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You've Had a Genetic Test. Now What?

A new project aims to incorporate the results of genetic screening into medicine.

By Emily Singer

Tests that look for genetic variations linked to a number of common diseases are now available over the Internet.



A study participant provides a saliva sample for DNA analysis. Credit: Coriell Institute

But a patient who walks into the doctor's office with a thick file of genetic information will probably find that the doctor has no idea what to do with it. A new project, launched in December by the Coriell Institute for Medical Research, in Camden, NJ, aims to change that.

The institute will offer free genetic testing to 10,000 local volunteers over the next two years. Participants will get information on their genetic risk for a number of diseases and a way to share that information with their physicians. The program will also help both

physicians and patients learn what to do with genetic information once they have it. Researchers will examine every step of the process to figure out how physicians can best incorporate genetic information into their practice to prevent and treat disease.

"Even though what's medically relevant is limited, the technology to do these studies is exploding," says Michael Christman, president and CEO of the institute. "It's early days, but we need to start thinking about this right now."

In the past two years, scientists have published a flood of studies linking specific genetic variations to heightened risks of contracting disease, such as diabetes or cardiovascular disease. The findings are becoming commercialized almost as quickly as they're announced. In theory, awareness of risk can help patients take measures to prevent disease onset. Those at higher risk of breast cancer might receive more frequent screening, for instance, or those at risk of cardiovascular disease could start taking statins.

But the very speed with which the tests have reached the market--and the fact that they are available online--has raised a number of concerns. Can consumers understand

the results without the help of professionals? Can their physicians? And most important, can the information truly improve patients' long-term health? "We need to evaluate the relevance of these genes in terms of their disease risk and clinical utility," said Ulrich Broeckel, director of the Individualized Medicine Institute at the Medical College of Wisconsin, in Milwaukee, at the Beyond Genome conference in San Francisco on Wednesday.

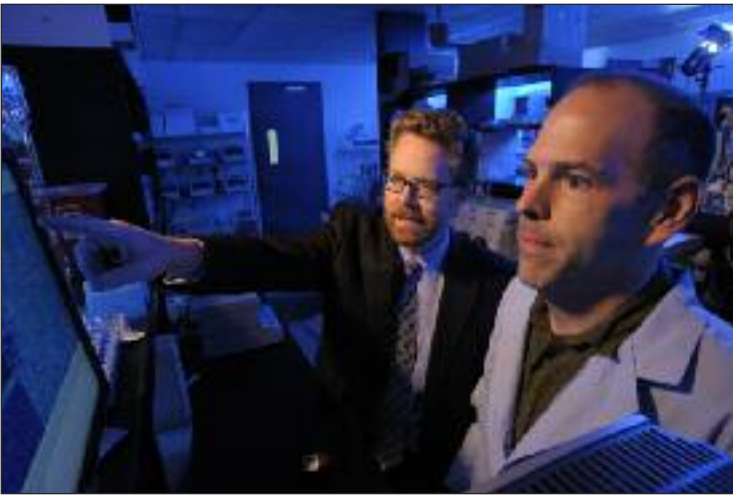
The Coriell Personalized Medicine Collaborative will offer services similar to those provided by private companies, such as Navigenics, DeCode, and 23andMe. All three of those companies use microarrays--chips studded with fragments of DNA--to screen DNA for specific genetic variations. But the Coriell project will offer a narrower range of results, and it will make a direct effort to involve medical professionals. "We want to build a model of an ethical, responsible, genome-informed medical practice," says Christman. "This is important because it's not being taken on by any government agencies." (Currently, few genetic tests are regulated by the federal government.)

While final details of the project have not yet been determined, it's likely that participants will initially be offered risk information on 11 different conditions, including breast and prostate cancer, type 2 diabetes, cardiovascular disease, and macular degeneration.

Coriell is also building a Web portal, due to launch in September, where participants can access their genetic-risk information and elect to share some or all of it with their doctors. They will also fill out regular health surveys to allow scientists to assess how useful this genetic information is in improving long-term health: lack of information on clinical utility has been a major criticism of direct-to-consumer genetic screening. The portal will also feature educational material targeted to



Researchers in Coriell's Genotyping and Microarray Center process DNA samples to determine individual genome profiles. Credit: Coriell Institute



Dr. Michael F. Christman and Dr. Norman P. Gerry discuss data analysis for correlating genetic variation with disease risk. *Credit: Coriell Institute*

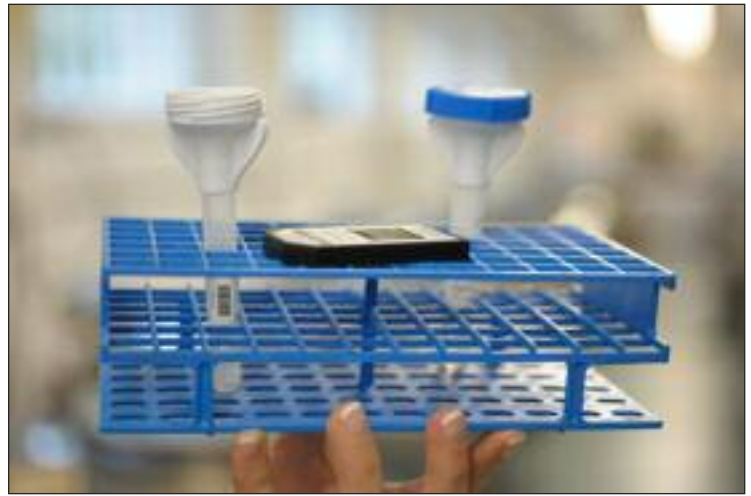
physicians, including continuing-medical-education courses.

When the site launches, will physicians know what to do with the genetic information their patients provide them with? “Absolutely not,” says Christman. “But it’s where we have to start. We can’t wait to train a new generation of doctors.”

Preliminary surveys that Coriell has conducted suggest that patients are also confused by genetics. “I’m alarmed by the lack of understanding,” says Christman. “People don’t understand the difference between Mendelian and complex genetic diseases.” In a Mendelian disease--named for the 19th-century priest Gregor Mendel, who did pioneering genetic research on fruit flies--the disease is linked to a single gene. Carrying certain variants of that gene virtually guarantees the eventual onset of the disease. With complex diseases, however, multiple genes interact with the environment to increase risk. Carrying any particular gene does not guarantee disease onset. “I don’t want people going home and saying, ‘I have the heart-attack gene,’” says Christman.

Unlike some private services, the Coriell project will give participants only that genetic information deemed by an advisory board to be medically actionable, meaning that there are medical or lifestyle interventions that can reduce risk of the disease or improve outcomes. (Of course, the definition of “medically actionable” can vary. Navigenics also limits testing to disorders for which interventions exist. But it includes Alzheimer’s disease on its list. Christman says that Coriell does not, since no interventions have been proved to have a significant impact on Alzheimer’s risk.) An independent review board will meet twice a year to discuss the latest research and decide which variants make the list.

As part of their efforts to engage the medical community, Christman and his team have reached out to local hospitals, which are offering the genetic-screening service to their staff, both as an individual benefit and as a way to learn about genomics. “The project is extremely important because it will add a dimension to patient care we haven’t had before--the genetic component,” says Simon Samaha, chief medical officer at Cooper University Hospital, in Camden, one of the participating organizations. Coriell has enrolled 2,000 participants to



A saliva collection kit and an Affymetrix GeneChip are used to collect DNA samples and to look for sites of genetic variation. *Credit: Coriell Institute*

date, about half of them medical professionals, and it aims to have 10,000 volunteers by 2009. Currently, only people who live locally, and thus can give consent to participate in the project in person, are eligible.

Because participating is free, Christman says, the project will reach a broader cross section of the population than do private companies, which charge \$1,000 to \$2,500 for their services. Scientists at Coriell have directly reached out to both African-American and Hispanic communities.

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Upcoming Events

EmTech08

MIT Campus, Cambridge, MA

Tuesday, Sept. 23, 2008 - Thursday, Sept. 25, 2008

<http://www.technologyreview.com/emtech/08/>

MIT Professional Institute

Cambridge, MA

Monday, June 09, 2008 - Monday, August 04, 2008

http://web.mit.edu/mitpep/pi/courses_topic.html?c1=banner&source=pi+TRevents

EurekaFest

Cambridge, MA

Wednesday, June 25, 2008 - Saturday, June 28, 2008

<http://mit.edu/invent/eurekafest.html>