

Invited review: Preimplantation genetic diagnosis

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Abstrak

the availabilty of assisted reproductive technologies and advances in molecular biology gave rise to preimplantation genetic diagnosis (PGD). PGD is an early form of prenatal diagnosis and determines the genotype of an embryo before implantation takes place to avoid the implantation of diseased embryos. this requires couples to adhere to a strict family planning and effective contraceptive strategy, and undergo in vitro fertilization (IVF) treatment. during IVF, sampling of the cells can be performed at different developmental stages from polar body biopsy to trophectoderm cells from the blastocysts. it is indicated in couples with a family history of monogenic autosomal or X-linked recessive or dominant disorders and in detecting chromosomal aberrations of the embryos. the genetic diagnosis is performed using appropriate molecular testing which might include polymerase chain reaction, fluorescent in situ hybridization, comparative genomic hybridization or microarrays. PGD is now a well-established procedure and offers an alternative reproductive choice for couples who are at risk of an affected child. in this review, the past, present and future aspect of PGD will be covered.