Cytogenetic analyses of male infertility

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INTRODUCTION

Infertility is a failure to conceive after at least one year of unprotected intercourse(1). It affects about 10-15% of couples in reproductive age(2). It is often suggested that a male factor problem is implicated in 50% of cases. The genetic landscape of male infertility is highly complex, and cytogenetic abnormalities represent an important cause.

The purpose of this study was to determine the type and the rate of cytogenetic abnormalities among infertile men attending the Pasteur Institute of Tunis.

RESULTS

A total of 630 Tunisian infertile patients with non-obtructive azoospermia (NOA) (n=447) and severe oligozoospermia (n=183) were included in this study.

Cytogenetic analyses were performed from peripheral blood lymphocyte culture. Chromosomes were analyzed by RHG-banding. For each patient, at least 20 well-spread metaphases were analyzed.

The chromosomal abnormality rate was 10.63 % (67/630).

Out of the 67 patients, 50 showed numerical aberration, 14 others patients presented structural abnormalities and 3 had a supernumerary marker chromosomes (SMC). Gonosomal aberrations were the most frequent in the NOA group, whereas, autosomal ones were the most frequent in the severe oligozoospermic group (Table I and Table II)

In the NOA group, 12.75% of patients had abnormal karyotype. The most frequent was Klinefelter syndrome (44/57) followed by autosomal translocation (6/57), XYY syndrome (2/57) and XX male (2/57)

Among the 183 oligozoospermic males, 5.46 % (10/183) showed an abnormal karyotype. Structural abnormalities were the most frequent (6/10) and consisted mainly in balanced translocation (3/10). Numerical aberrations were seen in only 2 patients with Klinefelter syndrome.

Table I: Gonosomal abberration

Karyotype	Non-Obstructive Azoospermia (n/%)	Severe oligozoospermia (n/%)
47,XXY	39 (8,72)	1(0,55)
46,XY/ 47,XXY	4 (0,89)	1(0,55)
47,XYY	1(0,22)	0
46,XY/47,XYY	1(0,22)	0
46,XX	2(0,44)	0
47,XXY/48,XXXY	1(0,22)	0
46,XY, del (Y)	1(0,22)	0

Table II: Autosomal abberrations

Karyotype	Non-Obstructive Azoospermia (n/%)	Severe oligozoospermia (n/%)
45 XY, der(13,14)(q10;q10)	1(0,22)	1(0,55)
45 XY -15,-22,+der t(15,22)	0	1(0,55)
46 XY, t(1,17)(q11;p12)	0	2(1,09)
46 XY, t(4,19)(p16;q11)	0	1(0,55)
46 XY, t(4,6)(p12;p22)	1(0,22)	0
46 XY, t(4,17)(q11;p11)	1(0,22)	0
46 XY, t(7,16)(p11;p13)	1(0,22)	0
46 XY, inv(7)(q22;q35)	1(0,22)	0
46 XY, t(9,22)(q11;p11)	1(0,22)	0

CONCLUSION

Chromosomal alterations found with a high frequency in our series of infertile men are a major cause of male infertility. This highlights the need for cytogenetic analysis for every infertile man, especially before assisted reproductive techniques.

REFERENCES

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