Gene tests bring agonizing choices

Patients, families, doctors wrestle with privacy issues

By Richard Willing
USA TODAY

Cover story

When a test to predict the risk of getting breast cancer became available four years ago, Liz Stierman was quick to get in line.

The Los Angeles woman was almost 40 — prime age for developing the deadly disease — and her family had a history of cancer. As a genetics counselor, she knew how the test worked: It searched DNA in her blood cells for an inherited mutation believed to help trigger the disease.

Stierman's doctor urged her to be tested. Her younger sister offered support. Her insurer pitched in, promising to pay the $2,500-plus bill. But a key person objected: Stierman's 12-year-old daughter, Claire Smith.

"Claire knew that if I tested positive (for the mutation), there would be a 50% chance she carried it, too," Stierman says. "Her thinking was, 'Mom, please don't do it. I don't want to be a teenager with that hanging over me.' "

Stierman's case offers a glimpse of the complex, deeply personal issues that families across the USA are confronting because of gene testing, which is beginning to live up to its promise of revolutionizing preventive medicine.

U.S. labs now do more than 200,000 gene tests a year, compared with just a few a decade ago. The explosion in gene testing is allowing patients to learn whether they are likely, or even certain, to contract breast, colon or ovarian cancer or hundreds of other serious medical conditions. Knowing such information far in advance allows patients to take frequent diagnostic tests, alter their diet and lifestyle and even make themselves available for experimental treatments.

But because gene tests track hereditary conditions, one patient's test results can bring devastating news to his or her entire family. That, medical professionals say, is raising unprecedented ethical and legal issues that are roiling

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By Bob Riha Jr., USA TODAY

"Learn as you go": Claire Smith, 12, did not want her mother, Liz Stierman, to be tested for cancer.

Gene tests pit patients' rights against relatives' rights to know — or not

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families, creating lawsuits against doctors and genetics counselors and forcing those professionals to make agonizing choices.

Although much of the national debate over gene tests has focused on their potential for abuse by employers, insurers and the government, health and legal analysts say the issues raised by cases such as Stierman's are at least as poignant. Patients who agree to gene tests must decide not only how much they want to know, but also whether to tell their relatives.

Their doctors and counselors, meanwhile, struggle with when and whether to break the hallowed rule of confidentiality, and inform a patient's relatives that they might be at risk. The law is unsettled, but some courts have leaned toward requiring professionals to disclose test results to a patient's relatives.

"The headlines (about genetics) are always about privacy or cloning or something wild like that," says Nathaniel Robin, a doctor and geneticist at University Hospital in Cleveland. "But in the real world, (gene testing) is where the nitty-gritty issues are being worked out. And so far, the technology is running miles ahead of the answers."

Rise in gene testing

DNA, a cell acid that a person inherits from both parents, is carried on microscopic packets called genes. The DNA forms body proteins that create most of a person's unique characteristics.

Occasionally, DNA sequences mutate. That fouls up the genetic code and creates diseases. These diseases can be passed from parent to child. Both parents have to pass on a copy of a bad gene to cause some diseases. Others only take one. Making matters more complicated: Some genetic diseases aren't fatal. With others, such as Huntington's, inheriting a single copy of the mutant gene is a death sentence.

Cover story

Scientists used to use family studies and other approximations to try to determine whether a person had inherited a mutant gene. But in 1990, the federally funded Human Genome Project began charting the exact sequence of DNA in each of the body's estimated 30,000-plus genes. This has enabled genetics researchers to see exactly where disease mutations have occurred, and to develop tests to find them.

Advances in testing have moved with lightning speed. Katherine Schneider, senior genetics counselor at the Dana Farber Cancer Institute in Boston, recalls that one disease gene had been sequenced in 1992, the year her career began.

Now tests for nearly 400 gene mutations are available, and more tests are being added constantly, says GeneTests, a Seattle consortium that tracks testing. Genzyme Genetics, a testing firm in Cambridge, Mass., has seen its work increase 40% a year since 1995. Myriad Genetics of Salt Lake City performed about 250 breast cancer tests in 1997. The lab is on track to perform 10,000 this year, spokesman Bill Hockett says.

Even so, Hockett says, "only a minority" of women at risk (for breast cancer) are being tested. Some, he says, fear employment or insurance discrimination if they test positive, even though insurers are barred by lab policy from learning test results.

"And some people," he says, "just don't want to know."

Clinic offers mutation-free embryos

A major reason that adults at risk for genetic diseases undergo testing is to ensure that disease-causing genes would not be passed along to any children they might have.

But those at risk for a fatal disease — such as Huntington's, which shuts down brain functions and is always fatal — often don't want to know whether they carry the mutation themselves. To avoid facing such devastating news, many simply choose to not have kids.

Now, a fertility clinic in Fairfax, Va., is providing a way for such people at risk for Huntington's disease to have children who are free of the disease-causing gene in a procedure that allows the would-be parents to avoid learning whether they have the mutated gene themselves.

The procedure done by Genetics and IVF Institute combines genetic testing of microscopic embryos with artificial implantation. The firm guarantees that parents at risk for Huntington's disease can produce and implant mutation-free embryos.

The clinic uses an at-risk couple's sperm and eggs to create embryos in the laboratory. When the embryos are 72 hours old, one cell of the eight-cell creature is extracted and its DNA searched for the Huntington's mutation. Only embryos that are free of the disease gene are implanted. The parents are not informed whether they carry the fatal mutation. Herman Stern, the clinic's director of genetics, says he has performed the procedure 15 times since 1996, leading to five pregnancies.

A drawback: At-risk patients who don't carry the Huntington's gene are never told that they are mutation-free. So if they want to have more kids, they're likely to return to the clinic for another anonymous and costly ($15,000-plus) procedure.

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"On alert": Liz Stierman, with Claire, doesn't regret being tested.

tington's, and that put her at 50% risk of carrying the gene.

"They knew that if I tested positive, they were saying that my mother was positive, too," says Wagner, 36. "They didn't want (to give) bad news to someone who wasn't their patient."

Wagner tested negative for the Huntington's mutation. Her mother, however, tested positive. That put Wagner's older sister and younger brother on the spot. Her sister chose to be tested, and was found to also carry the gene.

Her brother continues to resist testing. "It's a sore spot, so we don't bring it up much," Wagner says.

For doctors and counselors who give gene tests without trying to force communication between patients and their relatives, relying on patients to spread the news about test results doesn't always work.

In Los Angeles, a man who tested positive for a gene that causes a rare brain disease refused to tell his brothers, who also were at risk. Counselors say the man fled the testing clinic, ashamed at the news, rather than face his relatives.

Medical professionals say that courts have offered them little guidance on their responsibility to tell a patient's relatives that they are at risk of inheriting a disease. "In the law, it's (not) ... a question of whether there is a duty to warn of a potential condition, (but) a question of who that duty is to," says Marcia Weiss, a specialist in genetics law at Point Park College in Pittsburgh. "This is very much a work in progress."

In 1995, Florida's Supreme Court ruled that requiring a doctor to advise the children of a woman with inherited cancer would "place too heavy a burden on a physician." But in 1996, New Jersey's Supreme Court said doctors should take "reasonable steps" to advise "those likely to be affected" by a diagnosis of inheritable disease.

Both rulings were in response to lawsuits filed by people who got hereditary forms of cancer after their parents had. The lawsuits said the parents' doctors should have told family members they also were at risk for getting cancer.

Doctors and genetics counselors say they especially fear the potential impact of a more recent case.

In California three years ago, the parents of a baby born with Prader-Willi syndrome, a rare hereditary developmental disease, sued because their genetics counselor had not suggested testing for the condition before the child was born. The parents were offered tests for more common fetal conditions, such as Down's syndrome.

"This really sends a chill down the spine," says Sue Schmerler, a lawyer and genetics counselor at St. Joseph's Hospital in Paterson, N.J. "There was nothing to indicate that the child was at risk (for the rare disease), but since the test was available, they sued anyway."

The case was settled before trial. The terms were not made public.

The National Society of Genetics Counselors has issued guidelines under which a patient's relatives can be told of gene test results even if the patient objects. The guidelines require that the risk of disease to the relatives be "high" and "imminent" to override a patient's privacy rights. The guidelines have not been tested in court.

'I don't regret any of it'

For patients such as Liz Stierman, deciding whether to be tested — and whether to tell family members the results — was a "learn as you go" process.

After talking it through with her daughter, Stierman was tested for a common breast cancer mutation. The results were negative, meaning that daughter Claire doesn't carry the mutation, either. But then Stierman wound up getting another form of breast cancer. She had surgery, and now is in remission.

"I don't regret any of it," Stierman says. "Claire and I are waiting for them to discover whatever cancer gene I'm carrying. In the meantime, we are both on alert."

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Seeking 'a sense of peace'

The decision whether to seek testing often depends on what disease a patient is at risk for.

For Dixie King, taking a test for a mutation that greatly increases the likelihood of breast cancer was a no-brainer. King was 38, the daughter of a breast cancer survivor and by background a European Jew, a group more likely to carry the gene.

King tested positive in 1996 and opted to have her breasts and ovaries removed – radical procedures that greatly reduced her chances of getting cancer.

The testing "gave me a sense of peace," says King, of Oxford, Md. "It's like I traded in a car whose brakes were estimated to fail at some point about 85% of the time. I know there's still a small risk of an accident, (but) I can live with it."

For Stephanie Vogt, the question was whether to be tested for Huntington's disease, which shuts down brain functions and is caused by a single mutation.

The Kalamazoo, Mich., woman was at risk: Her father's father had died of the disease, which can begin in a patient's thirties and is always fatal. But her father had died in his early thirties without knowing whether he carried the gene. When she was younger, Vogt says, she had her "head in the sand" about what might be coming.

Only about 300 Americans take the Huntington's test each year, says the Huntington's Disease Society of America, which estimates that more than 200,000 people in the USA are at risk for the disease.

As she neared 30, Vogt decided that continuing not to know would be worse than hearing bad news.

"I figured if I was gene-positive, knowing or not knowing wouldn't make a difference – I was going to get (Huntington's) anyway," says Vogt, 29. "But I have things I wanted to get done in my life. If I was only going to have so much time, I was determined not to waste it."

Vogt tested positive last year – meaning she is destined to get the disease – but has "not looked back since."

The claims adjuster is going to college at night, gives lectures on Huntington's disease and also works as a radio newscaster.

Not all patients who take the Huntington's test react so well, even when the results are negative.

In Seattle, a man in his late thirties continued to tell friends he carried the Huntington's mutation three years after testing negative.

The man, says genetics counselor Robin Bennett, had led a partying lifestyle based on the assumption that he would die young.

Admitting that he did not carry the mutation would have forced him, in essence, to adjust his values, Bennett says.

In Pennsylvania, a woman who tested negative for the Huntington's mutation suddenly felt ashamed to be with her sister, who had tested positive.

"Survivor guilt is not uncommon," says Vivian Weinblatt, president of the National Society of Genetics Counselors.

The dilemma for doctors

Gene tests are creating challenges for doctors and genetics counselors, who are sworn to respect patients' privacy. Sometimes, medical professionals have patients who have tested positive for a gene mutation, but who do not want their relatives to know – even though the result means the relatives are at risk, too.

"It can be an excruciating problem," says Wendy Uhlmann, a counselor at the University of Michigan hospital. "Patient confidentiality is paramount. (But) how can you in good conscience leave somebody else at risk?"

To avoid being placed in such positions, some medical professionals have refused to give gene tests unless patients agree to share the results with their relatives. Last year, Michelle Wagner of Mesa, Ariz., had difficulty finding a lab to test her for Huntington's disease unless her mother agreed to be tested at the same time. Wagner's mother had lost a parent to Hun-

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