A microscopic photo of an embryo biopsy — a single cell is removed three days after egg insemination.

Microscopic Miracles

Modern technology screens embryos for Jewish genetic disorders.

Robin Schwartz
Special to the Jewish News

Human genes are a part of the makeup of each and every one of us; they're the blueprints in our cells that determine whether our hair is straight or curly, our eyes brown or blue. It's mind-boggling to think about, but scientists have discovered that each cell in the human body contains 20,000-25,000 unique genes, and we're made of trillions of cells. We're complex individuals, filled with microscopic bits and blips of information, yet we all start out the exact same way — as a tiny speck barely visible to the human eye.

"It's important to point out that even a minute error in the genetic code, like a single typo in a 1,000-page novel, can lead to devastating health consequences," said Dr. Michael Mersol-Barg, medical and laboratory director for the Center for Reproductive Medicine & Surgery in Birmingham.

"New technology can filter many of

Microscopic Miracles on page 24

A Story Of Life-Invigorating Echoes

It is a story of inspiration and imagination — a compelling account of medical advances providing hope to couples in the Jewish community who carry genetic-related diseases.

This intriguing story of assisted reproduction offers reverberating potential for healthy babies free of the identified diseases.

The *JN* felt the story had not been told on a wide scale, however.

So when the *JN* received a call from a local couple who had undergone successful counseling and treatment from a Birmingham reproductive endocrinologist, Dr. Michael Mersol-Barg, saying they wanted to share the story behind the birth of their daughter last year, we shifted into story-planning gear.

We felt it was important to help *JN* readers negotiate the two-step pathway to preventing passage of genetic disorders from parents to children.

Interested couples first submit to a screening to determine if *both* carry a diseased gene prevalent in the Jewish community. Carrier couples are directed to seek medical care from a specialist and to undergo in-vitro fertilization with genetic screening of embryos; only embryos without the disease for which carrier couples are at risk are placed into the womb.

When our host couple and their medical doctor, as well as another local couple with a similar story, were all on board, and we were assured access to the Jewish Genetic Disease Consortium for important context, we felt comfortable moving ahead with today's cover story by Contributing Writer Robin Schwartz.

A companion piece on the rabbinic perspective of assisted reproduction rounds out the *JN* presentation.

As always, we welcome your thoughts on this ever-evolving subject. Please direct submissions to: letters@thejewishnews.com.

— Contributing Editor Robert Sklar

these 'typos,'" he continued, "and help produce healthy babies."

Mersol-Barg, who is Jewish, is a reproductive endocrinologist who assists countless couples navigating the medical and emotional pitfalls of infertility. He's a member of numerous medical societies, president of the Michigan Reproductive Endocrinology and Infertility Society and serves as chair of the state's Fertility Advisory Committee. He's also an enthusiastic advocate for genetic testing, important for Jewish couples who may unknowingly be carriers for various genetic disorders like Cystic Fibrosis, Tay-Sachs, Canavan, Familial Dysautonomia and several other illnesses prevalent among Jews of Ashkenazi (Eastern European) descent.

According to the New York-based Jewish Genetic Disease Consortium, "approximately one in four individuals is a carrier of a gene for a condition that could be severe and may result in the early death of a child. Until we learn how to correct inherited genetic mutations, every generation of Ashkenazi Jews is at risk for passing on a gene for one of several diseases."

Carriers are healthy; their families may or may not have any prior history of disease. A simple blood test, which can be ordered by any physician, is all it takes to determine a person's carrier status.

"It can be a silent assassin," said Mersol-Barg. "These disorders are recessive so they're sneaky. Someone can be a carrier and not know it and fall in love with another carrier and have a one-in-four chance of having a child with the illness."

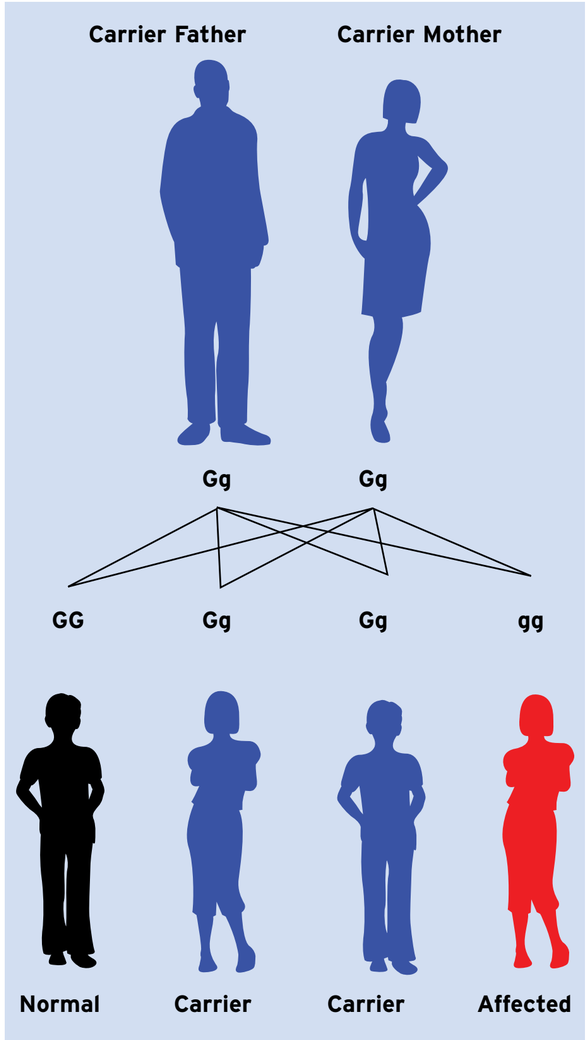
While he works with many couples struggling with infertility, Mersol-Barg points out that fertile couples, who may have no trouble conceiving at all, are really the best candidates for genetic testing.

"They're the ones who are going to get hurt because they have babies easily," he said. "This is a great opportunity to inform the community so they don't get blindsided. It's my mission to let people know about the technological advances available to assist them."

A 'Perfect' Match

So what happens when a person finds out he or she is a carrier?

It's an emotional rollercoaster a local couple in their 30s experienced firsthand. They asked not to be identified, but agreed to talk to the *Jewish News* with the hope of helping other families. Their story starts out like so many others — they met, fell in love, got married and began trying to have children. Because of a prior blood test, performed when she was single, the wife already knew she was a carrier for Gaucher's disease, an inherited disorder that affects many of the body's organs and tissues. The disease afflicts one in 500-1,000 Ashkenazi Jews, according to the U.S. National Library of Medicine. It can cause a wide range of problems, including anemia, easy bruising, bone pain and fatigue. In the most severe cases, life-threatening complications start before birth or in infancy, and most babies die a few days after birth. But both parents must be carriers and each must pass on their defective gene for a child to be affected. So you can imagine the local couple's surprise when the husband was tested and they learned he's also a carrier for Gaucher's disease. It's rare for two people to have the exact same genetic mutation, but they do.



If both parents are carriers of a recessive genetic defect, one in four (25 percent) children will be healthy and will not carry the genetic defect, two of four (50 percent) will be healthy, but will be carriers, and one in four (25 percent) will have the disease.

"These disorders are recessive so they're sneaky. Someone can be a carrier and not know it and fall in love with another carrier and have a one-in-four chance of having a child with the illness."

– Dr. Mersol-Barg

"It was very shocking to us; it was very upsetting," the wife recalled. "We were already dealing with infertility issues at this point, and we were already emotional and feeling overwhelmed. Now we had a 25 percent chance of having a child with Gaucher's. We didn't want to take that chance. We didn't think it was fair for us to roll the genetic dice and possibly have a sick child when the science exists for us to have a healthy one."

The couple made the decision to undergo a process called pre-implantation genetic diagnosis, where embryos are screened in a lab for the defective gene before they're implanted in the womb with the hope of creating a healthy pregnancy. It works much like in-vitro fertilization (IVF), where a woman's eggs are removed and fertilized in a lab. But once the embryos begin to grow, doctors have figured out a way to gather critical information about the baby-to-be as the cells are dividing in the earliest stages of development.

"On the third day, we use a laser to zip open the outer shell of the embryo," Mersol-Barg explained. "We remove one cell. That cell will represent the others to answer the question we want to know."

The microscopic cells are then sent to a lab for testing to determine whether the baby will have the disease, be disease-free or be a carrier (but not affected by the illness). Embryos that test positive for the defective gene are discarded; only those found to be disease-free or potential (unaffected) carriers are implanted.

As it turns out, one of the leading labs in the world specializing in this type of testing is right here in Detroit. The scientists at Genesis Genetics Institute pioneered pre-implantation genetic diagnosis almost 20 years ago. They're now able to screen for specific genetic disorders and get results back within 24 hours.

"We handle some of the rarest cases," said Shannon DeWall, a genetics counselor with Genesis Genetics. "This [technology] is not foolproof, it's not guaranteed; but it is something to help stack the odds in their favor. It's a positive thing for a lot of families, and it's nice to help people who are trying to expand their families and have healthy children."

The lab also is helping develop lines of embryonic stem cells with the University of Michigan to study disorders including Tay-Sachs disease, which causes the central nervous system to degenerate and often leads to death by age 4. Approximately one in 25 Ashkenazi Jews are Tay-Sachs carriers. In 2008, voters across Michigan approved the use of embryos discarded in fertility procedures for stem cell research.

The Florida-based group StemCellRegenMed announced in April that three children born with Tay-Sachs in Peru showed signs of improvement after receiving the groundbreaking gene therapy treatment.

Success Stories

Closer to home, the anonymous local couple referenced earlier can be counted among the pre-implantation genetic diagnosis success stories. They now have a healthy baby girl.

"I would give myself shots for three weeks and go for early-morning ultrasounds," the new mom recalled about the beginning stages of the process. "There's a silent sisterhood of women who go for these early-morning appointments and exchange encouraging glances.

"We had more than a dozen embryos tested," she continued. "Some had Gaucher's disease, and there were some where they couldn't determine enough information. We're not sure yet if our daughter is a carrier. But the genetic testing is phenomenal."

The procedures involved are costly, often in the tens of thousands of dollars. The genetic screening blood test alone is typically not covered by insurance and can cost several hundred dollars. But the new parents say, for them, there's no money better spent. They have some healthy embryos left, and they're planning to try again.

"This is one way we can end the chain of transmission," the new mom said. "That's why it bothers me when people talk about couples using labs to make 'designer babies.' For us, this technology helped create a healthy baby, and we will be forever grateful."

Hannah and Thad McCollum of Ferndale also count their blessings when they look at their healthy, growing, 1-year-old daughter, Molly. Without this technology, the two Metro Detroit attorneys are not sure what their lives may have been like. Hannah is Jewish; her husband is not. She has a brother who is developmentally disabled and requires 24-hour care.

After the couple got married several years ago, Hannah's mother is the one who insisted she go for genetic testing. She suspected her son has Fragile X syndrome, a chromosomal abnormality that often leads to mental retardation. The disease is not specifically tied to the Jewish community. Hannah was tested and found to be a Fragile X carrier.

"The condition is caused by too many repeats of a certain sequence on your X chromosome," she explained. "I have more than 600 repeats of the sequence. I have a very serious genetic disorder. I had about a 50 percent chance of passing it on to my child."

For that reason, the McCollums also underwent pre-implantation genetic diagnosis. After a long, emotional process, they welcomed their baby girl last year. Both the McCollums and the anonymous couple were treated by Mersol-Barg.

"This is truly a miracle," Hannah said. "It's stressful. We had a team in the room when I got pregnant. Parts of it are scary, and parts of it are kind of weirdly depressing. But it's amazing that you can do this. You can take what's essentially this organic process and you can defeat it so you have a child that's not carrying the disease you're trying to avoid." □

Thad and Hannah McCollum with their 1-year-old daughter, Molly

Rabbis Encourage Genetic Testing Before Marriage

Before they even agree to date or develop any kind of emotional connection, Orthodox Jews typically undergo genetic testing. The results are then used to determine whether or not the relationship should even be pursued in the first place.

"You don't want to get into a relationship that will not have such good endings," said Chaya Stein, 30, of Oak Park.

She and her husband are busy raising five children ages 6, 4, 2-year-old twins and a 9 month old. "I think it's imperative for everyone to check before they have children. It's very painful to watch a child who's born healthy deteriorate. What could be worse for a marriage?" she said.

Stein, like thousands of others, underwent anonymous testing through Brooklyn, N.Y.-based Dor Yesharim ("Generation of the Righteous" in Hebrew), also known as the Committee for Prevention of Genetic Diseases. The organization has offices in Israel and other countries.

"They basically give you a number," she explained. "When two people want to date, you call and find out if your numbers are compatible or not. It's almost like protocol; that's just what you do."

Dor Yeshorim tests for nine disorders, including Tay-Sachs, Familial Dysautonomia, Cystic Fibrosis, Canavan, Glycogen Storage Disease, Fanconi Anemia, Bloom Syndrome, Neimann-Pick and Mucolipidosis. It also tests for Gaucher's disease by request only, a source of controversy according to a 2008 article published by the New York-based *Jewish Week*. In the article, Dr. Stuart Ditchek, director of the nonprofit Jewish Genetic Diseases Consortium, accused Dor Yeshorim's founder, Rabbi Joseph Eckstein, of "playing God" because he dissuades people from testing for Gaucher's. According to the article, Ditchek also claimed Eckstein failed to inform people who tested positive for Gaucher that they were carriers. The rabbi denied the assertion, but said, "We don't believe it's right to say 'don't get married' because of Gaucher's" because there are treatments for it. The group reportedly tests about 17,000 people (primarily Orthodox Jews) each year.

"I personally have eight married children who went through the testing before scheduling a date," said Rabbi Elimelech Silberberg of the Sara Tugman Bais Chabad Torah Center in West Bloomfield. "This negates any emotional pain if the couple is not compatible. It's much more difficult to deal with these issues if they've already developed a relationship. Genetic testing has been the norm in observant circles for close to three decades."

Conservative and Reform rabbis we spoke with also encourage couples to undergo genetic testing, but generally do so later in a relationship as part of pre-marriage counseling.

"I once counseled a couple who were tested and found to be positive carriers of Tay-Sachs," said Rabbi Joseph Krakoff of Congregation Shaarey Zedek in Southfield. "Having this information in advance, doctors managed the pregnancy and everything worked out perfectly when it came to having children."

Rabbi Paul Yedwab of Temple Israel in West Bloomfield says he has been encouraging genetic testing for the last 25 years.

"Tay-Sachs especially is a devastating disease," he said. "It would be irresponsible for any rabbi or pre-marital counselor to fail to give couples the information they need." □

– Robin Schwartz