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EVALUATION OF CHROMOSOMAL ABNORMALITIES AND Y-CHROMOSOME MICRODELETIONS, WITH PRIMARY MALE INFERTILITY

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ABSTRACT

Background: As in general populace, 15% of all couples trying to conceive experience infertility. Around 50% of instances of impotence are caused by male dysfunction. In this research, patients of primary male failure in some kind of a single facility were examined to assess the rates of gene mutations and Y-chromosome particles having.

Materials and techniques: In 1696 instances of main male infertile, congenital malformations and Karyotype particles having were examined. In every instance, chromosomal defect analyses and En la particles having analyses [used by factor (AZF) areas] were carried out using the combination nucleic acid methodology and conventional morphological techniques, correspondingly.

Result:142 instances (8.4%; 142/1696) had gene mutations, according to the findings. In 46 instances (2.7%; 46/1696), Y chromosomal microdeletions were found. In 20 of 46 instances (43%), Y-chromosome microdeletions as in AZFc locus been discovered.

Conclusion: this research is one of only a few to examine a sizable variety of cases in Turks. It suggests that while in instances of primary male infertility, morphological and Y-chromosome 2 items tests should be carried out before choosing fertility treatment technologies.

Keywords: *Y-chromo-some microdeletion; basic male infertile; genetic syndromes; combinatorial PCR.*

Introduction

Infertility affects 15% of all spouses attempting to become pregnant in the general population. Male infertility is the root cause of around 50% of endometriosis. Many variables, such as autonomic dysfunction, varicocele, coital duct blockage, genital trauma, growth retardation, cardiovascular illnesses, genetic predisposition, etc. might contribute to male infertile. Y-chromosome depending on the activity, genetic mutation shortages, and congenital malformations are the three most common genetic origins of benign prostatic hyperplasia. The structural or statistical genetic anomalies associated with sex cells or autosomes, such as balance homologous recombination, may be present [1]. Rare hereditary is the most prevalent genetic abnormality causing male infertility (47,XXY). A subsequent most common molecular cause is Q u microdeletions. There are many markers responsible for normal testicular function in the used by element (AZF) locus of both the Y chromosome [2]. The AZF sector consists of three different loci with the labels AZFa, AZFb, &AZFc. Microdeletions of the AZF alleles are related to oligospermia and hypogonadism. Molecular morphological examination (secs) and Más social capital and social tests may be used to choose another best fertility treatment techniques (ART), like vitro fertilisation insertion (ICSI) and also in vitro fertilisation (IVF) techniques. This research aimed to assess the kinds and frequencies including both congenital malformations and Y-chromosome particles having

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affecting primary sexual dysfunction in order to offer precise and reliable genetic counselling prior to initiating ART.

Material and methods

Patients

The Genetic Diagnostic Center received 1696 referrals for patients of primary male infertility for this study. Records from every instance were reviewed. No case involved obstructive azoospermia, and all cases involved primary infertility. The investigation was approved as a retrospective study, however not all individuals provided informed consent. The patient records were used to gather the data.

Cytogenetic analysis

All cases' karyotypes were examined using the traditional approach in cultured peripheral blood cells. After trypsin-G banding procedures were used to stain at least 20 metaphase fields, they were examined. According to the International System for Chromosome Nomenclature Guidelines, the karyotype results were written.

Y-chromosome microdeletion analysis

After the gathering of chromosomal Dna evidence, the ChromoQuant AZF Qpcr kit (CyberGeneAf, Solna, Germany) was utilized in conformity with both the kit's recommendations to perform multivariate genotyping (PCR) screening for AZF aberrations. Series locations are amplified through PCR there in signaling pathways (SRY, ZFX/ZFY), AZFa (sY84, sY86), Suitors (sY127, sY134), Housed (sY254, sY255, sY160), and ‘ box (sY254, sY255). The outcomes of the Cpr were examined using an ABI Dual 3700 Pcr analysis (Applied Biosystems, Foster City, CA, USA).Utilizing the GeneMapper Software, data were examined.

Results

Of the 1696 cases of primary male infertility, chromosomal abnormalities were found in 140 cases (8.3%). (Table 1). Sex chromosomes (76.4%; 107/140) were found to have the highest chromosomal abnormalities. Autosomal inversions (4.3%; 6/140) and autosomal transloca-tions (19.3%; 27/140) were the other two. In 75 out of 140 cases, the most prevalent chromosomal anomaly was the Klinefelter syndrome (47,XXY). In 11 cases, the second one was identified as 47,XYY.

Table 1: Chromosomal abnormalities observed in study with primary male infertility

Karyotype	Number
Normal (46,XY)	1556 (91.7%)
Abnormal	140 (8.3%)

In 49 of 1696 examples (2.6%), También microdeletions also found. The most severely impacted location was determined to be the AZFc area (44.4%), trailed by that of the AZFb+c (31.1%), AZFa+b+c (11.1%), AZFa (8.9%), among AZFb (4.5%) locations. 11 of 45 patients (24.4%) exhibiting Y-chromosome germline mutations had altered markers (Table 2).

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Table 2:45 instances of Y chromosomal germline mutations showed 47 chromosomes.

Y chromosome microdeletion	Number	Karyotype
AZFc	20	46XY
AZFb+c	8	46XY
AZFb+c	2	46,X,del(Y)(q11.2)[80]/45,X[20]
AZFb+c	1	46,X,del(Y)(q11.2)
AZFb+c	1	46,X,derY
AZFb+c	1	46,XY[52]/45,X[48]
AZFb+c	1	45,X[94]/46,XY[6]
AZFa+b+c (SRY+)	4	46,XX
AZFa+b+c (SRY-)	1	46,XX
AZFa	4	46,XY

DISCUSSION

Male infertility is correlated with chromosomal disorders. Among infertile men, cytogenetic abnormalities are thought to occur between 2.1% and 28.4% of the time. In 140 cases (8.3%) in our series, aberrant karyotypes were discovered. This topic has been covered in a number of reports from India [3]. These studies' respective incidence rates were 11.2%, 1.6%, and 4.8%. The most frequent factor in infertility associated with chromosomes is abnormalities in the sex chromosome[4]. In this study, 107 of 140 cases (76.4%) had aberrant karyotypes found in the sex chromosomes. The most frequent hereditary reason for male infertility is Klinefelter syndrome. It is linked to severe spermatogenesis failure. In our study, 75 of 140 (53.5%) cases had the 47,XXY karyotype, which was the most common chromosome abnormality. It has been demonstrated that testicular sperm extraction may be used to recover sperm in patients with Klinefelter syndrome (TESE). Preimplantation genetic diagnosis and ICSI may be appropriate treatments in certain circumstances (PGD). In this study, 33 of 140 (23.6%) instances had balanced translocations and inversions, which were indicative of autosomal chromosomal abnormalities[5].While spermatozoa failure is commonly seen in balanced paternal rearrangement and reversal owners, the phenotypic of these individuals is typically normal. This because genetic recombination may affect the configuration of essential germ cell enzymes. Similar to inversions, recombination that may take place during gamete creation in inversions may result in genetically defective gametes, which in turn may result in imbalanced embryos. This might lead to repeated IVF failures or repeat miscarriages[6].Y-chromosomal depending on the activity (AZF regions) are the second most widespread heritable component of benign prostatic hyperplasia in humans, behind Klinefelter syndromes. [5]. The Y-chromosome sometime for is linked to extreme infertile men and azoospermia in benign prostatic hyperplasiascreening is an essential test to select optimal ART and to give appropriate genetic counselling [7]. As a result, it ought to be done frequently. In our investigation, 45/1696 cases (or 2.6% of the total) of Y-chromosome

microdeletions were found. Different incidence rates were reported by several investigations. However, the majority of them only included a few cases. In other study 2 of 60 cases (3.3%) of Y-chromosome microdeletion was present [4]. In a different investigation, Another study discovered that the prevalence was 1.3% (1 in 80 instances) [3] . In addition, 7.7% of the 187 cases in which found microdeletions in AZF areas [5]. The frequency of Y chromosome microdeletion was found to be 6.3% (4/63), according to these scientists. Similar studyinvestigation with a sizable number of patients. 1616 infertile males underwent the Y-chromosome microdeletion test. In 54/1616 cases, or 3.3% of the cases, it was found [8]. Similar to that, our study was one of the few that included a significant number of cases. Their outcome (2.6%; 45/1696) was comparable to ours. While the majority of sex chromosome abnormalities (with the exception of Turner syndrome) do not manifest clinically until adolescence, the majority of At birth, congenital homozygous errors may be identified. It is believed that the unbalanced embryos either fail very early as unidentified pregnancy (pruritus) or fail later in the breeding cycle. Families planning to get conceived are encouraged to undergo foetal either a (amniotic fluid biopsy or noninvasive prenatal) or PGD in situations with balanced evolutionary programming.

CONCLUSION

In conclusion, in order to properly give genetic counseling and choose an appropriate course of treatment for guys with primary infertility, The Y chromosomal relations test and flow cytometry should be performed. Throughout this respect, our research is one of only a few that covered a substantial quantity of cases of male dysfunction brought on by aneuploidy.

REFERENCES

1. Vogt PH. Molecular genetics of human male infertility: from genes to new therapeutic perspectives. Curr Pharm Des 2004;10:471- 500. [CrossRef]
2. Plaseska-Karanfilska D, Noveski P, Plaseski T, Maleva I, Madjunkova S, Moneva Z. Genetic causes of male infertility. Balkan J Med Genet 2012;15(Suppl):31-4. [CrossRef]
3. Balkan M, Tekes S, Gedik A. Cytogenetic and Y chromosome microdeletion screening studies in infertile males with Oligozoospermia and Azoospermia in Southeast Turkey. J Assist Reprod Genet 2008;25:559-65. [CrossRef]
4. Sargin CF, Berker-Karaüzüm S, Manguoğlu E, Erdoğru T, Karaveli S, Gülkesen KH, et al. AZF microdeletions on the Y chromosome of infertile men from Turkey. Ann Genet 2004;47:61-8. [CrossRef]
5. Akin H, Onay H, Turker E, Ozkinay F. Primary male infertility in Izmir/Turkey: a cytogenetic and molecular study of 187 infertile Turkish patients. J Assist Reprod Genet 2011;28:419-23. [CrossRef]
6. Vernaeve V, Staessen C, Verheyen G, Van Steirteghem A, Devroey P, Tournaye H. Can biological or clinical parameters predict testicular sperm recovery in 47,XXY Klinefelter's syndrome patients? Hum Reprod 2004;19:1135-9. [CrossRef]
7. Zhang H, Wang R, Li L, Jiang Y, Zhang H, Liu R. Clinical feature of infertile men carrying balanced translocations involving chromosomeCase series and a review of the literature. Medicine (Baltimore) 2018;97:e0452. [CrossRef]
8. Yapan C, Beyazyurek C, Ekmekci C, Kahraman S. The Largest Paracentric Inversion, the Highest Rate of Recombinant Spermatozoa. Case Report: 46,XY, inv(2)(q21.2q37.3) and Literature Review. Balkan J Med Genet 2014;17:55-62. [CrossRef]