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Original article

Cytogenetic analysis and endocrine profile in patients with nonobstructive azoospermia or severe oligozoospermia

M.H. Ali [a,*], **M. Soliman** [b], **A. Metwally** [a], **A. Ghobeish** [b]

[a] *Urology Department, Faculty of Medicine, Suez Canal University, Ismailia, Egypt*
[b] *Ismailia General Hospital, Ismailia, Egypt*

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Abstract
Objective: To study the prevalence of chromosomal anomalies in infertile males with severe oligozoospermia or non obstructive azospermia and its correlation with clinical and endocrine profile.
Patients and methods: Consecutive 30 male subjects (mean age 35.5 ± 7.1 years) with primary infertility attending at the infertility clinic, Urology department, Suez Canal University Hospital, Egypt were enrolled in the study. These patients had severe oligozoospermia (n = 9) or non obstructive azospermia (n = 21). Clinically testicular volume, scrotal Doppler ultrasound examination and endocrine evaluation (serum FSH, testosterone and prolactin) were determined. Cytogenetic analysis was performed by using the GTG (G-banded using trypsin and Giemsa) banding technique.
Results: Nine patients (30%) had chromosomal abnormality. Patients with Klinefelter Syndrome and de la Chapelle male syndrome represented 26.7% (n = 8) and 3.3% (n = 1) respectively. All patients diagnosed as Klinefelter group were azoospermic, while 57.1% of normal karyotyping were azoospermic and 42.9% were severe oligozoospermic ($p = 0.029$). Klinefelter group had significantly lower mean testosterone level than normal karyotyping group ($p = 0.016$). Also, Klinefelter group had significantly higher mean FSH and LH levels than normal karyotyping group ($p < 0.01$). The anomaly detected was 47, XXY chromosomal constitution, found in 8 (38%) out of 21 patients with non-obstructive azoospermia.
Conclusion: There is a high prevalence of chromosomal abnormalities in infertile males with non obstructive azoospermia. All patients with azoospermia and severe oligozoospermia (sperm count <5 million/ml) should undergo genetic screening. Our study indicates that even those presenting to infertility clinics can be heterogeneous in terms of karyotype and phenotype.

* Corresponding author.
E-mail address: hassanurol@yahoo.com (M.H. Ali).
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Introduction

Infertility represents a considerable health issue affecting up to 17% of couples. Male factors contribute to 30–50% of the problem [1]. Chromosomal abnormalities are more frequent in infertile males compared to general male population [2]. These abnormalities have been reported in a higher frequency in males with severe oligozoospermia and non obstructive azoospermia than moderate or mild oligozoospermia [3].

Variable numerical and/or structural abnormalities of sex chromosomes were encountered in infertile males [4].

Klinefelter syndrome (47, XXY, mosaic) represents the most common karyotypic abnormality in men with oligozoospermia or azoospermia and can be identified clinically by characteristic body proportions, gynecomastia, firm and small testis and associated endocrine and nonendocrine disease [5].

The chromosomal anomalies, and its prevalence and impact on infertility in the Egyptian population is not yet well studied. The present work was therefore an endeavor to verify the prevalence and patterns of chromosomal anomalies in these patients in our province at Suez Canal region.

We performed cytogenetic analysis in case of male infertility having severe oligozoospermia or nonobstructive azoospermia. The karyotype findings were correlated with physical and endocrine profile.

Patients and methods

This study was conducted as a descriptive cross-sectional study including 30 consecutive males with primary infertility during 12 month period (March 2012–March 2013). The mean age of patients was 35.5 ± 7.1 years with a range of 19–58 years.

Males with primary infertility (n = 30) and having non-obstructive azoospermia (n = 21) or severe oligozoospermia (sperm counts ≤ 5 millions/ml).; n = 9) were evaluated at Urology Department, Suez Canal University Hospital. An informed written consent was obtained from every patient participating in the study. The study design was approved by the Ethics Committee of the Faculty of Medicine, Suez Canal University, Ismailia, Egypt.

Patients were subjected to detailed structured history taking, physical examination, testicular volume measurement and digital rectal examination.

Every patient provided two semen specimens, each after 4 days of sexual abstinence. On the basis of their mean sperm concentrations, the patients were categorized as having azoospermia or severe oligozoospermia (≤5 million sperms/ml).

For all patients, blood samples were obtained in early morning for the measurement of serum testosterone, FSH and LH by radioimmunoassay.

Peripheral blood samples were collected from all patients into heparinised test tubes. Cytogenetic analysis was performed using the GTG (G-banded using trypsin and Giemsa) banding technique [6].

The karyotypes were described according to the ISCN (International System for Human Cytogenetic Nomenclature). Scrotal Doppler ultrasound was performed to evaluate the testicular size, vascularity and the evidence of varicocele. Testicular volume was calculated using Hansen formula for a prolate spheroid: length (L) × width (W) × height (H) × 0.52. Normal testicular volume is 12–30 ml [7].

Statistical analysis

Collected data were coded, entered and analyzed using Microsoft Excel software. Data were then imported into Statistical Package for the Social Sciences (SPSS version 16.0) software for analysis. Baseline characteristics of the study population were presented as frequencies and percentages (%) in qualitative data or mean values and standard deviations (SD) in quantitative data. Differences between frequencies and means were compared by Chi-square and paired samples *t*-tests, respectively. A p value of <0.05 was considered significant.

Results

There was no significant difference between both groups of normal and abnormal karyotyping regarding age distribution (p > 0.05). The mean age of Klinefelter group was slightly higher than normal group, but without significant difference (p > 0.05) (Table 1).

Azoospermia was reported in 21 patients (70%), while severe oligozoospermia was reported in 9 patients (30%). The mean sperm count of the patients with severe oligozoospermia was 1.98 ± 1.85 millions/ml with a range of 0.3–5 million/ml.

Nine out of thirty studied patients (30%) had chromosomal abnormality. Patients with Klinefelter Syndrome (n = 8; 26.7%) and one patient (3.3%) with de la Chapelle male Syndrome.

Table 1 Age distribution in normal karyotyping (n = 21) and Klinefelter (n = 8) groups.

Age (years)	Normal karyotyping (n = 21)		Klinefelter (n = 8)		Used test	p-value
	No.	%	No.	%		
19–<30	2	9.6	0	0.0	$X^2 = 1.7$	0.44
30–<40	15	71.4	5	62.5		
40–58	4	19.0	3	37.5		
Total	21	100.0	8	100.0		
Mean ± SD	34.3 ± 5.8		39.38 ± 9.1		t = 1.8	0.08
Range	19–44		31–58			