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Review

Management of the urological tract in children with anorectal malformations – a contemporary review

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***Abstract*:** Anorectal malformations (ARMs) consist of a broad spectrum of congenital anomalies that are associated with an equally wide variety of urological abnormalities, often with increasing incidence as the severity of the ARM increases. The importance of urologic involvement in the care of ARM patients has been noted for decades and is critical from birth to adulthood. Urology must be involved in the initial evaluation and operative care of the child as well as in monitoring and managing issues such as neurogenic bladder, renal disease, and eventually sexual function and fertility. Care of the ARM patient must be done through a multidisciplinary lens, with the urologist as a key player. This review will serve as an update on the management of the urologic tract in children with ARM.

Keywords: anorectal anomaly, cloaca, imperforate anus, urinary incontinence, urinary tract anomalies

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Introduction

Anorectal malformations (ARMs) consist of a broad spectrum of congenital anomalies that affect 1 in 5000 births.[1] The Wingspread classification is the original simplified system that divides commonly seen anomalies into high, intermediate, and low ARMs.[2] In 1995, Peña[3] devised a classification which grouped ARMs based on more specific anatomy and on the child's sex. In 2005, the Krickenbeck classification was created, building off of Peña's anatomic classification and adding more rare variants.[4] Table 1 consists of a summary of the ARM classification systems. These classifications are meant to guide medical and surgical management for each child and to offer a common language when reporting outcomes.

Patients with ARM have a wide variety of urological abnormalities, often with increasing incidence as the severity of the ARM increases.[5,6] Urological involvement in the care of all children with ARM is critical, as chronic kidney disease (CKD) and end stage renal disease (ESRD) are the most likely cause of decreased life expectancy in this population.[7] If ARM patients are found to have associated spinal anomalies, urology must be involved for management of potential neurogenic bladder (NGB), regardless of the involvement of the urinary system in the malformation itself.

The importance of urologic involvement in the care of ARM patients has been noted for decades in the published literature.[8–10] Historically, the 'team concept' of care, what is now known as multidisciplinary care, was touted as paramount to optimizing outcomes for these patients. This review will address the contemporary management of the urological tract in children with a range of ARM. It will discuss early evaluation and operative management as well as postoperative and long-term considerations.

Initial evaluation

Prenatally, there are very few interventions required of patients with ARM. In fact, prenatal diagnosis is often missed unless associated anomalies are present such as those found in VACTERL association (vertebral defects, ARM, cardiac

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Table 1. Summary of ARM classification systems.

	Krickenbeck (2005)[4]	Wingspread (1984)[2]				Peña (1995)[3]
		High	Intermediate	Low	Cloaca	
Male	Rectoperineal fistula			X		Perineal fistula
						Rectourethral fistula
	Rectourethral bulbar fistula		X			-Bulbar
	Rectourethral prostatic fistula		X			-Prostatic
	Rectobladder neck fistula	X				Rectovesical fistula
	Imperforate anus without fistula		X			Imperforate anus without fistula
Female	Rectoperineal fistula			X		Perineal fistula
	Rectovestibular fistula			X		Vestibular fistula
	Cloaca with short common channel (<3 cm)				X (int.)	Persistent cloaca (<3 cm)
	Cloaca with long common channel (>3 cm)				X (high)	Persistent cloaca (>3 cm)
	Imperforate anus without fistula		X			Imperforate anus without fistula
Complex	Rectal atresia/stenosis	X				Rectal atresia
	Posterior cloaca					
	Cloacal exstrophy					
	Associated presacral mass					
	H-type fistula					
	Rectovaginal fistula					
	Pouch colon					

ARM, anorectal malformation.

anomalies, tracheoesophageal fistula, renal anomalies, and limb abnormalities). Solitary kidneys, cross-fused ectopia, renal dysplasia, ureteral anomalies, and hydronephrosis may be urologic findings that need to be addressed after the fetus is born.[7,11–14]

After birth, an overall assessment of the patient's anatomy and any associated congenital anomalies is key. A spinal ultrasound and sacral X-ray as well as renal and pelvic ultrasounds to assess for hydronephrosis or hydrocolpos should be performed.[15–18] When the patient is brought to the operating room for management of the rectum and anus, either with primary repair or diverting colostomy, urology should be present to perform an exam under anesthesia (EUA). The EUA should consist of a cystoscopy and vaginoscopy for patients with vaginas. If a cloacal anomaly is present, the length of the common channel, urethra, and vagina are important for future operative decision making. This examination can also be helpful for the reconstruction as it usually allows for good visualization of the site of the fistula and enables accurate positioning of a urethral catheter.

A urethral catheter is often left in place for 24 h and once removed, post-void residuals (PVRs) should be obtained to ensure appropriate drainage. If there is concern for incomplete emptying, clean intermittent catheterization (CIC) may be initiated, although this can be challenging as the catheter will often preferentially enter the old fistula site. If there is persistent significant hydronephrosis or the presence of febrile urinary tract infections (UTIs) despite appropriate bladder drainage, a voiding cystourethrogram (VCUG) and initiation of antibiotic prophylaxis should be considered. In patients with recurrent UTIs prior to reconstruction, it is also important to ensure that fecal material is not able to enter the urinary tract through the distal stoma.

If hydrocolpos is present, this can be managed with a vaginostomy tube or intermittent catheterization of the vagina. Early drainage of the vagina is important, as a distended vagina can cause compression of the trigone and thus bilateral ureteral obstruction.[19] A study by Chalmers *et al.*[20] suggests that intermittent catheterization of the vagina may be preferred to vaginostomy tube as it minimizes scarring that could affect future total urogenital sinus mobilization (TUM).

Operative management

The role of the urologist in definitive operative management of ARM depends on the extent of urinary tract involvement. Regardless of the repair, the urologist must be present preoperatively to counsel parents on the possible need for CIC postoperatively. The presence of sacral anomalies as well as the anticipated extent of dissection may help to predict likelihood of CIC. However, setting the expectation that CIC and need for future continence procedures are possible is of great importance.

In patients with a rectourethral or rectovesical fistula, the urologist may aid in identification of the urethra and urethral or bladder repair if there is mucosal violation. However, the goal in dissecting the fistula in both posterior sagittal anorectoplasty (PSARP) and laparoscopic anorectoplasty is to be in a submucosal rectal plane to prevent urethral injury.

In patients with cloacal malformations, the urologist is often involved for the initial endoscopy and urogenital mobilization, whether partial or total, after the vagina is separated from the rectum. Patients with short common channels, defined as less than 3 cm, are often candidates for posterior sagittal anorectovagino-urethroplasty (PSARVUP). Those with long common channels, defined as greater than 3 cm, are more challenging and may require an open or laparoscopic approach with more complex genitourinary reconstruction.

TUM involves bringing the urethra and vagina to the perineum as a single unit. This approach, compared with separating the urethra and vagina, has been shown to decrease rates of urethrovaginal fistula.[21] To achieve greater length, the suspensory ligaments of the urethra as they attach to the pubic bone can be divided. This same approach without dividing the suspensory ligaments is known as partial urogenital sinus mobilization. Once the common channel is mobilized adequately, the channel is divided and can be used to reconstruct labia minora, if needed. The urethral meatus and vaginal introitus are matured to the perineum in anatomically appropriate locations.

When a long common channel is present, more complex decision making is often employed. If the length of urethra is shorter than a centimeter and a half, there may be little utility to attempting to reach the urethra to the perineum, as the likelihood of continence will be low.[22] Rather, the urethra can be separated from the vagina, and the common channel essentially becomes the distal urethra. The vagina can then be mobilized and pulled through to the perineum separately. During this dissection, the ureters may run very close to where the vagina approximates the bladder neck. In addition, aberrant ureteral anatomy is common in patients with ARMs and can complicate this dissection.[23] The use of ureteral catheters may help to identify and protect the ureters during this portion of the case.

In the immediate postoperative period, the urologist should dictate the duration of urinary drainage, which will depend upon the extent of urethral mobilization and difficulty of fistula dissection. As previously stated, they should be prepared to counsel the families on the need for CIC and should obtain PVRs after catheter removal. Although rare, some children may require antibiotic prophylaxis or anticholinergic therapy if there is concern for vesicoureteral reflux (VUR) or bladder dysfunction to minimize renal damage. Repeat renal ultrasound is recommended 4–6 weeks after

repair. If there is new or worsening hydronephrosis, consideration should be given for other ways to improve bladder drainage.

Bladder management and continence

It is estimated that 25% of children with ARM will have findings consistent with NGB, which may be due to multifactorial etiologies.[24,25] NGB is more common with concomitant spinal anomalies, which may be present in one third of patients with ARM.[26,27] Iatrogenic neuropraxial injury may occur during primary repair and is more likely the more severe the ARM and whether the repair was done transperitoneally. The risk of denervation is thought to be low if PSARP is available given the fistula location, although care is always needed as the neural innervation runs close to the fistula.[28,29] Some also hypothesize that there is an inherent predisposition to neurogenicity the higher the fistula location using the Krickenbeck classification.[30]

Monitoring with routine renal bladder ultrasounds to assess for bladder emptying and hydronephrosis is key to the urological follow-up of these patients. This may change over time, particularly in those with spinal and sacral anomalies, and must be followed as the child grows. The exact timing of repeat imaging and follow-up is unknown, as we do not have well-defined criteria for who is at more risk than others. Regardless of ARM level, at least yearly follow-up in a multidisciplinary clinic with an ultrasound is the practice at our institution to assess for continence, infections, and any other urologic concerns.

If there is concern for poor bladder emptying or spinal and sacral anomalies, urodynamics (UDS) should be performed to assess for severity of NGB. Management with CIC or vesicostomy, with anticholinergics added if needed, are mainstays of therapy. If the urethra is difficult to catheterize, utilization of Coude catheters or considering early continent catheterizable channel formation may be considered. If there is concern for unsafe filling pressures, anticholinergics, intravesical Botox injections, or enterocystoplasty should be considered to reduce upper tract damage. If there is concern for hydronephrosis on an ultrasound, evaluation with video urodynamics (VUDS) can assess for reflux. Any number of UDS findings can be seen in ARM patients and each patient should be treated with an individualized lens.

The definition of continence varies significantly among literature of all complex congenital genitourinary conditions and can be difficult to interpret. In general, most reports conclude that the more severe the malformation, the more likely to experience incontinence or require CIC and surgical intervention to achieve continence.[31–33] One study reported no difference in continence rates between rectoperineal fistulas and controls.[31] Comparatively, the rate of continence with rectobladder neck fistulas was only 15% without CIC and 43% if CIC is considered achieving social continence.[32]

For patients with common cloacas, the literature suggests that 20% of patients with short channels (less than 3 cm) and 80% of patients with long channels (greater than 3 cm) will need to perform CIC.[19] Warne *et al.*[33] reported that 80% of patients in their cohort achieved social continence, but only 22% of these girls voided spontaneously. Multiple groups have shown similar results, that in longer channels, children are more likely to require CIC and have increased neurogenic findings on UDS over time.[12,28,34] Creation of a continent catheterizable channel may be offered to patients as they are often sensate and prior surgical repair may make CIC difficult. In summary, emphasizing that achieving social continence may require CIC is important to appropriately manage expectations in ARMs, particularly with common cloacas.

Renal disease

CKD and ESRD are the greatest cause of decreased life expectancy in patients with ARM.[7,35,36] The risk may be as high as 6.4% in higher ARM compared with 1.1% in low ARM.[37] Patients with a history of cloacal anomalies seem to be at the highest risk of renal damage – it is estimated that 50% have some form of CKD by age 11.[7]

There are multiple contributing factors to kidney disease in this population. Obstructive uropathy and recurrent infections are primary causes of nephron loss. This may be due to VUR, NGB, or persistent obstruction. In these patients, routine follow-up with a urologist, correction of any ARM, and management of urologic anomalies will help preserve renal function. Rarely, primary renal dysplasia contributes to CKD and ESRD in ARM patients; however, some do present with renal agenesis, dysplasia, and multicystic kidneys.

It is estimated that 30–40% of children with ARM will have VUR found on VCUG.[7,25,38] Because of the increased risk of NGB, secondary reflux from elevated detrusor pressures must be considered. This reflux poses a high risk to renal function and the primary NGB should be addressed rather than the reflux itself. VCUG or VUDS should be obtained on ARM children with new or worsening hydronephrosis after primary repair to assess for VUR. Treatment focused on initially addressing bowel and bladder dysfunction, which is a significant concern in ARM patients, is paramount to successful management. Prophylactic antibiotics should be considered as well if there have been recurrent UTIs. If NGB and neurogenic bowel management are optimized and the child still has VUR and infections despite prophylaxis, ureteral reimplantation can be considered to reduce parenchymal scarring but only after ensuring that bladder pressures are normal.

Long-term considerations

With improvement in the management of patients with ARM, these children are living well into adulthood with almost normal life expectancies.[39,40] Therefore, focus on the long-term urological outcomes and management of these patients is critical to their care. Ultimately, the length of the common channel, length of urethra after repair, and concomitant spinal or sacral anomalies seem to have the most impact on long-term urinary and bowel continence.

Regarding continence and bladder function, Davies *et al.*[41] performed a questionnaire-based study of adults with a history of ARM and showed that higher malformations, particularly cloacas, had worse outcomes regarding continence and need for CIC. In their report, the general rate of incontinence was 41% and was as high as 80% in patients with common cloaca. All patients with low ARMs and 80% of high ARMs voided spontaneously per urethra – only 33% of common cloaca patients did so, the majority performing CIC through a continent channel. A more recent study by Chong *et al.*[42] noted a general incontinence rate of 32% in ARM adults, with increasing severity the higher the level of ARM.

CKD and ESRD remain considerations in adolescence and adulthood and should be monitored yearly by a nephrologist if there are any concerns. Annual urinalysis to assess for proteinuria, blood pressure, and renal bladder ultrasound are critical to maintaining kidney health. A recent study showed that nearly 30% of adults with ARMs live with CKD and almost 10% had undergone renal transplantation.[42]

The literature on sexual health and function in adult ARM patients is sparse; however, a few general conclusions have been suggested. Overall, it is thought that ARM patients have a later sexual debut than normal counterparts and that incontinence, both urinary and fecal, can significantly influence sexual well-being.[41,43–45] Davies *et al.*[41] found that both men and women with ARMs scored lower in body esteem scales, specifically regarding physical condition and sexual attractiveness. Among men, 17% reported erectile dysfunction, but the majority reported that they were very or mostly sexually satisfied. Both men and women with more frequent urinary and bowel incontinence had higher sexual anxiety scores, which highlights a key patient counseling and management point.

Fertility in ARM patients will be feasible by natural means for most patients with low fistulas. In men with high fistulas, particularly if the bladder neck is involved, these patients may be a high risk for retrograde ejaculation and may require assisted reproductive technology (ART) for conception. Holt *et al.*[46] showed that in a group of men with ARM being evaluated for infertility, iatrogenic injury to the pelvic nerves was responsible for infertility in at least 50% of patients.

For women, concomitant Mullerian anomalies may affect the ability to carry a pregnancy. For those with a uterus, rates of conception should be comparable with the general population; however, they may be at higher risk for miscarriage and pre-term delivery because of their anatomy.[47,48] Women with rectovestibular fistulas may be allowed to deliver vaginally, but those with common cloaca should be counseled on elective Cesarean section. And those with a history of urinary reconstruction should similarly be counseled on having a urologist present at delivery to help manage the surgical reconstruction intraoperatively.

Conclusion

The urologist plays a key role in the management of ARMs from birth through adulthood. Assisting in the corrective reconstruction, optimizing renal function, and managing NGB are critical

components of care of these complex patients. Continuing to address concerns about sexuality, fertility, and pregnancy through adulthood are also within the scope of urologic management. Partnering with pediatric colorectal surgery, pediatric and adolescent gynecologists, pediatric neurosurgeons and pediatricians, and eventually adult providers to give comprehensive and multidisciplinary care will help ensure optimal health outcomes and excellent quality of life.

Declarations

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Not applicable.

Consent for publication
Not applicable.

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Kelly T. Harris: Conceptualization; Writing – original draft.

Duncan T. Wilcox: Conceptualization; Writing – review & editing.

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