Gene Shopping: Parents Won't Pass on Deadly Diseases with New Procedure

With PGD, Mom Didn't Pass on Husband's Colon Cancer Gene

March 8, 2007 — - There's no doubt that blue-eyed Chloe Kingsbury is a special kid, but how she came into this world can only be described as exceptional. Chloe's father, Chad Kingsbury, carries a gene for a deadly genetic colon cancer that has taken the lives of four family members, including his mother and grandfather. "I have seen what this disease can do firsthand. I held my mother's hand while she died," he said. "I look at my grandma. Every time I see her there is an emptiness there. She lost all three of her children, and I just … I just couldn't do that." Since he carries the gene, Chad has an 85 percent chance of getting colon cancer and his offspring have a 50 percent chance of inheriting the gene.

But a new advance in genetics and reproductive medicine called preimplantation genetic diagnosis, or PGD, which uses in vitro fertilization has allowed the Kingsburys to bring Chloe into the world with no fear of getting the disease. "[PGD] is basically getting a diagnosis of an embryo prior to implantation or prior to pregnancy," said Renee Genovese, who works in clinical and molecular genetics at the Reproductive Genetics Institute.

When they decided to have their second child, the Kingsburys invited "Good Morning America" to join them as they went through the process again at Reproductive Genetics Institute in Chicago. The couple freely admits that despite the success with Chloe, going through with the procedure has not been an easy decision. "The whole thing sounded incredibly foreign and expensive and then there was this whole debate going on … about if we really needed to do this," Chloe's mother Colby Kingsbury said. The process starts with in vitro fertilization, where a mother's eggs are retrieved from her ovaries. In the Kingsburys' case, 16 of Colby's eggs were extracted, and each one was combined with Chad's sperm to form an embryo. The procedure was repeated 16 times, giving the lab 16 embryos to work with.

A few days later, when the embryo had grown to just eight cells, one cell was removed from each embryo for genetic testing. "We can test the embryo and we can test it with a single cell to see whether the embryo has any genetic abnormality or not," said Dr. Ilan Tur-Kaspa, who performs the procedure at the Institute. Still, it's a part of the procedure that can be alarming for some parents.

"I always reassure patients … the baby is not going to be missing an arm or another part of the body. It doesn't work like that," said Dr. Richard Paulson, the director of fertility at the University of Southern California. "The embryo is a blueprint that allows the formation of the whole pregnancy -- that means the baby and the placenta and the gestational sac -- all of that."

Just five days after the testing, the Kingsburys learn the results; of 11 fertilized embryos, three had the gene, which left eight healthy embryos. Two embryos were transferred back into Colby, and the remaining embryos were frozen, in case Chad and Colby want to try again. "You just want to make a happy life for your child, and that is all we are trying to do," Chad said. Colby and Chad successfully conceived and Colby is now five months pregnant.

Even now with their success, the couple says that the decision was difficult. "I mean we go to church, you know. … We have family that, you know, was thinking that, that it was expensive," Chad said. "I weighed the pros and cons of this and I couldn't, I couldn't pass that onto my daughter, knowingly.

"I have about an 80 to 85 percent chance of getting colon cancer. I get screened regularly. … I'm trying to combat it as best I can. I just want, like anybody does, I just wanted a healthy kid and not have to go through the worries that I go through." While some people equate the procedure with terminating a pregnancy, Colby disagrees.

"We have a beautiful daughter and we have another one on the way. … I don't see it that way," she said. "It's something that was right for us." "Good Morning America" medical contributor Dr. Tim Johnson drew a line between creating "designer babies" based on appearance and choosing embryos based on a life-threatening disease.

"I think there's a big difference between choosing, selecting for eye color or height or athletic ability or appearance, and choosing for potentially, and in some cases, a certain lethal disease, like the genes for Huntington's, for example," Johnson said. "I think we're capable, both professionally and societally, of making that distinction."

"There's a gray area in the middle where it's going to be more difficult," he continued. "But this kind of decision where you're talking about preventing colon cancer, to me is very clear."

To learn more about Dr. Ilan Tur-Kaspa go to http://www.reproductivegenetics.com/.

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