THE ABC's OF DNA

IS GENETIC TESTING A PATH TO WELLNESS—OR MORE INFORMATION THAN YOU WANT TO KNOW?

BY SUSAN R. MILLER

WHAT IF YOU HAD A CRYSTAL BALL AND COULD SEE THE FUTURE? And what if it told you that in a few years you might be diagnosed with breast cancer, diabetes, Alzheimer's, Parkinson's or another debilitating or life-ending disease?

Would you want to know?

Looking into the future and predicting the chances of developing a disease is not the work of crystal balls and hocus-pocus. It is the work of scientists who every day are developing better, cheaper and more accessible means of conducting genetic testing that can lead to earlier diagnosis, better treatments and, most importantly, saving lives.

While there are clear advantages to knowing your genetic makeup and whether you're predisposed to developing a disease, critics worry that in some cases it might open a Pandora's Box of confusion, anxiety, privacy issues, discrimination by insurance companies and even lead to unnecessary treatment. Worse yet, they argue, those found to be at a higher risk of developing a disease for which there is no cure would likely suffer vast emotional consequences leading scientists to ponder: Just how much information is too much?

SCIENTIFIC ADVANCES

DNA testing has been around since the mid-1980s, emerging as a way to use biological material—skin, hair, blood and other bodily fluids—to link criminals to their crimes.

For many years, mothers-to-be have undergone tests to determine whether their babies might be born with Down Syndrome, spina bifida or cystic fibrosis. Others have undergone pre-pregnancy testing to determine if they are carriers for a genetic condition that can be passed on to a child should they become pregnant.

Genetic testing also has been used to trace genealogy, helping adopted children locate parents and other family members and to learn more about their family medical history.

But it's only been in the last decade or so that scientists—with the unraveling of how the human genome operates—have been able to conduct the kind of gene sequencing that allows them to identify gene mutations that cause illness. Tests are various and can cost anywhere from around $100 to several thousand dollars, depending on how in-depth they are.

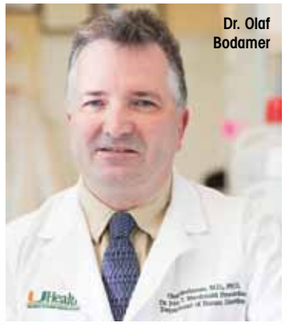
And as this paradigm shift empowers patients by providing them with information about their own medical makeup, questions are also being raised about validity and accuracy.

Case in point: Mountain View, Calif.-based 23andMe, which offers a genetic test for just $99, has caught the attention of the U.S. Food and Drug Administration. The test called for customers to mail a saliva sample in a vial, and in return receive about 250 health-related reports and information about their genetic history.

However, last November the FDA sent a warning letter to 23andMe demanding it cease selling its product noting that the company had not provided enough evidence that its tests were accurate, and raised concerns that patients were receiving genetic information without medical guidance. The FDA noted that products designed to diagnose, mitigate or prevent disease were medical devices that required its approval.

At press time, 23andMe agreed to stop providing consumers with access to its health-related genetic tests while the FDA completed its review. However, the company said it would continue to provide both ancestry-related information and raw genetic data without interpretation.

"We remain firmly committed to fulfilling our long-term mission to help people everywhere have access to their own genetic data and have the ability to use that information to improve their lives," Anne Wojcicki, co-founder and CEO of 23andMe, said in a press release. "Our goal is to work cooperatively with the FDA to provide that opportunity in a way that clearly demonstrates the benefit to people and the validity of the science that underlies the test."


Dr. Olaf Bodamer


Amy Byer Shainman and husband Jon

Amy Byer Shainman photo by Debby Gans Photography


Dr. Louise Morrell

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– Dr. Olaf Bodamer, chief of the Division of Clinical and Translational Genetics in the Dr. John T. Macdonald Foundation Department of Human Genetics, University of Miami Miller School of Medicine

The company isn't the first of its kind to go up against the FDA. In 2010, San Diego, Calif.-based Pathway Genomics announced plans to offer personal genetic tests through Walgreens. When the FDA got wind of its plans, Walgreens backed out of the deal. The company still offers genetic testing, but tests must be ordered through, and results delivered to, a qualified healthcare provider or genetic counselor.

According to the company's website, its tests cover a variety of conditions including cardiac health, cancer risk and inherited diseases. It also examines genetic markers known to impact metabolism and exercise, and provides strategies for reaching a healthy weight based on a patient's specific genetics. Further, it's said to test for drug responses to specific medications, including those used in pain management and mental health.

The company did not return phone calls for comment.

As the science continues to develop, experts urge caution.

"We have to be extremely careful. If genetic information becomes readily available to everybody, we have to make sure there is a level of knowledge and understanding and interpretation going along with these test results. Otherwise, I see people totally confused. It will be a nightmare," says Dr. Olaf Bodamer, chief of the Division of Clinical and Translational Genetics in the Dr. John T. Macdonald Foundation Department of Human Genetics at the University of Miami Miller School of Medicine.

WORD SPREADS

Angelina Jolie raised global awareness of genetic testing when she revealed her decision to have a double mastectomy after learning she had a gene mutation that placed her at high risk for developing breast cancer.

Since then, thousands of women around the country have sought similar tests for the BRCA1 and BRCA2 gene mutation that causes breast and ovarian cancer. Several South Florida hospitals that conduct genetic testing saw a spike in requests for the test.

"We were shocked at how unaware people were (before)," says Dr. Louise Morrell, medical director of the Lynn Cancer Institute at Boca Raton Regional Hospital. In February 2013, the hospital opened the Morgan Pressel Center for Cancer Genetics. It is named after the LPGA player whose mother died of breast cancer. What also surprised her was that the patients calling and coming in had done their homework and actually were at risk.

Dr. Bodamer saw a similar jump in requests. He says that while Jolie's case helped to raise awareness, it also caused confusion.

"Breast cancer is a complex disease and while genetics is a factor, in only a minority of cases does genetics play a predominant role," he says. "I think there was a lot of unnecessary activity due to the understandable anxiety of these women and the confusion over what role genetic testing plays in breast cancer."

Testing is the easy part, says Dr. Elizabeth McKeen, medical director of the Cancer Risk Assessment and Genetics Program at the Margaret W. Niedland Breast Center at Jupiter Medical Center.

"Identifying the people who should be tested and coming up with an individual plan for each patient is the hardest part," Dr. McKeen says. "That's why you want to go to an expert in this area of testing."

At Jupiter Medical Center's Cancer Risk Assessment and Genetics Program at the Margaret W. Niedland Breast Center, every patient who comes in for breast imaging is offered a cancer risk assessment. Questions relating to family and personal history help to determine if genetic testing is appropriate, says Conni Murphy, an advanced registered nurse practitioner. "A majority of people we test who are high risk—that is greater than 20 percent—are BRCA normal," Murphy explains.

This means that while they don't have the mutated gene, they still are at risk for developing breast cancer.

"These people are offered strategies to reduce their risk such as increased screenings and (preventive) medicines like Tamoxifen," Murphy says.

Faced with a family history of breast, ovarian and uterine cancers, Amy Byer Shainman of Jupiter underwent genetic testing in 2009 after seeing her sister battle ovarian cancer. At age 40, Shainman came up positive for the breast cancer gene mutation.

Her doctor laid out her choices: She could have preventive surgery, which meant having her breasts removed, as well as her ovaries, or her entire uterus. She could start taking Tamoxifen, or she could undergo enhanced surveillance and screenings on a regular basis for early detection.

"I felt like I had been run over by a truck. It's very overwhelming," she says. "I drank a lot of wine and ate a lot of chocolate," she jokes about how stressed she was at the time.

In March 2010, at 41, the mother of two decided to undergo surgery—first a hysterectomy. Six months later, she underwent a double mastectomy.

"It took longer for me to wrap my head around the mastectomy thing and I needed to do more research," says Shainman, now an outspoken proponent for genetic testing. She is outreach coordinator for the Palm Beach County Chapter of the national organization Facing Our Risk of Cancer Empowered (FORCE).

Shainman recently decided to test the validity of the 23andMe test to see if it confirmed her gene mutation. The results were accurate, she says. But does she recommend it for others?

"No," she says. "The information can be confusing. People think just because their BRCA test comes back negative they are not at risk. It gives you a false sense of security."

CALCULATING RISKS

While genetic testing for breast cancer is making headlines, there are numerous other tests available for a number of other conditions including age-related macular degeneration, Alzheimer's, heart disease and colorectal cancer, to name a few.

"We do colonoscopies by the recommended age of 50, but this is a cancer that can occur in a 25- or 30-year-old that you would never think to screen," says Dr. Morrell. "That's a whole area where we think it's important to do gene testing and we are expanding to get the word out and get people tested because it can make a big difference in survival."

As more research is conducted, and more tests become readily available, the costs have come down.


Dr. Elizabeth McKeen


Conni Murphy


Julie Matuzak and daughter Allie

The University of Miami's Dr. Bodamer points out that 10 years ago it took a consortium of four different laboratories two to three years to sequence a single human genome.

"With the equipment we have in our laboratory we can do it in 60 hours, so we have come a long way," he says. "And when you look at the cost 10 years ago we were talking about doing one human genome for $10 million to $20 million. Today, we can do it for $10,000."

Which brings us back to companies that provide genetic tests. Julie Matuzak adopted Allie from China nearly 17 years ago. The Detroit woman says her daughter became interested in the possibility of finding relatives or even her birth parents ever since learning about DNA in school. She also hoped to learn more about her medical history.

"She hates going to the doctor (because) when they ask her if she has a family history of something she has to say 'I don't know,'" Matuzak says.

Last summer, both decided to use 23andMe. Matuzak chose to look at the results that might have proved troublesome, such as whether she might carry the breast cancer gene mutation. "It caused me to speak to my retirement counselor again. I may be here a long time," she says with a chuckle.

While there have been several stories of children adopted from China finding other family members outside of China, Allie is not among them, at least not now. "I think she was a little disappointed, but I don't think there were great expectations," Matuzak says. "I think she sees it as a possibility for the future as more people register and more people get tested."

Since learning that 23andMe is no longer marketing the test, Matuzak says she hasn't changed her mind about the benefits.

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"The important function of the DNA test for us was for my daughter to be able to put herself out there on a DNA registry in the hope that someday she could find a birth relative," she points out. "That desire hasn't changed."

However, she adds: "I do appreciate the FDA figuring out about regulations to cover these kinds of new companies. If people were doing this primarily for the medical info I would want someone to be regulating and overseeing them to ensure accuracy." ○