

Duplicated urethra and a possible mullerian–renal–cervical spine (MURCS) association in a male child: a case report

Bir erkek çocuktaki üretra dublikasyonu ve olası MURCS (müllerian–renal–servikal–spinal) sendromu

Çağrı Güneri¹, Lütü Tunç², Fazlı Polat², Mustafa Özgür Tan², Kadri Karaer³, İbrahim Bozkırlı²

ABSTRACT

We report on a 2-year-old boy who was referred to our clinic with recurrent urinary tract infections, neurogenic bladder dysfunction and duplicated urethra. The child was unable to void spontaneously, and a suprapubic catheter had been placed. We performed an internal urethrotomy on his patent urethra, but the procedure did not successfully restore spontaneous voiding. After the surgery, we evaluated the child for the presence of congenital syndromes, especially the MURCS (Mullerian-Renal-Cervical Spine) association. Six months later, the patient underwent a left orchiopexy procedure. There was no evidence of the right testis. This case shows that the male variant of the MURCS association may accompany miscellaneous urogenital anomalies such as renal agenesis, cross renal ectopy, vesicoureteral reflux, neurogenic bladder dysfunction, posterior urethral valve, duplicated urethra, bilateral/unilateral undescended testes or vanishing testis.

Key words: Duplicated urethra; male; MURCS association; undescended testis.

ÖZET

Bu olgu sunumunda kliniğimize üriner sistem enfeksiyonu, nörojenik mesane ve üretra dublikasyonu ile refere edilen 2 yaşındaki erkek çocuğu rapor etmekteyiz. Hasta başka bir merkezde spontan idrar yapamama nedeniyle suprapubik olarak kateterize edilmiş idi. Bu hastada internal üretrotomi prosedürü gerçekleştirdik. Ancak bu yolla da hastada spontan miksiyon izlenmedi. Altı ay sonra hastaya sol orşiopeksi işlemi uygulandı. Sağ testis izlenemedi. Bu olgu MURCS sendromu erkek varyantına renal agenezi, kros renal ektopi, vezikoureteral reflü, nörojenik mesane, posterior üretral valf, üretra dublikasyonu, bilateral/tek taraflı inmemiş testis ya da testis yokluğu gibi çok çeşitli ürogenital anomalilerin eşlik edebileceğini göstermektedir.

Anahtar sözcükler: Erkek cinsiyet; inmemiş testis; MURCS sendromu; üretra dublikasyonu.

Introduction

The MURCS (mullerian-renal-cervical spine) association is a well-known but rare developmental disorder. Phenotypically, the features are widely variable, and the exact etio-pathogenesis is still unclear. Children with this association are generally female with a female karyotype, and the rest of the genitourinary organs, including the ovaries and fallopian tubes, are normal. The diagnosis of this condition in males is characterized by non-obstructive azoospermia instead of uterine malformations.^[1,2] In this case report, we aimed to make urologists aware of this condi-

tion, especially when multiple genitourinary anomalies accompany cervicothoracic anomalies and renal agenesis.

Case report

A 2-year-old boy was admitted to our clinic with duplicated urethra, urethral stenosis, bilateral undescended testes, neurogenic bladder dysfunction, vesicoureteral reflux, renal agenesis, cross ectopy of the right kidney, thoracic kyphoscoliosis and recurrent urinary tract infections. The patient had undergone a posterior urethral valve ablation procedure 12 months prior to presentation; 2 months after

¹Çankırı State Hospital, Çankırı

²Department of Urology, Faculty of Medicine, Gazi University, Ankara

³Department of Medical Genetics, Faculty of Medicine, Gazi University, Ankara

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Correspondence:
Çağrı Güneri
Çankırı State Hospital, Çankırı,
Turkey
Phone: +90 505 790 64 02
E-mail: drcagriguneri@hotmail.com

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that, the patient underwent an internal urethrotomy procedure for urethral stenosis in the bulboprostatic urethra, which was considered to be related to the previous posterior urethral valve ablation. The patient then underwent a perineal urethroplasty procedure 7 months after the urethrotomy procedure. During the follow-up period, a suprapubic catheter was placed because of a gradually increasing post-void residual urine volume, and the patient was referred to us for further work-up.

The patient was the first child of a non-consanguineous marriage and was born to a healthy 32-year-old mother and a healthy 38-year-old father. The patient was born at 40 weeks gestation via cesarean section because of fetal distress. His birth weight was 2700 g (3-10%), and his length and head circumference were not recorded. Difficulties with feeding and poor weight gain in the post natal period were noted. Psychomotor development was normal. The patient began to walk and speak single words at the age of 1.5 years old.

On physical examination upon this admission, the patient's height was 73 cm (>3p), his weight was 8.5 kg (<3p) and his head circumference was 47 cm (<3p). The patient also exhibited the following physical findings: relative macrocephaly, a flat face with sparse eyebrows and long eyelashes, dysplastic ears with a preauricular skin tag on his left ear, anteverted nostrils, a long philtrum and thin upper and lower lips (Figure 1a). On the urological examination, the size and shape of the penis appeared to be normal.

The patient also exhibited a duplicated urethra; the patent urethra was hypospadiac, and the obliterated one was in the correct anatomical position (Figure 1b). The left testis was palpable in the left inguinal region, but the right testis was non-palpable. A suprapubic catheter was placed due to neurogenic bladder dysfunction and urethral stenosis, which led to his inability to void spontaneously.

Investigations by his previous urologist revealed normal renal function tests and hematologic parameters. At that time, the patient was receiving intravenous antibiotic therapy because of the recurrent urinary tract infections. Chromosome analysis of peripheral leukocytes using the high resolution binding technique revealed a normal 46, XY male karyotype.

Urological investigations performed before the patient's admission to our clinic included urinary system ultrasonography, an abdominal CT scan, a radionuclide scan and a voiding cystography; these tests revealed thoracic kyphoscoliosis, a non-visualized left kidney, cross ectopy of the right kidney, a convoluted hydroureter, vesicoureteral reflux and radiological findings consistent with neurogenic bladder dysfunction (Figure 2a, b). Ultrasonographic investigation of the left inguinal region



Figure 1. (a) Dysplastic ears with a preauricular skin tag on his left ear. (b) Urethral duplication.



Figure 2. (a) Voiding cystourethrogram. Solitary kidney and a convoluted ureter. (b) Abdominal CT scan.

showed the left testis in the inguinal canal, whereas the right testis was not detected ultrasonographically.

Cystoscopically, urethral stenosis at 7 cm proximale from the meatus of the hypospadiac urethra was observed, and an internal urethrotomy was performed. There was no evidence of a posterior urethral valve except for the trabecular appearance of the bladder during cystoscopy. The other urethra in the correct anatomical position was obliterated. After the removal of the urethral catheter on the fourth post-operative day, the patient could not void adequately through his urethra, and we concluded that a cystostomy would be much more effective at preserving the patient's renal function.

Six months later, we performed a Dartos pouch orchiopexy procedure on the left testis. During the exploration of the left inguinal region, we noted that the left testis was small and soft. The post-operative period was uneventful, and the patient was discharged three days after surgery.

Discussion

The MURCS association is a rare developmental disorder that usually affects females. It was first described by Duncan et al.^[3] in 1979. The occurrence of the MURCS association is sporadic,^[4] and the etiology is still not clear; one hypothesis is that it results from an alteration of the blastema of the lower cervical and upper thoracic somites and pronephric ducts, which have an intimate spatial relationship in the fourth week of fetal development.^[5] In addition to this hypothesis, Pavenello et al.^[6] presented a family in which three sisters had Müllerian anomalies, and their brother was azoospermic. To the best of our knowledge, the number of reported cases of MURCS in a male is limited, and the clinical features are widely variable. Meschede et al.^[2] suggest that the more appropriate acronym might be ARCS (azoospermia, renal anomaly and cervicothoracic spine dysplasia). The reported clinical findings in males in addition to the cervicothoracic deformities and renal agenesis include azoospermia,^[1,2,7] detachment of the epididymis from the testis^[1], small sized testis with a soft texture^[1], hypospadias^[1], and agenesis of the vas deferens^[7]. We were not able to evaluate whether the present patient was azoospermic. However, during the surgical exploration, we noted that his only testis was obviously small and soft, and we expected him to develop azoospermia in the future. In this paper, we aimed to emphasize that in addition to the cervicothoracic spinal defects, the cases

of male MURCS may be associated with other miscellaneous genitourinary anomalies, such as renal cross ectopy, vesicoureteral reflux, posterior urethral valve and bladder dysfunctions related to prolonged infravesical obstruction, duplicated urethra, undescended testis and/or vanishing testis.

Conflict of interest

No conflict of interest was declared by the authors.

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