



# Department of Health and Human Services National Institutes of Health

## Fiscal Year 2013 Budget Request

Statement for the Record

House Subcommittee on Labor-HHS-Education Appropriations

March 22, 2012

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Director, National Human Genome Research Institute

### Mr. Chairman and Members of the Committee

I am pleased to present the President's budget request for the National Human Genome Research Institute (NHGRI) of the National Institutes of Health (NIH). The Fiscal Year (FY) 2013 NHGRI budget of \$511,370,000 includes a decrease of \$893,000 below the comparable FY 2012 level of \$512,263,000.

It is an extraordinary time for the field of genomics. Through recent scientific advances and technological developments, we are gaining a deeper understanding for how the human genome plays a central role in health and disease, enabling investigators across the biomedical research spectrum to pursue new avenues for translating this knowledge into clinical applications. NHGRI, guided by an ambitious vision for genomics research that the Institute published in February 2011, is poised to lead a research agenda in FY 2013 that will focus not only on basic genome biology and the genomic underpinnings of disease, but will also seek to develop strategies for applying genomics to advance medical science and, ultimately, to improve the effectiveness of healthcare.

### Ensuring a Strong Foundation

The unprecedented decreases in the cost of DNA sequencing — resulting from NHGRI-stimulated technology development coupled with myriad innovations by the NHGRI Large-Scale Genome Sequencing Centers — have fundamentally changed how genomic data is now generated as part of biomedical research. Whereas sequencing that first human genome during the Human Genome Project cost upwards of a billion dollars, sequencing a human genome using recently developed technologies will soon cost \$1000 (or less).

The recent renewal of the program supporting the NHGRI Large-Scale Genome Sequencing Centers ensures the productive continuation of flagship initiatives such as The Cancer Genome Atlas (TCGA) in addition to special projects with specialists focusing on specific disorders, such as Alzheimer's disease. These centers will continue to

develop innovative methodologies and information management systems, which will inevitably lead to further reductions in the cost of genome sequencing. With such reductions will come the opportunity to sequence the tens of thousands of individual genomes required to understand the small genetic differences that cumulatively confer risk for common diseases, such as diabetes and heart disease. Furthermore, the accessibility of low-cost DNA sequencing technologies will be essential for making genome sequencing a routine part of clinical care.

To facilitate the utilization of genomic tools and information for exploring biological questions and ultimately improving clinical care, the NHGRI Centers of Excellence in Genomic Science will conduct interdisciplinary research and training initiatives focused on the production, analysis, and utilization of genomic data. From these efforts, new insights into the complexity of human genome function are emerging, and these in turn are benefiting the research community at large. Similarly, the human-centric ENCyclopedia of DNA Elements (ENCODE) project and the companion model organism ENCODE project (modENCODE) will continue to build a 'knowledge base' that details the functional genomic elements underlying biological processes in humans as well as organisms that serve as important models for studying human biology.

To complement the requisite understanding of normal genome function established by these projects, tools for defining the genetic contributions to human disease are being developed. NHGRI continues to lead efforts within the international 1000 Genomes project to build a deep catalog of genomic variants among different human populations; in turn, this information will be used to identify the subsets of rare and common variants that confer risk for (or protection from) specific diseases or adverse drug responses. FY 2013 will also see the key maturation of the Human Heredity and Health in Africa (H3Africa) initiative, an NIH Common Fund project managed by NHGRI. The increased knowledge generated about genomic variation and the complex interactions between environmental and genetic factors in African populations will enhance understanding of disease predispositions and drug responses for all human populations.

If genomics is to be a powerful contributor to studies being performed across the biomedical research community, researchers must be able to process and analyze the massive amounts of genomic data that they can now readily produce. NHGRI will pursue the establishment of pioneering approaches for data management and analysis via the development and refinement of bioinformatic tools, resources, and standards.

## **Translating the Potential**

The Genome Sequencing Program continues to be a prominent and vibrant part of the Institute's research portfolio. Looking ahead, it will play an increased role in translating genomic-based capabilities to understand disease biology. The Program's renewal in FY 2012 included not only continued support for medical sequencing projects, but also a charge to conduct collaborative research projects with other investigators to broaden the application of genome sequencing as a tool for unraveling the genomic basis of human disease. The prototype for the latter is TCGA, a collaboration with the National Cancer Institute to identify the genomic basis of many different forms of human cancer.

The renewal of NHGRI's Genome Sequencing Program also included establishment of new Mendelian Disorders Genome Centers focused on rare, single-gene (called Mendelian) diseases. These new centers will seek to establish the genetic basis for thousands of rare disorders (affecting millions of Americans) for which the genetic defects remain unknown. Recent advances in genome sequencing offer the hope that the genetic underpinnings for most of these rare diseases can be identified through focused research efforts that were not possible or affordable with previous genome sequencing technologies.

## **Preparing for Genomic Medicine**

To capitalize on its growing foundation of basic and translational research, NHGRI recently launched the Clinical Sequencing Exploratory Research projects, a new component of the Institute's Genome Sequencing Program. The new projects will investigate how to utilize genomic knowledge in medical settings and begin to explore how healthcare professionals can routinely use genome sequence information for patient care. A related effort, the Electronic Medical Records and Genomics (eMERGE) Network, is pursuing how patients' genomic information can be linked to disease characteristics and symptoms in their electronic medical records, providing the ability to explore associations with disease pathologies and eventually to improve patient care.

Key to the ultimate success in all of these endeavors will be continued attention to the societal implications of advancing genomic technologies and understanding. Deliberate, ongoing engagement by laboratory, clinical, and social scientists and scholars in ethics, law, and philosophy with the public must remain a priority.

Through its portfolio of basic and translational research, the Institute is pushing forward the boundaries of our knowledge and defining the issues that must be addressed before genomics is routinely deployed as a standard element in medical care. NHGRI is leading this charge by funding ambitious research programs to understand the structure and function of genomes more fully, to use genomics as a central tool for understanding the biology of disease, and to establish the path for the implementation of genomic medicine. In all of these pursuits, the Institute maintains a laser-like focus on its ultimate mission — to improve human health through genomics research.

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## **Eric D. Green, M.D., Ph.D.**

### **National Human Genome Research Institute Bethesda, Maryland**

Eric D. Green, M.D., Ph.D. is the Director of the National Human Genome Research Institute (NHGRI) at the National Institutes of Health (NIH) in Bethesda, Maryland, a position he has held since late 2009. Previously, he served as the NHGRI Scientific Director (2002-2009), Chief of the NHGRI Genome Technology Branch (1996-2009), and Director of the NIH Intramural Sequencing Center (1997-2009).

Born and raised in St. Louis, Missouri, Dr. Green received his B.S. degree in Bacteriology from the University of Wisconsin-Madison in 1981, and his M.D. and Ph.D. degrees from Washington University in 1987. During residency training in clinical pathology (laboratory medicine), he worked in the laboratory of Dr. Maynard Olson, where he launched his genomics research program. In 1992, he was appointed an assistant professor of pathology and genetics as well as a co-investigator in the Human Genome Center at Washington University. In 1994, he joined the newly established Intramural Program of the National Center for Human Genome Research at the NIH, later renamed the National Human Genome Research Institute.

Honors given to Dr. Green include a Helen Hay Whitney Postdoctoral Research Fellowship (1989-1990), a Lucille P. Markey Scholar Award in Biomedical Science (1990-1994), induction into the American Society for Clinical Investigation (2002), the Lillian M. Gilbreth Lectureship for Young Engineers at the National Academy of Engineering (2001), an Alumni Achievement Award from Washington University School of Medicine (2005), and induction into the American Association of Physicians (in 2007). He is a Founding Editor of the journal *Genome Research* (1995-present) and a Series Editor of *Genome Analysis: A Laboratory Manual* (1994-1998), both published by Cold Spring Harbor Laboratory Press. He is also Co-Editor of *Annual Review of Genomics and Human Genetics* (since 2005). Dr. Green has authored and co-authored over 250 scientific publications.

From the early 1990s, Dr. Green's research program was at the forefront of efforts to map, sequence, and understand eukaryotic genomes. His work included significant, start-to-finish involvement in the Human Genome Project. More recently, Dr. Green established a program in comparative genomics that involved the generation and comparative

analyses of sequences from targeted genomic regions in multiple evolutionarily diverse species. His laboratory utilized its own mapping and sequence data to identify and characterize several human disease genes, including those implicated in certain forms of hereditary deafness, vascular disease, and inherited peripheral neuropathy. Most recently, Dr. Green led a number of efforts utilizing contemporary strategies for large-scale DNA sequencing to study genomic variation among humans (in particular, relating to genetic disease) and to examine the microbial communities (i.e., the microbiomes) that exist in and on the human body.

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