Day 5 embryos show reduced an euploidy rate compared to day 3 embryos in preimplantation genetic diagnosis for reciprocal translocation carriers

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(Introduction)For preimplantation genetic diagnosis (PGD) in Japan, chromosome analysis to identify chromosomal abnormalities is permitted only for translocated chromosomes without aneuploidy screening of non-translocated chromosomes. On the other hand, comparative genomic hybridization (CGH) provides accurate and comprehensive information about all chromosomes of embryos. Therefore, we investigated the incidence of chromosomal abnormalities originating from translocated and non-translocated chromosomes in day 3 and day 5 embryos undergoing PGD by using CGH.

(Material and Methods) This study was approved by the Japan Society of Obstetrics and Gynecology. In total, 31 PGD cycles from 17 carrier couples with reciprocal translocation, including 20 cycles of day-3 embryo biopsies and 15 cycles of day-5 embryo biopsies were studied. Average maternal age is 37.5 years for day-3 embryo biopsies and 36.2 years for day-5 embryo biopsies. Chromosome analysis was performed using CGH.

(Results)The incidence of chromosome abnormalities in day 3 and day 5 embryos was 86.5% (77/89) and 80.9% (38/47); 59.5% (53/89) and 63.9% (30/47), respectively, of abnormalities originated in translocated chromosomes. However, the incidence of abnormalities in non-translocated chromosomes in day 5 embryos (44.7% [21/47]) was significantly lower than that in day 3 embryos (76.4% [68/89]). The aneuploidy rate, except for translocated chromosomes, was 27.0% (24/89) and 17.0% (8/47) in day 3 and day 5 embryos, respectively.

(Conclusion)The incidence of abnormalities in non-translocated chromosomes shows a significant decrease from day 3 to day 5 embryos. These findings suggest that day 5 embryos are suitable for the exclusion of aneuploidy during PGD for reciprocal translocation carriers.