National Human Genome Research Institute

Fiscal Year 2003 Budget Request

Department of Health and Human Services
National Institutes of Health

Fiscal Year 2003 Budget Request

Witnesses appearing before the
House Subcommittee on Labor-HHS-Education Appropriations

March 13, 2002

and

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

March 21, 2002

Francis S. Collins, M.D., Ph.D.
Director, National Human Genome Research Institute

Kerry N. Weems
Acting Deputy Assistant Secretary for Budget

Mr. Chairman and Members of the Committee:

During Fiscal Year 2003, the field of genetics will observe a major anniversary, and the National Human Genome Research Institute will reach an unprecedented accomplishment. Fifty years ago, in the spring of 1953, Drs. James D. Watson and Francis Crick reported the discovery of the double helix structure of DNA, a landmark achievement in the annals of scientific research. In 2003 the Human Genome Project expects to complete the final DNA sequence of the human genome. NHGRI and their partners in the International Human Genome Sequencing Consortium announced the working draft of the human genome sequence in June 2000, published the initial analysis in February 2001, and since then have been working to correct all the remaining spelling errors and fill in all the gaps. The Human Genome Project is on target to meet that deadline and expects to finish the analysis in time for the 50th anniversary of the Watson-Crick paper.

The availability of the genome sequence of humankind could be said to mark the starting point of the genome era in biology and medicine. There is now much important work to do to deliver on the promise that these advances in genomics offer for human health. While sequencing the human genome has been NHGRI's most visible goal, the

Institute has also been conducting important genetic and genomic research in a variety of areas, including working to understand the way individuals differ from each other at the genetic level and the impact these variations may have on health. In addition, the Institute leads in the development of new technologies, such as DNA chips and tools for proteomics, and has been creating novel research strategies to study the function of genes and genomes.

A New Research Plan for NHGRI

The Human Genome Project has, since its inception, been guided by a series of overlapping five-year plans. These plans have laid out ambitious goals to advance our understanding of the human genome and the associated ethical, legal and social implications. The plans have been instrumental to the success of the Project by clearly enumerating our program objectives to the scientific community and the public, and by providing measurable objectives to guide our work and gauge our progress and success.

In December 2001, the NHGRI convened about 200 experts, including scientists, researchers in the ethical, legal, and social implications (ELSI) of the Human Genome Project, consumers, and policy experts to think very broadly and creatively about the future of genomics. Over the course of the following months, we will host several workshops to explore specific topics in detail and enumerate specific goals appropriate for NHGRI. We will take stock of where we are and where we have come from, critically evaluating the challenges and opportunities that lie before us and creating a bold new vision for the future of genomics.

Early and Stunning Results from the Human Genome Sequence

Obtaining an accurate reference version of the human sequence has always been the most compelling goal of the Human Genome Project. Between March 1999 and June 2000, the production of human genome sequence data in Institute-supported laboratories skyrocketed. During this time, scientists sequenced 1,000 DNA letters a second, 24 hours a day, seven days a week. The resulting working draft sequence covered over 94 percent of the human genome, with 33 percent in highly accurate finished form by February 2001. By January 2002 the amount in highly accurate finished sequence had risen to 65 percent. The final sequence will be completed in 2003, two years ahead of the original ambitious schedule.

The draft sequence of the human genome is already having a major impact on biomedical research. In the 12 months following the February 2001 publication in Nature of the publicly funded draft sequence, the paper has been cited in over 700 scientific reports, making it one of the most cited papers in all of science for the past year. These citations clearly demonstrate the widespread utility of the publicly available genome sequence and its enormous early impact to advance biomedical research in a wide array of areas.

The rationale for the Human Genome Project, and the strong and sustained Congressional support for it, has been the promise of improving human health. We are already beginning to see the fruits of that investment. Some of the citations of the Nature publication represent research that could not have been accomplished in nearly the same way or would not have been as profound were it not for the draft sequence of the human genome. More than 50 genes involved in human disease have been discovered, based on access to the public human genome sequence data. The examples cited below show the direct connection the genome sequence is having on improving human health.

Prostate Cancer

Using the draft sequence of the human genome, scientists at Johns Hopkins University and the NHGRI have found the first gene associated with an inherited form of prostate cancer. In a study of 91 high-risk prostate cancer families the researchers mapped the first hereditary susceptibility to prostate cancer to a region of chromosome 1 that they called the Hereditary Prostate Cancer 1 Region, or HPC1. They have now identified a specific gene - called RNASEL - in the HPC1 region that contains DNA misspellings associated with prostate cancer. Misspellings in this one gene

do not explain all forms of inherited prostate cancer, but the discovery of this gene is an exciting step towards understanding the causes of this common and devastating form of cancer. Ultimately, this discovery should bring us closer to being able to prevent the disease as well as better diagnostics and treatments.

Kidney Disease Gene

The recent identification of the gene for autosomal recessive polycystic kidney disease (ARPKD) by a team at the Mayo Clinic again shows the great power of the draft human sequence. The publicly available sequence of the human genome played an important role in the discovery of this disease-causing gene. With the identification of the responsible gene and the characterization of a rat model of the disease, rapid progress in understanding ARPKD can now be anticipated.

The Future of Genomics

The Human Genome Project and the NHGRI have always aimed to develop new information, tools and technologies that would enable scientists to gain a deeper understanding of the genetic contributions to disease, and to use this knowledge to improve human health. The imminent completion of the project's initial goals presents a compelling opportunity to focus aggressively on translating the spectacular research advances into medical advances. With the completion of the Human Genome Project soon at hand, much additional basic research, guided by a genomic approach, remains to be done to shed light on the many mysteries of life. At the same time, genome research offers a myriad of other opportunities for connecting detailed knowledge of the human genetic instruction book with important problems in clinical research. These basic and applied paths are not mutually exclusive, and finding the right balance between them, although challenging, will be the most effective approach in the end.

Comparative Genomics

To understand the function of the human genome sequence, scientists would like to compare it to the genome sequences of many other organisms. This approach relies on the fact that functionally important regions of DNA are conserved over long periods of evolutionary time. By comparing the human genome sequence with those of the rat, mouse, and other organisms, similar regions are readily apparent, indicating that something biologically interesting -- such as the existence of a gene or important regulatory element -- must be present at that location of the genome.

Simplifying the Study of Complex Genetic Diseases: The Haplotype Map of the Human Genome

Prior to the completion of the draft sequence of the human genome, most studies of diseases using genetics focused on single gene disorders such as cystic fibrosis and Huntington's disease. With the tools of the Human Genome Project, finding the genes for diseases caused by alterations in single genes has become relatively straightforward. Many common diseases, however, such as diabetes, cancer, heart disease, psychiatric disorders, and asthma are influenced by complex interactions between multiple genes as well as by non-genetic factors such as diet, exercise, smoking and exposure to toxins.

A key next step of the Human Genome Project will be the generation of a "haplotype map" of the human genome. This comprehensive resource for human biomedical research will capture the complete catalogue of the common genome ancestral segments - "haplotype blocks" - observed in the major human populations. This map will provide a new tool for scientists to scan the entire genome and identify more rapidly and effectively those genetic variations associated with disease risk and drug response in the human population. That, in turn, will help researchers develop an understanding of the complex biological processes that give rise to the disease and assist scientists in discovering treatments or cures for these illnesses. This new and exciting project is expected to be a public-private partnership and the data will be immediately and freely accessible.

Health Disparities Strategic Plan

From its inception NHGRI has been concerned about including individuals from various groups in its activities. As the Institute has grown in size and complexity the need for this has become even more imperative and a variety of initiatives have been started and continue to evolve to address this need. The NHGRI staff recognizes the inherent value of increasing diversity among the research workforce as well as engaging and empowering people from minority communities through joint research projects, information sharing, dialogue and the development of partnerships. In order to achieve these goals, NHGRI has developed a plan that lays out a multifaceted approach to address issues of health disparities. The plan encompasses research, training, and education/outreach activities.

Ethical, Legal and Social Implications

From its inception, NHGRI has taken on the responsibility to address the broader ethical implications of rapid advances in genetic information and technology. Since 1991, it has committed 5 percent of its budget to studying the ethical, legal and social implications (ELSI) of genome research.

The ELSI Research Program has continued to support significant and innovative research on the ethical, legal, and social implications of human genome research. Research projects supported in FY 2001 included projects in the areas of the privacy and fairness in the use and interpretation of genetic information; clinical integration of new genetic technologies; issues surrounding genetics research; and public and professional education.

As the Institute develops its new research plan, the ELSI issues will be carefully integrated. It will be extremely important to consider these issues as new fields of genomic discovery appear. It will also be essential for ELSI funded research to inform policy development in the area of genetics.

Education and Outreach

National Coalition for Health Professional Education in Genetics

In 1996, along with the American Medical Association and the American Nurses Association, the NHGRI founded the National Coalition for Health Professional Education in Genetics as a national effort to promote health professional education and access to information about advances in human genetics.

NHGRI/ORD Genetic and Rare Diseases Information Center

There are more than 6,000 genetic and rare diseases afflicting more than 25 million Americans, but many of these illnesses affect relatively few individuals. As a result, information about these rare disorders may be limited or difficult to find. In order to respond to this need, the NHGRI and the Office of Rare Diseases (ORD) have established the NHGRI/ORD Genetic and Rare Diseases Information Center to provide information on genetic and rare disorders to the public. The Information Center will meet the ever-increasing information needs of the general public, including patients and their families, health care professionals, and biomedical researchers by: 1) serving as a central, national repository of information materials and resources on genetic and rare diseases; 2) collecting and disseminating information on the diagnosis, treatment and prevention of genetic and rare disorders; and 3) coordinating with organizations and associations interested in genetic and rare disorders.

Conclusion

The investment in the Human Genome Project is already paying off in terms of advances in biomedical science that promise unprecedented advances in human health. We are moving into a new phase of genomics which will give us a deeper understanding of the genetic contributions to disease. Our vision is that by focusing on the applications of genetics to human health we will make great strides towards treating and curing many complex diseases.

NIH budget request includes the performance information required by the Government Performance and Results Act of 1993. Prominent in the performance data is NIH's second annual performance report which compared our FY 2001 results to the goals in our FY 2001 performance plan.

Mr. Chairman, I am pleased to present the President's budget request for the National Human Genome Research Institute for Fiscal Year 2003, a sum of $466,695,000, which reflects an increase of $35,977,000 over the comparable Fiscal Year 2002 appropriation.

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.

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 Management and Budget
BIOGRAPHICAL SKETCH

NAME: Kerry N. Weems

POSITION: Acting 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:

2001 - present: Acting Deputy Assistant Secretary for Budget, HHS

1996 - present: 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

Last Reviewed: February 23, 2012