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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 XX-XY 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, urethrorectal 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[2] 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]

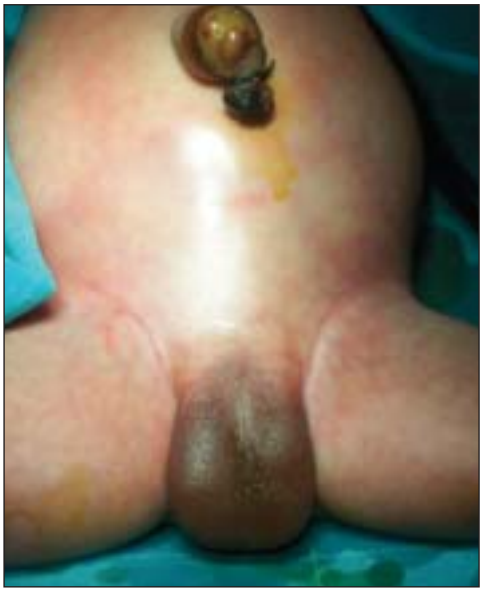

Figure 1: Photograph showing an absent penis with normal scrotum and descended testes

Figure 2: Cystogram depicting urethral fistulous communication with the rectum

The diagnosis of PA requires the absence of corpora cavernosa 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.[7,8] This rare entity should be differentiated from concealed penis, rudimentary penis, micropenis, epispadias, hypospadias pseudo hermaphroditism and intrauterine amputation of penis.[2,8,9] 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.[4]

In view of completely normal testicular hormone function in PA, Oesch et al[9] 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.[2] Stolar et al[10] were the first to use posterior saggital approach and recommended orchiectomy and diverting colostomy with or without vesicostomy before three months of age, followed by repair of the urethro - rectal 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.[3,11] 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.[12] 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.[3] 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.[13]

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