Congenital aphallia: management with urethral advancement


**ABSTRACT**

Congenital aphallia is caused by a failure of the genital tubercle to form. It is extremely rare and there are only eighty cases reported in the literature. Clinically the scrotum is well developed, the testes are descended, and the penis is absent. The urethra emerges adjacent to the anus and in some cases within the rectum. Patients should be genetically evaluated at birth and associated malformations should be looked for. It is important to propose early sex reassignment.

**Objective:** To present a case of congenital aphallia and its management with urethral advancement.

**Clinical case:** Patient is a 14-year-old male adolescent with no perinatal antecedents. Aphallia was detected at birth and sex reassignment was proposed after genetic study was carried out, but was refused by the mother. Patient presented with recurrent urinary infections and bladder lithiasis as a child. He sought medical attention due to straining, tenesmus, and intermittence. Physical examination revealed both testes in the scrotum, short perineum, and postspinhincteric urinary meatus. Patient had 46XY karyotype, urinalysis reported multiresistant urinary infection, and urodynamics reported infravesical obstructive uropathy.
obstructive uropathy. Imaging studies and cystoscopy showed no anatomical alterations, and no abnormal communications or Müllerian remnants. Patient underwent urethral advancement resulting in a perineal meatus with no complications and postoperative progress was adequate.

**Conclusions:** Congenital aphallia requires multidisciplinary management. Obstructive uropathy should be managed with urethral advancement techniques to improve micturition mechanics and to avoid damage to the upper urinary tract.

**Key words:** Aphallia, urethral advancement, presphincteric, Mexico.

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**INTRODUCTION**

Congenital aphallia is the result of a failure of the genital tubercle to form. It is an extremely rare form of genital ambiguity. It was initially described by Imminger in 1853 and presently there are approximately 80 cases reported in the literature. Incidence is estimated at 1 out of every 10-30 million live births.

During embryogenesis, genital protuberance appears on both sides of the cloacal folds. It migrates caudally coming together on the urethral fold and becoming fused at the perineal raphe to form the scrotum. The genital tubercle fuses to form the urethra internally and to form the penis and penile raphe externally. Penile agenesis occurs when there is deficient formation or a defect in the development of the genital tubercle in the fourth week of gestation (Image 1). ²

In the majority of cases the urethral meatus is somewhere in the perineal region or forms a fistula to the gastrointestinal tract, usually towards the rectum.

Skoog and Belman (1989) reviewed 60 cases of aphallia and defined 3 variants based on the position of the urethra and its relation to the anal sphincter: postphincteric, presphincteric (prostatorectal fistula), and urethral atresia. ²

In general, there is greater associated mortality and there are more malformations such as cryptorchidism, vesicoureteral reflux, horseshoe kidney, kidney agenesis, imperforated anus, musculoskeletal abnormalities, or cardiopulmonary abnormalities, when the meatus is more proximal (Image 2). ²,³

At least 60% of patients have a postsphincteric meatus located at the level of the anal margin. A total of 28% have a presphincteric meatus with meatal communication either as prostatorectal fistula, vesicorectal fistula, or rectal atresia. In cases of vesicorectal fistula and urethral atresia (12%), mortality is close to 100%. ²

The majority of patients have 46XY karyotype. Its clinical appearance is a well-developed scrotum with descended testes and absence of the penis. The anus is generally displaced in a forward position and the urethra may emerge from it, or be adjacent to the anal margin, or it may emerge within the rectum. ⁴⁻⁹ Diagnosis includes the complete absence of cavernous or spongious tissue with a urethral opening in the perineum near the anus or inside the rectum.

Aphallia must be differentiated from hidden penis, rudimentary penis, micropenis, male pseudohermaphroditism, or intrauterine amputated penis. ¹⁰⁻¹⁸ Boys with this malformation should be evaluated immediately at birth with karyotype and other studies to determine if there are associated malformations in the urinary tract or in other tracts and systems. Magnetic resonance imaging (MRI) has proven to be a useful imaging study for evaluating the defects and severity of this pathology. ⁵

Essential treatment objectives are early sex assignment and obstructive uropathy correction preferably within the same surgery, and obtaining results for long-term sexual function. ²

The intestine has been described in this type of patient. For older patients whose sexual identity has been determined, virilizing surgeries such as penile reconstructions have been reported with poor results, difficult techniques, and high morbidity rate that can cause more psychological damage, making this type of management controversial.

Other authors have focused on improving micturition mechanics and reducing the risk of urinary track deterioration that can lead to other complications and to kidney failure, and have proposed managing these patients with dismantling of the urethra from the anus-rectum and performing urethral advancement toward the perineum (urethrostomy).

The patient, as a male, may potentially be fertile. However, reconstruction of an esthetically acceptable phallus that has urinary function, sexual function, and adequate reproductive function is very difficult.

**OBJECTIVE**

To present a case of congenital aphallia and its management with urethral advancement.

**CASE PRESENTATION**

Patient is a 14-year-old boy with no pathological perinatal antecedents, product of the fourth, 38-week pregnancy that progressed normally, with adequate prenatal control, and adequate weight and height. Aphallia was detected at birth and after a genetic study sex reassignment was proposed but the mother refused it. At 12 years of age patient presented with bladder lithiasis managed with open cystolithotomy and he
sought urological evaluation due to recurrent urinary infections that had been treated with multiple antibiotic regimens. When providing the clinical history, patient stated he urinated through the rectum with straining, bladder tenesmus, and intermittence, and was 100% continent.

Physical exploration revealed male phenotype with apparent age similar to chronological age, infraumbilical scar from open cystolithotomy, secondary sexual characteristics, absence of penis, android pubic hair distribution, and both testes in scrotal sacs and normal in size and consistency. Anterior anal implantation was observed with presence of postphincteric urethral meatus with no spontaneous urine leakage (Images 3 and 4). Karyotype was 46XY, hormonal profile was normal, urinalysis reported urinary infection data and urine culture was positive for multiresistant *E. coli*. Ultrasound showed a thickened bladder wall with 30% residual urine, and urodynamics revealed infravesical obstructive uropathy. Excretory urography showed no urinary tract alterations.

Cystoscopy showed permeable 3.5 cm postphincteric urethra, bladder with cloudy urine and abundant sediment, orthotopic ureteral meatuses, and bladder walls with grade II trabeculations.

MRI revealed no evidence of Müllerian remnants and no communications or urethral fistulas with postphincteric meatus were observed (Image 5).

After evaluations by the departments of Urology, Plastic Surgery, and Child and Adolescent Psychiatry together with the patient and his parents, and due to past history of recurrent urinary infections from multiresistant organisms, bladder lithiasis, and obstructive uropathy with risk of upper urinary tract deterioration, it was decided that the patient would undergo urethral advancement, separating it from the anorectal sphincter and resulting in a perineal meatus (Image 6).

A 12 F transurethral catheter was left in place for 7 days and was removed with no complications and patient had spontaneous micturition.
After first postoperative month, urinalysis and urine culture reported no signs of urinary infection and bladder ultrasound showed 4% residual urine.

**CONCLUSIONS**

Congenital aphallia is an extremely rare condition that requires multidisciplinary and individualized management. The most accepted management is early sex reassignment with obstructive uropathy correction. However, the techniques of urethral advancement should be known so that micturition mechanics can be improved and upper urinary tract damage can be avoided.

**REFERENCES**