

Sometimes takes a touch of science

By Ayesha McAdams-Mahmoud
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Wendy and Chris Baker carry the recessive gene for Tay-Sachs, a fatal genetic disorder that took the life of their first son, Benjamin. Their daughter, Caroline, was born healthy, but rather than risk the chance another child would have the disease, the couple opted for a rare medical procedure called preimplantation genetic diagnosis. Their son, Jacob, will be born this week by C-section.

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In two days, Jacob will be born, but his will be no ordinary birth.

Jacob is one of hundreds of babies worldwide who are conceived with precautionary measures. They are miracle children, really, born disease-free to parents who think it impossible.

They are products of a rare medical procedure called preimplantation genetic diagnosis (PGD). PGD combines the technology of invitro fertilization and DNA analysis to allow parents to have children free of the genetic mutations they carry.

Jacob's parents, Wendy and Chris Baker of Rock Hill, sought the procedure because they are both carriers of a gene for Tay-Sachs disease, a recessive and fatal genetic disorder that causes severe deterioration of mental and physical abilities. Most children with Tay-Sachs die before the age of 4.

Any children the Bakers bring into this world have a one-in-four chance of having Tay-Sachs. The couple watched their first child, Benjamin, die from the disease.

"Each month, you would notice something else would be gone," Wendy said.

Benjamin developed normally for the first few months of his life. But the Bakers watched him struggle to pull up, take steps and stand. They began to worry when a pediatrician saw cherry-red spots in his eyes – a trademark of Tay-Sachs.

Wendy, a nurse practitioner and a supervisor at the York County Department of Health, remembers: "I knew that no matter what, if they saw cherry-red spots, it wasn't going to be a very good diagnosis."

All people are born with genetic mutations, but most never know it until they meet someone with the same mutation and try to have children. Wendy, 31, and Chris, 32, learned they were both carriers of the Tay-Sachs gene only after Ben was diagnosed.

Chris, who works at a local factory, started working part-time to spend more time with Ben. He and Wendy were unaware of how bad the disease could get when they decided to conceive their second child, Caroline. Fortunately, a test 10 weeks into Wendy's pregnancy showed Caroline, now 2, didn't have Tay-Sachs, and she was born healthy.

But Ben's condition worsened. He quickly became a toddler who battled seizures and struggled to sit up and swallow. Benjamin died in August 2004, when he was 3 1/2 years old.

The Bakers wanted Caroline to have a sibling, but they didn't want to risk another case like Benjamin's.

"After losing Ben and watching him go through the progression of Tay-Sachs," Wendy said, "I knew there was no way I could terminate my pregnancy due to an infected child."

They considered adoption and artificial insemination, but those options were too expensive.

Another couple they met through the National Tay-Sachs and Allied Disease Association told them about the PGD procedure. Their insurance covered part of the invitro fertilization process.

Taking what they learned from the couple they'd met and from the Internet, the Bakers headed to the Reproductive and Endocrinology Associates of Charlotte to ask for a PGD.

"Every couple has to make that decision about how they feel about growing their family," said Dr. Nancy Teaff, the Bakers' doctor at the center. Wendy remembers Teaff being surprised by how much she and Chris knew about PGD.

Teaff, a reproductive endocrinologist who has worked with couples like the Bakers for 14 years, said that when PGD works, it enhances a family's quality of life.

"My job as a doctor is to limit pain and suffering," Teaff said. "And I'm talking about psychological pain, too. I mean, it's difficult to see your baby die."

Dr. Mark Hughes, a molecular geneticist and founder of the Genesis Genetics Institute, led the team that developed the world's first PGD procedure 16 years ago.

"This technology came out of pure frustration," Hughes said from his Detroit office. "I'd meet couples like the Bakers and have to explain to them how this happened to them, and that they'd have a 25 percent chance of this happening again."

Hughes initially wanted to help partners who both carried genes for cystic fibrosis. Now his lab can conduct PGD analysis on 200 diseases -- including hemophilia, sickle cell anemia and cystic fibrosis. Hughes' lab receives data from couples at nearly 150 invitro fertilization programs worldwide.

Shortly after Benjamin's death, Wendy and Chris started the Ben Baker Memorial Fund through the Tay-Sachs & Allied Disease Association. The fund has raised \$23,000 for research in two years.

And the family remembers Benjamin in other ways.

Wendy said Caroline recognizes Benjamin in pictures and talks about him.

"We always laid him next to her," said Wendy, who is scheduled to deliver Jacob by C-section on Wednesday. The sociable blonde girl with bright blue eyes said she is excited to have a baby brother on the way.

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Sidebox 1: The Preimplantation Genetic Diagnosis Process

Background: All people carry genetic mutations. Some are more common than others according to ethnic and racial lineage. Sickle cell is more common in

blacks, while Tay-Sachs is more common in Jewish people. The couples who seek preimplantation genetic diagnosis, or PGD, can have children naturally, but opt against doing so to ensure the quality of their child's life.

Step 1: Once a couple know the genetic mutation they carry, a PGD lab conducts bloodwork so that a molecular basis of the genetic disease can be determined for the family.

Step 2: The couple begins the invitro fertilization process, in which the woman takes drugs to stimulate her egg production. Then, an average of 12 to 15 of her eggs are combined with the sperm from her male partner in a lab. After fertilization, the embryos are allowed to grow for three days.

Step 3: One cell is removed from each embryo for genetic analysis and sent off to the PGD lab. The embryos are watched closely to ensure continued growth.

Step 4: For 19 hours straight, a team of scientists conducts molecular DNA testing of the single cells that have been biopsied from the couple's embryos. The scientists can only look for the disease they know the couple carries, which means that genetic mutations unknown to the couple will not be found.

Step 4: The genetic lab sends a report back to the IVF center stating which cells have the disease, which are carriers of the genetic mutation, and which do not have the disease.

Step 5: The second half of invitro fertilization, in which the embryos are implanted in the woman's uterus, occurs. The embryos which are free of the disease and, possibly, those that are carriers are chosen for the transfer to the woman's uterus. Usually, 2 or 3 embryos would be transferred, depending upon availability. There is no guarantee that the woman will get pregnant through invitro fertilization, but 75 percent of patients under age 38 should "take home a baby" within three tries of invitro fertilization.

Cost: Invitro Fertilization costs about \$15,000, and PGD analysis costs about \$2,750 at Genesis Genetics Institute.

Sidebox 2: Local PGD and Tay-Sachs Resources

National Tay-Sachs & Allied Disease Association, www.ntsad.org.

Genesis Genetic Institute, www.genesisgenetic.com.

Reproductive and Endocrinology Associates of Charlotte, www.reachdrs.com

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