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Aphallia: An Adult Presentation

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Abstract: Aphallia or Penile agenesis is a rare urogenital anomaly with an estimated incidence of 1 in 30 million. We report a case of aphallia in a male, who had two well-developed testicles, but lacked a penis. An earlier goal of treatment for such presentations was female gender reassignment and feminizing reconstruction of perineum, more recent goal being phalloplasty.
Keywords: Aphallia, Penile agenesis, micropenis

INTRODUCTION

Aphallia or Penile agenesis is a rare urogenital anomaly with an estimated incidence of 1 in 30 million [1]. The first case of penile agenesis ever reported was in 1853 by Imminger [2], since then a number of cases have been reported [3].

Aphallia is an anomaly that occurs during the fourth week of embryonic development and is related to result from non formation of the genital tubercle or its failure to develop. Its diagnosis is easy at birth due to absence of penis [4]. Penile agenesis are found to be associated with other genitourinary anomalies like vesicoureteral reflux, cryptorchidism, horse shoe kidney, renal agenesis and imperforate anus [5]. Apart from these it should be differentiated from some congenital deformalities like severe hypospadias, severe epispadias, intrauterine penile amputation, pseudohermaphroditism, concealed penis and micropenis [6].

CASE REPORT

A 21year old patient came to the hospital with complaints of absence of penis and passing urine from the anal region. On examination patient had an absent penis with normal looking scrotum and bilaterally descended and well developed testis (Fig. 1). Perineum was normal and no opening was seen. Patient did not have any dysmorphic features or clinical features of any other anomalies. Anus was normally placed and an opening was noted on the external anal sphincter (Fig. 2). A 6 french infant feeding tube was passed through the opening and clear urine was drained (Fig. 3). Later for further evaluation of the patient was performed by various investigations. Karyotyping was done showing 46 XY typing (normal male karyotype). An Intravenous pyelogram (IVP) was done to look for associated anomalies of the kidneys- IVP revealed normal position and function of both kidneys (Fig. 4). Colonoscopy was done (Fig. 5).

Figure 1: Physical examination showing two well developed testicles and pubic hair with complete absence of penis

Fig. 2: Urethra opening anterior to anal verge

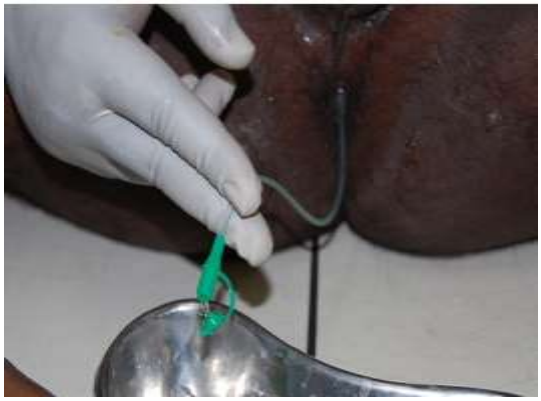

Fig. 3: A 6F feeding tube inserted in to the urethral opening and draining clear urine

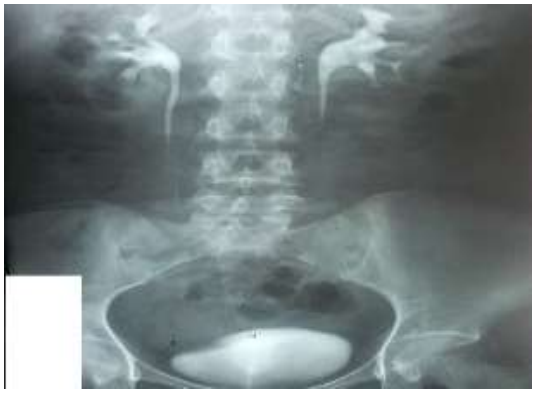

Fig. 4: Figure showing normal IVU

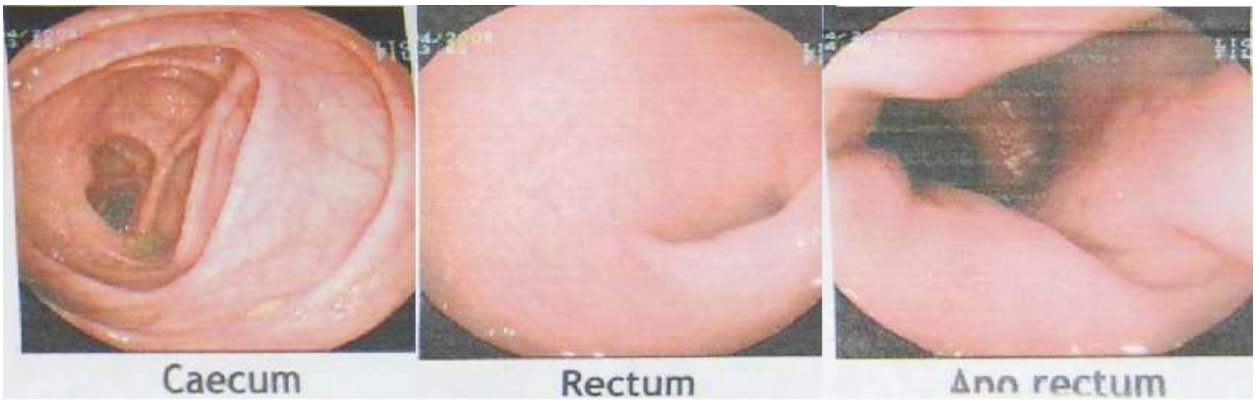

Fig. 5: Figure showing normal colonoscopy

DISCUSSION

Aphallia is a rare congenital anomaly with an estimated incidence of 1 in 10 to 1 in 30 million [7]. Aphallia develops as a result of failure of development of genital tubercle into phallus with absence of corpora cavernosa and spongiosum [8].

Aphallia is associated with communication of the urinary tract and rectum. Skoog and Belman [9] had classified aphallia into 3 types namely postsphincteric, presphincteric and urethral atresia [9]. Urethra opens near anal verge under a skin tag known as post sphincteric or into the rectum known as presphincteric which is less common [3]. It has been reported that a more proximal urethral meatus openings is associated with higher mortality rates [9].

The treatment consists of surgical treatment for female gender reassignment, bilateral orchiectomy in new born, vaginoplasty and estrogen therapy in adulthood [10]. Hormonal therapy is an integral part of management [3]. Concept of in-utero gender imprinting of brain and long term psychological effects of gender conversion has also been undertaken [11]. Phallus reconstruction and urethral reconstruction has been done successfully by surgeons [12].

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