Comparison of FISH and array CGH in the practice of PGD

<Objective>

Preimplantation genetic diagnosis (PGD) is a technology performed to determine whether specific mutation or an unbalanced chromosome has been transmitted to the embryo by analyzing biopsied cells from either cleaved embryos or blastocysts. PGD has been applied on recurrent miscarriage patients due to chromosomal translocation following the guideline of Japan society of Obstetrics and Gynecology (JSOG) from 2008 at our facility. PGD was initially performed by fluorescence in situ hybridization (FISH). However, we applied comparative genomic hybridization microarrays (aCGH) from 2012 after approval of using this technology by JSOG because of more accuracy. Therefore, the present study was conducted to compare the accuracy of diagnosis, subsequent pregnancy rate and miscarriage rate between FISH and aCGH.

<Method>

Retrospective analyses were performed on 35 PGD cases (66 IVF cycles) due to reciprocal translocation from March, 2008 to April, 2015. Twenty eight cycles of FISH group (204 embryos) and 38 cycles of aCGH group (180 embryos) were compared by the rates of normal embryo, clinical pregnancy and miscarriage. Although biopsy was performed on either cleaved embryo or blastocyst, no comparison was done between the embryo stages.

<Results>

The determination of unbalanced translocation on embryos was diagnosed with significantly higher ($P<0.001$) in FISH group (84.8%:173/204) compared to aCGH group (54.4%:98/180), but no difference on diagnosed normal chromosome between FISH (15.2%:31/204) and aCGH (17.2%:31/180) groups. There were no difference in pregnancy and miscarriage rates after transfer of the embryos diagnosed normal between FISH (33.3%:6/18 and 33.3%:2/6) and aCGH (42.9%:9/21 and 11.1%:1/9) groups.

<Conclusion>

Although FISH determined higher ratio of embryos with chromosomal abnormality than aCGH, subsequent pregnancy and miscarriage rates are similar in both group. Therefore, there must be more misdiagnosis or over diagnosis by FISH compared to aCGH. We can't conclude which is better FISH or aCGH due to small number of samples. FISH will be replaced by aCGH or even next generation sequencing (NGS) soon or later, not only due

to accuracy but also less cost and labor.