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Congenital cloaca: Long-term follow-up results with emphasis on outcomes beyond childhood

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ABSTRACT

Persistent cloaca remains a challenge for pediatric surgeons and urologists. Reconstructive surgery of cloacal malformations aims to repair the anorectum, urinary tract, and genital organs, and achieve fecal and urinary continence as well as functional genital tract capable for sexual activity and pregnancy. Unfortunately, even in most experienced hands these goals are not always accomplished. The endpoint of the functional development of bowel, urinary, and genital functions is the completion of patient's growth and sexual maturity. It is unlikely that there will be any significant functional improvement beyond these time points. About half of the patients with cloaca attain fecal and urinary continence after their growth period. The remaining half stay clean or dry by adjunctive measures such as bowel management by enemas or ACE channel, and continent urinary diversion or intermittent catheterization. Problems related to genital organs such as obstructed menstruations, amenorrhea, and introitus stenosis are common and often require secondary surgery. Encouragingly, most adolescent and adult patients are capable of sexual life despite often complex vaginal primary and secondary reconstructions. Also, cloacal malformation does not preclude pregnancies, although they still are quite rare. Pregnant patients with cloaca require special care and follow-up to guarantee uncomplicated pregnancy and preservation of anorectal and urinary functions. Cesarean section is recommended for cloaca patients. The self-reported quality of life of cloaca patients appears to be comparable to that of female patients with less complex anorectal malformations.

Repair of a congenital cloaca is a major challenge for pediatric surgeons. The condition is rare; reported incidences range around 1:20,000–50,000, therefore, most pediatric surgeons encounter only few cases of cloaca during their entire career. The management of cloacal malformations was revolutionized by the posterior sagittal approach (PSARP) and total urogenital mobilization procedures of Alberto Peña.[1,2] This approach enabled full reconstruction of the urinary tract, genital tract, and rectum and anus during one operative session. Before the PSARP era, many patients with cloaca initially underwent only anorectal reconstruction and the remaining urogenital sinus was left untouched or was repaired later. The outcomes of these operations were usually dismal.[3]

In the literature, there is scarcity of reports on long-term follow-up of cloaca. Furthermore, most reports present only small number of cases or the outcomes are displayed as a part of general population of anorectal malformations. No controlled or prospective studies on long-term functional outcomes are available, all reports in the literature are retrospective institutional reviews. Only a few reports include more than 100 patients.[4,5] Especially, there is very little information about functional status and quality of life of cloaca patients after the development of puberty and beyond their growth period.

The aim of this review is to summarize the reported late outcomes in patients with a congenital cloaca, with the focus on those who are at their puberty or beyond that. Moreover, the reported outcomes from the literature are compared with the author's own patients of a similar age range. Of a total of author's 89 patients with cloaca, 27 are adolescent or adults.

The importance of analyzing the outcomes at this age lies in the fact that at puberty or beyond that the development of anorectal and urinary tract functions are likely to approach its endpoint. The development of gynecological organs is also completed and the outcomes and prospect of sexual life and fertility can be assessed.

Anorectal function

Most of the reports describing anorectal function in patients with cloaca present the outcomes by pooling all age groups. The

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Table 1
Bowel function in adolescents and adults with cloaca.

	Continent (%)	Bowel management/ACE (%)	Medication (%)	Stoma (%)	Soiling (%)
Davies (2010)[12] ($n = 15$)	27	33	13	27	n.r
Couchman et al.[13] ($n = 19$)	58	5	n.r	37	n.r
Rintala (2015) ($n = 27$)	52	22	0	11	15

n.r, not reported.

usual lower cut-off age is 3–4 years, as these are considered the time points when children usually are toilet-trained and evaluable for bowel function.

It is very clear that the bowel function in a preschool-aged child is different from that of a patient who has gone through puberty. A certain degree of improvement in the bowel function in patients with anorectal malformations has been reported by numerous authors.[6–8] It is most likely that this holds true also in patients with cloaca. The small number of patients in almost every cloaca series have precluded the evaluation of the role of aging in relation to bowel functional outcomes.

Versteegh et al.[9] have recently published a systematic review on the functional outcomes in cloaca patients. This collective review has pooled the data on anorectal function for a total of 263 patients. The length of follow-up in this pooled patient series ranged from 5 months to 31 years. The largest series[10] included in the report had 156 patients with pertinent data on bowel function. In this series, the patients were older than 3 years. This systematic review reported bowel function by using Krickenbeck criteria.[11] None of the studies included in this systematic review were prospective, the data collection was a retrospective case note review in all.

Voluntary bowel movements were reported in 108 of the 188 patients (57%) in whom this factor of bowel function was analyzed. The presence of voluntary bowel movements ranged between 41% and 60%. Only one study in this systematic review reported patients with total continence. Peña et al. found 26 out of 156 (17%) patients to be totally continent at last follow-up.

More or less total fecal incontinence was found in 33% of the patients (range in the studies 14–41%) in this systematic review. The actual severity of incontinence was not graded in the articles included in the systematic review. The reports also did not describe any criteria for continence or incontinence. It is still obvious that many, if not most, of these patients required some sort of bowel management to stay clean. The reported bowel management modalities included regular enemas and formation of an ACE stoma. In 17% of the patients (range in the studies 5–22%) the quality of bowel control was so poor that the patients chose a permanent colostomy. Some degree of fecal soiling was reported in 71% of the patients (range in the studies 14–83%). Again, the severity of soiling was not graded. The consequences of soiling was not analyzed in the present reports.

Constipation was a very common problem in patients with cloaca. Overall, more than half of the patients suffered from constipation requiring medical management or enema program. The incidence of constipation ranged between 30% and 88%. Constipation was managed medically by oral laxatives (29% of the patients) or by an enema bowel management program.

Some kind of bowel management, either by retrograde bowel washouts or by antegrade washouts through an ACE appendicostomy were used on average by 38% of the patients. The need for bowel management ranged between 14% and 65% in the reports of the systematic review. The indications for bowel management were fecal incontinence in most cases or intractable constipation.

The reported bowel function outcomes in adolescents and adults are somewhat different than in the series that have pooled functional data from all age groups (Table 1). However, the data on bowel function in patients beyond their childhood is extremely scarce. Davies et al.[12] had 15 cloaca patients in their series of patients with anorectal malformations and a mean follow-up of 26 years. A total of 4 (27%) of their 15 patients were spontaneously continent, five (33%) had an ACE or used retrograde washouts, two (13%) used medication to augment continence, and four (27%) had a permanent stoma. Couchman et al.[13] identified 19 cloaca patients with a mean follow-up of 22 years. Fecal continence was reported by 11 (58%) of these patients. The remaining eight patients had an ileostomy or colostomy (7, 37%), or ACE conduit (1, 5%) for bowel evacuation. In the author's personal series, 27 patients are adolescent or adults. The mean age of the patients is 23 years (range: 13–40 years). Of these 27 patients, 14 (52%) have developed spontaneous continence and do not require any medications or protective aids. Six patients (22%) have an ACE formation for bowel emptying. Four patients (15%) suffer from soiling that occasionally requires use of protective pads. Three patients,[11] two with a complete sacral agenesis and another with a poorly functioning ACE conduit, have opted for a permanent stoma formation.

In the few studies concerning adolescent or adult patients with a cloaca, none have been reported to suffer from constipation. In the author's series this was also the case, none of the 27 patients reported constipation nor used any laxatives to augment bowel emptying.

It appears that the fecal continence rate in adolescents and adults is more favorable than in children. Several factors are likely to contribute to this finding. Adolescents and adults are maximally adapted to their initially deficient anorectal function and have usually developed strategies to stay clean. These include dietary modifications, optimized stooling habits, and sometimes, use of antipropulsive medication. The absence of constipation also promotes better continence because the risk of overflow incontinence is eliminated. In childhood, overflow incontinence is probably the most common cause of fecal soiling in these patients.

Urinary tract function

It is not unexpected that patients with cloaca commonly have urinary tract dysfunction. Urological as well as spinal and spinal cord anomalies are very common in cloaca patients. Urodynamic studies have revealed that a significant proportion of patients with anorectal malformations have neurovesical dysfunction prior to surgery.[14,15] This is especially true in patients with a cloaca.[16] Moreover, the extensive surgery required to repair a cloacal malformation can deteriorate the bladder function even more than in other patients with anorectal malformations.[16]

In the systematic review of Versteegh et al.[9] concerning cloaca patients of all ages, urinary tract function was reported in 332 patients. This review pooled patients from nine studies. Spontaneous voiding was achieved on an average by 46% (138/299) of the assessed patients. In the studies included in the systematic review, the percentage of patients with spontaneous voiding ranged from 22% to 54%. Intermittent catheterization to empty the bladder was reported by 42% (141/332) of the patients (range in the studies

Table 2
Urinary function in adolescent and adults with cloaca.

	Continent (%)	Continent stoma/CIC (%)	Incontinence (%)	Renal failure (%)
Davies (2010)[12] ($n = 15$)	20	n.r	80	n.r
Couchman et al.[13] ($n = 19$)	47	53	n.r	21
Rintala (2015) ($n = 27$)	63	19	15	11

n.r, not reported.

12–100%) and 22% (55/237) had undergone urinary diversion (range in the studies 18–27%). Urinary incontinence could be evaluated in 166 patients, 38 of these (23%) (range in the studies 9–41%) suffered from some degree of incontinence.

The reported urinary tract function in the general cloaca population, including patients of all age groups, appears to be quite similar as the anorectal function. Major defects in anorectal and urinary tract functions are seen in more than half of the patients. The bowel functional outcomes in adolescent and adult patients were markedly better than those in the series including patients of all ages. In terms of urinary tract, the functional outcome does not appear to be better in older age groups (Table 2). Davies et al.[12] found that only 20% of the adult or adolescent cloaca patients were fully continent for urine. The outcome was worse in patients that originally had had a long common channel. Couchman et al.[13] reported urinary tract function in 19 adolescent or adult patients with cloaca. Nine of 19 patients (47%) were able to void spontaneously; the remaining 10 required catheterization via the urethra or a continent Mitrofanoff vesicostomy. In the author's own series of 27 adolescent or adult patients, 17 (63%) voided spontaneously and were continent for urine. Three patients had undergone bladder augmentation and bladder neck closure. These three (11%) emptied the bladder via a Mitrofanoff channel. Three patients (11%) stayed dry by intermittent catheterizations and four (15%) had slight daily wetting that required the use protective pads.

There is very little data on renal outcomes of cloaca patients in the literature. Warne et al.[17] reported that 50% of the 64 patients with cloaca (mean age at the time of the study 11 years) developed chronic renal failure measured by glomerular filtration rate. Of these, 11 progressed to end-stage renal failure. Of these 11, four underwent renal transplantation and four died of chronic renal failure. Braga et al.[18] followed 12 patients with cloaca and found that 9 of the 12 patients (75%) had abnormally low glomerular filtration rate. In both these studies, the risk of chronic renal failure was higher in patients with significant urological malformations or presence of a single kidney. In the report of Couchman et al.,[13] 4 of the 19 (21%) adolescent or adult cloaca patients had impairment of renal function, one of these had undergone renal transplantation. In the author's series of 27 adolescent or adult patients with cloaca, only three (11%) have any evidence of chronic renal impairment. All the others have normal serum creatinine levels and radiologically stable upper urinary tracts.

It is evident that the patients with a cloaca are a special group in terms of need to continuously monitor the renal function. Associated urogenital anomalies that are present in most cloaca patients and presence of a long common channel increase the risk of renal impairment. Another risk factor appears to be the length of common channel[18]; the patients with a common channel length longer that 3 cm had worse renal outcome than those with a shorter common channel. The deterioration of renal function is the main problem that threatens the life of patients with a cloaca. Every effort should be given to treat the preventable causes of renal deterioration, i.e., correct obstructive urinary tract anomalies, prevent urinary tract infections, and treat bladder dysfunction. Regular monitoring of renal function is also warranted, probably throughout the patient's life. The monitoring should include ultrasound imaging of the kidneys and urinary tract.

Gynecological function

All patients with cloacal anomalies have abnormal genital tract. Approximately 40% of patients have duplicated Mullerian system with two vaginas and uteri. At birth, about 30% of the patients present with hydrocolpos[5]; most of these have also double vaginas and uteri. In some cases, the Mullerian duplication is not symmetrical; in these, one of the systems may be obstructed and sometimes very small, and may go undetected during primary surgery. The implication may be that the patient at the time of the puberty may accumulate menstrual blood to the obstructed system. This may cause severe cyclic abdominal pains with monthly exacerbations and retrograde flow of menstrual blood into peritoneal cavity. Following even perfect and uncomplicated genital repair during the primary cloacal reconstruction, several problems may remain. These do not became acute during childhood but very commonly require secondary surgical intervention following the onset of puberty.

In the literature, the reports on long-term gynecological outcomes in patients with a cloaca are even scarcer than those concerning anorectal or urinary tract function (Table 3). In the systematic review of Versteegh et al.,[9] gynecological function was reported by 71 patients that were pooled from three reports. Normal menstruations were found in only 35% of the patients. Obstructed menstruations were a common finding affecting 38% (24/63) of the patients. The causes of obstruction have been variable. Some patients had a stenosis in the persistent urogenital sinus; these patients had not undergone any genital tract reconstruction.[19] Another group are patients that had undergone genital reconstruction but still developed partial or complete obstruction of the menstrual flow.[20] Almost all of these patients had duplicated Mullerian systems that was mostly asymmetric. Amenorrhea

Table 3
Genital function in adolescent and adults with cloaca.

	Normal menstruation (%)	Obstructed menstruation (%)	Amenorrhea (%)	Abnormal menstruation (%)
Levitt et al.[20] ($n = 2$ 2)	32	41	27	n.r
Warne et al.[19] ($n = 41$)	32	37	24	7
Rintala (2015) ($n = 27$)	60	11	26	n.r

n.r, not reported.

affects approximately 25% of the patients with cloaca. Most patients with amenorrhea have absent or vestigial uterus. Most of these patients have also vaginal agenesis or very small blind-ending vaginas. In line with findings in the systematic review of Versteegh et al.,[9] Couchman et al.[13] found obstructed menstruation in 5 of their 19 (26%) adolescent or adults patients with a cloaca. In the author's own series of 27 adolescent or adult patients, 17 (60%) had normal mentstruations, four (15%) had amenorrhea due to Mullerian agenesis and three (11%) adolescents for unknown reasons despite otherwise normal pubertal development, and three (11%) had had obstructed menstruations.

The obstructed genital organs usually require surgical management that sometimes needs to be performed emergently. The usual presentation is a cystic abdominal mass. The anatomy is usually clearly delineated by ultrasonography and MRI imaging. Typical procedures for patients with obstructed double systems include resection of the obstructed hemi uteri and adnexa, and eradication of the abdominal collections of old blood and possible endometriosis.[19,20] Patients with retained urogenital sinus require usually pull-through vaginoplasty.

In addition to obstructions in menstrual flow, cloaca patients have commonly other cystic problems in the internal genital organs. Five (19%) of the author's 27 adult or adolescent cloaca patients have required surgical interventions for large cystic collections that involved uteri and adnexal tubes. These cyst were not related to menstruations and were histologically benign paraovarial cysts. All these operations could be performed laparoscopically.

The vaginoplasty that was performed at the time of the primary reconstruction by posterior sagittal approach or total urogenital mobilization requires revision in a significant percentage of patients to allow normal sexual activity. Couchman et al.[13] reported that 56% of the patients who underwent vaginal reconstruction in infancy required a secondary or even tertiary surgery. The authors questioned the rationale of total urogenital reconstruction during infancy and suggested that genital reconstruction could be delayed until there is full clarity of the anatomy of Mullerian structures. However, most pediatric surgeons and urologists who have experience in reconstructions of cloacas strongly advise full reconstruction in infancy.[4,5,10] It is logical to reconstruct the vagina at the same time as the rectum and urinary tract. The findings in the report of Warne et al.[19] support this as the patients with early vaginal repair had a stricture rate of 15% which compared favorably with the patients having postpubertal vaginoplasty and a stricture rate of 42%.

In the author's institution, adolescent patients with cloaca are offered definitive reconstruction of the vagina at the age of 14–16 years in order enable sexual activity and intercourse. Of 27 adolescent and adult patients, secondary vaginal procedures were required in 11 of the 22 patients (50%) who had elected the definitive vaginal repair. Of the 27 patients, three suffered from moderate neurological impairment, two of these were wheelchair-bound. The vagina in these three was non-obstructed but may later require introitusplasty. Two patients chose to delay the evaluation of the adequacy of the vagina for intercourse to a later stage. Of the 11 secondary vaginal repairs, eight were simple introitusplasties that in five were followed by a dilatation program. Two patients required a redo-colovaginoplasty due to severely strictured distal vagina following ileal vaginoplasty. One patient required a redo total urogenital mobilization to create an adequate vaginal introitus.

The rate of sexual activity in patients with cloaca has been reported only in three studies (Table 4). A total of 24 of Hendren's 154 patients[4] were adults and 17 (71%) of these had had sexual intercourse. Warne et al.[19] found that 12 of their 21 patients (57%) (age range: 17–34 years) had been sexually active. Adequate vagina for sexual intercourse as assessed by a gynecologist was found in 18 of the 21 patients (86%). In the study of Couchman et al.,[13] 8 of the 19 patients (42%) (age range: 13–35 years) were reported to be sexually active, of whom one had experienced difficulties in penetration. Of the author's 27 patients, (age range: 13–30 years) with a cloaca, 14 (52%) reported sexual activity. Two of these occasionally had problems with penetration requiring lubrication. Both of these patients had a very narrow pelvis due to near-total sacral agenesis. Another patient occasionally had painful intercourse due to a scar neuroma at the vaginal introitus. This responded well to local injection of long-acting anesthetic (bupivacaine). The rates of sexual activity in cloaca patients are somewhat less than in other females with anorectal malformation. Schmidt et al.[21] found that 65% of the females with anorectal malformations had active sexual life. In the report of van den Hondel et al.,[22] 87% of females with anorectal malformation were sexually active. In both these studies, the patients were older (all older than 18 years) than in reports concerning cloaca patients; this may influence the rate of sexual activity. Anyway, it is encouraging to find that a vagina capable for sexual intercourse has been reconstructed in most young females with a cloaca and about half or more of them are sexually active.

The fertility of cloaca patients is unclear. In the literature, reports concerning fertility and pregnancies in patients with a cloaca are largely lacking. Of Hendren's[4] 24 adult patients, six had gone successfully through pregnancies. Five of these delivered by cesarean section and one vaginally. In the report of Couchman et al.,[13] three of the eight patients with sexual activity were attempting to conceive with assisted conception methods and one patient had a complex preterm delivery. Of the author's 14 sexually active patients, three had delivered healthy babies by cesarean section. Although vaginal delivery is probably possible in many patients with a cloaca, cesarean section is usually recommended. All cloaca patients have undergone some sort of repair of the vagina that may be damaged following vagina delivery. Moreover anal sphincters and reconstructed urethra and bladder may suffer functionally from significant stretch caused by vaginal delivery.[23]

Quality of life (QoL)

Female patients and those with multiple associated anomalies have been reported to suffer from poorer quality of life than male patients and those without significant associated malformations.

Table 4
Sexual activity and fertility in adolescents and adults with cloaca.

	Sexual activity (%)	Adequate vagina for intercourse (%)	Problems in intercourse (%)	Number of patients with offspring
Hendren[4] ($n = 24$)	71	n.r	n.r	6
Warne et al.[19] ($n = 21$)	57	86	n.r	0
Couchman et al.[13] ($n = 19$)	42	n.r	13	1
Rintala (2015) ($n = 27$)	52	81	21	3

n.r, not reported.

Practically all cloaca patients have associated malformation and, obviously, are all females. Therefore, it would be logical to expect cloaca patients to have lower QOL than other female patients with anorectal malformations. The QoL of cloaca patients have been studied, in particular, only by Versteegh et al.[24] The QoL of children and adolescents with cloaca was compared with that of patients with rectoperineal and rectovestibular fistula using a standardized and validated scoring instrument for QoL evaluation (PedsQL 4.0 inventory). The patients with a cloaca and females with less complex anorectal malformations reported similar QoL. The reported QoL scoring did not differ significantly from the reference values obtained from a Dutch population with similar age distribution. On the other hand, parents of patients with cloaca reported more problems on several psychosocial domains compared with the healthy children and adolescents. There are no studies that have addressed the Qol of adult patients with cloaca.

Conclusions

Persistent cloaca is still a challenge for pediatric surgeons and urologists. Despite optimal operations that are performed by very experienced surgeons, significant functional disturbances remain in many adolescent or adult patients with cloaca. Approximately, half of the patients gain spontaneous fecal continence as they go through childhood. The remaining half require adjunctive measures to stay clean. Adolescents and adults report similar spontaneous urinary continence rates; the rest of the patients stay dry mainly by continent urinary stomas or intermittent catheterizations. Genital tract problems are common in puberty, many patients require secondary procedures to establish patent genital tract that enables uncomplicated menstruations and sexual life.

Encouragingly, the majority of cloaca patients who have gone through puberty are capable for normal sexual life. Persistent cloaca does not preclude pregnancies and offspring, but special measures are often required to attain conceivement and to guarantee that the functional status of the cloaca patient is not jeopardized by pregnancy. Self-reported quality of life of cloaca patients appears to be surprisingly good, but further studies are required to confirm these findings. Specialized centers with multidisciplinary services are essential for the care of cloaca patients beyond their childhood also.

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