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CASE REPORTS

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Isolated aphallia: a case report and review of literature

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Abstract

Background Aphallia is a rarest of rare congenital anomaly the incidence being 1 in 10 to 30 million live births. Seen at birth, it leaves the parents and family disturbed. Immediate counselling though done; it may take time for the parents to come to a decision.

Case presentation A newborn was brought for genital examination and since the penis was not present and the scrotum with testes were normal, aphallia was suspected. At 1 month after all the necessary investigations, isolated aphallia without any other congenital anomalies was confirmed. The options offered were gender reassignment before the age of 18 months or staged phallo and urethroplasty. Parents are so far undecided.

Conclusions Aphallia results from failure of the genital tubercle to develop, leading to the absence of all the penile components. Diagnosis of aphallia includes the absence of the phallus, male karyotype and normally developed scrotum with normal testicles. Usually, this anomaly is associated with multiple other anomalies involving the urinary, gastrointestinal and musculoskeletal systems. Management of this condition is challenging and requires a multidisciplinary approach. In addition to managing the associated anomalies, the options to treat aphallia will depend on the type, severity of associated anomalies, family background and socioeconomic status of the family. Psychological counselling along with surgery (in the form of gender reassignment or phalloplasty) are the mainstays of treatment. The literature of this anomaly and the treatment is reviewed.

Keywords Aphallia, Phalloplasty, Congenital anomalies

1 Background

Aphallia is a rarest of rare congenital anomaly the incidence being 1 in 10 to 30 million live births. Seen at birth, it leaves the parents and family disturbed. Immediate counselling though done; it may take time for the parents to come to a decision.

2 Case presentation

A newborn infant was brought for genital evaluation. The baby was delivered by a caesarean section a day back. On examination, there was no penis. The labial folds had retractile gonads. The urethral meatus was under the scrotum and the anus was normally placed.

The baby was sent for routine laboratory reports, an ultrasound examination of the abdomen/perineum and karyotyping. At 1-month follow-up, the laboratory reports were normal; the ultrasound abdomen was normal for all abdominal organs and there was the absence of the uterus. The perineal ultrasound showed both testes normal and no penile structure. The Karyotype report was 46XY. The genital examination now showed that the scrotum was well developed, both testes and vas were palpable and normal and there was no penile tissue palpable (Fig. 1). The urethral opening was away

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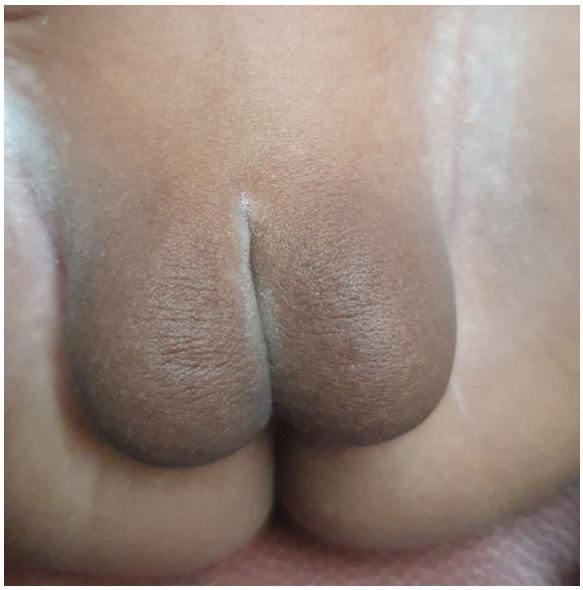

Fig. 1 Aphallia with normal scrotum and testes

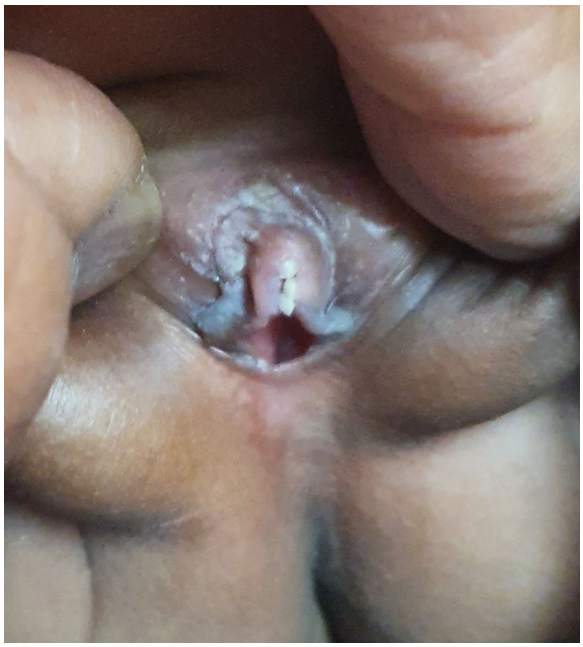

Fig. 2 Urethral opening below the scrotum with a dorsal skin hood

from the anus at the junction of the scrotum and perineum with a dorsal hood looking like a clitoris (Fig. 2). A diagnosis of aphallia was made and the parents were counselled for further treatment. The parents decided to rear the child as a boy and for superstitious reasons decided to wait before initiating any further treatment.

3 Conclusions

Aphallia is an extremely rare congenital anomaly. The incidence reported is 1 in 10 to 30 million live births [1]. The first case of aphallia was reported by Imminger in 1853 [2]. It results from failure of the genital tubercle to develop, leading to the absence of all the penile components. In the more severe aphallia associated malformations, the problem may originate with a defect in the induction of cloacal differentiation occurring in early embryogenesis as a defect in blastogenesis. A diagnosis of aphallia includes the absence of the phallus, male karyotype and normally developed scrotum with normal testicles. Occasionally, a small amount of erectile tissue may be present anteriorly [3]. Bindhu S. et al. reported a case of severe hypospadias with aphallia [4], but some penile tissue should be present to classify this as hypospadias. Usually in more than half the cases (54%), this anomaly is associated with multiple other anomalies involving the urinary, gastrointestinal and musculoskeletal systems. Isolated aphallia without any other anomaly is relatively rare. Location of the urethral opening depends upon the type of aphallia. Three types of classifications are described [8]. 1. Evans' prognostic classification based on the presence or absence of major associated anomalies. 2. Skoog's anatomical classification: Classified as pre-sphincteric, post-sphincteric and urethral atresia [3]. In the pre-sphincteric type, the urethral opening is inside the rectum, in post-sphincteric type it may be anywhere from the perineum to the pubis. The third type is urethral atresia which presents as a vesicorectal fistula. 3. As a disorder of sex development of the non-hormonal/non-chromosomal type [8]. Aphallia should be differentiated from micropenis, rudimentary penis, penile amputation, concealed penis and pseudohermaphroditism. Chakraborty reported a case where the urethral opening is at the pubis [6]. In such cases, micropenis or rudimentary penis should definitely be ruled out. The incidence of associated anomalies increases when the bladder outlet is more proximal. Skoog and Belman reviewed 60 cases of this anomaly and analysed the mortality depending on the type of aphallia. The post-sphincteric type has the least mortality of 13%, the pre-sphincteric type has a 36% mortality while urethral atresia has a 100% mortality.

Management of this rare disorder has been controversial. The reason being that the anomaly is extremely rare. In addition to managing the associated anomalies, the options to treat aphallia will depend on the type, severity of associated anomalies, family background and socioeconomic status of the family.

The spectrum of surgery ranges from feminising genitoplasty to staged penile reconstructive procedures. Till 1997, the recommended treatment was feminising genitoplasty because it was believed that 'raising these

patients as male can be disastrous', and that 'it is better to be incompletely female than inadequately male' [7]. However, these patients developed a high rate of gender dysphoria partly due to the prenatal androgen imprinting that had already taken place. It is now generally accepted that a normally genetic male with aphallia should be supported surgically as a male until the patient is old enough to gender identify.

Management can be divided into short, intermediate and long-term treatment phases: 1. Short term which comprises of treating any life-threatening complications if present. 2. Intermediate treatment comprises establishing a non-obstructing urethral tube along with a temporary phallus formation. 3. Long-term treatment comprises definitive phalloplasty and functioning urethroplasty. A successful neophallus should allow the patient to appear outwardly male, urinate standing up and perform adequately from a sexual perspective, usually with the help of a prosthesis. Phalloplasty should ideally be completed before puberty.

For phalloplasty, there are microsurgical and non-microsurgical procedures. Non-microsurgical are the abdominal, scrotal and groin flaps. The abdominal De Castro flap based on the superficial epigastric arteries is the most used [9] and it grows with time. Microsurgical procedures are complex with a much longer intraoperative time and necessitate multidisciplinary team management. They can be performed in adults [5] with concomitant insertion of a penile prosthesis. Whichever be the type of phalloplasty, the greatest challenge is establishing a successful urethroplasty.

However attractive the options of reconstructive surgery are, they have their pitfalls as the patient spends the better part of his life in hospitals undergoing surgery and fighting psychological issues [1].

Regarding gender, sexual designation should be directed to the gender that provides best prognosis in terms of reproductive function, capability of sexual function, normal appearance of external genitalia and self-identity with the gender. If gender reassignment is to be done, it should be instituted as early as possible (18 months) before the post-natal testosterone surge takes place as this would lead to the psychological imprinting of the child as a male.

Usually, these infants are lost for follow-up as the parents and family consider this a stigma and cannot decide on the treatment options offered to the child. The present case is unusual because there were no other anomalies detected, the anus was in the normal place and the urethral opening was much away and not at its verge. This case was offered option of phalloplasty or gender reassignment surgery at diagnosis but the family was undecided. The parents are waiting for a year anticipating a miracle through prayers and beliefs.

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Author contributions
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Declarations

Ethics approval and consent to participate
Written consent has been taken from the family. The Institutional ethics committee of M. P. Shah medical college, Jamnagar, India has informed that the approval for this case report is not required.

Consent for publication
Written permission from the parent of the child was obtained for presenting this case.

Competing interests
We confirm that there are no competing interests.

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