

Unilateral Agenesis of the Right Seminal Vesicle Revealed During a Couple's Infertility Workup

Aziz Lamghari^{1*}, Youness Boukhlifi¹, Zakaria Abide², Abdessamad Elbahri¹, Mohammed Alami¹, Ahmed Ameer¹

¹Department of Urology, Mohammed V Military Hospital of Instruction, Rabat, Morocco

²Department of Radiology, Mohammed V Military Hospital of Instruction, Rabat, Morocco

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*Corresponding author: Aziz Lamghari

Department of Urology, Mohammed V Military Hospital of Instruction, Rabat, Morocco

Abstract

Case Report

Absence of seminal vesicles is a very rare cause of male infertility which poses diagnostic difficulties. In addition, its treatment is complex. In this article, we report the case of a 31-year-old patient who consulted for primary infertility evolving for 2 years (genetic counseling was performed) and we discuss the diagnostic and therapeutic difficulties related to this particular form of infertility. Our patient was referred for assisted reproduction after testicular biopsy.

Keywords: seminal vesicles, therapeutic difficulties, testosterone, blood karyotype.

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INTRODUCTION

The absence of vas deferens and seminal vesicles is a very rare cause of male infertility. Unilateral agenesis of the seminal vesicles is frequently found among the abnormalities of the male genital tract discovered in adulthood during a workup for excretory azoospermia [1]. Infertility related to internal genital tract anomalies is rare; it is estimated to be 2% of male infertility cases [2]. We report the case of one patient and discuss the diagnostic and therapeutic difficulties associated with this particular form of infertility.

OBSERVATION

A 33-year-old patient, married for 2 years, presented in consultation for infertility associated with bilateral testicular pain of progressive onset. Examination of the external genitalia (penis, scrotum, hernial orifices, testicles) was without abnormality apart

from an exquisite pain on bi-manual palpation of both testicles. Palpation of the spermatic cord was performed to verify the presence of the vas deferens and was completely normal. A spermogram performed on two occasions indicated azoospermia. The hormonal balance (FSH, LH, testosterone) was normal. A testicular ultrasound revealed bilateral micro-calcifications of more than twenty per testicle. A pelvic magnetic resonance imaging (MRI), requested as an additional etiological examination, revealed agenesis of the right seminal vesicle, with dilatation of the seminal vesicle most probably by stenosis of its terminal portion. (Fig. 1). Cytogenetic analysis (blood karyotype) concluded that there were no chromosomal abnormalities on all mitoses examined within the resolution of the metaphase karyotype. The patient underwent a testicular biopsy which confirmed the presence of live spermatozoa, and he was referred to an assisted reproduction center for further management.

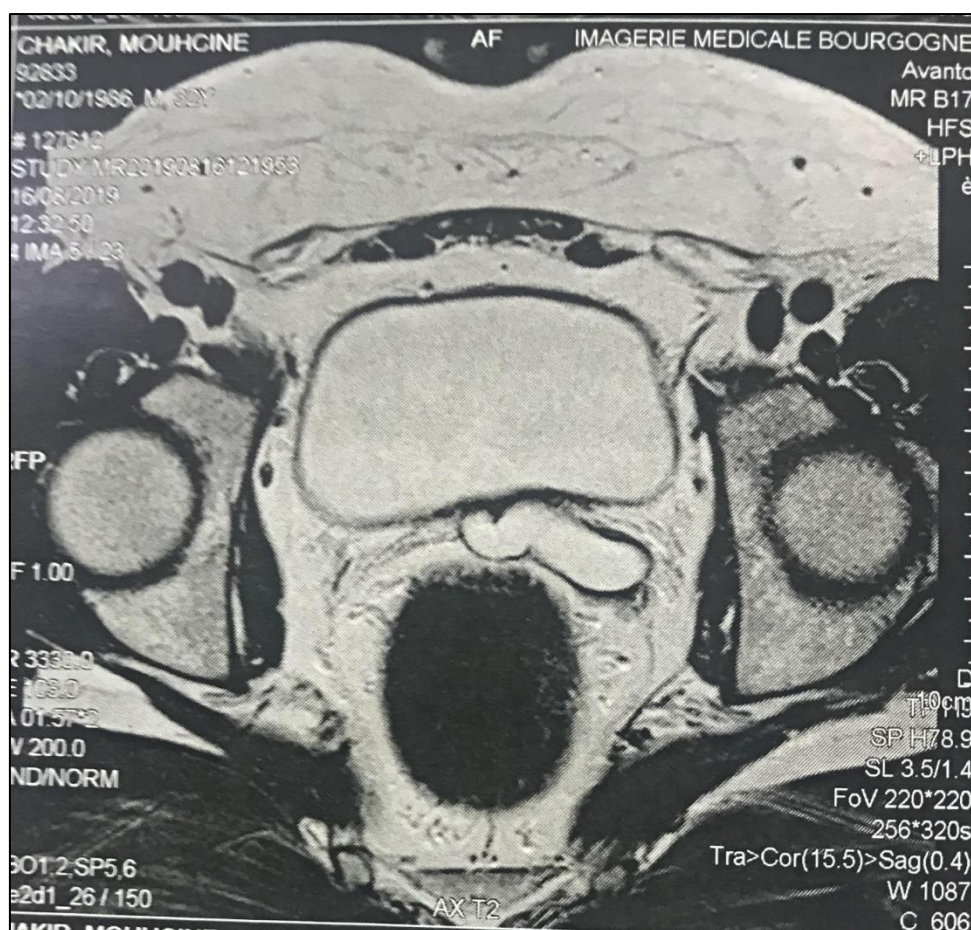


Figure 1: Magnetic resonance imaging of the deep seminal tract in coronal section in T2 sequence. Demonstration of the prostate. Absence of visualization of the right seminal vesicle with distension of the left seminal vesicle most probably related to stenosis of its terminal portion

DISCUSSION

Unilateral agenesis is often found during an infertility workup in the presence of hypospermia (ejaculate volume less than 2 ml). It should also be noted that an acidic pH (less than 7.2) associated with the absence of fructose in seminal biochemistry are characteristic elements of VS impairment in general. They can either correspond to an anomaly of embryological development of the mesonephrotic duct, or reveal a mutation of the ABCC7 gene in the context of cystic fibrosis.

They are frequently associated with developmental abnormalities of the vas deferens, the ureter and the kidney. If the anomaly occurs before the seventh week, a renal developmental anomaly (agenesis) may occur because the ureteral bud that induces the differentiation of the metanephros is always attached to the mesonephrotic duct. The developmental abnormality of a VS is always associated with an abnormality of the development of the vas deferens [3], either agenesis or ectopy. It may be an ectopy in a mullerian cyst, in the bladder or in the posterior urethra, causing recurrent epididymitis [4].

Clinical, radiological and genetic examinations allow us to consider different possibilities: unilateral or bilateral absence of the vas deferens, isolated or associated with renal agenesis, with or without absence of seminal vesicles, with or without cystic fibrosis mutation. Endorectal ultrasonography is a reliable examination to confirm the diagnosis of absent seminal vesicles suspected during clinical examination. Patients who have an isolated absence of the vas deferens associated with a cystic fibrosis mutation are at risk of transmitting the disease. This risk is estimated a priori at 1/50. In cases of isolated DKA associated with a cystic fibrosis mutation, the search for a genetic mutation in the spouse and genetic counseling are necessary before undertaking medically assisted procreation [5] [6]. In our case, the seminal vesicle agenesis was unilateral. The search for the genetic mutation in the patient was negative; there was no cystic fibrosis mutation gene. The search for a genetic mutation in the spouse was not necessary before undertaking medically assisted reproduction.

According to the study by Robin G *et al.*, [7], a transrectal ultrasound of the deep genital tract, routinely performed in the exploration of this infertility and complemented by pelvic MRI, showed isolated

unilateral agenesis of the left vas deferens in the pelvic portion. Given the presence of this anomaly, a search for mutations in the CFTR (cystic fibrosis transmembrane conductance regulator) gene revealed a composite heterozygosity (Delta F508/V938G). It was this study that first reported the association of these two mutations in the literature [7]. In our patient, the diagnosis of absence of seminal vesicles was made on endorectal ultrasound and confirmed by pelvic MRI. These two examinations confirm the diagnosis of seminal vesicle agenesis. Malformations of the male internal genitalia arising from the mesonephrotic duct, or Wolff's duct (epididymis, vas deferens, seminal vesicles, ejaculatory duct), are poorly known pathologies because of their rarity. Deferential agenesis is most often bilateral. It is characterized by the absence of the seminal vesicles and the ejaculatory duct. Mutation of the CFTR gene contributes to abnormalities in the development of the vas deferens [8, 9]. In the presence of deferential agenesis, cystic fibrosis should be investigated. In adults, the main presenting feature of bilateral agenesis is an abnormal sperm count [8]. Bilateral congenital absence of seminal vesicles and unilateral congenital absence of seminal vesicles are two causes of male infertility [10].

CONCLUSION

The diseases of the VS are rare, varied, and their diagnosis is often delayed because of their ignorance. Seminal vesicle agenesis, whether unilateral or bilateral, is a rare cause of infertility in couples. MRI, now indispensable for the diagnosis of VS disorders, allows a better study of the urogenital tract. As therapeutics evolve, the development of robot-assisted surgery will undoubtedly make it possible to standardize and facilitate dissection of this complex region in the future.

Abbreviations

FSH: Follicle Stimulating hormone.

LH: Luteinizing Hormon.

MRI: Magnetic Resonance Imaging.

CFTR: cystic fibrosis transmembrane conductance regulator.

VS: Vesicle Seminal.

ABCC7: ATP-Binding cassette C7.

DECLARATIONS

Ethics Approval and Consent to participate: Not applicable.

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