INTRODUCTION

Infertility is defined as the inability to conceive after one or more years of unprotected intercourse and occurs in approximately 15% of the population. Approximately half of the cases are due to male factors. In fact, infertility affects about 7% of all men. Male factor infertility is considered a complex disorder with a largely unknown etiology. In general, genetic abnormalities are thought to account for 15%-30% of this condition and chromosome abnormalities and Y chromosome microdeletions are frequently involved.

The study, based on the caseistic, aimed at contributing to a better understanding of the genetic causes of infertility, in order to improve genetic counseling of these conditions.

RESULTS AND DISCUSSION

In the 410 samples, 42 abnormal karyotypes (10.2%) were found, indicating an elevated frequency of chromosome abnormalities among the selected infertile men (Table I), when compared to that of newborn populations (~0.4%). This frequency is higher than that reported in most similar studies that pointed to frequencies ranging from 2.2%-14.2%. There are 27 sex chromosome abnormalities and 15 autosomal (structural) rearrangements.

6. Sex chromosome anomalies are present in 6.6% of total cases and represent 64.3% of the chromosome abnormalities. As expected, Klinefelter’s syndrome was the most common chromosome disorder (4.4%).

2 cases with 46,XX,der(X)(X;Y) occur. In these cases SRY is present in the X chromosome and regions AZFa,b,c were deleted (Figure 1). The X Y bivalent is particularly susceptible to errors in meiosis because of homology between the X and the Y chromosome. A case with a mosaic marker chromosome i(Y) presents two signals for SRY (Figure 2).

Reciprocal translocations were identified in 10 cases (2.4%), particularly in men with OLG0, OTA and NQ (Table I) and may have a strong impact on the spermatogenesis process lead to oligozoospermia or even azoospermic. In fact, chromosome translocations may cause reductions in testicular volume and testosterone level, which may impact spermatogenesis, resulting in male infertility. A review study reveals that have been found in approximately 1% of the infertile men and are more common in azoospermic males, and a value and a relation not found in the present study.

Y microdeletions cases were identified in 16 of the 247 Y microdeletions cases studied (6.5%) with a total of 23 deletions (Table II). There are more frequent in azoospermics (13.3% of this group), corresponding to 8/60 azoospermics. Among these cases, 7 presented deletions at the AZFc region and 3 presented deletion at the entire AZF region.

Oligozoospermics present exclusively deletions on AZFc region. Men with complete AZFc deletions have variable seminial and testicular phenotypes, with sperm products levels ranging from azoospermia to oligozoospermia.

- Sperm counts and other clinical data are essential for results interpretation and a genotype/phenotype correlation. The marked presence of chromosome abnormalities and Y microdeletions emphasizes the relevance of studying both factors in infertile men to improve genetic counseling, to allow the development of appropriate therapies, and to expand the knowledge about the etiology of male infertility.