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More than Gender Selection: PGD Can Decrease the Risk of Genetic Problems in Babies

Pre-implantation genetic diagnosis (PGD) is a cutting-edge reproductive technology that can be used in conjunction with IVF (in-vitro fertilization). In popular culture and movies, you may have heard of this procedure being used to determine embryo gender prior to pregnancy. Science fiction films have used PGD technology as a plot to create fictional storylines based on the ability of PGD technology to pre-determine the physical characteristics of babies, such eye color, build and height, but these "designer babies" are not what PGD is intended for. This incredible technology deserves credit for so much more than the ability to identify embryo gender (or even physical characteristics). In this article, we will explore the many different indications for PGD, how it can help decrease the risk of genetic diseases and what the considerations and limitations of PGD are.

The Science of PGD

In-vitro fertilization is necessary for PGD to be performed. In a typical IVF cycle, oocytes (eggs) are removed from a woman's ovaries and are placed in a petri dish in the lab; the partner's sperm is added to the dish where fertilization can take place. Once fertilized with sperm, the oocytes become embryos, and are placed in a warm incubator in the laboratory for three to five days. It is during this 3-5 day period of time that PGD can be performed. By removing 1-2 of these tiny, rapidly dividing cells from the embryo, PGD technology can be used to allow scientists to screen each embryo for genetic diseases, chromosomal abnormalities and even gender. There are several screening methods available for screening these extracted cells. One method, FISH (fluorescent in situ hybridization), is where the extracted cells are fixed onto a microscope slide and hybridized with fluorescent-labeled DNA probes. Each of these probes are specific for a portion of a chromosome, which can be

used to identify and localize certain DNA sequences. For example, probes used for the Y chromosome will fluoresce on male embryos (only males contain the Y chromosome), and probes used to detect Down syndrome will fluoresce in a particular pattern in embryos containing a third copy of chromosome 21 (the cause of Down syndrome). A newer screening technology is CGH (comparative genomic hybridization) which is even more effective since it screens all of the chromosomes in a developing embryo. Once embryos are screened, a couple can choose to transfer only chromosomally normal embryos back into the uterus in attempt to achieve a healthy pregnancy.

Why PGD?

According to the American Society for Reproductive Medicine, birth defects occur in nearly one in every 20 pregnancies, ranging from minor anatomic abnormalities to severe genetic disorders or mental retardation. Some couples may have a greater than average risk of having a child with a birth defect depending on their ethnic, family and medical background. For example, Caucasians have a 3-10% chance of carrying the defective cystic fibrosis gene. Even

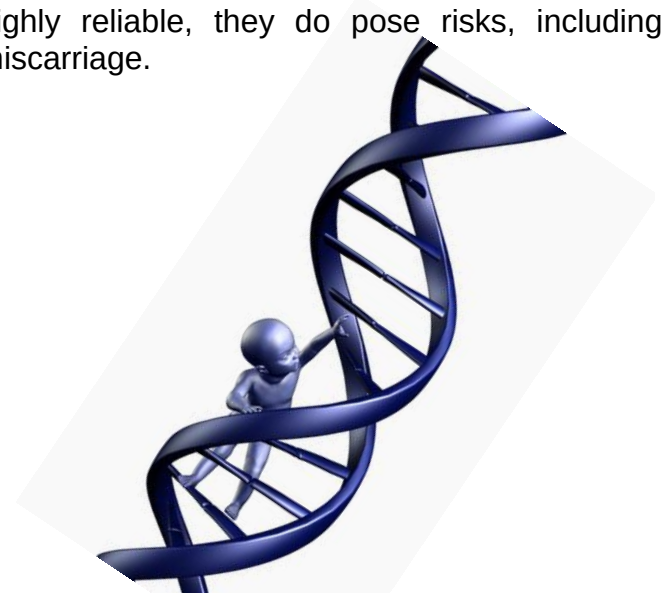
though carrying this gene is asymptomatic, if your partner also carries it you have a 1 in 25 chance of having a child affected by this devastating disease. Pre-conception blood testing can be performed on you and your partner to determine if you carry this gene, and if you are both found to be carriers, PGD can greatly reduce your chances of having an affected child. According to the American Society for Reproductive Medicine, other ethnic associations for specific diseases include:

- Sickle cell disease – as many as 1 in 10 African American's may be carriers
- Tay Sach's disease- Eastern European Jewish and French Canadian's have a higher chance of being a carrier
- Thalassemia- Greek, Italian, southern Asian and Mediterranean ethnicities have higher carrier rates.
- Spinal-muscular atrophy (SMA)- Caucasian, Asian Indian, Asian, Ashkanazi Jewish, Hispanic and African Americans can have as high as a 1:50 chance of carrying this disease even in the absence of a family history of the disorder.

If you have a family history of any inheritable disorders, you should seek genetic counseling prior to pregnancy to discuss what your chances are of having an affected child.

While PGD is not recommended as a routine procedure, it can be especially helpful for patients that have a known-carrier status for genetic diseases like cystic fibrosis, sickle cell or tay-sachs disease, have a history of recurrent pregnancy loss, or a family history of disorders or diseases like down syndrome, spinal-muscular atrophy, Huntington's disease, hemophilia or neural tube defects. Some women choose to use PGD in conjunction with IVF if they have failed several IVF cycles and/or are of advanced maternal age, since the chance of fetal chromosomal abnormalities increases with maternal age.

Prior to PGD technology becoming widely available, and still today, most women relied on pre-natal testing to determine the health of their unborn child. Ultrasonography, AFP, fetal cells in maternal blood testing, amniocentesis (removal of amniotic fluid from the womb for testing) or CVS (sampling and testing of placental tissue) are all excellent pre-natal tests that can help determine if an unborn child is affected by a birth defect, but can only be performed once pregnancy is achieved. If an abnormality is found, the woman must make the decision to either continue or terminate the pregnancy, depending on the severity of the fetal abnormality. Moreover, while invasive procedure like CVS and amniocentesis are highly reliable, they do pose risks, including miscarriage.



Because PGD screening of embryos is performed prior to implantation or pregnancy, it provides the information needed to transfer only "normal" embryos back into the uterus, thus increasing the changes for a healthy baby and reducing maternal stress in cases of recurrent pregnancy loss, known carrier status for a disease, or a family history of birth defects.

PGD and Gender Selection

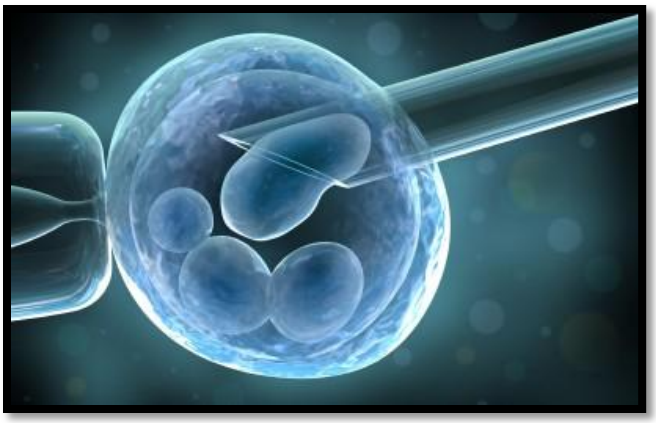
Knowing the gender of an embryo prior to implantation can be helpful especially if a parent carries an X-linked disorder, which means the disease in question is carried on the X (sex) chromosome. A child receives 23 chromosomes from the mother, and 23 from the father, however, the father is always responsible for the gender of the child.

The mother contributes an X sex chromosome to her child, and male sperm either contains an X or a Y chromosome. If a sperm containing a Y chromosome fertilizes an egg, the gender will be male (XY). If a sperm carrying an X chromosome fertilizes an egg, the gender will be female (XX). If the mother is a carrier of an X-linked disorder, such as hemophilia or Duchenne muscular dystrophy, and becomes pregnant with a boy, there is a 50% chance he will be affected since he only has one X chromosome inherited from his mother. In contrast, if a man carries an X-linked dominant disorder, like Alport syndrome, 100% of his daughters will have the disease. These gender-based genetic inheritance patterns make gender selection of embryos not only useful, but potentially life-changing, since the disease rates in certain genders can be so high. In these cases, rather than testing for the actual disorder, embryos can be selected based on gender to reduce the possibility of having a child affected by a debilitating disease.

Gender selection is not only used for medical purposes, but occasionally for the purpose of "family balancing". Some families-try as they might- seem to have one gender outnumber the other. In a family containing three sons, a daughter may be desired, and PGD can be used to specifically select a female embryo (or vice versa). Because gender preference and predominance in some cultures is a concern, PGD used for gender selection (in the absence of an X-linked disorder) is illegal in the UK and was banned in Canada until 2012. The United States doesn't have any strict laws pertaining to the use of PGD; however, the highly respected American Society for Reproductive Medicine discourages PGD used solely for gender selection in the absence of an X-linked disorder.

Limitations and Considerations

Like any technology, there are limitations to how much PGD can do. PGD technology is used to reduce the chances of debilitating diseases in offspring but is not always 100% accurate. Due to moisaicm (the one cell screened isn't representative of the other cells in the embryo) some embryos may be incorrectly considered "abnormal" and discarded, or may be deemed normal when they are not. In addition, PGD testing specifically looks for certain chromosomal patterns and abnormalities, but cannot screen every embryo for every kind of abnormality (there are thousands!). Pre-natal testing, such as AFP or amniocentesis may still be needed to confirm the absence of birth defects in pregnancies following PGD to confirm normal fetal development.



A single cell obtained from an embryo can be screened for gender or disease using PGD

Other limitations of PGD include the financial burden of the procedure itself; some medical insurances cover IVF if there is a medical indication for it, but PGD is typically not covered. Without any insurance coverage one IVF/PGD cycle can cost more than $10,000 (and pregnancy cannot be guaranteed). Further, there may be no "normal" embryos to transfer into the uterus to create a pregnancy after the screening-a result that is undeniably emotionally and financially devastating.

If you think PGD may be beneficial for you or someone you love, contact Fertility Solutions™. The physicians at Fertility Solutions™ have vast experience treating patients with recurrent pregnancy loss, genetic abnormalities, infertility and advanced maternal age. With over 50 years of combined experience, your Fertility Solutions™ physician can discuss your particular risk factors, help you navigate your financial and treatment options for pregnancy through IVF/PGD, and assist you in deciding if PGD is right for you.

News from Fertility Solutions

2013 was an amazing year for Fertility Solutions! We've helped bring hundreds of patients closer to their goal, and we've watched with joy as many left our office for the last time, headed for their OB office following successful treatment at FS.

A few highlights from this year:

Our IVF success rates (available online) from data collected from Jan, 2013 thru June 2013 show a 47% pregnancy rate in patients under 35, and 43% for patients age 35-37!

Dr. Carol Anania was nominated to Boston's Top Doctors for 2013, Dr. Huang was nominated to the Massachusetts Medical Society House of Delegates and Dr. Kowalik was selected to be in the Best Doctors database for 2014!

Dr. Moragianni has been reaching out to patients via radio- appearing on a half-hour medical show on the Greek-American radio program "Grecian Echoes"- offering truly fascinating and important information regarding the commonly asked questions and concerns of couples who are considering seeking treatment for infertility issues.

In early winter 2013 we announced our partnership with Fairfax Egg Bank, and we're excited for the opportunity to provide our patients the option of using frozen eggs from Fairfax Egg Bank- one of the nation's largest selection of frozen eggs.

While 2013 was amazing, we're sure 2014 is going to be even better. With the opportunity to do what we love and help patients achieve their dreams of building a family, we're aiming high and ready to exceed every goal and expectation in the year ahead! Cheers to a healthy and prosperous New Year!

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"Having a child is one of life's greatest gifts; we aspire to bring that joy to each and every one of our patients"

The doctors at Fertility Solutions are accepting new patients. Call 877-813-0159 to schedule a consultation with a board-certified specialist.

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