

To Pick and Choose

Preimplantation Genetic Diagnosis

By Heather Johnson Durocher

As a teenager, Michelle Steinhart of Irvine, Calif., was labeled a carrier of the genetic disease hemophilia. Even at that young age, she realized the dangers the disease presented to someday conceiving a child. That child could end up living a pain-filled life, similar to her father's.

Steinhart, 31, grew up watching her father suffer the effects of hemophilia, a disease so severe that he was not expected to live past 10 years old. In and out of hospitals throughout her childhood, her father succumbed to the disease in his 50s. "It meant a lifetime of pain and hospitalization and internal bleeding," she says. "It was horrible. It was a lot of pain for him and my family."

As a carrier of the disease, Steinhart would likely pass on an active form of the disease to a son. If she had a daughter, however, the girl would likely be a carrier or have a mild form of the disease. Hemophilia is a bleeding disorder caused by low levels of or absence of a blood protein that is essential for clotting.

After Steinhart married her husband, Michael Rappoport, she was eager to start a family. She was also deeply concerned about having a child with hemophilia. The couple explored their options, including adoption and the possibility of high-tech gender selection.

The couple ultimately opted for medical intervention. Through [in vitro fertilization \(IVF\)](#) and one of the newer advances in reproductive medicine – preimplantation genetic diagnosis (PGD) – Steinhart and Rappoport are now the parents of a baby girl, Samantha Ruby.

The procedure allowed the couple to select the female embryo, removing the risk of giving birth to a male child who would suffer from the disease. The process wasn't easy emotionally, financially or physically, but it did give them what they so desired: a healthy baby. "I would do it 100 times over to have my baby," Steinhart says. "I feel so blessed to have her."

PGD in Detail

PGD is used during IVF. According to Dr. Mark Leondires, medical director and lead physician with the Center for Advanced Reproductive Medicine in Norwalk, Conn., before the embryos are placed back into the mother, they are tested for chromosomal and genetic defects.

PGD was first used on human embryos in the late 1990s, says Dr. Leondires. At that time, the procedure was used strictly for genetic reasons – "carrier" couples with a high risk of transferring serious diseases to their offspring.

"There are over 40 single gene disorders that have been identified that can be tested for, from cystic fibrosis to sickle cell disease to Tay-Sachs," Dr. Leondires says.

New research from leading experts confirms that accurately diagnosing genetic abnormalities in the embryo reduces the risk of transferring an affected embryo by up to 88 percent. Dr. Leondires says that fertility experts are going a step further by saying that the procedure could prove beneficial for more couples, including those that have endured recurrent pregnancy loss.

"The majority of miscarriages – upward of 70 to 80 percent of miscarriages – are secondary to chromosomal problems within the embryo," he says.

Because these chromosomal problems usually come from the egg and not the sperm, screening the embryo could provide insight into potential problems before a miscarriage occurs. In other words, PGD would allow a couple to know which embryos secured for an in-vitro fertilization procedure are healthy.

One recent study evaluated the impact of PGD on women in high-risk pregnancy categories, including women who suffer from recurring pregnancy loss or two or more failed IVF cycles. The study revealed that PGD is promising.

"As a result of the study, we are now able to give couples answers never before made available regarding their chances for increasing pregnancy success and minimizing the risk associated with genetic disease," says Dr. Lawrence Werlin, the study's principal investigator and the founder and director of Coastal Fertility Medical Center in Irvine, Calif. Dr. Werlin also helped Steinhart and Rappoport have their baby.

The study's second phase is currently under way and further examines the effects that PGD has on women in high-risk pregnancy categories, including women of advanced maternal age. According to Dr. Werlin, preliminary results for advanced maternal age are looking more promising than previously discovered.

PGD Concerns

As promising as PGD is, experts express some concerns. Dr. Leondires has misgivings about using PGD when the reason for use is simply because a woman is older. He explains that not only does the patient have to undergo IVF, but then a 3-day-old, microscopic embryo must get a biopsy. "The procedure itself could cause harm to the embryo," he says.

The lack of long-term data also leads Dr. Leondires to be cautiously optimistic about PGD. While more than one million IVF babies have been born, less than 500 PGD babies have been born, he says.

"We have a habit in this field of saying, 'If this is good for these people, it should be good for others,'" he says. "Universal application of this procedure should be done with great caution."

For Dr. Werlin, studying PGD and using it to help couples in need is a personal long-held passion. He agrees that as the technology advances, medical guidelines will be needed.

"Part of what we'd like to do is put together outlines of ethical issues that need to be addressed and hopefully address those issues before they become problems," he says.

Experts also don't want to give false hope to couples. "Patients want to think that if they do PGD, they're guaranteed to have a normal or healthy baby, but there are no guarantees," Dr. Leondires says. "Even when we biopsy the embryo and find out it's normal and put it inside, there's a 10 percent incidence of the biopsy not being predictive of the pregnancy. The biopsy could still be wrong."

Dr. Werlin adds that the procedure is very exciting and promising, but he cautions that it's not for everyone and needs to be looked at on an individual basis.

PGD in the Future

The future looks very bright for reproductive technology, and you might not have to wait 20, 10 or even five years to benefit from current research efforts.

"There are studies going on that are beginning to look at all 23 [chromosomes] to rule out a problem," Dr. Werlin says. "When that becomes a reality, people who will do IVF will also more likely want to do PGD. The future is very exciting. As a technology right now, there are drawbacks. It only looks at nine chromosomes right now. Should one of the other 14 be abnormal, it may be a pregnancy that doesn't go on, or the patient doesn't get pregnant."

Also on the horizon are the findings of another study by Dr. Werlin assessing the alarmingly high multiple birth rates in the United States due to the success of IVF. According to Dr. Werlin, premature delivery of high-order multiples and all of their inherent risks need to be addressed. In an effort to seek a solution to the problem, researchers have designed a protocol using PGD as a tool to encourage placement of no more than two embryos at the time of transfer.

Steinhart and Rappoport are among those couples grateful for any research related to PGD. They are also interested in seeing what the future brings. Their daughter may face similar reproductive decisions if she tests positive as a carrier of hemophilia and could benefit from advances in PGD. "People like me, people like my daughter – this is our only hope of having a healthy baby," Steinhart says.