Genetics and Molecular Research

Translocation breakpoints of chromosome 1 in male carriers: clinical features and implications for genetic counseling

R.X. Wang, H.G. Zhang, Y. Pan, S. Chen, F.G. Yue, D.L. Zhu and R.Z. Liu

Center for Reproductive Medicine, Center for Prenatal Diagnosis,
First Hospital, Jilin University, Changchun, Jilin, China

Corresponding author: R.Z. Liu
E-mail: lrz410@126.com

Genet. Mol. Res. 15 (4): gmr.15048707
Received April 12, 2016
Accepted May 17, 2016
Published October 5, 2016
DOI http://dx.doi.org/10.4238/gmr.15048707

ABSTRACT. Reciprocal translocation is closely associated with male infertility and recurrent miscarriages. Balanced reciprocal translocations associated with reproductive failures are predominantly observed on chromosome 1. Additionally, infertile male patients present a number of breakpoints throughout chromosome 1. A translocation breakpoint might interrupt the structure of an important gene, leading to male infertility. Here, we report the breakpoints on chromosome 1 translocation and the clinical features presented in carriers, to enable informed genetic counseling of these patients. Balanced reciprocal translocations were found in 1.57% of the tested patients. Among 82 patients, 23 patients (28.05%) were carriers of the chromosome 1 translocation: 12 presented pre-gestational infertility with clinical manifestations of azoospermia or oligozoospermia, while 11 patients presented gestational infertility (able to conceive but with a tendency to miscarry or give birth to a stillborn). The breakpoint at 1p22 was

predominantly observed in these patients; additionally, breakpoints at 1p31.2, 1p10, and 1q25 were associated with gestational infertility. Breakpoints at 1p13, 1q12, and 1q21 were associated with pre-gestational infertility. These results suggested that breakpoints at 1p32, 1p13, and 1q21 were predominantly associated with pre-gestational infertility, while that at 1q25 was associated with gestational infertility. Chromosome 1 translocation carriers with infertility presenting as azoospermia or oligospermia should be counseled on chromosomal breakpoints and the different molecular technologies available to facilitate reproduction.

Key words: Male infertility; Chromosome 1; Balanced translocation; Breakpoint; Genetic counseling