Statement of

Nancy S. Wexler, Ph.D.

Chairperson

Joint NIH/DOE Working Group on the

Ethical, Legal and Social Issues

of the Human Genome Project

President

Hereditary Disease Foundation

Columbia University

Departments of Neurology and Psychiatry

before the

United States House of Representatives

Committee on Government Operations

Subcommittee on Government Information, Justice and Agriculture

October 17, 1991

INTRODUCTION

I am honored to be here this morning. You are to be congratulated for focussing the nation's attention on matters fundamental to each of us as individuals and as citizens, that is, protection of the privacy of medical and, particularly, genetic information. I would like to talk with you today about the rights of persons for the maintenance of privacy of genetic information. But I would also like to speak to the essential rights of persons to disclose genetic and medical information with impunity, without fear of harmful reprisals. And I would like to address the rights of individuals to choose whether or not to preserve their privacy or speak openly without negative repercussions.

I have the privilege of serving as Chairperson of the Joint NIH/DOE Working Group on the Ethical, Legal and Social Issues associated with mapping and sequencing the human genome. I am also speaking to you today as the President of the Hereditary Disease Foundation, and as Associate Professor of Clinical Neuropsychology in the departments of neurology and psychiatry, College of Physicians and Surgeons, Columbia University.

I have had personal experiences with respect to issues of genetic privacy and disclosure which I think are relevant. My mother was diagnosed with Huntington's disease when I was 22 years old. The illness had already claimed the lives of my maternal grandfather and three uncles. My father explained to my

older sister and me that we each had a 50/50 risk of inheriting it. The disease causes uncontrollable movements in all parts of the body, intellectual deterioration and severe emotional disturbances. It is invariably fatal and essentially untreatable.

I went to graduate school immediately after learning of this upheaval in our lives. Although I had done nothing to bring this on, I was at first embarrassed and ashamed to tell anyone about my mother's sad decline or my own risk. I was afraid people would treat me differently, watch me for symptoms, not want to date me, be overly distant or too solicitous. In graduate school I became involved with working with families with Huntington's disease but for some time I kept that world and my academic life quite separate. It felt slightly schizophrenic, literally commuting between my world of families with Huntington's disease in Detroit and my academic life in Ann Arbor, Michigan. Thanks to exceptionally understanding and wise faculty and friends at the University of Michigan, my two worlds meshed in very gratifying ways. But when I applied for an academic position after completing my doctorate, I asked all my advisors to rewrite their letters of recommendation because, although they had said nothing explicitly, I was afraid that it would be too obvious why I was interested in Huntington's disease. I was very concerned that I would never be hired if my risk status was known, and certainly never be considered for tenure.

The turning point came for me when the National Institute of

Neurological Disorders and Stroke invited me to serve as Executive Director of the Congressional Commission for the Control of Huntington's Disease and Its Consequences and later to join their staff. For the first time I could be totally open about the disease without fear of alienating my colleagues or losing my job security and employment benefits. The Neurology Institute was even courageous enough to offer me a civil service job from which it is difficult to extract people. This experience was enormously healing because my colleagues, experts in the disease, were willing to take a chance on a person with a one in two possibility of developing a neurodegenerative disease of the brain and body, with insidious onset, causing failures of judgment and memory and emotional instability. It was not privacy in this instance that was necessary - it was candor that cured.

The leadership of the Human Genome Project, both at the NIH and the DOE, were also willing to place a representative of genetic "consumer" groups as chair of their joint Ethical, Legal, and Social Issues Working Group. While many in genetic support groups have trepidations regarding the utilization of genetic information, the fact that the NIH and DOE have launched the biggest biomedical ethics program nationwide concomitantly with support of the basic research has helped to allay peoples' concerns. Much needs to be done, but at least there are resources available to do it thoughtfully, with careful planning, and there is interest at the NIH, DOE, and most importantly, on

Capitol Hill.

Let me emphasize that although no formal poll has been taken, the vast majority of families with genetic disorders are enormously grateful for advancing genetic technology. To us, it represents the best hope for an effective treatment and even eventual cure. Some in government or the general public have suggested slowing down the technology until the social support systems "catch up." This is antithetical to the best interests of patients and families as technology is the only hope that many have who are in a race against time before the effects of a lethal gene overtake them. When treatments exist, there will be fewer incentives to discriminate and the burden of revelation will diminish.

THE PATHWAY BETWEEN PREDICTION AND PREVENTION

It is important to understand the stages and time course by which most research on genetic disease progresses. First DNA markers must be identified which are localized precisely along a chromosome so that each is a small and specific distance from the other, like distance markers along a highway. Then, by studying families with a particular genetic disease and watching the disease gene being passed from generation to generation together with certain markers, the disease gene is "mapped" to a specific chromosome. Once close markers are discovered, presymptomatic and prenatal diagnosis is possible -- even decades before the

appearance of the illness. But the aim of the research is primarily therapeutic, not just diagnostic. Early diagnosis can be a benefit in many instances in which early intervention is critical, such as cancer or heart disease, or problematic if there is no prevention possible, such as for familial Alzheimer's disease.

The next step is the isolation and characterization of the abnormal gene itself. This phase can be rather rapid, a year or two, or can take a very long time. The HD gene was localized in 1983, the first instance in which DNA markers were successfully used to map a gene whose chromosomal assignment was unknown. Eight years later, we are still searching for the gene. The Human Genome Project will dramatically shorten the time it takes to find and characterize genes; at the successful conclusion of the Project one will only need to look it up in a book.

Observing altered genes and studying homologous genes in plants and animals can give rise to suggestions for therapy. Scientists are now exploring novel delivery systems using inhalants such as those used for treating asthma to place normal genes in the lungs in order to treat cystic fibrosis or alpha-1 antitrypsin disease. Some scientists are capitalizing on the affection which cold viruses, adenoviruses, show for the lung to harness them into the service of transporting normal genes in to correct damaged lungs.

The Human Genome Project greatly expedites finding and characterizing normal and abnormal genes in human and model

organisms and develops new technology that facilitates gene therapy. Gene therapy cannot be approached until the offending gene is identified and understood. Although understanding the molecular lesion is no guarantee of a cure, a sad lesson learned from sickle cell research, many new avenues are surely opened.

Another advantage of the Human Genome Project is that once critical genes modulating important biochemical or cellular pathways are discovered, they may be found to play a role in both the hereditary and sporadic forms of an illness. For example, both familial and sporadic forms of Alzheimer's disease may involve a disruption of the beta-amyloid gene. Treatment for the much more common sporadic form may be the same as treatment for the familial variant. Neurofibromatosis, retinoblastoma, colon and breast cancer all may involve disruptions of oncogenes, which produce tumors, or tumor suppressor genes. An environmental insult may work its effect by disrupting the action of these same genes in a cell, affecting somatic genes rather than germ cell genes. Although the disorder produced is not <u>hereditary</u> in that it is not passed on to the next generation, it is still <u>genetic</u> in that the functioning of a single or possibly multiple genes are affected. New treatments for cancer, for example, may address correcting the damaged gene, even though a noxious chemical might be the instigating agent of the disease.

Even some people's differential response to the AID's virus has been shown to be genetically mediated. Learning how these people resist the illness may lead to new therapeutic

understanding. Transplant rejection occurs due to genetic incompatibility between host and donor; if the actions of those genes could be modified, transplant success rates could improve dramatically. Of the approximately 100,000 genes that humans possess, probably the vast majority exist in some altered state through the effects of mutations through the millennia and these disordered states contribute to today's morbidity and mortality. The Human Genome Project should set the stage for the development of extensive new therapies both for disorders known to be hereditary and even those that are not. The benefits to human health are incalculable.

The risks are to some extent known. Dangers posed by genetic knowledge challenge systems already strained in our society today. These problems preexisted the Human Genome Project and they are too extensive to be ameliorated by our efforts alone. But the Human Genome Project can contribute toward their solution by convening and supporting some of the brightest and most creative thinkers to focus their efforts on devising policy and program recommendations.

THE JOINT NIH/DOE ETHICAL, LEGAL AND SOCIAL ISSUES WORKING GROUP

The mandate of the Joint NIH/DOE Ethical, Legal and Social Issues Working Group of the Human Genome Project is to anticipate problems attendant on this burgeoning technology and make programmatic and policy recommendations to assure that

information is used for the benefit of individuals and society. The National Center for Human Genome Research, National Institutes of Health, program on Ethical, Legal and Social Implications (ELSI) is directed by Dr. Eric Juengst, a philosopher trained in ethics with experience both in genetics and in the humanities. The ELSI program of the Department of Energy is directed by Mr. Michael Yesley, a lawyer who served as staff director of the National Commission for the Protection of Human Subjects in Biomedical and Behavioral Research and who has long-standing experience with issues of privacy and confidentiality. The joint NIH/DOE ELSI Working Group serves in an advisory capacity to the Human Genome Programs of both parent institutions. It is comprised of experts in medical genetics, ethics, law, biology, psychology and sociology. In each of its quarterly meetings focussing on different topics of interest, additional experts in diverse areas are convened.

The first priority of the Working Group is to facilitate the distribution of grant funds to support investigators and policy makers in the larger community. The testimonies of Dr. Bernadine Healey, Director of the NIH, and Dr. David Galas, Associate Director for Health and Environmental Research, DOE, cover the details of these programs. Certain areas are of such high priority that the Working Group itself has taken the initiative to develop programs in these areas. The ELSI Working Group enunciated four areas in which immediate attention is required:

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

REPERCUSSIONS OF DISCLOSURE -- INSURANCE COVERAGE

In the United States, with our current system of insurance coverage, unwanted disclosure of genetic information can result in the loss of critical health benefits, not only for an individual but for his or her entire family. It would be a bitter irony if people who can benefit from early diagnostic tests are dissuaded from availing themselves of the test because they may loose the very insurance they need to prevent the disease or protect themselves from it when it appears.

The medical community and the public were elated recently when two scientific groups announced the discovery of a gene causing polyposis coli or colon cancer. The early diagnosis of colon cancer can be life saving. If persons who carry one copy of this autosomal dominant gene have a colectomy before symptoms start or remove colon adenomas which are the first signs of developing disease, they can very effectively prevent colon cancer. If they cannot afford to pay for the genetic test, if third party carriers refuse to reimburse for it, or delete

coverage of the necessary preventive procedures, the advantages of early diagnosis will be for naught. The families who participated in research that led to these breakthroughs in gene identification, families in whom it is already known that this will be an effective and accurate test, are now cautiously weighing their options.

If you think that this is a fictitious concern, consider the following. A woman in Michigan had two siblings affected by colon cancer. She wisely had a colonoscopy as a prophylactic measure the results of which proved to be perfectly normal. She later applied for a health insurance policy. Although neither she nor any of her physicians discussed the family history of colon cancer with the insurance company, they somehow acquired this information. When her new policy arrived it contained a rider excluding all coverage of procedures relating to her colon. If she should develop colon cancer in the future, she would have to assume all treatment costs herself -- an impossibility. She cannot even afford to pay for the colonoscopies which might minimize the cost of any cancer through early detection.

Although her own genetic concerns should have been sufficient concern for one individual, this same woman married a minister who later developed Huntington's disease. The fatal HD gene was passed on to both their children who manifested the illness at a young age. Huntington's disease has a gradual onset. The neurologist diagnosing the illness in the son waited until he was age 19 to spare him the devastating news. What

doctor and mother did not realize was that had the son been diagnosed at age 18, when he was clearly symptomatic, he would have been covered as a dependent of the mother's on her insurance policy. As it was, he was independent but already ill and totally uninsurable. The mother's policy, although it had extensive mental health benefits for family members, was useless to her because Huntington's disease was defined as requiring custodial care and no amount of insurance would cover such care. This is often the situation confronting families with hereditary diseases that are chronic in nature. The mother was faced with the prospect of state hospital placement for her husband and son, both of whom were totally dependent on her modest earnings for their support.

The Insurance Task Force of the joint NIH/DOE Ethical, Legal and Social Issues Working Group is addressing critical questions with respect to how the insurance industry responds to the introduction of new genetic tests. Their recommendations, due in 1993, should help shape policy and practices in these areas, rather than merely being reactive to industry stances. (The Task Force is focussing on health insurance alone. Most people with or at risk for hereditary disorders are ineligible for life insurance.) As more new tests are developed, both for illnesses considered to be strictly inherited and for those in which some cascade of genes may play a role, public awareness of insurance industry practices will be heightened. An increasing number of individuals who took their insurance coverage for granted may

find themselves among the group whose insurance is in jeopardy.

At the moment, some individuals taking genetic tests are paying for them out-of-pocket to avoid any potential insurance repercussions. Presymptomatic testing for Huntington's disease is a "linkage analysis," studying DNA markers "linked" or very close to the HD gene to make a diagnosis. It is more accurate with more persons in the family tested using more markers. Some persons are paying up to $4,000 for testing; the expenditure is worth it to them to keep the information private. It is unclear if third party carriers would pay for the test and what actions they would take following a diagnosis of either positive or negative for the gene.

The insurance companies, in turn, are concerned that the public will pay personally for genetic tests and then alter their insurance coverage according to the outcome: increased insurance if health problems are predicted, decreased insurance for those shown to be healthy. This "adverse selection" might skew actuarial calculations in a deleterious fashion.

Some insurance companies are beginning to propose new programs for pooling risks across a large number of small employers and providing for more universal coverage, regardless of risk. They recognize that according to their current criteria, a large segment of the American public may become uninsurable with the advent of new genetic tests. Certainly the aim of the Human Genome Project is not to swell the ranks of the 37 million uninsured in this country. If, however, under the

auspices of the Human Genome Project representatives of the insurance industry, of consumer groups, academia, government and the public can meet to develop new policies and programs we can help to catalyze constructive changes for a much larger constituency.

REPERCUSSIONS OF DISCLOSURE -- EMPLOYMENT

Issues of insurance are integrally enmeshed with fears regarding employment discrimination. With health care costs soaring, there is a strong economic incentive for employers to screen out individuals who will develop costly illnesses. In some instances, the employee may be perfectly healthy but be a carrier of a deleterious gene that might combine with the same defective gene in a spouse to produce a devastating disease in offspring.

A recent report by the Office of Technology Assessment indicated that 12 companies among the Fortune 500 and 50 major utility companies reported using any form of genetic screening or monitoring in the workplane. Although the number is small, it may be because the full ramifications of genetic testing and its consequences have not seeped into public awareness sufficiently to entice employers. The attitude of insurance carriers toward genetic testing may help shape those of employers.

The recently passed Americans With Disabilities Act (ADA) forbids discriminating against qualified individuals with a

disability with respect to job application procedures; hiring, advancement and discharge practices; and compensation. The Act provides extremely important protections against employment discrimination for those either disabled by, presymptomatically identified to have, or asymptomatic carriers of a genetic disease. Taking its cue from the seminal Vocational Rehabilitation Act of 1974, the ADA protects three groups of individuals defined as disabled. The first definition, persons with a physical or mental handicap that substantially limits major life activities, will protect persons currently disabled due to a genetic illness. The second definition, those with a history of such an impairment, should protect persons with a treatable hereditary disease who may have been incapacitated but are now quite functional, such as persons with PKU, Gaucher's disease or hemochromatosis. The third definition of disability protects a group of people new to our citizenry but growing: the presymptomatic individual. The third definition includes persons regarded to have a such an impairment. This third definition should protect me against employment discrimination if an employer chooses not to hire me only because I am at risk. If I took a presymptomatic test for Huntington's disease and was shown to most likely have the gene (the test is not 100% accurate yet), my employer and I would know that the disease will at some point appear. And yet, as I am still functional and asymptomatic, my employment assessment should focus only on whether or not I can

do the job, not on the fact that some day I will no longer be able to function.

The ADA, and the EEOC interpretations of it as set forth in their regulations, specify that employers may only use job-related medical criteria in hiring decisions. Under the EEOC regulations, employers are permitted to perform a variety of medical tests once an offer of employment has been made, conditional on the outcome of a medical examination. Although employers are not legally restricted in the tests they request, they are only entitled to use job-related medical information.

The Joint ELSI Working Group and the Chairmen of the NIH and DOE Genome Advisory Committees wrote to Mr. Kemp, Chairman of the EEOC, requesting that permissable medical examinations be limited to assessing only job-related physical and mental conditions. We felt that even though employers were only entitled to utilize job-related medical information in their hiring practices and even though employment offers contingent on medical information must depend only on job-related medical results, permitting employers to perform any and all tests encourages surreptitious testing. If an employer legally cannot utilize certain medical information, why permit the employer to gather that information? Why would an employer pay to perform non-job-related medical testing, not at the voluntary request of the applicant or employee, if the information cannot be legally utilized in hiring decisions? To what use will that information be put?

The letter from the NIH/DOE ELSI program to the EEOC also

THE PRIVACY AGENDA

The ELSI Working Group has established a Privacy Task Force under the direction of ELSI member Ms. Patricia King, professor of law at Georgetown University, who has a long and distinguished career of government service as a member of the National Commission for the Protection of Human Subjects in Biomedical and Behavioral Research, and the National Institutes of Health Panel on Human Fetal Tissue Transplantation Research, among many positions. The Privacy Task Force is just being organized and setting its agenda. Among the areas in which research and policy recommendations are sought are the following:

Genetic Services

The ELSI supported Institute of Medicine study on "Predicting Future Disease: Issues in the Development, Application and Use of Tests for Genetic Disorders" will also focus attention on these most crucial questions of privacy and disclosure in the provision of genetic services. Genetic information, unlike most other medical information, immediately reveals private and personal information about others connected to the individual in question. For example, if you learn that my mother had Huntington's disease, you immediately know something intensely personal about my sister and me without our saying anything about ourselves. When I took my mother to a new doctor one day, he said to me, "Oh, Huntington's -- you have a one in

two chance of having it too, no?" He had no idea what I knew or didn't know.

There are controversies brewing within families which may spill over into courts with respect to ownership of genetic information. In certain genetic disorders for which close markers have been found but the gene not yet isolated, linkage tests using these closely linked DNA markers are the only means of providing diagnostic information. DNA samples from specified relatives are required. Linkage tests are now being used for presymptomatic diagnosis of Huntington's disease and polycystic kidney disease, among others. In some instances, parents have refused to give a blood sample for a test on the grounds that the counseling provided at a certain center was inadequate. In other instances, parents wished to provide genetic information for one offspring but not another. Once the information was given for the first, however, it was already known for the second. Could the information be used without a parent's permission to honor the request of a person at risk to learn his or her own genotype or should the request for the privacy of genetic information on the part of the parent be honored? The privacy of one person can be detrimental to the autonomy of another.

Another instance in which privacy may be violated is when a physician determines that there is a serious danger to others based on their relationship to someone with a known genetic problem. Not infrequently, the genetically affected individual requests that no one know the news, including prospective

recommended that the ADA specify that unaffected individuals who are heterozygous carriers for a gene causing a recessive (or X-linked) disease, that is carriers of one copy of a gene which, when two copies are present, causes disease in offspring, be explicitly protected under the Act. An example would be a person carrying one gene for sickle cell disease or cystic fibrosis. The carrier is asymptomatic. An employer may be tempted to discriminate, however, because if a carrier employee has a child with another carrier, each offspring has a one in four chance of having an expensive disease. (The annual cost of medical care for a patient with CF is about $20,000. Lifetime medical costs, costs based on a median lifespan of 27 years, are approximately $500,000.) The carrier rate for cystic fibrosis among Anglo Saxons is 1 in 25, while 8% of Afro-Americans carry the sickle cell gene. We are speaking of common disorders.

The ELSI program also recommended that the ADA deal with the privacy of genetic and medical information as a way to protect employees against discrimination. When insurance claims are made, usually an entire chart arrives in the company benefits office for other employees to peruse easily. Medical records are not "sanitized" so that only relevant material travels to other medical referrals or to benefits offices. We were informed by staff of the EEOC that our recommendation with respect to privacy exceeded the scope of the ADA.

marriage partners or siblings and cousins who may be equally at risk. The client is too ashamed or embarrassed, frightened or distraught by the information to inform others or even to allow the physician or genetic counselor to do so. The President's Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research in their Report on Screening and Counseling for Genetic Conditions discussed criteria for determining when patient confidentiality should be honored or abridged. Although complying with clients' desires for privacy, the report enumerated circumstances in which geneticists could responsibly contact relatives or others whom they deemed needed to know genetic information without permission of the client. This can occur when all efforts at persuasion of the initial client fail and if the condition in question poses a serious harm, such as an unbalanced chromosomal translocation with a high likelihood of resulting in severe disabilities in any child born with the genetic problem.

This problem of "contact tracing" to borrow a phrase from the public health model of infectious disease control, is likely to become more prominent as additional genetic tests are introduced. If about one in 25 caucasians carries an abnormal gene causing cystic fibrosis, once one family member is identified there will be an incentive to find the rest. But some individuals may prefer not to know if they are carriers and their wish for privacy must be respected.

Knowing and Not Knowing: Co-equal Rights?

Western culture is one in which knowledge is valued highly -- "knowledge is power," and to avoid knowledge is looked upon pejoratively -- "to hide your head in the sand". And yet, new genetic knowledge has enormous repercussions for individuals and families. In some instances, predictive testing exists for diseases for which there is no treatment or cure, such as Huntington's disease or neurofibromatosis. We can tell people that they will surely die of HD but we cannot tell them when the disease will appear. We can tell people that their children will have neurofibromatosis but we cannot tell them how severely the children will be affected, ranging from a few large "freckle"-like patches called cafe-au-lait spots to numerous disfiguring tumors all over the body.

For many who are at risk for untreatable late onset diseases, such as familial Alzheimer's disease or amyotrophic lateral sclerosis (ALS or Lou Gehrig's disease), the availability of presymptomatic testing forces them to contend with issues of timing. Should they know prematurely, before the disease begins or wait until symptoms start? Do they have the luxury of choosing for themselves or are there instances in which others require this knowledge? What if you were to know for certain that I am going to develop Huntington's disease. Would my university be reluctant to hire or promote me? Would Drs. Watson and Galas have entrusted this important committee to my leadership? If I were training to be a neurosurgeon, would you

feel it your ethical duty to persuade me to pursue a profession in which coordination, dexterity and judgment, lost early in HD, were not at a premium? Are there instances in which employers have a right to know genotypes or should they judge only on proficiency? If you knew for certain that I was going to develop Huntington's disease, would you feel any differently toward me now?

Testing Minors

Privacy issues can be particularly complex when one party is not able to provide informed consent to the abrogation of privacy. The current policy of testing centers for Huntington's disease, and probably for other late onset disorders as tests are developed, is not to test minors who are unable to provide informed consent for themselves. This policy defies common practices described by family law in which parents are entitled to medical information with respect to their minor children.

The clinicians and families developing the Huntington's disease testing protocol felt it was too onerous a burden for children to carry, knowing they are destined to die of Huntington's disease, and there is no medical advantage to knowing this information early. Testing centers also have refused to test children awaiting adoption, either at the request of prospective adoptive parents or the agency. One day, however, parents who feel justified in invading the privacy of their children for planning purposes may bring the issue to court.

Research on Large Families

One of the first activities of the ELSI Privacy Task Force will be to focus on the protection of privacy in the conduct of genetic research. Now that investigators are successfully localizing genes, they are faced with questions regarding recontacting individuals within the families participating in the research to inform them of their genotype, if they request it. Much of the confusion over recontacting could be alleviated by discussing the issue of providing feedback before the research begins, even specifying in informed consent forms whether and how such recontacting should take place. Some individuals will participate in the research only under conditions of anonymity and do not want to know their genotypes; others explicitly desire to know. Both should be accommodated.

In France recently, an interesting conflict occurred between a scientific group that had acquired information, through geneological studies, regarding which children in a small region were particularly at high risk for developing hereditary juvenile glaucoma. This genetic condition must be treated early or it will result in permanent blindness. The investigator's plan to contact high risk families immediately collided with French privacy law which forbade them to do so. The issue was finally resolved by the scientists mounting an intensive educational campaign in the relevant region, warning parents of the dangers and encouraging them to contact their local physicians who had

also been briefed by the researchers with all information except identifying names.

The frustration on the part of the investigator is to have information critical to someone's health, if early intervention is required, and be prevented from directly contacting the person. On the other hand, if the intervention is only marginal and the information devastating, individuals may not welcome such intrusions on their privacy.

Another area of concern is how research results are published. Following the tradition of accuracy in publishing, some groups have found themselves in the painful position of publishing genetically revealing information in the correct pedigree form so that it is recognizable to family members; should they ever see the publication, they will learn of a death sentence for certain relatives. And scientists should assume that eventually research subjects will see all relevent publications. Other groups disguise pedigree information, indicating that it is altered. Uniform standards for publishing sensitive information must be developed.

Data Bases and Banks

An important focus of attention for the Privacy Task Force is on banks and data bases either containing blood samples, DNA, or sensitive information pertaining to genotype. These banks range from the immense DNA banks being established for forensic purposes, to commercial banks, to academic and research banks, to

military banks, to sperm and ova banks, to public health banks, to "bottem drawer banks" of individual physicians or investigators, to voluntary health associations, to schools, to companies -- the list is expanding on a daily basis. With the advent of PCR techniques, spots of DNA used for testing babies at birth for a variety of metabolic disorders can now be re-utilized to check for hereditary diseases for which the parents never originally gave consent. There is growing discussion over how long samples should be kept, whether they be blood samples collected at birth or DNA samples of convicted felons. A critical issue is maintenance of security of the data and samples, particularly for interstate and international collections.

There is also potential controversy brewing over the use of DNA samples collected for one purpose and used for other, unrelated purposes. For example, if genes purportedly predisposing toward aggressivity, explosive discontrol syndromes or sociopathy were discovered, should investigators be permitted to search for these genes in DNA samples extracted anonymously from felons' DNA banks? The Center for Disease Control is now collecting transformed lymphocyte lines as part of its HAINES study examining general nutritional and population variables. To what use should these samples be put? We need to be very imaginative in our conjectures regarding the use of genetic materials: President Lincoln surely never suspected that his bones would be screened for the presence of a gene causing

Marfan's syndrome.

THE MISUSE OF GENETIC INFORMATION

Racism Under the Sheet of Genetics

Sociologist Troy Duster, in his recent book Backdoor to Eugenics, expressed the concern that since different ethnic groups are differentially affected by certain genetic disorders, racial prejudice will reemerge with renewed strength under the guise of genetic interest. One is not discriminating against Afro-Americans, an illegal activity, only people who are carriers of the sickle cell trait, who just happen to be predominantly Afro-American.

Screening for sickle cell anemia in the 1970s demonstrated how a program that was initially thought to be beneficial to a population resulted in doing harm. There was tremendous public confusion over the difference between those who were symptomatic and asymptomatic individuals with only a single copy of the sickle cell gene. Both patients and carriers lost their jobs, their insurance and suffered greatly. Even the U.S. military misunderstood the consequences of being only a carrier.

Genetic Education

Sensitive genetic information is entering a climate that is not very much more sophisticated today. There is a dearth of trained genetic professionals: fewer than 1,500 medical

geneticists and genetic counselors in this country. Many physicians have an inadequate understanding of genetics due to the paucity of genetic training offered in medical schools. Not only must complex notions of probability be communicated to clients requesting genetic information, but the great variability of genetic illness must be explained.

The ELSI programs of NIH and DOE are addressing these problems through the IOM study of genetic services and support of the development of new school curricula. Public awareness is being sharpened through support of two PBS television programs under development. Additional new incentives and programs will be a high priority for ELSI attention.

The Nazi Era

Dorothy Nelkin and Lawrence Tancredi, in their book Dangerous Diagnostics, discuss the possibility of developing a new "biological underclass" of people unable to obtain employment or insurance benefits, discriminated against in an increasingly "medicalized society". The authors present a possible worst case as a way of preparing us to provide the necessary protections against this outcome.

Nazi Germany has already provided us with a horrific example of the depths to which humans are capable of descending. (For information in the following sections, I am indebted to Dr. Peter Harper, editor, Huntington's Disease, WB. Saunders Company Ltd., London, 1991, pg.365-369.) The racial hygiene policies of the

Third Reich selected two medical groups for sterilization or elimination: the mentally ill and handicapped and those affected by genetic disorders. As persons with Huntington's disease fit both categories, they were doubly jeopardized. Nine disorders were listed for which sterilization was mandatory in the compulsory sterilization law of July 14, 1933: hereditary feeble-mindedness, schizophrenia, manic-depression, hereditary epilepsy, Huntington's chorea, hereditary blindness, hereditary deafness, hereditary malformations, and severe alcoholism. Patients to be sterilized were brought before a "Genetic Health Court" which ruled on diagnosis and recommended for or against sterilization.

Friedrich Panse, who died in 1972, was a professor at the Psychiatric-Neurological Research Institute in Bonn, whose director was Professor Kurt Pohlisch. Both were Nazi party members instrumental in establishing the race-hygiene laws and were actively involved in the genetic health courts and mass murder. Both were acquitted after the war and returned to prominent positions in their universities. Panse conducted the first survey of Huntington's disease in Germany, amassing a great volume of family record data. He also reported all cases and their families to the Nazi health administration where they were later sterilized or murdered.

There is currently a tremendous controversy in Germany regarding the use of Panse's register which has been recovered following the integration of east and west Germany. There is currently a moratorium on the use of these materials while debate

rages as to whether or not records gathered by a Nazi official for sterilization or elimination purposes can serve a legitimate research or medical function.

There were 350,000 to 400,000 people compulsorily sterilized during the Nazi regime. Beno Muller-Hill estimates that there would have been about 3,000 to 3,500 sterilizations for HD. When sterilizations no longer sufficed, mass murder ensued and Huntington's disease was on the list specifying disorders for which extermination was required. The numbers killed is unknown.

Local Politics

Although these atrocities occurred in Germany, the U.S. has nothing to be smug about. Charles Davenport, a prominent eugenicist who headed the Cold Spring Harbor laboratory before Dr. Watson's time, supported the German program of compulsory sterilization and recommended a similar policy at home:

> It would be a work of far-seeing philanthropy to sterilize all those in which chronic chorea has already developed and to secure that such of their offspring as show prematurely its symptoms shall not reproduce. It is for the state to investigate every case of Huntington's chorea that appears and to concern itself with all of the progeny of such. That is the least the state can do to fulfil its duty toward the yet unborn. A state that knows who are its choreic and knows that half of the children of every one of such will (on the average) become choreic and does not do the obvious thing to prevent the spread of this dire inheritable disease is impotent, stupid and blind and invites disaster. We think only of personal liberty and forget the rights and liberties of the unborn of whom the state is the sole protector. Unfortunate the nation when the state declines to fulfill this duty! (Davenport and Muncey, 1916).

CONCLUSIONS

We meet here today with the nation on a very different mission than that prescribed by Davenport. We meet to protect the privacy of individuals and families, to prevent the abuse of genetic information or the loss of rights and liberties.

Some people point to the Nazi catastrophe as evidence of the dangers of genetic information. But this barbarism occurred a decade before James Watson and Francis Crick discovered the structure of DNA, long before genes were localized on chromosomes or we were capable of sequencing them. The Nazis relied on observable symptoms, violations of the privacy of an individual emanating from the genes themselves.

It is chilling for me to see my name, so to speak, on the Nazi list slated for extermination or sterilization or read Davenport's cruel words. But it was my own torment to watch my mother in her personal concentration camp enslaved by a mind and body that no longer functioned. One in four persons with Huntington's disease attempts suicide and my mother was among this group. The Nazi activities were barbaric beyond imagination. But many hereditary disorders are also barbaric for those that suffer from them and those that love them. We cannot allow our concerns about the potential misuse of genetic information retard the search for alleviating the physical and psychological pain of these illnesses.

In summary:

1.) The Human Genome Project is an egalitarian search for all genes, many of which can cause or contribute to causing genetic disease.

2.) The Human Genome Project is an organized, coordinated and collaborative effort to find genes. The majority of Voluntary Health Organizations cannot support the piecemeal, expensive endeavors that may be required to find "their gene".

3.) The joint NIH/DOE Working Group on Ethical, Legal and Social Issues intends to be proactive and energetic in helping define a social and medical agenda in which people can take advantage of the benefits of new genetic knowledge without suffering from discrimination, economic, social or psychological loss, or stigmatization.

4.) New legislation may be required, either on a Federal or state level, to ensure the privacy of genetic information. ELSI programs will investigate the advantages and disadvantages of state versus federal legislation and help to develop policy options for introduction into new legislation.

5.) Discrimination based on genotype, just like discrimination based on race or gender which are expressions of genotype, should be prohibited. Legislation may be required to reinforce this basic civil right.

6.) The fruits of the Human Genome Project are a source of great hope for millions of Americans. When I ask people who are presymptomatically diagnosed with Huntington's disease what

sustains them, they usually answer "God and trust in science." Our political agenda is complex and will demand empathy, caution and courage. It cannot be carried out at the expense of science, but as two complementary programs to prepare for the 21st century.