

# With genetic testing, patients can see the future

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Denis Keegan was out of answers. The 30-year-old was suffering from kidney disease, but his doctors were struggling to pinpoint the cause. That's when Keegan turned to genetic testing.

Doctors at the Mayo Clinic in Rochester, Minn., extracted his DNA from a blood sample and examined his genome. There, embedded in his genetic code, they discovered the source of his kidney problems - a [mutant gene](#). The finding led them, at last, to a diagnosis: fibronectin glomerulopathy, an extremely rare kidney disorder. Armed with that knowledge, his doctors were able to tailor treatment for the condition.

"It was really reassuring," said Keegan, 30.

The human genome was mapped in 2003, revealing for the first time the entire genetic makeup of our bodies. Since then, genetic testing has become a booming industry - and an option for patients, such as Keegan, to learn more about their bodies' internal mysteries.

Advancements in the testing are coming rapidly, slashing the price and time it takes to get results. Just as the X-ray machine made it possible to peer inside the human body, genetic testing is changing the way we diagnose and treat diseases.

Testing is available through your doctor, or increasingly via direct-to-consumer kits that can be ordered online or purchased at a drugstore. By the end of the decade, Americans are expected to spend as much as \$25

billion a year on genetic tests for everything from diagnosing types of kidney diseases to determining [breast cancer](#) risk to screening prenatal health, according to United Health.

But for some patients, genetic testing represents a Pandora's box that, if opened, could cause needless anxiety among healthy people or sow discord in families if one member's test reveals troubling findings about the family's genetic makeup.

"In terms of how we apply it to our health, that's where the door has just been opened," said Melissa Truelson, a certified genetic counselor at the University of Minnesota Medical Center, Fairview.

To help navigate this brave new world, we turned to those on the front lines of this fast-evolving medical technology:

## **What is genetic testing?**

Genetic testing typically involves looking at a person's genes or chromosomes to help prevent, diagnose or treat a disease. It also can be used to determine whether a person is a carrier of a genetic disease and if there is a risk of passing it on to their children. Most often, the testing begins with DNA collected from a [blood sample](#), but many labs also can get DNA from a person's saliva.

Genetic testing jumped into the spotlight last year when Angelina Jolie, whose genetic test revealed a high risk of developing breast cancer, chose to undergo a double mastectomy in the hopes of preventing the disease.

"Everything our body does functions off of genes," Truelson explained. "When you think about diseases, many of them have an underlying genetic or inherited basis to them."

## How much does it cost, and does insurance cover it?

The cost varies widely, from \$99 at-home kits (such as the controversial 23andMe) to thousands of dollars for tests conducted at hospitals and clinics. Pricing also varies depending on what you are testing for, such as a newborn screening or colon cancer.

Insurance coverage also is dictated by the kind of testing and the individual's plan. Typically, insurance companies will cover tests that are ordered by doctors and usually involve diagnosing a disease. Patients who have a significant family history with a particular disease tend to receive coverage.

Direct-to-consumer genetic test kits are not covered. Hundreds of thousands of people have used 23andMe, which analyzes a segment of a person's DNA sequence for genetic alterations linked to cystic fibrosis, sickle cell anemia and even breast and [ovarian cancer](#). But critics - including the U.S. Food and Drug Administration - say the test is limited in its ability to diagnose health risks accurately and could lead users to undergo unnecessary medical procedures. FDA officials recently ordered 23andMe to halt marketing its DNA test kits.

## What can a test reveal?

For someone with a defined disease, the use of genetic testing can be very effective in determining what medicines work best in treatment, doctors say.

A 44-year-old woman with gall bladder cancer is among the success stories at the Mayo Clinic's Center for Individualized Medicine, said Dr. Alexander Parker, the center's associate director. Her tumor was not responding to the standard medicine used to treat gall bladder cancer.

Through genetic testing, doctors discovered that drugs used on leukemia patients might work for her. They tried it, and her tumor started to shrink, Parker said.

But for healthy people, whom he calls the "worried well," there is little to no value in having your genome mapped. It may cause harm by raising anxiety about the odds of developing a disease.

"At the end of the day, this is about risk," Parker said. "While we all want definitive answers to everything, the beautiful thing about our world is that there is random chance. Because of that, we can never say 100 percent that we know exactly what will happen to anyone."

## **What factors should go into your decision to get a test?**

Parker encourages those curious about genetic testing to have a conversation with their primary doctor first. Ask: "Is there any value in this for me?"

Genetic counselors also play a key role in helping reach a decision. They typically hold master of science degrees in programs that include training in medical genetics and counseling. They work in consultation with doctors, nurses and other health professionals.

Truelson said she encourages patients to ask these questions: What type of information will the test give me? How will this affect me from a medical standpoint? How will this affect me from an emotional standpoint?

Nada Maalouli, of Eagan, Minn., has wrestled with these questions for years. Her mother died of ovarian cancer before she reached menopause.

Her grandmother and an aunt on her mother's side died of breast cancer. Maalouli worries she may be next. A genetic test would tell her if she carries a mutant gene associated with higher risks of breast and ovarian cancer.

She has agonized over the decision to get tested, in part because she fears the test for breast cancer would be positive.

"I don't know if I can live with that," she said. "I feel like maybe it will (make) me depressed."

She has visited a genetic counselor three times in the past 10 years to weigh the pros and cons. At 51, she and her three sisters - all healthy so far - have made a pact. They will get regular mammograms and MRIs, and if any of them detects that there is something wrong, then the sisters will all go in for [genetic testing](#).

## **How will testing affect your family?**

One person's choice to get tested may also reveal health information about your relatives, who may prefer not to know, Truelson said.

For Keegan, who visited Mayo from Ohio, the decision to have his genome mapped affected several members of his family. Mayo doctors asked his older brother and father also to submit a DNA sample to find out if Keegan had inherited his father's disease. Keegan's father previously had a kidney transplant.

In addition, Keegan and his wife were thinking about starting a family and wanted to know what their chances would be of passing on the mutant gene. Turns out there is a 50 percent chance their offspring will inherit it. For the couple, the test results have led to many discussions about having children.

They talked about possibly taking a fertilized embryo and checking it to see if it has the mutated gene before deciding to continue with the pregnancy. "We stepped back, and thought, 'We're kind of playing God here,'" Keegan said. "We didn't think we wanted to do that."

In the end, they've decided that when the time comes, they'll roll the dice and have children the old-fashioned way.

Even if he passes the gene onto his child, there's a good chance that during the child's lifetime there will be medical advancements to improve treatment of the disease, Keegan figures.

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