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Preimplantation Genetic Diagnosis

A benefit of the state-of-the-art in assistant reproductive treatment requires among others, a definitive preimplantation genetic diagnosis (PGD) as an alternative to prenatal diagnosis, for couples with a family history of a genetic disease. In the past, parents had to take the risk for an inherited genetic disorder, or undergo prenatal diagnostic testing and pregnancy termination in case of a positive result, with all the accompanying psychological and injuring issues.

PGD was first indicated for single-gene disorders of known genetic diseases, sex-linked and chromosomal disorders, or for a human leukocyte antigen (HLA) matching prediction. As the capability of the technique expanded to additional diagnosis, such as diagnosis of adult-onset hereditary diseases like cancer-related mutations, the PGD offers the parents the opportunity to choose for implantation the absolute healthy embryo with diminished lifetime cancer risk and assured better quality of life. On the other hand, affected embryos may be discarded or used for research reasons. However the 'eugenic' aspects of PGD, raise ethical issues and social skepticism.

So far, ten most common diagnosed monogenetic diseases are diagnosed by PGD. However, parents will push in the near future for the diagnosis of cancer predisposition and chromosome translocations that could be transmitted in an unbalanced form to the embryo. This demand will create parental questions which will need answers and well-documented genetic counseling from the doctors, since the penetrance of the autosomal dominant conditions varies and not all affected persons will develop the same severity of disease. On the other hand, some embryos will eventually have a balanced chromosomal arrangement and will be born healthy. Uncertainties or limited knowledge for novel findings would be a significant challenge for doctors and couples as well and counseling sessions prior to starting PGD should be considered and tailored in accordance to the couple's needs.

The use of human leucocyte antigen PGD for

compatibility with an affected sibling, can give hope for cure to children with blood disorders, by employing haematopoietic stem cells transplantation, which may reflect the most optimistic view of PGD procedure.

PGD is a powerful tool that is expected to improve birth rate in couples with genetic defects and can cure diseases using the unique features of the pluripotent stem cells. The new high throughput technologies like whole genome sequence (WGS) and the omics approaches will overcome the limitations of PCR and as techniques in gene editing and gene therapy get more accurate, the PGD will be the gold standard in ART prenatal interventions. Still though, problems like emotional burden and ethical issues will also need to be addressed.

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