National Human Genome Research Institute

Fiscal Year 2004 Budget Request

Department of Health and Human Services
National Institutes of Health

Witness appearing before the
Senate Subcommittee on Labor-HHS-Education Appropriations

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Department of Health and Human Services
Statement by
Dr. Francis S. Collins
Director, National Human Genome Research Institute
On
Fiscal Year 2004 President's Budget Request
for the National Human Genome Research Institute

Mr. Chairman and Members of the Committee:

Due in great part to the visionary leadership and commitment of Congress, this month the International Human Genome Project (HGP), led by the National Human Genome Research Institute (NHGRI) of the National Institutes of Health (NIH), will have accomplished all of its original goals, ahead of schedule and under budget. This historic achievement, in the month of the 50th anniversary of Watson and Crick's seminal publication of the structure of DNA, opens the genomic era of medicine. April will also witness the publication of a bold vision for the future of genomics research, developed by the NHGRI. This vision, the outcome of almost two years of intense discussions with hundreds of scientists and members of the public, has three major areas of focus: Genomics to Biology, Genomics to Health, and Genomics to Society.

Genomics to Biology: The human genome sequence provides foundational information that allows development of a comprehensive catalog of all of the genome's components, determination of the function of all human genes, and deciphering of how genes and proteins work together in pathways and networks.

Genomics to Health: Completion of the human genome sequence offers a unique opportunity to understand the role of genetic factors in health and disease, and to apply that understanding rapidly to prevention, diagnosis, and treatment. This opportunity will be realized through such genomics-based approaches as identification of genes and pathways and determining how they interact with environmental factors in health and disease, more precise prediction of disease susceptibility and drug response, early detection of illness, and development of entirely new therapeutic approaches.

Genomics to Society: Just as the HGP has spawned new areas of research in basic biology and in health, it has created new opportunities in exploring societal issues. These include analysis of the impact of genomics on concepts of race, ethnicity, kinship, individual and group identity, health, disease, and "normality" for traits and behaviors, and defining policy options regarding the use of genomic information in both medical and non-medical settings.

New NHGRI Initiatives

The NHGRI has already begun several new initiatives, and is planning others, to meet the challenge of this new vision for the future of genomics. Below are examples of these cutting edge programs.

The Creation of a Human Haplotype Map

Multiple genetic and environmental factors influence many common diseases, such as diabetes, cancer, stroke, psychiatric disorders, heart disease, and arthritis; however, relatively little is known about the genetic basis of common diseases. The NHGRI has begun to create a "haplotype map" of the human genome to enable scientists to find the genes that affect common diseases more quickly and efficiently. The power of this map stems from the fact that each DNA variation is not inherited independently; rather, sets of variations are inherited in blocks. The specific pattern of particular genetic variations in a block is called a haplotype. This new initiative, an international public/private partnership led and managed by NHGRI, will develop a catalog of haplotype blocks, the "HapMap." The HapMap will provide a new tool to identify genetic variations associated with disease risk or response to environmental factors, drugs, or vaccines. Ultimately, this powerful tool will lead to more complete understanding of, and improved treatments for, many common diseases.

The ENCODE Project: ENCyclopedia Of DNA Elements

To utilize fully the information that the human genome sequence contains, a comprehensive encyclopedia of all of its functional genetic elements is needed. The identity and precise location of all transcribed sequences, including both protein-coding and non-protein coding genes, with their structure, transcription start sites, polyadenylation sites, and alternative splicing variants must be determined. The identity of other functional elements encoded in the DNA sequence, including promoters, enhancers, and other transcriptional regulatory sequences, and determinants of chromosome structure and function, such as origins of replication and hot spots for recombination, also is needed. The NHGRI has developed a public research consortium to carry out a pilot project, focusing on a carefully chosen set of regions of the human genome, to compare existing and new methods for identifying functional genetic elements. This **ENC**yclopedia **O**f **D**NA **E**lements (ENCODE) consortium, which welcomes all academic, government, and private sector scientists interested in facilitating the comprehensive interpretation of the human genome, will greatly enhance use of the human genome sequence to understand the genetic basis of human health and to stimulate the development of new therapies to prevent and treat disease.

Chemical Genomics

One novel way that the NHGRI plans to pursue translating genomics to human health is the development and deployment to the biomedical research community of libraries of small organic compounds. This is a fundamentally new approach for research in the public sector, and will accelerate understanding of the function of the human

genome and the development of new treatments. The NHGRI proposes to use the types of organic molecules in most marketed pharmaceuticals, "drug-like," or "small" molecules, as a core of this resource. In collaboration with other NIH institutes, the NHGRI is planning for a resource that includes: (a) large libraries of chemical compounds of appropriate structural diversity and properties; (b) assay development capacity; (c) robotic assay capacity, also termed high throughput screening (HTS); (d) medicinal chemistry capacity to transform "hits" identified by HTS into workable chemical probes; and (e) distribution capacity to disseminate the reagents to the biomedical research community efficiently.

Genome Technology Development

The NHGRI continues to invest in technology development that furthers the uses of genomics. Technical advances have caused the cost of sequencing to decline dramatically, from $10 to less than $0.09 per base pair, but this cost must decline even further for all to benefit from genomic advances. The NHGRI, along with many partners, will actively pursue the development of new technologies to sequence any individual's genome for $1,000 or less. Other areas of technology development are also ripe for expansion and the NHGRI plans to pursue them vigorously.

Studying the Genetic Basis of Health

Analytic methods to find genetic variants that contribute to disease can also help find genes and genetic variants that contribute to health. The NHGRI plans to support development of new tools and analytical methods to discover the genetic components of resistance to diseases, disorders, toxins, and drug reactions. By finding genetic variants that convey reduced susceptibility, researchers will better understand disease processes and how to slow, or even prevent, them. Promising approaches for identifying disease-resistant gene variants include studying people at high risk for a disease who do not develop it, relatives of people with disease who do not themselves have the disease, or individuals who reach extreme old age without serious illness.

Recent Scientific Advances in Genomics

Progress in Sequencing Model Organisms

From the Human Genome Project's outset, the NHGRI and its partners have included, among their research goals, mapping and sequencing the genomes of several non-human organisms, since they would be of great value in understanding the biological data encoded in the human DNA sequence and, thus, in combating human disease. Genomic sequences for a number of important organisms, beyond those initially identified by the HGP, have been determined. Primary among these is the laboratory mouse. In December 2002, an analysis of an advanced draft of the mouse genome was published and provided a key tool for interpreting the human sequence. The first assembly of the rat genome sequence was announced in the same month by the Rat Genome Sequencing Project. A peer review process now selects new genomes to sequence. To champion an organism, scientists write a "white paper" that presents arguments for prioritizing their proposed target for sequencing. After two rounds of white papers, this process determined the highest priority as: chicken, chimpanzee, cow, dog, a set of fifteen fungi, honey bee, sea urchin, and two protozoans. Sequencing of the chicken, chimpanzee, and honey bee has already begun.

Ethical, Legal and Social Implications of Genetic Research

The NHGRI devotes five percent of its annual budget to research involving the ethical, legal and social implications (ELSI) of genetics and genomics. Below are examples of this program's important work.

Genetic Discrimination

Most Americans are optimistic about the use of genetic information to improve health, but many are also concerned that insurers and employers will misuse genetic information. These concerns deter participation in important biomedical research and the clinical use of genetic information. The NHGRI has supported research efforts to elucidate this issue. Such research has helped inform legislative activity; over 40 states have passed genetic nondiscrimination bills.

Reducing Health Disparities

The NHGRI recognizes the critical importance of ensuring that the potential of genomic research benefits all racial and ethnic groups. The NHGRI has taken steps to engage and empower minority communities in genomic research. The rewards of genomic research will be realized only with active participation of all racial and ethnic groups. An important area of genomic research is investigating how DNA sequence variation affects differing susceptibility to disease among various populations. The significant societal ramifications of this research also need attention. Genomic research affects all populations; thus, all groups need to set the research agenda and examine the broader issues it raises. The NHGRI has intensified its efforts to address health disparities by developing a strategic plan that identifies goals in areas such as research projects, information sharing, development of partnerships, and increasing diversity of the research workforce.

Effects of Gene Patents and Licenses on Genetic Testing and Research

The NHGRI continues to be concerned about the issues of gene patenting and licensing. To gain a better understanding of these issues, it has funded case studies and surveys to describe and analyze the effects of patents that award proprietary claims to the use of DNA sequences. The NHGRI held a roundtable discussion in December 2002 with outside experts in gene patenting to explore the ramifications on healthcare delivery and research of patenting and licensing genetic sequence data and single nucleotide polymorphisms. The NHGRI will utilize the insights provided at this roundtable to define further research to inform the policy process.

Conclusion

This year marks a very exciting transition in the field of genomics, with the full sequencing of the human genome marking the successful achievement of all of the HGP's original goals, and thus the advent of the genomics era. When Congress decided to fund the HGP it did so with the justifiable belief that this work would lead to improved health for all. The ability to accelerate the realization of this vision now lies before us. At the same time, we must be sure that all our citizens have access to these technological advances and that this information is not misused. It is our sincere belief that the newly created discipline of genomics will make a profound difference on the health and well being of the people of this world. We are profoundly grateful for the support the Congress has given to this program.

Mr. Chairman, I am pleased to present the President's budget request for the National Human Genome Research Institute. The fiscal year (FY) 2004 budget includes $478,072,000, an increase of $13,467,000 over the FY 2003 enacted level of $464,605,000 comparable for transfers proposed in the President's request."

Francis S. Collins, M.D., PH.D

Director, National Human Genome Research Institute

April 14, 1950. Staunton, Virginia

Education:

University of Virginia, 1970 - B.S. (with Highest Honors);
Yale University, 1972 - M.S.; Yale University, 1974 - Ph.D.;
University of North Carolina School of Medicine, 1977 - M.D. (with Honors)

Professional History:

1977-1981, Intern, Resident, Chief Resident in Medicine, North Carolina Memorial Hospital, Chapel Hill, North Carolina. 1981-1984, Fellow in Human Genetics and Pediatrics, Yale University School of Medicine, New Haven, Connecticut. 1984-1993, Assistant, Associate and then Full Professor of Internal Medicine and Human Genetics, University of Michigan, Ann Arbor, Michigan. 1987-1993 Assistant, Associate, and then Full Investigator, Howard Hughes Medical Institute. 1993 to present, Director, National Human Genome Research Institute, NIH, Bethesda, Maryland.

Professional Organizations:

American Society of Human Genetics; American Society for Clinical Investigation; Association of American Physicians; Institute of Medicine; National Academy of Sciences; American Academy of Arts and Sciences.

Awards and Honors:

Morehead Foundation Fellow, 1973-1977; Alpha Omega Alpha, elected Junior year, President of UNC chapter, 1976-1977; Hartford Foundation Fellowship, 1985-1987; Paul di Sant'Agnese Award of the Cystic Fibrosis Foundation, 1989; Gairdner Foundation International Award, 1990; National Medical Research Award, National Health Council, 1991; American Academy of Achievement Golden Plate Award, 1994; The Baxter Award for Distinguished Research in Biomedical Sciences, Association of American Medical Colleges, 1994; Susan G. Komen Breast Cancer Foundation National Award for Scientific Distinction, 1995; Breath of Life Award, Cystic Fibrosis Foundation, 1997; Mendel Medal, Villanova University, 1998; Champions of Pediatric Research Award, Children's National Medical Center, 1998; Shattuck Lecture, Massachusetts Medical Society, 1999; Arthur S. Flemming Public Service Award, 1999; Association of American Physicians, George M. Kober Lecture Award, 2000; Scientist of the Year, National Disease Research Interchange, 2000; The Biotechnology Industry Organization and The Chemical Heritage Foundation Third Annual Biotechnology Award, 2001; Warren Triennial Prize Lecture, Massachusetts General Hospital, 2002; Lifetime Achievement Award, Virginia Biotechnology Association, 2002; 2002 Gairdner Foundation International Award of Merit; 51st National Prayer Breakfast Leadership Luncheon Speaker, 2003; Walker Prize, Museum of Science, Boston, Massachusetts, 2003.

Honorary Doctoral Degrees:

Emory University, Mary Baldwin College, Yale University, Mount Sinai School of Medicine, University of North Carolina, George Washington University, University of Pennsylvania, Brown University

Department of Health and Human Services
Office of Budget

William R. Beldon

Mr. Beldon is currently serving as Acting Deputy Assistant Secretary for Budget, HHS. He has been a Division Director in the Budget Office for 16 years, most recently as Director of the Division of Discretionary Programs. Mr. Beldon started in federal service as an auditor in the Health, Education and Welfare Financial Management Intern program. Over the course of 30 years in the Budget Office, Mr. Beldon has held Program Analyst, Branch Chief and Division Director positions. Mr. Beldon received a Bachelor's Degree in History and Political Science from Marshall University and attended the University of Pittsburgh where he studied Public Administration. He resides in Fort Washington, Maryland.

Department of Health and Human Services
Office of Management and Budget
Biographical Sketch

NAME: Kerry N. Weems

POSITION: Deputy Assistant Secretary for Budget

BIRTHPLACE: Portales, New Mexico

EDUCATION:
B.A., Philosophy, New Mexico State University, 1978
BBA, Management, New Mexico State University, 1978
MBA, University of New Mexico, 1981

EXPERIENCE:

January 24-Present
Acting Assistant Secretary for Budget, Technology and Finance, HHS

June 2002- Present
Deputy Assistant Secretary for Budget, HHS

2001 - 2002
Acting Deputy Assistant Secretary for Budget, HHS

1996 - 2002
Director, Division of Budget Policy, Execution and Management, HHS

1991 - 1996
Chief, Budget Planning Branch, HHS

1988 - 1991
Program Analyst, Office of Budget, HHS

1983 - 1988
Program and Budget Analyst, HHS
(Social Security Administration)

1981 - 1983
Staff Member, United States Senate

Honors and Awards:
2001 Presidential Rank Award
1995 Secretary's Distinguished Service Award
1993 HHS Senior Management Citation

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