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The Role of Karyotyping in the Diagnosis of Male Infertility

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Abstract

Introduction: Male infertility remains a taboo subject, often perceived as a source of shame in Algerian society. The aim of this study is to determine the prevalence of male infertility, describe its clinical and biological characteristics, and focus on genetic abnormalities that may lead to male infertility.

Materials and Methods: This is a descriptive and analytical cross-sectional study involving 118 infertile patients who consulted the CHU Ibn Rochd for infertility, contributing to couple infertility.

Results: The most represented age group was 30–35 years, accounting for 24.6% of the patients. Among the 118 patients, 46 were azoospermic (39%), of whom 15 patients (83%) had a pathological karyotype with a detected severe spermatogenic disorder (SSD).

Regarding the chromosomal origin of infertility, gonosomal abnormalities accounted for 83%, whereas autosomal abnormalities were found in only 17%. The same distribution was observed for numerical and structural chromosomal abnormalities.

Conclusion: In cases of male infertility, the challenge is twofold: to meet the couple's pressing desire for conception and to establish an accurate etiological diagnosis, particularly based on cytogenetic analysis. Indeed, certain chromosomal abnormalities may underlie impaired spermatogenesis.

Keywords: Chromosomal abnormalities,Ychromosome deletion, male infertility, cytogenetics.

Introduction

Male infertility, due to its frequency and impact on quality of life, constitutes a major public health issue. Its exact prevalence is difficult to determine because of the lack of precise global data [1].Male responsibility is systematically assessed, even in the presence of a female abnormality.

The role of the male partner in infertility has been the focus of numerous studies in recent years. According to some estimates. Infertility within a couple can be exclusively female or male in origin, but it is often the result of combined subfertility, where the interaction of both partners' factors significantly reduces the couple's ability to conceive [2], which, through a synergistic effect, impairs the reproductive capacity of the couple [3].

2.Materials and methods

This is a descriptive cross-sectional study, based on recruitment through clinical interviews and review of

complete medical records. A blood karyotype was performed for each patient, along with at least one semen analysis conducted during follow-up.

The study sample includes male patients experiencing infertility for more than one year, who were referred to our seervice.

The objective of the study is to analyze the karyotype results of infertile men in addition to their semen analysis, in order to better understand chromosomal abnormalities associated with male infertility and to address the research objectives outlined in our study protocol.

2.1.Patients

The sample consisted of male patients suffering from infertility for more than one year who were referred to our laboratory.

All patients provided informed consent for the performance of blood karyotyping.

Our study consisted of analyzing the karyotype results of infertile men as well as their semen analysis results.Only patients with infertility lasting more than one year were included in our study.

Inclusion Criteria

-Infertile couples married for more than one year without the use of contraception.

-Patients referred for preoperative evaluation (varicocele, ectopic testis, torsion, or testicular tumor) or for follow-up visits.

-Patients presenting with oligozoospermia who underwent blood karyotyping.

Non-Inclusion Criteria

-Infertile couples who had discontinued contraception for less than one year.

Exclusion Criteria

-Children and adolescents with **disorders of sex development (DSD)** in whom male fertility may be affected in the future.

2.2.Methods

2.2.1.Study design and data collection: In our study, patients were generally referred to our department to undergo karyotype analysis in the context of male or couple infertility. A structured interview was conducted to clarify the essential elements supporting the diagnosis. A standardized technical information form was established for each patient. This form consisted of 15 questions written in French and explained in Arabic during the interview. The questions were mainly closed-ended and mostly dichotomous. The form included several sections: an identification section (demographic data), general medical history, and a history of genital tract disorders.

2.2.2.Peripheral bloodkaryotyping technique: For cell culture, two heparinized tubes were prepared per patient. Five milliliters of RPMI culture medium were added to each tube, followed by 0.5 mL of peripheral blood sample with phytohemagglutinin. The culture tubes were incubated at 37°C for 72 hours. Each morning, all culture tubes were gently agitated. Cell division was arrested using colchicine. A hypotonic shock was induced using a KCl solution, followed by at least three fixation steps with methanol and acetic acid; after each fixation, the supernatant was discarded while retaining the cell pellet. The cell suspension was then spread onto slides and stained with Giemsa. The slides were examined under a light microscope. Generally, at least 20 metaphases with 46 chromosomes were counted, and 5 metaphases were karyotyped.

2.2.3.Semenanalysis(spermogram and spermocytogram): Semen analysis was performed after 3 to 5 days of sexual abstinence. The sample was incubated at 37°C and analyzed after two hours. Numerical and cytological parameters, as well as typical and atypical forms, were assessed according to WHO standards.

2.2.4.Statistical analysis: All data were entered and analyzed using SPSS version 20.0, EPIDATA 3.1, Excel 2016, and MedCalc® version 18.11.6.

Quantitative variables (continuous or discrete) were expressed as mean ± standard deviation, while qualitative variables (ordinal or nominal) were presented as totals and percentages.

The statistical tests used for comparisons between subgroups included:The Chi-square (χ^2) test for qualitative variables,Pearson's correlation coefficient to assess the relationship between two continuous variables,Fisher's exact test to evaluate associations

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between two categorical or binary variables,Student's t-test for comparison of quantitative variables.

The level of statistical significance (p-value) was set at 0.05. Associations between variables were considered statistically significant when $p < 0.05$, with a 95% confidence interval (CI).

3.Results

A total of 150 patients meeting the inclusion criteria were recruited ; only 118 patients could be included, while the others were excludedOur study included a sample of 150 patients.

The mean age was 35–45 years, with extremes ranging from 18 to 60 years. 17.8% of subjects were 45 years or older. Primary infertility was observed in 44.1% (n = 52) of cases, and in cases of azoospermia, it accounted for 23.7% (n = 28).

Among these patients, 52 requested a karyotype as part of a primary infertility workup, representing the main reason for consultation, whereas only 28 requested it in the context of azoospermia.

Regarding karyotype results (n = 118), 84.75% (n = 100) were normal, while 11.02%

(n = 18) were pathological. The pathological anomalies included: homogeneous Klinefelter syndrome (11.02%), mosaic Klinefelter syndrome (0.85%), gonadal dysgenesis (0.85%), and Robertsonian translocation (2.54%, 3 cases), bringing the total rate of chromosomal abnormalities to 15.26%.(see table 1).

Several types of chromosomal abnormalities were identified in our patients. The table above summarizes all these anomalies, indicating the number of cases found in the study sample.Among the 18 abnormal cases (n = 18), 83.3% were gonosomal abnormalities, including Klinefelter syndrome (both homogeneous and mosaic forms) and Turner syndrome (45,X / 46,XY). Only 16.7% of the anomalies were Robertsonian translocations (45,XY,t(13;14)), representing autosomal abnormalities.

In summary, among these anomalies, 18 cases involved numerical abnormalities, while 3 cases involved structural abnormalities. The homogeneous form of Klinefelter syndrome accounted for 72.3% (n = 13) of cases, whereas the mosaic forms accounted for 27.7% (n = 5). These abnormalities include autosomal, gonosomal, and numerical anomalies (see Table 2).

In our population, primary infertility accounted for 61.1% of cases (n = 11). Karyotype analysis showed that 41% of patients had a normal karyotype. Among patients consulting for azoospermia (n = 3, 16.7%), all presented a pathological karyotype.

Semen analysis, interpreted according to WHO criteria, Semen analysis, interpreted according to the WHO criteria, was performed in 118 patients. Quantitative abnormalities were dominated by azoospermia (38.8%), followed by a normal semen analysis (28.5%), oligoasthenoteratozoospermia (OATS) (19%), oligospermia (8.6%), and asthenospermia (5.2%).

Among the 46 patients with azoospermia, 83% had a pathological karyotype, while 31 patients had a normal karyotype. In patients with oligospermia, 11,1 % had a pathological karyotype. A significant correlation between chromosomal abnormalities and the karyotype was observed ($p = 0.0005$)

These results indicate that azoospermia is the most frequent spermatological abnormality and is strongly associated with chromosomal abnormalities, unlike other spermatological disorders, which show a weaker correlation with the karyotype.(see table 3).

4.Discussion

Several studies have investigated the issue of male infertility. In the present study, we compared our findings with those of major studies conducted in different geographic settings, including :the Mexican study by Meza-Espinoza [4] ;the Moroccan study by Addouroudj [5] ;the French study by Thonneau [6];the Tunisian study by Jaballah [7]

;the Brazilian study by Pina-Neto [8]and the Senegalese study by Sakandé [9]

However, other multicenter studies were not included in this discussion because of methodological differences and research protocols that were not comparable to those used in our study.

4.1.Biases and Limitations:despite its simplicity, this study has several limitations.

Infertility remains a sensitive health issue that touches on patients' modesty and privacy. Some individuals do not feel concerned and may refuse to acknowledge

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scientific evidence. As one Algerian woman explained: *"My husband refuses to admit the idea that he could be infertile or have problems of this kind."* This sociocultural factor may lead to underreporting and selection bias.Paradoxically, Algeria recorded a historic increase in births, reaching 880,000 in 2010. However, regarding fertility issues, Professor Mostapha Benzine, Head of the Social Studies Division at the National Economic and Social Council (CNES), confirmed the lack of statistical data: *"There is no census or study on infertility in Algeria."* This absence of national data limits the generalizability and contextual interpretation of our findings.The infertility rate is estimated to affect approximately 12% of Algerian couples. Among the couples seen by Professor Chitour, a specialist in endocrinology, male infertility accounts for 35% of cases, compared with a global average of about 40%.

The size of our study population is comparable to that reported by Sakandé et al [9] who included 220 patients, and to that of Jaballah [7] which involved 373 subjects. In contrast, the study by Thonneau et al. [6] ,published in 1999, included a substantially larger sample of 1,467 patients. Conversely, Belmokhtar [10]. reported a markedly smaller sample size (n = 27), while Addouroudj examined an intermediate sample size of 85 patients [5].

4.2.Characteristics of the Study population : Male infertility is a major public health issue worldwide. More than 70 million couples are affected globally, corresponding to an estimated prevalence of approximately 15%. In Algeria, about 12% of couples of reproductive age are affected, and it is estimated that male factors account for nearly 40% of infertility cases [11],this variability may be explained by differences in recruitment strategies and inclusion criteria across studies. However, several authors report that men are involved in nearly half of infertility cases, a finding that is consistent with the results of our study (n = 118).

4.3.age: The mean age of infertile men in our series was 35 years, with extremes ranging from 18 to 60 years. Age is well known to influence male fertility. In our study, 24.6% of infertile men were aged between 30 and 35 years, while 22.9% were between 35 and 40 years, with a mean age of 35 ± 8 years. This age distribution may be explained by the early identification of infertility and the inclusion criteria adopted in our study.Most studies have demonstrated that paternal aging has a significant impact on spermatogenesis, leading to a decline in sperm parameters, including sperm count, motility, viscosity, and morphology. Several contributing factors have been implicated, such as the increasing prevalence of comorbidities, including cardiovascular disease, hypertension, diabetes, obesity, hormonal disorders such as hypothyroidism, prostatitis, hydronephrosis, and renal microlithiasis,furthermore, many studies emphasize the deterioration of spermatogenesis and alterations in sperm DNA integrity when paternal aging is associated with additional risk factors. In contrast, Belloc et al [12] reported conflicting results, suggesting that the impact of paternal age on sperm parameters may not be as pronounced under certain conditions.

4.4.Normal or pathological karyotype. :Our findings regarding normal and pathological karyotypes are consistent with those reported by Rezgoune [11] but differ from results observed in other studies cited below, particularly the study by Pina-Neto [8]**.**

4.5.Autosomal and sex chromosome abnormalities :which reported a lower percentage of pathological karyotypes,Chromosomal abnormalities involving autosomes and sex chromosomes, whether numerical or structural, are among the most frequent causes of genetic infertility. In our series of 118 patients, 100 individuals (84.75%) had a normal karyotype. Homogeneous Klinefelter syndrome (47,XXY) was identified in 11.02% of cases, while mosaic Klinefelter syndrome (47,XXY/46,XY) accounted for 0.85%. Gonadal dysgenesis corresponding to Turner syndrome (45,X/46,XY) was observed in 0.85% of patients. Robertsonian translocations (45,XY t(13;14;15)) were identified in 2.54% of cases, corresponding to three patients.

An African study reported a rate of 8.2% of male infertility; among the 117 patients evaluated, 61 were aged between 35 and 44 years. In that study, 85.5% of patients presented both qualitative and quantitative sperm abnormalities associated with sex chromosome anomalies [13],Regardless of male age or the underlying genetic abnormality, any couple undergoing insemination should receive prior ovarian stimulation combined with pelvic ultrasound monitoring and hormonal assessment in order to

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minimize the risk of multiple pregnancies [14]. In infertile patients, these findings are consistent with those reported by Addouroudj. In that study,

4.6.Numerical and structural abnormalities :83.3% of cases presented sex chromosome abnormalities, mainly Klinefelter syndrome (homogeneous or mosaic) and Turner syndrome (45,X/46,XY). Robertsonian translocations (45,XY t(13;14)), representing structural chromosomal abnormalities, accounted for 16.7% of cases. Y chromosome microdeletions (del Yq) were also reported; however, this abnormality could not be detected using conventional cytogenetic methods.

A French study involving 14 infertile patients carrying Y chromosome microdeletions identified by PCR analysis divided participants into two groups, while 11 fertile men served as controls [15],Three patients from the subgroup (n = 6) underwent fluorescence in situ hybridization (FISH) using X- and Y-centromeric probes. The results revealed extensive and submicroscopic deletions of the long arm of the Y chromosome, associated with high rates of 45,XO cells in nullisomic spermatozoa, particularly involving the Y chromosome. In contrast, two isodicentric Y chromosomes were classified as normal using conventional cytogenetic banding techniques. These findings support the hypothesis that Y chromosome microdeletions may be associated with Y chromosome instability, leading to the formation of 45,XO cells [15],

4.7.The homogeneous and mosaic forms :The homogeneous form accounted for 72.3% of cases, corresponding to Klinefelter syndrome, while mosaic forms (autosomal and gonosomal) represented 27.7%. Although these chromosomal abnormalities are likely to result in sterility, it should be noted that in France, out of approximately 800,000 births per year, nearly 28,000 are achieved through assisted reproductive technologies (ART) [16].

4.8.Reason for consultation:Requests for karyotype analysis as part of the evaluation of primary infertility accounted for 44.1% of consultations, whereas requests related to azoospermia represented 23.7%. The study by Chennaf [17] reported a markedly higher consultation rate for primary infertility (94%) compared with secondary infertility (6%).

4.9.Reason for consultation and abnormal karyotype. :In the context of primary infertility assessment,61.11% of patients had an abnormal karyotype, compared with 41% who had a normal karyotype. In cases of azoospermia, 16.67% presented with a pathological karyotype, whereas 25% had a normal karyotype;spermatogonia represent the foundation for exploring the potential of germline stem cells [18].

It is hoped that advances in the understanding of stem cell biology may enable the restoration of spermatogenesis in infertile patients in the future. The two most frequent indications for consultation—azoospermia and primary infertility—may be explained by social factors and their impact on couples' lives, as affected couples tend to seek medical care more frequently in order to obtain optimal management [19],In France, the study by Thonneau et al. reported rates of primary and secondary infertility of 67% and 33%, respectively [6].

4.10.Chromosomal abnormalities and semen analysis:In our series, 46 patients presented with azoospermia; among them, 15 patients had an abnormal karyotype, whereas 31 patients had a normal karyotype. Two out of ten patients with oligospermia had a pathological karyotype. A statistically significant association (DSS) was observed between sperm abnormalities (oligospermia or azoospermia) and chromosomal abnormalities detected by karyotyping. However, mitotic studies in infertile patients have shown that approximately 12% of azoospermic patients have karyotypic abnormalities, whereas meiotic studies have reported cytogenetic errors in only 8% of patients [20] ;Conversely, the increasing number of mutant animal models presenting with male infertility has enabled the identification of additional genes, provided that detailed genotype–phenotype correlations are available [21].

4.11.Semen analysis: Among the 118 patients included in our study, 106 presented with abnormal semen parameters. Analysis of the frequency distribution of different sperm abnormalities in infertile patients showed that azoospermia was the most frequent abnormality, with a prevalence of 38.8%. These results differ from those reported in other studies, which may be explained by recruitment bias and sample size differences. Oligoasthenospermia

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accounted for 19% of cases, a frequency close to that reported by Thonneau et al [6] but higher than that observed by Belmokhtar [10] , while isolated oligospermia accounted for 8%, a result similar to that of Belmokhtar. In contrast, the study by Oudina reported oligoasthenospermia in 120 cases (13.8%) and azoospermia in 17.26% of patients [22].

Numerical and structural abnormalities of autosomes and sex chromosomes are involved in male infertility. The prevalence of chromosomal abnormalities ranges from 2% to 8% in the general infertile population and may reach up to 15% in azoospermic patients, representing a 10- to 20-fold increase compared with the general population, as reported by Coutton [23].

4.12.Semen analysis result and mean age: The analysis showed that patients with azoospermia had a mean age of 34 years, which was very close to that of patients with normal semen parameters (mean age: 35 years). According to our results, no significant association was found between patient age and semen analysis results, as no statistically significant difference was observed. This finding is not consistent with data from the literature and may be explained by the limited sample size,furthermore, karyotypic abnormalities observed in infertile patients include aneuploidies affecting the X and Y chromosomes, as well as structural abnormalities such as Y chromosome microdeletions. These alterations lead to mechanical disturbances during meiosis and germ cell arrest, resulting in the loss of genes involved in the azoospermia factor (AZF) regions [21].An African study involving 220 male subjects [9] aged between 23 and 64 years showed that semen analysis was normal in only 35 subjects (15.9%), whereas 84.1% presented abnormal semen parameters. Similar findings were reported in a Tunisian study by Jaballah [7] and the study by belmokhtar [10].Finally, structural studies of spermatozoa aim to improve patient management in the context of assisted reproductive technologies (ART) and to provide personalized genetic counseling to couples, thereby facilitating informed family planning decisions [24],It should be noted that genomic instability may occur at the chromosomal level, such as translocations and Y chromosome microdeletions, as well as at the gene level, including dynamic mutations and polymorphisms[25].All these abnormalities result from unrepaired DNA replication errors [26]. The nucleus plays a crucial role in maintaining genome integrity, and any disruption may ultimately lead to cell death [27].Furthermore, molecular biology techniques have enabled the detection of more subtle abnormalities, such as Y chromosome microdeletions, and have identified genes located on its long arm. Candidate gene analysis often relies on data obtained from animal models, particularly murine models [28].

A Turkish study demonstrated that chromosomal analyses are strongly recommended in the evaluation of fertility disorders; in that study, a numerical abnormality related to gonadal dysgenesis (46,XY/45,XO) was identified in 0.56% of cases [29] a slightly lower rate than that observed in our series (0.85%). Structural chromosomal abnormalities mainly contribute to the occurrence of non-obstructive azoospermia [27].In a study of 600 cases of Y chromosome aneuploidy, 45,XO cells were present in more than half of the cases [30].In patients with an apparently normal 46,XY karyotype, gonosomal mosaicism was detected only after reanalysis of 300 metaphases using FISH, which revealed microdeletions in the AZFc region [15],several studies have shown that gonosomal mosaicism may be a probable cause of assisted reproductive technology (ART) failure [24] ;Given the complexity of male infertility, the entire process cannot be fully modeled in vitro. Experiments in murine models have provided alternative approaches for genetic studies; however, thousands of genes are involved in the regulation of male fertility [31].In addition, molecular biology and cytogenetic techniques have enabled the detection of chromosomal abnormalities up to 100 times smaller, including fluorescence in situ hybridization (FISH), array comparative genomic hybridization (array CGH), and DNA microarray-based chromosomal analysis [32].A review briefly described the biological functions of the Y chromosome and its use in studying the genetic structure of different populations [33]. The analysis of the Y chromosome has also had a positive impact on forensic investigations and paternity testing.

5.Conclusion

The genetics of male infertility, and infertility within the couple, is a field of research that is continuously evolving. Although it opens new perspectives for scientific investigation, the desired outcomes are still far from being fully achieved.

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The genetic causes of male infertility are diverse and may be of chromosomal or gene-related origin. Progress made in understanding the underlying mechanisms has led to the identification of numerous factors involved in reproduction. Consequently, genetic investigation is essential in the evaluation of any case of male infertility.

Through this study, we highlight the crucial role of cytogenetic analysis in determining the origin of male infertility on the one hand, and in precisely identifying the chromosomal or genetic abnormality involved on the other. This approach enables the establishment of appropriate genetic counseling and the implementation of a multidisciplinary, tailored management strategy.

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Table 1: Distribution of karyotype results

Settings	Frequency	Percentage
46,XY	**100**	**84.75%**
47,XXY	**13**	**11.02%**
45 ,XY t(q13 ,q14)	**3**	**2.54%**
45,X0/46,XY	**1**	**0.85%**
46,XY/47,XXY	**1**	**0.85%**
Total	**118**	**100.0%**

Table 2: Distribution according to chromosomal abnormalities based on karyotype results

Settings	frequency	Percentage
sex chromosome	**15**	**83.3%**
Autosomal	**03**	**16.7%**

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Number	**15**	**83.3%**
Structure	**03**	**16.7%**
Homogeneous	**13**	**72.3%**
Mosaic	**05**	**27.7%**
Total	**18**	**100.0%**

Table 3: Distribution according to karyotype and semen analysis results

Résultt Spermogram	karyotype normal	%	karyotype Pathological	%	Total	%
AZOOSPERMIA	**31**	**31.0**	**15**	**83.3**	**46**	**39.0**
NORMAL	**33**	**33.0**	**0**	**0.0**	**33**	**28.0**
OATS	**22**	**22.0**	**1**	**5.6**	**23**	**19.5**
OLIGOSPERMIA	**8**	**8.0**	**2**	**11.1**	**10**	**8.5**
ASTHÉNOSPERMIA	**6**	**6.0**	**0**	**0.0**	**6**	**5.1**
TOTAL	**100**	**100**	**18**	**100**	**118**	

Tableau 4: répartition selon les résultats de speermogramme

	Effectif	Pourcentage
AZOOSPERMIE	**45**	**38.8%**
NORMAL	**33**	**28.5%**
OATS	**22**	**19.0%**
OLIGOSPERMIE	**10**	**8.6%**
ASTHÉNOSPERMIE	**6**	**5.2%**
Total	**116**	**100.0%**

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