Pediatr Surg Int (2003) 19: 689–692
DOI 10.1007/s00383-003-1042-1

CASE REPORT

Amar A. Shah · Anirudh V. Shah

Neonatal genital reconstruction in penile agenesis by anterior sagittal anorectovaginourethroplasty

Accepted: 19 June 2002 / Published online: 22 October 2003

Abstract Penile agenesis is a rare anomaly, occurring once in every 30 million births. Early gender reassignment and genital reconstruction helps the family to accept the childs altered gender and to reduce psychological problems. In the past, multiple operations were carried out to form feminized external genitalia. This report describes a single-stage gender reassignment and genital reconstruction by anterior sagittal anorectovaginourethroplasty in a neonate with penile agenesis.

Keywords Penile agenesis · Neonate · Single-stage reconstruction

Introduction

Agenesis of the penis occurs as a consequence of single-gene disorders, teratogenic effects, or malformation sequences and associations, and occurs in unrecognized patterns of anomalies. It thus should be considered a developmental field defect. Its concurrence with scrotal hypoplasia, absent raphe, and anal anomalies implies a major disturbance of the caudal mesoderm. In such cases, severe renal defects are usually seen and the prognosis is poor. When the patient has a patent urethra and normal scrotum, raphe and testes, however, penile agenesis may be a localized malformation of the genital tubercle [1, 2, 3, 4].

Genetic assignment as a phenotypic male is problematic because of the difficulties involved in constructing a functioning phallus and is not the assignment of choice [2, 4]. We report a case in which a female gender assignment and complete genital reconstruction was done by anterior sagittal anorectovaginourethroplasty (ASARVUP) in a neonate with penile agenesis.

A. A. Shah · A. V. Shah
Department of Pediatric Surgery, V.S. Hospital, Ahmedabad, India

A. V. Shah (✉)
"Anicare", 13 Shantisadan Society, Parimal Garden, Doctor House, Ellisbridge, 380 006 Ahmedabad, India
E-mail: anirudhshah@icenet.net
Tel.: +91-79-6442478
Fax: +91-79-6466855

Case details

A male infant was born with complete penile agenesis to a 28-year-old mother of poor socioeconomic status. The child was a full-term home delivery. No antenatal ultrasound examinations had been done for the mother. There was no history of any prenatal problems on the part of the mother or any antenatal drug history. No history of any congenital anomalies was noted in the other two children.

The physical examination revealed a 2.5-kg child with an absent penis, a normal scrotum with a median raphe and two palpable gonads. No urethral meatus was seen; urine was seen coming from the anal opening, which was anteriorly displaced and small (Fig. 1).

Ultrasound and radiological imaging revealed unilateral renal agenesis with a normally functioning right kidney. The bladder was normal and there was no VUR on micturating films. The postsphincteric urethral opening was present in the rectum (Fig. 2). Other systems were normal. Chromosomes were 46 XY.

It was decided to assign female gender to the child, and bilateral gonadectomy, urethral and vaginal reconstruction and anorectoplasty was performed on the 5th day of life.

The child was placed in a modified lithotomy position so as to expose the coccyx for a combined abdominoperineal approach. A curvilinear incision was placed in the hypogastric skin crease. Bilateral gonadectomy was performed through the inguinal route. The bladder was opened and a probe was passed through the urethral opening. A midline perineal incision was placed extending from the posterior margin of the scrotum to the posterior margin of the neoanus, which was marked with a muscle stimulator. The muscles were divided in the

Fig. 1 Neonate with penile agenesis with an anteriorly displaced small anal opening with urine seen coming out through it

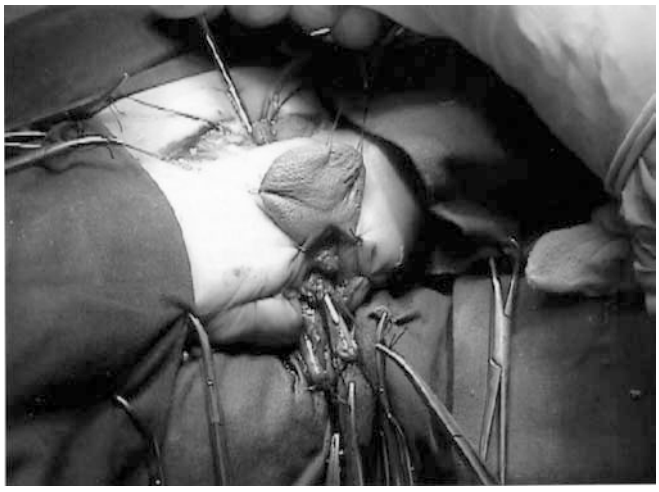

Fig. 3 The dissected urethra identified by a urethral sound passed transvesically

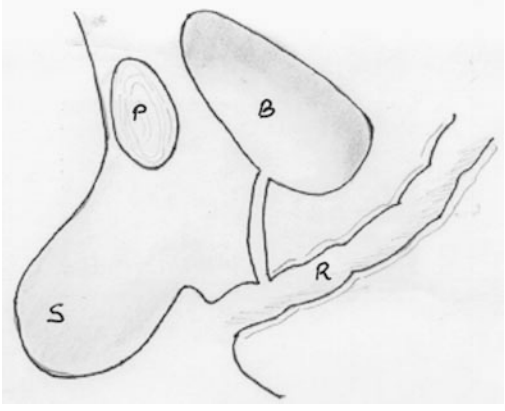

Fig. 2 Schematic diagram of the fistula opening into the rectum (*S* scrotum, *P* pubis, *B* bladder, *R* rectum)

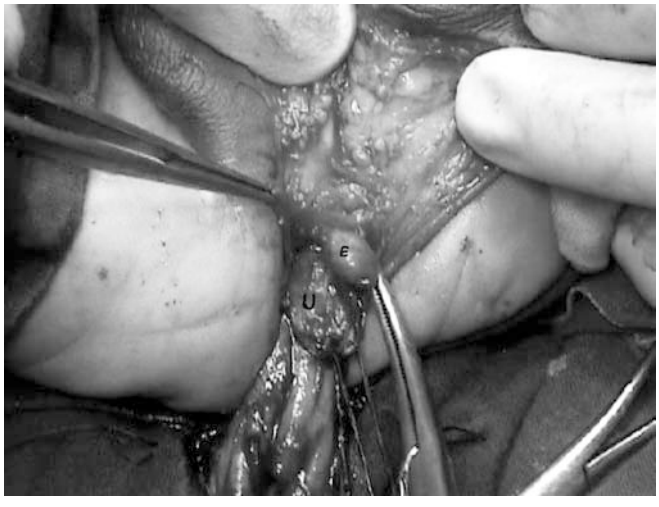

Fig. 4 The erectile tissue (*E*) anterior to the urethra (*U*) that was repositioned to form the clitoris in genitoplasty

midline until the rectum was reached. The rectum was mobilized circumferentially, and the recto-urethral fistula was identified, facilitated by a metal sound placed transvesically. The fistula was dissected from the rectum. The urethra was then dissected (3 cm in length) and brought to the perineum (Fig. 3).

The rectum was mobilized preserving the vascular supply from the bladder base. A small 0.5 cm erectile tissue was seen near the anterior wall of the urethra, which was preserved and repositioned to form a clitoris anterior to the urethra later in the genitalplasty (Fig. 4). The retro-rectal space was dissected through the levator sling. The abdomen was opened and the sigmoid colon was mobilized. The sigmoid colon was then transected just above the peritoneal reflection, and the distal rectal stump was closed in two layers with absorbable sutures.

The sigmoid colon was then pulled through the levator sling posterior to the rectum and placed within the confines of the external sphincter complex at the proposed neoanus site, and an anoplasty was performed after approximation of the levators. The perineal body was created, and the blind rectal stump was sutured to the perineum between the urethra and the pulled-through colon so as to form the neovagina. Vulvoplasty was done using the nonrugose scrotal skin. A suprapubic catheter was kept to drain the bladder and a fine Silastic tube was placed through the urethra as a stent. A protective proximal sigmoid colostomy was performed (Fig. 5).

The urethral stent was removed after 7 days and intermittent blockage of the suprapubic catheter was started. The suprapubic catheter was removed 2 days later. The colostomy was closed and a refashioning anoplasty was done 6 weeks after the procedure. The child is in regular follow-up, urinates from the neo-urethra and passes stools from the neoanus 7 to 8 times a day. Follow-up ultrasound examinations have shown no evidence of a post-void urinary residue in the bladder.

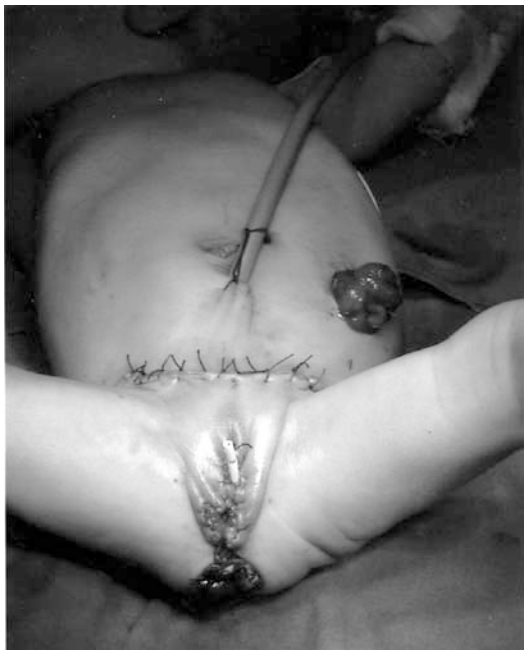

Fig. 5 Final picture after the complete genital reconstruction

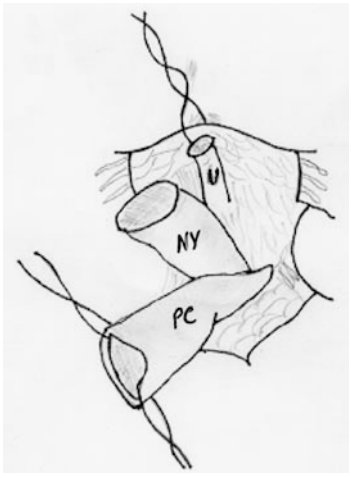

Fig. 6 Schematic representation of the anterior sagittal approach showing the urethra (*U*), rectal neovagina (*NV*), and pulled-through colon (*PC*)

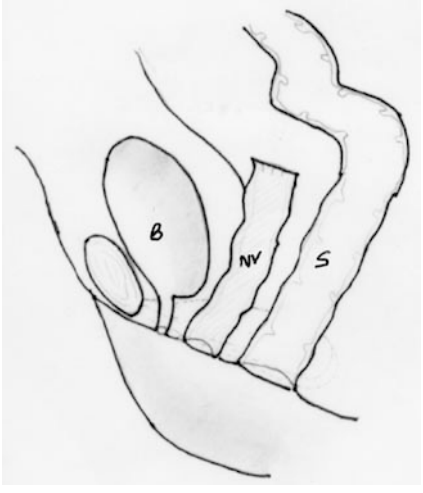

Fig. 7 Schematic diagram of completed repair (*B* bladder, *NV* rectal neovagina, *S* pulled-through sigmoid with no proximal anastomosis)

Discussion

The diagnosis of aphallia is evident at birth, and there is no reason for not initiating early treatment, as early gonadectomy and genital reconstruction help the family to accept the childs altered gender and reduce psychological problems. Aphallia dictates female gender assignment. Gonadectomy with preservation of the scrotal skin should be performed in the first few days of life to prevent male gender marking from the testosterone surge occurring between the 10th and 60th day of life [5]. Behavioral problems and male psychological orientation in childhood have been seen in patients in whom gonadectomy has been performed after 3 months of age, probably due to the problems of central nervous system testosterone imprinting [4]

A wide spectrum of associated anomalies may be seen in these cases, and patients can be classified into two groups: a severe form of anomaly with renal aplasia or dysplasia which would be incompatible with life, and a second group with fewer additional malformations and hence low mortality [6].

The failure of the genital tubercle to develop in very early embryogenesis results in the urethra often opening into the anus or rectum, although it has been described to open on the perineum, scrotum or overlying pubis [7]. Due to lack of penile and urethral development, the anus is displaced anteriorly and usually small in caliber.

In patients raised as females, genitourinary reconstruction is performed by correction of the urethral anomaly followed by vaginoplasty. Different approaches, including the conventional abdominal and perineal approach and the posterior sagittal approach, have been described for the reconstruction [7]. However, reports of complete gender reassignment and genital reconstruction as a single-stage neonatal procedure are few.

Our experience with single-stage neonatal gender assignment and genital reconstruction by the anterior sagittal approach has been encouraging. The patient is placed in a modified lithotomy position so as to facilitate a combined abdominoperineal approach without having to frequently change the position. The curvilinear hypogastric incision in the skin crease not only facilitates an inguinal gonadectomy (thus preserving the scrotal skin), but is also suitable for division of the urethrorectal fistula, vaginal reconstruction and colonic pull-through.

The urethral opening was at the anal verge, and division of the urethra from the anal canal could be performed without much difficulty. The ectopically placed anal opening prompted us to use the rectum for a vag-

inoplasty, pulling through the sigmoid colon posteriorly, thus avoiding an additional bowel anastomosis, which in turn saved time and reduced morbidity (Fig. 6 and 7).

As has been shown in patients with ambiguous genitalia, vaginal reconstruction as early as the first weeks of life can be carried out safely [8]. The cosmetic results are comparable to those achieved with a delayed operation, and early intervention may afford a far greater chance of appropriate psychosocial development.

This is one of the few reported cases of complete gender assignment and genital reconstruction in the neonatal period for penile agenesis. The combined anterior sagittal and abdominal approach definitely helps in the operative procedure. Further experience, comments and follow-up for this approach in the management of this rare condition are needed from multiple centers.

References

1. Evans JA, Erdile LB, Greenberg CR et al (1999) Agenesis of the penis: patterns of associated malformations. Am J Med Genet 84(1):47–55
2. Gautier T, Salient J, Pena A et al (1981) Testicular function in 2 cases of penile agenesis. J Urol 176:556–557
3. Gray SW, Skandalakis JE (1972) Embryology for surgeons. Saunders, Philadelphia, pp 613–15
4. Kessler WO, McLaughlin AP III (1973) Agenesis of the penis: embryology and management. Urology 1:226–28
5. Oesch IL, Pinter A, Ransley PG (1987) Penile agenesis: a report of six cases. J Pediatr Surg 19:172–74
6. Richart R, Benirschke K (1960) Penile agenesis. Arch Pathol 70:134–142
7. Sharp RJ, Holder TM, Campbell PH et al (1987) Neonatal genital reconstruction. J Pediatr Surg 22:168–171
8. Stolar CJ, Wiener ES, Hensle TW et al (1987) Reconstruction of penile agenesis by a posterior sagittal approach. J Pediatr Surg 22:1076–80