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Case Report

Penile Agenesis, Dysmorphic Features, Vesicoureteral Reflux and Rectovesical Fistula: A Rare Case Report

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Abstract

Aphallia, or agenesis of the penis, is a very rare congenital anomaly, with an estimated incidence of one in every 10-30 million births. More than half of aphallia cases have associated anomalies, including caudal axis, cardiovascular, genitourinary, and gastrointestinal anomalies. We present a case of aphallia associated with dysmorphic features, vesicoureteral reflux, and vesicorectal fistula.

Keywords: Aphallia, Penis, Development, Genitalia, Congenital anomaliesa.

Introduction

Aphallia, known as penile agenesis (PA), is a congenital anomaly in which the penis is absent [1]. It is a significantly rare anomaly of the genitourinary system, with an incidence of one in every 10–30 million births. Only a few reports of PA have been published so far [2].

PA is caused by an absence of the genital tubercle or failure in its development [3]. As a result, all three erectile columns of the penile shaft (the two corpora cavernosa and the corpus spongiosum) are completely absent [4]. The diagnosis of aphallia includes the absence of the penis, a male karyotype, and an abnormal urethral opening with a well-developed scrotum with bilateral palpable testes [3,5]. PA could either be an isolated anomaly or a complicated form with other associated congenital anomalies, which are usually incompatible with life [2].

In this literature review, we present the case of a patient with isolated aphallia associated with dysmorphic features, vesicoureteral reflux, and rectovesical fistula.

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Case

A neonate with aphallia was referred to the pediatric endocrine services for evaluation for any associated congenital anomalies. He was the first baby in the family who was born at term. The baby was delivered by spontaneous vaginal delivery and weighed 3 kg. The pregnancy was uneventful. No oligohydramnios was noticed during the follow-up period; and there was no history of use of any medication, ingestion of teratogenic substances, or exposure to radiation. The neonate was from a nonconsanguineous family with no history of a similar anomaly in the parents' families.

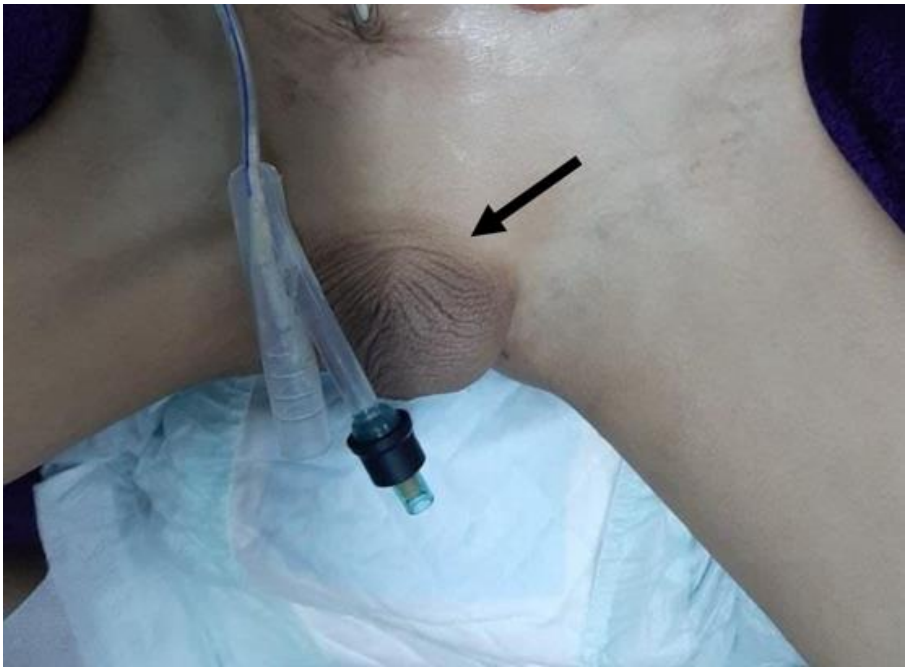

Figure 1: A 3 month of age revealed complete absence of the phallus, with a normal scrotum and bilaterally descended testes.

On general examination, the baby was found to have dysmorphic features: a triangular face, depressed nasal bridge, short philtrum, and long eyelashes. Examination of the external genital revealed complete absence of the phallus, with a normal scrotum, and bilaterally descended testes (Figure 1). No urethral opening was visible in the perineum, and urine was passed along with meconium through a perforate anus, which was normally situated (Figure 2). Examination of other systems was unremarkable, and no lower limb deformities were observed.

A micturating cystourethrogram (MCUG) revealed a rectovesical fistula and grade 4 right vesicoureteral reflux; karyotyping showed 46, XY karyotype; abdominal ultrasound showed no female internal organs; and scrotal ultrasound confirmed the presence of the testes. Future management plan will include urethroplasty with an opening in the perineum and phalloplasty after adolescence.

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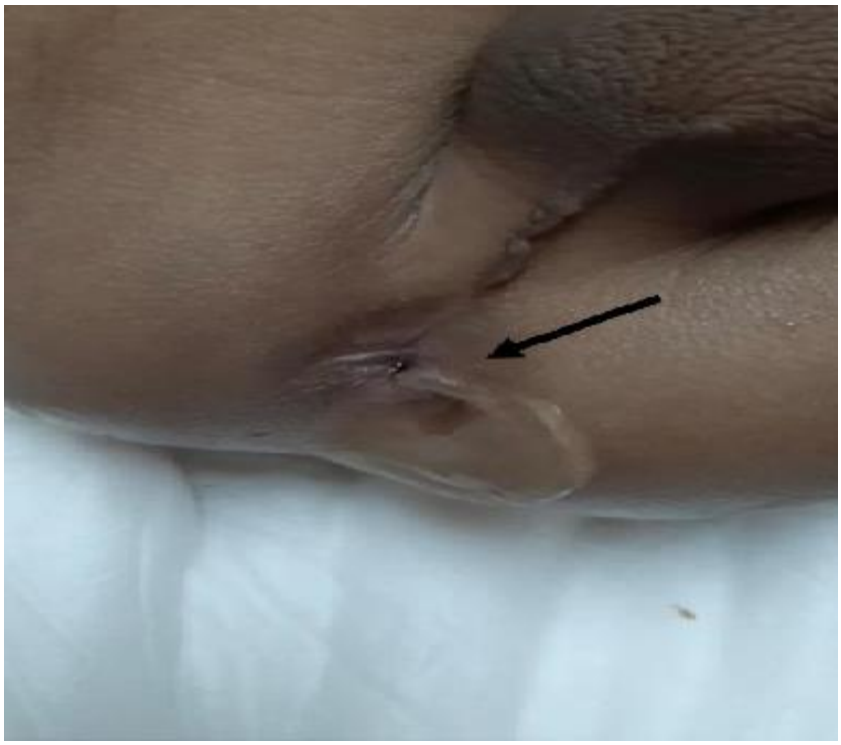

Figure 2: Urine passing through the anus due to absence of the penis.

Case Discussion

PA is characterized by the absence of the penis, as well as a urethral opening anywhere in the perineum, such as along the midline, on the anterior aspect of the scrotum, over the pubis, or anterior to the anus, or as an opening in the anterior wall of the rectum [5].

Dihydrotestosterone, which is produced from testosterone by the actions of 5-alpha reductase, stimulates the differentiation of the male external genitalia and the development of the urogenital sinus [4].

Embryologically, the penis is formed in the fourth week of gestation by the union of the labioscrotal folds as the genital tubercle expands and elongates due to testicular androgens. The solid epithelial plate is canalized, and this leads to the formation of a groove on the surface of the genital tubercle, which is bounded by the urethral folds. The urethra folds fuse to form the penile urethra [6].

PA is mainly caused by defects in the formation of the genital tubercle, or its inability to develop into a penis.

Althogh the diagnosis of PA is clinical, it is important to differentiate it from severe hypospadias, severe epispadias, intrauterine penile amputation, 46, XY disorder of sex development, concealed penis, and micropenis [5].

It has been estimated that more than 50% of patients with aphallia also have other anomalies, which may be incompatible with life. Such anomalies may include cardiac, genitourinary, gastrointestinal tract anomalies, and other developmental defects of the caudal axis [1].

The most common accompanying anomalies from our literature review are genitourinary anomalies (54%) [3]. Only few cardio-respiratory related anomalies have been reported, due to the high mortality rate associated with them.

The urethral opening is usually found in the perineum, between the scrotum and the anus, or as a fistula, opening into the gastrointestinal tract, most commonly the rectum [5].

Skoog and Bellman found that neonatal mortality and the incidence of other anomalies are higher when the meatus is more proximal [7]. They classified the relationship between the anal sphincter and ectopic urethral meatus into three categories: the post-sphincteric form, with an anterior perianal urethra; presphincteric urethrorectal fistula; and urethral atresia with vesicorectal fistula [2].

We present a rare case of aphallia associated with dysmorphic features (triangular face, depressed nasal bridge, short philtrum, and long eyelashes), rectovesical fistula, and vesicoureteral reflux. This case is clinically important because of its unusual presentation.

In 2011, Gérard-Blanluet et al. reported a case of aphallia associated with renal dysplasia, imperforate anus, clubfoot, right lung agenesis, and dysmorphic features (low-set ears and depressed nasal bridge) [8].

In 2015, Sharma et al. reported another case of aphallia associated with dysmorphism (low-set ears and depressed nasal bridge), respiratory distress, single umbilical artery, imperforate anus, endocardial cushion defect, pulmonary hypoplasia, and bilateral agenesis of the kidneys [9].

In 2016, Amiri et al. also presented the case of a premature baby with aphallia associated with dysmorphism (low-set ears and depressed nasal bridge), respiratory distress, clubfoot, tricuspid valve regurgitation, thick-walled bladder with bilateral reflux, and atrophic kidneys [10].

In all these cases listed above, the dysmorphic features and some of the associated congenital anomalies, including pulmonary hypoplasia, renal agenesis, and congenital heart defects, are related to the Potter sequence, which results from oligohydramnios.

In 2009, in a report of eight cases, two had dysmorphic features. The first case had aphallia associated with a urethrorectal fistula, congenital agenesis of the prostate, a beaked nose, low-set ears, and a high-arched palate. The second case was associated with a urethral opening in the scrotal area, with a cleft lip and palate [11].

Aphallia is a complex urogenital and psychosocial problem, and treatment requires a detailed evaluation of both clinical and psychosocial factors [1].

Apart from correcting life-threatening anomalies, the management of aphallia has two options [5]: the first is female sex reassignment, which is technically simpler, and results in a normal functional sexual life. It includes urethral perineal transposition, early bilateral orchiectomy with scrotal skin preservation, and feminizing genitoplasty during the neonatal period or at the time of presentation., followed by estrogen therapy in

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adulthood for breast development and other female sexual characteristics [12].

The second option is the male sex assignment, which involves the reconstruction of a normally functioning penis [5] and is the method that will be applied in this case.

Although it is more surgically challenging and has a relatively high rate of failure over time, mainly due to erectile dysfunction [13], this is the only available option because, based on his karyotype (46, XY), physical examination findings of bilaterally descended testes, and ultrasound results, which showed no female internal organs, our patient was a male neonate. Therefore, performing female sex reassignment is not an option in view of our Islamic rules. According to the fatwa given by the Islamic Fiqh Council under the Muslim World League, "changing sex, whether male to female or vice versa, is illegal because it amounts to changing the creation of Allah; except for abnormal sex organ development, where surgical intervention is considered as correcting a birth defect, rather than changing the organized nature of Allah" [14].

Moreover, Islam forbids homosexuality, and altering the external appearance of the male genitalia into that of the female would not change the fact that he is genetically male; taking into consideration some of the sex-specific Islamic rules, which require a distinguished application of certain laws based on sex. An example is the different portions of inheritance that a son and a daughter receive from their father's possession. Sex reassignment would violate this rule [15].

Conclusion

Supporting the Islamic rules Meyer-Bahlburg examined the long-term outcomes of 46, XY female-raised patients with PA, penile ablation, and cloacal exstrophy, and discovered that all patients raised as males continued to live as males (n=311), while 22% of those raised as females changed their sex, and 13% had evidence of sex dysphoria (n=77) [16].

Declaration Statements

This paper is the original work of the authors and not copied, in whole or in part, from any other work. The authors declare that this work has not been previously published or under simultaneous consideration by another journal.

Declarations of Interest

None.

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Author Contributions

Writing-original draft preparation, LA, and RA; writing-review and editing, LA, and RA and AA; supervision, AA; project administration, AA.

All authors have read and agreed to the published version of the manuscript.

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