Clinical Urology

International Braz J Urol

Vol. 37 (2): 244-251, March - April, 2011
doi: 10.1590/S1677-55382011000200011

Chromosomal and Molecular Abnormalities in a Group of Brazilian Infertile Men with Severe Oligozoospermia or Non-Obstructive Azoospermia Attending an Infertility Service

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ABSTRACT

Purpose: To determine the frequency of genetic alterations in a population of Brazilian infertile men with severe oligozoospermia or non-obstructive azoospermia.
Materials and Methods: Retrospective study of a group of 143 infertile men with severe oligozoospermia or non-obstructive azoospermia from the Andrology Outpatient Clinic of the Human Reproduction Service at the ABC School of Medicine. Of these patients, 100 had severe oligozoospermia, and 43 non-obstructive azoospermia. All patients underwent a genetic study which included karyotype analysis and Y-microdeletion investigation.
Results: Genetic abnormalities were found in 18.8% of the studied patients. Chromosomal abnormalities were found in 6.2% of the patients, being more prevalent in the azoospermia group (11.6%) than in the oligozoospermia group (4%). Chromosomal variants were found in 8.3%, and Y-chromosome microdeletions in 4.2% of patients.
Conclusion: The high frequency of genetic alterations (18.8%) in our series justified performing a genetic investigation in a population with idiopathic infertility, as results may help determine the prognosis, as well as the choice of an assisted reproduction technique. Moreover, a genetic investigation could minimize the risk of transmitting genetic abnormalities to future generations such as genetic male infertility, mental retardation, genital ambiguity and/or birth defects.

Key words: *male infertility; chromosome abnormalities; Y chromosome; microdeletions; oligozoospermia; azoospermia*
Int Braz J Urol. 2011; 37: 244-51

INTRODUCTION

Infertility is a very common health problem, affecting approximately 15-20% of couples who attempt pregnancy. In almost 50% of infertile couples, the problem is related to the male. About 15% of infertile men may carry a genetic abnormality, including chromosomal aberrations and single-gene mutations (1,2).

The frequency of chromosomal aberrations in subfertile males is estimated to be 2-3%, and in infertile patients with sperm counts below 10 x 106 spermatozoa/mL may reach 5-7%; among patients with azoospermia, the percentage of individuals with cytogenetic abnormalities increases by 10-15% (1,2).

Among the genetic abnormalities found in infertile men, those involving chromosome anomalies