National Human Genome Research Institute

Department of Health and Human Services
National Institutes of Health

Fiscal Year 2017 Budget Request

Statement for the Record
Senate Subcommittee on Labor-HHS-Education Appropriations

July 29, 2016

Prepared Statement of Eric D. Green, M.D., Ph.D.
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Mr. Chairman and Members of the Subcommittee:

Mr. Chairman and Members of the Committee: I am pleased to present the President's Fiscal Year (FY) 2017 budget request for the National Human Genome Research Institute (NHGRI) of the National Institutes of Health (NIH).

This past October, the scientific community celebrated a quarter century since the launch of the Human Genome Project. Now, roughly 13 years since the successful completion of that landmark effort, NHGRI looks forward to the next quarter century of genomics and its applications to advance human health. To this end, NHGRI is fostering a continuum of research from basic studies of the structure and function of genomes to work that advances medical science and seeks to improve the effectiveness of healthcare. Research funded by NHGRI, especially in the area of genomic medicine, has laid the foundation for major transformational efforts, such as the Precision Medicine Initiative and the National Cancer Moonshot.

As an example, NHGRI's partnership with the National Cancer Institute in leading The Cancer Genome Atlas for nearly 12 years has led to powerful new insights that will fuel the National Cancer Moonshot. Through the use of new genome-analysis technologies, data have been generated for over 10,000 tumor specimens, allowing the assembly of a rich data resource that is now freely available to any qualified researcher. This body of work is providing new insights about classifying and treating cancer, fundamental information for the National Cancer Moonshot.

Another major research program supported by NHGRI, the 1000 Genomes Project, has now established a powerful data resource with information about how humans differ with respect to their genomic blueprint. With its final summary papers published in *Nature* this past October, this international effort analyzed the genomes of over 2,500 people from 26 populations around the globe, identifying and cataloguing more than 88 million places in the genome that humans differ. Together, the generated information accounts for more than 99 percent of the common genomic variants (or 'spelling differences') that exist in the human population. By producing a global catalog of human genomic variation, researchers can now focus on which of the identified variants play a role in health and disease.

Despite the remarkable progress in genomics over the past quarter century, more powerful and accessible approaches for studying genome structure and function are needed. To help address this need, the Centers of Excellence in Genomics (CEGS) program supports multi-investigator, interdisciplinary teams that develop new approaches and technologies for using genomics in biomedical research. FY 2017 funds for NHGRI will support CEGS efforts that

combine genome-editing technologies and tissue-engineering methods to develop improved models of complex tissues such as the brain. The CEGS program exemplifies how basic research can be applied to specific problems, yielding solutions that can advance diverse fields of inquiry.

The promise of genomics as a clinical tool for improving patient care is now coming into focus. A program that vividly highlights this capability is the Undiagnosed Diseases Network (UDN). An extension of the Undiagnosed Diseases Program launched within the NIH Intramural Research Program in 2008 and funded by the NIH Common Fund, the UDN embodies the promise of genomic and precision medicine. Under NHGRI's leadership, both programs are helping patients (and their families) that have faced diagnostic odysseys and are discovering the genomic bases of extremely rare disorders. The value of UDN is multifaceted, with some aspects related to the care and support it provides to its patients and other aspects related to robust approaches being developed for the study of rare medical conditions. FY 2017 funds for the program will support an expansion in patient enrollment across the national network.

NHGRI is further maximizing the potential for public benefit from the Federal investment in genomics with programs such as UDN by promoting synergy across research communities. Leveraging leading experts across national consortia directly stimulates dialog and an exchange of ideas that can prove mutually beneficial to all groups involved. An illustration of this can be seen with the collaboration between UDN and NHGRI's Centers for Mendelian Genomics program, a large-scale effort that is elucidating the genomic causes of rare human diseases. Through their synergistic interactions, these consortia are producing new insights about basic biological pathways that are relevant to both rare diseases as well as more common ones. Utilizing the discoveries emanating from rare disease studies should propel forward basic understanding of common diseases, particularly in the characterization of disease mechanisms and the establishment of new treatment options.

NHGRI also recognizes the importance of examining and addressing the ethical, legal, and social issues associated with moving genomics initiatives forward and with implementing genomic medicine. In FY 2017, NHGRI will thus continue to fund research that examines the increasing accessibility of genomic information and technologies within society. Such studies will investigate questions related to biobanking, clinical genome sequencing, and broad data sharing - all issues of great relevance to the Precision Medicine Initiative. A particular focus will be placed on how such issues affect studies involving vulnerable or underrepresented populations. Genomic privacy and genetic discrimination, as well as the complexities associated with the return of results from genomics research studies, will also be explored.

Educating providers, patients, and the public about genomics is also imperative for the successful incorporation of genomics into healthcare. NHGRI dedicates significant time and attention to outreach efforts and the education of various stakeholders. The highly successful *Genome: Unlocking Life's Code* exhibition, developed in conjunction with the Smithsonian Institution, has now been seen by more than four million people as it travels to cities big and small across the United States.

NHGRI's well-rounded portfolio of basic, translational, and clinical research programs has the long-term aim of using genomics to advance the health of all Americans. FY 2017 funds will help to ensure that NHGRI continues to lead the genomics community, support the broader biomedical research community, and help realize a future of remarkable genomics-enabled healthcare innovations.

Eric D. Green, M.D., Ph.D.

Director, National Human Genome Research Institute

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), a position he has held since late 2009. NHGRI is the largest organization in the world solely dedicated to genomics research. 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 career in genomics research. In 1992, he was appointed 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 Research Program of the National Center for Human Genome Research, later renamed the National Human Genome Research Institute.

While directing an independent research program for almost two decades,
Dr. Green 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. These efforts eventually blossomed into a highly productive program in comparative genomics that provided important insights about genome structure, function, and evolution. His laboratory also identified and characterized several human disease genes, including those implicated in certain forms of hereditary deafness, vascular disease, and inherited peripheral neuropathy.

As Director of NHGRI, Dr. Green is responsible for providing overall leadership of NHGRI's research portfolio and other initiatives. In 2011, Dr. Green led NHGRI to the completion of a strategic planning process that yielded a new vision for the future of genomics research, entitled *Charting a course for genomic medicine from base pairs to bedside (Nature 470:204-213, 2011).* Since that time, he has led NHGRI in broadening its research mission; this has included designing and launching a number of major programs to accelerate the application of genomics to medical care. With the rapidly expanding scope of genomics, his leadership efforts also have involved significant coordination with multiple components of NIH, as well as other agencies and organizations.

Beyond NHGRI-specific programs, Dr. Green has also played an instrumental leadership role in the development of a number of high-profile efforts relevant to genomics, including the Smithsonian-NHGRI exhibition *Genome: Unlocking Life's Code*, the NIH Big Data to Knowledge (BD2K) program, the NIH Genomic Data Sharing Policy, and the U.S. Precision Medicine Initiative.

Posted: July 29, 2016