TURKISH JOURNAL OF UROLOGY Turkish Journal of Urology 2012; 38(2): 112-113 • doi:10.5152/tud.2012.024

PEDIATRIC UROLOGY

Case Report

Giant vesical diverticulum, unilateral renal agenesis and mental retardation: an unusual association

Dev mesane divertikülü, unilateral renal agenezi ve mental retardasyon: nadir bir birliktelik

Duygu Övünç Hacıhamdioğlu[1], Yusuf Kibar[2], Davut Gül[3], Murat Kocaoğlu[4], Faysal Gök[1]

ABSTRACT

Herein, an 11-year-old boy is described with giant vesical diverticulum, unilateral renal agenesis and mental retardation. This combination does not fit any known syndromes, and to our knowledge, this is the first report of the association of these features.

Key words: Bladder diverticulum; renal agenesis.

ÖZET

Bu yazıda dev mesane divertilü, renal agenezisi ve mental retardasyonu olan 11 yaşında bir erkek sunulmaktadır. Bu birliktelik bilinen herhangi bir sendroma uymamaktadır ve bilgilerimize göre bu özelliklerin birlikteliği ilk defa raporlanmaktadır.

Anahtar sözcükler: Mesane divertikülü; renal agenezi.

[1]Department of Pediatric Nephrology, Gulhane Military Medicine Faculty, Ankara, Turkey

[2]Department of Urology, Gulhane Military Medicine Faculty, Ankara, Turkey

[3]Department of Genetics, Gulhane Military Medicine Faculty, Ankara, Turkey

[4]Department of Radiology, Gulhane Military Medicine Faculty Ankara, Turkey

Submitted: 12.02.2011

Accepted: 21.05.2011

Correspondence: Duygu Övünç Hacıhamdioğlu Gülhane Askeri Tıp Akademisi, Çocuk Sağlığı ve Hastalıkları Anabilim Dalı, Etlik 06018 Ankara, Turkey
Phone: +90 312 304 43 74
E-mail: duyguovunc@yahoo.com.tr

Available online at www.turkishjournalofurology.com

Case

An 11-year-old boy was admitted to our department with a 4-month history of right flank pain and hematuria. The patient was born by uneventful delivery at 38 weeks of gestational age. No events were listed in his prenatal or postnatal histories until six months of age. The parents were healthy and nonconsanguineous. The patient is the younger of two siblings. His grandfather had renal atrophy of unknown etiology. No other family member was affected. In his medical history, the patient had 2 afebrile convulsions, at 6 months and 1 year old. His motor development stages were normal. The boy was enrolled in special education at 8 years of age following trouble with his lessons in primary school. The following tests were performed to determine the mental status of the patient. A hearing test, urine amino acid chromatography, and brain magnetic resonance imaging (MRI) were normal. A cytogenetic analysis revealed a normal 46, XY karyotype. The results of the WISC-R (Wechsler Intelligence Scale for Children-Revised) test are as follows: verbal score, 40 points; performance score, 45 points; and total intelligence, 41 points. According to these findings, the patient had moderate mental retardation. The patient did not have a history of urinary tract infections until four months prior to admission. The physical examination revealed mild wide-apart nipples and pectus excavatum. His weight and length were 32 kg (10-25th percentile) and 139 cm (10-25th percentile), respectively, and he had a head circumference of 53.5 cm. His blood pressure was 100/72 mmHg. The relevant physical examination was normal. Laboratory investigations showed a blood urea nitrogen level of 8.21 mg/dl (N: 5-25) and a creatinine level of 0.86 mg/dl (0.6-1.2). The blood cell count and other serum parameters were within normal limits. Ultrasonography showed that the right kidney was 83 mm in longitudinal axis and that the kidney parenchymal thickness was 10 mm. The left kidney was not visualized. The voiding cystourethrogram showed a large, smooth-walled bladder diverticulum that was located in the superior bladder, and no vesicoureteral

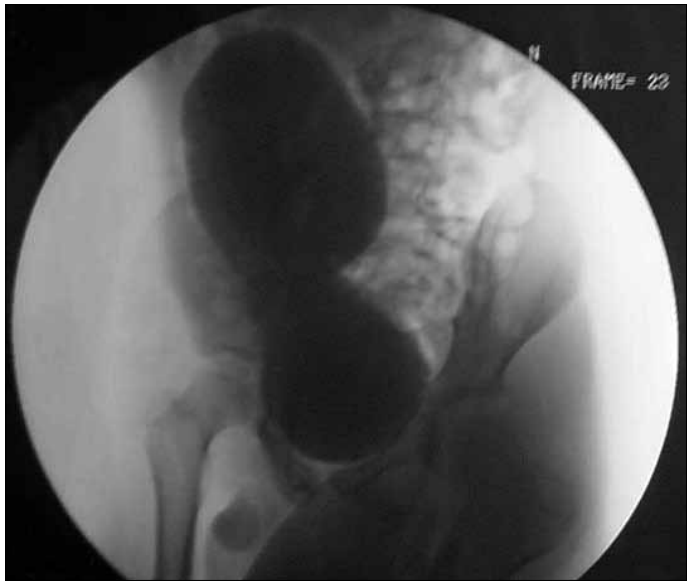

Figure 1. The patient's voiding cystourethrogram, smooth-walled bladder diverticulum was located in superior of the bladder.

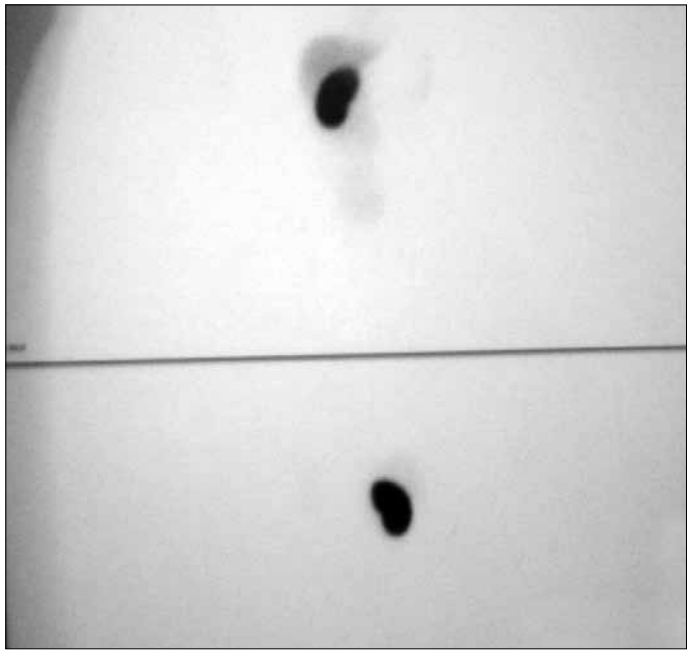

Figure 2. DMSA demonstrated no activation for left kidney.

reflux was observed (Figure 1). A renal 99mTc dimercapto-succunic acid (DMSA) study demonstrated normal scintigraphic findings for the right kidney, no activation for the left kidney (Figure 2) and radionuclide accumulation in the superior bladder in the area of the bladder diverticulum. A contrast renal MRI was performed, and it demonstrated a bladder diverticulum that was isointense with the bladder and 7x7x5 cm in diameter; it was located in the superior and anterior regions of the bladder (Figure 3). Additionally, the left kidney was not observed in the

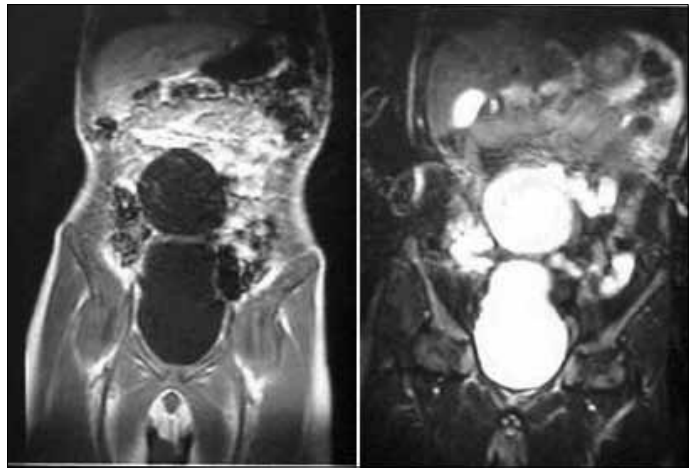

Figure 3. Contrast renal MRI showed a isointense diverticulum located in superior and anterior of bladder.

renal MRI. The patient had urinary tract infection symptoms because the diverticulum was not emptying during voiding. As such, surgery was performed. The area through the midline incision below the umbilicus diverticulum was resected, and the bladder was repaired. The postoperative period was uneventful. During the 11 months of follow up, the patient had no complaints and hematuria did not occur.

Discussion

A giant vesical diverticulum is not a common anomaly. It was reported as both a single anomaly and together with some syndromes, such as Type I Ehlers-Danlos Syndrome (#130000), Cat Eye Syndrome (#115470) and Beckwith-Wiedemann Syndrome (#130650). However, its occurrence with bladder diverticulum and renal agenesis is a rare. In 1992, three male patients have been reported with primary congenital bladder diverticulum, and the described 9-year-old patient also presented with unilateral renal agenesis.[1] We were unable to reach any patient previously reported with these anomalies. This is the first case in the literature of a patient presenting with bladder diverticulum, renal agenesis and mental retardation. However, all of these findings might be different abnormalities with a co-incidence or they may be part of a syndrome.

In conclusion, we offer that a patient should be better investigated for abnormal systemic associations when they present with any urinary tract anomaly.

Conflict of interest

No conflict of interest was declared by the authors.

References

1. Stage KH, Tank ES. Primary congenital bladder diverticula in boys. Urology 1992;40:536-8. [CrossRef]