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Association between chromosomal translocation imbalances and aneuploidies in PGT-SR in reciprocal translocation carriers

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【Introduction】

Reciprocal translocation carriers are known to be at high risk of conceiving genetically unbalanced embryos, which may result in miscarriage or an affected neonate. Preimplantation genetic testing for chromosomal structural rearrangements (PGT-SR), a method for embryonic chromosomal analysis, can detect not only translocation chromosome imbalance but also aneuploidy via comprehensive chromosomal analysis with whole-genome amplification (WGA), thereby potentially decreasing miscarriage rate. In this study, we aimed to investigate the relationship between chromosomal abnormalities derived from translocation chromosomes and aneuploidies and the frequency of chromosomal abnormalities among stimulation cycles in reciprocal translocation carriers receiving multiple PGT-SR cycles.

【Material and methods】

Between 2013 and 2017, we assessed 20 cycles of 8 couples (reciprocal translocation carriers in 5 females and 3 males; mean women's age, 35.1 years) who received 2 or more PGT-SR cycles at our clinic.

Ovarian stimulation was primarily controlled, and when chromosomally normal embryos could not be obtained, a different protocol for ovarian stimulation was adapted. All oocytes

were fertilized via intracytoplasmic sperm injection (ICSI). All embryos were cultured to the blastocyst stage. Approximately 5-10 trophectoderm (TE) cells were subjected to a biopsy using a laser system. Chromosomal analysis was performed using the microarray-based comparative genomic hybridization method or next-generation sequencing with whole-genome amplification.

【Results】

In all 20 cycles, the number of oocytes retrieved was 432 and the number of blastocysts subjected to biopsy was 90. Among 88 analyzable blastocysts, 19 had normal chromosomes (21.6%) and 69 had abnormal chromosomes (78.4%). Fifty-six embryos (81.2%) contained chromosomal imbalances derived from reciprocal translocation chromosomes and 37 embryos (53.6%) had aneuploidy, 24 embryos (34.8%) had both types of abnormalities. The incidence of chromosomal translocation-derived chromosomal imbalance was 60.0% (12/20) for male carriers and 64.7% (44/68) for female carriers, with no significant difference.

The incidence of chromosomally normal embryos per cycle was 0–67%. Normal embryos were not obtained in 9 cycles (45%); however, they were obtained at different stimulation cycles. In 9 PGT-SR cycles with three or more embryos of female carriers subjected to biopsy, there was no correlation between the incidence of the abnormality due to chromosome translocation and aneuploidy.

【Conclusion】

There is no association between the incidence of translocation chromosome imbalance and aneuploidy. However, approximately 80% of embryos with chromosomal translocation-derived chromosomal imbalance include aneuploidy. The incidence of chromosomal aberrations in embryos has a large difference among the PGT-SR cycles even in the same carrier.

In translocation carriers without normal embryos, performing different ovarian stimulation methods enhances the possibility of obtaining normal embryos.

Key words: Preimplantation genetic testing-structural rearrangement, chromosomal translocation, aneuploidy, ovarian stimulation