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The Ethical, Legal and Social Implications of Human Genome Research:

Preparing for the Responsible Use of New Genetic Knowledge

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EXECUTIVE SUMMARY

In House Report No. 101-591, the Committee on Appropriations requested that the National Center for Human Genome Research at NIH submit a report that identifies the major ethical and legal implications of human genome research and proposes policy options for addressing potential issues (page 85). The following is submitted in response to that request.

In this report we describe the objectives of the NCHGR program on the ethical, legal and social implications of human genome research, identify three sets of priority issues for the program, describe the plan for addressing these issues, and review current policy options upon which the program will build.

The goal of this NIH program is to anticipate and address the ethical, legal, and social implications of human genome research. The program has given highest priority to three sets of issues:

I. **Privacy of genetic information,** including questions of clinical confidentiality and data management.
II. **Effective Clinical Integration of new genetic testing options,** including questions of quality control and professional liability.
III. **Fairness in the use of genetic information,** including questions of insurance availability and employment screening.

Most of these issues have precedents in other professional and public deliberations from which potential policy options can be drawn for evaluation and development. The program is actively engaged in assessing these options in five different ways:

- Stimulating and coordinating policy development through workshops and commissioned papers identifying needs on high-priority issues.
- Facilitating rigorous research and policy analysis through competitive grants, contracts, and fellowships targeted to high-priority issues.
- Developing educational materials and programs for all levels.
- Soliciting input from the community-at-large through public meetings and testimony.
- Developing collaborative international efforts on the issues through liaison with relevant international organizations.

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INTRODUCTION

In its report on the Fiscal Year 1991 budget for the Department of Health and Human Services, the House Committee on Appropriations stated:

> "The Committee directs the National Center for Human Genome Research to transmit to Congress concurrent with submission of the fiscal year 1992 budget a plan which identifies the major ethical and legal issues raised by this project and proposes policy options to address these crucial issues." (House Report 101-591, page 85)

The following report has been prepared by the National Center for Human Genome Research of the National Institutes of Health, Public Health Service, Department of Health and Human Services in response to this request.

BACKGROUND

Many of the most stubborn health problems facing biomedical science are diseases in which our genes play a role. These include most of the 3,000 known disorders in which genes are the dominant cause, such as cystic fibrosis, sickle cell disease, or muscular dystrophy. But they also include other serious diseases, such as heart disease, diabetes, and cancer, which result from interactions among our genes, environments, and behavior. Therefore, understanding how specific genes function and malfunction has become a high priority all across the health sciences.

A basic resource for that research effort is the information that the Human Genome Project will produce: high-resolution maps of human chromosomes and human DNA sequence data. The genome research programs of the National Institutes of Health (NIH) and Department of Energy (DOE) were established to provide the support and coordination necessary for the scientific effort to produce that resource.

The new genetic information gained from genome research is likely to be useful in developing new diagnostic and predictive tests, in advance of corresponding therapeutic or curative advances. The process of realizing the benefits of such tests, however, will require individual, professional, and public deliberations over an important set of ethical and social questions--questions about the responsible use of new genetic information.

To ensure that information generated by human genome research can be of maximum benefit to individuals and society, the NCHGR established its Program on the Ethical, Legal and Social Implications of Human Genome Research to address these questions together with the DOE's activities in this area. This is a landmark opportunity to complement scientific progress with sound social policy-making.

PROGRAM OBJECTIVES

The goal of the NCHGR's program on ethical and social issues is to develop the safeguards that will be required as new genetic information is put to practical uses. In the NIH-DOE five year plan for the Human Genome Project, presented to Congress in 1990, four specific objectives were identified as critical to achieving that goal:

- Clarifying the ethical, legal, and social consequences of mapping and sequencing the human genome, through a program of targeted research.
- Developing policy options at professional, institutional, governmental and societal levels to ensure that genetic information is used to benefit individuals and society.
- Improving understanding of the issues and policy options, through educational initiatives at public, professional, and policy-making levels.
- Stimulating public discussion of the issues and policy options.

PRIORITY ISSUES

Over the next five years, these objectives and the activities they involve will be targeted to three sets of high-priority concerns:

I. **Privacy of genetic information**

II. **Effective clinical integration of new genetic testing options**

III. **Fairness in the use of genetic information**

All these issues involve policy questions that will continue to attend new advances in human genetics quite apart from the Human Genome Project. As genetic research accelerates, the range of health problems, health professionals, and patients affected by these questions will broaden. The increased complexity of that range often means that older answers to these questions must be reenforced, refurbished or reconsidered to achieve their intended aims. The NCHGR's activities addressing these questions are designed to catalyze and coordinate that process, to enable a sound and authoritative set of policy alternatives to be produced.

I. PRIVACY OF GENETIC INFORMATION

One way to help prevent information from being abused is to control its accessibility. Because genetic information is highly personal, its privacy is especially important to protect. But genetic information almost always has implications for other people's welfare as well: spouses, children, and extended-family members. The legitimate interests of insurers, employers, and researchers can also be affected. As a result, the appropriate ethical and legal limits of such protections are still unclear.

Thus, one of the NCHGR's highest priorities is to develop sound policies governing the confidentiality of genetic test results.

NCHGR Program Activities

Research: NCHGR has already initiated research projects addressing several facets of the privacy issues, including:

- The legal status of genetic information.
- The information-sharing practices of biomedical researchers.
- The professional ethical issues involved in maintaining confidentiality within families.

Policy Development: The program plans to host two policy-development meetings to address important facets of the confidentiality issues in FY 1991. Both of these meetings will yield policy analyses and recommendations:

- A conference to assess public policy options with respect to the protection of the privacy of genetic information, including proposed federal legislation.
- A conference to develop consensus within the biomedical research community on confidentiality practices in genetic research.

Policy Options and Precedents

In helping to develop policy on this issue, the NCHGR can build on deliberations over questions of privacy and equal opportunity from other spheres. Options include:

1. **Federal legislation** designed to protect the confidentiality of genetic information about individuals by defining information

management standards and information access procedures. Examples of relevant legislative initiatives include the Federal Privacy of Medical Information Act of 1980 (H.R. 5935, 96th Congress, 2d Session), and the recent Federal Privacy of Genetic Information Bill (H.R. 5612, 101st Congress, 2d Session).

2. **Federal research regulations** designed to control access to genetic information about identifiable individuals involved in the development and evaluation of new genetic tests. The most immediate population to be affected by new genetic tests will be the volunteers involved in their development. Precedents for protecting their privacy include the Federal legislation governing confidentiality of drug and alcohol abuse patient information (42 U.S.C. 290dd-3 and 290ee-3 and regulations at 42 C.F.R. Part 2), as well as Federal legislation providing for grants of confidentiality protection for health research (42 U.S.C. 241(d) and regulations at 42 C.F.R. Part 2A) and controlled substances research (21 U.S.C. 801 and regulations at 21 C.F.R. 1316.23).

3. **State legislation** designed to strengthen and clarify the limits of physician-patient privilege with respect to genetic information and otherwise control use of this information. States have traditionally helped to regulate physicians' ethical commitment to professional confidentiality through testimonial privilege statutes. New York's recent efforts to define the limits of medical confidentiality in the context of HIV testing may become relevant background as new genetic tests enter clinical practice.

4. **Professional standards** within the biomedical community governing the storage, retrieval, use, and disclosure of genetic information about identifiable individuals.

However, the ramifications of extending current policy to the case of genetic testing still require careful research and wide discussion. The thrust of the program's work over the next five years will be to analyze these policy options for their appropriateness to the case of genetic testing.

II. EFFECTIVE CLINICAL INTEGRATION OF NEW GENETIC TESTING OPTIONS

Human genome research is expected to increase greatly the number of gene-based diagnostic and prognostic tests available to health professionals. As a result, the diagnostic capabilities of clinicians will advance more rapidly than their ability to cure or treat detectable genetic disorders. Without proper guidance, the widespread clinical introduction of these new tests could lead to their misapplication and misinterpretation by professionals and patients alike.

The challenges involved in integrating those tests safely and effectively into clinical practice include:

- **Ensuring equal access to adequate education and counseling for patients.**
- **Establishing minimum qualifications for clinicians conducting tests.**
- **Assuring quality control for genetic tests.**
- **Establishing guidelines for genetic testing programs.**
- **Defining ethical and legal responsibilities of clinicians who perform tests and the rights of patients who undergo them.**

NCHGR Program Activities

Research: Much of the program's initial efforts will be devoted to exploring these clinical impact questions. Currently active projects include studies of:

- The adequacy of genetic knowledge and skills among American primary care physicians.
- The impact of new genetic tests on the professional norms and standards of genetic counselors.
- Commercial, legal, and professional influences on the development and use of new genetic tests.
- The impact of increased genetic services on public attitudes toward genetic risks.

In addition, a Request For Applications is being developed that will solicit specific research efforts aimed at answering the key factual and regulatory questions involved in the clinical integration of new genetic tests and services. Research proposals under this request will be evaluated and awarded during FY 1991.

Policy Development: These issues pose policy questions for those charged with improving public access to genetic health care services, regulating the development and use of new genetic tests, and establishing standards of practice within the health professions. The NCHGR's policy development activities with this issue are targeted accordingly, and include two professional workshops aimed at defining guidelines and professional policy needs with respect to the introduction of new genetic tests.

The first of these workshops, held on March 5-6, 1990, produced an influential report recommending against the routine use of new tests for cystic fibrosis for population screening. The second, held September 10, 1990, focused on cystic fibrosis testing as a starting point for identifying research and policy development needs. Complementing these workshops will be:

- A major study by the Institute of Medicine, aimed at producing professional recommendations for the integration of genetic services into mainstream medical practice.

- A working conference in 1991 to enable FDA staff to meet with geneticists to develop regulatory standards for new gene-based diagnostic tests.

Policy Options and Precedents

Again, in attempting to anticipate these questions, policy options at several different levels will be analyzed. Examples include:

1. **Federal legislation** expanding federal health care coverage to include payment for genetic counseling and education by appropriately qualified health workers, or providing incentives to private insurers to include such coverage.

2. **Federal regulations** establishing evaluation and quality control standards governing the development, manufacture, and marketing of new genetic tests.

3. **Professional policies** defining professional qualifications, testing program guidelines, and professional ethico-legal responsibilities for health professionals performing and interpreting tests.

III. FAIRNESS IN THE USE OF GENETIC INFORMATION

Discrimination based on genetic risk information can occur in two ways:

- Genetic risk information may be used unfairly to deny otherwise-eligible individuals opportunities for adequate health care coverage, life insurance, or employment.

- Genetic risk information may stigmatize individuals socially if it is misinterpreted by patients, their families, and the public.

Defining the acceptable role of genetic risk information in determining access to health care or employment is particularly important because of the legitimate interests at stake for both families at genetic risk and private insurance providers and employers. Moreover, the risks to both sides broaden as the genetic elements of more health problems are uncovered and gene-based tests for susceptibilities and carrier states are developed. At the same time, the primary problem that health professionals will face in the clinical use of genetic tests is the misinterpretation of their findings and the potential for psychological trauma, stigmatization, and social discrimination that can result.

Thus, one of the program's priority concerns is to determine how to best protect the social equality of tested individuals in the face of the biological diversity that genetic tests will uncover.

NCHGR Program Activities

Research: In order to combat genetic discrimination, more needs to be known about the social meanings of genetic risk, the practices of families, insurers and employers, and the relevance of current anti-discrimination protections. Projects already underway include studies examining:

- The technical prospects and legal limits of genetic testing for insurance and employment screening practices.

- The relevance of current anti-discrimination regulations for genetic discrimination.

- The implications of human genome research for international social policy issues, such as immigration policies, information-sharing, and cooperative health programs.

- The implications of human genome research for our cultural understanding of health, disease, and disability.

Policy Development: Many of the policy questions posed by the program's concern with genetic discrimination are questions concerning those with potential interests in using genetic information: most notably, insurance providers and families at risk for genetic health problems. To help address these issues at that level, a task force has been established to complement the expertise of the NIH-DOE Working Group with the expertise required to address the insurance-related issues adequately, including representatives of families at risk for genetic disease and the insurance industry. Starting in FY 1991, this task force will gather information, convene public meetings, and produce policy options.

Policy Options and Precedents

There is a range of public experience to build upon in considering policy approaches to protecting individual beneficiaries of genetic testing against insurance and employment discrimination. Policy options include:

1. **Federal legislation** designed to protect the insurance opportunities of individuals at genetic risk for health problems. For example, the Employee Retirement Insurance Security Act could be amended to extend existing regulations to self-insuring employers and pre-employment screening practices.

2. **Federal regulation** aimed at reducing employment discrimination on the basis of genetic risk information. For example, the recently enacted Americans with Disabilities Act provides protections for those already disabled or perceived to be disabled by genetic disease. It and other federal anti-discrimination initiatives such as the Rehabilitation Act of 1973 should also protect those known to be at risk for a genetically detectable health problem.

3. **State legislation** aimed at limiting the degree to which private insurers may classify risks, or limiting the kinds of risk information that insurers may solicit from applicants, or setting standards for insurance screening. Each of these options has precedents to study among different states' responses to the insurance problems raised by HIV testing during the 1980's.

4. **Professional and corporate policies** designed to address these issues within the appropriate industries and professional communities. For example, both the health insurance and life insurance communities have established internal working groups to assess the underwriting utility of genetic testing and to develop industry-wide guidelines for its use.

PUBLIC DISCUSSION AND EDUCATION

Ultimately, the best ways to decrease the risk of genetic discrimination and increase the beneficial uses of new genetic knowledge are to **promote better understanding of human genetics** and to **involve the public in deliberations over its use.** NCHGR is committed to an active program of public discussion and education and supports two important national projects aimed at improving professional and public understanding of genetic tests and their implications:

- **A series of community-based discussions** is being designed to provide the public an open forum for expressing their concerns and to improve public understanding of genome research. This series will begin in the Spring of 1991 and will involve meetings across the country.

- **A ten-part public television series** on "The Future of Medicine," slated to air in the 1992-93 season, will elucidate the clinical impact of genome research for a public audience.

CONCLUSION

As the biomedical sciences mature, biomedical scientists' social responsibilities also grow. Scientists work increasingly with policy-makers, health care providers, and the public to anticipate the human implications of the knowledge they provide. Over the past twenty years, professional and public discussions of these implications have become an integral part of the biomedical research process. Their role has been to help optimize the benefits to human welfare and opportunity that new knowledge can provide and to guard against its misuses. Over the next twenty years, advances in human genetics, accelerated by the Human Genome Project, will feature prominently in these discussions. By pursuing the study of the ethical, legal, and social implications of genome research in concert with their scientific initiatives, the NIH and DOE genome research programs take up their responsibility to help make those discussions as timely, well informed, and productive as possible.