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Comparison of the validity of next generation sequencing and array comparative genomic hybridization in a chromosome translocation patient

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[Objective] The use of next generation sequencing (NGS) has been rapidly increasing in preimplantation genetic diagnosis (PGD) including preimplantation genetic screening due to its accuracy. Here we compared NGS with array comparative genomic hybridization (CGH) in a reciprocal translocation woman using identical whole genome amplification (WGA) products.
[Methods] The patient with the 46,XX,t(2;4)(q22;q23) karyotype had two miscarriages. After obtaining approval from the Japan Society of Obstetrics and Gynecology, we performed PGD using CGH in the trophectoderm biopsy samples of 10 blastocysts. After all the embryos were diagnosed as abnormal, the reserved identical WGA products were re-analyzed using NGS.
[Results] The outcome of the chromosome analysis in all the embryos by both methods was abnormal; the concordance rate was 100%. In the 240 chromosomes from the 10 embryos, 16 (6.7%) were discordant in the two methods. In one of the 10 embryos, re-analysis by CGH after re-biopsy also produced uncertain results. However, NGS provided us a decisive analysis of the embryos. Furthermore, 4 of the 240 chromosomes that had different breakpoints from the patient's karyotype of G-banding in CGH showed the same breakpoint in NGS.
[Conclusion] NGS showed more accuracy in analyzing the chromosomes of embryos than CGH when the same WGA products were used. These results suggest that NGS is suitable for PGD in chromosome translocation cases.