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Cancer Screening Before Birth

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Genetic screening of days-old embryos for genetic disorders began in the late 1980s using in vitro fertilization (IVF). Fifteen years later, it's making headlines after preimplantation genetic diagnosis (PGD) gained worldwide interest in early May 2006 when British regulators announced fertility clinics may use the process to screen for certain adult cancers.

The Human Fertilisation and Embryology Authority in Britain expanded the use of PGD to not only screen for genetic mutations that cause fatal childhood diseases, but also mutations that confer a high risk for adult cancers, such as BRCA mutations, which greatly increase the risk of breast and ovarian cancer. Although the practice of PGD is regulated or banned in other countries, the United States has no formal guidelines or legislation restrictions. Of the approximately 50 centers in the world that perform the process, most are in the United States.

The process begins with a medical and family history of both potential parents, which is used to zero in on the search for certain mutations. Using IVF, several eggs are fertilized within a test tube. Once an early-stage embryo grows to eight cells, the embryo is genetically screened by removing and examining one cell. Embryos that do not contain the genetic mutation are implanted into the uterus to begin the pregnancy. Further research is exploring options to screen sperm and eggs even before fertilization.

In addition to the BRCA genes, other cancers screened with PGD include familial adenomatous polyposis, which confers a nearly 100 percent chance of developing colon cancer, and retinoblastoma, a type of eye cancer that often results in loss of sight and second cancers. People born with a mutated retinoblastoma gene have a 90 percent risk of developing cancer, often in the first years of life.

Because having certain genetic mutations only presents a risk—not guaranteeing a person will develop the disease—opponents of PGD caution that the process screens out embryos that may not necessarily develop cancer or may develop a cancer with a high cure rate.

For now, no ethical guidelines exist in the United States for what should or should not be screened using PGD. And while several medical organizations have ethical guidelines on sex selection, none have formally addressed cancer screening using PGD. But as biotechnology improves, it may be used more often, especially for potential parents with inherited cancer mutations.