CASE REPORT PEER REVIEWED | OPEN ACCESS

Congenital unilateral absence of the vas deferens

Winston Owens, Jonathan Hakim

ABSTRACT

Introduction: A 65-year-old male with a known history of unilateral renal agenesis was found to have concurrent absence of the vas deferens ipsilaterally.

Case Report: The patient presented with organic erectile dysfunction and wanted to proceed with surgical management. Routine pre-operative evaluation confirmed history of a left solitary kidney, and physical exam revealed absence of the right vas deferens. Previous imaging confirmed right renal agenesis, and with subsequent absence of the right vas deferens noted on physical exam, this confirmed embryological arrest as the cause of the anatomic abnormality.

Conclusion: Congenital unilateral absence of the vas deferens (CUAVD) is an uncommon finding that is largely found during infertility evaluations and vasectomy consults. It is important to understand genetic components and embryological origins of CUAVD to appropriately evaluate patients for additional defects.

Keywords: Absence of vas deferens, CFTR, Organic erectile dysfunction

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INTRODUCTION

John Hunter first described congenital unilateral absence of the vas deferens (CUAVD) in 1737 [1]. Congenital unilateral absence of the vas deferens is rare and has a prevalence of 0.5–1.0%. It is usually discovered during infertility work-up, or other urological pre-operative evaluations—most commonly vasectomy consultation [2, 3]. It is associated with either embryological arrest of the Wolffian/mesonephric duct or more commonly, cystic fibrosis transmembrane conductance regulator (CFTR) mutations [4–6].

We present a case of CUAVD with concurrent ipsilateral renal agenesis that was identified incidentally during pre-operative assessment and planning for an unrelated issue.

CASE REPORT

A 65-year-old white male presented with organic erectile dysfunction secondary to type II diabetes which was refractory to medical management. During routine pre-operative evaluation for an inflatable penile prosthesis, he admitted to solitary left kidney and no history of a right nephrectomy nor family history of cystic fibrosis. The absence of the right kidney was confirmed on a review of his previous magnetic resonance imaging (MRI) as seen in Figure 1. On physical exam, the right vas deferens was absent on palpation which confirmed an embryological arrest as the cause of the CUAVD.

DISCUSSION

Congenital absence of the vas deferens is a rare anatomical finding and is estimated to affect 0.5–1% of the male population [2]. This is usually found during evaluations for infertility or vasectomy consults. It can present with additional anatomical anomalies [2, 3]. Congenital unilateral absence of the vas deferens

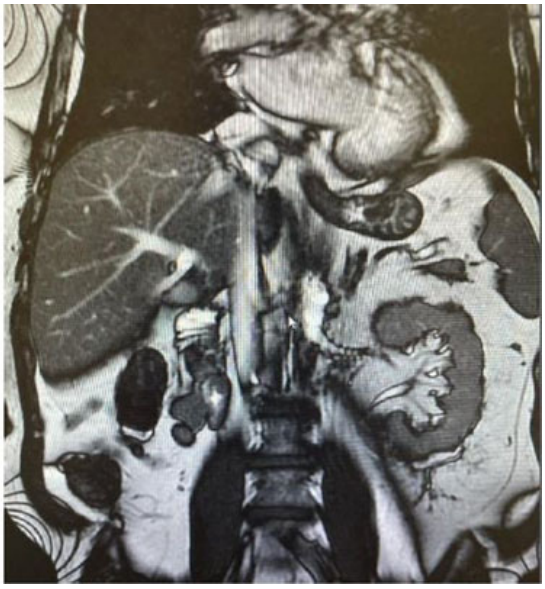

Figure 1: Review of previous MRI confirmed absence of right kidney.

is observed more commonly on the left side (66%); unilateral renal agenesis is seen more frequently with CUAVD (73.7%) compared to bilateral absence of the vase deferens (CBAVD) (11.8%) [2].

There are two subgroups of patients with CUAVD, fertile and infertile. Fertile patients with CUAVD have a patent vas deferens contralaterally and have a higher frequency of renal agenesis ipsilaterally. Infertile CUAVD patients either have partial or complete occlusion of the contralateral vas, contributing to their sterility. While infertility is possible with CUAVD, it is not the sole contributor to sterility in most cases. Weiske and colleagues reported only 0.4% of male infertility was attributed to CUAVD alone [2]. Additionally there is an association between CUAVD and cystic fibrosis (CF). A study found that 6 out of 14 infertile CUAVD patients had mutations in their cystic fibrosis transmembrane conductance regulator (CFTR) gene; other authors reported that 38% of CUAVD cases were associated with CFTR mutations [7, 8]. Compared to bilateral absence of the vas deferens, which has a more prominent association with CF. This was described in a study where of 106 CBAVD patients, 75% carried CFTR mutations or disease-associated variants [9].

The vas deferens is derived embryologically from the Wolffian duct (mesonephric duct). This duct also gives rise to the distal two-thirds of the epididymis, seminal vesicles, and ejaculatory ducts; also, it contributes to the ipsilateral trigone, and the proximal urethra up to the membranous urethra with its voluntary urinary sphincter. However, the Wolffian duct's key role here is to stimulate kidney development. The ureteric bud arises from the Wolffian duct and penetrates the metanephric blastema, inducing kidney development [10]. Disruption of this embryologic process at varying times will result in different congenital abnormalities. If the patient has a congenital solitary kidney, the cause could be due to a CFTR mutation or an embryological arrest.

Studies directed toward understanding a link between CFTR mutation variants and renal agenesis in the CUAVD patient population have been widely discussed and are controversial. Ultimately, this question requires additional analysis with a large sample size study [11]. A recent meta-analysis on this subject showed that the higher risk of renal abnormalities in the CUAVD population was not related to CFTR variants [4]. In this patient's case, the embryological arrest was confirmed by the physical exam finding of no palpable vas deferens in the scrotum on the affected side. Congenital unilateral absence of the vas deferens is not limited to presenting with renal agenesis. The literature has reported isolated CUAVD cases with anomalies of seminal vesicles, ejaculatory ducts, cryptorchidism, and inguinal hernias [3].

CONCLUSION

Congenital unilateral absence of the vas deferens is an uncommon finding that is largely found during infertility evaluations and vasectomy consults. It is important to understand the genetic and embryological origins of CUAVD and appropriately evaluate for additional congenital defects. Particularly significant is the CFTR mutation and the potential need for genetic testing and counseling for CF. This emphasizes the need to confirm palpation of the vas deferens bilaterally during physical exam.

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Author Contributions

Winston Owens – Conception of the work, Design of the work, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Jonathan Hakim – Conception of the work, Design of the work, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Guarantor of Submission

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Conflict of Interest

Authors declare no conflict of interest.

Data Availability

All relevant data are within the paper and its Supporting Information files.