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Phalloplasty and Urethroplasty in Children With Penile Agenesis: Preliminary Report

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Purpose: Female gender has been assigned to 46,XY newborns affected by aphallia, possibly resulting in subsequent gender dysphoria. Prenatal and postnatal effects of the androgens on the brain and sexual orientation cannot be modified later. Therefore, patients affected by aphallia should be raised as males. Because definitive forearm flap phalloplasty is generally not recommended before puberty, we performed a preliminary penile reconstruction during childhood in 4 patients.

Materials and Methods: Four patients with aphallia who had no sex reassignment at birth were treated at age 9, 17 and 36 months, and 12 years in a single operation. The urethral channel was dissected through an anterior-sagittal-transano-rectal approach, and then phalloplasty and urethroplasty were carried out using an abdominal skin flap and a bladder/buccal mucosa free graft.

Results: Immediate postoperative outcome was excellent in all the patients. One patient had excellent functional and cosmetic results at 5 years, while 2 had a partial dorsal urethral dehiscence resulting in an epispadiac urethra at 2 years, and 1 had necrosis of the distal urethra and was voiding through a scrotal urethrostomy at 9 months postoperatively. Phalloplasty survived and provided an adequate male appearance in all patients.

Conclusions: Opposite gender should not be assigned in patients affected by penile agenesis, who are better raised according to their karyotype and hormonal production. Definitive phalloplasty in adults may achieve good results. Nevertheless, this procedure is generally performed in postpubertal boys and it is not easily available everywhere. Therefore, we believe that social and psychological concerns justified this type of phalloplasty as a palliative preliminary procedure in 3 of our patients. In those countries where definitive forearm phalloplasty is not available our method may also be justified in older children (as in 1 of our patients) as an attempt at a definitive procedure.

Key Words: penis; urogenital abnormalities; urologic surgical procedures, male; reconstructive surgical procedures

The penis is an extremely complex organ, with urinary, fertility and psychosexual roles. Penile agenesis (congenital absence of the penis or aphallia) in an otherwise normal 46,XY male with functioning testes is a rare malformation that is reported to occur in 1 in 30 million births.^{1,2} Penile agenesis is a devastating condition for the patient and parents, and one of the most demanding and challenging problems for urologists.

Historically, gender reassignment was considered to be the most appropriate choice for these patients.^{3,4} Although adequate outcomes have been reported,⁵ in a long-term followup the majority of female raised patients with penile agenesis demonstrated a marked male-typical shift in psychosocial and psychosexual development,^{6,7} with most declaring themselves to be male. Psychosexual development, including gender identity, in genetic and hormonally male neonates seems to be strongly influenced by prenatal and early postnatal androgen exposure. Gender identity can be minimally modified by the environment and by endocrine treatments.^{6,7} Therefore, patients with aphallia should be

raised as males despite the particularly limited experience with phalloplasty in patients born with severe penile malformations, and the fact that definitive penile reconstruction is generally not recommended before age 15 years and is available in only a few highly qualified places worldwide.

CASE HISTORIES

In 4 patients affected by penile agenesis male gender was assigned. All patients presented with 46,XY normal male karyotype, a normal scrotum and palpable normal testes (undescended in patient 2). The urethra entered the distal anterior rectal wall in all cases. At referral 3 patients had not undergone surgery, and 1 had bilateral ureterostomies.

Following a complete medical evaluation, adequate elucidation of the aims, limits and magnitude of the operation and postoperative treatment, and proper bowel preparation penile reconstruction and urethroplasty were performed for each patient in a single procedure at age 9 months (Jeddah, Saudi Arabia, May 2001), 17 months (Bologna, Italy, February 2004), 3 years (Sao Paulo, Brazil, November 2005) and 12 years (Sao Paulo, Brazil, December 2005). One of us (RDC) took part in all the operations.

The procedure consisted of urethral division from the rectum (from the scarred perineal area close to the anus in patient 3) through an ASTRA with the patient in the prone

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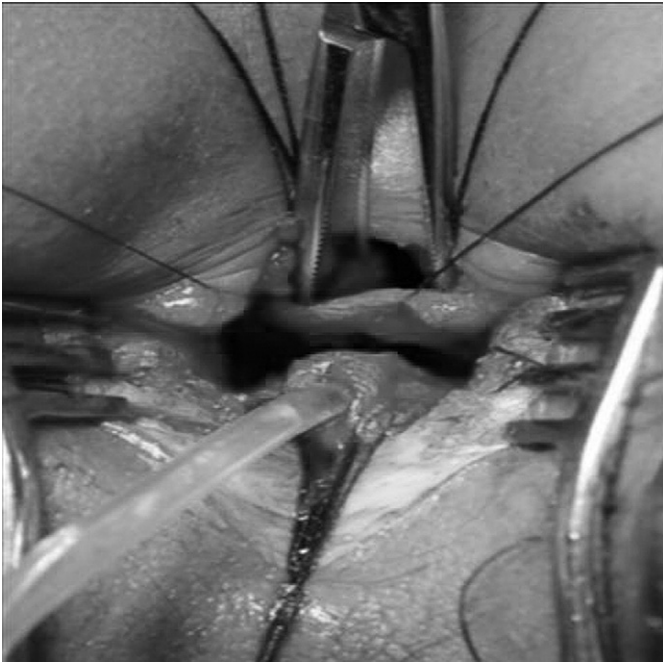


FIG. 1. Anterior-sagittal-transanorectal approach. First, with patient in prone knee-chest position, anterior rectal wall is divided in midline until urethral opening is reached. Next, catheter is passed in bladder and urethra is partially dissected off anterior rectal wall. Distal anorectal canal is then reconstructed.

knee-chest position (fig. 1).⁸ Phalloplasty and complete urethroplasty using a quadrangular abdominal skin flap (fig. 2) and bladder or buccal mucosa free graft were then performed with the patient in the supine lithotomy position (fig. 3). Skin flap dimensions were 4 cm wide \times 5 cm long in the 9-month-old patient, 5 \times 6 cm in the 17-month-old, 6 \times 7 cm in the 3-year-old and 9 \times 9 cm in the 12-year-old.

The new long distal urethra was created using bladder mucosa graft alone in 1 patient, a combination of bladder and buccal mucosa in 1, and buccal mucosa alone in 2. In patient 2 cavernous tissue was identified and saved for pos-



FIG. 2. Phalloplasty procedure. Quadrangular lower abdominal flap including skin and subcutaneous tissue is developed, as described previously.¹⁷ Dimensions of flap vary according to patient age.



FIG. 3. Urethroplasty procedure. New urethra is created tubularizing mucosa free graft (bladder mucosa alone in patient 1, bladder and labial mucosa in patient 2, and buccal mucosa only in patients 3 and 4). Mucosal tube is placed in space created in front of pubic branches, reaching abdominal skin flap distally and native urethra proximally. Neourethra is included within skin flap.

sible future tissue engineering and reuse. In this patient the ileoinguinal nerve was also identified and marked by metallic clip for possible use at the time of formal phalloplasty after puberty to facilitate new phallus sensation. In patient 3 the single left corpus cavernosum and glans penis were kept, and the glans was secured at the scrotal raphe to preserve erogenous sensation.

Penile size at the end of the procedures was 3, 4.5, 5.5 and 7.5 cm, respectively, in the youngest to oldest patients (fig. 4). A suprapubic catheter was placed in the bladder in 3 patients, with patient 3 excluded because of the presence of ureterostomies. The procedure lasted about 7 hours in each case. The immediate postoperative period was uneventful in all the patients. However, recovery differed significantly.

Case 1

This patient had classic aphallia and came from a poor family living in Saudi Arabia. No prenatal diagnosis had been made. There was no controversy regarding gender assignment at birth, ie the parents and physicians had no doubts about raising him as male. He presented at age 6 months, and the parents asked for penile reconstruction. He had normal kidneys and bladder, and was asymptomatic.

The urethral catheter was removed at 2 weeks postoperatively and VCUG was performed through the SPC, which was imprudently removed at the end of the evaluation. The child subsequently had progressive meatal and urethral stenosis, and consequently experienced trouble passing urine. He needed immediate insertion of a new percutaneous SPC.

Three weeks later removal of the distal urethra and scrotal urethroostomy were performed.

Five years after the first operation the patient is doing well, with no UTI and normal renal function. He is able to pass urine sitting on the toilet through the urethroostomy and has achieved normal bladder control. The new phallus is tender and in the correct position. It is 3.5 cm long and provides an acceptable male appearance. We are currently discussing with the father the opportunity to do a reoperation for cosmetic purposes only (scrotoplasty and glanuloplasty), or to perform a new distal urethroplasty.

Case 2

This patient had classic aphallia and came from a well educated Italian family. At prenatal ultrasound a right multicystic dysplastic kidney and megacystis were discovered. However, penile agenesis was not suspected, and, therefore, the parents were expecting a baby girl. Postnatally, many discussions and consultations were carried out before the final decision of male gender assignment. Nuclear magnetic resonance imaging confirmed the right multicystic dysplastic kidney, enlarged bladder and urethra ending in the rectum (fig. 5). The left upper urinary tract was normal and the presence of some cavernous tissue at the pubic branch was detected. The child presented with signs of urinary and fecal retention.

Three weeks postoperatively, with the patient under general anesthesia, the urethral catheter was removed, the SPC was replaced with a bottom device and VCUG was performed (fig. 6). He started passing urine through the new urethra. Three months later the suprapubic button was disconnected. The boy showed urinary retention secondary to the dysfunctional megacystis and not to urethral problems. A continent vesicostomy was created using a bladder



FIG. 4. Immediate final result in patient 4. Coronal sulcus is created incising skin and inserting skin free graft. Abdominal wall skin defect is covered by mobilizing and moving down upper portion of abdominal wall.



FIG. 5. In patient 2 preoperative nuclear magnetic resonance imaging reveals short urethra connecting large bladder with anorectal canal. Some cavernous tissue is present close to pubic branches.

wall flap with an umbilical stoma to allow clean intermittent catheterization.

At 2.5 years after the initial operation the patient is doing well. He has a good cosmetic outcome, no UTIs and normal renal function. From time to time he urinates normally through the new urethra, although he is currently on clean intermittent catheterization through the umbilical conti-

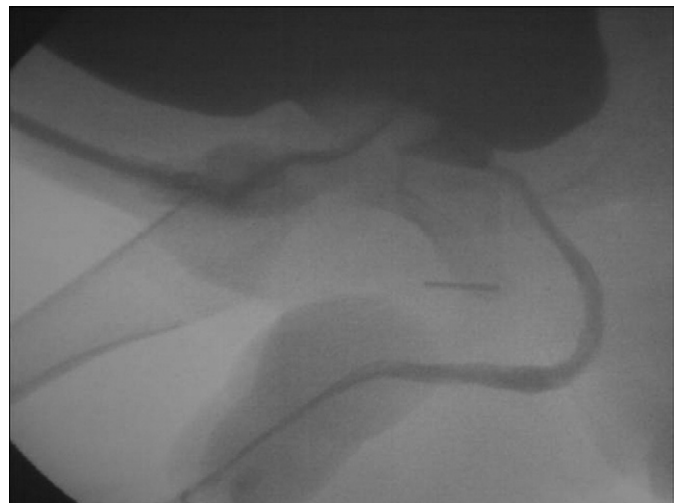


FIG. 6. Normal looking urethra is present on VCUG in patient 2 at 3 weeks postoperatively. Metallic clip marks ileoinguinal nerve.

ment stoma. Once daily, while the patient is sleeping, the parents calibrate the urethral meatus to prevent stenosis. Having problems with constipation, the patient was fully assessed and was observed to be affected with Hirschsprung disease, which was treated surgically. At the same time a redo granuloplasty was performed. Penile size is 4.5 cm (fig. 7).

Case 3

This patient from a Brazilian middle-class family had a variant of aphallia consisting of the presence of a minimal rudimentary microphallus with no penile urethra, a small single left corpus cavernosum and a small glans. No prenatal diagnosis was made and the parents were expecting a female. At birth, after strong familial conflicts, the decision to raise the patient as male was eventually made. At the age of 3 months he underwent a surgical procedure elsewhere, consisting of bilateral loop ureterostomies, which did not completely prevent urine from reaching the bladder, and an attempt to transfer the rectal urethra to the perineum, which resulted in a severe distal urethral stenosis causing recurrent right orchiepididymitis. The kidneys were normal, the partially defunctionalized bladder was not properly investigated because the severe urethral stricture did not allow catheter introduction, and the right testis was enlarged, irregular and hard.

At 3 weeks postoperatively VCUg was performed and the urethral catheter was replaced with the patient under general anesthesia. Two weeks later the catheter was definitively removed and the patient started passing the small amount of urine that reached the bladder because of the bilateral ureterostomies. Unfortunately, a partial dehiscence of the dorsal urethral sutures occurred, resulting in a stable but epispadiac urethral meatus.

At 9 months of followup the patient did not demonstrate any further complications. The new penis is in place with no wound problems, the epispadiac urethral meatus does not seem to have a tendency to contract, the new urethra is patent, and neither orchiepididymitis nor UTIs have been recorded. Penile size is 5 cm.

Case 4

This 12-year-old boy from a poor Brazilian family had classic aphallia. Normal kidneys and a good bladder with the ure-



FIG. 7. Long-term result in patient 2 at 18 months postoperatively

thra ending at the anterior anorectal wall were detected. He had reached normal bladder and bowel control at age 4 years. He did not present with any physical symptoms. He was a good student with excellent family support. However, he exhibited deep psychological problems related to the disease that were partially caused by the genuine, unfounded, but positive belief that surgery would eventually provide him with a normal penis. We extensively discussed with the child and his parents our operation, which was originally designed for infants as an early and palliative procedure. Considering the family social condition, and since a surgical team that was expert in adult phalloplasty was not available, we accepted the patient request for penile reconstruction.

A VCUg was obtained at 3 weeks postoperatively, and the urethral catheter was removed and replaced with the patient under general anesthesia. Two weeks later the catheter was definitively removed and the patient started voiding through the new urethra, and 2 months later the SPC was removed. Unfortunately, a partial dehiscence of the dorsal urethral sutures occurred.

At 9-month followup the outcome seems to be acceptable. The new phallus is in place, with no evidence of urethral or meatal stenosis. In addition, the patient is able to void through the epispadiac urethra, and he has normal bladder and bowel control. Penile size is 7.5 cm.

DISCUSSION

The classic picture of penile agenesis includes complete absence of the penis, a normal scrotum, a normal, frequently undescended, testis and 46,XY normal male karyotype. The most variable aspect of the disease is the urethral opening, which can be located in the perineal area, generally inside a foreskin appearing skin tag or in the anterior wall of the anorectal canal.¹⁻³

Penile agenesis is a morphological anomaly that is not caused by disorders of karyotype, gonadal development or sexual hormone metabolism.⁷ However, patients affected by this rare malformation usually present at birth with the same gender assignment dilemma as patients with ambiguous genitalia, and, therefore, can be included in the disorders of sexual differentiation. From this point of view congenital aphallia can be included with male cloacal exstrophy cases.⁶⁻⁹

In the past infants with congenital aphallia and those affected by cloacal exstrophy underwent gender reassignment surgery, including bilateral orchiectomy, urethral transposition, vaginal replacement and labial construction.¹⁰ This complex procedure may yield a good cosmetic outcome.⁵ However, frequently sexual dysphoria progresses with age, and eventually may culminate with patient initiated gender reassignment to male.⁶⁻⁹

Experimental data and several clinical observations have established the androgen role in male gender identity formation.¹¹ Prenatal and early postnatal androgen exposure deeply contributes to male psychosexual development and identity, which cannot be changed by neonatal castration, hormonal treatment, phenotypic adjustments or psychosexual manipulations.^{7,11,12} Therefore, male gender should be maintained in patients with penile agenesis.⁸⁻¹³

In the literature there are almost no data on total phallic construction in children with congenital malformations.

Conversely, phalloplasty for traumatic penile amputation has been reported in children and adolescents using different techniques.¹⁴ The most commonly used procedure for phalloplasty in postpubertal patients is the microvascular transfer radial forearm flap, particularly in the transsexual genetic female.¹⁵ This complex procedure requires a skilled team of physicians, including plastic surgeons, microvascular surgeons and urologists, and it is currently performed in only a few highly specialized centers that can guarantee high standard outcomes.

The main disadvantage of the radial forearm flap is its large and unpleasant donor site scar. Its use is generally not recommended in growing individuals.¹⁶ While we believe that this method should be considered a last step in patients born with aphallia, social and psychological concerns justify early palliative phalloplasty and urethroplasty in childhood. This is particularly true when our operation is performed in young children, as in patients 1 to 3. Our aim is to create male looking external genitalia, allowing children to void in a standing position from a urethral meatus placed at the tip of the penis. We also try to avoid any possible complication related to the rectal ending urethra. The use of our operation in older boys, as in patient 4, is more questionable. However, the relative simplicity of the procedure, which does not require a skilled team of plastic surgeons experienced in microvascular flap transfer, could make this operation an attractive option for the definitive treatment of penile agenesis in countries where these facilities are not readily available.

In our 4 patients we used ASTRA for separating the urethra from the rectum, a quadrangular lower abdominal skin flap for penile construction as previously described by Bettocchi et al,¹⁷ and bladder or oral mucosa free graft for urethral lengthening. The most original aspect of this procedure is the use of a free graft of mucosa for the urethroplasty, which is incorporated in the skin flap. This technique allows us to use the entire skin flap for the penis and glans penis replacement, avoiding all the complications related to long neourethras made of skin. On the other hand, it exposes patients to the well-known free graft tube problems that we experienced in our series, even if to a lesser extent than expected. Construction of an adequate urethral meatus at the tip of the new glans penis seems to be the most challenging part of the operation. We have the impression that the outcome might be better if the new urethra is constructed entirely of buccal mucosa.

In general, the procedure has had reasonable outcomes in our cases and it could be the right initial treatment for this rare and extreme genital malformation. Quadrangular lower abdominal flap is indicated when no scars are present on the lower abdomen, as in unoperated children affected by congenital absence of the penis.¹⁷ Until now, we have not investigated the feasibility of this procedure in cases of poor penile appearance in infants affected by the exstrophy-epispadias complex. When a midline scar is present a reasonable alternative may be the extended pedicle island groin flap described by Perovic.¹⁶ In view of the good results of phalloplasty, as compared to the relatively poor results of the urethroplasty, we are evaluating the opportunity to perform our method in 2 stages, or to include a temporary scrotal urethrostomy.

We wish to emphasize that we have reported only preliminary surgical outcomes in a limited number of patients. We do not know if our actual surgical method will be devel-

oped, improved or replaced in the future. However, we are convinced that retaining the male gender in 46,XY individuals born with aphallia is the right approach to this complex problem. We need a larger experience and longer followup to evaluate the potentialities of this method, taking into consideration the possibility of the new phallus growing with the child, reaching a satisfactory size and being able to accept the insertion of a penile prosthesis, which would allow erection and sexual intercourse.

Abbreviations and Acronyms

ASTRA	=	anterior-sagittal-transanorectal approach
SPC	=	suprapubic catheter
UTI	=	urinary tract infection
VCUG	=	voiding cystourethrogram

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EDITORIAL COMMENT

The authors present a series of 4 patients, all 46,XY males born with aphallia. Their approach in these children, who ranged from 9 months to 12 years old, was to do a single stage procedure that included phalloplasty and urethroplasty. The results of the phalloplasty were remarkably good in all 4 patients but the results of the urethroplasty are not as encouraging.

The concept of retaining the male gender in 46,XY individuals born with aphallia is certainly appealing. Their results with phalloplasty are convincing. Part of the problem with the urethroplasty is that they used free grafts of either buccal or bladder mucosa, which have little if any fixed vascular support structures to provide nutrition. Using free buccal graft has been observed in a number of studies to be less adequate than using onlay grafts or grafts affixed to vascularized tissues.

One might wonder about doing this procedure in 2 stages by first creating a perineal urethrostomy, as the authors describe, with ASTRA at the creation of the phallic skin tube,

and allowing that to mature for a period of time, then perhaps going back and creating a neourethra at a later date. This approach might provide more fixed vascularized tissue for the neourethra to achieve neovascularity and nutrition.

Another significant issue is whether the skin tube is going to grow along with the child. If the answer is "I do not know," then the authors have to justify the advisability of doing this procedure in a 9-month or 12-month-old youngster, as opposed to waiting until a later date. I suspect the skin tube will grow. However, if it does not, the secondary procedure is going to be more difficult because of the initial procedure.

The authors ought to be congratulated on their surgical tour de force. However, I think that the "preliminary report" part of the title has to be underlined and emphasized.

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