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Case Report
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Isolated Aphallia: A Case Report and Literature Review

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Abstract

Background: Aphallia is an extremely rare congenital disorder characterized by the absence of a penis. When realized at birth, parents and close family members are left devastated. Even if speedy counselling is given, parents of newborns may take time to come about to a decision.

Case Report: A 2-month-old male infant was referred with complaints of an absent penis. Physical exam revealed normal scrotum and testicles and no congenital anomalies. The anal opening was located normally, but the urethral opening was not visible anywhere in the perineum. Laboratory exams and ultrasonography were normal. The infant was delivered vaginally at home and the mother had not received any antenatal care.

Conclusion: Aphallia results from failure of the genital tubercle to develop, leading to the absence of all penile tissue. Diagnosis includes absence of phallus, male karyotyping and normally developed scrotum with normal testicles. Managing this condition is difficult and requires multidisciplinary approach. Psychological counseling and surgery (in the method of gender reassignment or phalloplasty) are the cornerstones of treatment. Despite counseling and referral to a high-level hospital, the infant was lost to follow-up, highlighting the challenges of managing congenital anomalies in resource-limited settings.

Keywords: Aphallia; Absence of penis; Resource-limited countries

Background

Aphallia, also called agenesis of the penis, is an exceptionally rare genitourinary anomaly occurring in about 1 in 30 million live births [1]. When seen at birth, it leaves the parents and close family members devastated. Even if immediate counselling is done, it may take time for parents of neonate to come about to a decision. Aphallia was initially described by Imminger in 1853, and currently, less than 100 cases have been reported worldwide [2]. Aphallia is caused by the failure of the genital tubercle to form or to fully develop. Clinical presentation is diagnostic; associated congenital anomalies involving both the genitourinary and other major organ systems are common [3]. This article presents a case of aphallia in a 2-month-old male infant and discusses the challenges of managing this condition in low-resource settings.

Case Presentation

A 2-month-old male infant was referred to our hospital for genital evaluation. The infant was borne-to-a-mother of five at home *via* spontaneous vaginal delivery. His mother was a 35-year-old multigravida who had not received any antenatal care. Her pregnancy and delivery were uneventful. The mother and her family lived in a remote area with limited access to healthcare services. The infant was taken to a local health center, from where he was referred to a secondary level regional hospital.

On arrival to the hospital, patient was healthy looking with no syndromic features. Physical examination showed a well-developed infant with appropriate milestones for his age. On examination of the genitourinary system, he had complete absence of a penis. The scrotum was well-developed and the newborn had bilaterally descended, well-developed testicles and normal vas. The anal opening was located normally, but the urethral opening was not visible anywhere in the perineum (Figure 1). The urethra opens to the rectum since the infant passed urine through the

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Figure 1: Aphallia with normal scrotum and testes.

anus.

The infant was sent for routine laboratory exams and ultrasound examination of the abdomen and perineum. Karyotyping was not available. Renal function tests and hormone analysis were normal. Ultrasonography of the abdominal organs was ordinary with normal-sized kidneys and no hydronephrosis and absent uterus.

A diagnosis of aphallia was made and the mother was counseled about the condition and referred to a tertiary hospital. However, she took the child home without visiting the referral hospital stipulating to raise the infant as a boy and for superstitious reasons.

Discussion

Aphallia is a rare congenital anomaly with reported incidence of 1 in 10 to 30 million live births [1]. It causes considerable bodily and psychological implications for the affected individuals. Aphallia arises due to an impairment in mesenchymal proliferation, resulting in the failure of the genital tubercle to develop normally. During the 4th week of embryogenesis, the proliferation of mesenchyme around the cloacal membrane forms a pair of swellings called cloacal folds. Cranial to the cloacal membrane, the cloacal folds unite to form the genital tubercle. Under the effect of testosterone and dihydrotestosterone, the genital tubercle enlarges and becomes cylindrical to form the phallus or primitive penis. Additional enlargement of the phallus forms the penis. The urethra may open at any point along the perineal midline raphe but most commonly opens onto the anterior wall of the anal verge [4].

In the more severe cases with associated malformations, the problem may stem with a defect in cloacal differentiation during development as a defect of blastogenesis. Isolated aphallia, without any malformation, is very rare. The diagnosis of aphallia can be made in the absence of the phallus, male karyotype, and normally developed scrotum with normal testicles. It should be differentiated from micropenis, rudimentary penis, penile amputation, concealed penis, and male pseudohermaphroditism.

Position of urethral opening depends on the type of aphallia. There are three categorizations described [5]. One is Evan's classification of prognosis that is based on the presence or absence of congenital malformations. Two is a classification is based on a disorder of sexual development [5]. Three is Skoog's anatomical classification as pre-sphincteric (urethral opening inside rectum), post-sphincteric (urethral opening between perineum and pubis) and urethral atresia (which presents as vesico-rectal fistula) [6].

Management of aphallia depends on management of associated anomalies, family background and socioeconomic status of the family. It also involves a multidisciplinary approach that includes counseling, hormonal therapy, and surgical interventions such as phalloplasty or gender reassignment surgery. The traditional mode of managing penile agenesis, until the end of 20th century, was to give female gender to the patient and perform multi-staged reconstructive procedures towards feminizing genitoplasty. Recently, however, concerns related to the long-term psychological effects of gender conversion such as gender dysphoria increased owing to prenatal androgen imprinting. This have led to recommendations that patients with aphallia should be supported surgically as a male until child is old enough to gender identify. It also has to be based on patient's karyotype and hormone production. The management decisions have to be made carefully, taking into consideration the age at presentation, rearing sex of the child (in cases of late presentation), psychological evaluation of the child, and acceptability of the family [7].

Managing congenital anomalies such as aphallia can be difficult in low-resource settings due to various challenges including limited access to healthcare services, social stigma, lack of awareness about the anomaly, and financial constraints. Diagnostic studies including karyotyping and imaging studies are difficult to perform in resource-limited settings.

The case reported is unusual because there were no other anomalies identified, the anus was in normal place and urethral opening was much away from the anal verge and into the rectum.

The patient lived with his parents and family in a remote area with limited access to healthcare services. Despite counseling and referral to a tertiary hospital, the patient was lost to follow-up, highlighting the challenges of managing this condition in low-resource settings.

Conclusion

Aphallia is a rare congenital anomaly that requires a multidisciplinary approach to management, and it can have life-changing effects on affected individuals. In low-resource settings, there are numerous challenges to managing congenital anomalies such as aphallia, including limited access to healthcare services, financial limitations, and social stigma. Despite these challenges, healthcare professionals must provide appropriate counseling, referral, and management options for people affected by aphallia in order to improve their quality of life.

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