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Case Series
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Multidisciplinary Management of Bladder Agenesis and Anorectal Malformations in Female Patients: A Case Series

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Background

Bladder agenesis is an exceedingly rare congenital anomaly with an estimated incidence of 1 in 600,000 [1,2]. Patients born with bladder agenesis carry a poor prognosis due to its association with renal impairment and defects of other organ systems, including the reproductive tract, vascular system, and neurologic system [2-4]. About 90% of those born with bladder agenesis are female, with an estimated female-to-male ratio of 30:1 [1,2]. Herein, we present two cases of bladder agenesis and one case of bladder dysgenesis in female patients; two underwent successful ileal neobladder creation and one underwent incontinent colonic conduit creation.

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Case 1

Our patient is a female, born at 37 weeks of pregnancy at an outside hospital. At birth, she was found to have a single perineal opening which appeared to be a vaginal introitus, absent urethra, and an imperforate anus. We did not have access to prenatal imaging, however, the patient's mother reported prenatal complications including oligohydramnios and antenatal hydronephrosis prompting induction at 37 weeks' gestation. This patient underwent a diverting colostomy procedure shortly after birth and was given the diagnosis of cloacal malformation. However, cystoscopy at the time of colostomy could not identify a urethral opening or ureteral orifices. The patient was voiding spontaneously but was also diagnosed with Stage 2 Chronic Kidney Disease (CKD) with a left atrophic kidney. At 14 months of age, she underwent a suprapubic tube placement into a fluid-filled structure believed at the time to be a small, contracted bladder. Cystoscopy through the vaginal introitus revealed a cervix at a depth of 4.5 cm. Upon retrospective review of the patient's chart, it is unclear what diagnostic studies and work-up the patient had between birth and 14 months of age.

At 15 months of age, an MRI Urogram showed an atrophic left kidney, duplex right kidney without hydronephrosis, stenosis of the mid-left ureter, and bilateral ureters that inserted low into what appeared to be the posterior bladder. Even so, contrast preferentially filled the vaginal vault (Figure 1a). Around this time, she also underwent a retrograde voiding cystogram. Contrast was injected through a suprapubic catheter, filling what appeared to be a small capacity bladder (Figure 1b). There is also reflux into the left ureter, which appears to drain into the inferior medial aspect of the cloacal channel (Figure 1b).

The patient developed recurrent UTIs. Upon further investigation with cystoscopy and genitogram, it was found that to the left of the cervix there was a small orifice connecting the common channel to an irregularly shaped rudimentary bladder, but no bladder neck or sphincter muscle was noted. The suprapubic catheter was found to be placed into the common canal and was removed intraoperatively.

Additionally, it was found that the distal rectum inserted posteriorly into the common cloacal channel. At the age of three years, she presented at our institution, where diagnostic laparoscopy revealed that the bilateral ureters inserted into the vaginal canal. At our institution, she underwent a Posterior Sagittal Anorectoplasty (PSARP) and takedown of the rectovaginal fistula. Three months later, she underwent a colostomy reversal with reanastomosis with distal colon. The decision was to use a portion of the end colostomy as a diverting stoma and the formation of ureterocolostomy. Currently, she is doing well and has been able to discontinue prophylactic antibiotics.

Case 2

A 46, XX female at 31 weeks gestational age presented at our institution at birth with imperforate

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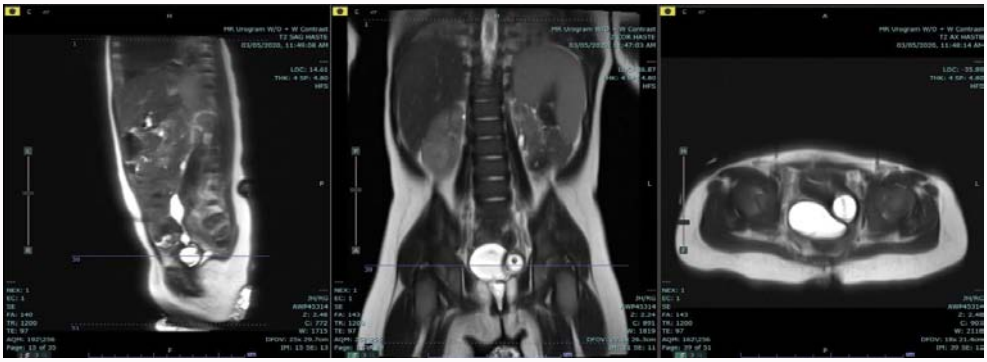

Figure 1a: MR Urogram at age 15 months (Case 1).
Left Image: T2 Sagittal view of location of presumed bladder. Middle Image: T2 Coronal View of presumed bladder location. Right Image: T2 Axial showing presumed bladder location with adjacent SP tube balloon.

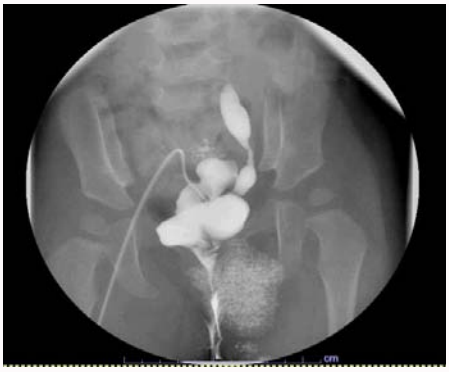

Figure 1b: Retrograde voiding cystogram at age 15 months, demonstrating contrast injected through suprapubic catheter filling common cloacal channel (Case 1). There is reflux into left ureter which appears to drain into inferior medial aspect of cloacal channel.

anus. The patient's mother underwent an urgent cesarean section due to pre-eclampsia. The pregnancy was otherwise unremarkable. A clinical exam revealed a single umbilical artery, a single-opening posterior to the clitoral hood that drained urine, and a blind-ending anal dimple. A subsequent endocrine workup was negative for congenital adrenal hyperplasia. An abdominal ultrasound on day 1 of life revealed a solitary fused pelvic kidney but providers were unable to delineate a bladder. On day 2 of life, she underwent abdominal exploration and creation of an end colostomy. An intraoperative examination revealed a blind-ending anal mucosa, two fallopian tubes, and small streak ovaries bilaterally. At the time, there was no clear delineation of the uterus from bladder. An MRI was deferred given the patient's small size.

At eight months of age, the patient underwent a cystoscopy and perineal examination with neuromuscular electrical stimulation of perianal musculature under general anesthesia to further evaluate her anatomy. On external perineal examination, she had a normal clitoris, labia minora, labia majora, and a hymen. Endoscopy revealed a common channel deep to the hymen, measuring 4 cm in depth. There was no vaginal canal or cervix identified. There was an orifice draining urine on the anterior wall of this channel. Neuromuscular stimulation of the perianal musculature revealed no contractions, but it is noted that the patient was able to contract perianal musculature voluntarily. A cystourethrogram was performed and 10 cc of contrast filled the common channel, forming a small triangular shape. No reflux was noted.

During her third year of life, she underwent an MRI Urogram. The findings confirmed ectopic fused kidneys draining into a single ureter, which drained into a triangular, bladder-like structure (Figure 2a). Two ovaries were visualized, and no uterus was identified. The patient had a colostomy takedown and PSARP. At age 10 years, the patient underwent hemodialysis catheter placement. The patient was still producing urine; therefore, she underwent Mitrofanoff procedure and creation of an ileal T-pouch. At age 13 years, the patient had a deceased donor kidney transplant, with the transplanted ureter reimplanted directly into the ileal T-pouch, with the rudimentary bladder left *in situ* (Figure 2b). Postoperative complications included acute rejection of donor kidney and acute kidney injury, secondary to

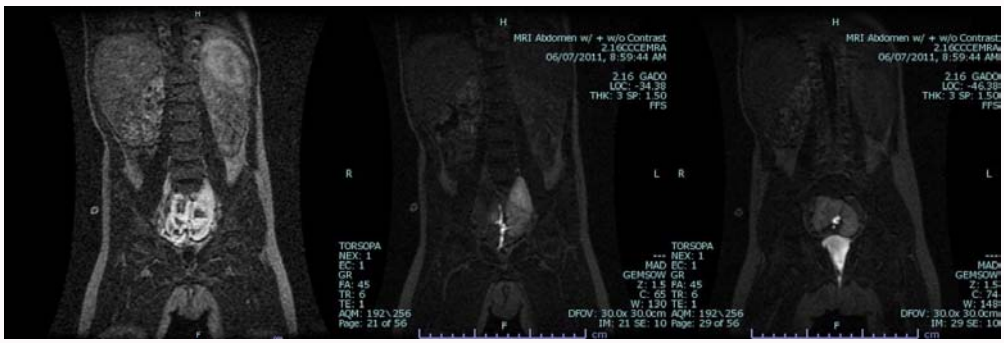

Figure 2a: A 46-XX female at 31 weeks gestational age presented at birth with imperforate anus and ambiguous genitalia (Case 2). MR Urogram at age 3 years is shown, demonstrating fused pelvic kidneys (left) with common drainage (middle) into an anomalous structure, later determined to be a rudimentary vaginal canal (right) and absent bladder.

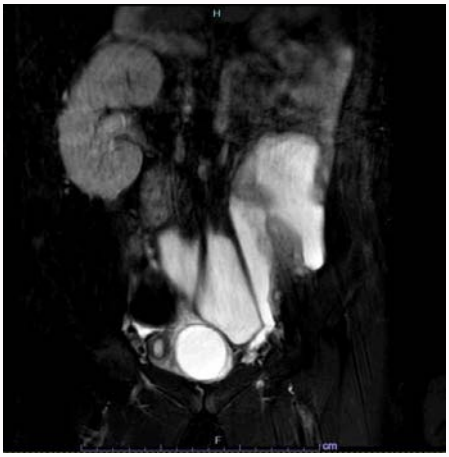

Figure 2b: MRI at age 13, s/p neobladder creation with Mitrofanoff, loculated fluid collection to the left of the neobladder, and transplanted right kidney.

a subtherapeutic immunosuppressive regimen.

Currently, the patient is doing well and uses a 14 French catheter through the Mitrofanoff every two to three hours during the day, with a collection bag for overnight drainage. She is also irrigating her bladder weekly. Due to the blind ending of the common channel deep to the hymen, postulated to be a rudimentary bladder canal, consideration will be made to dilate the single-opening ventral to the clitoral hood to establish a functional vagina pending further discussions with the patient and caregivers.

Case 3

A twelve-day-old twin female presented to the emergency department with decreased oral intake and lethargy. She was found to have both electrolyte abnormalities and hypoglycemia and was subsequently admitted for acute kidney injury. She had previously been feeding well until day seven of life. A renal ultrasound revealed left hydroureter and a right cystic kidney. A VCUG study revealed a bladder-like structure with no vesicoureteral reflux. Prenatal imaging studies were not available, though the patient's mother reported of a possible "hole in bladder" found on prenatal ultrasound. This suggests that some anatomic abnormality had been communicated by medical providers. Otherwise, she had an unremarkable prenatal workup, did not require NICU admission, and was able to go home with her family shortly after birth. On day 17 of life, an MRI Urogram confirmed ultrasound findings and revealed a duplex left renal collecting system with hydronephrosis and a solitary ureter draining the left kidney into the distal vagina.

On day 21 of life, a percutaneous nephrostomy tube was placed to decompress the left kidney due to acute kidney injury and hydronephrosis. An anterograde pyelography revealed a left kidney with a bifid collecting system, a severely dilated lower pole, and a solitary ectopic blind-ending distal ureter. At the time of the exam under anesthesia, a single perineal opening was noted. Cystoscopy revealed a common channel with a low confluence urogenital sinus that split off anteriorly and posteriorly. Further inspection revealed a small, rudimentary bladder with no ureteral orifices anteriorly, and a posterior vaginal canal. With the use of methylene blue and anterograde pyelography, it was established that the patient had no communication between the left ureter and rudimentary bladder or vagina.

At eight weeks of age, the patient underwent laparoscopic G-tube placement for feeding difficulties and anal dilation. At five months of age, the patient underwent a left cutaneous ureterostomy and left pyeloplasty for ureteropelvic junction obstruction. In her first year of life, she had recurrent UTIs, developed secondary adrenal insufficiency due to chronic steroid use, and had a laminectomy for tethered cord release. At age 17 months, she had another cystoscopy and cutback of the urogenital sinus flap to allow for better catheterization for a trial of bladder distension. Unfortunately, she was unable to tolerate bladder cycling. Thus, we created a Mitrofanoff procedure and created an ileal T-pouch on the patient at age six (Figure 3a), with placement of a peritoneal dialysis catheter. Six weeks later, the patient underwent a deceased-donor right kidney transplantation, right nephrectomy of native kidney, and peritoneal dialysis catheter removal (Figure 3b).

Discussion

We presented three rare cases of bladder agenesis, all of which were associated with cloacal malformations. All cases have undergone successful urinary diversion in the form of a ureterocolostomy, and two neobladder ileal T-pouch with continent catheterizable stoma.

Bladder agenesis is often incompatible with life due to its association with renal failure and other congenital anomalies. 90% of life-compatible cases occur in females [4]. This is hypothesized to be due to the conservation of renal function through drainage of the urine from the ureters into the uterus, vaginal canal, or other vestibule formed by the Mullerian structures [4]. It was previously suggested that in males with bladder agenesis, urine drainage must occur through a patent urachus or rectum to be compatible with life. However, Omil-Lima et al. presented a case report of a viable male child with left ureter implantation distally into the left seminal vesicle [4].

The pathophysiology of bladder agenesis is unknown but has been attributed to urogenital sinus maldevelopment at weeks five to seven of embryogenesis [5,6]. In normal development, this embryonic period is defined by the division of the cloaca into the anorectal canal posteriorly and the urogenital sinus anteriorly. The urogenital sinus is continuous with the allantois, which later regresses into the urachus. The superior urogenital sinus above the junction of the mesonephric ducts is what forms the future bladder. At this point, trigone development begins. The ureteric bud arises

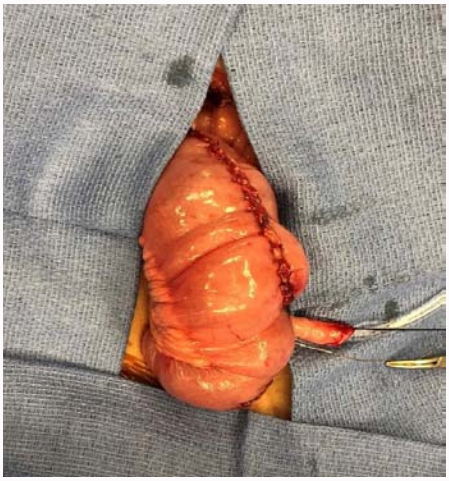

Figure 3a: Intraoperative image of neobladder with catheterizable stoma.

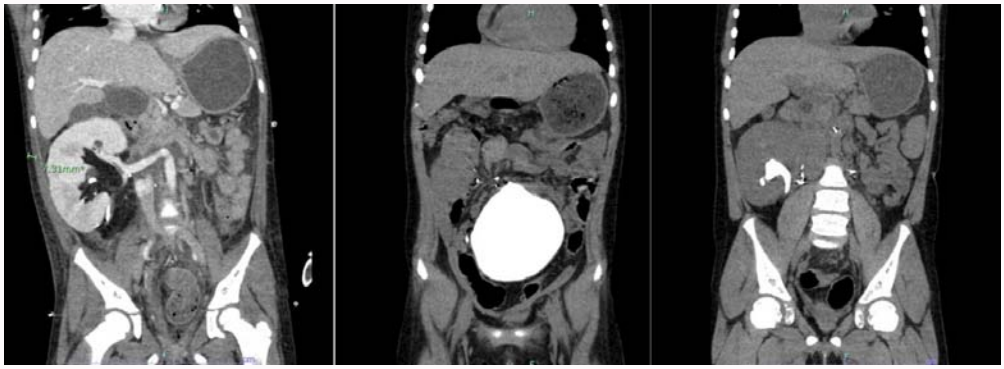

Figure 3b: CTAP of 6-year-old female (Case 3) with bladder agenesis and CKD on POD5 status post urinary diversion with Mitrofanoff, neobladder creation and right renal transplant (left). There is normal enhancement of transplant renal artery and vein. CT urogram of patient one-year post-transplant at age 7 after injection of contrast into Mitrofanoff (middle) and reflux of contrast into transplant renal collecting system with mild hydronephrosis (right).

from the mesonephric duct and grows superiorly until it contacts metanephrogenic mesenchyme. This induces differentiation of the metanephrogenic mesenchyme to form nephrons and triggers elongation of the ureteric bud to form future ureters. The section of mesonephric duct that is inferior to the ureteric bud, is now identified as the common nephric duct, which is continuous with the urogenital sinus. The common nephric duct contacts the developing bladder and undergoes apoptosis, which brings the primitive ureter in close contact with urogenital sinus, thus fusing the structures together. As development progresses, the ureteric orifices migrate superiorly and laterally to form the upper trigone [7].

In bladder agenesis, the ureteric bud fails to assimilate with the urogenital sinus and thus, the ureters fail to assimilate into the trigone [6]. This prevents filling and distension of urine in the bladder and may lead to underdevelopment of the bladder. As such, patients may have ureteral duplication or ectopic ureters, in which the ureteric bud may instead implant into the upper parts of the vagina or urethra in females- as was observed in our patients- or Wolffian duct derivates in males, which later form the seminal vesicles and vas deferens [7]. Development of the posterior anorectal canal is usually normal, but abnormalities have been reported as in the cases presented here. Other anomalies, such as genital ambiguity, uterine malformation, renal agenesis, multicystic dysplastic kidneys, spinal cord defects, and vascular malformation, often accompany bladder agenesis.

Bladder agenesis may present in-utero with oligohydramnios, hydronephrosis, or other genitourinary anomalies, but some cases may not be evident until early childhood. In infants and children, it may present with recurrent urinary tract infections. Diagnosis of bladder agenesis relies on physical exam findings, clinical symptoms, and imaging studies. The most frequently reported imaging study to confirm diagnosis is MRI, though CT, retrograde urogram and intravenous urography have also been reported [2,3].

Urinary diversion is crucial to prevent or delay the progression of chronic kidney disease in this patient population. In all three of our cases, appropriate urinary diversion in the form of a ureterocolostomy and two with ileal T-pouch with a continent catheterizable channel were performed after thorough counseling and discussion of options with caregivers.

Conclusion

In conclusion, our case series highlights the importance of early multidisciplinary intervention in female infants born with bladder agenesis and anorectal malformations. Bladder agenesis is a rare congenital anomaly that carries a poor prognosis due to its association with renal failure and anomalies of other organ systems. In cases compatible with life, successful optimization of genitourinary health with appropriate urinary diversion using a variety of techniques is of paramount importance. In the three cases presented here, all female patients with anorectal anomalies underwent successful urinary diversion utilizing a variety of techniques. The rarity of bladder agenesis and its association with other congenital anomalies make management challenging, but early recognition and multidisciplinary medical intervention can help improve outcomes for affected patients.

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