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OLGU

Bilateral seminal vezikül agenezisi ve soliter pelvik böbreğin eşlik ettiği bilateral konjenital vaz deferens agenezisi

Bilateral congenital absence of the vas deferens coexisting with bilateral seminal vesicle agenesis and solitary pelvic kidney

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Özet

Vaz deferens anomalileri yaygın değildir ve infertil erkeklerin % 1-2'sinde görülmektedir (1).Bu durumun olası nedenleri hala belirsiz olup Wolf kanallarının defektif gelişimi sonucu ortaya çıkmaktadır. Seminal vezikül ve böbrek anomalileri sıklıkla vaz deferens anomalilerine eşlik etmektedir. Bu vaka takdimi ile bilateral vaz deferens agenezisine eşlik eden bilateral seminal vezikül agenezisi ve soliter pelvik böbreği olan bir olgu sunulmuştur.

Anahtar kelimeler: Vaz deferens agenezisi, seminal vezikül agenezisi, soliter pelvik böbrek

Abstract:

Anomalies of vas deferens are uncommon and occurs 1-2% of the infertile men (1). The possible causes for this condition is still unclear, and it is caused by defective development of Wolffian ducts. Seminal vesicle and renal anomalies accompany to vas deferens anomalies frequently. In this report we present a case of congenital bilateral absense of vas deferens (CBAVD) coexisting with bilateral seminal vesicle agenesis as well as solitary pelvic kidney.

Key words: Absence of the vas deferens, seminal vesicle agenezis, solitary pelvik kidney

Bilateral congenital absence of the vas deferens

Introduction:

Seminal vesicle pathologies accompanying bilateral congenital absence of the vas deferens (CBAVD) and renal agenesis are rare congenital urological anomalies, which results from abnormal development of the Wolffian ducts and ureteric bud (2). CBAVD is usually diagnosed by physical examination for the investigation of male infertility. Seminal vesicle pathologies and renal anomalies can be detected by ultrasonography and computed tomography scans. Goldstein et al. reported 77% of SV anomalies, detected by CT, in patients with CBVAD (3).

Case report:

A 25 year old man was referred to our clinic after incidental diagnosis with CBAVD after initial consultation with the urologist. The physical examination confirmed bilateral absence of the vas deferens. Digital rectal examination revealed normal prostate. Scrotal examination demonstrated multiple bilateral epidymal cystic lesions. The semen analysis showed low volume (0.8 cc) azospermia and very low level of seminal fructose. Routine biochemical analysis of the serum and urine analysis did not demonstrate any abnormal findings. Transrectal ultrasonography showed bilateral seminal vesicle agenezis. Abdomino-pelvic CT revealed a pelvic solitary kidney with a unilateral ureter opening to the bladder base on the right side (Fıgure 1) and bilateral absence of vas deferens (Fıgure 2). Bilateral multiple epididymal cysts ranging from 3-4 mm size were detected by scrotal doppler ultrasonography. Serum follicle-stimulating hormone (FSH), luteinising hormone (LH) and total testosterone levels were normal. There was a slight increase in serum prolactin (PRL) level. Genetic evaluation evaluating cystic fibrosis transmembrane regulating (CFTR) gene mutation by reverse hybridization (Innogenetics Strip Assay) did not reveal any mutation for cystic fibrosis.

Figure 1. Pelvic CT-scan reveals solitary pelvic kidney

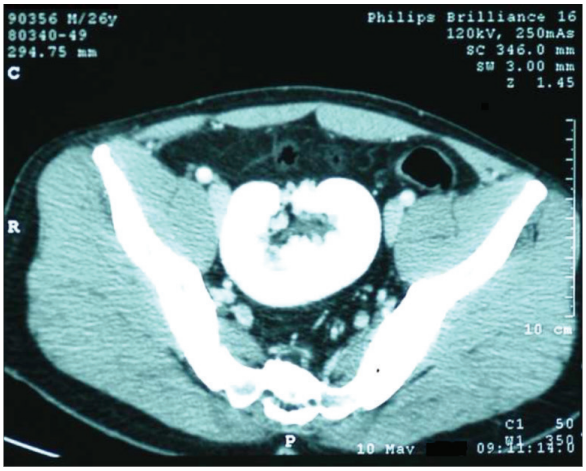

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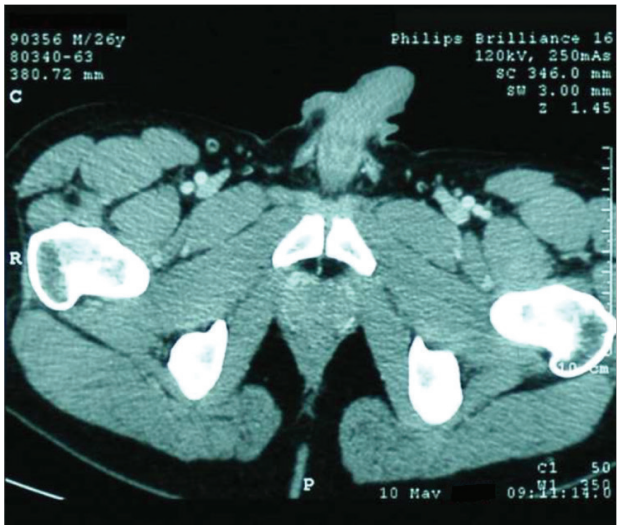
Figure 2. Pelvic CT-scan reveals bilateral seminal vesicle agenezis and bilateral absence of ductus deferens



Discussion:

The regression of the Wolffian duct before the fourth week of intrauterine life, in the absence of the ureteric bud results in agenesis of the vas deferens and the ipsilateral kidney. However, after the fourth week only absence of the vas occurs (4,5). CBAVD is a rare cause of male infertility (6) and is usually associated with low ejaculate volume (<2 mL according to WHO criteria), total absence of spermatozoa and an acid ejaculate composed of prostatic secretions, with low or absent fructose, concurring with the absence of seminal vesicle (SV) (7). Most of these cases are asymptomatic or sometimes detected as a cause of male infertility. Renal anomalies associated with CBAVD should be considered as supporting maldevelopment as a cause, but analysis of CFTR gene mutations in these cases should not be omitted. Because these patients are at greater risk (%50-60) for cystic fibrosis (8). Therefore, it is important to evaluate these couples for cystic fibrosis during consultation in the infertility clinics. In our case there was no sign of CF gene mutation. This supports the hypothesis that the combination of unilateral renal agenesis and CBAVD may be a result of another genetic defect affecting the Wolffian duct early in embriyogenesis prior to week 7, and not due to CF gene mutations (8). McCallum et al. reported that the percentage of men with a pelvic kidney was 10-fold higher in the unilateral renal agenesis and CBAVD group cohort than the CF and CBAVD group, and this was statistically significant (12% versus 1%, $p<0.01$) (8). From our findings we can conclude that infertile men with CBAVD should be thourogly investigated for renal anomalies. The genetic counselling in these patients are particularly important.

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