Genetic screening and evaluation for chromosomal abnormalities of infertile males in Jilin Province, China


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ABSTRACT. Chromosomal abnormality is the most common genetic cause of male infertility, particularly in cases of azoospermia, oligozoospermia, and recurrent spontaneous abortion. Chromosomal rearrangement may interrupt an important gene or exert position effects. The functionality of genes at specific breakpoints, perhaps with a specific role in spermatogenesis, may be altered by such rearrangements. Structural chromosome abnormalities are furthermore known to increase the risk of pregnancy loss. In this study, we aimed to assess chromosomal defects in infertile men from Jilin Province, China, by genetic screening and to evaluate the relationship between structural chromosome abnormalities and male infertility. The prevalence of chromosomal abnormalities among the study participants (receiving genetic counseling in Jilin Province, China) was 10.55%. The most common chromosome abnormality was Klinefelter syndrome, and the study findings suggested that azoospermia and oligospermia may result from structural chromosomal abnormalities. Chromosome 1 was shown to be most commonly involved in male infertility and balanced chromosomal translocation was identified as one of the causes of recurrent spontaneous abortion. Chromosomes 4, 7, and 10 were the most commonly involved
chromosomes in male partners of women experiencing repeated abortion.

**Key words:** Chromosomal abnormality; Male factor infertility; Azoospermia; Oligozoospermia; Recurrent spontaneous abortion