

Aphallia Presented as Nonneurogenic Neurogenic Bladder: A Rare Case Report

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The penis as a component of external genitalia, takes part in urinary, fertility, and psychosexual structure of males. We report a case of aphallia (penis agenesis) presented with nonneurogenic neurogenic bladder (hinman syndrome). Patient refused reconstructive procedure for aphallia and underwent mitrofanoff procedure for non-neurogenic neurogenic bladder and later trained in clean intermittent catheterization.

Keywords: Aphallia, Hinman syndrome, Mitrofanoff procedure, Non-neurogenic neurogenic bladder, Penile agenesis

INTRODUCTION

Aphallia or penile agenesis is an extremely rare genitourinary anomaly. Its incidence is about one in 30 million births.¹ It occurs due to absence or a failure in the development of genital tubercule.² It was originally described by Imminger in 1853, after that approximately 100 cases have been reported worldwide. Aphallia is classified into two major groups. The first one is the solitary malformation of aphallia. The second one is the complex form which is associated with other congenital anomalies and often incompatible with life.^{3,4} More than half of these patients have associated anomalies including genitourinary (54%) and gastrointestinal tract anomalies. Skoog and Belman⁵ classified these patients based on the relationship between the anal sphincter and the ectopic urethral meatus. They described three variations: The first one is postsphincteric form with anterior perianal urethra, the second one is presphincteric urethrorectal fistula, and the third one is urethral atresia with vesicorectal fistula.

Urethral opening can present either over pubis or at any part of perineum or most frequently in anterior wall of the rectum.⁶ The treatment of aphallia is controversial. Previously, gender re-assignment at infantile period of life

has been considered as the most appropriate choice for these patients.

CASE REPORT

A 16-year-old male patient presented with complains of fever with chills and rigor for 15 days and burning micturation, dysuria, incomplete emptying of bladder and constipation for 6 months. The patient was febrile at the time of admission. On per abdomen examination, the bladder was palpable. Genitalia examination revealed agenesis of penis (Figure 1), normal scrotum, bilateral normally positioned testis and vas deferens, external meatus opening present in perineal region covered with skin tag and well developed male secondary sexual characters (Figure 2). On digital rectal examination, patient had normal anal sphincteric tone. Urine routine suggested plenty of pus cells and serum creatinine was 3.8 mg/dl. The patient was catheterized with 12 French Foleys catheter and 400 ml of urine drained. Ultrasonography of abdomen and pelvis showed, increased bladder wall thickness with mild bilateral hydroureteronehrosis. Cystourethroscopy performed which suggested normal capacity bladder with severe trabeculations. Urodynamic study was done suggested atonic bladder. Magnetic resonance imaging of lumbo sacral region done and was normal. Buccal mucosa smear was consistent with male genotype and revealed a normal karyotype 46XY. All serum hormone levels were in normal limits. On co relation of clinical and investigation findings patient was diagnosed to have aphallia with nonneurogenic neurogenic bladder (hinman syndrome) with recurrent urinary tract infection and renal failure. Patient denied reconstructive procedure for aphallia. Patient

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underwent mitrofanoff procedure for hinman syndrome (Figure 3) in which appendix is taken as a conduit and one end is anastomosed with urinary bladder and other end brought out at umbilicus. Further patient was trained in every 4th hourly clean intermittent catheterization (CIC) through umbilicus end of mitrofanoffs procedure with 10 French infant feeding tubes. Post-operative period was uneventful. On follow-up, serum creatinine was 1.6 mg/dl.

DISCUSSION

The failure of development of genital tubercle with incomplete separation of the urogenital sinus from the hindgut by the urorectal septum results in aphallia or penile agenesis. The classic diagnosis of aphallia includes complete absence of the penis, well-developed scrotum, normal descended testis with 46XY normal male karyotype. External urethral opening may be located in the perineal area, mostly inside a foreskin appearing skin tag. Aphallia must be differentiated from micropenis, concealed penis, rudimentary penis, and disorders of sexual development.⁷ Other congenital anomalies such as cryptorchidism, renal agenesis/dysplasia, musculoskeletal, and cardiopulmonary anomalies are also common (>50% cases), hence evaluation of patient for internal anomalies is mandatory. In the past, infants with aphallia underwent gender reassignment surgery, including bilateral orchiectomy, urethral transposition, vaginal replacement, and labial construction. However, the majority of these patients demonstrated male typical shift in psychosocial and psycho sexual development in a long-term follow-up.⁸ The phallus construction has been successfully done by some surgeons. Since the patient's gender identity is formed after the second year of life several urologists have advised to perform masculinizing operations, in order not to disturb the patient and his parents psychologically.⁹ Treatment of aphallia presents many challenges and it involves multidisciplinary approach. The team should include an urologist, pediatrician, geneticist, and mental health expert.

Our patient presented with urinary tract infection and renal failure and diagnosed to have hinman syndrome with aphallia. Patient relatives were counseled regarding penile reconstructive surgery and its complications and chances of failure, they denied for reconstructive procedure. Mitrofanoffs procedure for nonneurogenic neurogenic bladder was performed and subsequently patient was trained in CIC.

CONCLUSION

Aphallia or penile agenesis is extremely rare anomaly and may be associated with other congenital genitourinary or gastrointestinal abnormalities. It has a great psychosocial and psychosexual impact on patient and relatives. A multidisciplinary approach is required to treat it and



Figure 1: Aphallia

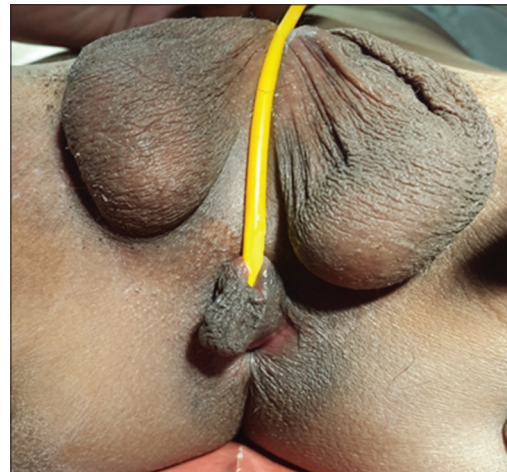


Figure 2: Aphallia with urethral opening in perineum and covered with skin tag

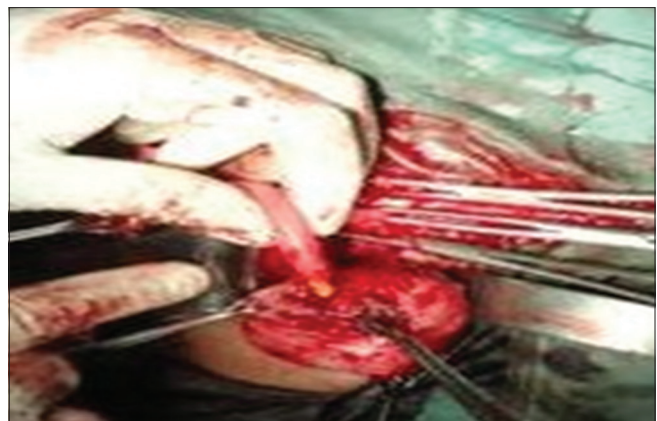


Figure 3: Mitrofanoff procedure (anastomosing appendix to urinary bladder)

depending on the age of presentation of patient different treatment options (either gender re-assignment or penile reconstructive procedure), and their pros and cons should be discussed with patient relatives.

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