



**Testimony  
Before the  
Subcommittee on Health  
Committee on Energy and Commerce  
United States House of Representatives**

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**The Threat of Genetic Discrimination to the  
Promise of Personalized Medicine**

*Statement of*

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Good afternoon, Chairman Pallone and members of the Subcommittee. Thank you for the opportunity to speak with you today. I am Francis Collins, Director of the National Human Genome Research Institute (NHGRI) at NIH, part of the National Institutes of Health (NIH) within the Department of Health and Human Services.

It is my pleasure to be appearing before you today as you consider the Genetic Information Nondiscrimination Act of 2007. We stand at a critical time in the development of medicine: the mapping of the human genome has provided powerful new tools to understand the genetic basis of disease, but our ability to fully realize the promise of personalized medicine is limited by legitimate fear of how this powerful information could be abused. Many people are afraid that their genetic information will be used against them and are unwilling to participate in medical research or be tested clinically, even when they are at substantial risk for serious disease. More than ten years ago, expert advisors to the genome project concluded that federal legislation is needed to provide all Americans with protection against genetic discrimination in health insurance and employment. Without it, we may never realize the full potential of genomic research, and, more importantly, of individualized approaches to health care.

### **New Tools and Technologies**

Since the completion of the Human Genome Project (HGP) in 2003, major advances in our understandings of the causes of disease have been appearing at an accelerated pace. As one example, the HGP enabled the development of the “HapMap,” a detailed map of variations in the spelling of our DNA instruction books. Research supported by NHGRI has also led to orders of magnitude reduction in the costs of sequencing an individual’s complete genome for medical

purposes. It is the vision of NHGRI that within the next ten years, the cost of sequencing the complete genome of an individual will be \$1,000 or less. Should an individual so choose, this information could then be used as part of routine medical care, providing health care professionals with a more accurate means to predict disease, personalize treatment, and preempt the occurrence of illness.

### **New Findings in Genetics of Common Disease**

Even before the \$1000 genome becomes a reality, advances from genome research are already leading to important new understanding of the role of genetic factors in a number of common diseases. For instance, the HapMap made possible research that recently identified two major genes that influence risk for developing adult macular degeneration, a leading cause of vision loss in the elderly, with those at lowest risk having less than 1% chance of developing the disease, and those at highest risk a 50% chance. Other similarly derived recent discoveries include identification of variants in different genes that elevate risk for developing type 2 diabetes, Crohn's disease, prostate cancer, and Alzheimer's disease. Other new findings include the identification of genetic variants that predict whether or not a particular individual will respond well to drug treatment for disease, or will suffer a side effect. Each of these discoveries opens a new path toward diagnosis, prevention, and treatment, but the public will be reluctant to travel these paths if fair and reasonable protections against the improper use of genetic information are not in place.

NHGRI is currently involved in other groundbreaking initiatives, such as the Genetic Association Information Network (GAIN) and the Genes, Environment, and Health Initiative

(GEHI), that will accelerate understanding of the environmental and genetic causes of common diseases such as asthma, schizophrenia, cancer, bipolar disease, diabetes, and Alzheimer's disease. Increased understanding will in turn lead to better strategies for individualized prevention and treatment and enable the development of personalized health care. NHGRI has also joined with NIH's National Cancer Institute in funding a joint project called The Cancer Genome Atlas (TCGA) to accelerate understanding of the molecular basis of cancer through application of genome analysis technologies. TCGA will provide new insights into the biological basis of cancer, and will help to optimize treatment and prevention strategies.

Already, healthcare providers can test whether some of us carry DNA variants that pre-dispose us to certain diseases, and new research efforts could help to expand this capability and possibly offer better opportunities for preventive measures. If illness does occur, doctors will have more powerful tools to identify the molecular causes, and to prescribe medicines based on individualized genetic information. This is our chance to transform medicine from "one-size-fits-all" to a potentially personalized approach.

### **Fear of Discrimination**

As you can see, the science of genomic medicine is rocketing forward. But fear of genetic discrimination threatens to slow both the advance of such groundbreaking biomedical research and the integration of the fruits of that research into our nation's health care. If individuals continue to worry that they will be denied health insurance or refused employment because they have a predisposition to a particular disease, they may forego genetic testing that could help guide medical professionals to lessen their risk, simply because the test identifies them as having

such a predisposition. This is about all of us, as there are no perfect specimens at the DNA level; each one of us carries numerous gene variants that increase our risk of developing one disease or another. Therefore, each one of us is at risk for genetic discrimination.

Public concerns about the possible misuse of their genetic information by insurers or employers have been documented. A recent NIH study of families at risk for hereditary nonpolyposis colorectal cancer (HNPCC) (a particular form of colon cancer) revealed that the number one concern expressed by participants regarding genetic testing was about losing health insurance, should the knowledge of their genetic test result be divulged or fall into the “wrong hands.” Nearly half of individuals with a 50% chance of having the HNPCC mutation cited fear of insurance discrimination as their greatest concern surrounding their participation in this study. Similarly, a recent survey of the personal attitudes of cancer genetics specialists showed that 68% of respondents would not bill their own insurance company for HNPCC or breast and ovarian cancer (BRCA) genetic testing due to fear of genetic discrimination, and 26% of respondents said they would use an alias when being tested.

NHGRI remains deeply concerned about the impact of potential genetic discrimination on both research and clinical practice. Unless Americans are convinced that their genetic information will not be used against them, the era of personalized medicine may never come to pass. The result would be a continuation of the current one-size-fits-all medicine, ignoring the abundant scientific evidence that the genetic differences among people help explain why some of us benefit from a therapy while others do not, and why some of us suffer severe adverse effects from a medication, while others do not.

In 2005, the Bush Administration issued a Statement of Administrative Policy supporting Senate passage of S. 306, the “Genetic Information Nondiscrimination Act of 2005.” That bill never came to a vote in the House. In January of this year, the President visited the NIH and again called on Congress to pass a bill to protect Americans from genetic discrimination. We share the President’s concern and commitment to this issue, and we are delighted to see this issue being taken up early in the 110<sup>th</sup> Congress. We are hopeful that this will be the year when the American people are given a gift that is long overdue – federal legislative protection against genetic discrimination.

Thank you, Mr. Chairman. I would be pleased to answer any questions that the Committee might have.

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**Education:**

University of Virginia, 1970 - B.S. (with Highest Honors);

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.

**Biographical Information:**

Dr. Collins is a physician-geneticist noted for his landmark discoveries of disease genes and his leadership of the Human Genome Project. With Dr. Collins at the helm, the Human Genome Project consistently met projected milestones ahead of schedule and under budget. This international project culminated in April 2003 with the completion of a finished sequence of the human genetic blueprint. From its outset in 1990, the public sequencing effort swiftly deposited all of its data into free, public databases for use by scientists around the world. Building on the foundation laid by the Human Genome Project, Dr. Collins is now leading the NHGRI effort to ensure that this new trove of sequence data is translated into powerful tools and thoughtful strategies to advance biological knowledge and improve human health.

Dr. Collins' own research initiatives have included the discovery of a number of important genes, including those responsible for cystic fibrosis, neurofibromatosis, Huntington's disease and the gene that causes Hutchinson-Gilford progeria syndrome, a dramatic form of premature aging. In addition to his scientific achievements, Dr. Collins is known for his continuing emphasis on the importance of ethical and legal issues in genetics. He has been a strong advocate for protecting the privacy of genetic information and has served as a national leader in efforts to prohibit gene-based insurance and employment discrimination.

**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.