National Center for Human Genome Research

Program on the Ethical, Legal, and Social Implications of Human Genome Research

Progress Highlights

Public Discussion and Education

Seventeen open meetings devoted to the public discussion of the social implications of genome research will have been held during the 1990-91 academic season, in 11 states and the District of Columbia. 15 of these meetings are supported by NCHGR staff involvement or financial support.

These meetings are complemented by NCHGR support for a 10 part public television series now in production entitled "The Future of Medicine", which will explore the public impact of genome research.

ELSI Research and Policy Development Cycle

The NCHGR provided support for 16 projects addressing ethical and social issues in FY 1990, for a total of $1.6 million. In FY 1991, NCHGR expects to provide approximately $2.4 million in support of additional projects. The investigators leading these projects will produce their own reports and recommendations concerning the specific issues they are exploring. Meanwhile, in order to coordinate the work and catalyze results, the NCHGR will bring the investigators concerned with high priority issues together on a regular basis to assess progress and ascertain areas of consensus. The reports of these meetings will analyzed by the NIH-DOE ELSI Working Group, and recommendations for policy development by government agencies, legislators, professional groups or industries will be formulated and communicated directly to the relevant audience.
The first such meeting, on Sept. 10-11, 1990, addressed the clinical introduction of new genetic tests, and produced a recommendation for pilot studies to develop professional standards for the delivery of genetic tests for cystic fibrosis. This recommendation has since been adopted by the Director of NIH, and a plan for such studies is being developed. Subsequent workshops are focusing on regulatory and legal approaches to protecting access to and use of genetic information.

Genetic Discrimination Protections

At the ELSI Working Group Workshop on January 10-11, 1991, grantees and consultants identified two profitable avenues for the development of public protections against the unfair use of genetic information:

A. The recently enacted Americans With Disabilities Act has the potential to prevent the abuse of genetic information in the context of employment. The language and intent of the act apparently extends its protections to persons with genetic disorders, persons presymptomatic for genetic disease, and persons who are simply carriers of single copies of disease genes. However, these protections should be strengthened by being explicitly recognized in the regulations developed by the E.E.O.C. for the implementation of this Act.

ELSI researchers are currently surveying state legislation intended to prevent genetic discrimination and developing specific recommendations to the EEOC about how best to incorporate genetic protections into the ADA.

B. The private insurance industry in the process of establishing its own policies with respect to genetic testing and genetic information. It is unlikely that any particular genetic tests would be cost-effective enough to be used as a routine insurance screen. However, as more people become aware of their genetic risks, insurers will have to decide whether to include such information in their underwriting decisions. The Health Insurance Association of American has invited the NIH-DOE ELSI Working Group to work with them in developing their policy, and the Group has established an independant multi-disciplinary Insurance Task Force to examine the issues commercial insurers and their clients face and to develop recommendations for policy.

Much of the health insurance in the U.S. is not provided by commercial firms, but by self-insuring large employers. While representatives of these corporate benefit plans are included on the Task Force, these insurance plans are not regulated by the laws that already govern the activities of commercial firms. In order to increase the reach of these legal protections, the state ERISA legislation could be extended to cover these self-insurance plans. NIH and DOE ELSI researchers are now gathering information and working with state governments towards the development of model legislation in this area.

Genetic Privacy Protections

The privacy and confidentiality of information about individuals' genetic health risks are currently protected by an overlapping network of federal legislation and regulation governing access to medical records, state laws protecting the confidentiality of physician-patient communication, and judicial decisions concerning professional confidentiality standards. The recent Federal Privacy of Genetic Information Bill (H.R. 5612, 101st Congress, 2d Session) appears to duplicate many of the current protections. It is being examined by ELSI researchers for its potential to improve upon them, and will be discussed with other policy proposals in this area at an upcoming Working Group Workshop.

Guidelines for Clinical Uses of Genetic Tests

Determining the protocols that should be considered the professional "standard of care" for the delivery of genetic testing and counseling services is increasingly important as new genetic tests are integrated into mainstream medical practice. To help develop these standards, the NIH and DOE have initiated a two-year study by the National Academy of Sciences/Institute of Medicine designed to assess and propose professional policies regarding the clinical introduction of new genetic tests. At the same time, at the recommendation of the ELSI Working Group, the NCHGR is taking the lead in organizing an NIH-wide initiative to assess the clinical delivery of DNA-based testing for the cystic fibrosis gene through pilot clinical studies.

Social Policy Development and Scientific Advance

One of the lessons of the history of bioethics and science policy-making is that good ethics (and sound policy) requires good facts. In the absence of ongoing scientific research, policy-makers are left addressing scenarios which may have no basis in reality. Thus, those concerned about the "biohazards" of recombinant DNA research in the 1970's were reassured when further scientific research showed such risks to be low. Similarly, those prepared to advocate widespread screening for cystic fibrosis upon the discovery of "the CF mutation," were confounded when further research identified up to 50 other mutations in DNA which could produce the disease. Without ongoing research and the insights of those engaged in it, the foresight that policy-makers require to anticipate the true risks of new knowledge is limited in both directions. By complementing genome research with the development of social policies governing its applications, the human genome project insures that advances in each area help the other to be optimally informed, responsible and beneficial.

NIH-DOE Joint Working Group on Ethical, Legal, and Social Implications of Human Genome Research
FY 1991 Activities**

Introduction

The mission of the NIH-DOE Working Group on the Ethical, Legal, and Social Implications (ELSI) of Human Genome Research is to:

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

Toward these goals, the ELSI Working Group delineated four high priority areas for program activities by NIH and DOE:

1. research on issues of quality and access in the use of genetic tests
2. research on the fair use of genetic information by employers and insurers.
3. research on privacy issues involving genetic information
4. public and professional education

These four areas served as the basis for the NCHGR report which was submitted to Congress, in January 1991, entitled, "The Ethical, Legal, and Social Implications of Human Genome Research: Preparing for the Responsible Use of New Genetic Knowledge." This memo describes the ELSI Working Group's 1991 activities in each of the high-priority areas.

Issues of quality and access in the use of genetic tests

1. **Studies of CF testing and counseling initiated.** An NIH Request for Applications (RFA# HG-91-01) "Studies of Testing and Counseling for Cystic Fibrosis Mutations" (See Tab N) was published in April 1991. Applications submitted against the RFA will be reviewed in early August, and funding is planned to begin at the end of FY 1991.

This RFA was developed in response to the report from the ELSI Working Group's September meeting, which focused on the introduction of new genetic tests. After discussion of this priority research area with clinical geneticists, genetic counselors, industry representatives, and representatives from CF voluntary organizations, the Working Group endorsed the need for studies of CF testing and counseling, so that the implications of CF testing on individuals, families, and society, may be addressed before testing becomes wide-spread.

*Understanding our Genetic Inheritance, The U.S. Genome Project: The First Five Years FY 1991-1995.

The need for NIH supported pilot studies was reported to the NIH Program Advisory Committee on the Human Genome on December 3, 1990. The PAC agreed that the NCHGR should take a lead role in soliciting and supporting proposals for CF pilot studies.

2. **NAS/IOM study commissioned.** The NIH and DOE are co-funding a study by the National Academy of Sciences/Institute of Medicine, "Predicting Future Disease: Issues in the Development, Application, and Use of Tests for Genetic Disorders." This study is aimed at producing professional recommendations for the integration of genetic services into mainstream medical practice. The first meeting of the study panel will take place on July 23-24, 1991.

3. **White paper commissioned.** The ELSI Working Group commissioned a white paper to provide an expert review of the state of research in this priority area, "Cystic Fibrosis Heterozygote Detection: The Introduction of Genetic Testing into Clinical Practice" (Benjamin Wilfond, M.D. and Norman Fost, M.D., University of Wisconsin).

Fairness in the use of genetic testing

1. **Insurance Task Force established.** The ELSI Working Group Insurance Task force, co-chaired by ELSI members Tom Murray and Jon Beckwith, met in May 1991, in Cleveland, OH. The Task force is comprised of representatives from the Insurance industry, corporate benefit plans, consumer and health voluntary groups, and scholars actively researching insurance issues. The group developed a plan of action for developing guidelines for insurance policy by 1993. The next meeting will be held in the Washington, D.C., area, to meet with insurance underwriters and actuaries, to learn about the use genetic data in risk assessment procedures.

2. **White paper commissioned.** The ELSI Working Group commissioned a white paper to delineate policy options in the area of insurance and employment testing, "The Ethical, Legal, and Social Issues Concerning the Use of Genetic Tests by Insurers: Toward the Development of Appropriate Public Policy" (Nancy Kass, Sc.D., Johns Hopkins University)

3. **Statement on Americans With Disabilities Act developed.** In a statement to the EEOC (see attached), the members of the ELSI Working Group identified three areas in which significant changes to the EEOC regulations should be made to improve the ADA's protections against genetic discrimination, and recommended the following amendments to the regulations:

a. discriminatory actions based on an individuals genotype, including the possibility of having affected children, should be covered by the ADA.

b. post-offer, employment entrance medical examinations should be limited to assessing job-related physical and mental conditions.

c. in order to limit access by employers to genetic information in medical records and health insurance claims:

1. an employer should be permitted to require that an employee or applicant authorize his or her health care to respond to specific, job-related questions, rather than sign a blanket release form.

2. rule-making proceedings should be initiated to determine the most effective way of protecting the privacy of health insurance claims. One option is for each employee to have a separate medical claims number and submit claims by number only.

These recommendations will be presented for discussion by the NIH-DOE Program Advisory Committee on the Human Genome on June 25, 1991.

Privacy issues involving genetic testing

1. **Privacy panel.** A panel meeting of experts will convene in Boston on June 28, 1991, to discuss policy approaches to genetic privacy. The Working Group members agreed that the Group needs to become better informed about the implications of the Human Genome Privacy Act, which was recently re-introduced into the House by Representative Conyers.

2. **White paper commissioned.** The Working Group commissioned a paper on the issues of privacy and discrimination, "Genetic Discrimination: Use of Genomic Information to Exclude Persons from Employment, Insurance, and Access to Health Care" (Larry Gostin, J.D., Harvard University)

3. **ELSI Working Group Workshop.** The next ELSI Working Group Workshop, scheduled for September 11-12, 1991, in Washington, D.C., will focus on privacy of genetic information, and continue the discussion of issues raised at the June privacy meeting.

Public and Professional Education

1. **Outreach meeting held.** Leaders of voluntary health organizations and genetic disease support groups were addressed by members of the scientific community and the ELSI Working Group at the NCHGR's first public outreach meeting, in January, 1991. The purpose of the meeting was to encourage these groups to participate in the on-going discussion of the ethical, legal, and social implications of the Human Genome Project. The meeting closed with a request for further discussion of educational issues.

2. **Education Consultation.** As a follow-up to the January Outreach meeting, consultants who have demonstrated expertise in K-12, undergraduate, graduate, professional, and/or public education were invited to an NCHGR Education Consultation, to discuss the state of genetics education in the United States. Plans to work with Alliance of Genetic Support Groups, CORN, and the National Issues Forum, to promote public education were initiated.

TIMELINE OF ELSI ACTIVITIES IN FY 1991

September 1990 — **ELSI Working Group Workshop, Rockville, Md.**
"The Introduction of New Genetic Tests"

ELSI Working Group Accomplishments:
- delineated the four high-priority areas
- stated the need for pilot studies addressing CF testing
- commissioned three white papers to address high-priority areas

January 1991 — **ELSI Working Group Workshop, Crystal City, VA**
Privacy and Confidentiality

ELSI Working Group Accomplishments:
- became informed on the state of legal protections for the confidentiality and privacy of genetic information
- chartered the Insurance Task Force
- received updates on the activities of NIH and DOE grantees

April 1991 — **ELSI Working Group Workshop, Los Alamos, NM**
"Policy Approaches to Genetic Discrimination"

ELSI Working Group Accomplishments:
- recommended amendments to the EEOC regulations of the ADA
- received updates on the activities of NIH and DOE grantees
- requested that a panel to address privacy issues be convened

May 1991 — **Meeting of the ELSI Insurance Task Force**
Cleveland, OH

ELSI Working Group Subcommittee's Accomplishments:
- constructed a plan of action for developing guidelines by 1993
- set a time frame for future meetings
- plan to meet with insurance underwriters and actuaries

**For complete summaries or reports of these meetings and workshops, please contact Eric Juengst, ELSI Program Director, NCHGR.

Activities of the NIH-DOE Joint Working Group on Ethical, Legal, and Social Implications of Human Genome Research
July 1992

Introduction

The mission of the NIH-DOE Working Group on the Ethical, Legal, and Social Implications (ELSI) of Human Genome Research is to:

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- examine the ethical, legal, and social consequences of mapping and sequencing the human genome;
- stimulate public discussion of the issues;
- develop policy options to assure that the information is used for the benefit of the individual and society.*

Toward these goals, the ELSI Working Group delineated four high priority areas for program activities by NIH and DOE:

1. research on issues of quality and access in the use of genetic tests
2. research on the fair use of genetic information by employers and insurers.
3. research on privacy issues involving genetic information
4. public and professional education

These four areas served as the basis for the NCHGR report which was submitted to Congress, in January 1991, entitled, "The Ethical, Legal, and Social Implications of Human Genome Research: Preparing for the Responsible Use of New Genetic Knowledge." This memo describes the ELSI Working Group's 1991 activities in each of the high-priority areas.

Issues of quality and access in the use of genetic tests

1. **Studies of CF testing and counseling funded.** Eight studies were funded from applications submitted to the NIH Request for Applications (RFA# HG-91-01) entitled "Studies of Testing and Counseling for Cystic Fibrosis Mutations."

> Three components of the NIH--the NCHGR, the National Institute of Child Health and Human Development, and the National Center for Nursing Research--have launched a three-year research initiative to define the best methods for educating and counseling individuals who want to be tested for CF mutations. Seven research teams across the country will conduct eight studies to address issues in testing, education, and counseling for the CF mutations. The NCHGR has taken the lead role in supporting these studies. In addition, in order to facilitate communication among the research teams, the principal investigators will meet regularly as a consortium.

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2. **NAS/IOM study.** The NIH and DOE have co-funded a study by the National Academy of Sciences/Institute of Medicine, "Predicting Future Disease: Issues in the Development, Application, and Use of Tests for Genetic Disorders." This study is aimed at producing professional recommendations for the integration of genetic services into mainstream medical practice. The first, agenda-setting, meeting of the study panel took place on July 23-24, 1991. The second meeting, scheduled for February 12-13, 1991, will focus on issues concerning the quality control of laboratory practices.

3. **White paper commissioned.** The ELSI Working Group commissioned a white paper to provide an expert review of the state of research in this priority area, "Cystic Fibrosis Heterozygote Detection: The Introduction of Genetic Testing into Clinical Practice" (Benjamin Wilfond, M.D. and Norman Fost, M.D., University of Wisconsin).

Fairness in the use of genetic testing

1. **Insurance Task Force.** The ELSI Working Group Insurance Task Force, (ITF) co-chaired by ELSI Working Group members Tom Murray and Jon Beckwith, met in May 1991, in Cleveland, OH. The Task force is comprised of representatives from the Insurance industry, corporate benefit plans, consumer and health voluntary groups, and scholars actively researching insurance issues. The group developed a plan of action for developing guidelines for insurance policy by 1993.

The second meeting of the ELSI Insurance Task Force was held in Washington, D.C., where the ITF met with insurance underwriters and actuaries, to learn about the use of genetic data in risk assessment procedures. Before the next meeting, subgroups of the ITF will: (1) devise a mechanism in which the policy implications of existing discrimination cases can be examined, and (2) draft a set of principles concerning the use of genetic tests in insurance underwriting and coverage practices. The next meeting is scheduled for March 23-24 in Boston. The group plans to meet with representatives of the Medical Information Bureau, a database used by insurance companies.

The third meeting of the ELSI Insurance Task Force was held in Bethesda, MD, on March 23-24 (the meeting site was changed from Boston due to restrictions in the NCHGR travel budget). The ITF met with staff of State Insurance Commissioners offices, as well as experts in employment discrimination law and self insured corporate benefit plans. A number of issues were discussed which will form core principles to be addressed in the Insurance Task Force's official report, slated for May of 1993. In addition, the ITF subgroup chartered to address alleged cases of insurance discrimination presented a series of background papers. These papers recognize work that has already been done in this area and lay the groundwork for the tasks of this subcommittee. The next meeting of the Insurance Task Force will take place on May 31-June 1, 1992.

The Task force met again on May 31-June 1. The meeting was devoted to creating an outline of the final report. A subcommittee was formed to research and report on the flow of genetic information from tests to records to insurance companies, and another is

investigating the nature of adverse selection. Adverse selection happens when insurance applicants know they will become ill from a genetic disorder, then conceal the genetic test results and purchase additional insurance at a low premium. The insurance companies will pay out claims to these people at a higher than expected rate. Fear of adverse selection is one of the major reasons cited by the insurance industry for reserving the right to access the results of genetic tests of its applicants. Reports of these subcommittees will be heard at the next meeting of the Task Force, which will be held in San Francisco, CA, November 9-10.

2. **White paper commissioned.** The ELSI Working Group commissioned a white paper to delineate policy options in the area of insurance and employment testing, "The Ethical, Legal, and Social Issues Concerning the Use of Genetic Tests by Insurers: Toward the Development of Appropriate Public Policy" (Nancy Kass, Sc.D., Johns Hopkins University)

3. **Statement on Americans With Disabilities Act developed.** In a statement to the EEOC, the members of the ELSI Working Group identified three areas in which significant changes to the EEOC regulations should be made to improve the ADA's protections against genetic discrimination, and recommended the following amendments to the regulations:

a. discriminatory actions based on an individuals genotype, including the possibility of having affected children, should be covered by the ADA.

b. post-offer, employment entrance medical examinations should be limited to assessing job-related physical and mental conditions.

c. in order to limit access by employers to genetic information in medical records and health insurance claims:

1. an employer should be permitted to require that an employee or applicant authorize his or her health care to respond to specific, job-related questions, rather than sign a blanket release form.

2. rule-making proceedings should be initiated to determine the most effective way of protecting the privacy of health insurance claims. One option is for each employee to have a separate medical claims number and submit claims by number only.

These recommendations were presented for discussion and endorsed by the NIH-DOE Program Advisory Committee on the Human Genome on June 25, 1991. An EEOC representative responded to these concerns, saying that non-job related testing was in fact legal as long as the results of such tests are not used for discriminatory purposes. They also indicated that the ADA does not protect the privacy of medical information. The Working Group is awaiting further clarification of other aspects of the EEOC response.

Privacy issues involving genetic testing

1. **Privacy panel.** At the request of the Working Group members, a panel meeting of experts convened in Boston on June 28, 1991, to discuss policy approaches to genetic privacy. The Working Group needed to become better informed about the implications of the Human Genome Privacy Act, which was re-introduced into the House by Representative Conyers.

2. **White paper commissioned.** The Working Group commissioned a paper on the issues of privacy and discrimination, "Genetic Discrimination: Use of Genomic Information to Exclude Persons from Employment, Insurance, and Access to Health Care" (Larry Gostin, J.D., Harvard University)

3. **ELSI Working Group Workshop.** The September ELSI Working Group Workshop focused on privacy of genetic information, and continued the discussion of issues raised at the June privacy meeting. This discussion included the consumer perspective, as well as privacy of stored biological, computer files, and research data. An initiative on privacy, was chartered to look in depth at privacy protections for genetic data.

4. **Congressional hearing on privacy.** The House Committee on Government Operations held a hearing on October 17 to hear testimony on the privacy of genetic information. Bernadine Healy, James Watson, David Galas, and Nancy Wexler were among the panelists who advocated the need for preserving the right to privacy of genetic information while preventing discrimination against those who choose not to keep their genetic make-up private.

Public and Professional Education

1. **Outreach meeting held.** Leaders of voluntary health organizations and genetic disease support groups were addressed by members of the scientific community and the ELSI Working Group at the NCHGR's first public outreach meeting, in January, 1991. The purpose of the meeting was to encourage these groups to participate in the on-going discussion of the ethical, legal, and social implications of the Human Genome Project. The meeting closed with a request for further discussion of educational issues.

2. **Education Consultation.** As a follow-up to the January Outreach meeting, consultants who have demonstrated expertise in K-12, undergraduate, graduate, professional, and/or public education were invited to an NCHGR Education Consultation, to discuss the state of genetics education in the United States. Plans to work with Alliance of Genetic Support Groups, CORN, and the National Issues Forum, to promote public education were initiated.

3. **Public Forum.** The ELSI working group held its first public forum in Iowa City, IA on April 21, 1992. University of Iowa faculty gave background information on genetics and the significance of Human Genome Project, emphasizing its implications for health

and health policy. Representatives of families affected by genetic disorders, state genetics coordinators, clinical genetics services providers, and clergy testified on issues related to delivery of and access to genetic services, including ethno-cultural sensitivity and personnel numbers and education needs; the need for protection of the privacy of genetic information; and the risk of discrimination on the basis of genotype. Many of those who testified stressed the importance of involving families affected by genetic disorders in the ELSI program activities. The members of the working group were impressed by the breadth and depth of the testimony and discussion that followed. Copies of the written testimony are available on request from NCHGR.

TIMELINE OF ELSI ACTIVITIES

September 1990 **ELSI Working Group Workshop, Rockville, Md.**
"The Introduction of New Genetic Tests"

ELSI Working Group Accomplishments:
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- commissioned three white papers to address high-priority areas

January 1991 **ELSI Working Group Workshop, Crystal City, VA**
Privacy and Confidentiality

ELSI Working Group Accomplishments:
- became informed on the state of legal protections for the confidentiality and privacy of genetic information
- chartered the Insurance Task Force
- received updates on the activities of NIH and DOE grantees

April 1991 **ELSI Working Group Workshop, Los Alamos, NM**
"Policy Approaches to Genetic Discrimination"

ELSI Working Group Accomplishments:
- recommended amendments to the EEOC regulations of the ADA
- received updates on the activities of NIH and DOE grantees
- requested that a panel to address privacy issues be convened

May 1991 **Meeting of the ELSI Insurance Task Force**
Cleveland, OH

ELSI Working Group Subcommittee's Accomplishments:
- constructed a plan of action for developing guidelines by 1993
- set a time frame for future meetings
- plan to meet with insurance underwriters and actuaries

September 1991 **ELSI Working Group Workshop, Bethesda, MD**
"Privacy of Genetic Information"

ELSI Working Group Accomplishments:

- Privacy Task Force chartered
- plan of action for continued discussion of privacy

December 1991 **Meeting of the ELSI Insurance Task Force**
Washington, DC

ELSI Working Group Subcommittee's Accomplishments:

- became informed about the use of genetic data by insurance companies
- plan to draft set of principals by next meeting
- plan to examine existing cases of genetic discrimination

February 1992 **ELSI Working Group Workshop, Bethesda, MD**
"Priorities for Program Initiatives"

ELSI Working Group Accomplishments:

- advised ELSI program staff to address four priority areas: the psychosocial impact of prenatal diagnosis, testing and counseling for the p53 mutation, ethical issues involved in pedigree research, and public and professional education.
- set April 21 as the date for the ELSI public forum in Iowa City, IA
- set September 14-16, 1992, as the date for a workshop of all NCHGR and DOE ELSI grantees.

March 1992 **Meeting of the ELSI Insurance Task Force**
Bethesda, MD

ELSI Working Group Subcommittee's Accomplishments:

- became informed about legislative and regulatory issues concerning genetic testing and insurance
- drafted set of core principals for further discussion and refinement
- wrote set of background papers on past work in this area, existing policy statements, and industry positions on genetic testing.

April 1992 **ELSI Public Forum**
Iowa City, IA

ELSI Working Group Accomplishments:

- heard testimony from persons affected by the introduction of genetic tests into clinical practice
- drafted set of core principals for further discussion and refinement
- wrote set of background papers on past work in this area, existing policy statements, and industry positions on genetic testing.

May 1992 **Meeting of the ELSI Insurance Task Force**
Bethesda, MD

ELSI Working Group Subcommittee's Accomplishments:

- drafted outline of final report
- assigned subgroups to gather information on adverse selection and the flow of genetic information between genetic tests, medical records, and insurance companies