

Case Report

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A Rare case of Aphallia

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Introduction

Aphallia is a rare disorder with multiple associated anomalies. It is found in one in 20-30 million live male births. Fewer than 100 cases have been reported in the literature. More than 50% of the cases are associated with anomalies. Its management requires a stepwise approach. In addition, gender reassignment should be performed preferably before puberty, taking into account psychological and psychiatric evaluation and counseling. The most challenging issue is penile reconstruction. Several reports of neophalloplasty techniques in infancy have been published in the last 10 years. Review articles

Aphallia (total absence of penis) is an extremely rare abnormality that can be part of the urorectal septum malformation sequence.

We are reporting a 40-day-old boy who was referred to our nephrology clinic due to the absence of the penis and urinating through the rectum. He was born to a 17-year-old mother and a 24-year-old father, and was delivered term via normal vaginal delivery.

The pregnancy was uncomplicated with no maternal toxin or medication exposure. Both parents were healthy and there was no family history of congenital abnormality. The parents were also unrelated.

Physical examination revealed agenesis of the penis, a normal scrotum, and bilateral normally positioned testises. Moreover, the heart, lungs, abdomen, head and neck, and spinal column were all normal on examination. The karyotype was 46XY and the gender was male. Initial ultrasonography one week after birth revealed moderate bilateral hydronephrosis but the last ultrasonography 45 days later revealed only mild fullness of both kidneys.

Keywords: Aphallia; Gender; penile agenesis.

Running Title: A Rare case of Aphallia

have been reported on this topic in recent years. [1]. It was first described by Imminger in 1835 and only 100 cases have been reported to date [2]. Most of the affected cases have 46 XY karyotypes and therefore early gender reassignment is recommended to prevent consequent psychological impacts [3]. Aphallia is the results of either nonformation of the genital tubercle or its failure to develop normally during the 4th week of gestation; hence, it is frequently associated with other malformations. The urethra is frequently opened at the anal verge near to a small skin tag; in other cases, it opens directly into the rectum [4]. According to the kind of the relationship

between the urethral orifice and the anal sphincter, several observations have been mentioned. More proximally situated meatuses have a higher incidence of other associated anomalies and greater rate of neonatal death [5]. A diagnosis is made clinically by physical examination, which shows a completely absent penis, abnormal urethral opening, and normally developed gonads inside a well developed scrotal sac [6]. Penile agenesis (PA) can be classified in two major groups: solitary malformation of PA, and a complex malformation that is so severe that is often incompatible with life [7]. More than 50% of these patients have associated anomalies including genitourinary (54%) and gastrointestinal tract anomalies, or developmental defects of the caudal axis [8]. It is recommended to first treat life threatening associated anomalies. Definitive management of genital anomaly is controversial; it was recommended in the past to create a female gender reassignment by urethral perineal transposition, early bilateral orchiectomy and preservation of the scrotal skin, and feminizing genitoplasty in the neonatal period followed by vaginoplasty at puberty. However, the trend is now to focus on continuing the male sex gender using urethral transposition and performing delayed phalloplasty which is more difficult surgically [4].

Case Report

A 40- day-old infant (weight: 4200g, height 55cm) was born by normal vaginal delivery (NVD) to a 17-year-old woman (gravid 1, para 1, abortion 0) term at 38 weeks gestation in Urumie, northwest of Iran, without prenatal or peripartum difficulties. The parents were unrelated and healthy with no history of teratogenic drug or toxin exposure and no family history of congenital anomalies.

Physical examination revealed aphallia (Fig. 1), a normal scrotum, and bilateral normally positioned testes and vas deferens. The anus was positioned normally and the urethral opening was not visible anywhere in perineum. The heart, lungs, abdomen, and head and neck were all normal on examination. The infant was in an overall good condition and did not have any clinical evidence of other associated anomalies.

The results of laboratory investigations including liver and renal function tests, serum electrolytes, thyroid tests and FSH, LH, testosterone, DHEAS, ACTH, and hydrocortisone were in the normal range for age and sex.

Chromosomal studies revealed a male 46XY karyotype. His mother's prenatal ultrasonography revealed aphallia without any other anomalies. Ultrasonography one week after birth revealed moderate bilateral hydronephrosis but the last ultrasonography 45 days later revealed only mild fullness of both kidneys with a normal renal and parenchymal size without any anomalies in the bladder. Both testes were in the scrotum with a normal size and echo and without hernia or hydrocele. There was no phallus and cavernous tissue. The neck of the bladder was displaced posteriorly and the urine exited through the rectum. The abdominal and pelvic ultrasound examination showed normal internal anatomy with no evidence of the uterus or ovaries. There were no abnormalities on chest X-ray and echocardiography. Cystourethrogram (VCUG) by passing a small catheter through the urethral orifice inside the rectum showed a normal appearing bladder with a vesicorectal fistula opening to the anterior wall of the rectum without any reflux (Fig. 2). The pediatric urologists at our clinic suggested a sex change to female with genitoplasty, but his family did not agree. Therefore, we recommend a prophylactic antibiotic regimen to prevent UTI (because of the presence of kidney fullness) and follow-up.



Figure 1. Aphallia in Physical examination

Discussion

One of the rare complex anomalies of the urogenital system that has a severe clinical and psychological impact on the child and parents is aphallia [9]. It is a highly stressful situation for the parents to have a newborn baby with aphallia, and it is our responsibility to reassure them and suggest possible solutions like feminising genitoplasty or phalloplasty.



Figure 2. VCUG shows vesicorectal fistula opening to the anterior wall of the rectum

Feminising genitoplasty and bilateral orchiectomy before three years of age along with starting estrogen at puberty and continuous psychiatric support were recommended in the past [10]. It has been recently recommended to perform early phalloplasty to preserve sex characteristics. If the condition is not diagnosed until puberty, it is better to perform phalloplasty surgery to minimize the psychological burden. Finally, the ultimate choice rests upon parents at earlier ages but later on, it is not recommended to assign an opposite gender because of the karyotype and hormonal production [11]. Our plan for this baby is to follow him up along with urethral transposition and delayed phalloplasty according to the parents' decision.

Conflict of Interest

All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and the 1964 Helsinki Declaration and its later amendments or comparable ethical standards. Informed consent was obtained from all individual participants included in the study

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