

## Preimplantation Genetic Diagnosis <sup>[1]</sup>

By: Antonios, Nathalie Keywords: Fertilization <sup>[2]</sup> Genetic testing <sup>[3]</sup> Bioethics <sup>[4]</sup>

Preimplantation genetic diagnosis (PGD) involves testing for specific genetic conditions prior to the [implantation](#) <sup>[5]</sup> of an embryo in the uterine wall. This form of genetic screening has been made possible by the growth of *in-vitro* [fertilization](#) <sup>[6]</sup> (IVF) technology, which allows for the early stages of development to occur in a laboratory dish rather than *in vivo* <sup>[7]</sup>. The purpose of PGD is to identify what are considered to be abnormal embryos in order to select the most desirable embryos for [implantation](#) <sup>[5]</sup>. Diagnosis is comprised of two steps: extraction of one or two cells from an IVF-produced embryo, and application of the PGD test. PGD is important to [embryology](#) <sup>[8]</sup> because it has advanced IVF results and allowed couples more opportunities to deliver a child to term; however, it has also created much controversy.

Preimplantation genetic diagnosis was developed in the early 1980s as an alternative to post-[implantation](#) <sup>[5]</sup> prenatal testing. Before this technology, the only other forms of [prenatal diagnosis](#) <sup>[9]</sup> available involved amniocentesis and chorionic villus sampling (CVS). Both of these technologies involve the examination of embryos during later stages of development, when the embryo is already developing inside the mother. Couples who choose either of these tests must decide whether or not to terminate the [pregnancy](#) <sup>[10]</sup> if test results come back positive for a particular genetic disease.

In October 1989, Alan H. Handyside, who later became a preimplantation genetics consultant, performed the first successful PGD test for [cystic fibrosis](#) <sup>[11]</sup>, an X-linked disease. Initially, PGD was used as a form of gender selection to avoid having a child with a sex-linked disease. Since males are more often affected by X-linked genetic disorders, couples using PGD often choose to have only girls. PGD quickly came to be used for three main groups of inherited disease: single-gene mutations such as [cystic fibrosis](#) <sup>[11]</sup> and sickle cell [anemia](#) <sup>[12]</sup>, sex-linked disorders such as [hemophilia](#) <sup>[13]</sup>, and chromosomal abnormalities such as [Down syndrome](#) <sup>[14]</sup>.

With PGD testing, embryos created through IVF are cultured in a laboratory for three days until they reach the 8-cell stage. At this time a [blastomere](#) <sup>[15]</sup> biopsy is performed in which one or two of the blastomeres are removed by inserting a micropipette through the [zona pellucida](#) <sup>[16]</sup>, which surrounds the embryo. Removing these cells does not damage the embryo, as the cells can easily multiply to replace themselves and allow for the development of a normal [fetus](#) <sup>[17]</sup>. Once the totipotent [blastomere](#) <sup>[15]</sup> is removed, it is prepared for genetic testing.

Blastomeres are usually examined for chromosomal abnormalities when eggs are extracted from mothers past the age of thirty-five, as these women tend to be at higher risk of having a child with abnormalities. In order to examine the genetic components of a [blastomere](#) <sup>[15]</sup> the DNA must be removed, thereby destroying the cell. Fluorescent probes are added to the DNA, which then bind to specific chromosomes in a technique called fluorescence *in situ* hybridization (FISH). These fluorescent markers allow the chromosomes to become visible, enabling researchers to determine not only the sex of the embryo, but also the pairings of the

chromosomes. Embryos with pairings such as ?XXY? or ?X? can be eliminated to avoid [aneuploidy](#) [18], an incorrect number of chromosomes. If a [blastomere](#) [15] is diagnosed with [aneuploidy](#) [18], the embryo from which it was removed is expected to have the condition as well.

In order to test for dominant and recessive disorders, a polymerase chain reaction (PCR) is used. This process allows for the examination of single-gene mutations. DNA is extracted from the [blastomere](#) [15], and then a particular DNA sequence is amplified by PCR to facilitate its analysis. This is a very rapid and convenient method for technicians to test DNA. Through PCR, one molecule of DNA can be replicated into one billion molecules of DNA. Because the PCR reaction is very sensitive, it requires pure, high-quality DNA samples from only one cell. Although not very common, errors can occur and result in misdiagnosis leading to the discarding of a normal embryo, or an affected embryo being transferred into the mother.

The individual blastomeres of the embryo are totipotent, meaning that they have the same developmental potential as an embryo. Therefore, isolating a [blastomere](#) [15] involves the creation of a second, duplicate embryo, which will be destroyed during the diagnostic procedures. When the first successful [blastomere](#) [15] separation was announced it was openly acknowledged and initiated significant controversy due to that fact that a clone was created.

PGD has advanced the field of genetic testing significantly, but has also created concerns regarding [eugenics](#) [19] and genetic discrimination. The fact that there are no present federal or state laws to regulate the use of PGD brings in even more ethical ambiguity. Although PGD was originally thought to be a mechanism to discard embryos affected by a particular disease, some couples are now using this technology to actually choose embryos affected by a particular disease to be implanted in the [uterus](#) [20]. Such cases have occurred with parents with dwarfism and congenital deafness. Some parents have used PGD as a means of creating siblings who can provide transplants for their older siblings, or as a means of sex selection. Because there are no governmental regulations, parents could in theory use PGD in nearly any capacity to create a child.

## Sources

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