PGD

Preimplantation genetic diagnosis

An option for couples at high risk of passing on a serious hereditary condition to their children

Bioteknologinemnda
The Norwegian Biotechnology Advisory Board

What is preimplantation genetic diagnosis (PGD)?

PGD is a method which allows for genetic testing of a fertilized egg before an actual pregnancy is established. The use of assisted reproductive technology (ART) is required. PGD may be a realistic option for couples who have a known risk of passing on a serious hereditary condition to future children. With this method, fertilized eggs are tested to identify those eggs which may result in a pregnancy not at risk for developing the specific condition in question. Since PGD does not test for other health problems or birth defects, PGD cannot guarantee a healthy child.

PGD was introduced in the early 1990's and has resulted in the birth of approximately 4000 children worldwide.

PGD may also be considered by couples interested in having a child not at risk for a specific hereditary condition who also may donate stem cells to an ill sibling (such children have been referred to as "savior siblings"). A fertilized egg not at risk for developing the hereditary condition which also has the matching tissue type to the ill sibling, is selected. Matching tissue types between donor and recipient is important to minimize the chance that the recipient will reject the donated stem cells. This application of PGD is called PGD/HLA testing, where HLA refers to tissue typing.

PGD may be an option for couples who:

- have a known risk for having children with a serious hereditary condition
- already have children with a serious hereditary condition
- have had several spontaneous pregnancy losses due to a chromosome abnormality (one member of the couple carries a chromosome rearrangement)

Alternatives to PGD:

- Diagnostic testing of an established pregnancy (prenatal diagnosis) with the option of pregnancy termination if the fetus is determined to have the specific condition in question using:
  - CVS (chorionic villus sampling) usually performed between pregnancy weeks 10–12 or,
  - amniocentesis usually performed between pregnancy weeks 15–18
- Use of a sperm donor (if the susceptibility for the specific hereditary condition is inherited through the father)
- Adoption

Cells are removed from a fertilized egg for genetic testing. Photo: S. Ziebe. Rigshospitalets Fertilitetsklinik, Denmark

Where can I learn more?

Read more about PGD, PGD/HLA, and prenatal diagnosis: www.bion.no

Read more about prenatal diagnosis (in Norwegian only): www.helsedirektoratet.no

Read about the Preimplantation Genetic Diagnosis Board (PGD-nemnda) (in Norwegian only): www.klagenemnda.no/pgd-nemnda

When is PGD allowed by Norwegian law?

In Norway, the Biotechnology Act regulates access to PGD.

PGD is only offered to couples legally residing in Norway, where one or both carry a genetic susceptibility for a serious hereditary condition and, there is a high likelihood that this couple will have a child who will develop this condition.

The seriousness of a hereditary disease is evaluated on criteria including: reduced life expectancy, pain and suffering caused, and existing treatment options (palliative or those increasing life expectancy).

PGD is not available for couples interested in routine prenatal testing for chromosome abnormalities due to advanced maternal age.

Access to PGD/HLA is even more restrictive than for PGD alone.

The Norwegian Biotechnology Advisory Board (Bioteknologinemnda) has published a 36 page pamphlet describing the PGD process and surrounding ethical issues in greater detail (available in Norwegian only). Important contact information is also included in this detailed pamphlet.

The pamphlet is free of charge and can either be ordered or downloaded at www.bion.no. Medical genetics departments and clinics specializing in assisted reproduction across Norway should also be able to provide this pamphlet.

Photo: iStockphoto

Ethical questions to consider

- When exactly is a condition serious enough to allow for PGD?
- Does PGD contribute to a society where individuals with certain traits or characteristics are selected out?
- Does offering PGD stigmatize those individuals who currently live with conditions that PGD can test for?
- Is it more or less acceptable to select out fertilized eggs with the help of genetic testing prior to pregnancy or, to become pregnant naturally, have genetic testing during pregnancy (with prenatal diagnosis) and terminate a pregnancy if the fetus is shown to have the hereditary condition?
- Is it acceptable to involve other family members in genetic testing if this is required to establish the genetic test necessary for PGD? What about others' rights not to know about genetic status?
- Is it acceptable to use PGD/HLA to select for a child whose stem cells may be used to treat an ill sibling? How might this influence this child's development and childhood? What are the limits for how much this child will be expected to contribute?

The PGD process

Preimplantation genetic diagnosis (PGD) is a complex and time consuming process. This process can take one to two years and may be a burden for some couples. Far from all couples will be successful having a child using this method. The chance to have a child varies between 10–25% per attempt.

Part of the PGD process is available in Norway and part must be done abroad. Couples interested in PGD begin first with a consultation at a medical genetics department. In order for PGD to be a possibility, the specific hereditary condition must be correctly identified and genetic testing available. Couples considering PGD also receive counseling and information on the involved risks, benefits, and drawbacks of this method, the chances to be successful with PGD, and relevant alternatives to PGD (see inside brochure).

PGD requires egg fertilization outside of the body using assisted reproductive technology (ART) even if the couple are able to conceive naturally (are fertile). The couple must be evaluated by specialists to check that they are suitable for such treatment. This evaluation also includes evaluating the couples' suitability for parenting and a consideration for what is best for the child.

Couples interested in PGD must apply formally to the Preimplantation Genetic Diagnosis Board. This board evaluates applications and decides where the actual PGD treatment will take place.

If an application is approved, the PGD process starts with the development of an appropriate genetic test at a facility abroad suited for the specific condition and situation involved. Genetic testing of several family members may be required.

When the genetic test for PGD is available, the woman can begin hormone treatment in order to stimulate egg cells. This hormone stimulation takes place in Norway, usually at a clinic specializing in assisted reproduction.

When the egg cells are mature, the couple travels to the PGD treatment center abroad. These egg cells are removed from the woman's ovaries. Each egg cell is fertilized with a single sperm from her partner in the laboratory. The fertilized eggs grow and divide. When there are 8–10 cells in each fertilized egg, one or two of these cells are gently removed from each egg for genetic testing (see photo inside brochure). The remainder of each egg is allowed to grow and divide while the genetic testing takes place.

After genetic testing, one fertilized egg not susceptible to develop the hereditary condition in question is placed into the woman's uterus (womb) if available. Women who become pregnant through PGD receive routine prenatal care in Norway. The couple is also offered genetic testing through prenatal diagnosis to confirm PGD test results.

The Norwegian state covers the costs of up to three PGD attempts, with the exception of costs for hormone stimulation and clinic co-pays.

The couple must decide how to handle unused fertilized eggs. Fertilized eggs may be frozen for later use, destroyed, or donated for research purposes.

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P. O. box: 522 Sentrum, 0105 Oslo. Tlf: 24 15 60 20
Fax: 24 15 60 29. Email: bion@bion.no. Internet: www.bion.no
Editor-in-chief: Sissel Rogne
Editing by: Abby L. Grant, Marit Kise, Grethe S. Foss and Norunn K. Torheim
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