Female pseudohermaphroditism with urethral duplication: A delayed presentation in adulthood

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INTRODUCTION

Female pseudohermaphrodites are genotypical females (46, XX) with two ovaries, however, their external genitalia may have varied levels of masculinisation. The commonest cause of female pseudohermaphroditism is congenital adrenal hyperplasia, which is usually identified in the neonatal period as ambiguous genitalia, presenting as either partial or complete fusion of the labioscrotal fold. It may, at times, also present as clitoromegaly with a phallic urethra.

As such, the duplication of the urethra is a rare congenital anomaly in females and the definitive embryological origin of this condition is still unclear. The most accepted classification for the duplication of the urethra, proposed by Orotolano-Nasarallah, is based on the embryological origin of the accessory urethra and its connection with the primary urethra.[1] The urethra is commonly duplicated in the sagittal plane and may be associated with the duplication of the urinary bladder or some anomaly of the upper tracts such as ureteral ectopia.[2] A combination of female pseudohermaphroditism with urethral duplication is rare, with only a few isolated case reports in neonates published in the literature.[3] To our knowledge, delayed diagnosis in adulthood is extremely rare.

CASE REPORT

A 26-year-old female had multiple failed attempts of catheterization during a planned LSCS (Lower Segment Caesarean Section) and underwent intraoperative suprapubic catheterization. She was later referred to us with a suprapubic catheter in situ. The physical examination revealed a hypertrophied clitoris with a phallic urethra and the labial folds were fused along with a posteriorly displaced vaginal introitus [Figure 1]. Ultrasonography revealed normal kidneys and bladder and the retrograde urethrogram could only visualise the urethra partially [Figure 2]. Subsequently, evaluation under anesthesia and cystoscopy revealed a phallic urethra with multiple false passages in the urethra with a normal but curved urethral tract [Figure 3]. Vaginoscopy revealed a roomy vagina with a normal cervix. However, no abnormal urethral opening was identified.

The patient was planned for urethral reconstruction and labial separation. Circumcoronal incision at the clitoris was made and extended in the midline in an inverted Y shape around the vaginal introitus. On separation of the spongiosum from the cavernosa, an accessory urethral tract opening into the anterior vaginal wall was identified [Figures 4 and 5].

ABSTRACT

Female pseudohermaphroditism with urethral duplication presenting as urinary retention in adulthood is extremely rare. We report the case of a 26-year-old female who had multiple failed attempts of per urethral catheterization during a planned cesarean section. She had labial fusion and clitoral hypertrophy with a phallic urethra and underwent labial separation with urethral reconstruction.
urethra did not have any connection with the rest of the urethral tract; hence, it was excised in toto. The primary urethra was narrow distally, hence was split opened ventrally till the normal caliber was reached. The dorsal wall of the urethra formed the mucosal lining of the genital introitus. The clitoris was reconstituted without corporoplasty and labial separation was performed so as to restore the appearance similar to a normal female external genitalia [Figure 6]. She had an uneventful recovery. The perurethral catheter was removed on the 14th postoperative day and on subsequent follow-up, the lady was voiding well. The histology of the excised urethra showed a tubular structure lined by squamous epithelium with nonspecific inflammation.

**DISCUSSION**

Duplication of the urethra in females is one of the most uncommon congenital anomalies of the lower urinary tract. It is often associated with the duplication of the urinary bladder or ano-rectal anomalies. As per the hypothesis put forward by Patten and Barry, the duplication of the urethra is the after-effect of an unusual connection between the primordial genital tubercle and the ventral end of the cloacal membrane. The clinical presentation depends on the patency of the duplicated urethra. Those with a patent complete duplication of the urethra may be asymptomatic and may not be diagnosed till adulthood. However, few may present with double stream of urine, incontinence of urine, recurrent urinary tract infection, or urinary retention.

Based on the magnetic resonance imaging, the duplication of urethra in females can be classified into the following types:

- **Type 1**: Duplication of the urethra and the bladder
- **Type 2**: Duplication of the urethra with a normal urinary bladder
- **Type 3**: Accessory urethra posterior to the primary urethra
- **Type 4**: Duplication of the proximal urethra with a single distal urethra
- **Type 5**: Normal proximal urethra with a duplication of the distal urethra.

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**Figure 1:** Fusion of labial folds with the hypertrophied clitoris

**Figure 2:** Retrograde urethrogram showing partial visualization of the urethra

**Figure 3:** Cystoscopy image showing phallic urethra (with guide wire) and false passages (marked with arrow) in the urethra

**Figure 4:** Intraoperative photograph showing an infant feeding tube passing from the primary urethra into the bladder (marked with black arrow) and a second infant feeding tube passing from the accessory urethra into the vagina (marked with yellow arrow)
Our patient had type 5 female urethral duplication and presented in adulthood. In addition, she also had clitoromegaly with a phallic urethra and a posteriorly placed vaginal introitus. She had normal mullerian structures on ultrasound with a normal reproductive history. To our knowledge, very few cases of urethral duplication and virilization have been described in the literature. Most of these patients are diagnosed in the neonatal period with ambiguous genitalia and presentation as adults is extremely rare.

Depending on the extent of virilization, the female pseudohermaphroditism can be classified into Group 1 – Congenital adrenal hyperplasia, Group 2 – Masculinisation by maternal hormone, Group 3 – Idiopathic, Group 4 – Special (associated with complex ano‑rectal anomalies), and Group 5 – Familial. As in our patient, idiopathic female pseudohermaphroditism includes features such as clitoral hypertrophy, presence of a phallic urethra, accessory vaginal urethra, incomplete fusion of the labia or urinary retention. Since the pathology is not due to hormonal causes, the other components of virilization are missing in these cases.

The treatment of this rare condition depends on the symptomatology and the presentation of the patient. A thorough physical examination assessing the degree of virilization is the mainstay of treatment planning. The work up of these cases includes ultrasonography of the pelvis, micturating cystourethrography, retrograde cystography, and cystoscopy. The most common indication for surgery includes difficulty in micturition, either duplication of the urinary stream or urinary retention. Other indications are recurrent lower urinary tract infections, correction of the ambiguous genitalia, or the associated genitourinary malformations. Surgical planning aims to preserve the primary continent urethra, reconstruct a patent urethra by excising the accessory urethra, if required Rectification of the masculinized genitalia.

CONCLUSION

The delayed diagnosis of female pseudohermaphroditism with urethral duplication in adulthood is rare. The anatomical diagnosis of this condition is based on the radiological investigations such as micturating and retrograde cystography as well as the intra-operative findings. The treatment of this condition should be tailored as per the symptomatology and the presentation of the patient.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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