A rare case of penis agenesis (Aphallia) with associated multiple urogenital anomalies

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A B S T R A C T
The penis as a component of external genitalia, takes part in fertility, urinary and psychosexual structure of males with its complex character. We report a case of penis agenesis with associated left renal agenesis, left superior segment ureteral agenesis, prostate agenesis, left ureterocele, right vesicoureteral reflux and high urethrorectal communication above the rectal sphincter. The patient refused any surgical intervention because of his religious beliefs.

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1. Introduction

The penis agenesis (PA) is an extremely rare genitourinary anomaly with an incidence of about one in 30 million births [1,2]. It is believed to result from the absence or a failure in the development of genital tubercle [3]. Since it was originally described by Imminger in 1853, approximately 100 cases have been described worldwide. PA can be classified in two major groups. First one is the solitary malformation of PA. Second one is the complex form which is often incompatible with life because of accompanying congenital anomalies [4,5]. More than half of these patients have associated anomalies including genitourinary (54%) and gastrointestinal tract anomalies and developmental defects of caudal axis [6].

Skog and Bellman [6] have classified these patients based on the relationship between the anal sphincter and the ectopic urethral meatus. Three variations were described by them: the first one is post sphincteric form with anterior perianal urethra, the second one is presphincteric urethreorectal fistula and the third one is urethral atresia with vesicorectal fistula. Urethral opening could be either over pubis or at any part of perineum or most frequently in anterior wall of the rectum [6,7]. Other anomalies and death are seen related with more proximal opening of the bladder outlet [6].

The treatment of PA is under debate. Historically, gender reassignment at infantile period of life has been considered as the most appropriate choice for these patients [8,9]. Surgical intervention included urethral transposition and bowel vaginoplasty via posterior sagital approach plus scrotoplasty and orchiectomy. Although adequate outcomes have been reported [10], the majority of these patients demonstrated male-typical shift in psychosocial and psychosexual development in long time follow up [11,12].

2. Case report

A 22 year-old patient was referred to us due to absence of penis and urination through rectum. His history included recurrent suprapubic and right flank pain and persistent urinary tract infections. Physical examination revealed agenesis of penis, normal scrotum, bilateral normally positioned testis and vas deferens, well male secondary sexual characteristics, normally located and at tone anus and anal sphincter. Prostate gland was not palpable at his digital rectal examination (Fig. 1).

Ultrasonography showed the absence of left kidney, superior segment of ureter and prostate gland. Left ureterocele, increased bladder wall thickness due to persistent urinary tract infections and dilated right ureter were also seen. Buccal smear was consistent with male genotype, and chromosomal studies revealed a normal karyotype 46XY. All serum hormone levels were normal for an adult male.

Cystoureterogram performed through rectosigmoidoscopy showed normal appearing bladder, a vesicorectal fistula opening
to the anterior wall of the rectum, left ureteroceles and grade 2 right vesicoureteral reflux (Fig. 2).
99mTc-Dimercaptosuccinyl acid (DMSA) renal scintigraphy, computerized tomography (CT), urography and magnetic resonance imaging (MRI) were performed in order to plan the surgical intervention. These imaging procedures also confirmed ultrasonographic and cys-tourethrogramic findings (Fig. 3).
Although the patient and his parents were informed about the masculinizing operation, they refused any surgical intervention because of their religious beliefs. We acquainted the patient with the possible health problems and invited him to regular follow up.

3. Discussion

The embryology of this rare anomaly has been described as a result of the genital tubercle failure to develop with incomplete separation of the urogenital sinus from the hindgut by the urorectal septum [1,6].
The classic PA diagnosis includes 46 XY normal male karyotype, complete absence of the penis, a normal scrotum, normal and frequently undescended testis. Urethral opening can be located in the perineal area, mostly inside a foreskin appearing skin tag or the anterior wall of the rectum [6].
PA must be differentiated from rudimentary penis, micropenis, concealed penis, interuterine amputation of penis and disorders of sexual development [13]. Patients usually present at birth. In the past these infants with PA underwent gender reassignment surgery, including bilateral orchiectomy, urethral transposition, vaginal replacement and labial construction. Although adequate outcomes have been reported [10], the majority of these patients demonstrated male typical shift in psychosocial and psychosexual development in a long time follow up [11,12]. Since the patient’s gender identity is formed after the second year of life several authors have advised to perform masculinizing operations, in order not to disturb the patient and his parents psychologically as we have advised in our case [5,14,15].
In conclusion, treatment of PA presents many challenges and it involves multidisciplinary approach. The team should include a urologist, a pediatrician, an endocrinologist, a geneticist, and a mental health expert. Some patients and parents may refuse sur-

Fig. 1. Penis agenesis, bilateral normal testis and scrotum (A), perineal and anal examination (B,C).

Fig. 2. Grade 2 right vesicoureteral reflux (A), vesicorectal fistula and left ureterocoele (B), distal part of left ureter (C).

Fig. 3. Left renal agenesis (A), agenesis of prostate (B), vesicorectal fistula opening to anterior wall of the rectum (C).
gical intervention. In this case, they should be informed about the devastating outcomes of this anomaly and invited to regular follow up controls by the urologist.

Disclosure

We declare that the authors have obtained informed consent from the patient to report this case.

Conflict of interest

No conflict of interest was declared by the authors.

Financial disclosure

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