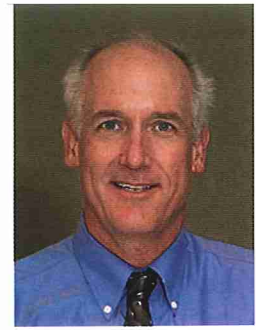


## Advanced Fertility Options: Preventing Genetic Disease through

# Embryo Biopsy

by William Meyer, MD

Dr. Meyer received his medical degree from the University of Virginia in 1983. He completed an obstetrician/gynecologist residency at Emory University, and then went to Yale University Hospital, where he received advanced training through a reproductive endocrinology and infertility fellowship in 1990. He is board certified in both Ob/Gyn and Reproductive Endocrinology and Infertility. In 2006, Dr. Meyer formed Carolina Conceptions with Dr. Grace Couchman. Along with Dr. John Park, the three physicians comprise an experienced medical team offering exceptional success rates, innovative treatments and compassionate care to earn a reputation as one of the fastest-growing infertility clinics in the southeast. To learn more, visit [www.carolinaconceptions.com](http://www.carolinaconceptions.com).



Recently two areas of in vitro fertilization (IVF)—egg freezing and preimplantation genetic diagnosis (PGD)—have dramatically changed the options physicians at Carolina Conceptions can offer their patients. While Carolina Conceptions offers both techniques, PGD, better known as embryo biopsy, has more well-defined uses and has rapidly changed our conception of disease prediction, detection and prevention.

PGD is the newest of the reproductive technologies that allows for diagnosis of a disease even before the embryo is implanted back into the uterus. Couples who are carriers of diseases, such as cystic fibrosis, spinal muscular atrophy or familial cancers, now have the ability to prevent their children from inheriting such genetic disorders with the use of this micromanipulation technique.

Carolina Conceptions performs embryo biopsy in our on-site lab after the cleaved embryo has been in culture media in a low-oxygen tension incubator for three days. At this time a single cell, or blastomere, is removed from the eight-cell embryo after using a laser to create an opening in the shell, or zona pellucida. With gentle traction the blastomere is squeezed through the aperture and then either fixed on a slide or suspended in a solution and sent for genetic analysis. Only the healthy unaffected embryos, now called a blastocysts, are placed back into the woman's uterus under ultrasound guidance.

Cystic fibrosis and spinal muscular atrophy are two of the most commonly inheritable diseases. It is estimated that 1 in 30 to 40 people are carriers of either disease. Before PGD, couples in which both parents were carriers had several options. One was to try their luck by conceiving and then use antenatal testing to determine if the pregnancy was affected, and then the pregnancy could be terminated or carried to term. Other options included doing no testing or using donor sperm.

In the last two years, Carolina Conceptions has performed eight cases of PGD in couples who are carriers of either cystic fibrosis or spinal muscular atrophy. Currently, 6 out of the 8 couples have succeeded in delivering a healthy unaffected child.

Overall, Carolina Conceptions has currently performed 27 embryo transfers after PGD for such conditions as cystic fibrosis, spinal muscular atrophy, Trisomy 21 and Robertsonian, and various reciprocal chromosomal translocations in couples with repetitive miscarriages. A total of 14, or 55 percent, of the transfers have resulted in pregnancy. Currently, we have couples who are carriers of sickle cell, Tay-Sachs and specific single-gene mutations undergoing preparation for ovarian stimulation and IVF.

One of our most recent PGD success stories comes from a couple in Macon, N.C., who discovered they were both carriers for spinal muscular atrophy. They spontaneously conceived their first child, who at six weeks of age was diagnosed with the fatal disease and subsequently passed away four months later. Together, the couple shared a 25 percent chance of having another affected child, a 50 percent chance of conceiving a carrier and a 25 percent chance of delivering a perfectly healthy baby. After being presented several options, the couple decided to pursue IVF with PGD at Carolina Conceptions. The couple conceived during each of the two attempts at IVF. They now have two healthy children: a 2-year old and 4-month old.



Although most diseases screened by PGD manifest themselves at birth or in early childhood, it is now possible to test for diseases that manifest themselves later in life. We have now expanded the use of PGD to include BRCA1 and BRCA2 and the testing of genes like p53 mutations that may predispose one to familial cancers.

Currently, more than 3,000 IVF cycles have been accompanied by PGD internationally, resulting in approximately 700 live births. With the completion of the human genome project, virtually any genetically inherited disease can be identified in a single cell. So, the applications of PGD appear endless.