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Case Report / Olgu sunumu

Penile Agenesis

Penil Agenez

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Abstract

Penile agenesis is a rarely seen congenital anomaly with dramatic psychological outcomes. In more than half of the patients, other genitourinary anomalies can accompany penile agenesis. We herein present a newborn with rectovesical fistula associated with vesicoureteral fistula.

Keywords: Penile agenesis, rectovesical-fistula

Öz

Penil agenezi dramatik psikolojik sonuçları olan oldukça nadir konjenital bir anomalidir. Hastaların yarısından fazlasında diğer genitoüriner anomaliler eşlik edebilir. Biz burada veziko-üreteral reflünün eşlik ettiği ve rekto-vezikal fistülü olan bir yenidoğanı sunduk.

Anahtar Kelimeler: Penil agenezi, rekto-vezikal fistül

INTRODUCTION

Development of male genitourinary system is a complex phenomenon. Penile agenesis is a rarely seen congenital anomaly with dramatic psychological outcomes, which is encountered in 1 of 30 million births. This condition is believed to arise from absence or incomplete development of genital tubercle, and more than half of them can be associated with other genitourinary anomalies. This anomaly was firstly reported by Imminger in 1853.[1] To date, almost 100 patients have been reported in the World literature.[2]

We herein report a neonatal case with rectovesical fistula associated with vesicoureteral fistula because of its rarity.

CASE REPORT

A 7-hour-old newborn was referred to our clinic because of penile agenesis. Birth weight, head circumference, and height of the term baby who was born to a gravida 1,parity 1 40-yearold mother were 2750 gr, 33 cm, and 44 cm, respectively. On clinical examination, penis was not detected. Scrotum and both testicles were normal in appearance. Anus was at its normal anatomical location and urethral orifice was not detected on perineal region (**Figure 1**).

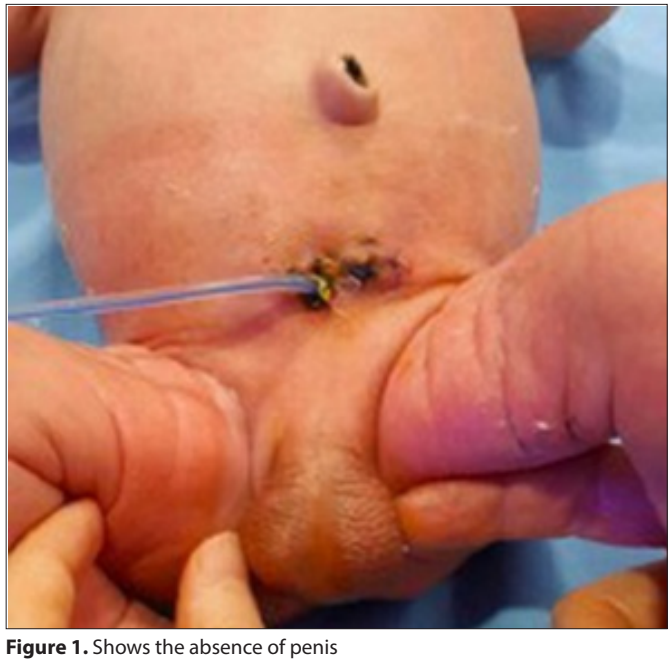

Figure 1. Shows the absence of penis

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Any evidence of dysmorphism, and clinical characteristics of any possible comorbidity were not found. Direct cystourethrogram revealed presence of a rectovesical fistula. On voiding cystourethrogram grade 3 vesicoureteral reflux was observed. Results of other imaging modalities (abdominal X-ray, echocardiography, cranial MR) were within normal limits. Baby had a 46 XY karyotype. Three days after the birth, the patient underwent cystostomy. Ovaries not visualized on pelvic ultrasound. ACTH: 27,4 pg/ml, cortisol: 9,97 µgr/ dl, FSH: 1,09 mIU/mL LH: 0,2 mIU/Ml, SHBG: 42,7 nmol/L, DHEAS: 133 µgr/dl, 17.aOHP: 2,66 ng/ml was normal. During his monitorization, symptoms of irritability, vomiting, and fever. Consequently, levels of acute phase reactants increased, growth of Escherichia coli was observed on urine culture media. Antibiotherapy was initiated. When the general health state of the patient improved, construction of colostomy from the proximal part of the rectovesical fistula was planned, and the patient was transferred to the department of pediatric surgery. For genital reconstruction the departments of pediatric urology, pediatric endocrinology, pediatric genetics, and pediatric psychiatry were consulted.

DISCUSSION

Penile agenesis is a rarely seen anomaly. At 4th week of gestation, it is a dysfunction of genital tubercle which results in incomplete separation of urogenital sinus from hindgut by urorectal septum.[2] This leads to total absence of all three components of penile shaft ie, both corpora cavernosa and spongiosum. Kessler and Mc Laughlin reported the incidence of genitourinary anomalies as 50 % which included cryptoorchidism, renal agenesis and dysplasia, prostate and bladder agenesis, as well as rectovesical and rectourethral fistulas. Besides non-genitourinary anomalies such as spina bifida, VATER–related anomalies, gastrointestinal anomalies, prune-belly syndrome, Potter syndrome, Treacher Collins syndrome, and mental retardation have been reported.[3] Diagnosis is made by meticulously clinical examination, karyotyping, and radiological examinations including a cystogram, and a magnetic resonance imaging.[1] Comorbidities of the patient were examined, and vesicoureteral reflux was observed.

In the literature, penile agenesis has been classified in various ways. Evans et al.[4] classified them according to the presence or absence of major anomalies. Penile agenesis is classified in two major groups as isolated penile agenesis and its complex form associated with congenital anomalies, including genitourinary (54%) and gastrointestinal anomalies, as well as developmental defects of caudal axis.[2] It was included in the complex group as genitourinary abnormalities accompanied our case. Skoog and Bellman[5] divided these patients into 3 groups according to the relationship between anal sphincter and the ectopic urethral meatus; namely, postsphincteric form with anterior perianal urethra, presphincteric urethrorectal fistula, and vesicourethral fistula associated with urethral atresia. Urethral orifice can be localized on perineal region, skin tag resembling foreskin or anterior wall of rectum.[5] Position of the urethral orifice is related to the prognosis. Proximal urethral orifice and presence of other associated anomalies are associated with poor prognosis.[6]

Penile agenesis should be differentiated from disorders of sexual development, including hypospadiasis, severe forms of epispadiasis, intrauterine penile amputation, pseudohermaphroditism, rudimentary penis, concealed penis, and micropenis.[6]

Treatment of penile agenesis is debatable. Urethral transposition should be performed at an early stage to achieve excretion of urine and feces through separate routes. Due to fluctuations in serum testosterone levels between postnatal 10th and 60th days, early stage gonadectomy is recommended to prevent psychological stress of the child.[1] During long-term follow-up of these patients, a shift towards male type has been demonstrated in physiological and psychological development of the child.[7] In recent years, hormonal production and raising of the patients according to their karyotypes have been advocated to prevent sexual dysphoria.[8,9] However, construction of a functional fallus is still the most problematic issue. Therefore, a viewpoint suggesting postponing sexual assignment till the child expresses his/her sexual identity has also been proposed.[8]

Our patient was the first child born to an advanced-aged mother who was living in a patriarchal community. The family was given information about this anomaly of their baby by a council composed of a neonatologist, a psychiatrist, an endocrinologist, and a pediatric urologist.

The family declined sexual reconstruction at an early age, so construction of a colostomy was planned to separate excretion routes of urine and feces.

CONCLUSION

In this rarely encountered case of penile agenesis, we wished to emphasize the process of management and multidisciplinary approach for these patients.

ETHICAL DECLARATIONS

Informed Consent: Written informed consent was obtained from all participants who participated in this study.

Status of Peer-review: Externally peer-reviewed.

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