Preimplantation Genetic Diagnosis

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What is preimplantation genetic diagnosis?

Preimplantation genetic diagnosis (PGD) is the use of reproductive technologies for genetic analysis of *embryos* prior to their transfer and implantation. In patients with *hereditary genetic disorders*, such as BRCA carriers, this allows for the screening and transfer of embryos unaffected by the genetic disease in question.

How is preimplantation genetic diagnosis different from preimplantation genetic screening?

While PGD involves testing an embryo for a specific genetic disorder, preimplantation genetic screening (PGS) involves testing an embryo for structural chromosomal abnormalities (e.g. trisomies like Down Syndrome) in couples presumed to have normal chromosomes. It is recommended that PGS is done concurrently on all embryos having PGD testing.

Why consider preimplantation genetic diagnosis?

PGD can reduce the risk of having a child with a genetic abnormality carried by one or both parents if the abnormality can be identified. Patients who carry mutations in BRCA1 may consider PGD to prevent the risk of transmission to their offspring who have a significant increased risk for the disease.

What does preimplantation genetic diagnosis involve?

Counselling must first be provided by a genetic counsellor to ensure that the patient understands the risk of having an affected child, the impact of the disease on the child and the benefits and limitations of PGD.

PGD requires *in vitro fertilization (IVF) with intracytoplasmic sperm injection,* even though many patients using this technology may have no known difficulties conceiving a pregnancy. A *biopsy* of the embryo is then taken for sampling of its DNA and genetic testing is performed. The selected unaffected embryo is then put back into the patient's uterus.

What are the risks of preimplantation genetic diagnosis?

There are possible risks involved at all stages of the process including risks associated with IVF (including *ovarian hyperstimulation,* multiple gestation including twins or triplets, bleeding and infection), risks associated with embryo biopsy, risk of misdiagnosis and the possibility that no embryo may be transferred if all embryos are affected.

What is the cost associated with preimplantation genetic diagnosis?

There are costs associated with both PGD and IVF. Although certain medications may be covered by insurance, neither of these technologies are subsidized in Canada, except for Ontario. Contact your local fertility centre for pricing information as this can differ based on site of treatment.

Glossary

***Embryo*:** The product of a fertilized egg

Hereditary genetic disorder: A disease that can be passed down from the parents' genes

In vitro fertilization (IVF): The process of combining egg and sperm, creating embryos and then transferring the embryo to the uterus

Intracytoplasmic Sperm Injection: The process of injecting the sperm into the egg to improve the fertilisation rate

Biopsy: A procedure during which a small sample of tissue is removed for analysis

Ovarian hyperstimulation: Exaggerated response to the use of ovarian stimulation medications

References

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4. Preimplantation Genetic Testing: A Practice Committee Opinion. Fertility and Sterility. November 2008.