

## REPORT OF THE WORKING GROUP ON ETHICAL, LEGAL, AND SOCIAL ISSUES RELATED TO MAPPING AND SEQUENCING THE HUMAN GENOME

The plan to map and sequence the human genome has profound implications for the alleviation of human suffering due to genetic disease. Genes directly causing or predisposing to human disease will be placed on the map for all to investigate. Additionally, normal genes which may be involved in the pathways leading to the development of new treatments will be captured and fundamental biological lessons in genetic regulation and functioning will be learned through the Human Genome Initiative.<sup>1</sup>

Any scientific endeavor of this magnitude must be developed in concert with a plan to ensure that the public has access to the benefits in improved health care, which should be a result of the research. It is also imperative to protect individuals and society from possible hazards which may be a consequence of our improved ability to detect and predict hereditary illness. The use of genetic information, for good or ill, has long been an issue in our society. But the quantity and complexity of genetic information that should become available requires that special precautions be taken.

Accordingly, the National Center for Human Genome Research is giving high priority to the development of a program to address the ethical, legal, and social implications of the Human Genome Initiative. This plan will attempt to anticipate the impact of the Human Genome Initiative and address what protections need to be in place so that the information generated can be of maximum benefit to individuals and society.

Although initially the Human Genome Initiative will produce information that will lead to the detection and diagnosis of genetic disease, the long-range goal will go beyond this to providing improved treatment, prevention, and ultimately cure. The interim phase, before adequate treatment is available, is the one in which the most deleterious consequences can occur, such as discrimination against gene carriers, loss of employment or insurance, stigmatization, untoward psychological reactions and attention. Once effective treatment is available for an illness, most of these

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<sup>1</sup>The Human Genome Initiative is discussed in detail in the National Academy of Science's 1988 report, *Mapping and Sequencing the Human Genome* and the Office of Technology Assessment's 1988 report, *Mapping Our Genes--The Genome Projects: How Big, How Fast?*



problems disappear. As the fruits of the Human Genome Initiative are realized, there will be an increased need for improved professional and public education to take advantage of the information gained.

In responding to the desires of the scientific community to understand the social, ethical, and legal implications of research on the human genome, the Office of Human Genome Research developed a program announcement, which appeared in the March 3, 1989 NIH Guide to Grants and Contracts. Applications were requested to address questions such as: (1) What are the concerns to society and to individuals?; (2) What questions in the areas of ethics and law need to be addressed?; (3) What can be learned from precedents?; (4) What are the policy alternatives and the pros and cons of each?; and (5) How can we inform and involve the public?

At its January 1989 meeting, the Program Advisory Committee on the Human Genome established the working group on ethics to develop a plan for this component of the human genome program. After considerable informal discussion within the group and with other scholars in ethics, law, and related fields over subsequent months, the working group had its first formal meeting on September 14-15, 1989. A roster of the members is attached.

At this meeting, the working group began to define and develop a plan of activities to address the ethical, legal, and social issues arising out of the application of knowledge gained as a result of the Human Genome Initiative. Representatives of the National Science Foundation (NSF) and the National Endowment for the Humanities were invited to present their grant programs for research on ethics, science, and society, and the working group noted that there was considerable opportunity for collaboration with these agencies, taking advantage of their expertise and experience in managing grants in this field.

The working group agreed that the purpose of the ethics component of the human genome program should be to:

- o anticipate and address the implications for individuals and society of mapping and sequencing the human genome;
- o examine the ethical, legal, and social consequences of mapping and sequencing the human genome;
- o stimulate public discussion of the issues; and
- o develop policy options that would assure that the information is used for the benefit of individuals and society.



The working group was strongly supportive of a program that would anticipate problems before they arise and develop suggestions for dealing with them that would forestall adverse effects. The approach to accomplishing these objectives should be several fold:

- o to stimulate research on the issues through grants;
- o to refine the research agenda through workshops, commissioned papers, and invited lectures on specific topics selected by the working group;
- o to solicit public input from the community-at-large through town meetings and public testimony;
- o to support the development of educational materials for all levels; and
- o to encourage international collaboration in this area.

### **Stimulate Research**

The working group is eager to encourage investigators in the research community to explore the wide range of issues pertinent to the human genome program. Outcomes of this research may be used to develop educational programs, policy recommendations or possible legislative recommendations.

In discussing the ethical, legal, and social consequences of the Human Genome Initiative, the working group deemed the following topics to be of particular importance and will strongly encourage research in the following areas.

1. Fairness in the use of genetic information with respect to:
  - o insurance (acquisition and maintenance of health, life, disability, catastrophic, long-term care, and automobile insurance coverage)
  - o employment (equal access)
  - o the criminal justice system
  - o the education system
  - o adoptions
  - o the military
  - o any other areas to be identified



2. The impact of knowledge of genetic variation on the individual, including issues of:
  - o stigmatization
  - o ostracism
  - o labelling
  - o individual psychological responses, including impact on self image
3. Privacy and confidentiality of genetic information regarding:
  - o ownership and control of genetic information
  - o consent issues
4. The impact of the Human Genome Initiative on genetic counseling in the following areas:.
  - o prenatal testing
  - o pre-symptomatic testing
  - o carrier status testing, especially for very common disorders such as cystic fibrosis
  - o testing when there is no therapeutic remedy available, such as for Huntington's disease
  - o counseling and testing for polygenic disorders
  - o population screening versus testing
5. Reproductive decisions influenced by genetic information:
  - o effect of genetic information on options available
  - o use of genetic information in the decision-making process
6. Issues raised by the introduction of genetics into mainstream medical practice:
  - o qualifications and continuing education of all appropriate medical and allied health personnel
  - o standards and quality control
  - o education of patients
  - o education of the general public



7. Uses and misuses of genetics in the past and the relevance to the current situation, e.g.:
  - o the eugenics movement in the U.S. and abroad
  - o problems arising from screening for sickle-cell trait and other recent examples in which screening or testing sometimes achieved unintended and unwanted outcomes
  - o the misuse of behavioral genetics to advance eugenics or prejudicial stereotypes
8. Questions raised by the commercialization of the products from the Human Genome Initiative in the following areas:
  - o intellectual property rights (patents, copyrights, and trade secrets)
  - o property rights
  - o impact on scientific collaboration and candor
  - o accessibility of data and materials
9. Conceptual and philosophical implications of the Human Genome Initiative on:
  - o the concept of human responsibility
  - o the issue of free will versus determinism
  - o the concept of genetic disease, particularly in view of the high rate of human genetic variability and the large numbers of people who will be found to have genetic vulnerabilities

Most of this research can best be accomplished through the support of scholarly research and conferences. The working group recommended that support for conferences be limited to those that are highly focussed and produce a specific product, such as recommendations or policy options. The types of research to be supported should be varied and involve many of the disciplines traditional to the humanities. General surveys for purposes of information gathering are not recommended at this time.

### **Refine the Research Agenda**

The working group is intentionally small so that others with specific necessary expertise can be recruited to join the effort as needed. To accomplish its task, the working group plans to invite individuals from a variety of disciplines to help refine



the research and policy agenda. This activity will include small workshops, commissioned papers, and invited lectures by knowledgeable individuals. In an effort to gather needed information in a timely manner, the working group will convene two to three times annually to collect information and discuss how this new knowledge will be integrated into a plan to refine the research agenda and propose future action.

Initial plans for the first workshop are underway. The format of a focus group is envisioned. Participants will include prominent individuals from various occupations and professions on which the Human Genome Initiative will have an impact such as, insurance companies, industry, labor unions, geneticists, "consumers" of genetic information and services, constitutional law, newsmedia, and the arts. The intent is to invite individuals who may not have been actively involved in the Human Genome Initiative or genetic research or services, but who can view the issues from a fresh perspective.

Participants will be provided background materials compiled by members of the working group and will be encouraged to discuss, on the basis of their experience and expertise, the most salient ethical, legal, and social repercussions of the plan to map and sequence the human genome and suggest areas of research, policy development, or legislation that they feel should be in place. From these discussions, the working group will formulate specific recommendations to bring before the advisory committee.

### **Solicit Public Input**

The working group unanimously agreed that a critical component of its mission is to inform the general public (in the broadest sense) about the Human Genome Initiative and to solicit from them their questions and concerns about human genome research.

The town meeting format was considered appropriate for soliciting public input. However, to be effective such meetings must be carefully planned, taking into consideration the need to reach a broad cross-section of the public, and factors such as site, selection of participants, and wide publicity. A meeting of this type is tentatively planned for early 1991, or the end of the first year of this plan.

### **Support of Education**

The human genome program should include a strong educational component involving both formal and informal education targeted to all educational levels.



It is suggested that NIH collaborate with NSF to develop model curricula that would be appropriate for the following groups: students at all levels, the newsmedia, medical practitioners, genetic counselors, scientists, teachers, and groups targeted for genetic services. Because NSF has experience in curriculum development, the working group believes that co-funding of appropriate NSF programs would be an efficient way for NIH to accomplish its goals in this area. In addition, a program of individual postdoctoral fellowships, such as those funded in the scientific components of the human genome project, are recommended for support of individuals who have doctoral degrees in biomedicine and want to pursue studies in the ethical, legal, or social aspects of human genome research or vice versa.

Additional activities that should be pursued are:

- o short courses in ethical, legal, and social aspects of human genome research for scientists; and
- o short courses in genomics for scholars from the humanities who want to do research on the ethical, legal and social implications of the genome project.

### **International Collaboration**

The working group supports the concept of international collaboration in this area under guidelines similar to those for biomedical research on the human genome. Collaborative projects should be supported by funds from all the participants in the collaboration. The Human Genome Organization (HUGO) could play an obvious role in this area, which would be welcomed.

The Human Genome Initiative will have a profound impact on the lives of people in all countries, including those without genome research programs. Ideally, representatives from all interested countries should participate in considering the issues that will arise. An international organization, such as UNESCO, could facilitate cooperation in this area.

Diseases and the suffering they cause respect no geographical boundaries. The sharing of results from the Human Genome Initiative across geographical barriers must be encouraged. Although differences exist cross culturally in the use of genetic information, the working group hopes there are also sufficient similarities so that its efforts can be useful to all.

**Contributors to the Report on Ethical, Legal, and Social Issues  
Related to Mapping and Sequencing the Human Genome**

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## **Contributors to the Report on Ethical, Legal, and Social Issues**

**Jonathan R. Beckwith** is a bacterial geneticist interested in genetic screening. For more than 20 years, he has been interested in and concerned about the long-range implications of genetics and behavior and genetics and intelligence quotient.

**Robert Cook-Deegan** is a clinician whose interest in genetics dates back to his research on Alzheimer's Disease. While on an Office for Technology Assessment fellowship, he prepared two reports on human gene therapy and public policy related to the human genome project. Dr. Cook-Deegan is currently writing a book on how the Human Genome Initiative got started in the United States.

**Patricia King** is an attorney and academician whose legal career has been in civil rights law. She has served on the National Committee for Protection of Human Subjects, the Recombinant DNA Advisory Committee, and the Presidential Committee on Ethics. Ms. King is a Fellow of the Hastings Center and is interested in genetics and how it affects minorities.

**Victor A. McKusick** is an internationally recognized geneticist who has been active in human genetics research for over 40 years. More than any other person, he has been responsible over the years for collecting data on inherited diseases. Since 1973, he has collected and coordinated data on the human gene map, which in 1988 included 2,000 genes.

**Robert F. Murray, Jr.** is a clinical researcher who directs a clinical genetics program in sickle-cell disease. He became involved initially with the ethical aspects of screening for sickle-cell disease. Dr. Murray is concerned about individuals who want to plan their destiny based on new technologies emanating from genetics research.

**Thomas H. Murray** is a social psychologist who has written extensively about genetic screening in the work place. He has undertaken fellowships with an emphasis on humanities at Yale University and The Hastings Center. Dr. Murray was recently elected a Fellow of The Hastings Center and is currently co-authoring a publication with a geneticist for the British Medical Association.

**Nancy S. Wexler** is a clinical psychologist and a researcher. Her mother died of Huntington's Disease and she is a potential consumer of the information generated by the Human Genome Initiative. Many of her current efforts are to get individuals, interest groups, and the federal government to anticipate how information generated from the Human Genome Initiative can be used maximally to benefit the individual.