HEARING
BEFORE THE
COMMITTEE ON
LABOR AND HUMAN RESOURCES
UNITED STATES SENATE
ONE HUNDRED FOURTH CONGRESS
SECOND SESSION
ON
EXAMINING RECENT DEVELOPMENTS IN GENETICS RESEARCH, PUBLIC POLICY ISSUES WITH REGARD TO ACCESS TO AND USE OF GENETIC INFORMATION, AND THE IMPACT OF GENETIC TECHNOLOGIES ON CERTAIN SECTORS OF INDUSTRY, HEALTH CARE DELIVERY SYSTEMS, AND THE PUBLIC

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(III)
have had no training; similarly, other health professionals as well are in that category.

I am happy to tell you that we concluded yesterday a day-and-a-half meeting to organize what will be called the Coalition for Health Professional Education in Genetics, with enthusiastic support from the American Medical Association and the American Nursing Association. This coalition is set up to generate curricula, to distribute that information, to give health care providers of all sorts access to this kind of information.

So, Madam Chairman, we are in the early days of a genuine revolution in medicine, spurred on by these revelations in genetics and the Human Genome Project. All of us are likely to be touched by the consequences since all of us carry specific susceptibilities to future illness. There are, I regret to say, no perfect genetic specimens, not even in these hallowed halls.

Will this revolution, however, realize its promise to alleviate suffering and improve human health in dramatic new ways or, as Senator Domenici has sounded the alarm, will the irresponsible misuse of genetic information deprive individuals of peace of mind, health care or employment, leading to a painful backlash which will be very difficult to recover from?

I want to assure you that I am bullishly optimistic that we can achieve an outcome that will truly benefit the American public and grateful that this committee and you especially, Senator Kassebaum, by organizing this hearing, have recognized the importance of legislative protections against this kind of misuse. The hope of a better future for Jane and for all of us depends on the success of that effort.

Thank you.

The CHAIRMAN. Thank you very much, Dr. Collins.

[The prepared statement of Dr. Collins may be found in the appendix.]

The CHAIRMAN. I am sorry there is a vote in progress, so if Dr. Rothenberg and Dr. Holtzman could wait for a few minutes, we will be back shortly.

[Recess.]

The CHAIRMAN. The hearing will please come to order.

I appreciate very much your patience, and I hope we will not have any more votes for a while.

Dr. Rothenberg—or Professor Rothenberg. I do not know which you prefer.

Ms. ROTHENBERG. Either one is fine. Thank you.

Madam Chairman and members of the committee, thank you for the opportunity to be here this morning. I have a prepared statement for the record. What I thought I would do is to start by talking a little bit about what I consider probably the most important public policy question that needs to be addressed, and that is: Is genetic information really different? What makes genetic information different than medical information?

Then, I would like to talk very briefly about the major public policy goals that I think we have to reach some consensus on before we are able to figure out the details of which way to go with legislation.
Then, maybe very briefly—and all in my 5 or 6 minutes here—I would like to give you a framework of some of the activities that have been going on at the State level, both with respect to insurance and employment discrimination, which appear to be two of the major concerns already raised this morning, and then leave you with a few public policy challenges that hopefully will lead into Dr. Holtzman's remarks.

Genetic information is very personal and unique. It is powerful in that it can change the course of our lives once we get this information. It is potentially predictive about our future. It is pedigree-sensitive, meaning that it is not only information about us, but information about our families, our siblings, our cousins, blood relatives that we may not have spoken to in 20 or 30 years, but we are now connected to. It is permanent in that it cannot be changed, and finally, it is prejudicial. We have a history in this country of using difference about genetics and eugenics, and unfortunately, we have a bad history in our country and throughout the world of trying to use difference in genetics to discriminate.

So that in the end, a lot of these “Ps” that I have just emphasized are not really so unique in that they are only genetic, medically. I think that as a scientific matter, it might be very difficult, and any good lawyer, never mind any good scientist, could figure out an argument to say, well, but this other sort of medical information is like that, too. But in the end, and perhaps at the margin, which may be most important when we decide which way to go, it does have different social implications in our society. We know this both historically with respect to our past; we know this with sickle cell in the seventies; we know this with Jews around World War II. There are numbers of ethnic groups right now that are concerned about genetic information that we are discovering at this point. The Native American communities have concerns, the African American communities, the Ashkenaze Jewish communities. So we have to be very sensitive about how this information gets used and what its implications may be for the future of our program.

That then raises for me the question, what are the policy goals of trying to take some action in this area, and I want to just highlight six.

First of all, would we want the fear of those issues I have just raised, or those concerns for discrimination, to prevent research? If genetics research is a positive thing, and genetic information is a positive thing, we would want to figure out ways to get at these fears of being hurt by genetic information.

Two, we would want to be concerned that we do not coerce people to be tested because we might want to use this information for social purposes.

Three, we want to be very concerned about preventing what I call “genetic myopia,” that everything not be explained by genetics. There are many other things in our environment and in our histories and in the way we live our lives that are not genetically-based, and we cannot just look and put blinders up to everything that is going on around us.

We have to make sure that the information that we do get does not get misinterpreted, so we need good training, not only of people
who are giving information but of people who are hearing informa-
tion, both providers and consumers.

And finally, we need to be concerned about the quality of this in-
formation—is this good information—because if we are going to
take social risks—and there are going to be social risks with the
more information that flows—the more this information and ge-
ettologic testing gets into the market, the more we let it into the mar-
et, the more flow of genetic information will be out there. So we
want to make sure that it is good information, that it is worth tak-
ing that risk.

That finally leads me to how do we then prevent these harms as
best we can. Let me just focus on two. First of all, there is the issue
of health insurance. This is an issue that has developed increasing
interest over the last few years, and now we have close to a dozen
States at least that have to varying degrees set up a legislative or
analytical framework for prohibiting to varying degrees discrimina-
tion based primarily on genetic test results.

It started in 1991, this new set of legislation, in Wisconsin. They
prohibit requiring or requesting a genetic test to get coverage; they
prohibit requiring or requesting the results of a genetic test—what
I call the privacy component or the confidentiality component of
this legislation. They prohibit conditioning coverage or benefits on
genetic testing and prohibit considering the testing in the deter-
mination of rates.

So it is not only whether you get and keep your coverage, but
what implications it has on your rates, because if the rates are so
high, for many, that basically means you do not get coverage.

The problems with this approach on a national level are that,
first of all, it focuses narrowly on a test result rather than more
broadly on genetic information. And as you have already heard, ge-
ettologic information is a lot more than a DNA test. So it does not in-
clude information that could be obtained by other means, such as
a family history or medical examination or the records. And just as
important for those of you in Congress, self-funded plans are ex-
empt from these State insurance laws. The ERISA exemption in
some States allows over half of all of those employees who are cov-
ered through their employers, those insurance plans are not cov-
ered under any of these State prohibitions.

So we have a patchwork, and at this point we have a momentum
going, but certainly not any attempt at a consistent public policy
with respect to health insurance discrimination. At present, in Wis-
consin, Ohio, California, Colorado, Georgia, Minnesota, New Hamp-
shire, Oregon, Virginia and Maryland, and waiting for the Gov-
ernor’s action in New Jersey, we have this legislation to varying
degrees. Also, about 20 States just between 1995 and 1996 have in-
troduced legislation in this area.

So the momentum is moving, and it has made for what I call in-
teresting bedfellows between the biotech industry, which is very
concerned that we need to address discrimination if we are going
to be able to move the technology into the market, the scientists,
the researchers, and of course, the consumers, particularly the
women’s health advocates, who have been very active at the State
level and at the Federal level in trying to push this legislation.
In part, this came out of the working group, the National Action Plan for Breast Cancer and the Ethical, Legal and Social Implications Working Group of NIH and DOE last July, that made this series of recommendations that were published in Science. They pretty much follow that framework at the State level, but they add one or two important differences.

One is that they do not use genetic test; they use the more broad definition of genetic information. Second, they make very clear that written disclosure for any disclosure of genetic information is critical and that it needs to be made specific for each disclosure so that you cannot do blanket informed consents and you cannot do blanket disclosures or authorizations; they have to specify that you are determining as the individual where that information is going to go.

Now, just briefly, in my 30 seconds that I have left, if I might, I will proceed to genetic information in employment and in the workplace environment. In the workplace environment, there are three issues that you have to be concerned about. One is how is the genetic information going to be used to affect somebody's employment possibilities. Are they going to get a job? Are they going to be able to keep a job? Are they going to be promoted? Is it going to demote them if the employer has genetic information? How is it going to affect the terms and conditions of their employment?

Second, what implication does it have for health insurance since, as I just mentioned, most of us get our insurance through employers, and a large percentage of us are in self-funded plans.

And third, what implications does it have for privacy? Even if you are not going to be discriminated against in a particular context, who has the right to know this very private information?

The EEOC in 1995 did issue a guidance in its compliance manual that did specify that its interpretation of the Americans with Disabilities Act does apply to individuals subjected to discrimination on the basis of genetic information relating to illness, disease or other disorders, but that has some limitations. First of all, it is not a rule; it is an interpretation. It has not yet been interpreted in a court. We have not in fact had a case yet. It does not cover unaffected carriers of recessive or x-linked disorders, and there is still really nothing that prevents employers from getting a general medical release once there has been a pre-offer of employment.

There have been a few States, just like insurance, that have also addressed employment discrimination or the use of genetic information in the workplace. Not by accident, many of these are in fact the same States often that have integrated these protections at the same time they have integrated protections in insurance, and they typically also prevent the use of genetic testing as a condition of employment; you cannot test without consent, and it cannot be used in any way to affect the terms, conditions or privileges of your employment. We have had action in that area as well. Again, the problem is that they tend generally—with the exception of the State of New Jersey, where we are waiting for the Governor's signature—to primarily focus on the genetic test. Again, the employer is not prohibited from requiring a general medical release, and they focus on the use, not the access to the information, so the burden
is on the employee to have to prove that he or she has been discriminated against, which is often very difficult.

The States to varying degrees—Oregon, New York, Wisconsin, Iowa, Rhode Island, New Hampshire, and waiting for the signature in New Jersey—have in fact passed legislation on employment, and a number of other States have introduced bills.

I will skip at this point but will be open for questions about additional issues with respect to privacy. I think it is clear to all of you to know at this point that we presently have a patchwork of privacy protection with respect to medical information and with respect to genetic information throughout the United States and that there is an attempt to try to get at a Federal solution. But I need to warn you that any attempt to get at a Federal solution should not preempt the stronger State laws currently on the books which have been integrated into these anti-discrimination statutes. We have some—not great, but some—privacy protection at the State level which I would hate to lose with any Federal solution.

Let me finally make the transition to what I consider the most important public policy challenge. As we move into increasing proliferation of genetic testing in the marketplace, or if we should move, when is it appropriate to move this testing? How good does the testing need to be? How good does the information need to be before we generate all this flow of information? When is it appropriate to make the transition from research to the market? Is it appropriate before we have these protections in place, and if so, how can we protect individuals from benefiting from this information, not from being harmed?

At that point, I will stop and make the transition to Dr. Holtzman.

Thank you.

The CHAIRMAN. Thank you very much.

[The prepared statement of Ms. Rothenberg may be found in the appendix.]
of the sort you describe, which I agree are critical if we are going to get better answers.

So everyone enrolled in this network will be followed prospectively, but probably a significant subset will then enroll in some comparison of this approach versus that approach in order to get follow-up information. Without the kind of network that I was describing, it is difficult to know where the patients will come from for the trials that you propose and I think rightfully so will be needed to get really rigorous answers.

The CHAIRMAN. Professor Rothenberg, from your experience with the different States that are getting into this, is there a common trend that you have noted? Is this something that the insurance commissioners are working with? Have they developed some standards yet on this?

Ms. ROTHENBERG. It is interesting. There has been an interest of the health insurance commissioners. The life insurance companies are also very interested in this issue. The common trends have basically been, since Wisconsin's law passed, to varying degrees and affected in part by the workshop recommendations last summer and have followed these four components to varying degrees that I explained—the prohibiting or the requiring or the testing, the results of the testing, something about privacy, about disclosure and about not having an impact on coverage or on rates.

However, what you tend to see is that a number of States where laws have passed are States that are also doing health care reform, so they have already done some things about community rating or portability, and there is not a lot of medical underwriting that is continuing to go on in certain groups, but depending on what State you are looking at, they might have that reform for large insurance groups; other States will have it for small groups or for individuals. And I remember talking to the delegate of New Hampshire a few years ago when they passed both an employment and an insurance bill at the same time, and they said the insurance companies had dealt with community rating—the year before, they had had some reforms—so the health insurance companies were not that concerned.

In the States that have attempted to address life insurance at the same time, that tends to be where some of the battles are, and even in the State of Maryland, which this year passed a law just with respect to health insurance, the life insurance companies were very concerned and testified.

So one theme that I see is that if you attempt all at once to do everything, you tend to have more people coming out of the woodwork—but some States have done that successfully or almost successfully. New Jersey did attempt and is awaiting the Governor's signature to try to handle a number of things at once—employment discrimination, health discrimination, life and disability—although they handled it somewhat differently—privacy issues, samples, and enforcement.

On the other hand, New York, which has also been quite active in this area, attempted an omnibus bill as did a number of other States attempt to establish commissions to study it, and then they come back and come up with how we are going to solve everything. New York had to "chop it up," as they have said to me; they
chopped it up into a lot of different bills—a health insurance bill, something dealing with life and disability and health, something dealing with privacy, and something dealing with employment. The Governor signed an employment discrimination bill just a few weeks ago. They were not successful in New York in any major reform in insurance at all. They got a minor reform through with respect to informed consent. So it is hard to generalize about whether to go with an omnibus or a very specific bill.

I think that continuing over the next year, probably the biggest push will be in health insurance, and I think we will continue to see a number of States, assuming the Federal Government does not do something first, continuing to address it. There is increasing momentum for health insurance, but it is easier in States that have already attempted to address some level of health care reform.

The CHAIRMAN. Thank you very much.

Senator Jeffords?

Senator JEFFORDS. Ms. Rothenberg, I would like to talk to you about ERISA—everybody else will tune out now.

The CHAIRMAN. Senator Jeffords is one of the few people who likes to talk about ERISA.

Senator JEFFORDS. It is not that I like to, but—

Ms. ROTHENBERG. As I tell my law students, anybody who is interested in ERISA will get a great job.

Senator JEFFORDS. Can I sign up for your course? [Laughter.]

Ms. ROTHENBERG. Oh, I do not teach it.

Senator JEFFORDS. It seems to me that when we created ERISA, the purpose was to try to ensure good protection for employees, but also to make it unnecessary for employers to comply with all the different States' laws.

Now, with respect to genetics, we are talking about more than two areas, but certainly one is discrimination. So my first question is: Is it necessary to reform ERISA in order to protect against discrimination, or do we have to modify the EEOC, or are the State laws protecting employees sufficiently?

Ms. ROTHENBERG. Well, political reality is part of your question, as well as a practical reality and an ideal world. It seems to me that in the ideal world, we would try to do something at the Federal level not only to get at your ERISA concerns, but also because we have a tremendous patchwork. If you see discrimination as a civil rights issue, and you deal with it at this patchwork level through a variety of either insurance laws or employment laws or whatever, and somebody lives in one State and works in one State and gets his insurance in another State, it gets pretty confusing. So if we even just put ERISA aside, it might be a justification for trying to do something at the Federal level; but when you add ERISA to it, if you have two people living next door to each other, like on my street in the State of Maryland, and one of us in a self-insured plan and one of us is not, right now, one of us might get some protection, and one of us might not.

So you need to take some Federal action if you are concerned about making sure that everybody in that State or everybody in this country gets some sort of protection.
Now, with respect to the EEOC or the ADA, I might say, there might be parts of the ADA that could be strengthened to make sure that there is not discrimination in the workplace, but if we could even get at the health insurance issue and have a level playing field with respect to the use of genetic information, then everybody would play the same, and there would be fewer incentives perhaps to try to pick out who would be the cheaper people to have in an employment setting where the employer to a large extent is paying for the health insurance claims.

So I guess my short answer to that is that, short of a Federal action, there is not going to be a way for every employee to be treated the same.

Senator Jeffords. So from our perspective, you are saying we should act.

Ms. Rotherenberg. Yes—however—and I know you have a lot of other things to do, but I was going to say yes, you need to act, but I do not want to say we need to throw the baby out with the bath water. There are some good things that some States are doing, and we have to learn from what those States are doing, and we have to build on it. We do not want to create more conflict, but where we can avoid patchwork, and it gives people protection, that is what our goal should be.

Senator Jeffords. Well, we do not plan to do anything this year, I am sure, but anyway—

Ms. Rotherenberg. Oh, I do not know. [Laughter.]

Senator Jeffords. I am very interested in this area. Especially in the insurance area, I think there are ways that we can handle this. But it will take concepts that we have not really utilized, such as establishing reinsurance to take account for certain risks so insurers have no incentive to discriminate. There are other ways to deal with risk through reinsurance and things like that. But I am extremely interested in this, and you have answered my question as far as the future goes, so I will stop at that point.

Thank you, Madam Chairman.

The Chairman. You will work on it in the next Congress, if not before.

Senator Jeffords. Right, yes.

The Chairman. Senator Frist?

Senator Frist. Thank you, Madam Chairman.

I have several fairly quick questions. Dr. Collins, one of the provisions being considered today in the privacy legislation and in a number of the bills that have been proposed or suggested is the written authorization for the collection and analysis of DNA samples and long-term storage for samples in preparation for potential use in the future. It is a very controversial issue. Could you paint that picture for us, the controversy involved, and I guess even more specifically, what effects that will have, potentially adversely or maybe positively, on research in the future?

Dr. Collins. I think that is a very important question, Senator. I think the generally principle that if genetic information is private, it should not be given to some third party without your permission underlies a lot of the efforts to protect genetic privacy. But having said that, I think one has to consider the consequences of any particular language that seeks to achieve that goal. And I
think in particular, we must be careful in trying to put those protections in place that we do not do severe damage to research enterprises that great benefit the American public particularly in the fields of epidemiology and pathology.

Pathologists, for instance, have made many important observations by studying tissues that have been stored, sometimes for decades, from individuals who have had operative procedures in the past and from which much can be learned, particularly in the field of cancer, about what molecular problem gave rise to their disease.

It is totally impractical to imagine going back and contacting each one of those individuals and seeking their permission in order to do a simple analysis about whether or not this particular oncogene had an alteration or not. And if we were to put legislation in place that would require that kind of re-contact, we would destroy a very important area of research.

Many of us believe that those issues can be dealt with by the simple process of rendering samples anonymous, that is, stripping them of all the identifiers that would allow you to determine who did this come from prior to carrying out the analysis. A number of groups that have met to consider this issue have all come up with that same general conclusion, and that in fact is consistent with the current guidelines for the protection of human subjects as codified in 45 CFR 46.

However, I think not everybody has completely grasped onto that, and I do think we should be very careful in the process of putting together legislative efforts to be certain that those issues are thought about.

Concern has been raised as to whether in fact you can ever render a sample anonymous if you are analyzing DNA, since our DNA sequences are unique. And while that is a serious issue if you are planning to do a great deal of analysis where you might learn enough detailed sequence information that that sample could have only come from one person on the planet, in general that is not the task which pathologists and epidemiologists have in front of them. They are interested in looking at a very small number of sequence changes, and given that 99.9 percent of all of our sequences are identical, the likelihood that you are going to reveal the identity of a particular individual inadvertently through this kind of research is in most instances rather remote.

Senator FRIST. Is that one of the principal objections?

Dr. COLLINS. That is one of the principal objections.

Senator FRIST. That, in spite of what we had in legislation, the identification could still be made.

Dr. COLLINS. It could still be made.

Senator FRIST. Ms. Rothenberg, did you want to comment?

Ms. ROTENBERG. Yes, I think that that is a major concern, but I think there also is the importance of educating the public because I think if you asked most people who went into an operation and had some of their tissue stored over a long period of time—and they went in for a clinical purpose, not for a research study—their expectation would be that it was being stored for them. I do not think most individuals know or understand that some of the tissue specimens or DNA samples that are being stored are being used for other purposes and for research. Not that we would not do this, but
we need to be very careful that we educate the public so that there again is not a backlash about using genetic information to hurt us, particularly ethnic groups. So that if I go in as an individual because I have breast cancer, and my specimen is being stored, and then somebody later on wants to figure out if my particular ethnic group either has a higher propensity for a disease or some behavioral genetic study, we have to be very careful about what that is going to be used for. And that is partly the reason why there is so much interest prospectively in getting consent about it being used because it is an attempt to also educate the public about the future uses.

Senator FRIST. Have the States addressed this any better at a State level—clearly, some States are much further along in this whole issue, and you made that point quite eloquently—in this particular arena, are there certain States that have addressed this legislatively that we can look at and learn from?

Ms. ROTHENBERG. Some States have begun to address it, not in any real comprehensive way. Some have addressed it in a way of saying that your sample will be thrown out unless you give informed consent. That is a concern to many researchers and to many pathologists and other groups.

I think we need to have a lot more dialogue—there has been a lot of dialogue, but I do not think the dialogue is necessarily having to pass a lot of procedural laws first; I think we really have to do a lot more to educate people about expectations, and there needs to be a discussion also at the institutional review board level about what research needs to be done and what does not and what the implications are, not only for individuals, but for their groups as well.

Senator FRIST. Thank you.

Just real quick, 45 seconds, Dr. Holtzman, on the top part of your chart and the work of the Task Force on Genetic Testing, when you say “available for medical practice” in terms of the criteria before and what needs to be present, what does that mean, practically?

Dr. HOLTZMAN. Let me give an illustration. There is a test that looks for a common variant of a protein, the apolipoprotein-E4 test, that has been found to be associated with Alzheimer’s disease. In fact, this test has been available for use in conjunction with heart disease diagnosis for a number of years, but it is now being used occasionally in standard practice outside of investigational settings for people who may want to know what their risk of getting Alzheimer’s disease in the future is. And my definition of “medical practice” is one where the test is not being used under a research or investigative setting, that there are physicians out there who know about the availability of the test and can order this test in what they perceive to be either their benefit or the benefit of the patient, without this test having been rigorously demonstrated to provide meaningful and safe and effective information. And I think that that is the problem with this apo-E4 test for Alzheimer’s disease. We have not yet had a sufficient investigational phase, which I talk about at the top part of the poster, to assure us that this test should be out there for predictive purposes.
Professional and public education: As more genetic tests become available to the public, the use and interpretation of those tests and the information they generate will no longer be managed by genetic specialists alone. There will be too few professionals with the advanced training in genetics to meet the expected demand for genetic testing and services. As patients ask more questions about genetic tests and disease risk, responsibility for the use and interpretation of tests and genetic information will increasingly fall to primary care physicians, nurses, physician assistants, and public health professionals who are not specialized or trained in genetics.

Therefore, it is imperative that all of our Nation's health care professionals have the knowledge, attitudes, skills and resources they need to effectively integrate genetics into the diagnosis, prevention, and treatment of disease, and to address the related ethical, legal, and social issues. In 1995, NCHGR held a meeting of health professional and education experts to define education priorities for the ELSI grant program. These experts concluded that while public education is important, professional education is of the highest priority, because most individuals will seek genetic information from medical professionals. To this end, NCHGR is now working with professional organizations and consumer groups on a proposal to establish a National Coalition for Health Professional Education in Genetics. Yesterday, we concluded the first planning meeting of the Coalition and I believe these successful discussions will provide the necessary direction for our health professionals in this important area.

CONCLUSION

Congress, and particularly this Committee, has long been the leader in supporting genetic research and the Human Genome Project. Therefore, it is fitting that this Committee is now taking the lead in addressing the important policy issues raised by this research. Protecting privacy and preventing the misuse of genetic information is essential not only for us to reap the benefits of this research but indeed, in order to carry on the research at all. Increasingly we hear of people unwilling to participate in genetic research out of fear that information about their genes might be used against them by insurers or employers. Genetic discrimination has been hailed as the "civil rights" issue of this decade. We have the unique opportunity to address genetic privacy and discrimination issues now as the scientific information unfolds, before we find ourselves in a full-blown crisis. I look forward to continuing to work closely with the Congress to develop sound policies to ensure that the Human Genome Project and new genetic information is used to benefit, not harm, the American people.

This concludes my remarks. I would be pleased to answer any questions you may have.

PREPARED STATEMENT OF KAREN H. ROTHENBERG

Good morning Madame Chair and members of the Committee. I am pleased to appear before you today to address the public policy challenges raised by the use, misuse, and access to genetic information. Toward this goal, I will first examine whether genetic information is different than other types of medical information and whether it requires a special public policy approach. I will then examine what role legislative approaches may play in addressing the use, misuse and privacy of genetic information, particularly in the health insurance and employment context. I will conclude with highlighting emerging public policy challenges posed by the future proliferation of genetic tests in the marketplace.

Genetic information is personal, powerful, predictive, pedigree-sensitive, permanent, and prejudicial. As a result, it is information people commonly wish to keep private, although DNA databanks and computer technologies make protecting people's privacy increasingly difficult. Most individuals expect that all medical information should be protected. The potentially harmful risks associated with genetic information may demand that we pay special attention to its use, misuse, and privacy.

Genetic information has implications not only for the individual, but also for his or her blood relatives, including parents, siblings, cousins and future offspring. Thus, the intergenerational impact of genetic information (and heritability) makes the risk for misuse, including stigma and discrimination, significant and unique. Genetic information may be linked to certain ethnic and racial groups, many of whom have suffered from discrimination and eugenic policies that historically were "justified" by genetic findings. For example, restrictive immigration laws against Eastern Europeans in the 1920's, sterilization policies, Nazi atrocities, and insurance and employment discrimination against carriers of the sickle cell trait were justified by the power of genetic information. Even the recent discovery of specific gene mutations that may be associated with higher rates of breast and ovarian cancer
in the Ashkenazi Jewish community has raised concerns about how this information may be used to discriminate against them. The African American and Indian communities are also very concerned about behavioral genetic studies on violence and alcoholism.

An individual's genetic makeup is unique and cannot be altered. Even though a predictive test result is not a diagnosis, it is still powerful information and there is risk for misinterpretation by both providers and patients. People may believe that their fate is predetermined genetically and there is nothing they can do to change it.

The fear of genetic discrimination in the health insurance context cannot be underestimated. It is argued that individuals who might otherwise choose genetic testing will decline it based on their fear that they or their family members will not be able to obtain or maintain health insurance coverage. As a result, the future of research on the benefits and risks of testing for genetic conditions, including susceptibility to such common diseases as cancer and heart disease, may also be inhibited. Thus, as the mapping of the human genome continues to progress and new genetic tests proliferate, policy makers need to evaluate the development of legislative and regulatory strategies to address these concerns.

In the 1970's, a few states began to pass legislation that addressed genetics issues. North Carolina, for example, passed legislation prohibiting health insurers from refusing to issue insurance or charging higher premiums based on sickle cell trait or hemoglobin C trait. By 1991 a new generation of state legislation began to evolve with the passage of a Wisconsin law prohibiting health insurers from: requiring or requesting an individual or a member of the individual's family to obtain a genetic test; requiring or requesting directly or indirectly into the results of a genetic test; conditioning the provision of insurance coverage or benefits on genetic testing; or considering genetic testing in the determination of rates.

This approach attempts to integrate protection against discrimination in insurance practices, coverage, benefits, and rates with some privacy protection for the individual and his/her family. Similar approaches have been incorporated to varying degrees in recent legislation passed in about a dozen other states.

The development of public policy to address genetic information and health insurance must be analyzed in the context of a complex and inadequate health insurance system, the uncertainty about the future scope and impact of genetic testing, and the political realities of a pluralistic society. The current patchwork of state legislative approaches does not provide a comprehensive solution to genetic discrimination and health insurance.

First, with the exception of a few states, these laws focus narrowly on genetic tests, rather than more broadly on genetic information generated by family history, physical examination, or the medical record. Although insurers are prohibited from using the results of a chemical test of DNA, or the protein product of a gene, they may still use other physical/physiological (phenotype) indicators, patterns of inheritance of genetic characteristics, or even a request for genetic testing as the basis for discrimination. Thus, meaningful protection against genetic discrimination requires that insurers be prohibited from using all information about genes, gene products, or inherited characteristics to deny or limit health insurance coverage.

Second, a large proportion of the population receives health benefits from self-funded plans not subject to state insurance laws. The federal ERISA preemption prevents a statewide approach to regulating the use of genetic information by all plans providing health benefits. Furthermore, there are no federal laws specifically addressing genetic discrimination in health insurance that would extend to self-funded plans.

Recent health insurance proposals at both the state and federal level focus primarily on modest reform in the areas of accessibility, portability, and renewability of coverage, prohibiting insurers from denying coverage based on health status or medical condition, but often permitting exclusions for pre-existing conditions for limited time periods. Because it is unclear whether and to what extent genetic information would be covered, recent federal proposals, including the "Health Insurance Reform Act," introduced by Senators Kasiebaum and Kennedy, explicitly prohibit health plans from denying health coverage based on "genetic information." With these policy considerations in mind, the following recommendations were developed by the National Action Plan on Breast Cancer (NAPEC) and the Working Group on Ethical, Legal, and Social Implications of the Human Genome Project (ELSI) for both state and federal policy makers to protect against genetic discrimination:

1. Insurance providers should be prohibited from using genetic information, or an individual's request for genetic services, to deny or limit any coverage or establish eligibility, continuation, enrollment or contribution requirements.
2. Insurance providers should be prohibited from establishing differential rates or premium payments based on genetic information, or an individual's request for genetic services.

3. Insurance providers should be prohibited from requesting or requiring collection or disclosure of genetic information.

4. Insurance providers and other holders of genetic information should be prohibited from releasing genetic information without prior written authorization of the individual. Written authorization should be required for each disclosure and include to whom the disclosure would be made.

The recommendations further provide that genetic information be defined as "information about genes, genetic products, or inherited characteristics that may derive from the individual or a family member." An insurance provider is defined as "an insurance company, employer, or any other entity providing a plan of health insurance or health benefits including group and individual health plans whether fully insured or self-funded." As you know, a number of members of the Senate and the House with us today have taken a leadership role in introducing federal legislation that integrates these recommendations.

Another concern is how genetic information may be used in the workplace setting. It may have an impact on employment possibilities, health insurance and privacy. In order to protect against employment discrimination, the U.S. Equal Employment Opportunity Commission (EEOC) issued a guidance in its compliance manual in March 1995 stating that the Americans with Disabilities Act covers an individual with positive predictive genetic test results (citing cancer as an example) from discrimination in employment. This interpretation is yet to be tested in court, and does not address the employer's access to medical (and genetic) information. A few states have now passed legislation that prohibits genetic testing as a condition of employment; prohibits genetic testing without informed consent; prohibits the use of genetic test results to affect the terms, conditions, and privileges of employment; and prohibits payment or benefits to employees in return for taking a genetic test. As with most state legislation addressing health insurance and genetic discrimination, these laws also tend to focus narrowly on the genetic test. They do not prohibit employers from requiring a general medical release; further, the ERISA exemption also allows self-insured employers to alter benefit plans to reduce or eliminate coverage for specific conditions and procedures. Since most employers will continue to have access to genetic information, the burden will be on the employee to prove that the employer used genetic information to discriminate. Recent legislation in New York and New Jersey, however, does provide stricter limits on an employer's access to and use of genetic information. To date, there is no federal law that specifically addresses the use, misuse and access of genetic information in the workplace, although a few of the recent proposals integrate employment issues to varying degrees. The National Action Plan on Breast Cancer will join together again with the Working Group on Ethical, Legal and Social Implications of the Human Genome Project for a one-day workshop on October 4, 1996 to specifically address genetic discrimination in the workplace. It is hoped that, as a result of this workshop, we will develop guidelines for both state and federal policy makers in this area.

Anti-discrimination statutes also integrate various levels of privacy protection. However, there is currently a patchwork of legislative sources that address genetic privacy and confidentiality, including medical records confidentiality statutes, public health data bases and registries, public health genetic programs, criminal investigation, parentage and adoption statutes, and research regulations. There is no uniform approach to addressing genetic privacy. It is critical that any federal legislation to regulate genetic (and medical) privacy not preempt stricter privacy protections integrated into state anti-discrimination statutes. Furthermore, medical privacy legislation must specifically address protection of genetic information. Currently, federal proposals vary with respect to how they address these issues.

Perhaps our greatest public policy challenge will be to determine when, if at all it will be appropriate to make the transition from predictive testing for high-risk individuals and families within a research context to testing the general population. Should we have in place anti-discrimination protections and privacy legislation before we continue to expand genetic testing? How can we better quantify and qualify social risks? We must strive to resist a genetic "quick-fix" mentality that promotes genetic testing in the health care market, until we have a better understanding of the risks of genetic testing. Perhaps it is even more important that we continue the public policy debate and develop the strategies to ensure that genetic information is used to benefit, not to harm, individuals and their families.

[Additional material may be found in the files of the committee.]