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Preimplantation Genetic Diagnosis for the Better Management of Couples During Assisted Reproduction

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ABSTRACT Intracytoplasmic Sperm Injection (ICSI) offers the real prospect of genetic parenthood for men with profound oligozoospermia and azoospermia. However, it may result in transgenerational transmission of genetic defect, which substantially increases the recurrence risk of infertility in the offspring of couples treated with ICSI. Recent developments in the field of genetics and assisted reproduction have led to emergence of preimplantation genetic diagnosis (PGD). PGD helps in the negative selection of the aneuploid and abnormal embryos. For PGD, the embryo is biopsied and 1-2 blastomeres are removed from the 6-10 cell embryo. For establishing the diagnosis, the genetic analysis is carried out using FISH (for structural and numerical chromosomal anomalies) or PCR (for single gene disorders). Only the normal embryos are transferred back to the uterus, thus ensuring a normal pregnancy. Spare or arrested embryos obtained following ICSI were used for the study. FISH was performed on 5 embryos using CEP X spectrum green probes. PCR was done for cystic fibrosis common mutation DF508 and for b-thalassaemia IVS1®5 and 619 bp deletion using blood as positive control. Mosaicism was noted in two of the five embryos using FISH. The blastomeres were found to be normal for cystic fibrosis and b-thalassaemia. PGD has an important role in helping patients to avoid the risk of transmission of genetic defect or abnormalities and also helps in avoiding repeated medical termination of pregnancy.

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