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### Aphallia (Penile agenesis): A preliminary report of three cases

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#### ABSTRACT

Aphallia (penile agenesis) is an extremely rare abnormality with the reported incidence of 1 in 30 million births. The cause of this anomaly is associated with no genital tubercle formation or its development impairment. The majority of patients have 46XY Karyotype. The scrotum, testes and testicular function are usually normal. We report the preliminary experience with 3 cases of aphallia in different age groups along with a review of the literature.

**Key Words:** Aphallia; penile agenesis; anorectal malformation; phallus embryology; anoplasty.

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#### Introduction

Aphallia or penile agenesis is an extremely rare congenital anomaly occurring in 1 in 30 million births [1]. Either the absence of the genital tubercle or failure of genital tubercle to fully develop is responsible for this anomaly. Genitourinary anomalies such as renal agenesis or cystic kidney, horseshoe kidney, vesicoureteral reflux (VUR), prostate agenesis are found in about fifty-four percent of cases of aphallia [2-4]. Additionally, aphallia has also been reported with other abnormalities such as skeletal and neural disorders, annular pancreas, clubfoot and heart problems [2-4]. Imminger

was the first to report a case of aphallia in 1853 [5], and till date, less than 100 cases have been reported in literature. The karyotype of patients with aphallia is normal (46 XY); the scrotum is well formed, and the urethra is short and opens abnormally to the rectum or anus [6].

In this study, we report the preliminary experience with 3 cases of aphallia in different age groups along with a review of the literature.

#### Case 1

A 1-day-old neonate was referred to us with absence of the phallus and passage of little meconium since birth. On examination, the baby was active, pink, and weighed 3.0 kg. There was absence of the phallus and a bucket handle defect at the anus. The urethral opening was located on a skin appendage near the anus [Fig. 1].



**Fig. 1.** Neonate with aphallia, urethral opening located on a skin appendage near the anus ,well-developed scrotum with bilateral palpable testis in scrotum.

Well-developed scrotum was present in the normal position with both the gonads palpable in scrotum. Karyotype was 46 XY. Ultrasound study of the abdomen showed ectopic right kidney in pelvis and normal left kidney and urinary bladder. 2-D echo showed 4 mm Atrial Septal Defect (ASD). After these investigations, the baby underwent a Y-V cut-back anoplasty. The infant was discharged with regular anal dilatation .Mother was taught clean intermittent catheterization through urethral opening in the perineum. Definitive reconstruction is planned at a later date. The child is now 6 months old. He has normal renal parameters and a normally functioning both kidneys. We intend to do phallic reconstruction at a later time.

## Case 2

A 6-month-old male infant referred to us with a history of rudimentary phallus and passage of

stools and urine from normal anal opening since birth. General physical examination of infant was normal. He weighed 6.0 kg and appeared pink and active. On local examination, a rudimentary phallus was palpable. The scrotum was well developed containing bilateral testes. The anal opening was located normally but was very small and a pyramid shaped soft tissue was present at anal verge [Fig. 2].



**Fig. 2.** Male child with aphallia , anal opening located normally, a pyramid shaped soft tissue present at anal verge.

Renal ultrasonography showed mild hydroureteronephrosis on right side. Serum testosterone, HCG, LH levels were normal. Karyotype was 46 XY. On examination under anesthesia rudimentary glans & prepuce with hypoplastic corpora seen after releasing preputial adhesions present at the site of phallus. No urethral meatus or plate seen. Urethral opening seen on anterior wall of rectum 2-3 cm from anal margin which could be catheterized with No.6 infant feeding tube [Fig. 3]. Anus small stenotic admitting No.14 straight anal dilator only with urine coming from it. Y-V cutback anoplasty was done.

Micturating cystourethrogram done by catheter passed through urethral opening showed right grade II VUR, good bladder capacity & normal contour. Urethra was not seen [Fig. 4]. Baby discharged with regular anal dilatation and chemoprophylaxis.



**Fig. 3.** Rudimentary glans & prepuce with hypoplastic corpora seen after releasing preputial adhesions present at the site of phallus. No urethral meatus or plate seen.



**Fig. 4.** Micturating cystourethrogram showed right grade II VUR, good bladder capacity & normal contour. Urethra was not seen.

### Case 3

6 years child, reared as male was referred to us. He had perineal urethroplasty done in infancy. Patient had aphallia, poorly developed scrotum on left side, and left testis was palpable in left groin [Fig. 5].



**Fig. 5.** Patient with aphallia, poorly developed scrotum on left side, and left testis palpable in left groin with normal anal opening.

He had normal anal opening. A skin tag was present between urethroplasty and anus. He is being planned for left side orchiopexy and phalloplasty after complete work-up.

### Discussion

Aphallia occurs within 4 weeks of embryonic development accompanying other anomalies [7,8]. The embryological defect of genital tubercle to develop and incomplete separation of the urogenital sinus from the hindgut by the urorectal septum have been proposed as a cause for this anomaly [1,9]. This leads to the total absence of all three components of penile shaft ie, both corpora cavernosa and spongiosum [10].

In its classic presentation, aphallia includes 46 XY karyotype, complete absence of the penis

with urethra opening at any point on the perineum in midline, a normal scrotum, normal and frequently undescended testis [9]. The differential diagnosis of aphallia includes rudimentary penis, micropenis, concealed penis, intrauterine amputation of penis epispadias, hypospadias and pseudo hermaphroditism [5,11,12]. Aphallia may also be present as a component of a more severe complex malformation syndrome that includes the perineal area, such as sirenomelia, cloacal exstrophy or the urorectal septum malformation sequence [13].

Skoog and Belman classified aphallia into three types as presphinctic, postsphinctic and urethral atresia regarding urethral opening position [14]. Patients can present at birth but usually at a later age due to social stigmata associated with aphallia. In the past these infants with aphallia were changed phenotypically. They underwent feminizing genitoplasty with bilateral orchiectomy, urethral transposition, vaginal replacement and labial construction. Although satisfactory cosmetic outcomes have been reported [15], the majority of these patients demonstrated typical shift in psychosocial and psychosexual development towards masculine character over long term follow up [16,17] as a result of male type of genetic imprinting due to normal levels of testosterone in these patients. Patient's gender identity is formed after the second year of life hence several authors have advised masculinizing operations at appropriate age to maintain harmony between sexual appearance and psyche [18,19].

An analysis of neophalloplasty series reveals identification of two major groups. One consists of microsurgical techniques, which are complex procedures, takes longer operative time, and require multidisciplinary teams. These procedures include concomitant

prosthesis insertion and are designed for surgery at adulthood. The second group consists of non-microsurgical techniques, including flap techniques. These techniques have the advantage of simplicity and feasibility to be performed at younger age, but are necessarily planned according to a two-staged strategy, combined with prosthesis implantation in adolescence [6].

In conclusion, aphallia is a rare malformation usually associated with other congenital anomalies. Management includes teamwork of an urologist, a pediatrician, an endocrinologist, a geneticist, and a mental health expert. The general consensus is that the sex assignment should be done on the basis of the karyotype of the child. Constructing a functioning phallus is still a major surgical endeavor. Delaying the sex assignment until the child spontaneously declares the sexual identity is an acceptable option.

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