Penile agenesis

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ABSTRACT

Penile agenesis is an extremely rare disorder with profound urological and psychological consequences. The goal of treatment is an early female gender assignment and feminizing reconstruction of the perineum.

KEY WORDS: Aphallia, Penile agenesis, Ambiguous genitalia

Penile agenesis (PA) is an extremely rare developmental anomaly with the reported incidence of 1 in 30 million births.[1] PA is believed to result from either the absence of the genital tubercle, or its failure to develop.[2] Several investigators claim the absence of corpora cavernosa and corpora spongiosum as a prerequisite for the diagnosis of penile agenesis.[3] Except for the reported XXXY mosaic, patients have 46 XY karyotypes.[4] More than half of these have associated anomalies, including developmental defects of the caudal axis, genitourinary and gastrointestinal tract anomalies.[5] The scrotum, testes and testicular function are usually normal.[2]

CASE REPORT

A two-day-old 3.2 kg genotypic male (46XY) neonate was brought, by a social organization, to our hospital with the complaint of absence of penis, and passage of meconium mixed with urine through rectum. On examination patient had an absent penis, with a normal looking scrotum and bilaterally descended testes [Figure 1]. Anus was normally placed and the urethral opening was not visible anywhere in the perineum. Patient did not have dysmorphic features or clinical evidence of any other associated anomaly. Urethra was later diagnosed, by cystography, to open up high in the anorectum anteriorly by a fistulous communication [Figure 2]. Ultrasound examination revealed normal looking kidneys and urinary bladder. Divided sigmoid colostomy was performed for fecal diversion, and patent urachus noticed during exploration was excised. Three weeks later, bilateral orchiectomy was done, urethreocolostal fistula disconnected and urethra mobilized and transposed anteriorly and placed between the scrotal folds which were preserved for subsequent genital reconstruction.

DISCUSSION

The earliest case report of aphallia was by Imminger in 1853[5] since then only 75 cases have been reported in the literature[6]. Skoog and Belman[5] suggested three variants, based on urethral position in relationship to the anal sphincter, as: Postsphincteric; Presphincteric (Prostatorectal fistula) and Urethral atresia. More proximal the bladder outlet, greater is the likelihood of other anomalies and death.[5]

Figure 1: Photograph showing an absent penis with normal scrotum and descended testes
The diagnosis of PA requires the absence of corpora cavernosa and copora spongiosum with urethra opening at any point on the perineum in midline, over pubis, anterior aspect of the scrotum, or, most frequently just anterior to the anus and anterior wall of the rectum. This rare entity should be differentiated from concealed penis, rudimentary penis, micropenis, epispadias, hypospadias pseudo hermaphroditism and intrauterine amputation of penis. Anorectal anomalies such as imperforate anus, congenital rectal strictures and rectovesical fistula, cryptorchid testis, hydrocele, hernia, renal dysplasia, horseshoe kidneys and agenesis of prostate could be associated malformations.

In view of completely normal testicular hormone function in PA, Oesch et al emphasized the need for an early bilateral orchiectomy to prevent psychological imprinting of the child as a male because of the postnatal testosterone surge that occurs between the 10th and 60th day of life. Stolar et al were the first to use posterior sagittal approach and recommended orchiectomy and diverting colostomy with or without vesicostomy before three months of age, followed by repair of the urethrorectal fistula and vaginoplasty at one year of age.

There is a consensus that infants with PA should undergo early gender assignment and be raised as girls, despite male karyotype. However, its relevance to the Indian society is still debated. In Indian villages and towns it would be easier to live as a sexually incompetent male in the society rather than a single unmarried girl. Delay of diagnostic and therapeutic measures resulting in male gender assignment has led to severe psychological and anatomic problems because of difficulties in constructing a functioning phallus. In contrast, it is easier to establish a normal female appearance. However, no reports concerning long term physiological and psychological results exist in immediately gender reassigned patients. Perhaps they are as dissatisfied as girls as they would have been as boys.

REFERENCES
Editorial comments

Gender assignment is a common dilemma in newborns with aphallia. Though in most cases, the urethra is short and opens in the anorectum yet it may open in the perineum with good urinary control. Female gender has often been assigned for technical reasons and also for the belief of psychosexual neutrality at birth with female genitoplasty.

Patients with aphalia are truly males and are expected to behave so with male mental makeup in their adult life. If they are assigned a female gender, the child may not be expected to behave normally in society due to abnormal sexual orientation. The brain gender makeup has been currently proved to be affected by antenatal androgens which can not be altered by psychosexual and endocrine treatment. Also the long term results in patients converted to females are not available in the literature.

Henceforth, the present consensus is that patients affected by aphalia are better raised as males. Recently, successful phallic reconstruction in two patients with aphalia were presented from Bologna (Italy), using the lower abdominal wall skin flap for making the shaft and the bladder/labial mucosa free graft for making the urethra. The procedures were completed at 9 and 17 months of age.

However, all is not that well. Male genitoplasty in such cases requires a long urethroplasty and it is not supported by corpus spongiosum. Due to lack of urethral resistance, the long and the short term results of the long urethroplasty are not satisfactory. Patients are not able to expel urine at the meatus in a forceful stream. This problem was encountered in one out of the two cases from Bologna and a scrotal urethrostomy had to be resorted to later on. We also had the similar experience in one case in whom male genitoplasty was performed.

The initial management should include dismantling of the urethra from the anorectum and placing the same in the perineum (as preliminary urethrostomy). This may be done even during the newborn period or infancy so as to avoid urinary complications in future. One should not allow the child brought early to go without a urethrostomy as death due to chronic renal failure has been reported. If the parents are not sure of the gender, they may be given time to think it over.

In Indian society, the family accepts an inadequate male better than an incomplete female along with psychosocial disturbance. If a male gender assignment is planned, the phalloplasty should preferably be done after puberty as the results following phalloplasty performed in infancy and childhood may not be satisfactory due to inadequate growth of the reconstructed phallus. If the child presents late after puberty or the parents had refused treatment in the past, as may be the case in our country, phallic and urethral reconstruction should be done with the hope to achieve satisfactory results.

In our experience, three cases of aphalia presented during the newborn period, in infancy and at 7 years of age (with urethra opening in the anorectum in the last case). One seen during the newborn period was converted to female gender. The second case was from Madagascar (Africa) and insisted only for male gender assignment. In this case, a successful but a smaller sized phalloplasty using lower abdominal wall flaps was performed. The urethra is short but quite effective in expelling the urine. There were no backpressure changes noted during follow up at 2 and 5-year age. The third one did not agree to female sex assignment and refused treatment.

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