

Research pictures the ultimate portrait in personalized medicine

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by **Shannon Bond**

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How much would you pay?

For people who don't want to wait—and who can afford it—some companies now offer full genome sequencing.

Cambridge, Mass.-based Knome Inc. charges \$350,000 to map an entire genome. Customers receive a copy of their sequenced genome, a report analyzing the results and access to counseling and interpretation services, said Knome CEO Jorge Conde.

“The cost of sequencing today is the primary cost in providing these services,” Conde said, and the high price tag limits the company's clientele.

“We would welcome technological advancements that would lower costs,” he said, and thereby attract more customers.

Is more testing good for us?

While genetic testing is an important part of modern medicine, “it's not necessarily all in the genes,” said Julie Burger, assistant director of the Institute for Science, Law & Technology at IIT's Chicago-Kent College of Law.

“People might not be prepared for the results that they're going to receive. They might not know how to interpret their results,” she said.

Burger said it's important to remember that just because you have a genetic mutation that increases your chances of developing breast cancer, that doesn't mean you are going to get sick. But it may change how you view your body and how you are viewed by insurers.

“There are a lot of psychological and health issues that come into play,” Burger said, so taking advantage of genetic counseling is important.

The wider availability of testing does not guarantee that patients will use genetic information wisely, she cautioned. “Are people really going to be using the results to improve their lives or better protect their health?”

The goal is to develop a device that laboratories and doctor's offices can use to quickly screen a patient's complete genetic makeup.

With this information, experts say, physicians could personalize treatment and prevention plans for each patient.

Researchers at the University of Illinois at Urbana-Champaign are taking the first step by streamlining DNA sequencing to rapidly map your entire genome for only \$1,000.

That's a far cry from the \$3 billion budget of the Human Genome Project, which took 13 years to fully sequence human DNA.

At present it can cost \$1,000 and more to screen for a single gene, and one company charges consumers \$350,000 for complete DNA sequencing.

The Illinois scientists hope to slash that price tag through a technique called nanopore sequencing.

The method forces DNA molecules to pass back and forth through a tiny hole, or nanopore, in a silicon chip. This produces an electrical field that can be read to decode the unique sequence of chemical "letters" that make up an individual's DNA.

Computer simulations showed the technique could work, physics professor Aleksei Aksimentiev said. His collaborator Gregory Timp, a professor of electrical engineering, is building a prototype that would put the theory into practice.

While they have a prototype model, they are still refining development for a marketable device.

"We are doing it because we would like to eventually get to a device that would perform DNA sequencing very fast and affordably," Aksimentiev said. The method could cut costs because silicon chips are inexpensive to produce, he said.

The project is funded by a \$2.1 million grant from the National Human Genome Research Institute (NHGRI), part of the National Institutes of Health.

NHGRI funds research for technologies that reduce genome sequencing costs (which can be as high as \$5 million) to \$1,000 or less. "The ability to sequence an individual genome cost-effectively could enable health care professionals to tailor diagnosis, treatment and prevention to each person's unique genetic profile," NHGRI said in a statement on its web site.

While genetic testing is becoming increasingly common as scientists identify genes that may play a role in conditions such as breast cancer and heart disease, the high cost of screening makes routine genome sequencing impractical.

Genetic testing generally focuses on specific genes, not the entire genome, said Scott Weissman, a genetic counselor at Evanston Northwestern Healthcare's Center for Medical Genetics. Prices vary depending on the test, the size of the gene and whether the patient has a family history of disease, he said.

"Typically, it's more expensive to screen someone for the first time because we have to screen the entire gene," he said. If a patient's family member has already been diagnosed with a hereditary condition, only the part of the gene where that mutation occurs needs to be tested.

Weissman said single-gene tests cost an average of \$1,000, but some tests run as high as \$7,000. As technology improves, he said, those prices will come down—and the approach to health care will change.

"I think the ultimate goal of the Human Genome Project and all this genetic testing going on is really to change the paradigm for medicine" to a preventative model, Weissman said.

A doctor could use a patient's genetic profile to advise the patient on how to modify her diet and lifestyle, and prescribe drugs if necessary to prevent the onset of disease.

"We can prevent disease rather than treat it," he said.

