

**Institute for Alternative Futures**  
**Foresight Seminars on Health and Innovation**

**SUMMARY**

FORESIGHT SEMINAR ON  
ETHICAL ISSUES IN GENETIC TESTING  
June 21, 1988

**ABSTRACT**

The mapping of the human genome portends an onslaught of ethical issues that will confront medical practitioners, consumers, employers and public policy makers. Already, tests that show predispositions to disease are entering a market a society that is poorly equipped to deal with issues raised by the tests. One such issue is who should control and use information generated by the tests? Patients, their families, doctors, employers and insurers all can use genetic information when making important decisions, and the conflict between their interests will be difficult to resolve. Decisions influenced by genetic tests can affect careers, medical reimbursement, marriage and even the choice to have a child or an abortion. Yet despite the powerful implications that access to genetic information may hold, public policy has not defined responsibility for how new genetic information will be used.

Part of this responsibility involves education about the meaning of results from genetic tests. While the tests will provide a great deal more information that will govern decisions, the statistical nature of genetic test results will create difficulties for many people. More information will not mean more certainty. It will reveal probabilities, and if people are not prepared to interpret the probabilities or to deal with their psychological impact, there may be devastating consequences. Counseling and education will be required; yet they can be very expensive, and the debate about who should pay the bill has not been resolved. Support groups can play an important role in helping people understand and cope with information about diseases, but they are not always utilized. Currently, no public policy regulates the use of the new information that will be garnered by genetic testing.

Beyond the many issues that affect different individual interests, public policy will also confront the impact of eugenics on the human gene pool. Many contend that scientists are playing God by creating technologies that can govern human evolution. People have been governing, but not controlling evolution in many ways, however, and genetic testing will provide simply one more means to eliminate disorders. A new eugenics is rising. These issues will have more to do with how people learn to make choices derived from incomplete information, and how they make better decisions even as they realize the limitations of the new knowledge from genetic testing.

**BACKGROUND**

Dr. Philip R. Really, Director of the Eunice Kennedy Shriver Center University Affiliated Program began the seminar by discussing presymptomatic and direct gene testing, as well

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as the ownership rights of these genetic results. Dr. Neil Anton Holtzman, Professor of Pediatrics, and Epidemiology, at Johns Hopkins University next described the scope of the technology available for genetic testing and the policy implications thereof. Dr. Walter Truett Anderson elaborated on the policy implications and also discussed the political and ethical dialogues that will be needed to shape ethical public policies.

#### **Philip J. Reilly, J.D., M.D.**

Imagine that a test exists that can tell you 20 years in advance whether you will or will not die of a certain genetic disorder. Imagine that a battery of tests exist that can identify whether or not you are predisposed to come down with certain kinds of disorders that are at least partially genetically influenced, but also depend on your life style. Imagine that very powerful tests exist that can directly inform you about genetic information that is very relevant to whom you marry and with whom you decide to have children. What are the social and economic consequences of these tests?

Rapid technological progress has resulted in the identification of a very large number of genetic markers. The markers are a basis for determining risks about the likelihood of a disorder occurring. However, before a diagnosis is made presymptomatically in an individual, a family must agree to be tested and to share the resulting information. Family-based testing may generate intrafamilial conflicts and raise many ethical questions: What are the circumstances under which one individual should be able to compel another to undergo genetic testing? What are the circumstances under which a physician would be justified to violate the age-old principle of confidentiality and alert a third party about a genetic risk that he or she faces?

The results of predisposition testing, if released to Individuals other than the patient or client, could introduce such problems as insurance discrimination and problems in the workplace. Under the principles of freedom of contract, an Individual employee could waive the right of privacy in exchange for employment. The employee's doctor could then be asked to supply the requested information. Some argue that because the physician is more aware of the impact of this information than their patients, he/she may be compelled to refuse to disclose the information to the employer or insurer. What impact would this have on the doctor/patient relationship?

Historical evidence suggests that certain precautions must be taken if mass screening for carrier status is to be made widely available. For example, in the early 1970's, thirteen different states wrote laws requiring certain individuals to undergo testing for sickle cell carrier status. However, there was no actuarial data to show that carriers had an increased risk of morbidity.

Some tests promise to be both economically and ethically sound. For example, one could build a compelling case for universal cystic fibrosis (CF) carrier screening. Individuals could be alerted in advance to avoid the risk of having a child with a serious, and as of yet,

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poorly treated genetic disorder. The economic benefits are obvious and the ethical foundation for prevention of unnecessary suffering is solid.

#### **Neil Holtzman, M.D., M.P.H.**

Prenatal tests can help identify fetuses with genetic diseases like cystic fibrosis and Huntington's disease. These disorders are only the tip of an iceberg of conditions for which genetic tests will be possible as a result of recombinant DNA technology. Presently, this technology permits us to localize a gene for a disease and identify it without knowing anything about its function, provided we have enough families with the disorder in question who will participate in the research. After this step, direct tests can be developed for the disease-causing gene or abnormal gene products. This knowledge permits the development of tests for the entire population.

Different disease-causing genes are responsible for a small proportion of people who suffer from Alzheimer's disease, manic depressive disorder, and heart disease. Genetic predispositions have been discovered in people for breast cancer and possibly lung cancer. Until large epidemiological studies are performed, we will not know the proportion of people who carry a specific mutation and who are likely to develop the disease. This consideration raises some significant policy questions about how to apply a population-wide test and then determine the number of people who will eventually be affected.

In the area of clinical testing, problems of new-test validity must be addressed. If there is a very short time lag between the application of the predictive test and the appearance of the disease, the validity of the test can be determined. However, diseases which appear long after their detection make it difficult to validate the predictive test. It is possible that backup tests will be developed which can affirm the predictive test. The FDA's current procedures and resources may not be sufficient to address the problem. (They have not yet been applied to DNA-based tests for human genetic disorders.)

Another area of concern involves laboratory reliability. As the technology of genetic testing becomes more accessible, an increasing number of laboratories and companies have come into existence which develop and perform their own tests. Tests that are not marketed as kits are not submitted to the FDA. Most states had inadequate authority to establish the reliability of new tests. As a result, the laboratory errors may be added to the inherent uncertainty of new tests.

For most common genetic disorders, there are unlikely to be tests for all of the mutations capable of causing the disease. Thus, it would not be possible to identify all people who are likely to get the disease. Also, the etiology of the majority of cases is much too complex for the disorder to be identified by a single test. There are multiple factors that interact, e.g., environmental, nutritional, and genetic factors. Given these limitations, genetic testing is limited in its ability to detect all those at risk for a disease.

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New technology will allow us to localize genes on specific regions of specific chromosomes, identify the gene products, and develop predictive tests. But we will not have therapies available, possibly for many years after the identification of the genes. We will not know how to intervene and successfully prevent the onset of symptoms in people who have a disease-producing gene type.

There are several groups in society that will have a great interest in these tests, including insurers, employers, and third party payers. This is particularly appropriate in light of pending health legislation which places a great deal of responsibility for providing health care on employers. Employers are likely to want a work force that is, and will stay, healthy if they are asked to absorb increasing health care provision costs. Thus, in addition to using genetic testing in the workplace to look for exposures to workplace hazards, it may be used to identify potential employees who are at greater risk for future disease. As some people become less employable, they will become the responsibility of the public sector; costs to society are not decreased but merely shifted. Existing and proposed legislation contains no provisions to prevent this misuse of genetic testing.

Also arising from our ability to predict the occurrence of a disease but not the ability to treat it, is the increasing use of strategies to avoid the conception or the birth of a child with such a disease. Using genetic tests, one could identify fetuses who have a high probability of developing a disease. Society would save a considerable amount of money by providing abortions, rather than absorbing the costs of long term care for those fetuses. An economic drive may push us toward a new eugenics, particularly as we get increasingly concerned about health care costs. This contrasts with classical eugenics, for we now have a firm biological basis to predict who will be affected with a disease. Like classical eugenics, the new eugenics poses very serious questions for consideration.

#### **Walter Truett Anderson, Ph.D.**

We need to analyze our arguments. We must consider why and how we argue and what we argue about. It is important to devote attention to the language we are using and note how it shapes our controversies. In the domain of genetic testing and screenings many people contend that scientists are "playing God". These individuals also believe that the "natural" process of evolution is being disrupted by these interventions. But the underlying structure of their arguments is misleading. The emphasis should not be placed on whether individuals should have increasing responsibility for their own health and reproductive lives, but how they deal with that responsibility. We are reluctant to accept that we are already governing evolution, not controlling it. Governance, even in a totalitarian society, is never completely in control. We are responsible for evolutionary developments in countless ways and in various countries, and we are becoming more responsible for it every day.

We are confronted by an astounding escalation of rates of evolutionary change which has been going on for millennia. The escalation is occurring so rapidly that it is impossible for

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us to ignore the responsibility we have for governance of the biosphere and of living things within it. People will have to handle more and more power, information, and choices. The capacity to use and understand information is an area of prime concern. Information, contrary to what we might have thought, does not create certainty. The information that scientists give us may be profound and brilliant, but it cannot give us absolute certainty about the future, nor can it give us easy answers about how to prepare for it. Using information wisely requires us to develop new skills and new values.

There is a gap between science and the lay culture. For example, it is difficult to translate a scientist's interpretation of information derived from genetic screening into concepts familiar to a lay person. We do not have absolute information nor absolute conclusive proof that once a marker is identified in a patient, they will progress toward the culmination of the disease and will understand the implications and ramifications of their predisposition. Yet frequently some decisions must be made based on incomplete information.

So much of the dialogue and so many of the concepts that we use are inadequate for our realities. Eugenics at one time was very popular in American society. Suddenly, due to the activities of one Adolph Hitler, it became a taboo phrase. The fact of the matter is, we are practicing eugenics and we are going to be practicing more of it in the future.

We need to begin to examine the substance, motivation and language of our arguments and issues. As never before, socially concerned people are going to have to take in new information, synthesize it, and realize the limitations of the new knowledge. This will enable them to make better decisions about it.

#### **QUESTIONS AND ANSWERS**

Rather than using medical and genetic information only to screen certain people, would it be possible to place genetic testing in a broader context, which would shift our concepts more to public responsibility?

Dr. Reilly responded that, for complex disorders such as heart disease, one can have a certain sense of relief in knowing that no one is to blame; it was preordained. To be informed that your child has a genetic disorder and that the child inherited the gene from you can be a devastating piece of information. By and large, most people are not prepared to internalize and understand that information without some effort. This generates the need for a whole new kind of information analysis and education. Dr. Reilly added that the cost of properly communicating this information would cripple the ability to deliver the service.

Concern should not only be centered on how an individual would handle a negative diagnosis, but also on how others would perceive that individual. If we are going to do widespread screening, then we will have to deal with a tremendous gap between screening and educating people about the meaning of the tests.

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Dr. Holtzman added that, the de-mystification of medicine is changing the nature of the patient/physician relationship. People expect explanations from physicians and reserve the right to make their own decision about medical care, including whether to be tested and what to do about abnormal results. Additionally, society must decide who should have access to test results, and who should have the power to demand that information from individuals. Is it the physician's, the employer's, or the insurance company's information or the patient's? Should there be a much greater sharing of information? Most people who submit to genetic screening will not be sick patients and they will have to decide whether they want their test result released to others. One has to look beyond the physician/patient relationship and prepare people for the possibilities and limits of the tests.

Dr. Anderson concurred and added that we must be clear about what information would be made available to a patient or client. Clearly there are many kinds of information. For example, parents who are deciding whether or not to give birth to a child who will have Down's Syndrome must be Informed of the genetic odds Involved and the human or social information about other people's experiences in similar situations. The available resources and effects of the syndrome on others' lives should be included in that information.

If the spectrum of diseases or predispositions that can be screened for are extended, will it make sense for these tests to be cost driven -- especially if there are tests which would affect most of the population ?

Dr. Holtzman responded that cost considerations may promote an employer to deny an employee certain benefits, On the other hand, an employer who is hiring a worker at the age of 25 may not be concerned about the risks of that person developing heart disease. As a matter of fact, in tents of pension plans, an employer might be more interested in finding someone who is likely to develop heart disease at the age of 55 because he/she will spend less on pensions.

If we participated in widespread genetic testing, what percentage of the population would be relatively likely to be dropped by their insurance carrier? Could testing move ahead enough to make this possibility a serious and significant argument in favor of a national health insurance program?

Dr. Holtzman responded that some insurers or employers would find it Inappropriate to hire people who were at risk for heart disease, and they are likely to use HDL/LDL ratios or cholesterol tests which are already available. Of the people who have the highest 10 percent of cholesterol in the population, only about a quarter of them will get heart disease; they constitute only about 30 percent of all the people who will get heart disease In a given time. In other words, those tests are not very good. A genetic test increases their predictive value; however, their sensitivity will be reduced. Unless guidelines are established for the use of genetic testing in employment, this will become a problem of significant magnitude. The exclusion of many people from private or employer-paid health insurance argues for some form of national health insurance that will cover everyone.

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Comment from the floor: There is a possibility that insurance companies could rate predispositions, not as an exclusion, but could use them to increase the premium on an individual basis, making those premiums prohibitive. People at the highest risk for some of the most expensive conditions, could be saddled with the highest insurance bills. So, these people are then left without insurance, although they are the ones who are most In need of protection.

Dr. Holtzman added that, once a test is developed to predict the future risk of disease, the insurance companies will be terribly worried about adverse selection. When a person knows they have an increased risk of developing some disease, that person will want more private health insurance. The insurance companies, to protect themselves, need the same information that people have.

Thus, it is likely that as these tests become widespread, insurance companies will have to require tests to the extent that the knowledge is available.

What are the forensic applications of genetic testing? Would the comparison of a suspected criminal's blood to the blood or tissues left at the scene of a crime infringe on his/her rights?

Dr. Reilly responded that forensics is a hot area in DNA testing. The Supreme Court (Smerber v California) has established that drawing blood for probable cause is not a violation of the Fourth or Fifth Amendments. There is going to be tremendous interest concerning the appropriate use of forensic testing. It has already been used in about twelve criminal cases. Mr. Hicks from the FBI is predicting that in three years, every State Crime Lab In the country will have a DNA testing lab.

There may be some potential conflict in the use of DNA derived from a sample found at the scene of the crime. But, there is a greater conflict concerning to what the sample will be compared. This posits the existence of massive genetic databases, DNA banks where people could run a DNA sequence derived at the scene of the crime against a national crime data bank. These issues raise more interesting questions about controlling entry into a reference database.

What has to happen scientifically before we are actually able to conduct commercial tests in a reasonable way? What kind of progress in genetic research still has to be made before that becomes a reality, and how far away is it? Are we likely to see population-based screening for many rare diseases? If so, what will it do to insurance coverage for those diseases?

Dr. Holtzman responded that, in the case of Huntington's disease, which has a relatively low incidence rate, there will probably never be population-wide screenings - only familial testing. For those within families in which disease has already occurred but who are not found by the test to be at risk, it would be possible to obtain insurance at the standard

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rate. For population-wide screening using DNA testing, tests will be introduced within 5 to 10 years.

Dr. Anderson stated that there will be an escalation of concern about the need for a large-scale rethinking of our whole national approach to health service and health insurance. This would be in response to the technology and problems that are not going to be solved with the types of programs, policies and responses that currently exist.

There has been considerable discussion in public health literature about the uselessness of screening for disorders that cannot be treated, or screening for disorders where the results of the screening test has no benefit to the individual. Is there any potential for distinguishing between those things for which there is utility in screening and those things for which there is not? Would this distinction be better controlled under a national health insurance system than in a free market medical care system?

Dr. Holtzman responded that there is no certainty that when a genetic factor is identified, for example LDL receptor defect, the person with that receptor defect will develop coronary artery disease by 65 years of age. Within the general population, we are unsure whether the presence of that disorder, which in certain families does follow the rules of Mendelian inheritance, is always going to predict the disease. This is a very important bit of information that needs to be collected prospectively. Determining the magnitude of risk at the genetic level should be done before testing. Even for single gene defects (e.g., PKU), there is not a one hundred percent expressivity of the disorder.

There is always some utility in screening. There will always be some people at risk for certain disorders who will want to know they are at risk even when nothing can be done other than avoiding conception or birth of affected offspring. It is going to be painful for that individual, regardless of the age at which the disease becomes manifest.

Dr. Anderson commented that there are biotech companies looking for an avenue into the market. There will be substantial development of tests by entrepreneurial firms, wherever their market research reveals that another source will pay for it.

Dr. Holtzman added that some commercial developers may try to do market research to find out if a market exists for a test for Alzheimer's disease based on absence of therapy and only the possibility of avoidance. They may discover that the market is so small that the test will not be developed. Also, some companies which are trying to make "fast" money may nevertheless promote a test through advertising even though the test is an imperfect predictor of disease. At the rate progress is being made, the doctor may be removed as a middleman in this procedure, and home testing for genetic predispositions may soon be possible. Companies will advertise directly to consumers.

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Dr. Anderson concurred and added that the policy response will be in the area of consumer protection with regard to advertising regulations, interventions, and licensing of tests.

Will the genetic information for predispositions toward a disease make it easier for someone to avoid some substances or situations that may exacerbate their conditions, or will people be able to alter their lifestyles?

Dr. Holtzman responded that, both would be possible, but that people should be counseled about what the risks are and the extent to which avoiding substances or changing lifestyles will lower risk associated with the results of a predictive test. Therefore, communication of the Information must be done carefully. The information on predispositions is not a statistical average. It varies, depending on the individual.

Many cases involve the issue of Living Wills through which an individual can elect to refuse treatment if they ever reach a certain stage or quality of life. If parents receive information about their unborn children, will they be able to use a Living Will to refuse treatment or medication for their child?

Dr. Holtzman stated that we will be able to accurately detect disorders in the fetus early enough in the pregnancy to permit the mother to decide to terminate the pregnancy. There is already a precedent of parents and even the infant recovering damages, when prenatal diagnosis was not performed in a high-risk situation. The child can claim damages under the doctrine of wrongful life.

The doctrine of fetal abuse, supported by some lawyers and ethicists argues that society has a right to intervene and require prenatal diagnosis for severe disorders that would cause pain and suffering after birth. The implication, therefore, is that a prenatal diagnosis puts a responsibility on the parents to terminate the pregnancy. A decision must be made as to who has the right to determine prenatal diagnosis and the fetal outcome, Dr. Holtzman stated his belief that It should be the parents and the mother in particular. The difficulty lies in trying to convey to people who have never had experience with a particular handicap what it is like to raise a child with a disorder so that they can make informed decisions.

A participant commented that insurance will cover the cost of tests available for communal situations, but it will in no way cover a so-called "therapeutic abortion" unless the mother's life is endangered. We want to protect the infant from abuse whether it's still in the fetal form or after it has been born, and yet the abortion option is often excluded for people who cannot possibly afford insurance coverage.

When sickle cell disease is detected, penicillin prophylaxis can be instituted to protect the infant from early death. Are there any other areas where a genetic test will identify such a valuable therapeutic treatment, making it ethically clear that it should be pursued?

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Dr. Holtzman said that as tests are developed and the clamor to use them grows, researchers will continue to develop therapies that offer more alternatives. There may be a shift from prenatal testing to neonatal testing, as effective treatments that must be started early are developed. But there is a danger that the widespread adoption of prenatal diagnosis (and abortion) may dampen research into finding effective treatments. After screening began for Tay-Sachs in the early 1970's, a relatively high proportion of people at risk, Ashkenazic Jews, were screened and there was a tremendous decrease in the occurrence of the disease, as couples at risk took advantage of prenatal diagnosis and abortion. The prevalence of the disease has fallen and, hence, the research interests in developing treatments have decreased.

Dr. Anderson queried, how much of an impact must be made upon the gene pool before a legitimate concern arises?

Dr. Holtzman responded that it would depend upon how the disorder is inherited. For most fairly simple recessive Mendelian disorders, the transmission of the gene that is capable of causing disease from one generation to the next requires two carriers. But the frequency of the gene in the population depends primarily on matings between carriers and non-carriers. Thus, a very small proportion of the total gene pool is affected by testing, and many generations would be needed to have any effect. Late-onset dominant disorders could be eliminated in a single generation as a result of prenatal diagnosis if every fetus at risk is aborted and if known carriers do not conceive.

A participant contented that the issue of genetic disease has a mystique about it because of a perception or a misperception that the disease is not treatable. Some genetic diseases are more treatable than environmental diseases. There probably is no disease that is strictly genetic or strictly environmental. Thus, all disease may be thought of as a confluence of multiple genetic and multiple environmental interactions -- ecogenetics.

Self-help organizations offer patients and their families a resource of information and education that exceeds by far what a doctor can do in a one hour session.