



Male carriers of balanced reciprocal translocations in Northeast China: sperm count, reproductive performance, and genetic counseling

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ABSTRACT. Balanced chromosomal translocations in men can cause failure of spermatogenesis owing to meiotic impairment. Male carriers may exhibit normozoospermia, although clinical manifestations can include oligozoospermia or azoospermia, oligozoospermia or normozoospermia. Here, we reported the characteristics of balanced reciprocal translocations in men from northeastern China, and explored the relationship between sperm count and reproductive performance, to enable informed genetic counseling. The frequency of balanced reciprocal translocations was found to be 1.62%. Semen analysis showed that 5.9% of male carriers had azoospermia, 43.1% had oligozoospermia, and 51.0% had normozoospermia. Of the 25 men with a balanced reciprocal translocation and azoospermia or oligozoospermia, chromosome 1 was the most commonly often involved in the translocation. However, in the 26 normozoospermic men with a balanced reciprocal translocation and normozoospermia, chromosome 3 was most commonly implicated. Fifty percent of men with a balanced reciprocal translocation conceived a pregnancy that went to term. Our data suggest that of all chromosomes,

chromosomes 1 and 3 are the most commonly involved chromosomes in balanced reciprocal such translocations in northeastern Chinese men. Karyotype analysis should be performed for men with azoospermia, oligozoospermia, and those in couples having suffered recurrent miscarriages. Natural conception should be discussed during genetic counseling for male carriers of balanced chromosomal translocations with normozoospermia.

Key words: Sperm count; Balanced translocation; Genetic counseling; Reproductive performance