Landmark gene studies released

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The mapping of the human genome was one of the great moments in the history of science

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WASHINGTON -- Science passed another milestone on Monday with publication of the first description of the human genome, an advance likely to revolutionize the understanding and treatment of disease.

While acknowledging that the potential benefits of the genome map are staggering, experts at Monday's announcement agreed that significant breakthroughs in understanding and treating diseases are likely at least a decade away.

"Both of the sequences being presented today are still drafts," said Dr. Bob Waterston, head of the Washington University Genome Sequencing Center, who is helping to lead the Human Genome Project. "We are moving swiftly to complete the whole genome."

Dr. Francis Collins of the National Human Genome Institute in Washington said the studies were significant both for their discoveries and the speed by which scientists have unraveled at least part of the genetic mystery.

"These revelations arrive almost five years early from the original predictions of not having this information until 2005, and here we are. We have the first draft of our own book of life and we've read it from cover to cover, and we've discovered some pretty amazing surprises," Collins told CNN.

Scientists say they have learned that humans don't have as many genes as previously thought -- not all that many more than in a fruit fly.

The research also confirmed that males can take the blame -- or credit -- for creating most inherited genetic mutations.

Genes determine everything from eye and skin color to vulnerability to illness. Human have about 30,000 genes, the scientists found.

Few genetic differences between people

Scientists have also learned that the genetic differences between any two people are relatively small.

The analyses were performed by the two teams that made headlines last year for determining nearly all the "letters" of the human DNA code.

That three-billion-letter code, called the genome, is a chemical sequence that contains the basic information for building and running a human body.

"We suddenly have the global view, the view of the Earth from the moon, and it's pretty thrilling," said Dr. Harold Varmus, a former director of the National Institutes of Health who now heads the Memorial Sloan-Kettering Cancer Center in New York.

Celera Genomics, a private company based in Maryland, is publishing its findings in Science. A public international effort, led by the United States, is publishing its analysis of the genome in Nature, a British journal.

The two teams, which worked independently, estimated roughly the same number of human genes: About 26,000 to 39,000 according to Celera, and about 30,000 to 40,000 according to the consortium. Scientists with both groups said the best bet is something fewer than 35,000.

That's surprisingly low, leaders of both scientific teams said.

Cracking the genetic code could help scientists and doctors find disease and illness.

"I think it means that we'll be able to track down the actual causes of disease," said Eric Lander of the Whitehead Center for Genome Research. "Most folks don't realize we don't know the cause of asthma, of heart disease, of diabetes or hypertension."

Collins said it will take "a long time" to apply the information toward fighting -- even curing -- some diseases. "But we are that much closer having this foundaton now in front of us," he said.

'...a time to be very excited and optimistic...'

"I think it's a time to be very excited and optimistic," Collins said, predicting that science will zero in on the genetic factors involved in diabetes, heart disease and other common disorders "within the next five to 10 years."

Already, cancer drugs are targeting the disease at the molecular level, he noted.

On another front, Collins said federal legislation was needed to protect against the misuse of genetic information in the work place or by health insurance companies.

"We can see the path that needs to be traveled," he said. "Let's get on with it and put that protection where it needs to be so the public doesn't get injured by very valuable information that we all may wish to have."

"We need effective legislation to make those kinds of misuse illegal," Collins said, referring to a recent lawsuit over a private employer-sponsored genetic testing, apparently without employee knowledge or permission.

Dr. Craig Venter of Celera Genomics also urged passage of an anti-discrimination bill.

"If anything, we've learned that we don't think this data's going to be as deterministic as was previously thought," Venter said. "There is even more need for this legislation until science catches up with our collective ignorance."

Disease, addiction traced to DNA

Mapping the chemical sequences for human DNA -- the chemical "letters" that make up the recipe of human life -- is a breakthrough that scientists expect will lead to new cures for cancers, heart disease, drug addiction and mental illness.

One in three people in the western world will develop cancer, and one in five will die from the disease, so the search to find the maybe 20 abnormal genes in any of the 200 types of cancer is a daunting task, but one that will be facilitated by the genome sequence.

"All cancers are caused by abnormalities in DNA sequence," said Dr Michael Stratton, the head of Britain's Cancer Genome Project.

As for addressing addictions, medical research suggests that about 50 percent of the risk is genetic.

"That would make addiction more inheritable than diseases we commonly think of as genetic, such as adult onset diabetes or common hypertension," said Eric Nestler, the head of the department of psychiatry at the University of Texas Southwestern Medical Center.

About 100 genes and their products have been shown to influence the process by which animals become addictive. Nestler believes the sequencing of the human genome, as well as the completed genomes of the mouse and rat, which are expected soon, will narrow the search.

But so far scientists haven't been able to identify any genetic abnormalities in humans that contribute to the risk of addiction in humans.

'More powerful tools'

The biggest initial impact of the human genome is expected to be on drug development, customizing drugs to individual genetic profiles and earlier diagnosis of disease.

Currently there are fewer than 500 targets for all the drugs on the market. Scientists predict the sequencing will increase that number to several thousand, sparking a boom in genomic research in the pharmaceutical industry.

"There are potentially a huge number of targets that can be investigated for potential drugs. There is also the personalization of medicine," said Tim Hubbard of the Sanger Center in Cambridge, England.

He likened the human genome to an automobile manual used by mechanics to determine what is wrong with a car that isn't running properly.

"We're going to provide doctors with much more powerful tools to diagnose exactly what is wrong with somebody."

Dr. Michael Dexter, director of the Wellcome Trust, which funded the British part of the Human Genome Project, has said before that mapping the human genome "has been compared with putting a man on the moon, but I believe it is more than that.

"This is the outstanding achievement not only of our lifetime but perhaps in the history of mankind," he declared.

Specific sequences of DNA characters form the genes that make us what we are, govern our biological functions and determine our susceptibility to illnesses like cancer or diabetes.

The Associated Press & Reuters contributed to this report.