Total vaginal replacement with sigmoid colon for partial testicular feminizing syndrome in pediatric age

Jehad Abu Daia, MBCH, FRCS. Roberto De Castro, MD. Carmine Del Rossi MD, Kamal A. Henash, MS (Urology), FACS.

ABSTRACT

A 46XY patient, raised as girl, affected by androgen insensitivity syndrome with an incomplete receptor block (partial testicular feminizing syndrome), underwent a total vaginal reconstruction, bilateral orchietomies and vulvoplasty at the age of 11 years. At the time of the operation, the clitoris was mildly enlarged and a short urogenital sinus was present with a normal urethral opening and a very small vaginal introitus. The vagina was tiny, blind-ending and useless and, obviously, no female internal genitalia were found. Two large hemiscrotums with full size testes were present. The patient has a normal female attitude. The vaginal replacement was performed using a segment of sigmoid colon. Eight days after the operation the patient and her mother were taught how to dilate and irrigate the neovagina once a day. At one month follow-up, the neovagina showed a good looking introitus, the mucorrea was significantly decreased, a size 22 Hegar dilator was easily introduced inside the vagina and a vaginogram was satisfactory. Moreover, the two labia majora, made with hemiscrotum flaps, gave a normal appearance to the external genitalia. In conclusion, the preliminary result of this case confirms the feasibility of vaginal reconstruction in pediatric age, with advantage of early restoration of a normal genital anatomy in a one stage operation before the usual age of puberty. The sigmoid neovagina seems to be the procedure of choice for total vaginal replacement in children, for good cosmetic and hygienic results and for simple and temporary postoperative management.

Keywords: Pseudohermaphroditism, androgen insensitivity syndrome, testicular feminizing syndrome.


The incomplete receptor block of androgen insensitivity syndrome, known as partial testicular feminizing syndrome (PTFS) is less uncommon than was previously thought.1 PTFS is a male pseudohermaphroditism variant, with 46 XY karyotype and, obviously, no female internal genitalia. Compared to the complete receptor block of androgen insensitivity syndrome, PTFS presents a mildly more virilized whole body and external genitalia: none or modest breast development, hypertrophic clitoris, labia majora similar to hemiscrotum, earlier outside appearance of the testes.

At the extremities of the spectrum of the malformation there are apparent normal female external genitalia and apparent normal male external genitalia.1 However, the majority of the patients with PTFS at birth are phenotypically female and they are rightly raised as female, but their vagina is frequently tiny, blind-ending, useless and needs to be replaced.

Vaginal reconstruction can be performed by numerous methods: the non-surgical perineal dilatation (Frank's technique),3 the skin grafts McIndoe operation4 and the use of bowel segment (small bowel or sigmoid colon).4

From the Department of Urology, Pediatric Urology Section (Abu Daia, De Castro, Henash), and the Department of Surgery, Pediatric Surgery Section (Del Rossi), King Faisal Specialist Hospital & Research Center, Riyadh, Kingdom of Saudi Arabia.

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Address correspondence and reprint request to: Dr. Jehad Abu Daia, Department of Urology, King Faisal Specialist Hospital & Research Center, MBC 83, PO Box 3354, Riyadh 11211, Kingdom of Saudi Arabia. Fax. 01 - 442-4301.
We report the case of an 11 year old patient with PTFS, who underwent a total vaginal replacement with a segment of sigmoid colon, bilateral orchietomies and vulvoplasty in one stage. The aims of this report are to show the feasibility and the advantages of early vaginal reconstruction and to support the choice of the sigmoid colon for vaginal replacement in children.

**Case Report.** An 11 year old patient reported recently to the Pediatric Urology Outpatient Clinic of King Faisal Specialist Hospital & Research Centre for a surgical program of genital reconstruction. The patient had been followed in this hospital from early childhood by the Pediatric Geneticists and Pediatric Endocrinologists for a case of PTFS with labial masses. The karyotype is 46XY. She was raised as a girl and has a normal female attitude. Several abdominal and pelvic ultrasound evaluations showed a normal urinary tract and absence of uterus and ovaries. A recent genitogram showed a short urogenital sinus with normal bladder and urethra and a tiny, blind-ending vagina (Figure 1). She has, from the same parents, 5 normal brothers, 8 normal sisters and 2 younger sisters with the same disease.

As the first step, she underwent an examination and endoscopy under general anesthesia: the whole body shape looked like a boy of the same age, there was no breast development, a mild hypertrophic clitoris was present, the labia majora were similar to hemiscrotum, with full-size testicles inside (Figures 2a and 2b). The endoscopy confirmed the presence of a short urogenital sinus and a very small vagina. The vagina was considered as useless and a surgical program of vaginal replacement, vulvoplasty and removal of the testicles was suggested for the patient and her family.

The preoperative treatment consisted of a thorough cleansing bowel program, antibiotic therapy from the day before surgery, i.v. fluids and clear fluids orally from 24 hours before surgery.

The surgery was performed under general anesthesia and insertion of a catheter in the peridural space. The peridural catheter was also used in the post-operative period for analgesia. A Foley catheter is placed in the bladder with the patient placed in a lithotomy position, the abdomen and the perineum are prepped in a single field.

Through a suprapubic-Pfannenstiel incision, the 2 testicles were removed (Figure 3a). The abdomen was opened through the same incision with the muscles split in the midline from the umbilicus to the pubis. A 14 cm length of sigmoid colon was isolated, pedicled on a distal sigmoid artery and vein, preserving the primary vascular arcade (Figure 3b). The proximal part of the graft was closed in 2 layers with absorbable suture. The bowel continuity was re-established with a single layer anastomosis of interrupted Vycril 3/0 sutures, with the knots inside the lumen; the mesenteric defect was closed, keeping the neovagina and its mesentery at the left side of the field.

The vulva region was then incised in a cruciate fashion and a tunnel is created bluntly between the bladder and the rectum. The peritoneum was incised pushing upward a Heger dilator from the perineal introitus and the dissection was continued until a large plastic tube can be passed from below through the perineum (Figure 3c).

The neovagina was pulled through the tunnel by a Allis forceps, paying particular attention to the blood supply of the graft to make sure that it was not twisted or under tension. A single layer anastomosis was performed with Vycril 3/0 at the vulva region. A few fixation sutures were applied between the neovagina and the retro-peritoneum to prevent volvulus or prolapse of the graft. No drainage was left and the neo-vagina was stented with a Vaseline gauze for 5 days to facilitate the adhesion of the graft to the surrounding tissue. The labia majora were then recreated using the 2 hemiscrotum flaps.

Eight days after the operation the patient and her mother were taught how to calibrate, dilate, and irrigate (to remove the mucus) the neovagina once a day (Figure 4). Calibration and dilation of the neovagina has to be performed daily for about 6 months. Irrigation will be necessary for at least 4 months. A set of Heger dilators (size 18 to 26) are left with the patient.

The post-operative course was uneventful. The pathology report was compatible with incomplete variant of androgen insensitivity because the features present were those of adult testis with spermatogenesis, and actually full maturation of germ cells to spermatozoa is very unusual in patients with complete receptor block of androgen insensitivity syndrome.

At the one month follow-up appointment, the neovagina showed a good looking introitus, the mucora was significantly decreased, a size 22 Heger dilator was easily introduced inside the vagina and a vaginogram was satisfactory. The 2 labia majora, made with hemiscrotum flaps, gave a normal appearance to the external genitalia.

**Discussion.** Partial testicular feminizing syndrome (PTFS) is a variant of androgen insensitivity syndrome with partial receptor blocks. The vagina in patients with PTFS is often a distal useless remnant, that frequently needs to be replaced. The number of vaginal replacements reported in the literature in patients with complete or incomplete androgen insensitivity syndromes is small, compared with the number of cases affected by Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome. In the extraordinary Hendren's series of 65 vaginal reconstructions in children and young adults, the majority (34 cases) were affected by cloacal
Figure 1 - Genitogram showing a tiny blind-ending vagina and a short urogenital sinus.

Figure 2 - Clinical photographs showing (a) A male habitus with no breast development; (b) Labia majora similar to hemiscrotum with two large testes.

Figure 3 - Operative photographs showing (a) Full size testicles; (b) A 14 cm long segment of sigmoid colon used as neovagina; (c) A large plastic tube inserted into the perineum to dilate the tract of the neovagina, and a Foley's catheter inserted into the urethra.
malformations, 12 by MRKH syndrome and only 2 patients were in the intersex group. In the 2nd largest series of vaginal replacement in pediatric and adolescent ages (31 cases), Hensle and Reiley report 20 patients with MRKH syndrome and 5 with androgen insensitivity syndrome. At the University Hospital of Bologna, (attended by R.D.C) 19 cases of vaginal replacement were carried out and 7 of them were affected by androgen insensitivity syndrome. Two of us (C.D.R and R.D.C) both had previous experience with 10 vaginal replacements in young ladies in Bangladesh, but all of them were affected by MRKH syndrome.

Compared with the perfect female external genitalia of MRKH syndrome and the almost normal appearance of complete receptor block of androgen insensitivity syndrome, the external genitalia of patients affected by PTFS frequently presents some anomalies like a hypertrophic clitoris, incomplete labia majora (often similar to hemiscrotum) and common opening of urethra and vaginal remnant in a urogenital sinus. Therefore, the vulva and the clitoris have to be reconfigured before the vaginal reconstruction. The ectodermal distal portion of the blind-ending vagina, very useful in the MRKH patients to create distal vaginal flaps to be anastomosed to the neo-vagina, is rarely utilizable in PTFS. Therefore, the vaginal draft has to be removed and skin flaps have often to be created to avoid direct external appearance of the neo-vagina. All these surgical preliminaries, in our opinion, are very important to achieve an esthetically right result, that will help very much to achieve functional and psychological excellent results.

Regarding the technical choice of vaginal reconstruction, at present, pediatric urologists and pediatric surgeons seem to prefer the use of bowel and in particular the use of sigmoid colon. Actually, the Frank technique of non-operative vaginal dilatation seems not to be adequate for patients with a short vaginal dimple and for the strict follow-up required. The McIndoe technique using a skin graft, a very common procedure, requires a continuous and prolonged dilatation, night time stenting, with a high rate of stenosis, graft shortening and dyspareunia. The small bowel not infrequently has a short vascular pedicle, inadequate to reach the perineum and the mucus production is greater and does not decrease with time. On the contrary, the choice of sigmoid colon as a graft for creation of a new vagina is extraordinarily effective for the length that can be obtained and for the good blood supply that prevents complications such as contraction, shrinkage, narrowing or stenosis at the perineal introitus. The thick wall of the colon tolerates trauma better than small bowel or skin grafts. The sigmoid neo-vagina offers simple and temporary post-operative management (mean 6 months). The mucus production after 3-4 months decreases dramatically and calibration, dilatation and irrigation are temporary and well tolerated.

In conclusion, the good preliminary result obtained in this patient affected by PTFS supports the feasibility of vaginal replacement in the pediatric age. The restoration of a normal genital anatomy before the standard age of puberty, achieves certainly great psychological benefit. The sigmoid neo-vagina seems to be the procedure of choice for vaginal replacement in children because of good cosmetic and hygienic results, and for simple and temporary post-operative management.

References