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NIH Launches Comprehensive Effort to Explore Cancer Genomics
The Cancer Genome Atlas Begins With Three-Year, \$100 Million Pilot

The National Cancer Institute (NCI) and the National Human Genome Research Institute (NHGRI), both part of the National Institutes of Health (NIH), today launched a comprehensive effort to accelerate our understanding of the molecular basis of cancer through the application of genome analysis technologies, especially large-scale genome sequencing. The overall effort, called The Cancer Genome Atlas (TCGA), will begin with a pilot project to determine the feasibility of a full-scale effort to systematically explore the universe of genomic changes involved in all types of human cancer.

“Now is the time to move forward with this pioneering initiative. Thanks to the tools and technologies developed by the Human Genome Project and recent advances in using genetic information to improve cancer diagnosis and treatment, it is now possible to envision a systematic effort to map the changes in the human genetic blueprint associated with all known forms of cancer,” said NIH Director Elias A. Zerhouni, M.D. “This atlas of genomic changes will provide new insights into the biological basis of cancer, which in turn will lead to new tests to detect cancer in its early, most treatable stages; new therapies to target cancer at its most vulnerable points; and, ultimately, new strategies to prevent cancer.”

NCI and NHGRI announced today at a news conference in Washington, D.C., that they have each committed \$50 million over three years to the TCGA Pilot Project. The project will develop and test the complex science and technology framework needed to systematically identify and

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characterize the genetic mutations and other genomic changes associated with cancer. The pilot will involve a few types of cancer that will be chosen for their value in helping to determine the feasibility of a possible larger-scale project. The process for determining the types of cancers to be studied is currently underway.

Cancer is now understood to include more than 200 different diseases. In all forms of cancer, genomic changes -- often specific to a particular type or stage of cancer -- cause disruptions within cellular pathways that result in uncontrolled cell growth. TCGA will delve more deeply into the genetic origins leading to this complex set of diseases, and, in doing so, will create new discoveries and tools that will provide the basis for a new generation of cancer therapies, diagnostics, and preventive strategies.

“The goal of studying the human genome has always been to improve human health. The Cancer Genome Atlas Pilot Project represents another bold step in that direction,” said National Human Genome Research Institute Director Francis S. Collins, M.D., Ph.D. “Such an ambitious venture requires significant planning. Given the genetic complexity of cancer, we are certain to face many daunting challenges in this pilot. But by pulling together some of the best minds in the cancer and genomics research communities, I am confident that the pilot will succeed, and we will go on to develop an atlas that will accelerate cancer research in ways we cannot even imagine today.”

NCI Deputy Director Anna D. Barker, Ph.D., said, “The Cancer Genome Atlas Pilot Project is a revolutionary step in cancer medicine that leverages advances in cancer biology, genomics technologies, biorepositories, and bioinformatics for the ultimate benefit of cancer patients. Key challenges for the TCGA Pilot Project include not only addressing cancer’s complexity, but also developing the technologies to advance the science of cancer genetics. A better understanding of cancer genetics is part of the overall effort to eliminate the suffering and death due to cancer.”

Data and technologies produced by other genomic projects have provided the tools necessary to produce new insights into how and why genetic changes cause cancer. The Human Genome Project, an international effort led in the United States by NHGRI and the Department of Energy, was completed in April 2003 and provided a reference DNA sequence of the human genome. The Human Genome Project also helped to advance sequencing technologies and paved the way for other genome-based research tools, including a comprehensive map of human genetic variation, or haplotypes, recently produced by the International HapMap Consortium.

Genetic mutations linked to breast cancer, colon cancer, melanoma, and other cancers already have led to diagnostic tests that can point to the most effective intervention. Recent

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discoveries in cancer genomics have helped to identify several treatments that work by targeting cancer cells with a specific genetic change, such as Gleevec[®], a drug for chronic myeloid leukemia and gastrointestinal stromal tumors, and Herceptin[®], a drug for one form of breast cancer. These successful developments support further examination of the molecular origins of cancer to more quickly develop new tools to diagnose, treat, and prevent cancer.

In the TCGA Pilot Project, a Human Cancer Biospecimen Core Resource will support the collection, processing, and distribution of cancerous and healthy, control tissue samples to Cancer Genome Characterization Centers and Genome Sequencing Centers. The genes and other genomic targets identified will be sequenced by the Cancer Genome Sequencing Centers using high-throughput methods similar to those employed in the Human Genome Project. The Cancer Genome Atlas Pilot Project seeks to identify genetic mutations in the DNA code that are specifically associated with the type of cancer being sequenced. In addition, the Cancer Genome Characterization Centers will work to identify other types of larger-scale genomic changes, such as copy number changes and/or chromosomal translocations, that contribute to cancer development and/or progression.

These data from TCGA Centers will be deposited in public databases supported by NCI's cancer Biomedical Informatics Grid (caBIG[™]) and the National Library of Medicine's National Center for Biotechnology Information. As in the Human Genome Project, TCGA data will be made available to the worldwide research community. This data will provide researchers and clinicians with an early glimpse of what is hoped will evolve into an unprecedented, comprehensive "atlas" of information describing the genomes of all cancers. This atlas will enable researchers throughout the world to analyze and use the data in their own research to develop new diagnostics and therapies for different cancers.

Recognizing that not all technologies needed for high-throughput, cost-effective analysis of the cancer genome are already in hand, TCGA also will support new technology development. Some of these methods will focus on improving current genomic analysis technologies, while others will emphasize new approaches, such as epigenomics. Epigenomics looks at how various small molecules, such as methyl groups, when added or removed from DNA, can have profound effects on gene function.

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Each component of the TCGA Pilot Project will have clear milestones and goals. Only if the pilot achieves its goals will the full-scale project to develop a complete atlas of the cancer genome move forward.

The Cancer Genome Characterization Centers, Genome Sequencing Centers, and Biospecimen Core Resources will be selected in 2006. Applications and proposals will be reviewed by experts in the field, and awards will be based on merit and programmatic needs of The Cancer Genome Atlas Pilot Project.

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NCI and NHGRI are two of the 27 institutes and centers at NIH, an agency of the U.S. Department of Health and Human Services.

For more details about The Cancer Genome Atlas, including Q&As, a graphic, a glossary, a brief guide to genomics, and a media library of available images, please go to <http://cancergenome.nih.gov>.

For more information about cancer and the National Cancer Institute, please visit the NCI Web site at <http://www.cancer.gov>, or call NCI's Cancer Information Service at 1-800-4-CANCER (1-800-422-6237).

Additional information about NHGRI can be found at its Web site, <http://www.genome.gov>.