

Preimplantation genetic screening

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Aneuploidy is common in human embryos and is a principal cause of implantation failure and miscarriage. Aneuploidy rates increase with maternal age. Preimplantation genetic screening (PGS) is a process by which chromosomal abnormalities in embryos can be diagnosed before transfer. The demand for this screening has increased in recent years because of the rise in maternal age as well as the development of accurate diagnostic technologies.

In PGS by fluorescence in situ hybridization (FISH), randomized controlled trials did not show any benefit of PGS in terms of the accuracy of the technique and chromosomal mosaicism in cleavage-stage embryos. Advanced technologies based on comprehensive chromosomal screening (CCS) methods with whole genome amplification (WGA) can provide accurate and reliable genetic information, where CCS includes array comparative genomic hybridization (CGH), and next-generation sequencing (NGS). Of these technologies, NGS can also detect embryo mosaicism.

Embryo biopsy was carried out at the blastocyst stage rather than at the day-3 embryo stage, since the multicellular blastocyst biopsy reduces misdiagnosis due to mosaicism and improves diagnostic accuracy in WGA.

In contrast with the increased application of PGS worldwide, the Japan Society of Obstetrics and Gynecology does not allow PGS in Japan owing to ethical problems; however, clinical trials on PGS are planned for several IVF facilities in Japan.

Finally, in the context of reproductive medicine, PGS is considered an indispensable technology in obtaining genetic information of selecting viable embryos.