavulanate. Urine cultures were sent from the bladder which grew Klebsiella pneumonia and Stenotrophomonas maltophilia, sensitive to amoxicillin/clavulanate. Despite culture directed treatment she remained febrile and hyperdynamic. She was admitted to the PICU. Because of her instability, bilateral percutaneous nephrostomy tubes (PCNs) were placed. A nephrostogram showed no contrast opacification of the right ureter (Figure 1d). Cultures from both PCNs and the bladder at the time of placement grew Pseudomonas aeruginosa. She was started on meropenem. Subsequent cultures showed persistent growth and due to lack of sensitivity in-vivo and she was switched to piperacillin/tazobactam. Given her complicated infectious picture she was transitioned to ciprofloxacin prophylaxis despite its risks in the pediatric population.

Her right kidney function was further evaluated with a 24-hour urine creatinine clearance. Notably, the right kidney produced 200 mL with Urine Cr < 2.50 mg/dL. While admitted, this PCN similarly made significant urine. These results indicated while the right moiety had poor clearance, it still produced high volume and was therefore at risk for infection. Definitive drainage options versus partial nephrectomy were carefully considered. Given the variability in number, origin and size of vasculature for horseshoe kidneys, the isthmus possibly containing 80% functional renal parenchyma, and parental preference to preserve kidney function, all attempts were made to avoid partial nephrectomy.3

Figure 1

Figure 2
Ectopic and Blind Ending Ureteral Insertion (continued from previous page)

To delineate the anomalous anatomy, a follow up MR urography was completed and showed bilateral ureters with unclear ectopic insertion points. She also underwent cystoscopy, vaginoscopy, left retrograde pyelogram, right antegrade pyelogram and methylene blue injection. During this, the left ureteral orifice was found in the posterior proximal urethra (Figure 2a). No right ureteral orifice could be identified.

After bilateral urinary decompression and on ciprofloxacin prophylaxis, she remained infection free. At 8 months she underwent definitive surgery. Intraoperatively, we identified a complete right UPJ obstruction with complete distal ureteral atresia into the bladder-neck. In addition, an intervening section of patent ureter existed between the two (Figure 2a). Consistent with her UDS study, she had a small capacity bladder, presumed to be defunctionalized. Right pyeloplasty and a bilateral non-refluxing reimplant were completed through a Pfannenstiel incision (Figure 2b). The vasculature of the ureter was well visualized and preserved during surgery. Post-operatively she tolerated PCN clamping and stent removal without further infections or admissions. RBUS completed 19 weeks after PCN removal showed marked improvement of the hydronephrosis when compared to original studies.

DISCUSSION

This case illustrates the broad spectrum of ureteral-renal anomalies in children with VACTERL, as well as the difficulties in accurate anatomic diagnosis despite advances in imaging, and the value of temporizing by decompressing the urinary system prior to definitive surgical intervention allowing for patient stabilization.

References


Crossed-Fused Ectopia with Prenatal Hydronephrosis

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CASE

A 31-year-old G1P0 woman underwent first trimester fetal ultrasound which revealed fetal cystic hygroma and fetal hydrops. Noninvasive prenatal testing and chorionic villus sampling confirmed a diagnosis of fetal Turner syndrome. The 18-week anatomy ultrasound showed right hydronephrosis with an anterior-posterior diameter of 8.6 mm and possible left renal agenesis (Figure 1). Bladder and amniotic fluid levels were normal. Other findings included resolved hydrops, small left sided heart structures, and vermian dysgenesis. Fetal echocardiogram showed possible coarctation of the aorta or bicuspid aortic valve. The patient was referred to urology for prenatal counseling for right hydroureteronephrosis and possible left renal agenesis. Recommendations included antibiotic prophylaxis at birth, renal ultrasound at 48 hours of life and voiding cystourethrogram (VCUG).

The right hydronephrosis remained stable through pregnancy and the baby was born at term. Postnatal echocardiogram confirmed coarctation of the aorta, and she underwent repair on day of life 5. Post-

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nital kidney ultrasound showed severe right hydroureteronephrosis with marked parenchymal thinning and a left moiety near midline without dilation; the precise configuration of the renal units was not clear based on ultrasound alone. Creatinine was elevated to 1.3 mg/dL immediately after birth but nadir was 0.28 by day 12.

MRI revealed left-to-right crossed-fused ectopia in an L-configuration, with severe right hydroureteronephrosis and thin parenchyma (Figure 2, A and B). A VCUG demonstrated left grade 3 vesicoureteral reflux (VUR) into a midline collecting system, no right VUR, and a normal bladder (Figure 2, C). Dimercaptosuccinic acid (DMSA) renal scan suggested very limited function of the right, hydronephrotic kidney (Figure 2, D).

The patient was discharged home on day of life 20 and had no urologic issues during her admission. Concern remained for the risk of sepsis should she develop a urinary infection in a severely obstructed, dilated system. Considerations for management were observation on antibiotic prophylaxis, cutaneous ureterostomy, and refluxing or non-refluxing ureteral reimplant. After discussion of options with parents, she underwent a right distal loop cutaneous ureterostomy at 2 months of age. Cystoscopy at the time of surgery revealed an orthotopic left ureteral orifice; the right could not be visualized, indicating the presence of a right ectopic ureter. Her post-operative course was uneventful.

Post-operative ultrasounds have shown only mild right pelviectasis. The patient continues to do well clinically at 14 months postoperatively, with limited urine output from the cutaneous ureterostomy. She remains on antibiotic prophylaxis, without any urinary tract infections (UTIs).

**DISCUSSION**

Around 35% of patients with Turner syndrome have congenital anomalies of the kidney and urinary tract (CAKUT), including horseshoe kidney, hydronephrosis, renal ectopia, duplex collecting system, and solitary kidney. However, there does not appear to be an increased incidence of chronic kidney disease in Turner syndrome patients with CAKUT. Crossed ectopia accounts for around 6% of CAKUT in Turner syndrome. In the general population, VUR occurs in 20% of crossed ectopia cases and ectopic ureteral orifices occurs in 3% of cases.

This case demonstrates the use of a temporizing cutaneous ureterostomy to avoid sepsis in infancy in the setting of an obstructed right collecting system associated with a L-configuration crossed-fused ectopic kidney. Temporizing cutaneous ureterostomies are a safe and effective option for management of significant hydronephrosis in infancy, most often reported on in the setting of megaureter. Kitchens et al. reported on 29 patients with significant hydronephrosis and primary or secondary megaureter who underwent temporary end cutaneous ureterostomy prior to reimplantation.

The risk of stomal stenosis was 3%. There were 9 febrile UTIs during 786 total months diverted, corresponding to a 1% risk of having a febrile UTI per month of diversion. A more recent series by Shrestha et al. reported on the use of end cutaneous ureterostomy in patients with primary obstructive megaureter and either febrile UTIs, palpable hydronephrosis, or renal failure. They reported improved renal function and only one febrile UTI while awaiting reimplantation, with no stomal complications noted.

In addition to reducing the risk of febrile UTI until definitive intervention is pursued, we also believe the diversion will allow more accurate functional assessment of the obstructed moiety in the future. The plan is to re-evaluate with a DMSA renal scan to inform the decision between right ureteral reimplant versus nephrectomy.

**References**

Anorectal Malformation with Ureteral Ectopia and Renal Failure

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INTRODUCTION

Cloacal anomalies are congenital malformations with genitourinary and gastrointestinal confluences, resulting in a singular orifice to exit from the perineum. These abnormalities are present more commonly in female patients with an incidence of 1 per 50000 live births.1 Patients with anorectal malformations present with ectopic ureters at a rate of 3.5%, and commonly have renal dysfunction and higher transplant rates (2.5%).2 Early cutaneous ureterostomies can be performed to help preserve renal function.3

CASE

The patient was born prematurely at a 33-week gestation at 1.6 kg. Prenatal ultrasounds showed thickened nuchal fold, bilateral hydroureronephrosis, cystic hygroma, and oligohydramnios. She was noted to have anorectal malformation with high cloaca, no pelvic floor musculature, and sacral agenesis. Renal ultrasound demonstrated bilateral grade 4 hydroureronephrosis with tortuous, dilated ureters, possible bilateral duplication, and probable dysplastic, cystic kidneys (Figure 1). Spinal ultrasound also showed tethered cord. Additional imaging revealed sacral agenesis; VCUG showed small bladder with no reflux (Figure 2a). Nuclear medicine scan MAG3 showed 9% right renal function.

On day of life (DOL) 2, a sigmoid loop colostomy was performed. Exam under anesthesia and retrograde pyelogram (Figure 2B) revealed an atrophic bladder with an ectopic right ureter opening to the bladder neck, with no obvious left ureteral orifice with suspicion of ectopic insertion. Additionally, a high cloaca was observed with imperforate anus.

On DOL3, Creatinine was noted to increase steadily to 2.6 despite intermittent catheterization of the common channel. Thus, bilateral loop cutaneous ureterostomies were performed on DOL4, showing improvement in urine output. Empiric clean intermittent catheterization was started due to severe tortuosity of the ureters and stopped after 24 hours due to adequate spontaneous drainage.

BUN/Cr values improved (Figure 2d) and repeat RUS showed improvement of hydronephrosis. At the patient’s 4 month follow up, there was resolution of the left hydronephrosis and improved right hydronephrosis. MRI (Figure 2c) showed sacral dysgenesis, anal atresia, rectal pouch at the level of pelvic floor musculature, and absence of right pelvic floor muscles. The patient also had tethered cord surgery performed in subsequent months.

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Currently, both cutaneous ureterostomies are draining well, which was documented by a renal ultrasound performed on 6/21/22 and continued visits during follow-up.

DISCUSSION

50% of cloaca patients have chronic kidney disease, with increased risk of kidney failure, making renal preservation of utmost importance. Children with anorectal malformations and ureteral ectopia have a higher incidence of kidney transplantation compared to those without. In our case, early bilateral cutaneous ureterostomies in the perinatal period helped normalize renal function, averting need for immediate dialysis.

CONCLUSION

The initial aggressive management with urinary diversion utilizing cutaneous ureterostomies in our patient with anorectal malformation and ectopic ureters highlights the need for vigilance with this multidisciplinary team in helping prevent early dialysis, hopefully preserving long term renal function. Long term surgical planning will continue to be a multidisciplinary team approach with the family to achieve the best outcome for the child.

References

Two Patients with 46, XY Sex Reversal: How to Decide on Gender Identity and What to do With the Gonads?

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INTRODUCTION
Disorders of sexual development (DSD), classified into sex chromosome; 46, XX and 46, XY DSDs; XX and XY, sex reversal; and ovotesticular disorders are congenital conditions characterized by atypical chromosomal, gonadal or phenotypic sex. The fate of the bipotential gonad into either testis-specific or ovary-specific pathway is determined by a cascade of molecular events controlled by several genes expressed in the gonadal XY and XX chromosomes. Phenotypic differentiation into internal and external genitalia is subsequently influenced by locally secreted or circulating sex hormones. Advances in genomic sequencing have improved the identification of genes including WW0X, DMT1, NR5A1, HHG, DMR2 associated with gonadal dysgenesis and SRD5A2, CYP21A2, CA21H, FGFR2 associated with disorders of sexual differentiation. Here, we report the cases of two patients with 46, XY chromosome on cell free DNA and discrepancy in perinatal physical examination and imaging.

CASE
Patient 1 was born at 37 2/7 weeks gestational age to a 36-year-old mother. Cell free DNA demonstrated 46, XY fetus and ambiguous genitalia was noted on antenatal ultrasound. Amniocentesis showed a heterozygous pathogenic variant in the NR5A1 gene. C841T missense mutation resulting in Arg281Cys was confirmed on single gene analysis on day 1 of life. Postnatal examination was consistent with female phenotype with normal appearing clitoral hood, labia minora and hymenal ring. Postnatal ultrasound identified abdominal gonads resembling testicles, Mullerian structures and fallopian tubes (Figure 1). Anti-Mullerian hormone and inhibin were undetectable and ACTH stimulation test was unremarkable. Further work up at 2 months of age showed low testosterone and LH and elevated FSH. Given concerns for likely streak gonads, especially with presence of Y chromosomal material, decision was made to perform bilateral laparoscopic gonadectomy at 9 months old. Operative findings were remarkable for normal appearing prepubertal uterus and fallopian tubes, bilateral streak goads involving the fallopian tube and patent right inguinal ring without gas tracking into the right labia. Histological features on final pathology confirmed streak gonads with dissecting gonadoblastoma.

Patient 2 was born at 29 1/7 weeks gestational age to a 20-year-old mother. Cell free DNA demonstrated 46, XY fetus but female genitalia on fetal ultrasound. Ultrasound showed bilateral inguinal gonads without Mullerian structures (Figure 2). Testosterone levels were low and inhibin and AMH were in low end of normal range for infant males but elevated for infant females. Patient failed ACTH stimulation test and required hydrocortisone supplementation for adrenal insufficiency. DNA analysis demonstrated homozygous missense mutation in the CYP11A1 (exon 8:c.C1393T:p.R465W) gene associated with congenital adrenal insufficiency. Patient remained in NICU for 3 months during which family decision was made to raise female. Evaluation at 4 months revealed normal appearing external female genitalia but with palpable structures in bilateral inguinal canals. Patient underwent bilateral inguinal orchietomy and inguinal hernia repair at 4 years old. Operative findings were notable for right abdominal testicle seen entering the internal ring with healthy-appearing gonadal vessels and vas deferens. On the left, the internal ring was noted to be closed with healthy-appearing gonadal vessels and vas deferens entering the closed ring. The testicle had descended beyond the internal ring and was ultimately removed through an inguinal exploration. There was absence of Mullerian structures and normal labia, clitoris, hymenal ring and urethral meatus. Pathology confirmed Sertoli cell only testicle and benign epididymis and spermatic cord. Patient’s developmental course is notable for severe short status, craniosynostosis, and dysmorphic features.

DISCUSSION
These cases demonstrate the variable manifestations of 46, XY DSDs. Gonadal dysgenesis in both cases is likely a result of lack of androgen and AMH expression during development. Pathogenic variants in NR5A1 or steroidogenic factor 1 have been associated with 10-20% of 46, XY DSD cases. NR5A1, by regulating the transcription of critical genes for gonadal and adrenal development, activates expression of AMH and stimulates testosterone biosynthesis by Leydig...
Two Patients with 46 XY Sex Reversal (continued from previous page)

cells. Homozygous NR5A1 mutation is associated with adrenocortical insufficiency as in patient 1. CYP11A1 is a cytochrome P450 protein that catalyzes the conversion of cholesterol to pregnenolone, the rate limiting step in adrenal and gonadal steroidogenesis. Mutations result in varying degrees of primary adrenal insufficiency and 46, XY DSDs, typically with external female genitalia. Studies suggest an increased risk of gonadoblastoma in 46, XY DSDs, however, there is no consensus on timeline for gonadectomy. Phenotypic appearance at birth often determines sex of rearing which often correlates with gender identity over time. A European study with 1040 DSD adults reported gender changes from birth assignment in 5% of participants. Overall, given the heterogeneity of DSD cases, patient management should involve interdisciplinary care teams and shared decision making with families.

References:

It’s a…Baby! Ambiguous Genitalia in Mosaic Turner Syndrome with Y Chromosomal Material

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INTRODUCTION
Turner syndrome (TS) is one of the most common chromosomal abnormalities affecting 1 in very 2500-3000 newborn females. Characterized by short stature and gonadal dysgenesis, 50-60% of the patients have a 45X monosomy karyotype; the remaining karyotypes demonstrate a mosaic pattern. Y chromosome material is detected in about 5% of Turner syndrome patients with mosaicism. We report a case on a patients with Turner syndrome 45X/46XY mosaicism identified by conventional cytogenetics and course of diagnosis and treatment, which the authors believe underscores the importance of complete genetic testing prior to initial gender assignment.

CASE
Our patient was delivered at born at 37 2/7 weeks gestation to G3P3 24-year-old woman. Prenatal female sex was assigned with no prior cell free fetal DNA testing performed. Upon birth, there was ambiguous genitalia noted requiring further evaluation. Notable physical exam findings included: palpable right gonad, bifid scrotum and 2 cm phallic structure with hypospadiac meatus and no vaginal orifice. Pelvic ultrasound demonstrated hydrometrocolpos and presence of uterus. Scrotal ultrasound demonstrated normal-appearing right testicle within scrotum, a small hydrocele containing internal debris, and non-visualization of left gonad (Figure 1). Laboratory testing determined normal electrolytes levels and a normal 17-hydroxyprogesterone level of 71 ng/dL. Results included a dihydrotosterone of 14 ng/dL, testosterone at 22 ng/dL, and Anti-Müllerian Hormone at 29 ng/mL. Genetic testing was conducted, confirming Turner Syndrome (abnormal mosaic pattern of monosomy X with a 19.552 Mb duplication of Yp11.32q11.222 – 45,X [53.5%] and 46, Xyyp [46.5%] with 2 copies of SRY). Parents initially assigned female sex prior to NICU discharge.

Subsequently, the patient was noted to have a symptomatic left inguinal hernia at follow-up visit. They underwent repair at 2 months of age along with endoscopy of urogenital sinus and left gonad biopsy. A finding of high confluence urogenital sinus with normal vagina was notable. The hernia contained gonad that grossly appeared consistent with ovary and fallopian tube. (Figure 2) A wedge biopsy revealed no gonadal tissue.

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It’s a Baby! Ambiguous Genitalia (continued from previous page)

Patient was found to have an elevated testosterone of 101 ng/dL at 2 months and 88 ng/dL at 4 months during mini puberty of infancy consistent with normal male levels. Due to the normal male testosterone levels, the parents have opted to maintain gender neutrality until child is able to participate in medical decision-making.

DISCUSSION

It has been shown that Y-chromosome mosaicism in TS patients predisposes them to increased risk of developing a gonadoblastoma, a rare gonadal neoplasm composed of aggregates of germ cells and immature Sertoli cells or granulosa cells. These tumors are found in approximately 15%–20% of 45X/46XY individuals. Typically, gonadoblastomas are benign tumors unless malignant overgrowth of germ cells occurs and it evolves into an invasive dysgerminoma, which can be seen in up to 60% of cases. For this reason, it is recommended that TS patients with Y-chromosome mosaicism have should prophylactic removal of the dysgenic gonad.2,3

Disorders of Sexual Dysfunction (DSD), such as Y-chromosome mosaicism in TS patients, are complex conditions that require a multidisciplinary approach to addressing both the needs of patient and parents. Often most of these disorders present postnatally with the rare few occurrences unexpectedly appearing on prenatal ultrasound. A method to evaluate for possible genital ambiguity is to utilize non-invasive prenatal testing (NIPT), such as cell free fetal DNA (cffDNA) testing. With earlier detection, this provides for early management and counseling for patients, especially with concern to gender assignment of the child.4

Increased use of NIPT provides the benefit of time, specifically more time for parents to process and to prepare mentally and emotionally for the pregnancy outcome. It provides an increased sense of control over the course of the pregnancy leading to improved decision making.5 This is vital in reducing premature gender assignment, because it allows for a full work-up to be conducted prior to the parents having to make decisions on the fetal sex, which is not often the case with the discovery of a DSD post-birth. This case further reinforces the importance of utilizing methods such as cell free fetal DNA (cffDNA) testing for earlier identification in pregnancy prior to gender assignment.

References

Intraperitoneal Perforation During VCUG in Premature Infant

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INTRODUCTION

Voiding cystourethromgs (VCUGs) are a crucial tool in the urologic workup of infant and pediatric populations. While reported complications are rare, the potential for complications should be considered. Here we present a rare and serious complication of intraperitoneal perforation during VCUG catheter placement with subsequent management.

CASE

A 43-day-old male born at 29 6/7 weeks developed E. coli sepsis during NICU stay at an outside hospital. Prior 20-week prenatal ultrasound was reportedly normal. Renal ultrasound obtained during his sepsis evaluation demonstrated bilateral SFU Grade I hydrenephrosis. He was treated with IV antibiotics with good clinical response. After the infection was adequately treated, a VCUG was obtained to assess for underlying urologic anomaly; however, contrast was seen instilling into the peritoneal cavity and scrotum during the study (Figure 1). Due to this finding, the patient was immediately transferred to our Children’s Hospital. As the patient was stable on arrival, a renal bladder ultrasound was obtained after transfer which showed the foley balloon posterior to the bladder (Figure 2). The patient was taken to the operating room where cystoscopy demonstrated perforation through the prostatic urethra. The injury appeared consistent with catheter placement using a stylet, which was later confirmed with the outside treatment team. A new catheter was placed over a wire with intraoperative cystogram confirming position. The patient remained stable post-operatively. After 2 weeks, peri-catheter retrograde urethrogram showed no extravasation. VCUG at that time demonstrated bilateral III vesicoureteral reflux without urethral anomaly. The catheter was removed, and the patient was discharged home on Bactrim prophylaxis.

DISCUSSION

VCUGs represent one of the most common imaging studies obtained in pediatric urology. The most common complications include minor side effects such as patient discomfort and hematuria. Urinary tract infections after VCUG have also been reported with rates ranging from 1-4%, though given the patient population (i.e., many getting this study for UTI history) one must interpret these numbers with a critical eye.1-3 Regarding intraperitoneal perforations, complications (continued on next page)
Intraperitoneal Perforation During VCUG (continued from previous page)

along this magnitude are exceedingly rare and typically are represented in random case reports. Generally, these have been due to bladder perforations in the setting of very small, noncompliant bladders and prior surgeries. In our case, since the patient was stable, obtaining the ultrasound prior to proceeding to the OR allowed us to confirm the diagnosis of a urethral injury as the catheter was seen coursing posterior to the bladder; this was not evident on outside imaging. These findings refined our preoperative planning and approach. While the patient’s prematurity raised concerns about the feasibility of endoscopic intervention due to associated small urethral caliber, we were able to successfully perform cystourethroscopy. Upon reaching the area of injury we had strong suspicion that the metal stylet that comes with the 6 French Rusch catheter had been used to place the catheter completely through the prostate into the abdominal cavity. This instance reinforces that though these stylets are available in the 6 French Rusch foley kit, they should not be routinely due to risks of significant injury. The patient appears to have no long-term complications at this time, although he will continue to be observed to rule out late-term stricture. In summarizing this case, we feel there are a few key management points to highlight: 1) First and foremost, ensure stability of the patient; 2) Consider your approach and have a backup since premature infant urethras may not accommodate an infant cystoscope; 3) Perioperative antibiotics are recommended given the intraperitoneal contamination; 4) Ensure adequate urethral rest with maximal urinary drainage (foley, SPT, etc.) post-operatively and obtain follow up imaging to ensure resolution; 5) Don’t forget to complete imaging work-up once the injury is stable to rule out other urologic pathology.

CONCLUSIONS

VCUGs, though common, can still result in complications such as the case detailed here. We describe successful conservative management and are working to develop a standardized protocol for VCUGs in high-risk patients from outside facilities.

References

INTRODUCTION
Among all congenital urinary tract anomalies, ureteropelvic junction obstruction (UPJO) is the one of the most common etiologies. However, UPJO in the setting of crossed fused renal ectopia is a rare anomaly with few cases reported in the literature. Therefore, there are limited data on presentation, diagnosis, and management of UPJO in the setting of crossed fused renal ectopia. We aim at reporting a case of right UPJO with crossed fused renal ectopia and our experience its diagnosis and management.

CASE
Right hydronephrosis was initially identified on prenatal ultrasound at 27 weeks of gestation. APRPD was 27mm (Figure 1A). The left renal fossa was empty (Figure 1B). A reniform structure was located near the midline of the fetal pelvis (Figure 1C). There was no hydroureter or dilated bladder. Repeat prenatal ultrasound revealed similar findings without resolution of right hydronephrosis.

The child's delivery was uneventful. Postnatal ultrasound again demonstrated severe right hydronephrosis and crossed fused ectopia. VCUG was performed at 1 month of age, and there was no evidence for reflux (Figure 1D). MAG-3 renal scan at the time revealed delayed drainage of upper renal moiety (right), with a t½ of 33.5 minutes. Split function was fairly equal (Left kidney: 46%; Right kidney: 54%) (Figure 1E). On 6-month follow up, a MRU was obtained for detailed anatomical and functional information. It demonstrated crossed fused ectopia with upper moiety hydronephrosis and cortical thinning with a Patlak differential function of 32% (Figure 1F, 1G, 1H). Upper moiety revealed less intense cortical enhancement, and delayed contrast excretion. The patient had 2 episodes of febrile UTIs during his first several months of life requiring hospitalization. Given his decreased upper moiety renal function and febrile UTIs, a dismembered robotic pyeloplasty was performed.

Intraoperative retrograde pyelogram showed crossed fused ectopia, with high insertion of the right ureter and stenotic right UPJ (Figure 2A, 2B). These findings are confirmed after docking the robot and dissecting the right ureter. Right renal pelvis was dilated, and right ureter inserted more cephalad (Figure 2C). After transecting the ureter at the level of insertion, the UPJ was noted to be narrow (Figure 2D). The ureteropelvic anastomosis was then performed at a more dependent position. Patient was discharged on postoperative day 1. Ureteral stent was removed on postoperative week 5. Patient has been doing well. Postoperative 3-month ultrasound showed significant improvement in right hydronephrosis (Figure 2E, 2F).

CONCLUSION
UPJO in the setting of crossed fused ectopia is a rare combination of anomalies that may not always present itself clearly in the antenatal setting. Postnatal evaluation including cross sectional imaging with MR Urogram may provide detailed anatomic and functional information that are useful in guiding surgical planning.

References
Bilateral Ectopic Ureters Presenting as Prenatal LUTO in Newborn Female

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INTRODUCTION

Lower urinary tract obstruction (LUTO) is more commonly seen in males with congenital anomalies such as posterior urethral valves (PUV), Eagle-Barrett Syndrome and urethral atresia. LUTO with associated oligohydramnios in a female fetus is an exceedingly rare but serious pathology. Here we present a case of LUTO with associated oligohydramnios in a female fetus secondary to bilateral ectopic ureters with subsequent management.

CASE

A 36w0d female was born via c-section at a large freestanding tertiary care pediatric facility. Her prenatal history was complicated by Lower Urinary Tract Obstruction (LUTO) diagnosed by prenatal ultrasound at 24 weeks. A multidisciplinary prenatal consult was obtained including the Maternal Fetal Medicine (MFM), cardiology, nephrology, and genetics teams. Fetal MRI obtained at 26 weeks’ gestation demonstrated bilateral hydroureter with apparent ectopic insertions and associated ureteroceles, cystic kidneys and oligohydramnios (Figure 1). The mother was admitted to the MFM unit and followed with serial ultrasounds. At 36 weeks’ gestation she underwent cesarian section for breech presentation after developing anhydramnios. Given her prenatal history, Urology was consulted in the NICU immediately after birth. She had been placed on extracorporeal membrane oxygenation (ECMO) due to severe pulmonary hypoplasia, which also provided renal replacement therapy. Renal bladder ultrasound was obtained on DOL#0 showing bilateral hydroureter, bilateral echogenic cystic kidneys and a decompressed bladder without clear ureteral insertion into the bladder (Figure 2). On DOL #9, she was successfully transitioned off ECMO to continuous renal replacement therapy (CRRT). She was taken to the OR for peritoneal dialysis (PD) catheter placement and gastrostomy tube placement by pediatric surgery at 6 weeks of life with plans for long-term PD as a bridge to renal transplantation. Unfortunately, she developed sepsis at 9 weeks of age and died from fungemia 4 days later.

DISCUSSION

An ectopic ureter is defined as any ureter that does not enter the trigone. They occur in approximately 1 in 2000 newborns and are more commonly seen in females, where they may enter multiple sites such as the bladder neck, vagina, or perineum. Classic presentation in females is con-
Bilateral Ectopic Ureters (continued from previous page)

tinuous incontinence, whereas in males continence is maintained due to ectopic ureteral insertion proximal to the external urethral sphincter. Bilateral single system ureteral ectopia is extremely rare and may be associated with a hypoplastic bladder and renal dysplasia, which occurred in our patient.¹ These cases, though rare, represent difficult treatment challenges. As in this case, though urology providers may not be as directly involved in the newborn management, particularly if renal replacement therapy (RRT) is needed, it is important to provide appropriate counseling - as there is high morbidity and mortality associated with neonatal dialysis.² For those with associated pulmonary hypoplasia, though ECMO can serve as temporary RRT, it can be very difficult to transition from ECMO to other forms of RRT. Another treatment challenge is to get patients to a size and age in which they can accommodate a renal transplant, which is typically 10 kg (20lbs) and 2 years of age. To do so, adjuncts such as gastrostomy tubes are often used to help with nutritional growth requirements. For those who have functional native kidneys, additional challenges include determining timing and location of ureteral reimplantation in a hypoplastic bladder. In such cases, controversy also surrounds considerations such as concomitant augmentation and continence procedures such as bladder neck reconstruction or closure.³

CONCLUSIONS

In summary, we present the case of a newborn female with prenatally diagnosed LUTO with associated anhydramnios secondary to bilateral ectopic single system ureters temporarily managed with ECMO in the setting of concomitant pulmonary hypoplasia. While she was successfully transitioned to PD, this patient ultimately succumbed to line sepsis in early infancy. This case highlights treatment considerations as well as controversies and challenges in management of these complex patients.

REFERENCES


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