Seminars in Pediatric Surgery 25 (2016) 108–111

Contents lists available at ScienceDirect

Seminars in Pediatric Surgery

journal homepage: www.elsevier.com/locate/sempedsurg

Long-term urological outcomes in cloacal anomalies

Brian T. Caldwell, MD*, Duncan T. Wilcox, MD, MBBS

Department of Pediatric Urology, Children's Hospital Colorado, 13123 East, 16th Ave Box 463, Aurora, Colorado 80045

ARTICLE INFO

Keywords:
Cloaca
Long-term urologic outcomes
Continence
Chronic kidney disease
Chronic renal insufficiency failure

ABSTRACT

Cloacal anomalies are the most complex and severe form of congenital anorectal malformations (ARM) and urogenital malformations, and it has been well documented that increased severity of ARM leads to worse outcomes. While short-term data on persistent cloaca are available, a paucity of data on long-term outcomes exists, largely because of a lack of uniform terminology, inclusion with other ARM and evolution of the operative technique. On comprehensive review of the published literature on long-term urological outcomes in patients with cloacal anomalies, we found a significant risk of chronic kidney disease and incontinence, however, with improvements in surgical technique, outcomes have improved. Continence often requires intermittent catheterization and in some cases, bladder augmentation. The complexity of cloacal malformations and associated anomalies make long-term multidisciplinary follow-up imperative.

Introduction

Cloacal anomalies are rare congenital malformations that serve as the most severe and complex form of anorectal (ARM) and urogenital malformation, accounting for 10% of all ARM. Patients born with persistent cloaca are uniformly female with the rectum, urethra, and vagina draining into a common channel that opens to the perineum; therefore, the clinical presentation is that of a female with a single perineal opening. Cloacal anomalies are thought to occur worldwide at a rate of 1:50,000 births[1]; however, the true incidence is difficult to predict because of differences in classification and inclusion with other ARM.

Within the past century, major advancement has occurred in the surgical correction of cloacal anomalies, with separate correction of the ARM and urologic intervention modified to a comprehensive repair.[1] Continued evolution of the technique led to significant improvement in 1982 with the posterior sagittal anorectovagino-urethroplasty (PSARVUP).[2] In 1997, the surgical technique was simplified by adopting the total urogenital mobilization (TUM), that alleviated the challenging and time consuming separation of the urethra and vagina.[3] Regardless of the approach, the goals of therapy remain steadfast: separate drainage of fecal and urinary streams with protection of renal function, while achieving social continence of both urine and feces. Another important goal is the repair of the vagina and the preservation of internal genitalia, to allow the patient to become sexually active and, if possible, to be fertile.

Cloacal malformations are often diagnosed on postnatal exam; however, there are certain prenatal findings that can help to identify the diagnosis. Early knowledge can assist with setting parental expectations and planning of the postnatal course. Cloacal anomalies should be considered in any female fetus where prenatal ultrasound reveals bilateral hydronephrosis with a poorly visualized bladder and cystic structure arising from the pelvis.[4]

Early intervention within the first few days of life is warranted because of need for fecal diversion, as well as evaluation and decompression of urinary obstruction. Hydrocolpos is found in 30% of patients and often the culprit for urinary obstruction because of urine preferentially collecting in the dilated vagina via the confluence. The distended vagina compresses the trigone and ureters, causing ureteral and bladder outlet obstruction, ultimately endangering renal function.[5]

The clinical presentation of cloacal anomalies is on a spectrum of severity and depends on the level of embryologic arrest in early development of the urorectal septum. With a high rectal fistula, mullerian ducts are unable to fuse leading to anomalies such as vaginal duplication and uterus didelphus,[6] commonly associated with cloacal anomalies.[7] Various other associations are common in cloacal anomalies such as VACTERL, but most notably spinal and urological abnormalities including solitary kidney, ectopic and

Abbreviations: ARM, anorectal malformation; CIC, clean intermittent catheterization; CKD, chronic kidney disease; CRF, chronic renal failure; GFR, glomerular filtration rate; PSARVUP, posterior sagittal anorectovagino-urethroplasty; VUR, vesicoureteral reflux; TUM, total urogenital mobilization.

* Corresponding author.
E-mail address: brian.caldwell@childrenscolorado.org (B.T. Caldwell).

http://dx.doi.org/10.1053/j.sempedsurg.2015.11.010
1055-8586/

fused kidneys, renal dysplasia, hydronephrosis, and vesicoureteral reflux (VUR).[8–10] The severity of the associated condition, in addition to the length of the common channel, can be predictors of overall patient outcome.[5]

Improved management during childhood has led to increased cloacal patient longevity well into adulthood, with a near normal life expectancy.[11] Although the short-term outcomes for cloacal patients are well-documented, long-term outcome data remains sparse. This review will discuss the available long-term urological outcome data for cloacal anomalies, especially focusing on continence and renal function.

Challenges of the literature

When reviewing a rare entity such as cloacal anomalies, analysis of the data can be challenging for several reasons: small numbers, lack of uniform terminology, and definitions, variable disease severity, inclusion of cloaca with similar but distinct anomalies, and evolution of the intervention technique. With an incidence of one in 50,000 births worldwide,[1] even specialized centers have relatively low numbers of cloacal patients. To compound the problem, much previous data was combined with other ARM[11] or with disorders utilizing similar surgical approaches, such as TUM for urogenital sinus.[1,12,13] Although this was done to bolster the power of individual studies, meaningful conclusions are difficult to extract from these combined data.

With such small numbers of cloacal patients treated at individual institutions, the only foreseeable way to achieve viable results is by using the same verbiage and collecting the same data across institutions for comparison. Complexity of the anatomical anomaly has been shown to have substantial effect on outcomes[5]; however, a consistent standard is not represented in previous studies. Recent classification of ARM has provided some direction to this, with persistent cloaca divided into those with common channel >3 or <3 cm.[14] Other studies have used high/low defect or short/long channel[11]; whereas, some refute this classification because it over-represents the channel length in patients with a phallic phenotype.[8]

Bladder outcomes are not solely a result of channel length or operative technique; on the contrary, spinal abnormalities have a significant association with cloaca and bladder dynamics.[15] Most of the studies to date have not presented information about the presence or type of spinal abnormality associated with cloaca anomalies. Directly related to this is the lack of consistent definition of continence, a common problem among other disease entities as well. Social continence seems to mean being dry during the day by means of spontaneous voiding or clean intermittent catheterization[16]; however, urinary continence itself has been defined on a continuum from completely dry all the time to 4, 3, or 2 h dry intervals.[8,13,17]

Likewise, chronic kidney disease (CKD) outcomes suffer from lack of uniformity. Glomerular filtration rate (GFR) is the most accurate measure of renal function and can be estimated from nuclear renal scan and in some cases, cystatin C.[9,10,18] Due to the retrospective nature of most studies, surrogate measures are utilized such as age adjusted serum creatinine or serial serum creatinine; however, these can underestimate the degree of CKD because of muscle mass differential in patients with spinal anomalies.[8]

As with many complex disorders, methods of repair continuously evolve, and the same is true for cloacal anomalies. Much of the long-term data available come from the early experience with newer techniques for reconstruction (TUM) or are combined with older operative techniques. This complicates true evaluation of long-term outcomes. In the ever evolving care of cloacal patients collaboration, maintaining consistent terminology, and collecting similar outcome data across centers will be especially important for meaningful future studies.

Upper urinary tract

Associated anomalies

A large proportion of cloacal patients have associated general urinary tract (83%)[9] and renal abnormalities (61–68%),[2,9] often negatively impacting bladder and renal function. In several of the largest series on cloaca, the structural renal anomalies most commonly identified were solitary kidney (13–26%), renal dysplasia (13–27%), ectopic kidney (8–14%), duplex kidneys (4–9%), hydronephrosis (16–58%), and ureteropelvic junction obstruction (5–8%).[2,8–10]

To complicate the discussion further, vesicoureteral reflux (VUR) is found in 41–57% of cloacal patients.[2,8–10] In recent non-cloaca literature, controversy exists regarding the role of VUR in renal deterioration and therefore surgical correction is controversial as well. Whereas several studies commented on the prevalence of VUR, only one reported the grades of VUR in five patient series: two with grade II, two with grade III and one with grade IV.[10] Quoted rates of VUR requiring repair are between 50 and 80%, yet only one study defined the requirement: severe VUR with frequent febrile UTI's despite antibiotic prophylaxis.[6]

Independent predictors of chronic renal failure were found to be VUR (23/34), dysplasia (14/17), and new renal scarring (15/19) in a 20-year retrospective study of patients with cloaca ($p < 0.05$). Additionally, all eight patients with solitary kidney had chronic renal failure (CRF).[9] It is complicated to assess long-term renal function specifically associated with a cloacal anomaly in light of these confounding intrinsic renal abnormalities that may variably predispose the patient to renal deterioration.

Beyond the intrinsic renal dysfunction associated with structural abnormalities, hydrocolpos is present in up to 30% of cloacal patients. Despite being continuous with the urethra, the vagina does not evacuate the trapped urine and debris spontaneously. An obstructed vagina can lead to infection or extrinsic compression of the trigone and bladder. Several options exist for genitourinary decompression including vesicostomy, vaginostomy, ureterostomy, nephrostomy, and clean intermittent catheterization of the common channel.[5] Although there is not a consensus on the best management of hydrocolpos and the resulting uropathy, a recent retrospective cohort study of 25 cloacal patients showed preservation of renal function with clean intermittent catheterization comparable to early surgical decompression.[19]

Renal function outcomes

Although renal function is dependent on a variety of factors including the bladder status, intrinsic abnormalities and other associated anomalies, several studies have evaluated the overall long-term effect of cloacal anomalies on renal function. In the Great Ormond Street experience with 64 cloaca patients at a mean age of 11.2, fully half the patients had chronic renal failure with glomerular filtration rate (GFR) less than 80 ml/min per 1.73 m^2. The actual mean was much lower at 41 ml/min per 1.73 m^2. Included were 17% of patients who progressed to end stage renal disease, 6% who underwent renal transplantation and 6% who died of end stage renal disease. Based on clinical parameters, only 60% of the patients had GFR calculated with the remainder assumed normal, so the actual rate of renal failure may have been higher.[9]

In patients with posterior urethral valves, nadir serum creatinine has been shown to be a good prognostic indicator[20];