

## New era of gene-based 'personalized medicine' dawning

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Six years ago, scientists announced the completion of the Human Genome Project, a historic effort to decipher each of the 3 billion letters in the genetic instruction book for our species. A single anonymous male from Buffalo, N.Y. - code name RP11 - provided the bulk of the DNA used for the project.

Now, many thousands more people are contributing DNA samples for a wide array of follow-on studies designed to turn the project's findings to practical use in health care, genetics and biological research.

Researchers and doctors have opened a new era of "personalized medicine" that seeks to tailor therapies to patients based on their unique genetic makeups and medical histories.

According to the National Cancer Institute, the days are passing when most <u>cancer tumors</u> were thought to be essentially the same and patients got the same drugs.

"We're not very good at selecting therapies for individual patients," Dr. Rick Hockett, the chief medical officer of Affymetrix, a genetics firm in Santa Clara, Calif., told a conference on personalized medicine this month in Washington. "Targeted therapy," he said, can "improve the benefit-risk ratio for patients."

For example, Hockett said that heart patients who took the popular anticlotting drug Plavix had a greatly increased risk of serious problems,



including death, if they had two tiny mutations in their genes.

Massachusetts General Hospital in Boston and the Memorial Sloan-Kettering Cancer Center in New York have begun to screen lung tumors for genetic abnormalities that could reveal whether a particular treatment is likely to work or should be avoided.

Last year, Dr. Richard Wilson, the director of the Genome Sequencing Center at Washington University in St. Louis, compared 20,000 genes in cancer cells from a woman who died of leukemia with healthy cells also taken from her body. Wilson identified 10 mutations \_ or genetic mistakes \_ related to her cancer, including one that blocks <a href="mailto:chemotherapy">chemotherapy</a> drugs from getting inside the cancerous cell.

Other developments in the approaching world of personalized medicine include:

\_A "Personal Genome Project" led by George Church, a geneticist at Harvard Medical School in Boston, aims to recruit 100,000 people of diverse backgrounds to analyze their genomes and medical histories.

The first 10 participants already have published their personal data \_ including their pictures, weights and smoking and drinking habits, as well as their DNA \_ on the Internet for anyone to see at www.personalgenomes.org/pgp10.html.

To take part, volunteers donate hair and saliva samples for DNA analysis. So far, 13,000 people have asked to be enrolled. For privacy, their data will be encoded by number, not by individual names.

The goal is to discover which genetic variations are related to which diseases, so that targeted therapies can be designed. According to Church, at least 1,449 genes have been linked to potential illnesses. For



instance, a gene called ApoE is associated with a higher risk of Alzheimer's.

\_The National Human Genome Research Institute in Bethesda, Md., has launched a "1000 Genomes Project," which has started to collect DNA from 1,000 individuals from Africa, Asia and Europe. The goal is to provide much broader and deeper information than the original, limited Human Genome Project could.

Researchers are seeking clues to individual differences in susceptibility to disease, response to drugs and sensitivity to the environment.

"Just as astronomers see farther and more clearly into the universe with bigger telescopes, the results of the 1000 Genomes Project will give us greater resolution as we view our own genetic blueprint," Francis Collins, the former director of the National Human Genome Research Institute, said in a statement explaining the plan. "We'll be able to see more things more clearly than before, and that will be important for understanding the genetic contributions to health and illness."

\_The National Geographic Society is collecting DNA from about 300,000 people for a "Genographic Project" that traces their distant ancestries from continent to continent all the way back to their African roots.

Researchers at 10 centers around the world collect DNA samples from local populations. A team led by Spencer Wells, a genetic anthropologist and the magazine's "explorer in residence" in Washington, will analyze the samples.

In addition, anyone who wishes to can buy a \$100 self-testing kit and provide a saliva sample to the Genographic project. Participants will get back reports that describe in general terms the migration of their



ancestors from their original roots in Africa some 60,000 years ago.

\_A booming, but controversial, genetic testing industry also has sprouted, offering to analyze a person's DNA \_ at a price \_ for possible susceptibility to cancer, Alzheimer's and dozens of other diseases.

More than 1,000 such tests are on the market, Kathy Hudson, the director of the Genetics and Public Policy Center at Johns Hopkins University in Washington, told a congressional committee.

According to the Human Genome Project's Web site, some genetic tests have "greatly improved or even saved lives," but there are concerns that they might be used by insurance companies to deny coverage or by employers deciding whom to hire or promote.

To meet these concerns, Congress passed a "Genetic Information Non-Discrimination Act" last year, which is just starting to have an impact.

"The portions of it relating to health insurance just went into effect on May 21, and so will apply to next year's (health) plan for most people," said Susannah Baruch, of the Genetics and Public Policy Center. The parts of the new law that relate to the use of genetic information on the job won't take effect until Nov. 21, but the Equal Employment Opportunity Commission already has drafted regulations.

More on personalized medicine: www.personalizedmedicinecoalition.org

Personal Genome Project: www.personalgenomes.org

1000 Genomes Project: <a href="https://www.1000genomes.org/page.php">www.1000genomes.org/page.php</a>

National Geographic's Genographic Project: genographic.nationalgeographic.com



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