

Psychological characteristics of and counseling for carriers of structural chromosome abnormalities

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ABSTRACT. Infertility as a psychological problem has gained increasing attention. Male partners among infertile couples have elevated levels of psychological distress, which could affect semen quality, result in hormonal abnormalities, and increase the occurrence of early miscarriage. Infertile women are more vulnerable to psychological distress and require psychological support. Subfertile women who conceive after assisted reproduction have higher stress, anxiety, and depression levels. Psychological interventions have been shown to have beneficial effects on infertility patients. However, psychosocial characteristics of carriers of structural chromosome abnormalities have not been studied. We report the characteristics of carriers of structural chromosome abnormalities and their influence on psychological counseling. Seventy-five patients were carriers of reciprocal translocations, 25 carried Robertsonian translocations, 17

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carried inversions, 10 carried deletions, and 3 carried isochromosomes. The main clinical characteristics were recurrent spontaneous abortion, oligospermatism, azoospermatism, primary amenorrhea, and fetal death. Self-rating anxiety scale (SAS) and self-rating depression scale (SDS) scores of women with structural chromosome abnormality were significantly higher than those scores of women with normal karyotype. SAS and SDS scores of men with structural chromosome abnormality were significantly higher than those of men with normal karyotype. SAS and SDS scores of women with structural chromosome abnormality were significantly higher than their scores of men with normal karyotype. SAS and SDS scores of women with structural chromosome abnormality were significantly higher than their scores of men with structural chromosome abnormality. Women carriers with structural chromosome abnormality were more vulnerable to psychological distress. Psychosocial counseling for carriers of structural chromosome abnormalities should focus on self-confidence and treatment with assisted reproductive technology.

Key words: Infertility; Structural chromosome abnormality; Anxiety and depression; Psychological counseling

INTRODUCTION

Infertility is a multidisciplinary problem, and infertile patients require the best diagnostic, therapeutic, and psychosocial services (Li et al., 2014). Recently, infertility as a psychological problem (Podolska and Bidzan, 2011) has received an increasing amount attention (Bak et al., 2012). The frustration of infertile couples leads to not only anxiety and depression but also negative effects on relationships (Bak et al., 2012). Depression was reported by 48% of women and 23.8% of men (Noorbala et al., 2008). Infertile couples' rates of significant anxiety symptoms (24.2%) and depressive symptoms (20.0%) were higher than the corresponding rates of 13.7% and 9.5% for controls (Fatoye et al., 2009). Male partners among infertile couples experience elevated levels of psychological distress (Dyer et al., 2009). Infertile men had lower self-esteem, higher anxiety, and showed more somatic symptoms than fertile men (Kedem et al., 1990). Gana and Jakubowska (2014) reported that their findings confirmed the predictive effects of infertility-related stress on both emotional and marital distress. Psychological interventions have been shown to have beneficial effects for patients with infertility (Cousineau and Domar, 2007).

Psychological distress could affect semen quality, resulting in hormonal abnormalities, and increase the occurrence of early miscarriage. Gollenberg et al. (2010) reported that stressful life events may be associated with decreased semen quality in fertile men. Serum total testosterone correlated negatively with hospital anxiety and depression score (HADS), but luteinizing hormone (LH) and follicle-stimulating hormone (FSH) correlated positively. Sperm count, motility, and morphologically normal spermatozoa were lower in persons with abnormal HADS. Psychological stress primarily lowers serum total testosterone levels, with secondary increases in serum LH and FSH levels altering seminal quality (Bhongade et al., 2015). Depression in men is possibly related to decreased sperm concentration, and poor coping with stress is associated with increased occurrence of early miscarriage (Zorn et al., 2008).

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Psychosocial characteristics of couples who conceived after assisted reproduction have gained more attention. After first-trimester miscarriage, subfertile women who conceived after assisted reproduction had higher stress, anxiety, and depression levels, and they experienced more traumatic effects from the event than did women who conceived naturally (Cheung et al., 2013). *In vitro* fertilization (IVF) failure predicts subsequent psychological distress, but pre-IVF psychological distress does not predict IVF failure. These findings suggest that instead of focusing efforts on psychological interventions specifically aimed at improving the chance of pregnancy, attention should be given to help patients prepare for and cope with treatment and treatment failure (Pasch et al., 2012).

However, psychosocial characteristics of carriers of structural chromosome abnormalities have not been studied. The effects of structural chromosome abnormalities depend on whether they are present in balanced or unbalanced forms. The carriers of balanced forms may show variable sperm counts, ranging from normal counts to low counts, or even a total absence of sperm in the ejaculate (Venkateshwari et al., 2011). Hence, psychological characteristics and counseling of these patients may be more complex. In this study, psychological characteristics of carriers of structural chromosome abnormalities in Jilin Province, China, and their implications in psychological counseling are reported.

MATERIAL AND METHODS

Patients

Couples who sought prepregnancy counseling in the Andrology Outpatient's Clinic of the First Hospital of Jilin University from January 2012 to June 2015 were consecutively recruited for this study. One-hundred thirty couples were recruited, including 72 men and 58 women with structural chromosome abnormalities. One partner was the carrier of the structural chromosome abnormality among all couples. Couples with normal karyotypes were recruited as a control group, which included 102 men and 82 women.

Questionnaire

An elaborate questionnaire was provided to each subject to collect their information, including age, marital history, educational status, monthly income, smoking history and frequency, alcohol consumption, occupational status, and medical history.

Evaluation of anxiety and depression

Anxiety and depression were evaluated by the self-rating anxiety scale (SAS) and self-rating depression scale (SDS), respectively. Two indices were included by using the Chinese versions of SAS and SDS, which Gao et al. (2013) reported. According to Gao et al. (2013), those with SAS \geq 50 and SDS \geq 53 were diagnosed as having anxiety and depression, respectively.

Cytogenetic analysis

Peripheral blood from all subjects was collected in sterile tubes containing 30 U/

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mL heparin. G-banding was performed using cultured peripheral blood lymphocytes, and the karyotypes of metaphases were analyzed for each subject (Zhang et al., 2015).

Statistical analysis

All data were analyzed using SPSS v.17.0 for Windows (SPSS, Inc, Chicago, IL, USA). Parametric variables were compared by independent sample *t*-tests. All results were reported as means \pm SD. P < 0.05 was considered to be statistically significant.

RESULTS

The types of structural chromosome abnormalities and clinical characteristics are shown in Table 1. Of the 72 men, 58.3% (42/72) carried reciprocal translocations, 22.2% (16/72) carried Robertsonian translocations, 11.1% (8/72) carried inversions, 6.9% (5/72) carried deletions, and 1.4% (1/72) carried isochromosomes. Clinical characteristics included 43.1% (31/72) with recurrent spontaneous abortions, 23.6% (17/72) with oligospermatism, 19.4% (14/72) with azoospermatism, and 13.9% (10/72) with fetal deaths. Of the 58 women, 56.9% (33/58) carried reciprocal translocations, 15.5% (9/58) carried Robertsonian translocations, 15.5% (9/58) carried inversions, 8.6% (5/58) carried deletions, and 3.4% (2/58) carried isochromosomes. Clinical characteristics include 58.6\% (34/58) with recurrent spontaneous abortions, 31.0% (18/58) with fetal deaths, and 10.3% (6/58) with primary amenorrhea.

Table 1. Type distribution of structural chromosome abnormalities and clinical characteristics.							
Gender (N)	Structural abnormality		Clinical characteristics (cases)				
	Туре	Cases					
Male (72)	Reciprocal translocation	42	Recurrent spontaneous abortion (31), oligospermatism (17), azoospermatism				
	Robertsonian translocation	16	(14), fetal death (10)				
	Inversion	8					
	Deletion	5					
	Isochromosome	1					
Female (58)	Reciprocal translocation	33	Recurrent spontaneous abortion (34), fetal death (18), primary amenorrhea (6)				
	Robertsonian translocation	9					
	Inversion	9					
	Deletion	5					
	Isochromosome	2					

Table 2 shows that SAS and SDS scores of women with structural chromosome abnormalities were significantly higher than the scores of women with normal karyotypes (P < 0.001). Table 3 shows that SAS and SDS scores of women with structural chromosome abnormalities were significantly higher than the scores of men with structural chromosome abnormalities (P < 0.05 and P < 0.01 respectively). Table 4 shows that SAS and SDS scores of men with structural chromosome abnormalities were significantly higher than the scores of men with structural chromosome abnormalities (P < 0.05 and P < 0.01 respectively). Table 4 shows that SAS and SDS scores of men with structural chromosome abnormalities were significantly higher than scores of men with normal karyotypes (P < 0.001).

 Table 2. Self-rating anxiety scale (SAS) and self-rating depression scale (SDS) scores of women with structural chromosome abnormalities.

Characteristics	Women with normal karyotypes (82)	Women with structural chromosome abnormalities (58)	Р
SAS scores	47.80 ± 6.19	54.41 ± 8.52	0.000*
SDS scores	50.41 ± 6.72	58.34 ± 7.70	0.000*

*P < 0.001 compared to women with normal karyotypes.

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Table 3. Self-rating anxiety scale (SAS) and self-rating depression scale (SDS) scores of men and women with structural chromosome abnormalities.

Characteristics	Men with structural chromosome abnormalities (72)	Women with structural chromosome abnormalities (58)	Р
SAS scores	51.42 ± 5.84	54.41 ± 8.52	0.025*
SDS scores	54.43 ± 5.27	58.34 ± 7.70	0.002**

*P < 0.05 and **P < 0.01 compared to women with structural chromosome abnormalities.

Table 4. Self-rating anxiety scale (SAS) and self-rating depression scale (SDS) of men with structural chromosome abnormality.

Characteristics	Men with normal karyotypes (102)	Men with structural chromosome abnormalities (72)	Р
SAS scores	46.70 ± 6.72	51.42 ± 5.84	0.000*
SDS scores	48.76 ± 7.29	54.43 ± 5.27	0.000*

*P < 0.001 compared to men with normal karyotypes.

DISCUSSION

The inability to conceive children is a stressful situation experienced by individuals and couples all around the world (Cousineau and Domar, 2007). Infertile couples express the need for psychosocial support, but they often feel that support is not available (Read et al., 2014). Timely support and psychological intervention would be beneficial in the management of infertile couples (Cheung et al., 2013).

Physicians and clinicians should acknowledge the immense psychosocial effects of the diagnosis of infertility in men on both men and their fertile women partners (Bak et al., 2012). Understanding the psychological mechanisms observed in patients treated for infertility might help in their diagnoses and might help them face this new and extremely difficult situation (Podolska and Bidzan, 2011). In this study, we examined the psychological characteristics of carriers of structural chromosome abnormalities. Seventy-five patients were carriers of reciprocal translocations, 25 carried Robertsonian translocations, 17 carried inversions, 10 carried deletions, and 3 carried isochromosomes. For male carriers, the main clinical characteristics were recurrent spontaneous abortion, oligospermatism, azoospermatism, and fetal death. For female carriers, the main clinical characteristics were recurrent spontaneous abortion, fetal death, and primary amenorrhea.

Infertile couples seeking psychological help are characterized by high levels of psychological distress. Infertile women are more vulnerable to psychological distress and require psychological support (Wischmann et al., 2009; Klemetti et al., 2010; Omoaregba et al., 2011). Infertile women experience various problems, including being childless, negative self-concept, and psychological symptoms (Karaca and Unsal, 2015). For Malaysian infertile couples, significantly higher frequencies of depression, anxiety, and stress-related difficulties were reported by wives than by husbands (Musa et al., 2014). Severe depressive symptoms were reported by 11.6% of women and by 4.3% of men, and they were significantly associated with increased infertility-related distress at individual and partner levels (Peterson et al., 2014). In this study, SAS and SDS scores of women with structural chromosome abnormalities were significantly higher than the scores of women with normal karyotypes. SAS and SDS scores of men with structural chromosome abnormalities. Female carriers of structural chromosome abnormalities are more vulnerable to psychological distress and require psychological support.

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Research concerning the psychosocial aspects of infertility and infertility treatment more often focuses on women than on men (Fisher and Hammarberg, 2012). Infertile men who are socially isolated, have an avoidant coping style, and appraise stressful events as overwhelming are more vulnerable to severe anxiety than men without these characteristics. Men prefer oral to written treatment information and prefer to receive emotional support from infertility clinicians rather than from mental health professionals, self-help support groups, or friends (Fisher and Hammarberg, 2012). In this study, SAS and SDS scores of men with structural chromosome abnormalities were significantly higher than scores of men with normal karyotypes.

Psychosocial counseling for carriers of structural chromosome abnormalities should focus on increased self-confidence and timely treatment with assisted reproductive technology. High infertility-related stress levels before beginning fertility treatment can negatively affect the stability of marital relationships and lead to repartnering (Martins et al., 2014). Psychosocial interventions for couples undergoing treatment for infertility could be efficacious for reducing psychological distress and for improving clinical pregnancy rates (Frederiksen et al., 2015). The cumulative live birth rate is 64.3% for couples in which either partner has a chromosomal anomaly and who have experienced repeated miscarriage (Flynn et al., 2014). The success rates of natural pregnancies range from 30% to 70% for men carrying translocations (Ozawa et al., 2008). Based on the effectiveness of psychiatric interventions for increasing pregnancy rates, it is crucial to mandate psychiatric counseling at all fertility centers to diagnose and treat infertile patients with psychiatric disorders (Ramezanzadeh et al., 2011). IVF accompanied by preimplantation genetic diagnosis increases pregnancy rates for carriers of balanced chromosomal translocations (Vozdova et al., 2011).

CONCLUSIONS

In summary, the reciprocal translocations were most commonly structural chromosome abnormalities in Jilin Province, China. The main clinical characteristics were recurrent spontaneous abortion, fetal death, oligospermatism, azoospermatism, and primary amenorrhea. Psychological distress should be taken seriously for carriers of structural chromosome abnormalities. Psychosocial counseling for carriers of structural chromosome abnormalities should focus on increased self-confidence and timely treatment with assisted reproductive technology.

Conflicts of interest

The authors declare no conflict of interest.

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