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IMPACT OF ADVANCES IN GENETIC TECHNOLOGY ON HEALTH CARE AND PUBLIC POLICY

James W. Hanson, M.D. *

Ms. Weber: Dr. Hanson has assured me that there is virtue in brevity, but I believe that it's important for you to hear exactly where Dr. Hanson comes from.

Dr. James Hanson is professor of Pediatrics and of Preventive Medicine and Environmental Health, and is director of the Institute for Health, Behavior and Environmental Policy at the University of Iowa.

A native of Jefferson, Iowa he attended the Johns Hopkins University and subsequently received his doctor of medicine degree from the University of Iowa College of Medicine in 1969. His Internship and residency in pediatrics were at the Johns Hopkins Hospital.

Thereafter, he spent two years as a medical epidemiologist in the birth defects section at the U. S. Centers for Disease Control and Prevention in Atlanta, Georgia. From 1974 through 1976, he was a post-doctoral fellow in dysmorphology at the University of Washington in Seattle prior to returning to the University of Iowa to be on the pediatrics faculty.

From 1977 to 1992, he was director of the Division of Medical Genetics. In that capacity, he helped to develop a statewide system of genetics health care services in Iowa which included a Regional Genetics Consultation Service, a Newborn Genetic and Metabolic Screening Program, a statewide Teratogen Information Service, and the Iowa Birth Defects Registry, which he also directed.

He helped to develop and served on the executive committee of the Center of Health Effects of Environmental Contamination at the University of Iowa and became director of the University's Institute for Health, Behavior and Environmental Policy in 1993.

He has served as a consultant to numerous federal and state agencies, including the National Institutes of Health, the Centers for Disease Control and Prevention, the Maternal and Child Health Bureau, and the Office of the Surgeon General, regarding genetic health care services and matters relating to birth defects.

He has also been a consultant to several private and public organizations concerned with child health and related topics, including the March of Dimes Birth Defects Foundation, the Institutes of Medicine of the National Academy of Sciences, and the National Center for Education and Maternal and Child Health.

In 1991, Dr. Hanson was a fellow in public policy of the Joseph P. Kennedy, Jr. Foundation, assigned to the staff of the U.S. Senate Subcommittee on Disability Policy, Senator Tom Harkin, chairman.

During his fellowship, he assisted with the development of legislation concerning education, health and disability policy. He is currently on leave from the University of Iowa, assigned as senior advisor to the National Vaccine Program Office, Office of the Assistant Secretary of Health in the Department of Health and Human Services.

His research interests are so long and so involved and his membership in societies so

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extensive that it would probably take the entire time for his lecture to go through them all, but is my distinct pleasure to present to you Dr. James Hanson.

Dr. Hanson: This is the first time I've had the pleasure of speaking to a mixed group like this of lawyers, law students, people interested in public policy, physicians, and so forth. So it's a little hard for me to know exactly what will capture your interest about this subject.

I'd like to start off by asking the question, "Who cares about genetics and public policy?" In the course of this answer, I need to give you a few definitions or terms, or at least specify the way I might use certain terms. When you start to talk about genetics and public policy, most people lose interest because of preconceived ideas.

I want to make it clear that I think that these impressions that are prevalent among the general population are really "genetic misperceptions." Several of these genetic fallacies that have been identified.

Genetic disorders, first of all, are not rare conditions. Genetic conditions clearly affect a substantial proportion of our population. A term that often is confused with "genetic conditions" is "birth defects." Birth defects are conditions which have at least two known categories of causes. There are those that are due to alterations in our genetic material, and there are those that are due to environmental effects on the developing fetus; things that the pregnant mother may be exposed to during pregnancy.

And as a consequence, people get very confused about the difference between genetic disorders and birth defect problems. Nonetheless, birth defects, a substantial number of which are genetic, can be used as a way of indicating the magnitude of the problem.

Any calculation of that, any studies that have been done, show that between three and four percent of newborns can be identified as having a birth defect within the first year of life. About half of these are really quite serious and potentially fatal. In fact, lethal birth defects are the most frequent cause of infant mortality in the United States at the present time.

This is one way of measuring the impact of genetic conditions and birth defect problems on our population. But it's only one way to look at this. If you follow that same cohort of children up to their young adult years, you find that the number rises to about fifteen to twenty percent of individuals being identified by the time they are adults as having some kind of genetic or birth defect problem, at least a third of which, or five percent overall, are of major medical or cosmetic significance.

As a consequence, in addition to problems such as infant mortality, these conditions contribute disproportionately to morbidity in our population. That is, they produce ongoing states of ill health, whether they are problems that lead to disability in children or adults, or other later onset conditions.

This percentage of those affected would grow if we include the adult onset kinds of conditions that we now know have major genetic determinants, conditions like cancer, like coronary artery disease or diabetes, affective disorders like schizophrenia or bipolar illness. Many of the older age onset conditions which afflict large numbers of people in our population clearly have major genetic determinants to them.

So the burden of genetic conditions, as well as of birth defects in general, is really a substantial one and if for no other reason than the size of that problem, it's clearly one that

should compel interest on the part of policy makers.

What has been done by health care professionals and public health specialists, to address these kinds of disorders?

The last twenty years especially has seen the advent of what I think of as a group of genetics health care services in the United States. These birth defects diagnoses and treatment programs include genetic counseling programs, genetic and metabolic screening programs, prenatal diagnosis and treatment programs, and environmental hazards avoidance.

One might object that the last isn't really genetics. However, genetics is often defined as the study of variability, and not all variability is heritable. Geneticists need to understand about the non-heritable components to variability and their interactions with the heritable components. Whereas we sometimes cannot manipulate our genetic endowment with ease, we certainly have the possibility of manipulating our environment more readily, at least in some situations. So programs to reduce environmental hazards, especially as they may relate to fetal growth and development, clearly reduce the burden of this same group of conditions.

We've started to develop and expand the range and scope of treatment approaches for birth defect problems and of genetic disorders. This relates to another of the genetic fallacies, that is, that genetic disorders are not treatable. They may not be curable, in the ordinary sense of the word, but they are clearly treatable, and that applies to the vast majority of them in one way or another. Sometimes the treatments are not fully successful, but there are certainly treatments that can be brought to bear in a high percentage of these conditions.

Finally, I also include health education about genetic disorders and about genetics in general as a kind of a genetics health service. If you don't have knowledgeable health professionals and knowledgeable consumers, it's impossible to really assure access to the treatment programs or the care services for these kinds of conditions. As these new technologies that you've been hearing about have been developed, we have seen an increasing number of applications of this technology in the health care arena.

Screening programs, a topic which deserves special attention to, are a long-term approach that has been applied for at least thirty years in this country. There are several types of screening programs. Screening programs are not intended to establish specific diagnoses. Screening programs are intended to be applied to large populations of individuals to identify a much smaller subgroup of individuals who are at high risk of a particular kind of problem.

We have seen the development of several kinds of screening programs that relate to genetic services. There are newborn metabolic and genetic screening programs in every state in this country. These are programs of which most of you are aware, at least if you have children. A little drop of blood was drawn from the heel of your baby when the baby was born, and in different states it was tested for a different number and types of conditions.

Almost every state tests for phenylketonuria (PKU) and for hypothyroidism, but some states may test for a much larger number of conditions including hemoglobin disorders like sickle cell anemia, or galactosemia, which is a disorder of a type of sugar metabolism. Other screened disorders may include maple syrup urine disease and biothinidase deficiency adrenal hyperplasia syndromes and so forth.

The screening programs that have been developed to test newborns for conditions that

meet several criteria. First of all, they are conditions for which there is no really good alternative for sufficiently early identification. A screening program isn't needed if during routine health care the person was going to be recognized and treated appropriately, anyway. So these are programs that are designed to pick up individuals that would customarily be missed or at least missed until too late. Secondly, they're programs that are designed for disorders for which there is a treatment. If treatments are not available, a question could be raised about the value of screening, at least with public dollars. Third, these are conditions that are frequent enough that you actually would find some affected children. Screening programs have a potentially large cost. Even if costs are only a few dollars per individual, screening 100,000 births is expensive,and requires justification. Such newborn metabolic and genetic screening programs are prevalent throughout the country at the present time.

A second type of screening program that has developed are the high-risk pregnancy outcome screening programs. They go by a variety of different names. I prefer this term, because what they do is they identify women during pregnancy who are at an increased risk for a variety of adverse reproductive outcomes, including early fetal death and stillbirth, presence of certain fetal birth defect problems, and a variety of other conditions and situations. In many of these cases there are interventions that can improve maternal or fetal well-being.

In some states, these programs are called neural tube defect screening programs or birth defect screening programs. I discourage those terms, because, first of all, those outcomes are among the less frequent of the outcomes that you can identify; and, secondly, they often focus (in my view inappropriately so) the attention of people on controversial societal issues, like termination of pregnancy in the presence of a child with a birth defect. I believe that it is inappropriate to label a program with regard to only its most controversial and, in some respects, least frequent type of outcome. In my view, really one should focus on the more general purposes for which the program was developed.

There are other kinds of screening programs that have been proposed such as screening for individuals who may by at increased risk if they're exposed to certain kinds of environmental hazards in the work place. In addition, NIH is planning investigational programs to screen people for high risk of breast, ovarian and colon cancer development.

Those experimental programs are just now being developed. However, it is clear that we will see researchers looking at the ability to use new molecular genetic technologies to identify high-risk populations. These will be populations with identifiable specific health outcomes for which the risk is remarkably different than that of the general population.

It makes little difference whether it is breast cancer or whether colon cancer or Alzheimer's disease or any number of other conditions. So long as there is some kind of relatively clear medical intervention that could be proposed, there will be enormous interest in the possibility of doing mass screening. The purposes of these screening programs are prevention of adverse health outcomes or early interventions to improve outcome.

The second area of application of these new genetic technologies is in the area of diagnosis of conditions. It includes both genetic and non-genetic disorders. This is not generally appreciated by the public. In fact, I think it is not generally appreciated by most health professionals.

This point relates to the question "what is a genetic test?" The term "genetic test" has

two distinctly different definitions. One definition of a genetic test is a method by which the genetic material of the body is examined. An example is a chromosome test in which the chromosomes are actually counted or studied in some way. Other examples involve taking DNA from cells in the body and analyzing it for the presence or absence of certain genetic markers.

If that kind of definition of a "genetic test" is used, it may relate to any living organism which has genetic material, such a test could be used to study questions related to any kind of organism, whether it's a human being with questions about the likelihood of a genetic condition such as muscular dystrophy, or a germ causing an infectious disorder.

One of the applications currently under development is for rapid diagnosis of infectious organisms that cause various diseases. Examples might include rapid simple office-based tests for deciding if a person has a strep throat, or some other infection. Indeed, these technologies have been used to develop screening approaches for HIV. As can be seen, this leads to a wide range of applications.

The other definition of the term "genetic test" is a test for the presence of a genetic condition in a person or, for that matter, any organism. Essentially, then, any kind of test known to be of use in diagnosing the presence of a disorder or characteristic might be covered. It might be an eye test in which a clinician does visual fields, looking for evidence of macular degeneration.

Currently, the genes for degeneration have not been identified. Doctors can't identify these conditions through some kind of DNA technology at the present time, but can make a diagnosis by having an ophthalmologist do a funduscopic examination of the eye and doing visual field testing.

Likewise, if a doctor saw a patient with a skeletal disorder that made a person short, an x-ray could become a "genetic test" in that it would help to determine which form of disturbance of growth he is dealing with.

As can be readily seen, the definition for the term "genetic test" is important. Careful consideration of goals is crucial to ensure the desired outcome, and also to avoid unintended consequences. These diagnostic tests that are being developed have an enormous range of applications, and corresponding implications for policy.

Finally there are the applications of genetic technology to the treatment of both genetic and non-genetic types of conditions. These include both the possibility not only of doing gene transplantation or genetically altering tissues and using them in individuals (which we're beginning to see examples of now) and also to use the ability to identify a genetic characteristic and to design a chemical that would recognize a particular genetic pattern and then to hook something onto that chemical, thereby targeting a therapeutic agent to only the cells or the organisms that contain that particular genetic marker in them. This approach is thought to have a great deal of promise for the development of new agents to fight cancer, to fight infectious diseases, and so forth.

Likewise, this technology is the basis for the dreams about developing a very simple one-shot vaccine. That is, scientists might be able to take the genetic material from several different organisms and put it together in one simple viral package. Then this single dosage form could be given to the individual perhaps to simply swallow, not even to inject. If it were also possible to attach regulatory genes that would cause the expression of different

parts of that genetic material at different times as the child's immune system became able to respond, perhaps one dose given at birth of such a comprehensive vaccine might be able to immunize children against a whole range of different genetic conditions.

Imagine what a wonderful advance that would be in terms of expense, the ability to reach the entire population, simplification, making things easier on parents, and probably for safety issues as well.

As can be appreciated these applications now in the area of treatment are really quite wide ranging. It is important to consider them broadly and not to simply focus on the single approach of transplantation of genes and changing the human genome.

These genetics technologies, have enormous implications not only for the treatment of genetic disorders, but for treatment of cancer, treatment of other common types of disorders, whether they are infectious diseases or cardiovascular conditions, even possibly for behavioral disorders.

It may be possible to design therapeutic agents which would in some way modify neural tissues, leading to correction of problems in the way those nerve cells communicate with each other or how they grow and develop in the fetus. There are also implications for occupational and environmental health, in terms of being able to identify who is susceptible or especially susceptible to a given environmental agent. With such knowledge it becomes possible to protect that individual so that he or she can continue to be employed, without the alternative of adopting some kind of discriminatory policy.

Because this new genetic technology has such wide-ranging implications across many health care fields, there is a need to think about health policy issues.

How then should screening and testing programs be developed appropriately? The issue of appropriateness raises questions about access to care and equity. That is, are people going to have equal access to these programs? These are issues that the next speaker will address in terms of who gives consent and under what circumstances can informed consent be appropriately assured, as well as issues of how much society can afford and who pays. Many such issues will have to be addressed with regard to these kind of programs.

In terms of therapeutic implications, how can appropriate therapeutic outcomes be ensured? What are the prices that society can afford to pay for screening rare conditions if these technologies are used widely (the "rationing issue")? As an example of this issue, recall the earlier point that screening programs could be rather expensive items. Take the example of cystic fibrosis (CF).

CF, as you know, is a relatively common (as individual genetic conditions go) severe disorder in the past customarily thought of as a childhood condition. Now, it is known that there are some adults that have cystic fibrosis, and not everybody who has cystic fibrosis has a terribly severe outcome.

Nevertheless, traditionally it has been thought of as a condition that affected young children. CF produces bowel and digestive problems. It produces severe lung disease, recurrent infections, and usually leads to death at an early age, typically before the age of twenty, at least in years gone by. It's a bad condition. It is a condition that results in enormous health care expenses for families. These children, at least the more severely affected ones, spend a lot of time in the hospital.

Now, for this reason, people have suggested that the general population should be

screened in order to identify the children early on and get them into care earlier. Perhaps lives could be extended. Perhaps some of the more severe complications could be prevented. Maybe better approaches to treatment can be developed.

As a consequence, there was a search for the cystic fibrosis gene, which was successful. Scientists were somewhat surprised to find that the gene for cystic fibrosis is very long. With any gene, the longer it is, the more places there are where its message can be altered (mutated). We know that there are over a couple of hundred different changes in that gene that can be associated with disease, e.g., a variation of cystic fibrosis.

Now, one implication of that finding is that a genetic test becomes more complex to perform. Current estimates of cost of doing this test on an individual run from very optimistic suggestions of $100 to over $300 per person, just for the laboratory part of the test alone.

If you were to consider applying such a test to the general population this could have substantial cost implications. (By the way, the laboratory part of the screening program is never the most expensive part. The most expensive part is actually having somebody go to the child, draw the blood, do the paperwork, send the blood in, collect the results when they come back and talk to the family.)

Were a policy to be adopted that all four million newborns in the U. S. be screened each year, the health care system could be looking at what Everett Dirkson used to call "real money" ("A billion here, a billion there, pretty soon you're talking about real money."). Remember also, this is before we ever get to the point of suggesting that we might try some kind of therapeutic intervention.

It is apparent that such ill-considered policies easily have enormous implications for the health care system. Concerns about cost shifting or cost increases will inevitably raise serious questions about societal choices as to how and for whom we should spend our money.

This is certainly not to suggest that children with cystic fibrosis should not have access to treatment. They obviously should. They deserve care as much as any other child or any other person deserves health care. But we must recognize that the indiscriminate use of ill-considered strategies can have enormous cost implications. Alternative approaches should be considered.

Another area of great concern is in terms of collection of information and how we monitor the population. One aspect of this relates to the lack of understanding that we have about the causes of birth defects. As I mentioned, there clearly are environmental factors as well as genetic factors that interact to cause birth defects. The cause of most birth defects is presently unknown and, as a consequence, we have developed, in a few states, large-scale birth defects surveillance programs in the hopes that through the systematic collection of information on the occurrence of these events we would be (1) able to facilitate studies that might identify the causes and allow us to prevent those defects from happening; and, (2) able to more appropriately direct services to those individuals who have these conditions.

Unfortunately, the minute you develop a widespread data system, whether it's for surveillance of birth defects or for surveillance of the frequency and distribution of genetic disorders, even for the most noble of purposes (like being sure these people get appropriate health care at the right time) you end up with the potential for abuses to occur. Concerns immediately arise about invasion of people's privacy and confidentiality. Again, that will

be a set of topics that will be discussed in the next presentation.

This situation begs for clear public policy formulation. But in the rush to deal with these justifiable concerns, there arises a paradox which has received little attention. That is that without these kinds of data, we may never be able to actually fully enjoy the fruits of the genome project and many other kinds of genetic research. Why might this be?

Remember, most of the health conditions (or, for that matter, human characteristics) that have a genetic component to them, have other components as well. Some scientists would argue that there is no such thing as a purely genetic disorder; just like they would argue that there's no such thing as a purely environmental disorder. They argue that every set of conditions has genetic components and environmental components. Furthermore, when you start to really study those disorders, you discover, just like we did with cystic fibrosis, that it isn't one single disease, but it's a whole complex group of disorders, each of which is a little different in its clinical characteristics.

Likewise, if you carefully investigate the environmental contribution to disease, you discover that in a person with one set of genetic information, the environment may not play very much of a role in predicting outcome at all, but another person with a slightly different set of genetic characteristics may be greatly affected by several different factors in the environment.

The result is that we often end up concluding that these genetic test systems typically give only "risk" information. They tell you that you're more likely to develop a particular health outcome, but not that you will definitely develop that health outcome. The question then, of course, is how much is that risk and what are the other factors that play into that equation.

It's one thing to say to a young woman, "You have the breast cancer gene and you're at an increased risk to develop breast cancer during your lifetime." But, if you can't tell her when that increased risk really becomes operational, whether age 13 or at age 57, or throughout that span, the implications are quite different.

If the risk doesn't become significant until she is 45, for instance, why would she go to a lot of special diagnostic or treatment programs early in life? On the other hand, if the person who has that increased risk (genetic factor) has an even higher risk if they're exposed to certain factors in the environment, the possibility of preventive interventions through risk factor avoidance is raised, suppose, for the sake of argument, that with mammography screening, the little bit of extra radiation might increase the risk even further. Were this true, intensified clinical surveillance of persons with this gene could have the unintended consequence of actually increasing risk further in a selected subcategory.

Now, I want to emphasize, nobody has suggested that this scenario could be factual. My intent is to demonstrate that if we don't look at partitioning the risk for these health outcomes into heritable and non-heritable components, and try to dissect out which apply to which individuals, we could give people greatly misleading kinds of information. What were will-intended health care or preventive health care strategies could be, in fact, misguided.

Now, why did I raise that issue in the context of health data and surveillance? The genetic material of individuals varies enormously from person to person. Likewise, environmental exposures throughout a lifetime vary enormously. Thus, the very thought

of trying to develop a sound epidemiologic study to understand and measure the contribution of the multitude of different exposures and genetic risk factors that may apply to a given health outcome becomes an enormous problem. Even if you simplify such a study greatly by making a variety of assumptions and by lumping people together in broad categories, it is easy to see that you have to have an absolutely enormous kind of population to study.

This raises a question about those who refuse to be a part of such a study. If people refuse, does the very fact that they're more likely to refuse mean that they have a genetic characteristic that affects their behavior and thereby changes their risk in some way? If it does, then, of course, your study will be misguided by the very fact that you allowed people to refuse to participate.

This kind of question arises when people talk about taking population-based samples of DNA, for example, obtaining samples from newborn screening programs. You could take those little blood spots, using left over material, extract the DNA, (this has been done) and study an entire population.

Theoretically, one could track the individuals throughout their lifetime, find out through regular health interviews and data collection what they've been exposed to, what they've been eating, what infections they've had, who ran over them or what accident they were in and so forth, and then investigate the contribution of all these factors to a variety of different health outcomes.

This is a potentially very powerful way of learning what that genetic risk information really is; what you should really tell to people. But it's also very troublesome, because you may learn lots of things. You certainly would have information about people that they may view as being their own, and something they don't wish shared with anybody else.

A good example of such concerns has been found in the area of health insurance. Again, I won't get into that because it's going to be discussed later. But clearly there is the potential for people to identify an individual at increased risk or even at a suspected increased risk and exclude them from health care or exclude them from employment based on the finding.

Another serious policy issue is, who are those health providers that must understand these issues and where are they going to receive training? Can they appropriately diagnose and treat and appropriately communicate information to the people whom they serve? It's quite clear at the present time that, with very limited exceptions, our medical schools do not do a very good job of educating physicians regarding basic principles of human genetics and their application to the diagnosis or treatment of human health conditions.

It's clear, also, that the same thing can be said about schools of nursing. And I would venture to say it's probably true about schools of law, even though we don't think of lawyers as health providers. I won't, out of courtesy, say what we do think of them as.

This set of concerns branches across a wide range of public policy issues. These are issues for the nation and for states as well.

It is clear that this new genetic technology touches not only the health care delivery arena, but it also affects insurance regulation, employment issues, and civil liberties. It touches issues of public safety and justice, because these tests can be used to identify individuals. You may know the Army now has a proposal to investigate the use of DNA for a "genetic dog tag." Questions have already been raised about how long the military

services should retain this information, and who else could have access to that genetic database.

Likewise, there are similar forensic and legal issues in the application of genetic technologies for the identification of people who have left biologic specimens behind at the scene of a crime.

There are also the environmental issues which arise regarding individual variations in susceptibility, or the concern mentioned in the previous presentation. These concerns focus on issues of environmental justice and whether or not we're doing things to our environment through genetic manipulation that may not be appropriate and are most likely unintended.

In this last regard, I want to make a point that I didn't hear raised in the last session. That is, is it better to make genetic changes deliberately or by accident? As human beings, we've been doing genetic "experiments" (directing our own evolution) in many unintended ways probably the beginning of humanity.

Human beings don't mate at random, though to many it may seem like that. In fact, short people tend to marry short people. Deaf people tend to marry deaf people. People of one ethnic group tend to marry within that group more than they marry between ethnic groups. All of these kinds of factors, as well as war, genocide, famine, and cultural practices, affect genetic drift and selection; mutations which occur in one population may be confined to that population, which is one of the reasons why we see that conditions like Tay-Sachs disease are disproportionately frequent in people of Ashkenazi Jewish descent, why conditions like cystic fibrosis are more frequent in northern European Caucasian populations, and so forth.

We have been using principles of genetics for a long time with regard to animal and plant breeding, and all of these things to one degree or the other may have effects like our new technology. One might think of them as unwitting genetic technologies, one way or the other. Their consequences, like mutation, result in change by chance. It is not clear to me that this is morally or scientifically preferable.

So it's not likely that this is going to go away. We cannot legislate away genetic technology and knowledge. Nor can society prevent changes in the human condition. What we might be able to do is to say that the only acceptable ways for genetic changes and evolution to occur in human population is by accident, that is we just wait for random mutations and see what happens.

Maybe humans disappear as a consequence. maybe we get stronger, more healthy or whatever. But the truth and fact of the matter is that this is not a simple predictable process. For me, an important question concerns choice. Is it better to have some control or is it better to rely on the whim of chance and forces of nature. I'm not sure that I know the answer to that, but I think each of us ought to ask ourselves such questions and encourage open debate.

There are questions in the education arena that our society should be addressing. If genetic technologies are available which could identify children at risk for various kinds of educational problems, can they and should they be used to make appropriate services available to those children in order to allow "better" outcomes (however that is defined). What if, instead, they were used to segregate and deny services to at-risk categories of individuals? These are serious questions for our society.

A similar set of issues arises with regard to disability policy. Should the presence of a disability or the predisposition to develop a disability that can be determined through a genetic test, be allowed to be used if that discriminates against individuals? That would be quite contrary to the philosophy behind the Americans with Disabilities Act, and much of the disability movement in this country would be very, very distressed by such an approach.

I must comment briefly on professional standards. For the most part, the health care community has not considered or developed appropriate standards for the application of these kinds of tests. We have also not adequately investigated quality assurance processes for these tests.

These tests are not as simple to apply as they might seem. Many of my colleagues who run research laboratories seem to have a simplistic notion of quality assurance. If they can do the test and get more or less the same result three different times in a row, they regard a test as a highly quality controlled kind of process.

Unfortunately, it doesn't work that way. When there have been attempts to compare results using the same test in different laboratories, it has often been discovered that the results do not correlate well.

The whole set of principles that will be used to regulate genetic testing devices is almost completely undeveloped in this country by comparison to what we do with other medical devices. The Food and Drug Administration has developed very little in the way of capability to deal with this set of issues, and at times even seems reluctant to address the issue.

And the same is true of Health Care Financing Administration (HCFA). HCFA has responsibility under the Clinical Laboratory Improvement Act and amendments to regulate various kinds of medical testing devices and services. But they, also, are ill prepared to understand the issues. CDC is not much further along, though there is an attempt within that agency to develop a strategy and capability. Likewise, most state agencies are ill-prepared.

However, some state agencies have made real strides in these areas. Our public health laboratory in Iowa, for instance, has a number of people with special training in molecular biology who are really very knowledgeable in this area, and some other public laboratories have started to develop their capabilities around the country. But the rule is that most of these personnel are inadequately prepared to understand the concepts for the issues, or to apply them with respect to regulatory or quality assurance kinds of policy.

I will close by reminding you of the number of policy issues awaiting resolution. The fact is that we lack any coherent comprehensive national strategy to address this new technology and its implications across a wide range of public policy areas. We have failed to address and coordinate the roles of federal agencies regarding genetic health services. We certainly have inadequate state and local resources to address these problems at that level. There is an absence of laboratory and practice standards with regard to the application of these technologies, and there is insufficient knowledge among the public in general and among policy makers in particular.

It seems clear to me that we have a wide range of needs now, and that we need to begin to think in terms of research directions. Some of these are beginning to be tackled through the Ethical, Legal, and Social Implications branch of the Human Genome Project, but

there are far more of them to be tackled than there are resources for that program. It's clear that many of them could and should be tackled, policy makers and people from the academic and public arena in all kinds of agencies and institutions. We should be drawing together, ethicists, lawyers, the general public, scientists, physicians and people from all walks of life.

I'll leave you with a few questions about these disorders and their application. Hopefully they'll prompt you to think about your own questions and your own contribution to the answers which we all need.

(1) Our new health care reform system, whatever that's going to look like, will children and adults with genetic disorders have access to basic and appropriate health care? (2) Will genetic technologies be included among the health care services in our efforts to reform the system? (3) What will be the financial impact of these technologies? (4) How will we move to preventive health care strategies using this technology? (5) Who will have access to health-related genetic information? (6) Where will health care providers receive the necessary training to integrate genetics into our health care system? I'll be happy to have questions.

Mr. Cooper: Would you wish to comment on the need for pretest counseling and confidentiality of genetic information?

Dr. Hanson: Well, with regard to pretest counseling, it seems to me that whether we're talking about a genetic test or any other kind of test, it's always a good idea to tell people what it is you're doing and why you're doing it.

That, of course, is a simple answer, but in the case of applying a genetic test to an individual, I think it becomes especially important. People don't necessarily understand what kind of information they're likely to get out of having that test done, and there are a lot of unintended consequences that can occur.

Let's use the example of a woman going in for prenatal diagnostic test. I think most women have heard of doing amniocentesis for the prenatal detection of a chromosome disorder in a baby, but I think most women don't know what the wide range of chromosome disorders that may be found.

They may have heard of Down syndrome and they may have an impression that it's very severe or conversely not very severe. But let me ask you whether or not you think they've heard of trisomy 13, which is generally lethal within the first year of life, or trisomy 18, which is also lethal in early childhood. Have they considered Turner syndrome, which is a disorder that would make a woman short and infertile, but otherwise be quite compatible with a healthy life or very good quality of life? Have they heard about small genetic variations in chromosomes which we don't fully understand? That's a relatively common kind of outcome.

If a woman hasn't thought about those possible outcomes in advance of doing the test and she gets a result that is different than what she'd heard of, or if she gets indeterminant kind of advice (e.g. "Well, we think there's something a little odd here but we really don't know what it means!") do you think she's going to be happy that she had the test?

Because of our lack of full understanding of the implications of these tests and the

frequency of unintended outcomes, it becomes especially important to try to do some kind of pretest counseling. Unfortunately, if you don't have knowledgeable providers who know what those outcomes may be, it's difficult for them to do really meaningful counseling.

The other side of this, of course, is that if you ask providers to do pretest counseling, it takes a precious commodity: time. Time is money in this society, and anything that we do like this to really try to ensure knowledgeable choices on the part of people, will either make the system less efficient or more costly. I think we need to balance those priorities.

Now, there was a second part of your question?

Mr. Cooper: Confidentiality.

Dr. Hanson: Confidentiality, of course, is an issue about which we're all very concerned. I endorse maintaining confidentiality.

The problem is that with genetic tests, you learn something about people that are not present. If I do a test on someone in this room, I may discover that they have a genetic condition, but this implies that probably one or both their parents have that same condition or genetic factor that their brothers and sisters are at a high risk for that same condition or genetic factor as well as their offspring.

What does the clinician do if a parent says, "I don't want to know," but the child says, "I do want to know," and has the test done; and has a result and it's positive? Suppose the grandfather had the same disorder. We would know that the intervening person has got the gene and they already said they didn't want to know. Now, how can you protect their confidentiality or their privacy?

A more common situation is misstated or misidentified paternity. It's not uncommon to discover, in the course of genetic testing in a family, that there were unacknowledged people who contributed to this family.

Mr. Cooper: I'm sorry I brought that up.

Dr. Hanson: It's grist for a lot of lawyers, I'll tell you that. Some people have suggested that's where lawyers come from.

Ms. Weber: It seems to me after hearing a lot of the discussion this morning that the science seems to be ahead of our national approach to some sort of cohesive system.

Do you have any suggestions or are there any national approaches underway that would pull this together and give us some sort of federal approach, or is this going to be in a state of disarray and allowed to be left to go to every laboratory in a state?

Dr. Hanson: Well, I fear the latter. I'm not aware of a plan. We're going to hear one federal policy maker address the group this afternoon about his suggestions as to how we manage some of the data issues. I'm always pleased when somebody takes the time and effort to think their way through a complex set of issues like that.

But when you try to look across these policy arenas that I've tried to highlight here, I don't really know of any proposal or authorized process. Now, the ELSI project certainly

has people coming from a wide range of backgrounds who have acquired an overview o many of these different issues, but that particular advisory group for that particular project, which is based at the National Institutes of Health, has essentially no authority to do anything other than to conduct research or to identify problems and suggest policy recommendations or options.

They can't compel attention, and many other agencies are paying very little attention to what they're doing. Other agencies may know that something is going on, but they have not evinced any very serious interest in coming together in any coherent or planned way.

There are discussions between the Department of Justice, and EEOC and other groups. CDC is talking to NIH a little bit and vice versa. But I think at the present time there is no coherent plan.

Member of the Audience: My name is Hans Goerl and I'm the director — this is sort of in response to what you're saying. I'm the director of the Genetics Center, which is currently located in Hagerstown, and we are a public service agency, nonprofit, which is dedicated to the prevention of and eventually the response to genetic discrimination, which is discrimination by insurers, educators, employers, on the basis of somebody's genetics as opposed to their behavior or their actual physical disease or condition.

I have found that a lot of people just don't understand this issue, and I've proposed a series of hypothetical questions and I'd like to ask you one which is more designed to stimulate thought than anything else.

Dr. Hanson: Should I hide now?

Member of the Audience: Somebody's got to start thinking about this stuff. Somebody's got to start getting people motivated and that's what I'm trying to do.

In 1927, after a number of states had enacted eugenics laws which called for the mandatory sterilization of people who were designated to be criminals or detectives, the Supreme Court ruled that those statutes were not unconstitutional and that it was not unconstitutional for a state to require sterilization of someone who was likely to have a genetically defective child.

What happened basically, the case that Supreme Court ruled on was that a young lady by the name of Carrie Buck was the child of a retarded mother. Carrie had a child at the age of 17 or so. Some doctors from the state of Virginia looked at this child, who at the time was three months old, said, yeah, the child looks kind of like she has a funny look in her eye, she must be retarded, too.

The state of Virginia then said, okay, Carrie, under our eugenics laws you have to be sterilized. That case went to the Supreme Court and the Court ruled that the state had a compelling interest in saving itself the expense and inconvenience of becoming responsible for such children.

Therefore, it was constitutional for the court to sterilize these children. Judge Oliver Wendell Holmes wrote the opinion and came up with the immortal legal words that "three generations of imbeciles are enough."

My question is: Do you perceive a serious danger that current genetic technology, with the interplay of the complete absence of any constitutional protections, could result in

politically motivated genetic discrimination in mature individuals of this society?

Dr. Hanson: The answer is yes, of course.

Member of the Audience: What can we do about it?

Dr. Hanson: I would point out, of course, that potential for the discrimination has been present for a long time. As your example illustrates, in the absence of any new genetic technology, people were making bad public policy and unethical choices, based on "bad" science. I think "bad" science and "bad" information rarely lead to good public policy. It if ever does, it's really very fortuitous.