AMERICAN SOCIETY FOR REPRODUCTIVE MEDICINE
Formerly The American Fertility Society
1209 MONTGOMERY HIGHWAY • BIRMINGHAM, ALABAMA 35216-2809 • TEL (205)978-5000 • FAX (205)978-5005 • E-Mail: asrm@asrm.org

FACT SHEET
PREIMPLANTATION GENETIC DIAGNOSIS

Preimplantation genetic diagnosis (PGD) is a technique that can be used during in vitro fertilization (IVF) procedures to test embryos for genetic disorders prior to their transfer to the uterus. PGD makes it possible for couples or individuals with serious inherited disorders to decrease the risk of having a child who is affected by the same problem. This technique is controversial and raises issues of sex selection and genetic engineering. At present, PGD is only offered in a few centers, usually under the supervision of an institutional ethics review board, but its use may become more widespread in the near future.

WHY IS PGD PERFORMED?

PGD can be considered in couples who have been psychologically traumatized by repeated pregnancy loss due to genetic disorders. It is also appropriate in couples who already have one child with a genetic problem and are at high risk of having another. Below is a partial listing of conditions for which this technique has been successfully applied. Additionally, there are over 200 disorders that could potentially be prevented by gender selection of embryos.

HOW IS PGD PERFORMED?

Embryos are obtained two ways: either by combining an egg and sperm in the laboratory (i.e. IVF), or by flushing out the uterus five to seven days after fertilization. Once the embryo(s) is isolated, a single cell is removed from each embryo under microscopic guidance and analyzed for the presence of genetic disorders. A diagnosis is obtained within a day or so of the test, and only the unaffected embryos are replaced in the female's uterus. Not all disorders can be diagnosed with this technique.

WHEN IS PGD PERFORMED?

PGD was first performed in 1989. It has since been successfully applied to a wide variety of genetic diseases, either single gene disorders or chromosomal abnormalities. In single gene disorders where the gene structure is known, such as cystic fibrosis or Tay-Sachs, the actual genes of the sampled embryo can be examined for the presence of the condition. Other genetic conditions, such as Duchenne muscular dystrophy or hemophilia, affect only males. In these cases, while the exact gene defect may not be known, the DNA of the biopsied cell can be examined to determine the sex of the embryo and only female embryos replaced. Finally, in cases of severe recurrent chromosomal diseases, such as Down syndrome (which has an extra chromosome), the number and character of several chromosomes of the sampled embryo can be determined.

PGD CAN DETECT:

- α-l-antitrypsin deficiency
- Cystic fibrosis
- Fragile X syndrome
- Lesch-Nyhan syndrome
- Charcot-Marie-Tooth disease
- Down syndrome
- Tay-Sachs disease
- Duchenne muscular dystrophy
- Hemophilia A
- Retinitis pigmentosa
- Gender
- Turner syndrome