Case Report

Rare case of bilateral vassal agenesis and AZFc microdeletion: A case report

Mohammad Ali Sadighi Gilani[1,2*], Hesamoddin Sajadi[1]

1. Department of Andrology at Reproductive Biomedicine Research Center, Royan Institute for Reproductive Biomedicine, ACECR, Tehran, Iran
2. Department of Urology, Shariati Hospital, Tehran University of Medical Sciences, Tehran, Iran

Received: January 2018; Accepted: May 2018; Published online: 20 July 2018

Abstract:

Introduction: Bilateral vassal agenesis (CBAVD) is a rare cause of male infertility. Also one of the nonobstructive cause of azoospermia and infertility in men is microdeletion in long arm of Y chromosome. In this case report we have reported a rare case of CBAVD, LT renal agenesis and AZFc microdeletion.

Patient Information:

28 years old man, who was farmer with history of 6 years of infertility presented to our andrology clinic in Royan institute. In physical examination there was not any palpable vas deferens bilaterally, also The semen analysis was azoospermia. In sonographic evaluation, LT kidney wasn't seen in its anatomic location. The patient candidate for diagnostic PESA and bilateral PESA were negative. After that, the patient candidate for bilateral TESE and fascinatingly TESE was negative too. After genetic evaluation microdeletion was detected in AZFc subregion of Y chromosome.

Conclusion:

In men with obstructive azoospermia mostly testicular spermatogenesis is normal but this is not the rule because seldom spermatogenic dysfunction maybe present like our patient. We emphasize about the importance of consideration of obstructive and nonobstructive azoospermia together in a patient for urologists.

Keyword: Vassal Agenesis; Azoospermia; Azoospermic Factor

Cite this article as: Sadighi Gilan MA, Sajadi H. Rare case of bilateral vassal agenesis and AZFc microdeletion: A case report. Men's Health Journal 2018; 2 (1); e3.

1. Introduction

Bilateral vassal agenesis (CBAVD) is a rare cause of male infertility (1-2%) (1).

infertility treatment in CBAVD men include percutaneous sperm retrieval from epididymis and microinjection that almost ever is effective, because spermatogenesis usually is normal in this group of patients. (2) In bilateral vassal agenesis two point is critical:

First: because in men with CBAVD the chance of mutation in CFTR gene is high and about (60-90 %), so genetic consultation of CFTR gene is indicated (1). Genetic screening of men with CBAVD and their wives identifies the most common severe form of CFTR mutations like F508 that may result in clinically cystic fibrosis in their offspring (3).

***Corresponding Author:** Mohammad Ali Sadighi Gilani, Unit 502, Sahebgharanieh Medical building, Niloofar St., Aghdasieh, Tehran, Iran. Tel: +989125184529. Email: ali.sadighi@royaninstitute.org

Second: in vassal agenesis the lack of wolffian duct development on that side is possible (4).

Renal ultrasound in men with CBAVD is indicated because 11% of men with CBAVD have renal agenesis (5). One of the nonobstructive cause of azoospermia and infertility in men is microdeletion in long arm of Y chromo-some. Y chromosome is the determinant of male gender and is the only chromosome transmitted to the son from father (6). There is an area in the long arm of the Y-chromosome that is essential for spermatogenesis, this region is called AZF (7). Microdeletion in 3 regions of Y chromosome is associated with oligo or azoospermia which became known as AZF a, b, c (8). DAZ gene which is associated with spermatogenesis is located in AZFc region (9). In this case report we have reported a rare case of CBAVD, LT renal agenesis and AZFc microdeletion.

This topic reminding the coincidence of obstructive and nonobstructive azoospermia in a patient and propose more attention about mixed azoospermia in infertile men for urologists.

2. Patient Information:

28 years old man, who was farmer with history of 6 years of infertility presented to our andrology clinic in Royan institute.
In physical examination RT testis was 12 cc volume and LT testis was 20 cc volume and there was not any palpable vas deferens bilaterally.
The semen analysis was azoospermia with low volume (0.1 cc) and lack of fructose (0) with NL PH (7.8). hormonal tests were LH:10.9(relatively high), FSH:9.1 (NL), T:3.5(NL).
In sonographic evaluation, RT kidney was bigger than normal with compensatory hypertrophia and LT kidney wasn't seen in its anatomic location. Additionally, bilateral vassal and seminal vesicles agenesis was reported.
The patient candidate for diagnostic PESA and bilateral PESA were negative too.
After that, the patient candidate for bilateral TESE and fascinatingly TESE was negative.
The pathologist report was maturation arrest 70% and complete seminiferous tubule hyalinization (25%) and sertoli cell appearance (5%).
Then the patient was evaluated for genetic tests. (karyotype & AZF).
Karyotype was NL but microdeletion was detected in AZFc subregion of Y chromosome.
Finally, the patient selected for MDTESE and RT MDTESE was positive and sperm cryopreserved.

3. Methods:

Genetic evaluations in this case was include:
1.Peripheral blood specimen study on the basis of GTG technique at 500-500 band resolution revealing 46 chro-mosomes. No chromosomal aberration detected.
2.DNA extraction & analysis of the most common Y chromosome microdeletions, by using multipolymerase chain reaction and gel electrophoresis was detected microdeletion in AZFc sub-region of the long arm of Y-chromosome.

4. Discussion:

We have reported a rare case of CBAVD and renal agenesis (obstructive azoospermic cause of male infertility) asso-ciated with AZFc microdeletion (nonobstructive azoo-spermic cause of male infertility).
In our patient sperm retrieval with PESA and TESE was negative. Contradictory to vassal agenesis and obstructive azoospermic men that testes usually have normal spermatogenesis.
After genetic evaluation, it was determining that the patient had AZFc microdeletion and CBAVD.
Some studies suggest that in CBAVD men spermatogenesis is normal like Goldstein and Schlossberg (1998). (10)
But Maxwell reported that 33 CBAVD men underwent testis biopsy, and 4 of these patients (12%) had impaired spermatogenesis. After genetic evaluation one of them had AZFb microdeletion. (11)
Also Okada et al studied that 30% of CBAVD men had histological evidence of hypo spermatogenesis. (12)
In our patient we can find out that in men with CBAVD, testicular spermatogenesis dysfunction may be accompanied.
We believe our patient was a case of mixed azoospermia (obstructive and nonobstructive), so in known cases of azoospermia we must consider other causes of male infertility.

5. Conclusion:

In men with obstructive azoospermia mostly testicular spermatogenesis is normal but this is not the rule because seldom spermatogenic dysfunction maybe present like our patient.
So if the sperm retrieval was negative in this group of patient, genetic testing includes karyotype analysis and AZF is indicated.
We have reported this case because of unique and rare presentation.
At last we will emphasize about the importance of consideration of obstructive and nonobstructive azoospermia together in a patient for urologists.

6. Acknowledgment

None.

7. Conflict of interest:

All authors declare that there is no conflict of interest in this study.

8. Funding support:

All authors declare that this study was accomplished without any funding or support.

9. Author's contributions:

All the authors have contributed to drafting/revising the manuscript, study concept, or design, as well as data col-lection and interpretation.

10. Reference

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