

Aphallia: A Rare Congenital Anomaly

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Abstract

Aphallia or absence of penis is a very rare congenital anomaly. Clinical presentation is diagnostic, but immediate and long term management poses great dilemma. The issue like gender reassignment needs parental counseling. Due to social reasons parents have difficulty in giving consent for the classical management of gender reassignment. However with better penile reconstruction techniques, now there is hope for such parents who want to bring up their child as a boy. We present such a rare case with associated dilemmas.

Key Words

Aphallia, Congenital anomaly, Penile Reconstruction

Introduction

Aphallia or absence of penis is a very rare congenital anomaly. It is estimated to be occurring in 1 in 10 million live births. A total of about 80 cases have been documented in the literature so far (1). Clinical presentation is diagnostic, but there are associated various other ano-rectal anomalies and other systemic anomalies which needs to be investigated. Both immediate and long term management of the patients of aphallia pose great dilemma. We present here a case of aphallia and its management. Also discussed in brief are the dilemmas associated with the management of such patients.

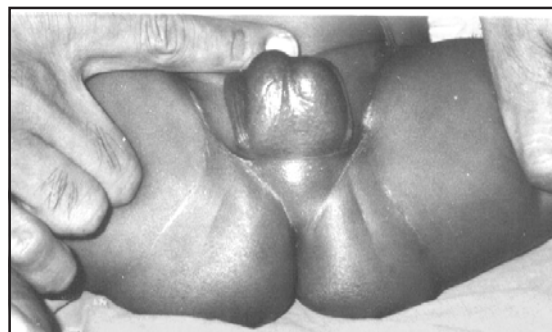
Case Report

A 6 month old was brought to the OPD by mother with complaint of absence of penis since birth and passing urine per rectally. Patient was healthy child with well developed scrotum and bilateral normally descended testes (Fig1). Anal opening was anteriorly placed and no urethral opening was visible in the perineum or anal verge (Fig 2). During examination squirt of urine was seen coming from the anal opening. There was no other identifiable external anomaly. Ultrasound abdomen & pelvis showed normal internal anatomy and no female internal genitalia. Ultrasound scrotum showed presence of both normal testes & epididymes. Karyotyping was not done in view these findings. Voiding cystourethrogram (VCUG), done by suprapubic puncture and instillation of contrast, showed normal capacity bladder and a thin tract

Fig 1: Showing Absence of Phallus with well Developed Scrotum and Fully Descended Testes



Fig 2: Showing no Urethral Opening in perineum & Anteriorly Placed Anal Opening

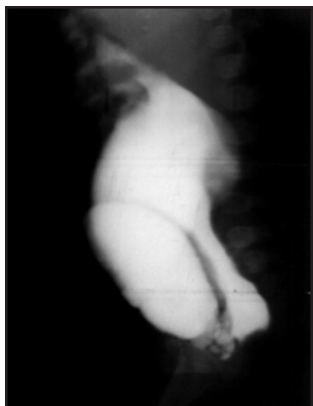


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Fig 3: Micturating Cystogram (lat. view) Showing Urinary Flow into Rectum & Sigmoid Colon



(Prostatic urethra) opening into the rectum, filling rectum and sigmoid colon with dye (recto-vesical fistula) (Fig. 3). The treatment options were discussed with parents and female gender reassignment was decided. Bilateral Inguinal Orchiectomy and Urethral Perineal transposition was performed. Feminine genital reconstruction was planned for the later date.

Discussion

Aphallia is a very rare congenital anomaly with an estimated incidence of 1 in 10 million. Eighty cases have been reported in literature so far (1). The anomaly develops as a result of failure of development of genital tubercle into phallus with absence of all three components namely both corpora cavernosa and spongiosum. Karyotype is typically XY. The scrotum is usually normally developed. The testes are usually normal with normal testosterone production in response to human chorionic gonadotropin stimulation(2). Urethra opens near anal verge (post sphincteric) under a skin tag or less commonly into the rectum (presphincteric, as in our case).

Other congenital anomalies like cryptorchidism, renal agenesis/dysplasia, musculoskeletal and cardiopulmonary anomalies are also common (>50% cases), hence evaluation of patient for internal anomalies is mandatory(3, 4). The treatment, classically, is multi staged, and consists of female gender reassignment. Bilateral orchiectomy urethral perineal transposition and feminizing Genitoplasty is done in new born period / at presentation. Vaginoplasty is done at older age (5, 6). Hormonal therapy is an integral part of management, starting at the expected time of puberty and continuing life long.

Recently the concept of in-utero gender imprinting of brain and long term psychological effects of gender conversion has gained acceptance among clinicians (7).

Also the phallus construction has been successfully done by some surgeons. So the treatment now is individualized according to age at presentation, rearing sex of child and psychological evaluation of child by pediatric psychologist and acceptability of the family (8).

In our case the patient had presented at 6 months. Both classical and newer treatment approaches were discussed with the parents. The parents initially were reluctant for gender reassignment and wanted to maintain male gender and phallic construction. But keeping in view, the future socio sexual aspects of child and fertility, they decided for female gender reassignment. As the social bias for male gender is still very high in Indian families, it is difficult for the parents to decide for the gender reassignment. Hence the parents need lot of counseling. The female gender reassignment is easier and the child can have a normal sexual life. Whereas phallus construction to maintain male gender has high failure rate when it comes to erectile function, hence these children may have sexual problems in their latter life. This aspect should be discussed with the parents in detail so as to resolve their dilemma regarding the treatment and their decision should be respected while planning the treatment of their child. This further stresses the need for counseling and individualizing the treatment of the patients with aphallia.

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