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CLINICAL AND GENETIC STUDY IN MALE INFERTILITY WITH AZOOSPERMIA

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Introduction

- Worldwide, it has been estimated that about 7% of all men experience infertility. About 20% of the causes of male infertility are of genetic etiology.
- The most common genetic causes reported are chromosomal abnormalities and Y chromosome microdeletions.

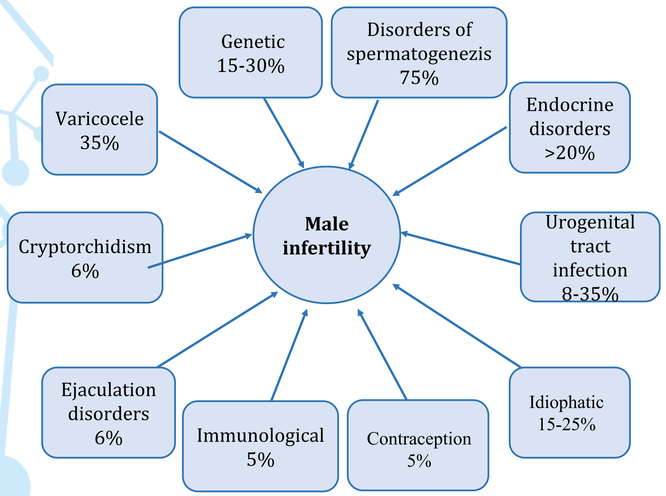

Figure1: Distribution of the most common causes of male infertility

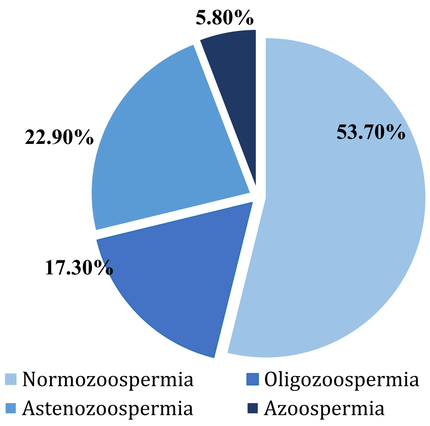

Figure 2. Semen structure to infertile men during 2019-2020 from RM

Keywords

- male; infertility; azoospermia; karyotype; Y Chromosome

Purpose

- Study of chromosomal variations, Y chromosome microdeletions and mutations in the CFTR (Cystic fibrosis transmembrane conductance regulator) gene in men with azoospermia.

Material and methods

- 96 men with azoospemia were investigated by cytogenetic classical G-banding technique.
- The genomic DNA was isolated and used for the analysis of AZF microdeletons and mutations in the CFTR gene by the PCR technique. The AZFα regions (sY84, sY86, DBY1, sY620), AZFb (sY117, sY127, sY134, SY143), and AZFc (sY254, sY255, sY153, SY158) were analyzed. ΔF508 and G542X were tested for the CFTR gene.

Results

- Of 96 cases of men with azoospermia, 35 (36.4%) showed genetic variations and 61 (63.6%) without changes.
- In the 35 patients, in 24 (25%) cases chromosomal abnormalities were found, in 10 (9.6%) patients the microdeletions of the Y chromosome in the AZF region, of which in 8 cases they presented normal karyotype 46,XY and in 2 cases variations in karyotype. In 3 (3.1%) men were diagnosed as carriers of mutations in the CFTR - ΔF508 gene; for calculating the risk of recurrence in offspring were also investigated their wives, who were homozygous.

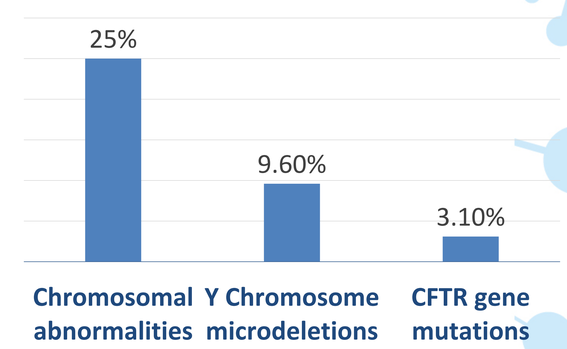

***Figure 3. Frequency of* genetic variations in 96 men with azoospermia**

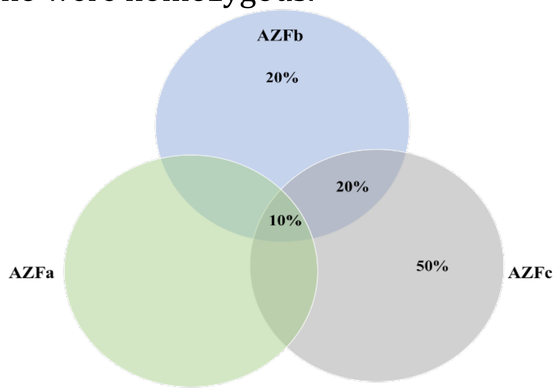

Figure 4. Venn diagram illustrating frequencies of different type of detected Y microdeletions in AZF regions. AZFc deletion were involved in 50% (5/10) of total deletions respectively. AZFb deletion were detected in 20% (2/10), combined deletions including AZFb and AZFc were also detected in 20% of all total deletions. Large deletion of all regions AZFa, AZFb and AZFc were found in 10% (1/10).

Regions	Number of men with azoospermia	Frequency of microdeletions from the AZF region
Moldova	**96**	**9.6%**
India	119	7.6%
Spain	57	14%
China	137	8.7%
	945	11.5%
Algeria	49	2.0%
Japan	60	11.7%
Tunisian	76	11.8%
Turkey	52	1.3%
USA	385	10.4%
Netherlands	37	8.1%
Brazil	60	6.6%
Mexico	50	12%
Jordan	34	8.3%

Figure 5. Frequency of Y chromosome microdeletions in selected populations

Conclusion

- Clinical-genetic evaluation of couples with male infertility associated with azoospermia is necessary, not only for the correct establishment of the diagnosis but also for their treatment.