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Trauma and Reconstruction

Two-Stage Urethroplasty with Buccal Mucosa for Penoscrotal Hypospadias Reconstruction in a Male with a 46,XX Karyotype

Pieter D'hulst [a,*], Jochen Darras [a], Steven Joniau [b], Pieter Mattelaer [a], Linsey Winne [c], Diederik Ponette [a]

[a] Dept. of Urology Az Damiaan Oostende, Gouwelozestraat 100, 8400 Oostende, Belgium
[b] Dept. of Urology UZ Leuven, Herestraat 49, 3000 Leuven, Belgium
[c] Dept. of Endocrinology Az Damiaan Oostende, Gouwelozestraat 100, 8400 Oostende, Belgium

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ABSTRACT

We present a case regarding a 32-year old African male with penoscrotal hypospadias, left cryptorchidism and a left inguinal hernia. There were moderate masculinization characteristics. He underwent a Lichtenstein hernia repair with perioperative biopsies of the left inguinal testis and epididymis. Microscopic examination showed a Sertoli-only left testis with Leydig-cell hyperplasia and the left epididymis consisted of ovarian tissue with corpora albicantia and maturing follicles. Endocrinological evaluation showed a sex-determining region Y (SRY) negative 46,XX karyotype. We successfully performed a two-stage urethroplasty with buccal mucosa graft to reconstruct his penoscrotal hypospadias.

Introduction

46,XX testicular disorder of sex development is a rare condition that affects approximately 1 in 20000 newborn males. Affected people have male external genitalia and children are typically raised as males. However, they mostly have small testes and may also have abnormalities such as cryptorchidism or hypospadias.

At puberty, development of male secondary sex characteristics such as facial hair and deepening of the voice masculinization is slow or absent and gynecomastia is frequently present. Furthermore, the affected males are infertile.

Case presentation

A 32-year old male presented at the emergency department because of swelling and pain in the left groin since a couple of days. There was no fever. Clinical examination showed left cryptorchidism, penoscrotal hypospadias, bilateral gynecomastia and a left inguinal hernia. Ultrasound revealed a left epididymitis. Non-steroidal anti-inflammatory drugs and quinolones were prescribed. Following consultation at abdominal surgery, a Lichtenstein hernia repair was performed. Peroperatively the surgeon noticed a small, reddish-brown left testis and a hardened, cystic left epididymis. Biopsies were taken for analysis.

Microscopic examination showed a Sertoli-only left testis with Leydig-cell hyperplasia. The left epididymis consisted of ovarian tissue with corpora albicantia and maturing follicles. The patient was referred to the endocrinology department for further investigations. A blood sample revealed a suppressed testosterone level and genetical analysis with fluorescence in situ hybridization (FISH) and karyotyping showed a SRY negative 46,XX karyotype. These findings are consistent with a SRY negative 46,XX testicular disorder of sex development. An MRI of the pelvis showed intrascrotal gonadal tissue on the right side and gonadal structures in the inguinal canal on the left side. There was also a small uterus without a vaginal component.

The patient wanted a surgical reconstructive procedure regarding his penoscrotal hypospadias and a two-stage urethroplasty was planned and performed. During the first stage, the hypotrophic urethral plate was augmented from the glans until the bulbar level. We harvested and grafted a buccal mucosa patch and a perineostomy was constructed for urinary diversion. Eight months later we performed the second stage. The buccal mucosa graft had healed perfectly and the anterior urethra was reconstructed by tubularizing the newly formed urethral plate (Figs. 1–3).

* Corresponding author. Department of Urology Az Damiaan Oostende, Gouwelozestraat 100, 8400 Oostende, Belgium.
E-mail address: pieterdhulst@icloud.com (P. D'hulst).

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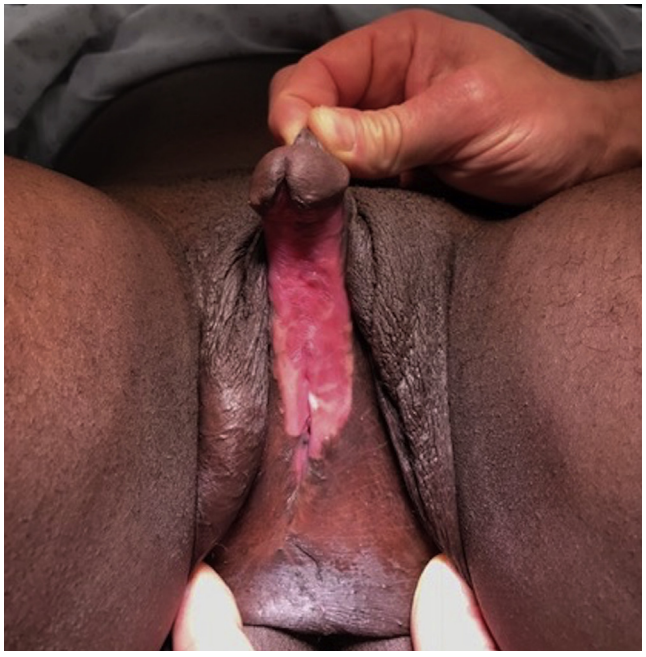

Figure 1. Status at the time of the second stage urethroplasty: nicely healed buccal mucosa graft.

We planned to leave the catheter in situ for 2 weeks, but first cystography showed minimal distal leakage so we left the catheter in situ for 2 more weeks. Cystography 1 month postoperative showed no leakage and the catheter was removed. Six weeks after surgery he had a good uroflowmetry with a maximum flow rate of 18 ml/s with no post-void residual urine. He was pleased with his improved quality of life.

Discussion

46,XX testicular disorder of sex development is characterized by various clinical presentations like cryptorchidism, hypospadias and gynecomastia, but normal male external genitalia may also be present.[1] Diagnosis is made by gonadal biopsy, testosterone deficiency and molecular studies with FISH and karyotyping.[1,2]

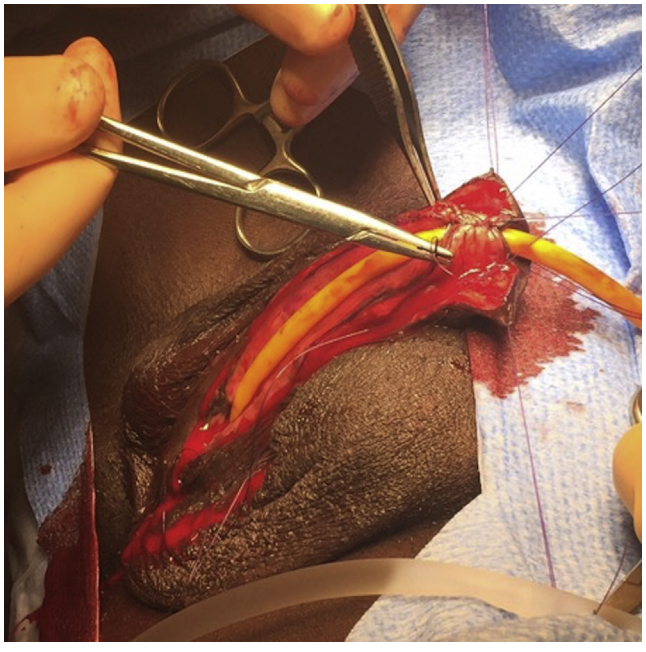

Figure 2. Tubularization of the BMG into a neo-urethra.

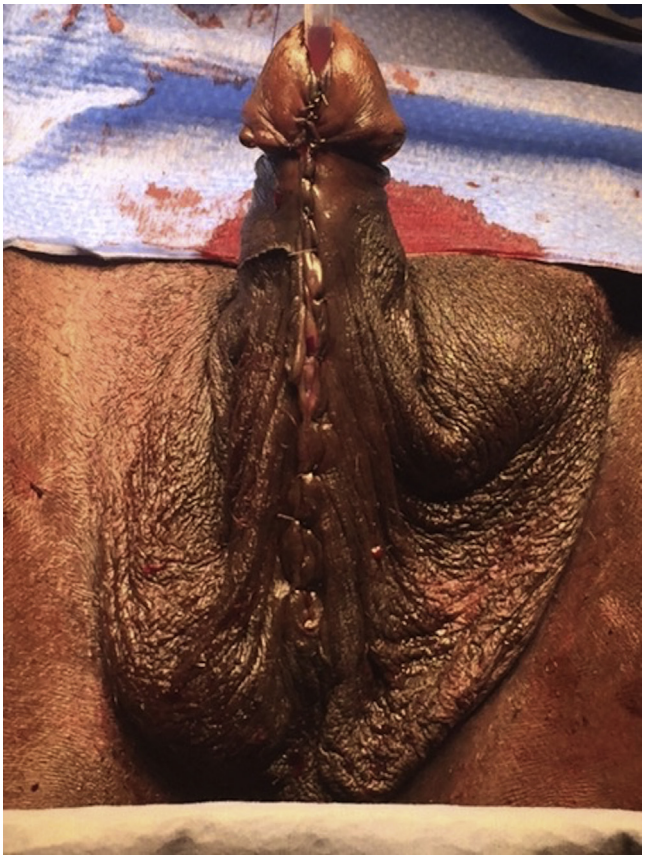

Figure 3. Final result after closure.

The SRY gene is responsible for making the SRY protein. The expression of this protein causes the initiation of male sex determination in humans. There are two types of 46,XX testicular disorder of sex development: SRY-positive and SRY-negative.

In about 80% there is an abnormal exchange of genetic material between chromosomes which causes the SRY gene to be translocated, almost always onto an X chromosome. A fetus conceived from a sperm cell with an X chromosome bearing the SRY gene will develop as a male, despite not having a Y chromosome. This form is called SRY-positive 46,XX testicular disorder of sex development.[1,2]

In about 20% of the affected people the SRY gene is absent. This form of the condition is called SRY-negative 46,XX testicular disorder of sex development. The cause of the disorder in these individuals is often unknown. They are more likely to have ambiguous genitalia,[1,2] which was the case in our patient.

The penoscrotal hypospadias was reconstructed with a two-stage urethroplasty using a buccal mucosa graft. Primary hypospadias repair is rarely performed in adults and most studies in the world literature are focused on pediatric patients. We preferred a two-stage approach because a full circumference urethral reconstruction was necessary.

The use of buccal mucosa is widely accepted in the reconstructive literature as an excellent graft tissue.[3,4] At the first stage, we harvested and grafted a large buccal mucosa patch as a neo-urethral plate, considering the length of urethral defect. After a period of eight months, excellent tissue healing was obtained and reconstruction of the neo-urethra was performed in a second stage. Longer follow-up after this surgery is still needed to fully assess the patient's quality of life.

Concerning the residual scrotal and inguinal gonadal tissue, further surgery has to be performed in the nearby future because of the elevated risk of development of germ cell tumors. Testosterone treatment should be initiated to stimulate the development of male secondary sex features and to improve quality of life. Esthetical reconstructive surgery could be performed to treat the bilateral gynecomastia. Since disorders of sexual development (DSD) can be associated with important psychological impact, patients with DSD should also be offered psychological support.

Conclusion

We describe a rare case of 46,XX testicular disorder of sex development in a patient with severe penoscrotal hypospadias. Urologists should be aware for genetical disorders in patients with suspect clinical features like cryptorchidism, hypospadias and gynecomastia.

A multi-disciplinary approach is needed to fully assess the patient's disease, treatment and follow-up. The reconstructive urologist and endocrinologist both play a major role in achieving an improvement of the patient's quality of life.

Consent

Patient consented with publication.

Conflict of interest

None declared.

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