

Screening of embryos helps avert miscarriage

By Carey Goldberg, Globe Staff, 6/13/2003

It began more than a decade ago as a form of futuristic baby making: Researchers had found a way to screen fertilized eggs for some genetic flaws, allowing parents to implant only those that were free of certain inherited diseases.

Ethicists sounded alarms about an impending era of "designer babies," but use of the technique, called preimplantation genetic diagnosis, remained rare. It was only last year that the 1,000th genetically screened baby was born, by doctors' worldwide count.

Fertility specialists say that preimplantation screening is now becoming common at cutting-edge clinics. In about three out of four cases, it is used not to choose disease-free babies, but to help women who are older or who have had repeated miscarriages to produce viable babies.

The earlier philosophical debates over preimplantation screening are giving way to more mundane concerns: In Massachusetts, a new bill would mandate that health insurance coverage for infertility be expanded to include treatments for recurrent miscarriage, including preimplantation screening.

"It seems to me that as medicine evolves in this area, the laws should keep up," said state Senator Andrea F. Nuciforo Jr., Democrat of Pittsfield, who is a sponsor.

Current law requires insurance coverage for fertility treatment only for those unable to conceive, not for women unable to carry a baby to term. New Jersey and Illinois have already passed similar bills requiring expanded coverage.

In preimplantation genetic diagnosis, doctors take an embryo conceived in the lab and remove one cell to examine its genes. The technique was first used to check for disorders like cystic fibrosis, which is caused by a defect affecting a single gene. But today the screening can detect broader genetic flaws that could keep the embryo from implanting and developing normally.

For one North Shore couple who had suffered four miscarriages, preimplantation genetic diagnosis has already made a difference. The wife is now five months pregnant after the screening determined that two of eight embryos she produced had sufficiently normal chromosomes to implant.

"If not for PGD, I don't think she could have gone through more of the unknown," the husband, who spoke on condition of anonymity, said of the screening. "You just lose hope."

They couldn't persuade their insurer to cover the procedure, so they paid about \$3,600 out of pocket. Now they are among those pushing for the bill to expand infertility coverage.

The Massachusetts Association of Health Plans opposes the bill, arguing that state-mandated infertility coverage is already expensive and that some preimplantation screening techniques remain experimental.

"We're not at a point where it's wise to take this on as a mandate, whether from a financial, ethical, or even medical point of view, because it's still very much a procedure in development," said Dr. Marylou Buyse, the group's president.

Preimplantation diagnosis usually adds about \$3,500 to the cost of an in vitro fertilization cycle, which is generally about \$10,000.

Currently, about 2 percent of the tens of thousands of in vitro procedures done each year nationwide use such screening, estimated Dr. Santiago Munne, director of preimplantation genetic diagnosis at Saint Barnabas Medical Center in Livingston, N.J., one of three top centers that accept samples for analysis from around the country.

Use of the technique has increased rapidly, and about 1,000 screenings are now done each year in the United States, said Dr. Yuri Verlinsky, a specialist at the Reproductive Genetics Institute in Chicago.

However, researchers are still trying to determine how effective the screening is for various types of patients. The first clinical trial in which women seeking fertility treatment were randomly given the genetic screening found that it significantly helped women who have suffered repeated miscarriages. Of 57 patients in the study, 64 percent of women with miscarriage problems who used the screening became pregnant, compared with 38 percent in the control group. The study will be published in August in the journal *Fertility and Sterility*.

The study indicated that preimplantation genetic diagnosis may also help women who have fertility problems because of age, but more data is needed to be sure. Some leaders in the field report even broader success with the technique. Dr. Munne estimated that 80 percent of his preimplantation genetic diagnosis patients are older women and that it improves their in vitro fertilization pregnancy rates by up to 50 percent. Without the screening, successful pregnancies result in less than a quarter of in vitro fertilization attempts in women over 38.

Dr. Lawrence B. Werlin, principal investigator of the study to be published in *Fertility and Sterility*, is optimistic about the future of the technique. In time, he said, "you can picture that everyone who does in vitro fertilization would want to know they're putting back embryos that have normal chromosomes, and it will dramatically change IVF for the better."

Others remain skeptical. Preimplantation genetic diagnosis clearly helps many women who suffer from repeated miscarriages, said Dr. Mark R. Hughes, a pioneer of the screening at Wayne State University in Michigan. But "the data are simply not available yet regarding the value of routine chromosome testing for everyone seeking infertility treatment," he said.

Even as the debate over such screening continues, the technology is leaping ahead. Right now, labs must choose between checking for a single-gene defect like Huntington's disease or for the nine most common chromosome defects. Soon, perhaps in a couple of years, specialists say, they will be able to check every single chromosome. Already, they say, the screening can help with one of the biggest problems of fertility treatments, the increased likelihood that a mother will bear twins or higher multiple births because doctors implant several embryos to improve the odds that one grows into a fullterm fetus.

Because preimplantation genetic diagnosis allows doctors to be sure they are implanting genetically sound embryos, they are likely to implant fewer of them. Ethical questions remain, however. One of the hottest questions is whether parents should be allowed to use preimplantation screening to choose the sex of a baby. Most clinics decline to do gender selection, but parents still request it.

Some ethicists continue to sound warnings about the pitfalls of introducing such a conscious process of selection into reproduction. Some say it takes evolution out of nature's hands, with unpredictable consequences. The screening also has an error rate estimated at between 5 and 10 percent, so women worried about having an abnormal baby still need to undergo some kind of prenatal screening like amniocentesis. And there is a small chance that the test itself, which involves removing one cell when the embryo is only eight cells, could harm the embryo.

Even enthusiasts don't recommend preimplantation screening for everyone. Dr. Joseph Hill at The Fertility Center of New England, which does about 20 such screenings per month, recommends them for couples with known chromosomal abnormalities or a record of repeated miscarriages. Hill also does such screening for women whose in vitro attempts have failed despite healthy-looking embryos and for women over 38 who are getting in vitro fertilization anyway, because so many of their eggs are likely to be genetically abnormal. Rosanne Klovee, a 32-year-old family lawyer from Medford, fits the bill. She has endured the misery of five miscarriages, the apparent results of genetic flaws in the embryos.

With her next pregnancy, Klovee plans to pay for preimplantation genetic diagnosis out of pocket. She remains confident of becoming a mother, but is hoping that the screening will improve her chances and give her some answers.

"I'm a very strong person, but there's a limit to how much I can take," she said.

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