

The role of preimplantation genetic diagnosis in diagnosing embryo aneuploidy

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Purpose of review

Use of preimplantation genetic diagnosis to improve *in-vitro* fertilization outcomes is reviewed.

Recent findings

Many embryos produced *in vitro* contain chromosomal abnormalities and have little potential for forming a viable pregnancy. The most commonly used method for preimplantation genetic diagnosis involves embryo biopsy on day 3 of development, followed by fluorescence in-situ hybridization analysis of 5–12 chromosomes.

However, positive results have been more common with single-cell biopsy and the analysis of nine or more chromosomes, including 15, 16, 21, and 22. Comparative genomic hybridization, array-comparative genomic hybridization, and single-nucleotide polymorphism arrays analyze all chromosomes and, although technically demanding and requiring experience for successful use, improve the selection potential of preimplantation genetic diagnosis and minimize error rates. Recent data suggest that biopsy at the blastocyst stage may allow sampling of representative genetic material without compromising embryo viability. The optimal strategy for aneuploidy screening using preimplantation genetic diagnosis seems to be blastocyst biopsy at 5 days and comprehensive chromosome analysis (comparative genomic hybridization, array-comparative genomic hybridization, single-nucleotide polymorphism array).