Recurrent urinary tract infections in a child with ambiguous genitalia

Ambigus genitalyalı bir çocukta tekrarlayan idrar yolu enfeksiyonları

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Abstract

Ambiguous genitalia is described as the external genitalia that do not have the typical anatomic appearance of either a boy or a girl. After providing clinical and metabolic stability in these children, distinct problems such as recurrent urinary tract infections can also be a cause of irritability. Here, a 22-month-old infant with the diagnosis of XLAG syndrome (X-linked lissencephaly and ambiguous genitalia, with 46XY genotype) who suffered from recurrent urinary tract infections due to many different microorganisms including bacteria and fungi, is presented. Previous radiologic evaluations of the patient, when he was 10-month-old, such as urinary ultrasonography (US), voiding cystourethrogram (VCUG), and ^{99m}Tc-DMSA renal scintigraphy were reported as normal. However, when previous VCUG examination was re-evaluated, entire urethra could not be seen in the images. A new VCUG examination which performed as retrograde urethrography showed prostatic utricle as a cause of recurrent urinary tract infections. Although VCUG is sufficient for the evaluation of the anatomy and abnormalities of the bladder and urethra, a detailed imaging of the urethra is usually considered less than the bladder. We want to emphasize that this case is a reminder in depicting the importance of urethral anomalies which can be a cause of recurrent urinary tract infections in children with ambiguous genitalia.

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Key words: Urinary tract infection, urogenital abnormalities, urethral diseases

Özet

Ambigus genitalya bir kız ya da erkek çocuğun dış genital yapısının tipik anatomik görünüme sahip olmaması olarak tanımlanır. Bu çocuklarda klinik ve metabolik denge sağlandıktan sonra, tekrarlayan idrar yolu enfeksiyonu gibi farklı problemler huzursuzluk nedeni olabilir. Bu yazıda, bakteriler ve mantarların dahil olduğu bir çok mikroorganizmaya bağlı tekrarlayan idrar yolu enfeksiyonu ve XLAG sendromu (46 XY genotipi ile birlikte, X'e bağlı geçişli lizensefali ve ambigus genitalya) tanısı ile izlenen 22 aylık bir süt çocuğu sunulmuştur. Hastanın 10. ayında yapılan ultrasonografi, işeme sistoüretrografisi (İS) ve DMSA'lı böbrek sintigrafisi gibi radyolojik değerlendirmelerinin normal olduğu bildirilmişti. Bununla birlikte, önceki İS tekrar değerlendirildiğinde üretranın tamamı görülemedi. Yeniden çekilen İS' de tekrarlayan idrar yolu enfeksiyonlarının nedeni olarak prostatik utrikül gösterildi. İşeme sistoüretrografisi üretra ve mesane anatomisi ile anomalilerini değerlendirmede yeterli olmasına rağmen, üretranın detaylı görüntülenmesi genellikle mesaneden daha az dikkate alınır. Bu olgunun ambigus genitalyalı çocuklarda tekrarlayan idrar yolu enfeksiyonuna neden olabilen üretral anomalilerin önemini vurgulayan bir hatırlatıcı olduğunu düşünmekteyiz.

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Anahtar sözcükler: Üriner kanal enfeksiyonu, ürogenital anomaliler, üretral hastalıklar

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Introduction

The condition that the external genitalia do not have the typical anatomic appearance of normal male or female genitalia is described as ambiguous genitalia which results from a variety of congenital abnormalities of sexual differentiation. Many authors agree about that the etiology underlying genital ambiguity in newborns impacts management including recommendations for sex rearing. Hereby, appropriate therapeutic strategies are planned and developed, after a careful and complete evaluation, by an experienced team of pediatric endocrinologists, geneticists, and surgeons in the follow up period. However, after providing clinical and metabolic stability in these children, some problems which can be a cause of irritability may be overlooked. In this paper, a child with XLAG (X-linked lissencephaly and ambiguous genitalia) syndrome suffered from irritability and recurrent urinary tract infections due to prostatic utricle for a long time is presented.

Case report

A 22-month-old infant was referred to Pediatric Nephrology Department for recurrent urinary tract infections resistant to medical treatment. According to the past medical history, he had a diagnosis of presumably XLAG syndrome (X-linked lissencephaly and ambiguous genitalia, with 46XY genotype) at birth, and was followed by departments of pediatric endocrinology and pediatric neurology. He has been treated with several antiepileptics such as phenytoin, clonazepam, phenobarbital and fludrocortisone for a long time. His parents were first degree cousins. He has 3 siblings: a 7-year-old healthy boy, a 4-year-old healthy girl and a 2-month-old boy with XLAG syndrome. His medical records also revealed that he suffered from recurrent urinary tract infections due to many different microorganisms including bacteria and fungi. A voiding cystourethrogram (VCUG) was performed and reported as normal at age of 10 months. At the same time, ^{99m}Tc-DMSA scintigraphy and renal ultrasonography (US) examinations were reported as normal, too.

On physical examination, the child was afebrile but irritable. He was 81 cm long (between 25th and 50th percentile) and he weighed 11 kg (between 25th and 50th percentile). His head circumference was 36.5 cm (below the 3rd percentile). Clinical examination identified microcephaly, lack of visual contact, axial hypotonia and motor-mental retardation according to age. On genital examination, ambiguous genitalia was identified (impalpable gonads, phallus of 0.5 cm single urogenital meatus and scrotal hypoplasia with normal skin pigmentation) (Figure 1).

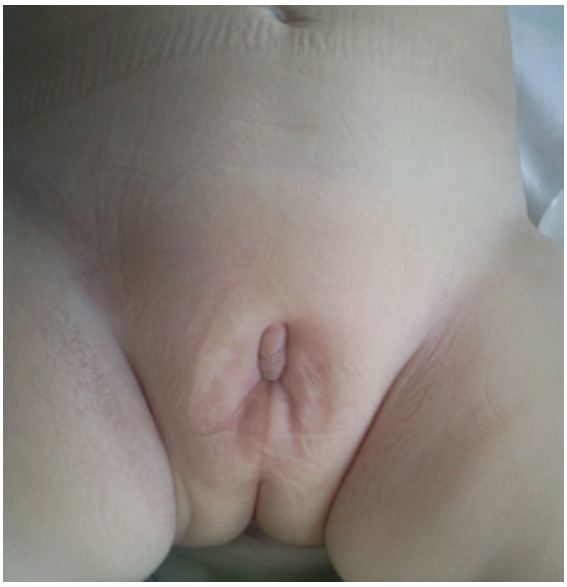

Figure 1. The photograph obtained from the patient shows ambiguous genitalia.

Urinalysis revealed pyuria, and the urine culture yielded *Escherichia coli*. Complete blood count, renal function tests and acute phase reactants were all normal. Ultrasound examination of the urinary tract showed normal kidneys (size and echogenity), without dilatation of the upper urinary tract and normal bladder. ^{99m}Tc-DMSA scintigraphy was also normal. Urinary tract infection was treated with appropriate parenteral antibiotics. VCUG was performed about one year ago in our case. When we re-evaluated previous VCUG, we saw that entire urethra was not displayed. A new VCUG which performed as retrograde urethrography showed prostatic utricle with normal bladder and without vesicoureteral reflux (Figure 2). The utricle excision was planned.

Discussion

The Müllerian duct is present in all human embryos at the early stages of the development. In male subject, secretion of Müllerian inhibiting factor causes regression of the Müllerian system. Utricular anomalies result from the incomplete regression of the Müllerian duct remnant or incomplete androgen mediated closure of urogenital sinus caused by an error of sensitivity to local testosterone or Müllerian

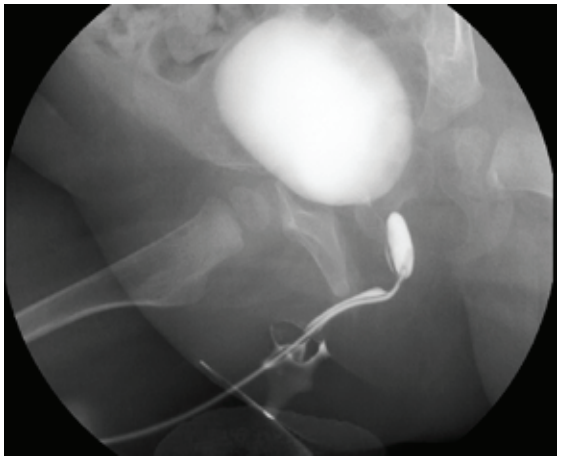

Figure 2. VCUG image shows a typical prostatic utricle and normal bladder without vesicoureteral reflux.

inhibiting factor [1-4]. Therefore, the prostatic utricle is a short, blind-ending pouch located on the verumontanum [2]. It occurs most often in males with perineal or peno-scrotal hypospadias [5]. Why prostatic utricle has been associated with various stages of inter-sex situations may be explained by the pathogenetic mechanisms, mentioned above.

Although some utricles are asymptomatic, it may present clinically as lower urinary tract irritative symptoms, postvoid dribbling, urethral discharge, recurrent urinary tract infections, stone formation in pouch or pseudo incontinence due to secondary trapping of urine in the pouch, and retention of urine particularly when the utricle is enlarged. In the differential diagnosis, ectopic ureter, dilated ejaculatory duct, Müllerian duct cyst, and diverticula in any part of the urethra should be kept in mind [6].

VCUG or retrograde urethrography define the utricular size and its origin from prostatic urethra [7]. VCUG was performed about one year ago in our case. VCUG is usually sufficient for the evaluation of the anatomy and abnormalities of the bladder and urethra. However, a detailed imaging of the urethra is usually considered less than the bladder in VCUG. It depends on many things such as irritability of infants, the need to take more number of X-rays and focus on vesicoureteral reflux. For these reasons, VCUG should be repeated as retrograde urethrography to detect the urethral abnormalities, especially in uncooperative child.

Although non-surgical approaches include transurethral utricle catheterization and aspiration, dilatation of utricle orifice, sclerotherapy and electro fulguration, surgical excision is reserved for symptomatic utricles [8].

In conclusion, the children with ambiguous genitalia are uncommon in pediatricians' practice, but they may suffer from very important problems such as recurrent urinary tract infections, voiding disorders. If there are recurrent urinary tract infections in these children, the urethra, from bladder to external orifice, must be carefully evaluated by VCUG or retrograde urethrography. We thought that this case is a reminder in depicting the importance of urethral anomaly in children with ambiguous genitalia.

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