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Case Report OMICS International

Congenital Unilateral Absence of the Vas Deferens as an Incidental Finding During Radical Prostatectomy – Case Report Literature Review and Recommendations for Clinical Management

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Abstract

Congenital unilateral absence of the vas deferens (CUAVD) is a disorder affecting up to 1% of males. As these patients are often asymptomatic the diagnosis is usually made incidentally in the context of imaging studies performed for other indications or during urologic surgical procedures like vasectomy or radical prostatectomy. Besides a potential clinical manifestation in terms of infertility due to obstructive azoospermia, CUAVD gains clinical importance because of its association with renal anomalies as well as mutations of the *CFTR* gene. Here we describe the case of a patient who was diagnosed with CUAVD during radical prostatectomy, review the currently available literature and give recommendations on the clinical management of patients with CUAVD.

Keywords: CUAVD; Prostatectomy; Vasectomy; CFTR-mutation; Infertility; Renal agenesis

Case Report

A 74 year-old man diagnosed with organ-confined prostate cancer presented to our department for laparoscopic, robot-assisted radical prostatectomy. The patient's medical history included hypertension, cholecystolithiasis as well as a congenital solitary kidney on the right side. Due to a relatively low PSA level (4.5 ng/ml) and a Gleason-Score of 3+4=7a in only two biopsy cores, no preoperative staging was performed. During the surgery neither a ductus deferens nor a seminal vesicle could be identified on the left side despite thorough preparation. Histopathological examination of the resected specimen showed a complete lack of the semen-transporting anatomical structures (ductus deferens, seminal vesicle, ejaculatory ducts) on the left side leading to the diagnose of CUAVD associated with a solitary kidney on the ipsilateral side. The patient reported no complaints regarding erectile or ejaculatory functions. CUAVD is a disorder affecting up to 1 in 100 males [1]. It was first described by John Hunter in 1737 [2] while Reverdin was the first one to observe an association between absence of the vas deferens and ipsilateral renal agenesis in 1870 [3]. Since then several case series have been published analyzing the clinical and genetic characteristics of this disease [4-9]. As CBAVD is a well-known abnormality often associated with hetero- or homozygous mutations of the cystic fibrosis transmembrane conductance regulator gene (*CFTR*), it was plausible to also look for the CFTR-status in CUAVD patients. In one of the largest studies comprising 21 CUAVD patients, Mickle et al. [5] observed heterozygous CFTR mutations in 8 cases (38%). Notably, all of these 8 patients had non-iatrogenic occlusion of the contralateral vas deferens at either the inguinal or pelvic level. Renal anomalies were present in 5 patients (24%; 4 ipsilateral renal agenesis, 1 ipsilateral pelvic renal ectopy) – all of them showing CFTR wild-type in genetic testing. Similarly, Schlegel et al. [6] described ipsilateral renal agenesis in 26% and heterozygous CFTR mutations in 25% of CUAVD patients. Again, all patients that had accompanying renal agenesis were negative for CFTR mutations while all those with CFTR mutations had normal renal anatomy. The results of these reports were confirmed by Kolettis and Sandlow [7] as well as Schwarzer and Lane [8-10] in recent studies. Based on these findings one can assume that there are two distinct subpopulations of CUAVD patients with different genetic and etiological backgrounds [5,7]: The first subgroup comprises patients with heterozygous mutations in the *CFTR* gene as the underlying pathophysiologic mechanism, which do not show accompanying renal anomalies. However, these patients may become clinically symptomatic due to obstructive azoospermia causing infertility. In the second subgroup of patients a hitherto unknown genetic event (not involving the *CFTR* gene) leads to an abnormal development of the entire mesonephric duct at a very early stage in embryonic development. Consequently these patients often show ipsi- or (in rare cases) contralateral renal agenesis in addition to CUAVD while being mostly asymptomatic. Other congenital anomalies like renal ectopy have also been described [5]. Taking the above mentioned pathophysiologic mechanisms and clinical features of the disease into consideration, all patients with a newly diagnosed CUAVD should undergo imaging of the upper urinary tract-primarily by sonography. If there is no evidence for variations of renal anatomy, genetic testing for CFTR-mutations might be reasonable especially for patients of reproductive age due to the potential risk of transmission of renal anomalies and CFTR mutations [7].

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