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Case Report

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Unveiling Hidden Infertility: A Case of Congenital Bilateral Absence of the Vas Deferens (CBAVD)

Madushanka U. D. P.[1], **Pragalathan B.**[1], **Idhrees I. H.**[1], **Nandasena A. C. N.**[1]

[1]Department of Urology, Colombo North Teaching Hospital, Ragama, Sri Lanka.

Abstract

We report a rare case of male subfertility due to congenital bilateral absence of the vas deferens (CBAVD) in a 38-year-old man with primary infertility. Semen analysis showed azoospermia with normal hormonal profile and testicular biopsy revealed normal spermatogenesis. Imaging confirmed the absence of vas deferens bilaterally and an absent left kidney. CBAVD, often associated with Wolffian duct anomalies, highlights the importance of thorough evaluation in male infertility. While physical examination and TRUS may be inconclusive, diagnosis enables fertility through sperm retrieval and assisted reproductive techniques. Genetic counselling is essential due to potential mutation-related implications.

Keywords: Congenital bilateral absence of the vas deferens (CBAVD), primary infertility, absent left kidney, cystic fibrosis

Introduction

Congenital bilateral absence of the vas deferens (CBAVD) is clinically characterized by the absence of the bilateral vas deferens; the main clinical manifestation is infertility, accounting for 1-2% of male infertility cases (1). CBAVD is often associated with developmental abnormalities of the uro-genital system and cystic fibrosis (CF)-related clinical manifestations. The primary characteristic of this condition is the absence of both vas deferens, which prevents the transport of sperm from the epididymis to the ejaculatory ducts, thereby leading to male infertility (2). In addition to CF-related symptoms, other congenital genitourinary abnormalities, mainly including dysplasia or the absence of the kidneys and seminal vesicles, can accompany CBAVD (1).

Physical examination, particularly palpation aimed at differentiating the vas deferens from the other cord-like structures, are often not reliable, especially in obese patients or those with high-riding scrotums. However, both transrectal and scrotal ultrasonography serve as reliable imaging modalities for identifying abnormalities of the seminal vesicles and epididymis in individuals with CBAVD.

This case report describes a patient who initially presented with subfertility and was subsequently diagnosed with congenital bilateral absence of the vas deferens (CBAVD), along with an overview of his clinical management.

Case presentation

A 38-year-old married man presented with a history of primary infertility for two years to the urology clinic. He denied history of smoking, drug abuse, genital trauma, childhood or other systemic diseases. Physical examination of the scrotum raised doubt about the presence of palpable bilateral vas deferens, while the testes and external genitalia were of normal size. A digital rectal examination was unremarkable. Semen analysis revealed normal volume, fructose-positive azoospermia. A hormonal profile of prolactin, luteinizing hormone (LH), follicle-stimulating hormone (FSH) levels and serum testosterone

levels were within the normal range. Transrectal ultrasound scan (TRUS) revealed bilaterally present seminal vesicles (Figure 1). Testicular biopsy showed normal spermatogonia (Figure 2). Contrast enhanced CT showed absent L/S kidney, and the rest of the study was unremarkable.

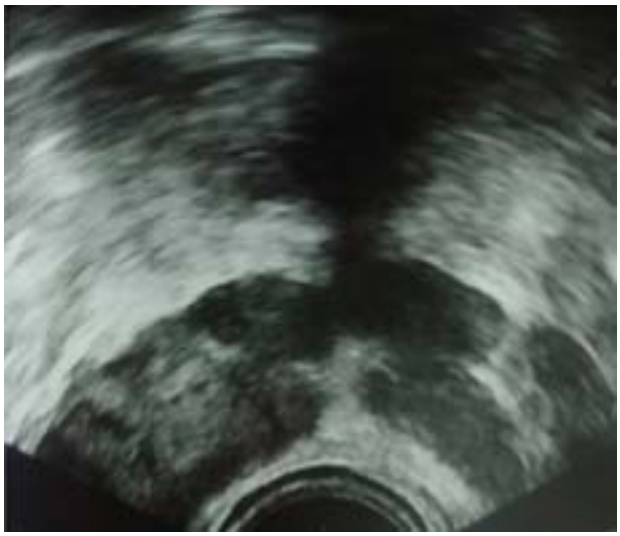

Figure 1. *TRUS shows normal B/L seminal vesicles.*

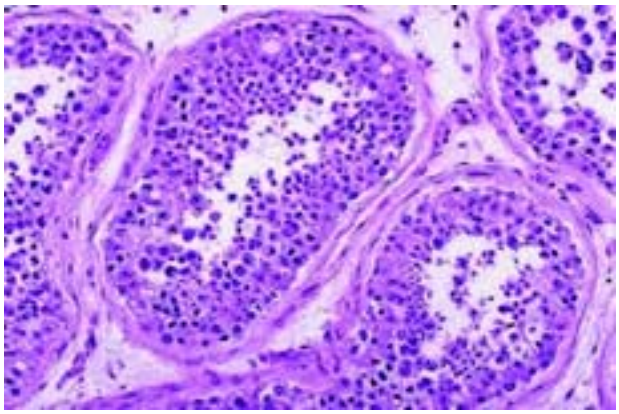

Figure 2. *Testicular biopsy showed normal spermatogonia.*

Under the impression of an obstruction in the vas deferens planned for vasogram and bilateral spermatic cord exploration done.

Bilateral inguinal exploration done under spinal anesthesia with the intention of performing vasogram and cord structures identified and explored. There were no spermatic cords present bilaterally. Subsequently, the patient and his wife received counseling and were referred for intracytoplasmic sperm injection (ICSI) as an assisted reproductive technique.

Discussion

Congenital bilateral absence of the vas deferens is a rare but significant cause of male infertility, characterized by the complete absence of both vas deferens. The testes typically function normally and produce sperm, but the absence of the vas deferens prevents sperm from being included in the ejaculate, resulting in azoospermia and infertility.

CBAVD accounts for approximately 1-2% of male infertility cases and about 6-8% of obstructive azoospermia cases (1). It is found in nearly all males with CF, with a prevalence of about 98% in this group, and can also occur as an isolated condition without other CF symptoms. The condition is typically diagnosed in adult men during infertility evaluations or systematic CF assessments. The primary cause of CBAVD is genetic mutations, most notably mutations in the CFTR gene, which is also implicated in cystic fibrosis (3). CFTR mutations are the main pathogenic factor, while mutations in the ADGRG2 gene are the second most common cause (1). CBAVD can present either as a mild manifestation of CF (CF-CBAVD) or as isolated CBAVD without other CF symptoms (4). Because of the genetic basis, there is a risk of passing these mutations to offspring, making genetic counseling essential for affected individuals and their partners (1). CBAVD is frequently accompanied by other congenital abnormalities, particularly involving the urogenital system. These include absence or hypoplasia of the seminal vesicles (present in about 50% of cases) and unilateral renal agenesis (5-10% of cases) (5). Such anomalies arise from developmental defects in the mesonephric duct during embryogenesis.

Physical examination may be insufficient in diagnosing CBAVD, especially in obese patients or those with high-riding scrotums, as the vas deferens may not be palpable (6). Transrectal ultrasonography (TRUS) and scrotal ultrasonography are versatile imaging tools to detect seminal vesicle and epididymal anomalies. Nevertheless, the definitive diagnosis may sometimes require surgical exploration, especially in cases with inconclusive findings, both clinical and imaging.

Men with CBAVD typically present with infertility characterised by low semen volume (<1.5 mL), acidic pH (<7.0), absence of fructose in semen, and

azoospermia. Despite the absence of the vas deferens, spermatogenesis within the testes is usually normal, allowing for the possibility of biological fatherhood through assisted reproductive technologies (ART).

Although CBAVD causes obstructive azoospermia, fertility can be achieved via sperm retrieval techniques such as testicular sperm extraction (TESE) or epididymal sperm aspiration, combined with ART like intracytoplasmic sperm injection (ICSI) (7). Genetic counselling and mutation screening, particularly for CFTR mutations, are recommended before ART to assess the risk of transmitting genetic defects to offspring.

Conclusion

In patients presenting with primary subfertility, a meticulous genital examination is essential. Diagnosing CBAVD requires high clinical suspicion, as physical examination of the cord structures and TRUS can offer important diagnostic clues. However, the condition is often diagnosed retrospectively. Fortunately, with preserved spermatogenesis, affected individuals can achieve biological parenthood through intracytoplasmic sperm injection (ICSI), making early recognition and appropriate referral crucial for successful fertility management.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient has given his consent for his images and other clinical information to be reported in the journal. The patient understand that their names and initials will not be published, and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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