U.S. DEPARTMENT OF HEALTH AND HUMAN SERVICES

SECRETARY'S ADVISORY COMMITTEE ON GENETICS, HEALTH, AND SOCIETY

Fifth Meeting

Monday,
October 18, 2004

Congressional Ballroom
Bethesda Marriott
5151 Pooks Hill Road
Bethesda, Maryland

IN ATTENDANCE:

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C O N T E N T S

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P R O C E E D I N G S (8:35 a.m.)

DR. KINGTON: My name is Raynard Kington, the deputy director of NIH. Good morning and welcome. It's a pleasure to be here representing Secretary Thompson and Dr. Zerhouni, the director of NIH, in opening the fifth meeting of the Secretary's Advisory Committee on Genetics, Health, and Society. This committee's work is increasingly important in light of ongoing developments in genetics and genomics. Your service on this committee helps in the Department, as well as other agencies represented around the table, to address and stay abreast of the broad array of complex medical, scientific, ethical, legal, and social issues raised by the development and use of genetics and genomic technologies, and we greatly appreciate your commitment to this work.

I'm here not only to welcome all of you to the meeting but also to mark several important changes in the committee's membership and leadership, and to honor the service of three exceptional people, only two of which, I believe, are here.

First I'd like to begin by recognizing Dr. Edward McCabe, the first chair of the committee. The Secretary appointed Dr. McCabe for his expertise and knowledge of the field and exceptional leadership qualities, and he did not disappoint. He was also appointed to provide a smooth transition between the predecessor committee, the Secretary's Advisory Committee on Genetic Testing, and this committee, which Dr. McCabe chaired for three years.

On behalf of the Secretary, let me thank you, Dr. McCabe, for your service over the past five years to the HHS mission to improve health and for your dedication as chair of the committee. You have ably guided the committee through its first year of life, through a systematic review and priority-setting process that led to the committee's study priorities, and through the development of consensus recommendations to the Secretary on genetic discrimination, genetics education for health professionals, and direct-to-consumer marketing of genetic tests. Your leadership really has provided the committee with a firm foundation for future work, and we've appreciated your service and all of the extra time the position of the chair involves.

We want to acknowledge your leadership demonstrated in the transition of the previous committee to the current committee, and as a token of our appreciation it's my pleasure to give you this certificate from the Secretary recognizing your service to the committee.

(Applause.)

DR. KINGTON: Another plaque, I'm sure.

(Laughter.)

DR. KINGTON: We'd also like to thank two members of the committee whose service is ending. Kim Zellmer and Brad Margus were appointed for their knowledge of consumer issues, bringing to the proceedings their own families' experiences with genetic disorders. They've each devoted an extraordinary amount of time and energy to the work of the committee both during and between meetings, and they have contributed in many ways to advancing our understanding of the impact of genetic and genomic technologies on families, individuals, and society.

Ms. Zellmer, on behalf of the Secretary, let me thank you for your work and commitment. I know that your personal experiences provided you with insights that have been extraordinarily important to the committee's deliberations. Your contributions to the committee's work on priority setting and genetics education have been especially valuable, and

we've appreciated your service enormously. We know that these are incredibly time-consuming appointments to committees like this, and we appreciate your time.

If you could come forward, another plaque.

(Applause.)

DR. KINGTON: Mr. Margus is not here. However, let me still thank him for all of his work and commitment to the committee. He, too, has brought important insights and perspectives, personal and professional, to the deliberations of the committee, particularly in the area of work on direct-to-consumer marketing. That's been particularly invaluable, and we have a plaque for him as well which we will deliver to him when he arrives.

Both of you can consider yourselves emeritus members of the committee, which means that you can be called upon at any time to work more.

(Laughter.)

DR. KINGTON: The challenges posed by genetic and genomic technologies are in many ways just beginning, and this committee is going to be incredibly valuable for the foreseeable future. As we look forward, we are pleased that we were able to identify an extraordinary person to follow as chair.

As I was driving over from the NIH, I realized that I knew Dr. Reed Tuckson for almost 20 years. Dr. Tuckson was two years ahead of me in the Robert Wood Johnson Clinical Scholars Training Program at the University of Pennsylvania, and when I arrived fresh from my medical residency, all of the cohort of people who began when I began were told in no uncertain terms that the bar had been raised considerably in terms of our performance, and that Reed in particular was going to be a tough act to follow.

For the last almost 20 years, 19, I've counted on Reed as a colleague and friend and mentor and have not hesitated to call upon him whenever necessary. We crossed paths repeatedly when he was president of the Charles Drew University in Los Angeles and I was at the RAND Corporation, and then later. Most recently he agreed to serve on one of the most difficult committees we have had recently, and that's the NIH Blue Ribbon Task Force on Conflict of Interest policies, which required an extraordinary time investment over a very short period of time in advising us on how to deal with an issue that's of great importance to the future of the agency.

Clearly, Dr. Tuckson has a tough act to follow as well here, but we know that you're up to the task and we have every confidence that you will, as you always do, perform admirably and make us all feel that we should work harder. Welcome, and I'll pass it off to you, Reed.

DR. TUCKSON: Thank you very much. Thank you for that.

I just want to give a couple of seconds to see if, even though he's going to be with us in deliberations for quite a while, but Ed did such a terrific job. As somebody who was on the first committee and watched this transition, I just marvel at what he was able to achieve as our leader both on the first and now this committee.

But, Ed, would you like to say a couple of comments, please?

DR. McCABE: Well, thank you.

It's been truly an honor to serve on the Secretary's Advisory Committee on Genetic Testing, and then the Secretary's Advisory Committee on Genetics, Health, and Society. One of the things that you will find, Reed, is that you have fantastic staff with Sarah in charge, and you will be wise to listen to what Sarah tells you. If you follow Sarah's

directions, you will look extremely good. If I've had any success, that's been the secret to it.

(Laughter.)

DR. McCABE: But also, the committee members are just fantastic on both of those committees. From some experiences on the first committee, I learned that you always had my back, and I hope that I can do equally as well for you. Thank you.

(Applause.)

DR. TUCKSON: Well, we have a lot to do, and thank God for each one of you. It's going to be very interesting to watch as so many of you are going to have a chance to really produce and showcase the work that you've been doing as committee chairs of these subcommittees and so forth. I'm just amazed, as I read through all of this material again, at how hard each one of you are working on this committee's behalf. We owe you a grade of thanks.

The public was made aware of this meeting through notices in the Federal Register, as well as announcements on the SACGHS website and listserv. I will tell you that as I've come to review a little bit about how do you evaluate the quality of advisory committees to the government, I am reminded as I have looked through some of those criteria that the number-one way in which these committees demonstrate their value to the nation is through its ability to reach out to the public. So I'm very pleased, and I think we're even going to try to do some other things which we can talk about later in terms of redoubling our efforts to have all of our work into the hands of the public that is most interested in this and those who ought to be more interested in what we're doing. So this meeting was made available through those notices.

We were rechartered again through August of 2006, so you're stuck.

(Laughter.)

DR. TUCKSON: The following ex officio agency representatives, we want to thank all of them that are here, but I want to make particular note that we are joined today also by Ms. Cari Dominguez, chair of the Equal Employment Opportunity Commission. She will be serving as the EEOC's ex officio.

Chair Dominguez, we're very, very pleased that you are taking the time. It really shows how important the issues are that you're here about, I know most that are of interest to you, how important it is to our country. So thank you for joining us.

Mr. Richard Campanelli, director of the HHS Office for Civil Rights, is also expected with us today, and that is also worthy of note.

Dr. Francis Chesley, director of AHRQ's Office of Extramural Research, Education, and Priority Populations, has been appointed to serve as AHRQ's new ex officio. Thank you very much, Dr. Chesley, for joining and serving.

We're glad for all of our other ex officios who have been maintaining their commitment to this committee.

Chris Hook and Joan Reede will not be attending. Hunt Willard will be joining us tomorrow. So we thank them for that.

One of the most important things, at least to me coming on, and I'm sure to you, was that given that we are advisory to the Secretary of Health, does the Secretary of Health and does his office care about what we are doing. I wanted you to know that I did put in a call to that office and spoke with, as the news would say, an unnamed senior official.

(Laughter.)

DR. TUCKSON: I was encouraged that the unnamed senior official was well aware of what we are doing. I was insistent and assured that within a couple of days of my

conversation the Secretary would be briefed about the fact that the new incoming chairman of the committee wanted to be sure that it was important to him and that he was paying attention to what we are doing.

So I will just simply leave it there and say that I think it is important because you all are working so hard on this committee that you've got to know that it's not just being filed on a shelf somewhere, that this is important that it's getting done, and I'm going to take my responsibility, as Ed hands the baton off to me, to really ensure that that is happening. So I will give you an update on that, on the Secretary's conversation, the next time that we meet.

Let me say that the status of the committee's work product since June, what has been going on, we have the resolution on genetics education and training of health professionals. That copy is available at the desk. It was transmitted to the Secretary in August. The roadmap for the integration of genetics and genomics in society, the study priorities of the Secretary's advisory committee, will be transmitted to the Secretary very shortly, and that will happen really in the next matter of days and weeks.

The letter to the Secretary on direct-to-consumer marketing of genetic tests and technologies is in the final stages of review by our committee and will be transmitted to the Secretary soon. It's in the table folder, and there is still time, if you have any last, last, last minute, short, brief, non-controversial comments.

Sarah is kicking me under the table as I say that.

Finally, a very brief overview of the agenda. Of course, first we will start out with learning about important HHS initiatives to promote family history-taking. We will then dive very deeply into the issue of genetic discrimination and information about the nature, magnitude and scope of the discrimination problem in society. We will move to completing our deliberations and finalizing our recommendations on coverage and reimbursement. Then finally, we'll begin planning future work on two high-priority issues, large population studies and pharmacogenomics.

Public comments are scheduled for today and tomorrow. So far we have about seven or so who have registered to provide comments. Any others who may be interested should sign up at the registration desk. Again, this is a relentlessly public experience. So if you have comments, we welcome you to do that.

With that, let me turn over to the czar, the general, Sarah Carr, who will take care of some very important technical stuff.

MS. CARR: Thank you, Reed.

As you know, the members of this committee are appointed as special government employees in order to serve, and at each meeting I always remind you about the rules of conduct that apply to government employees and to you. These rules are in a document called "Standards of Ethical Conduct for Employees of the Executive Branch," and each one of you got a copy of this document, and I know you've reviewed it very carefully.

I'm just going to highlight two of the rules today. One is about conflicts of interest, and the other is about lobbying.

Conflicts of interest. Before every meeting, you provide us with information about your personal, professional, and financial interests, and this is information that we use to determine whether you have any real, potential, or apparent conflicts of interest that could compromise your ability to be objective in giving advice during the committee

meetings. While we waive conflicts of interest for general matters because we believe your ability to be objective will not be affected by your interests in such matters, we also rely to a great degree on you to be attentive during our meetings to the possibility that an issue will arise that could affect or appear to affect your interests in a specific way.

In addition, we have provided each of you with a list of your financial interests and covered relationships that would pose a conflict for you if they became a focal point of the committee deliberations. If this happens, we ask you to recuse yourself and leave the room.

Lobbying. Government employees are prohibited from lobbying, and thus we may not lobby, not as individuals and not as a committee. If you lobby in your professional capacity or as a private citizen, it is important that you keep that activity separate from your activities associated with this committee. Just keep in mind that we are advisory to the Secretary of Health and Human Services. We don't advise Congress.

Thank you for being so attentive to the rules of conduct. We appreciate your conscientiousness very much.

DR. TUCKSON: Terrific.

Well, to kick us off on our first session on the importance of family history in health, to get that started, I'm going to introduce Francis Collins. We have to be concerned that Francis flew on the redeye all night, so we're in great danger that he will fall off at any moment. So, Ellen and Daniel, if you'll keep an eye on him, we'll all appreciate it.

DR. COLLINS: Thanks for telling just about everybody about that, Reed. That's much appreciated, and I probably will need the support.

Well, it's my pleasure to introduce Alan Guttmacher, who is going to give this presentation on the importance of family history in health. Alan has a distinguished career as a physician in the area of medical genetics and pediatrics. For some time, he was the only board-certified medical geneticist in the State of Vermont, and also ran the first newborn intensive care unit in the State of Vermont.

The State of Vermont went into severe mourning when I recruited Alan to come to NIH because, as you can imagine, their census dropped rather dramatically in terms of these kinds of capabilities. But since he has been here for the last five years, he's become an absolutely essential part of what we're trying to do in the genomic arena, and he runs our Office of Policy Education and Communication, as well as a host of other issues, and he has been the point person in our discussions with the Surgeon General about a particular initiative on family history that I think he is going to tell you something about.

I would also just like to say, because it happens to be the very day when this is being announced, that Alan has been elected to the Institute of Medicine as of today. So congratulations, Alan.

(Applause.)

DR. GUTTMACHER: When Francis said he wanted to introduce me, I knew there was some way he was going to try to embarrass me. Since I recently introduced him, I was quite concerned. I thought it might be even worse than that. But thank you.

This is a very important issue. In fact, the Surgeon General had hoped to be here to be able to give this talk. He's very sorry that because of another commitment he couldn't be here. I'm very sorry that because of another commitment he couldn't be here. I hope you won't be too sorry that because of another commitment he couldn't be here to talk

about a trans-departmental initiative that the Surgeon General is really spearheading, and that has to do with family history.

So first I thought I'd talk a little bit about the importance of family history and health. I realize that this will be largely preaching to the choir, but I'm going to end the sermon by letting everyone know how we can use our voices together to sing and preach to a larger choir than just the folks in this room.

If this is the age of genomics, as we much trumpet it, why should family history be important? It seems sort of old-fashioned, the kind of thing you might, heaven forbid, use a pencil and piece of paper about and not that sophisticated genomic tool, et cetera. Well, of course, we know, in the age of genomics, that most diseases are due to an interaction of multiple genes and environmental factors. So we know that, but what do we do with it?

Well, we should remind ourselves that today, almost every patient that is seen has available a free -- I would underscore "free" since that's unusual medicine -- personalized tool that captures many of those genetic and environmental interactions and can serve as a cornerstone for individualized disease prevention. That tool, of course, which has stood the test of time, the other thing to be said for it, is the family history.

Now, even though we will, of course, continue to gain important new genomics tools in the years ahead, and those are the kinds of things that this committee has been thinking about and their impact on health care, family history is going to remain relevant for many years to come, and in fact will become more useful because it has to do with such varied health concerns as you see here -- heart disease, colorectal cancer, breast cancer, ovarian cancer, osteoporosis, asthma type 2, diabetes, suicide. You can go on and on. Most of the common, significant causes of morbidity and mortality that we have, in fact, family history can contribute to our understanding of them and certainly our approach to individual patients.

Yet nonetheless, most people, many people certainly, are unaware of their relevant medical histories, and many of us health professionals under-utilize this information in advising our patients about how to maintain good health.

For instance, you can use family history information to affect the way that you do population screening for all of these different kinds of conditions, a large array of conditions to which family history is pertinent. Then beyond screening in terms of our management of various conditions, again a large slew of conditions for which we have good data to show that family health history can in fact make a difference in management.

So if this is so useful, why aren't health professionals using it more consistently and more effectively? All of us who have a clinical background would be embarrassed probably to talk about the way we individually use the family history but would certainly be embarrassed to talk about how all health care providers in general, our professions tend to do that.

I think one bar is that clinicians tend to underestimate the actual utility of the family history. The way that we can get past that hump I think is really better teaching and more pervasive role modeling of the effective use of the family history. If those of us who have some particular background in thinking about family history and using it in health care were to demonstrate to others how it can be helpful, I think that the message would spread more effectively.

Of course, another significant factor which becomes more significant with each year's health care changes is the insufficient time, really in the clinical setting often, to

obtain, to organize, and to analyze the family history. It's one thing to say family history can be pertinent, but it takes a lot of time to actually collect the information and to be able to set it down in some way that is available in the future and to be able to really analyze and do something with it.

Now, how do we get past that hurdle? Well, there are some creative approaches out there that people have been working on in recent years that I think are just beginning to come to fruition that will help practitioners who are busy do this using less of their time. But if we do that, in fact, do the patients care about this? Well, there are soon to be published data that come from the CDC -- they're not published yet, so I'm not going to tell you about them, but you can look forward to them soon from Muin and his colleagues -- that show that the American public actually is well aware that family history is important to health. But despite that, relatively few people in the general public have actually ever collected health information from relatives to obtain a family health history, which suggests that there, too, is a bar, an obstacle which needs to be overcome, and that is that even though people are convinced family history can be pertinent and important for their own health, they haven't actually acted on that, which means there are probably some impediments in terms of this being difficult for families and individuals to do.

So what can we do about that was something that a number of agencies within the Department have been trying to deal with over the last few years, and those include the Surgeon General's office, HRSA, AHRQ, very importantly the CDC, and various parts of the NIH as well. So we've come together to form what's called now, or will be called officially in a little while, the Surgeon General's American Family Health Initiative. So let me tell you a little bit about this initiative.

The goals of the initiative are, first of all, to increase the American public's awareness of the importance of family history and health, though we again have some data that they're fairly aware of this already, but not so true at the moment is to give the American public tools to be able to gather, understand, evaluate, and use family history to improve their health; to increase the awareness of health professionals about the importance of family history; to give those health professionals tools to be able to gather, evaluate and use family health information, and tools to communicate with their patients about family history; to use this as a way to increase both the genomics literacy and health literacy in general. If we're going to get the public ready to use some of these new genomic tools as they become more available, genetic testing, et cetera, it would be a good idea to have folks become more familiar with some of those concepts by using the old tried and true family history. And preparing both the American public and their health professionals for this coming era of genomics in which we believe that will be a regular part of health care.

So what can we do? The first little product of this initiative to point to is something called My Family Health Portrait. You see this is the banner for it. This is a dummy of somewhat what it will look like. If you look closely you'll see that it says "My Family Health Portrait" at the top. In the middle it says "Welcome to Your Family Health Portrait." This is such a dummy that it hasn't been updated to reflect the official name, My Family Health Portrait.

This is going to be a web-based tool that will be unveiled very shortly -- I'll tell you a bit more about that in a moment -- where individuals and families will be able to download directly to their computer so that this information lives only on their computer, not

on some government site, which would be illegal amongst other things, and allow people -- not just allow people, but give people, we believe, an easy, interactive kind of way to gather their family health information. Then once they've gathered it, in fact, give them some guidance about what they might do with that information.

So November the 8th, Monday in a few weeks, there will be a formal announcement of the initiative, and there will be the release of the My Family Health Portrait website. There will be a big even downtown. Several agencies within the Department will be cooperating in that. We're going to have some other media around that date, so we're hoping that you will hear a fair bit about it in your local media at that time, as well as national media.

This is the official logo of the thing. There will be this press conference at the National Press Club on November 8th to announce this.

Then on Thanksgiving Day, this is really the focus and this is part of what will be talked about at this press conference. The idea is to make Thanksgiving Day that day when American families by and large traditionally gather together to eat a lot, to watch the Packers on TV and do other kinds of things, to use that family event to actually talk about family history and to gather family history information, the idea that people would have this web-based tool. They could use it that day, they could gather some of the information before, they could gather it afterwards. But the time when the family is really together, when you have Aunt Gladys around who can actually tell you about what you thought you had heard about Uncle Joe or something like that, to get more accurate information.

That is, of course, the other problem with doing this in the office, is that often you have the person who comes in has pretty imprecise information. All the clinicians in the room are nodding their heads. That's the experience of all of us. In fact, when you have the family gathered around is the best time to get more accurate information. To be able to build that over the course of time, we're hoping to make this into an American annual event, that Thanksgiving would be that day. We've all seen that picture, the traditional Norman Rockwell. "Freedom From Want" is what it used to be called. The way I think of it is now that we've finished the family history, let's eat.

We hope to encourage the American public to think that Thanksgiving is the day, before you sit down to raise your cholesterol levels, that you actually talk about family history and gather this information.

The initiative will continue past this year, past this Thanksgiving. We're very eager to have other interested parties, be they federal agencies, and particularly non-federal agencies, other organizations, et cetera, that we've had some communications with previously to the degree that we're legally allowed to do that, we're very much hoping that this will be something that not just the genetics community, certainly the genetics community but other communities will participate in it and seize an important idea to bring this tool that we really know can make a difference in health care and can be fairly easily accomplished, that we be able to do that in a much better way.

So with that, I'm going to stop. Mr. Chairman, I don't know if there's time for any questions or comments. If folks have them, I'd be happy to take any.

DR. TUCKSON: Oh, we actually do have just a second. This is an important presentation.

Let me just turn to Ed. I think he has a comment first.

DR. McCABE: Well, I just wanted to comment that I teach the medical

students. I had three hours with them last week and I told them that this was the cheapest but perhaps the best genetic test that we had today.

The other thing is I was shaking my head because I recalled that as a geneticist, as a board-certified medical geneticist, I was always asking my patients for their family history, and I realized that I was asking them for more than I could provide myself, and sat down with my mother and went over the family history and learned quite a bit about myself that I had not known. Soon thereafter, she developed Alzheimer's disease, so it was very important that we were able to record that information and store it away in the family Bible so that it can be then utilized by generations to come.

So this is an extremely important tool. I think too few of us take the time to really get that information, and I would encourage everyone here today to be sure that you can do for your families what we as geneticists ask our patients to do for us.

DR. TUCKSON: Any other quick questions or comments?

(No response.)

DR. TUCKSON: Alan, thank you so much.

Let me just ask two follow-ups, then, because I think you've got two things that I think we need to sort of get some more information about. First is that whatever is going to go out on this in terms of genetics and literacy, which is part of this, the whole idea of why is this important in a genomic age to the public and how do you use this information, we sure would like to, I think, see that and benefit from whatever is being sent out, because that may help us through some of our efforts to improve the public literacy around the issues of genetics. So I think it would be a great FYI for us.

Secondly, if there's any part of this tool that's going to be used as part of any of the agency's efforts around information technology, electronic medical records, I think it would be useful for us to also have that as an FYI follow-up, because we're going to obviously wind up having to deal with those issues downstream. So if this is going to be integrated in any way into any of the new health information technology efforts that are coming out of the government, it will be interesting to see.

DR. GUTTMACHER: Well, certainly, we welcome the opportunity to keep the committee aware of this and also to ask input from the committee and through the committee, the various folks that the folks the committee is connected to, because again, this is the first stage of what we hope will be a long-term kind of initiative. So we'd be very happy -- for instance, we are developing not just a computer base but also pamphlets and those kinds of things that we would love organizations interested in distributing those, which is often the biggest challenge, of course, with the printed word, how to distribute them.

We're developing many of these things. For instance, the computer base too, I should tell you, will be available not just in English but also in Spanish before Thanksgiving, so that we'd love the opportunity to interact with the committee as this goes forward. Thank you.

DR. TUCKSON: Well, to bring this to closure, let me offer three quick possibilities that we might be able to do. Unless there's a major controversy, we might try to get an endorsement.

First, that we endorse the importance of family history as a tool in medicine apropos Ed's comments.

Number two, that we report to the Secretary that we are encouraged to see

HHS agencies working together to bring this to fruition.

Third, that we would encourage those agencies that are not part of this to get involved.

So we would at least be supportive of this with those three recommendations.

Is there any discussion about those three?

Emily?

DR. WINN-DEEN: I just wanted to ask a question. Are you going to provide some kind of a mailout so that you could take a pamphlet or something and mail it to your elderly relatives and ask them at their leisure, for those of them that aren't computer literate and aren't able to deal with that, to send information back?

DR. GUTTMACHER: There will be a pamphlet which will explain sort of the role of family history, why it's important for specific disorders, and has a template in which you can record information that we made available.

DR. TUCKSON: So with that, by a show of hands, would you support endorsing the importance of family history as a tool in medicine, that we report to the Secretary that we're encouraged to see HHS agencies working together to get this done, and third, that we would encourage other agencies to get involved and support this initiative?

All those in favor of that transmission, say aye, or raise you hand, same thing.

(Chorus of ayes.)

DR. TUCKSON: Anybody opposed?

(No response.)

DR. TUCKSON: Terrific.

For a follow-up, Alan, I hope we do get those two follow-ups so that we can see what actually does go out.

DR. GUTTMACHER: Very good.

DR. TUCKSON: Thank you.

DR. GUTTMACHER: Thank you, appreciate it.

DR. TUCKSON: All right. For the next part of our effort, we're going to devote from now until 12:45 on a pretty serious discussion on genetic discrimination. You will remember that this is a top priority for our committee. We've written two letters to the Secretary urging support for the enactment of federal nondiscrimination legislation. At our March and June meetings we discussed the question of whether there were additional efforts that we could take to inform the debate regarding federal legislation in this area. The United States Senate has subsequently unanimously passed a genetic nondiscrimination bill a year ago, but the House Subcommittee on Employer/Employee Relations has held a hearing on this in July without further action that we are aware of. There is a summary of those hearings in your Tab number 4.

The purpose of this session is to gather additional public perspectives on genetic discrimination and information about the nature, magnitude, and scope of this problem in society. The session was planned and organized by our Task Force on Genetic Discrimination. Agnes Masny, who has been terrific as chair, and Barbara Harrison and Debra Leonard and Emily Winn-Deen and Robinsue Frohboese and Tim Leshan, former EEOC Commissioner Paul Miller, Joann Boughman of the Genetic Fairness Coalition, all were

extraordinarily involved in this, and we thank them for it. Amanda, I want to thank you for your lead staff work on this. I also want to thank the members of the panel for participating and taking note of the written comments that have been submitted. They have been compiled in a separate binder, which I hope you all have. A copy is available for review at the registration desks.

Now to lead us through this, let me introduce Agnes. Thank you, Agnes, for your hard work.

MS. MASNY: Thank you very much.

I'd also like to thank Dr. Tuckson, who is going to help co-facilitate this session, and again thank Dr. McCabe for his leadership, bringing us thus far, and to the staff, Sarah and her staff, for helping coordinate, and especially Amanda Sarata, who has been working by my side in helping to pull all this together; and most especially to thank all of the speakers who have come from far and wide today to be able to give testimony today so that we will be able to document for the record the experience of the public, health professionals, and others regarding this very important issue of health discrimination, genetic discrimination.

As Dr. Reed mentioned, what led up to forming this particular task force and the members that he already indicated have been presented, and then as was mentioned is that one of the reasons why we have actually established having a public hearing today is that because from the past we've recognized that the evidence about genetic discrimination, and particularly the fear of genetic discrimination, has been very hard to document. There are very many reasons that have been brought forth to the committee regarding the public's concern about genetic privacy, and presenting their information in legal cases is very hard to do. So that's one of the reasons why we wanted to have this hearing.

Also, because to date there has been lack of sufficient evidence, and this lack of evidence has been actually brought up as one of the reasons as a barrier to going ahead with legislation for anti-discrimination. So at the 2004 June meeting, we did decide to undertake this public hearing in order to provide the Secretary with additional information that might be useful in addressing the potential obstacles regarding genetic discrimination and moving it forward for legislation.

So the purpose of this session, as Reed has already mentioned, is to learn more about the magnitude and scope and the nature of genetic discrimination throughout society, and to provide all of us with this public forum to be able to both hear the testimony and then to be able to discuss the issues that will come forth from this hearing.

Then the last step will be for us as a committee to then look at what are the next steps that we have to take to hopefully move the legislation forward.

So what we're going to be doing today is we're actually going to hear from three panels: members of the public, health care providers, and then additional stakeholders. The patient panel will consist of members of the public who have experienced genetic discrimination or who have altered their health care because they have a concern about genetic discrimination either from insurance or from employment, or those who have had to pay out of pocket because of their concern for genetic discrimination.

We also took time to look at, as a task force, getting members of the health care community providers who have that first-hand experience with the patients, hearing their concerns about genetic discrimination, to also be able to come and testify. So again, we hope to hear from the providers about some of their patients who may have altered their health care

decisions or who have not participated in genetic research because of genetic discrimination and other concerns that they have regarding discrimination.

Then the third panel will be additional stakeholders that will hopefully give us an additional perspective, and this from employers, from the health insurance perspective, and from the society at large, and this primarily through a representative from the Center for Genetics and Public Policy who, over the past several months, have held town meetings across the country on the issues of genetics, and genetic discrimination was one of the issues. So we'll be hearing that particular perspective as well.

The committee and the SACGHS had requested written public comments through the Federal Register notice, through the SACGHS website, through the major distribution lists of SACGHS, and then we also targeted specific medical groups like the American College of Obstetrics and Gynecology to be able to go directly to their membership to ask about this particular issue, whether they as providers have come in contact with genetic discrimination or the fear of discrimination from their patients, and then also various listservs from health professionals, physicians, nurses, and genetic counselors to request public comments.

So we did receive 43 public comments. Twenty-two were from the public, 11 were from health care providers, 9 from professional organizations, and one very nice, thoughtful testimony from Representative Louise Slaughter, who has co-sponsored the House version of the bill 1910.

So we're going to begin, then, with the most important part, to actually hear from the public, and we're going to start first with the patient panel. We're really very fortunate to have with us today a panel of seven members of the public who would like to share their personal stories and experiences and their own perspective on genetic discrimination. They come to us from across the country.

What I'll do is I'll introduce the whole panel as a whole, and you can just kind of go in order across from the table. We will ask the speakers that they'll have 10 minutes to present their public testimony. We'll try to keep track of the time for you. Then after everyone has had the opportunity to present, then we will open it up for questions from the advisory group, and the ex officios will have an opportunity to ask any additional questions after the end of all of the testimony, okay?

So again, we'd like to thank and welcome our first panel. We have Heidi Williams from Cecilia, Kentucky. Phaedra Malatek is here with her family, her two sons, from Chicago, Illinois. Rebecca Fisher from Oakton, Virginia. Tonia Phillips from Roanoke, Virginia. Paula Funk from Little Rock, Arkansas. Maria Carolina Hinestrosa from Washington, D.C. Phil Hardt from Phoenix, Arizona.

We're going to turn it over to you. Phaedra, would you be willing to start us off? Oh, no. Heidi, do you want to start? And we'll go down the line.

MS. WILLIAMS: Thank you.

Mr. Chairman, members of the committee, my name is Heidi Williams, and my children, Jayme, 8, and Jesse, 10, were recently victims of genetic discrimination. In August of 2003, I saw a commercial on television advertising affordable health care insurance for individuals through Humana, Inc. I called the toll-free number and talked with a young woman who quoted me a price for a policy that would cover both of my children. I was told that the monthly cost to insure my children would be approximately $105, and I immediately

told the young woman I would like to complete an application so that the coverage would begin as soon as possible.

I was asked a series of questions about my children, including whether or not they had a preexisting condition. I relayed to the young woman, under a threat of a fine and incarceration for falsifying information, the fact that my children were carriers of the genetic disorder called alpha-1 antitrypsin deficiency, or AAT, a liver deficiency that can progressively affect the lungs, the liver, or both, but that my children, unlike their mother, who is lung symptomatic, would never suffer from any aspect of the disorder.

The young woman, who wasn't quite sure what to do with this information, asked me to hold on the line while she contacted her supervisor. As I spoke with her supervisor, I again explained how my children were only carriers of the AAT gene and that my children themselves would never suffer from any aspect of the disorder as I am suffering, and that they are exceptionally healthy and active children. Again, I was told to hold the line because, as this gentleman was uncomfortable with the information I had imparted, he needed to contact his supervisor.

As I spoke to the senior supervisor, I once again relayed the information about alpha-1 and how my children were only carriers. To be born what is considered symptomatic, you must have two parents who are at least carriers of the AAT gene and be of a certain phenotype. I am what is considered the symptomatic phenotype of ZZ, and my husband is considered to be of a normal phenotype. Therefore, my children can only be carriers and, as research supports, will never be susceptible to the various problems symptomatic AATs face, including lung and liver failure.

Once the senior supervisor and I finished speaking, I was given back to the young woman who initially interviewed me and, after finalizing the application, was told by her that I would receive a reply to my children's application for health insurance within 24 hours. After five days of waiting, I knew instinctively that there had been a problem with my children's application. I received a letter two days later, exactly one week after the phone application, stating that my children were being rejected for their health care insurance through Humana, Inc., due to their AAT status and for no other reason.

After much self-recrimination, I shared my woes with the Alpha-1 Lungs and Life Chat Group, a large alpha-q Internet community that is extremely concerned about genetic discrimination, relating my frustrations and my fears for my children having been twice rejected for health insurance coverage. Nancye Buelow, who suffers from alpha-1 herself and was a representative of the Genetic Alliance at the time, heard about my problems with Humana, Inc. and approached me about publicly coming forward with my story through the auspices of the Genetic Alliance. I agreed, and together with the Genetic Alliance and the help of a prestigious Washington, D.C. law firm and a wonderful and very knowledgeable AAT doctor, an appeal to the August 2003 letter, a letter which rejected my children for health insurance coverage on the basis of their genetic status, was drafted and sent to Humana, Inc.

Enclosed within the letter to Humana, Inc. was research information from both the National Institutes of Health and the Alpha-1 Foundation supporting my argument that both Jesse and Jayme, as carriers, would not become symptomatic of alpha-1 antitrypsin deficiency and that both would remain free of AAT's debilitating destructiveness throughout their lifetime. In February of this year, I received my response to the written appeal and was once again shocked to read that my children were being rejected for health insurance coverage

only on the basis of their AAT carrier status and nothing more.

It was only after Humana, Inc. had been approached by a reporter for a well-known and well respected newspaper that they reversed their decision and offered my children full coverage under their company prorated from August 2003 and paid in full until April 2004 by Humana, Inc. themselves. Fortunately for me, my children are now covered by a company that understands that everyone is entitled to affordable health care coverage, and not Humana, Inc.

Needless to say, Humana, Inc.'s reversal of their decision felt like a hollow victory. No one should have to force an insurance company to cover perfectly healthy children. In fact, I don't believe it should have mattered what their genetic status was to begin with. We are all viable members of a community with contributions to make and shouldn't have to be afraid that our genetic anomalies, in whatever form they arise, will be held against us. I should not have had to spend the better part of six months wondering if the decision to have my children's genetic status verified by their pediatrician was a huge mistake. I should not have to wonder if my children's genetic status is going to follow them into the workforce and render them unable to become employed in their chosen fields. And I certainly should not have to feel guilty for unknowingly passing this genetic anomaly on to my children.

Humana, Inc. made me feel guilty and ashamed for needing to know my children's genetic status. Furthermore, they made me feel guilty for needing a parent's peace of mind in regard to my children's future health, and for that I am angry. Today, there is a current of fear reverberating throughout the genetic community. It is not just a fear of loss, but it is a fear of retribution. It is a fear that forces many within this particular community to accept what should be unacceptable, discrimination by genetic status. Many people are afraid to come forward and fight for their rights to employment and health insurance coverage because they are afraid of the retribution that may not only be taken against them but could be taken against their families as well.

Therefore, it is because of the callous treatment of my children and the countless others before them that I want to make sure that this sort of policy practice never happens to anyone ever again. I want to make sure that I will never again exchange emails with someone who has been phased out of a position due to her genetic status. I never again want to hear the story of someone who has been denied health care coverage, had their health insurance coverage canceled, been passed over for promotion, demoted, fired, or simply not hired due to their genetic status.

National legislation that would make it illegal for insurance companies and employers to use someone's genetic status against them has indeed been drafted. The Senate passed S. 1053, the Genetic Information Nondiscrimination Act, unanimously last October, yet one year later this very important piece of legislation that would protect many Americans is still stranded in the House of Representatives. As each day passes and the genetic community waits for the House to bring this bill to a vote, scores of people across this nation are being persecuted on the basis of their genetic status. It is completely reprehensible that any type of discrimination still exists and has to be legislated against in this day and age. But since discrimination still exists, it must be swiftly eradicated in any form that it is found before its destructive force has had the chance to harm anyone else.

Finally, my family and I were extremely lucky. We had the backing of several people and organizations to help us fight our battle in the war against genetic

discrimination that very few people in the genetic community win. Only through legislation and education will genetic discrimination loosen its hold on a community of people who are suffering from its devastating effects.

Thank you.

MS. MASNY: Thank you, Ms. Williams, for your very powerful testimony.

Now we'll hear from Phaedra Malatek.

MS. MALATEK: Good morning, Chairman Tuckson and members of the committee. Thank you for giving me the opportunity to speak with you today. My name is Phaedra Malatek. I am a wife, mother, sister, daughter, and friend of people who care very deeply about what we're doing here today.

I live in Aurora, Illinois with my husband and two sons. My primary occupation is adjunct faculty at two local community colleges. Otherwise, I'm involved nationally and locally on issues related to women's cancer. But I'm here today to talk to you about the hope that lies in genetic testing, more specifically the Genetic Information Nondiscrimination Act, S. 1053. I talk about hope because, as Ralph Waldo Emerson says, "We judge of a man's wisdom by his hope," and today I'm feeling pretty wise.

For me, genetic testing and the protection offered by S. 1053 can be compared in an analogy to weather tracking or storm prediction. Imagine, if you will, that we had no knowledge of the storms that recently swept through the southeastern United States and the Caribbean. How would the death toll change? How would the damage assessment change? How would the insurance industry have changed? Now imagine never having any information about any storm ever. Well, I think our understanding and consideration of genetic testing can be compared to those "what ifs."

What if people were given the knowledge of the potential storms in their lives? How would they be able to protect themselves? What would serve as the plywood for the windows, and what evacuation routes would be made available to them? More importantly, how many lives would be saved? Because that really is the question, isn't it? How many lives can we save by what we're doing here and through the enactment of S. 1053?

Continuing the storm analogy and the concept of discrimination, let's consider a situation where you know that a storm is coming, or that may come, and you take the precautions such as boarding up your windows and putting the lawn furniture in the pool, but you're unable to get insurance simply because you know a storm may come? Your neighbor, on the other hand, has no knowledge of the storm and doesn't take the necessary precautions. If the storm doesn't hit, that's all fine and good. Everybody wins. However, if the storm does hit, the insurance company provides full financial reimbursement for your neighbor's severely damaged or lost property, but there's no coverage for your property no matter how small or large the loss.

It seems to me that that's what we're talking about with genetic discrimination, a situation in which no one wins and everyone loses. Those with advance warning are actually harmed rather than helped by the knowledge they possess. Insurance companies and those without advance knowledge even larger losses because they have been given the knowledge to protect themselves. If fair warning is given to all parties through genetic testing, the people who are able to protect themselves and the insurance companies who agree to be at risk for any loss suffered all have a much larger measure of protection.

Those who are informed about their risk can be proactive and take either

prophylactic measures or be monitored more closely, increasing their ability to entirely avoid developing a disease or having it detected in its earliest, most treatable and survivable stages. As we all know, this not only saves lives but dramatically decreases the cost to employers, insurers, and the individual.

A storm such as this hit my life a number of years ago. In 1991, my father gathered together his children, siblings, nieces, nephews, cousins, to discuss a disease that he had been diagnosed with. The disease is hemochromatosis, which is often referred to as iron overload. My father had been exhibiting symptoms prior to diagnosis, including arthritis, heart arrythmia, and a change in skin tone. At the time he was diagnosed, his heart and liver were fully involved. At the same time, his physicians conjectured that my grandfather and great-grandfather may have also carried and suffered the effects of this disease.

Within a year of our meeting, my father suffered a heart attack and within 10 years had died from the disease. Since my father's diagnosis, two of my siblings have developed complications of the disease. None of them, my father or my siblings, have had genetic tests for hemochromatosis. Because of my family's history with hemochromatosis and the fact that it is the most common inherited disease in the United States, my husband and I are concerned for the welfare of our two children.

Mitchel and Trevor, who are here with me today, throughout their lives we received conflicting information on how to approach their risk for hemochromatosis. We were told to have periodic blood tests to measure their iron level. We were told to do nothing. We were told to constantly monitor their diet. While all of this may have been good advice, none of it replaces knowing for certain that Mitchel or Trevor carried the genetic mutation for the disease that contributed to my father's death and is an issue in the life of my siblings. With that knowledge, we could have taken proven knowledge to lessen the impact the predisposition for this disease might have on their lives.

Like storm predicting and tracking capabilities, genetic testing seems to offer an opportunity to learn more about the constitution of diseases and their potential serious damage. It can help us track the progression of a disease, as well as determine treatment or even protective measures to avoid the storm that may result from a genetic mutation. For my children, this could be life-altering information, altering in that it will decrease the likelihood that they will be incapacitated by hemochromatosis.

For diseases such as ovarian cancer, it can mean the difference between prophylactic treatment that could allow women at high risk to lead long, successful lives, and the stark contrast of the often futile and very painful death-prolonging treatment.

Throughout the recorded history of hurricanes, experience has gone from storms that came out of nowhere as recently as 100 years ago to those that we were able to track minute by minute 100 days ago. The dramatic change is not a result of the decrease in the power of the storm but rather an increase in technology and our understanding of hurricanes. Along the same lines, technology and advances in the area of genetic testing can similarly provide predictability and a greater level of protection for those at risk, and that risk, or even the knowledge of the potential risk, can be protected through S. 1053. While the technology for physical protection through genetic testing seems to be in place, or at least advancing at a relatively rapid clip, the social and economic protections are not.

As it stands right now, if my children undergo genetic testing for hemochromatosis, they risk not being able to obtain health insurance when they're no longer

covered under my husband's policy and possible discrimination when they seek employment. So we're given a choice: protect their health or protect their livelihood. It's troubling to me that as Americans we're placed in a position where we have to make such a terrible choice. It also troubles me that S. 1053 offers protection that would eliminate the need for my husband and I to decide such a difficult thing, and it's not been brought to a vote in the House of Representatives.

All of this is reminiscent of a series of choices that were being made 40 years ago. In the late '50s and early '60s, my parents fought diligently for the rights of people who were genetically different from them. They were not different in that they were at higher risk for obtaining hemochromatosis or ovarian cancer but that their skin was a different shade of beautiful. My parents, along with many others, won that fight. The Civil Rights Act amendments are there to protect people from discrimination based on genetic makeup that we can see, be it skin tone, gender, or disability. A person's genetic makeup that isn't visible should be equally protected under the same terms and can be through S. 1053.

It's remarkable for me to realize that the work my parents did for the Civil Rights Act in the '60s was not complete. Here I am, 40 years later, working on the same issue, equal rights and protection under the law no matter the genetic makeup of a person. The fact that we can look inside the DNA of a person to know more about them should not preclude them from the protection that was fought for so valiantly. As I see it, genetic testing is the weather tracking device of health. Just as we rely on weather tracking technologies to predict and to allow us to protect ourselves from hurricanes or other weather-related storms, I urge you to allow us to do the same for genetic diseases.

We must move forward in protecting people from the potential storms in their lives. You can do this by urging Secretary Thompson and my representative, Speaker Hastert, to bring this bill to a vote in the House of Representatives. I'm sure you can agree with me when I say that protecting lives is equally or more important than protecting property. If we can, we should, and S. 1053 will.

MS. MASNY: Thank you, Ms. Malatek. That was such a beautiful analogy between the genetics and storms.

We'll hear next from Rebecca Fisher.

MS. FISHER: I don't think I have anything to add to that. It seems almost like preaching to the choir, but I would like to thank the committee for everything the committee members have done in the past to address genetic discrimination concerns, and I hope that our remarks will help to inform your actions going forward.

In a provocative October editorial in the journal Science last year, Nobel laureate Sidney Brenner wonders what medicine will be like in the year 2053. "Perhaps," he wrote, "the prime value of our work to society will be the creation of a new public health paradigm in which those who have a genetic predisposition to disease will learn how to take extra care." Dr. Brenner needn't wait 50 more years to see this prediction realized. Some of us, those who possess BRCA1 or 2 mutations, known to predispose us to breast and ovarian cancer, are already taking extra care.

A recent study established that 67 percent of women with this mutation are diagnosed with breast cancer by the time they're 50 years old. But I have a cousin who died of it when she was 28. I have another who is battling Stage 4 ovarian cancer as we sit here today. She has a 4-year-old. My mother had breast cancer at 35. Her mother died of ovarian cancer

at 41. Her sister had breast cancer at 32. I was 31 when I was diagnosed with Stage 3 breast cancer. My daughter, a 21-year-old, is in this line, too. She tested positive for BRCA1. She will also have to learn how to take extra care.

But the care that Katie will have to learn how to take includes not only the low-fat diet she's already eating and the daily exercise regimen she's undertaken. It includes more than the breast self-exam she's required to perform monthly, and believe me, I do remind her. It even goes beyond the MRIs of her breasts she will start receiving when she turns 25. The extra care she will have to learn how to take demands that she, like me and like everyone in our family who has this mutation, hide -- that is, hide, H-I-D-E -- her genetic information even, and perhaps especially, from those health care providers most likely to help her manage her lifelong predisposition to disease.

Unfortunately, that's what we're reduced to. Hiding integral health information is the only fail safe way we can avoid discriminatory practices such as the loss or denial of health insurance or the loss or denial of employment, because there simply is no comprehensive federal legislation that patently forbids insurance or employment discrimination on the basis of genetic information.

The argument has been advanced most recently and very publicly in the Wall Street Journal last March that seeking to ban DNA discrimination isn't really necessary because discrimination simply doesn't exist. Actually, it does exist, but the fact that it exists only sporadically and anecdotally is a function of the newness of the technology and the fact that useful predictive genetic information like ours is not yet ubiquitous. It is not a function of insurance companies' and employers' decisions to take the moral high road and, out of the kindness of their hearts, remain disinterested in this information in the same way that they are legally obliged to remain disinterested in information such as race, gender, creed, or sexual preference.

I know from experience that insurance companies don't work this way. When I was sick, I worked as a medical librarian for a small community hospital in south Florida. The hospital was self-insured. Pay attention to this part. The hospital was self-insured, and a third-party administrator managed our insurance plan. About a year after my last treatment, I was sitting at my desk when the phone rang. The flustered young woman at the other end of the line told me I was the fourth person she had been transferred to, and someone along the line had suggested to her that perhaps I could give her the information she needed.

Perhaps I can, I offered. Well, she began, I am calling about Rebecca Fisher. Her bone marrow transplant and other health care costs exceeded the calendar year cap last year, and I'm calling to find out if that's going to happen again this year. I'm Rebecca Fisher, I said, and I really hope not.

This experience taught me something. It taught me that there are people who are paid to look at me and see not my ability to contribute to a community, not my honesty, my integrity and my faith, not my education, hard work, and social conscience, not my family members and the ways in which I have helped each of them succeed, but dollar signs, costs, increased liability, and the odds of my dying an expensive death.

Let us face the fact that financial incentives to use genetic information are already present. The Washington Post reported just last month that employer-sponsored health insurance premiums rose 11.2 percent this year and are expected to rise 13 percent next year. With these increases in mind and no enjoinder against using genetic information to predict

future losses, it is a failure of stewardship, and I feel terrible that this committee has done this repeatedly, sent letters to Secretary Thompson repeatedly without any action. It is a failure of stewardship to expect companies and employers to simply do the right thing, and when they don't lavish precious man hours, health care hours, and litigation costs to undo the damage.

I fear for my children, especially for my daughter, who must live not only with an exponentially higher risk of developing a terminal disease but also with the burden of never knowing whether or when she will legally be asked to take a genetic test as a condition of employment, be lawfully fired from a job because she's very likely to get breast cancer, or be legitimately denied health insurance or life insurance on the basis of her genetic predisposition to disease.

We live in a world that has no safety net for us, not even HIPAA. Many people simply do not understand that HIPAA is no panacea for all that ails health privacy. The HIPAA gap means that HIPAA addresses none of our workplace concerns, and ERISA rules exempt, exempt, employer-based health plans like the one at the small hospital where I worked from mandatory HIPAA compliance. If my BRCA1 positivity had been known in 1994 and the HIPAA protections of today were in place then, the young woman on the other end of the phone could well and legally have recommended to her superiors, and probably gotten a bonus for doing it, that I not be extended further health insurance coverage.

The HIPAA gap is deep and wide. Of the 137 million private sector American employees who have health insurance, a whopping 45 percent -- this is from Steve Donohue at the Department of Labor -- a whopping 45 percent, that is 63 million Americans, fall into it. The genetic information of each one of these individuals, together with the information of every uninsured American -- that's another 45 million people -- is fair game.

In my opinion, genetic information is no different from any other essential distinguishing information about any human being, all of which is by law kept off the bargaining table that bears up this human rights-based society. But if this argument is truly different -- okay, I'll give you this. If this argument is truly different, if because of its fiscal component, as the United States Chamber of Commerce might argue, we must locate this debate within the framework of an implicit utilitarianism, I would point to professional contributions I and other genetically vulnerable people have been able to make because we've been lucky enough to remain considered employable.

I would point to the contributions my daughter, 21 years old, hopes to make with her two degrees in public policy and economics from Duke University. I would point to the way in which our family's completion of innumerable psychological questionnaires, the donation of tissue from our bodies, and the giving of our blood have advanced medical science. I would argue that we are, in fact, making a difference for the health of all people, everyone in this room, that we've lived up to our end of the social contract and deserve the same fundamental legal protections that are extended to all Americans.

Last summer, attorney Lawrence Lorber, representing the U.S. Chamber of Commerce, the loudest voice speaking against federal genetic information protections, told the House Education and Workforce Committee that the possibility of employers being accused of engaging in genetic discrimination would be disastrous for them from both a legal and public relations perspective. He offered this as proof that genetic discrimination legislation is unnecessary.

I would like Mr. Lorber to tell that to my friend Susan, a 38-year-old

woman whose sister is being treated for breast cancer, whose mother had pre-menopausal breast cancer, and aunt who died of it. We sat together at one of our son's ice hockey games last winter and she shared her story. Without wanting to push, I gently asked her whether she had considered speaking with a genetic counselor. Oh no, she exclaimed, I would never want to risk losing my insurance. This woman is a master's prepared therapist who watches CNN and reads the paper.

Fear and innuendo surround the brave new world of genetic information. People are afraid. Their fear keeps them from being tested, even when this test might make the difference between whether they live or die. And at the risk of sounding paranoid, I would go on to suggest that none of us present today can afford the luxury of writing off this problem to high-risk individuals like me. The stage is already set for a problem of catastrophic proportions. Guthrie spot programs whereby every newborn infant's blood is collected, screened and stored are found in all the states and territories of the U.S. and provide what is potentially the largest and most complete genetic bank and library available in the country.

The continued non-use of genetic information implied by insurance companies' and employers' lack of interest to date does not provide safeguards for any of us, high-risk or not.

MS. MASNY: We'd ask you in one minute to wrap up, please.

MS. FISHER: I'm almost done.

We with strong family histories of disease in which the baton of illness has been passed from generation to generation are simply the first line of defense against a staggering spectrum of possible abuses. We want to be heard, we want to be protected, and we don't want to sit in the back of the bus anymore.

Thank you.

MS. MASNY: Thank you very much. I disagree with your opening statement that you didn't have anything more to add, because you definitely had a lot more to add. Thank you for your very powerful testimony.

Now we'll hear from Tonia Phillips.

MS. PHILLIPS: Good morning, ladies and gentlemen of the committee. My name is Tonia Phillips, and I'm here to tell my story. It is short and sweet.

I work for a small company of about four people, including my two bosses, the owners. We are a tight-knit family. They have been with me through my mother dying of ovarian cancer in April of 2002 and my own genetic testing for BRCA mutations. I was very open with my experience just because we are a small company and there was no way to hide it.

After finding out I was positive for the BRCA1 mutation in March of 2003, which means I have an 80 percent lifetime chance of getting ovarian cancer and a 45 percent lifetime chance of getting breast cancer, I began preventive steps. I had a hysterectomy in October of 2003 and a prophylactic mastectomy in March of this year, and I'm still in the middle of reconstruction.

About four months ago, our group health insurance bill came in the mail, and it had gone up $13,000 a year for four people. My boss got the bill and yelled it through the office. I knew that she was directing that towards me. I was immediately asked to switch to my husband's health insurance policy because my situation was the reason the insurance premium went up so much, and they said that if I was taken off the policy, it would not go up. I was even told they would raise my hourly rate if I switched.

I told her I was not comfortable with switching insurance companies at the time because I was still in the reconstruction process. It was like pulling teeth to get the insurance company to pay for these procedures, and switching would confuse and complicate everything. I didn't think it was in my best interest to switch while I still needed more surgery. My feeling is that anyone in the company could be diagnosed with anything tomorrow and that it's not fair that I be asked to drop insurance that is important to me. I was doing something that would prevent me from going through a horrible disease that would cost much more than these preventive surgeries I was having.

We finally came to an agreement that employees would have to start paying half of their premium, which was fine and fair with me, but I'm sure the other employees weren't too happy with me. It seems unfair to me that I am taking steps to keep myself healthy and to prevent cancer in the future, and I am being singled out and made to feel I am a liability. I also don't smoke, I work out, I eat right most of the time. If someone in the company were diagnosed with cancer or some other disease, they would not have been asked to switch insurance companies as I was asked. I hope that me coming here and telling my story will help with defining the problem and passing laws against genetic discrimination of any kind.

Thank you.

MS. MASNY: Thank you, Ms. Phillips. You definitely do help to define the problem.

Now we'll hear from Paula Funk.

MS. FUNK: Good morning. Thank you so much for inviting me to come and talk about my story. When you're going through the process of finding out your genetic predisposition, sometimes you feel alone. Last night I was thinking this isn't anything I should be nervous about because you're here to help me, and I really appreciate that invitation.

My name is Paula Funk. I'm a mother of twin 3-year-old daughters, and I have a husband that's here with me to support me today. My family has a strong history of breast and ovarian cancer. I'm going to tell you a little bit about my family.

My dad is one of ten children, and he has five sisters. All five of his sisters have had breast cancer, and the current count right now is that eight of my cousins have had breast cancer as well. The number breakdown there, that is 13 women out of 24 that have had breast cancer. This disease is something that the women of my family have to constantly think about. There's a constant threat.

My first memory in life is taking my aunt Dorothy to her weekly treatments to fight breast cancer. It's a sketchy memory, but I remember clearly a sweet lady lying down in the back of my mom's station wagon in a pink nightgown. We took her to treatment every week, and that was the beginning of my understanding of what my future had for me.

My sweet aunt Dorothy has survived breast cancer twice and is now fighting an aggressive form of ovarian. This month she's going to have to have a surgery to repair tissue that all of the treatments for cancer have torn a hole in her chest area, and they're going to have to do treatment just to patch that area. This makes me really sad. It makes me feel like I have to aggressively fight my possibility of cancer.

Ten years ago I started realizing this, and I decided to pursue genetic testing. At that time, genetic testing required several women from one family to give a blood sample to determine if there was in fact a genetic mutation. I approached my aunts and my cousins about this, and they talked to their physicians, and their physicians recommended that

they not pursue genetic testing because at the time people could deny insurance and the discrimination could be even worse in the future as more was learned about being genetically positive.

Because of this, I chose not to have the test at the time. I was 23, and 30 seemed like a long way away. Well, I'm 33 now, and I'm in the middle of where most of the women in my family begin to get breast cancer. Two of the women were 30 when they were diagnosed. Because I am in the middle of that stage where most of the women get breast cancer, in May of 2003 I decided to consider being genetically tested again. I talked with a genetic counselor, and she asked me a lot of questions about insurance. It was an unfortunate time for me to want to pursue it. My husband and I had just opened our own small business. We were the two people in the business.

As I researched about individual insurance, I learned that even with the gaps that HIPAA provides, it's a biggest protection than what individual insurance provides. There's no protection. It's considered free market, and they can deny you for anything. As a matter of fact, I was denied from individual insurance because I had had a C-section.

Because of this, we began looking to see if we could find a group insurance policy for two people. Almost all of the insurance companies required three to five people to qualify as a small group. I finally talked to UnitedHealthcare, which allowed a two-person company to be considered a group. If they had not accepted this, my husband and I were going to have to close our small business, and he was going to go to work for a large company so we could have protection in a large group.

I'm thankful that they accepted me as a small group, but I live with the fear every day that I could be rejected.

During the time that I was trying to find insurance, I asked my father if he would be tested first because the genetic disorder was going through his side of the family, not my mother's, which is what we usually hear. He took the genetic test and came back positive. I knew from my research that I had a 50/50 chance of carrying this mutation as well. It was tortuous waiting the three months to find out if I could have insurance, but I finally was able to take the test, and I took came back BRCA Strain 1 positive. This means that my twin daughters have a 50 percent chance of having the mutation as well, and I have up to an 88 percent risk of breast cancer in my lifetime, and up to a 44 percent risk of ovarian cancer as well.

Ovarian cancer is particularly alarming to me because I've heard that there's a 50 percent mortality rate once you are able to be diagnosed with it.

I'm so grateful that I have an opportunity to save my own life, though. I hope to have a prophylactic mastectomy this fall, and I've been told that it gives me a 95 percent chance that I will never have breast cancer. After I'm finished having children, I plan on having my ovaries removed as well. That too will give me a 95 percent chance that I will never have ovarian cancer.

There was a point where the fear of death just outweighed my fear of discrimination. That's why I pushed through with being genetically tested in spite of my fears. I have had several problems along the way because I was tested genetically. The first one I mentioned earlier. We put off being tested for 10 years because of what the physicians recommended because of the potential discrimination. Countless women in my family during the last 10 years have been diagnosed with breast cancer, and several of them have lost their battle to breast cancer. That could have all been prevented if we had pursued testing then.

That really grieves me thinking about the loss of life there that could have been avoided. I've decided that knowledge about my health is a gift. I want everyone to feel the freedom to have that gift. My dad and I paid for our own tests because of our fear, and then my doctors, after I was diagnosed with BRCA Strain 1 positive, changed my diagnosis code for each procedure they requested. When I had my breast MRI, they didn't write that I'm BRCA Strain 1 positive. They didn't feel safe for the protection of me. They simply wrote that I had a strong family history, and I know that that means that there is discrimination out there or they wouldn't do that. Being BRCA Strain 1 positive is a stronger case than having a strong family history.

I am in the process of sending out information packets to 86 different addresses of my direct relatives and thinking about the fact if there are 86 different addresses, how many different people live there, because this is a disease that affects men and women. Men have the same chance of carrying this strand as women do. As I talk to my family members, the amazing thing to me is they have more questions about genetic discrimination than they do about how it affects their health. That is so sad to me, and most of the relatives that I've talked to have refused to take the test because of that fear. I can't help but think that just in my family, if we could all band together, how much we could do for the research of genetics if they felt the freedom to be tested.

I have one cousin that desperately wants to take the test, but her husband is a preacher at a small church and they have an individual insurance policy. So she can't take the test. There is no protection at all for her. She's 35. She's two years older than me. Of the 13 women in my family that have had breast cancer, most of them have been in their 30s. It makes me so sad that she has to wait until she has cancer until insurance will pay for a procedure.

One last area of concern is a problem that I'm having with insurance currently. I have had my insurance for less than a year. Other than the basic screening tests that I've had to determine whether I currently have cancer or not, such as CA125 counts, ultrasounds, mammograms, a breast MRI and a needle biopsy, there have been no other expenses this year, and I was just informed that my health insurance has been raised $100 a month with no explainable reason. I've talked to several medical people and they say that this is unusual and looks suspicious.

Another problem that I'm having currently is that prophylactic measures are not something that they automatically cover. Over two months ago I requested for them to agree to cover a prophylactic mastectomy for me. It has been over two months and no progress has been made on this issue. My father had bypass surgery five years ago, and there was no board of review that he had to go to for that surgery, and I don't understand why I have to sit and wait during that two months.

One thing that I would like to leave you with is I so appreciate you listening. My medical management and the medical management of many in my family have greatly been affected both by genetic discrimination and just the fear and the possibility of it. Unwitting discrimination has become a major part of my daily life. Discrimination worries me so much for the future as well.

Last Saturday my husband Jonathan and my two daughters, Audrey and Anna, and I walked in the Race for the Cure. My 4-year-old daughters had so many questions about what was going on and what was it about, and my answers had to be simple because they were so young. But I couldn't help but think what a complex issue this has been for me. My

prayer is that when they are old enough to decide whether they should be tested genetically, that discrimination isn't even part of their decision process.

Finding out your genetic status is permanent. You can't take it back, and it isn't something that you can change your mind on. What I really need, and what we all need, is a law that clearly defines the safety and the fact that you cannot be discriminated against genetically. We don't know what the future holds or how society is changed, so at this point I'm very vulnerable depending on the direction that that goes.

Thank you so much for your time, and thank you for inviting me to tell my story.

MS. MASNY: Thank you, Ms. Funk.

We'll now hear from Ms. Hinestrosa.

MS. HINESTROSA: Good morning. My name is Carolina Hinestrosa. I am a 10-year, two-time breast cancer survivor. I'm a mother of a 13-year-old daughter. I'm also the executive vice president for programs and planning of the National Breast Cancer Coalition.

My first diagnosis with breast cancer was at the age of 35. My second diagnosis was at the age of 40. My younger sister was also diagnosed twice, first at age 29, and then at 34. Over Christmas last year, two of my cousins and an aunt were diagnosed with breast cancer as well. Of course, we suspect there is a genetic mutation that predisposes members of my family to breast cancer.

I sought genetic counseling as part of a study. After carefully weighing the potential benefits and harms of genetic testing, I decided not to undergo testing for fear of potential consequences to my daughter. My fears are two-fold, first that the information may not be protected and might even be misused. I also worry that if I test positive, my daughter might be obligated to disclose the presence of a genetic mutation and that she might suffer future discrimination in health insurance and employment as a consequence.

I have four sisters and a brother. We all worry about our risk for breast cancer and the potential risk for our daughters, yet none of us feel safe enough to undergo genetic testing. My family experience illustrates why our nation needs strong nondiscrimination laws.

Since its founding in 1991, the National Breast Cancer Coalition, of which I am a member and am executive vice president, has changed the world of breast cancer in public policy, science, industry and advocacy by empowering those with breast cancer, our families and friends, and creating new partnerships, collaborations, research foundation opportunities, and avenues for quality access to health care.

The National Breast Cancer Coalition is now over 600 strong in terms of organizations who are members, and we represent several million patients, professionals, women, our families and friends. Coalition members include cancer support information and service groups, as well as women's health and provider organizations.

The mapping of the human genome has brought with it the promise of reducing human suffering by targeting interventions for those at risk for disease. The National Breast Cancer Coalition believes that strong legislative and regulatory strategies must be established to address the protection of individuals from the misuse of genetic information at the national, state and local levels of government. Genetic information is uniquely private information that should not be disclosed without authorization by the individual. Improper disclosure can lead to significant harm, including discrimination in the areas of employment,

education, health care, and insurance.

The 1996 Health Insurance Portability and Accountability Act, HIPAA, took significant steps toward extending protection to individuals from genetic discrimination in the health insurance arena by creating privacy standards, but this law does not go far enough. It is time to extend protections against genetic discrimination to everyone. The development of new genetic tests necessitates legislative and regulatory strategies to address the issue of how to protect individuals from the misuse of their genetic information.

Fear of potential discrimination threatens both a woman's decision to use new genetic technologies and to seek the best medical care. Women are also afraid to enroll in research and clinical trials that involve genetic studies, and this in turn threatens the viability of the scientific community to conduct the research necessary to understand the cause and find a cure for breast cancer. Many of the women testifying at present in this audience today have experienced exactly those concerns.

NBCC strongly supports the enactment of legislation that would protect millions of individuals against discrimination not only in health insurance but also in the workplace and that will provide strong enforcement mechanisms that include the private right of action. For this reason, NBCC supports H.R. 1910, the Genetic Nondiscrimination Health Insurance and Employment Act authored by Congresswomen Louise Slaughter. This legislation prohibits health plans from requesting, requiring, collecting or disclosing genetic information without prior specific written authorization of the individual; from using genetic information or an individual's request for genetic services to deny or limit any coverage, to establish eligibility, continuation, enrollment, or contribution requirements; and from establishing differential rates or premium payments based on genetic information or an individual's request for genetic services.

This legislation also prohibits employers from using genetic information to affect the hiring of an individual or to affect the terms, conditions, privileges, benefits, or termination of employment unless the employment organization can prove this information is job related and consistent with business necessity. Also, from requesting, requiring, collecting or disclosing genetic information prior to a conditional offer of employment; or under all other circumstances requesting or requiring collection or disclosure of genetic information unless the employment organization can prove this information is job related and consistent with business necessity.

It also prohibits from accessing genetic information contained in medical records released by individuals as a condition of employment in claims filed for reimbursement for health care costs and other services. Also, it prohibits releasing genetic information without specific prior written authorization of the individual.

Most importantly, H.R. 1910 contains strong enforcement language and provides individuals with a private right of action to go to court for legal and equitable relief if they are a victim of genetic discrimination whether they are subject to discrimination by the health plan or the employer.

NBCC does not support the Genetic Nondiscrimination in Health Insurance and Employment Act, S. 1053, passed by the Senate in October 2003, because it does not contain sufficient enforcement provisions. Unlike H.R. 1910, S. 1053 does not provide individuals with a private right of action should they become a victim of genetic discrimination in the individual insurance market. NBCC believes that a right with no enforcement is really

not a right at all. It is for that reason that no matter how carefully a bill is worded, no matter how much effort is put into it, including protections that breast cancer patients need, if that bill does not have a strong enforcement mechanism, then NBCC will not support it.

As we clearly can see from the witnesses here today, genetic discrimination is a real and growing problem that needs an immediate solution, not one that should wait until we have further cases of women and men who have experienced this type of discrimination that is so detrimental to the ability to seek quality health care.

Thank you for the opportunity to share the views of the National Breast Cancer Coalition.

MS. MASNY: Thank you very much for your own personal experience, as well as for the views of the National Breast Cancer Coalition.

Lastly, we'll hear from Phil Hardt.

MR. HARDT: Good morning. It's a privilege to be here today, and I want to thank the committee for inviting me to share my thoughts and personal experiences with everyone on the critical subject of genetic discrimination.

I have two genetic diseases, hemophilia B, a bleeding disorder, which I inherited from my mother, and also Huntington's disease, a degenerative brain disorder, which I inherited from my father. My two biological daughters and granddaughters are all carriers of hemophilia B, and as a result I now have two handsome grandsons who must also infuse with clotting factor each time they get hurt. All three of my biological children were at risk for Huntington's disease, but I am happy to report that none of them carry the destructive gene and cannot pass it on to subsequent generations. One tested publicly, and two tested anonymously to conceal their outcomes.

I mention biological children because I also have five adopted children, four of whom have severe handicaps.

Nevertheless, our story is one of continuing genetic discrimination even though we have laws that are supposed to protect me, my children, and my grandchildren.

"It was the best of times, it was the worst of times," as Dickens said in "A Tale of Two Cities." Because of advancements with the Human Genome Project, we now stand on the brink of having more useful information that has the potential of helping literally millions of individuals prepare early for various diseases. However, the reality is the knowledge that you are carrying any particular genetic disorder, in my case hemophilia and HD, is just as devastating to you, your children and your grandchildren as the disease will be later. This is further exasperated in Huntington's disease because of the severity of its symptoms and the absolute necessity for those who face the 50/50 chance of inheriting it to prepare early and thoroughly in order to minimize its overall destruction.

Tens of thousands of individuals with Huntington's disease have lived and died and are already in the insurance company's profitability calculations. However, it wasn't noted on their death certificates because of genetic discrimination fears. It is ludicrous now to believe that because you can know early that you might inherit a genetic disorder that all of a sudden we're going to create higher medical costs. This is not the case. We are living examples of the Tiresias complex. If you remember, the blind seer Tiresias confronted Oedipus with the dilemma, "It is but sorrow to be wise when wisdom profits not."

Huntington's disease is an inherited progressively degenerative brain disorder that results in loss of both mental faculties and physical control. It causes brain cells

to die prematurely. Loss of these brain cells causes very specific impairment and eventually death. Every child of an affected parent has a 50 percent chance of inheriting the gene and developing the disorder themselves. If HD is passed on by the father, another risk exists of anticipation occurring and each gene-positive child becoming symptomatic, even as early as a young infant or in their teenage years.

HD symptoms debilitate a person when they least expect it, usually in the prime of their lives, around 40 years of age, when they still have children at home and are actively pursuing careers. Living with HD is like living with Alzheimer's, Parkinson's, MS, and going insane all at the same time. Genetic testing has been available for Huntington's disease for longer than any other adult-onset disorder, since 1993. The discovery of the genetic mutation causing Huntington's disease made possible the use of predictive testing to identify current unaffected carriers. In 2000 Cohen said, "Genetic testing is intended to give families with a family history of HD the opportunity to assess their own risk for developing the disease more specifically, monitor their health status closely and, if a predictive mutation is present, make informed choices about reproduction and lifestyle."

It is interesting to note here that before 1993, the almost quarter of a million individuals who are at risk for HD in the United States were polled, and overwhelmingly about 90 percent of them said that they would take advantage of the test to find out if they were carrying the destructive HD gene. However, since the definitive test became available, fewer than 10 percent have tested as a direct result of genetic discrimination.

I'd like to now tell a little bit about my family history. In 1971, I was diagnosed with hemophilia B. In 1989, I was hired by Allied Signal Automotive and told by the HR manager there not to tell my boss about my hemophilia or I would never be promoted or trained because he wanted to get the biggest return on investment for his bucks, and if he knew I might have a disability, I would never go anywhere in the company. Consequently, all future bleeding episodes had to be hidden from him.

In 1996, a claim I filed for credit insurance on a car I had purchased for my daughter was denied because I had recently seen a neurologist regarding problems that I was having. In 1997, I was diagnosed as having Huntington's disease. In the year 2000, my oldest daughter married and applied for mortgage life insurance. She was turned down by every major insurance company because of Huntington's disease. Copies of several rejection letters are included in your packets, and note that the insurance companies don't even have fear of putting their rejections in writing.

Each of her rejection letters state two pertinent facts that are important. Number one, they each state that they will not insure her until she has tested for Huntington's disease, and two, that she is found to be negative. Then the insurance agent on one of the letters where they insure her husband writes a note at the bottom that says when you find out your status for HD, then we can insure your children, showing that the discrimination is down to the third generation now.

In 2002, my grandson, Enoch Maximillion, is denied health insurance coverage because of hemophilia that he inherited from me, and a copy of this denial is also included in your handouts. They must now earn less than they are capable of to qualify for state welfare in order to get coverage.

In 2002, my daughter Michelle and son Phillip tested anonymously for HD to protect them in case either of them tested positive. I am over the Huntington's Disease

Society of America, Arizona affiliate in the State of Arizona, and in 2001 a geneticist and I established anonymous genetic testing to protect individuals so that they can use a bogus name and social security number and address and all other information, and pay cash. But the problem is it's very expensive. It's around $900 out of pocket to find out. But it is completely concealed. But it's a shame that we have to do this.

Last year I applied for long-term care insurance and was rejected on the basis of my HD after becoming divorced and realizing that I would probably need someone to take care of me later.

Now, here is a list of ways that open genetic discrimination adversely affects those with HD over and above the negative effects of the disease itself. Those who are at risk are reluctant to participate in research, even anonymous research, because they fear being found out. For example, the PHAROS study for HD could have almost a quarter of a million at-risk individuals in it, but they have only been able to recruit about 1,000. Imagine the decrease in numbers. Other important research tests are no different. Because of our small numbers, unfortunately, we need every bit of data possible to make things significant.

Proper medical and mental health care are not sought on a timely basis that could have (inaudible) help reduce suffering and raise everyone's quality of life. Open communication is almost non-existent between parents and their at-risk children regarding how they can better prepare to minimize the destruction of HD if they do have it. HD must be kept shrouded in secrecy to protect everyone. For the same reason, at-risk children are not encouraged to seek good education, college education, careers with companies who offer group benefits, marriage and childbearing options, including adoption. Misdiagnosis and the same thing with medication occur because one doesn't know, or knows but can't be honest with their doctors and other health care providers for fear of being discovered. Healthy living habits aren't adopted either early on to postpone onset.

Now, using our negative experiences with being wise and our wisdom not only doesn't profit us but is even used against us. How many other future discoveries that have the potential to bless the lives of millions of others by predicting other diseases soon enough for individuals to take positive action against them will be thwarted because of flagrant genetic discrimination?

Thank you very much.

MS. MASNY: Thank you, Mr. Hardt.

And thank all of you for your very profound testimony.

Now we're going to open up to the committee a question and answer period to be able to direct questions to our panelists, if any members of the committee would like to direct any questions to them.

DR. TUCKSON: I just want to, just as we get into this, just say to each of you that on behalf of all of us on the committee, we really appreciate your taking the time to come and talk to us. I can imagine that it's not easy to do what you all just did. I will assure you from the chair's desk here that as we go forward in our work, we're going to make sure we do everything in our power to make it worth your while, that something will come of this.

I know you'll go home and people will say what happened? We're going to try to make sure that something happens as a result of your doing this. So before my colleagues get into the specifics, just a real big thank you to you all.

DR. COLLINS: I also want to thank all of you for the very powerful and

moving stories that you have told, which certainly underline in stark and compelling terms the need to do something about a situation which grows worse every day. It is, I'm sure, a great disappointment for all of you that we haven't fixed this by now, when the arguments are compelling, when you can see that the likelihood of more and more genetic testing being offered is inevitable, and therefore the likelihood of more and more people facing up to the dilemmas that you have faced also becomes inevitable.

I must say that after a year ago, just about exactly, seeing this bill pass the Senate unanimously, it seemed as if this was finally going to get solve, and yet here we are.

Becky Fisher, I know you have been a very effective voice in terms of carrying this message on the Hill. What do you hear when you speak to people, particularly on the House side, about the importance of doing this? What's the roadblock that is getting in the way right now given how compelling the arguments are, as all of you have presented? What do you see as the reason why this hasn't been solved, and what do you see as the way around that?

MS. FISHER: Thank you for asking me that, Dr. Collins. Someone else asked me that, actually, at the D.C. City Council. They were considering the legislation for the City of Washington, and my response wasn't exactly politically correct, but it is what I believe. The United States Chamber of Commerce is the strongest, loudest voice speaking out against this legislation. Without going into too much detail, they have a lot more money than a medical librarian housewife living in northern Virginia, and they have a lot more clout than we do.

The only problem is they don't have any moral authority. So I still continue to believe that we will get it done. With all due respect to the National Breast Cancer Coalition, I think their support of the Senate bill would be a huge, huge help for us, because most of us don't really want private right of action, we just want the protection. So I would like to go on the record as expressing that for myself and for literally hundreds of people that I know who are in the same boat.

MS. MASNY: Ed?

DR. McCABE: This is really two parts. Again, I wish to share what has been said before. These are powerful, very important statements that all of you have made, and I appreciate all of the sacrifices that you have gone through before, you and your families, and the sacrifices that you make just to appear before us today. So thank you very much.

We as a genetics community, and also as members of the public, have been told that genetic discrimination does not exist. We've been told that over and over. In fact, scholarly articles have been written and are referenced in the genetics literature where the authors made inquiries to insurance companies, and guess what? They said there is no genetic discrimination. Yet, all of us know that it exists, and that's why this is so important today.

I would ask perhaps Mr. Chairman that the National Chambers of Commerce be invited at some future meeting to appear before us so that they could explain why it is that they value individuals as commodities more than they value them as individuals, because I am very concerned that these members of our communities have this as such a profound policy position, so profound that they have protected this bill from leaving the desk in the House of Representatives. I don't know if it has left yet.

But to be able to freeze a bill that passed unanimously in the Senate without leaving the desk, we heard in a previous meeting from a House of Representatives staff person that the House has to have separate hearings. They can't simply ride on the coattails of the Senate. There have been no hearings because it did not leave the desk, and I would like to hear

from the most powerful group that has protected that.

We've also been told in previous meetings and op-ed pieces, in the literature, that there is no need for legislation because we are already protected by legislation, and there's a litany of laws that are cited, including the ADA, to protect individuals against genetic discrimination. I would ask, and I'm sorry to put you on the spot, Dr. Majidi, but I would ask has the Department of Justice ever gone through a systematic review of the current legislation to document that, in fact, as citizens of this country we are protected by the existing laws?

DR. MAJIDI: I'm afraid we haven't been quite approached to do that review yet. So as of now, I don't have any specific information for you.

DR. McCABE: Who would need to approach you to do that review?

DR. MAJIDI: Basically, the Secretary for Health and Human Services would be a good starting point.

DR. McCABE: Well, given that we have been, and that I until recently have been writing laws to two Secretaries of Health and Human Services, if another letter from this committee could move that agenda forward and determine whether in fact we are protected or are not protected, I would ask the committee to give consideration to yet another letter.

MS. MASNY: Yes, we will take that into consideration.

Also, for the record, just to mention that the Chamber of Commerce was invited to this meeting with a specific invitation today, but actually referred us to another group who we'll be hearing from in the stakeholder panel, the third panel.

DR. McCABE: What is the name of that group, please?

MS. SARATA: The Society for Human Resource Management.

DR. McCABE: And we have a document from them also documenting the laws that exist, which was part of what precipitated my question. I would ask if the Society for Human Resource Management considers themselves a spokesperson for the National Chambers of Commerce. So perhaps they can be prepared because I will be asking that question when they speak.

DR. FROHBOESE: Good morning. I join my colleagues on the committee in thanking each and every one of you for your courage and important advocacy in this important area.

Following up on Dr. McCabe's question about various laws and their coverage, I know several of you, I believe Ms. Fisher and Ms. Hinestrosa -- I'm sorry if I'm mispronouncing your name, Ms. Hinestrosa -- mentioned a HIPAA gap, and I wondered if you could address that a bit more from your perspective. I'm particularly curious as to whether it has to do with the privacy rule, which my office within HHS Office for Civil Rights is responsible for enforcing, or if it has to do with other aspects of HIPAA.

MS. HINESTROSA: Well, the protections that HIPAA brings about really are for people who are members of a group plan and don't extend to people in the individual market. So that is an important group of people who are not well protected by HIPAA.

MS. FISHER: I would just like to add to that, that my understanding of the HIPAA gap is that -- well, the biggest problem is it doesn't address any workplace concerns at all, biggest problem. It only deals with insurance. If you're just considering the insurance pie, then that pie is sliced up again into people who are protected and people who aren't protected by HIPAA provisions. Those who are protected are protected under group health insurance

plans.

This is important because most people don't understand this. HIPAA does not extend to ERISA-exempted state plans. Seventy-seven percent of employers offer at least one state self-funded insurance plan. So that means that if my husband works for Bank of America, which he does, we have the choice every November of getting 10 different choices of plan that we could subscribe to. We could go with a Blue Cross or an Aetna and be in a group health plan, or we could go with Bank of America self-insurance plan. That's Bank of America betting that they're going to do better than Blue Cross is with that money.

Blue Cross happens to administer that plan, so I don't really have a clue that I'm not protected under HIPAA. This is a big problem. There are 100 million people in this country who are having the same problem, and they don't even know it. So I think this is a really important piece of information to get out there.

Dr. Collins asked earlier what was the pushback on the Hill. The pushback is, "No problem, we did HIPAA." Well, guess what? HIPAA doesn't work for a lot of us, and I'm glad to have the opportunity to tell you all this because my guess is that it's never gotten that granular with any of you either. But those of us who know what we're up against know this.

Thanks for asking that.

MS. MASNY: Once again, we'd like to reiterate what we've heard from every one of the members here, to thank you very much for your testimony. We certainly have heard you and we already have some actions that we'll be ready to take as we discuss further.

Yes, Ms. Williams?

MS. WILLIAMS: I'd like to reiterate the fact that a lot of these people come forward and take a chance, that when they go back to wherever they come from that they could be summarily fired, that they could have their insurance policies pulled from them. I come here with nothing to lose, okay? The VA takes care of me and I'm on disability. I have nothing to lose. But these people who sit to my left have a lot to lose when they go home, and they know that, and they come here at great risk to themselves and their families, and I want everyone to understand this.

I want the House to understand this, that there are people here at risk, and my children who are here today are still at risk. As they get older and they choose their careers, my daughter is a competitive gymnast. She's 8 years old. She wants to grow up and be a gymnast. But there is a chance that she could be discriminated against because she is a carrier of alpha-1. My son, he wants to be a research scientist. He wants to build habitats on the moon. He's 10 years old. There's a chance that when he gets to that point, he may not be hired in his chosen field. He may have to, God forbid, flip burgers at McDonald's.

Let's see, at age 9 he was considered to have an estimated 138 IQ, and it's a lot more than that now. I mean, he may have to let his considerable IQ go to waste. And nothing against McDonald's. They fund college educations all over this nation. But my son has the potential to do great things, and that may not happen. But I am more concerned with the people to my left right now because I know what can happen to them. I've heard it, I've seen it, and that's what I want everyone to understand.

Thank you.

MS. MASNY: Thank you, and we will definitely work today not to let any of your own experiences go to waste. Thank you.

What we're going to do is we're going to take a break now, and that will be

until 10:45, and then we'll reconvene for Panel 2, which will be the health care providers.

(Applause.)

(Recess.)

MS. MASNY: Our second panel, then, is of health care providers. The committee, then, will be hearing from them regarding their own patients' experiences that they have been in contact with, and they'll be telling us about their patients concerns, about the impact of genetic discrimination.

So our panel, then, will be three, and again they'll each have 10 minutes to present, and we'll have 10 minutes for questions after their presentations. So we'll be hearing from Jeff Shaw, who is an oncology genetic counselor from Penrose Cancer Center in Colorado. Then we'll be hearing from Don Hadley, who is a researcher at the National Institutes of Health in Bethesda, Maryland; and from Mark Brantly, who is the director of the alpha-1 antitrypsin testing program at the University of Florida.

So we'll begin with Jeff Shaw.

MR. SHAW: Thank you. My name is Jeff Shaw, and I am the director and genetic counselor for the Hereditary Cancer Service of Penrose Cancer Center in Colorado Springs, Colorado, elevation 6,300 feet. I don't quite know what to do with this much oxygen.

(Laughter.)

MR. SHAW: I would like to thank you for giving me the opportunity to present information from our program to this committee. The clinicians here feel that you all were a very difficult group to follow, but one thing we've got is numbers.

I provide genetic counseling for individuals and families in every area of medical genetics, from prenatal to adult-onset conditions. For the last seven years I've worked exclusively with patients concerned about hereditary cancer predisposition, much like the women who have presented here today. The purpose of our program is twofold. One is to provide the best estimate of cancer risk so that screening can be appropriately modified so that if a cancer occurs, it can be caught as soon as possible when survival is the highest and treatment is easiest to go through. Two, it's to provide appropriate implementation of medical and lifestyle interventions to drastically reduce the risk of cancer, especially in those with an inherited predisposition.

Our program is clinically based and merges the gap between research-based programs and the implementation of genetic testing into general medical practice. Although we are clinically based, at the outset of our program we created a large database to collect information we felt would be helpful to the provision of our service. The data presented today covers seven years of clinical service to over 900 individuals for hereditary cancer predispositions.

I'll start with the fear of genetic discrimination. Genetic counseling for presymptomatic cancer predispositions is complex and time consuming. It involves, of course, a detailed family history that we try to confirm with medical records, education regarding the differences between sporadic, familial and inherited cancer predispositions, and also the psychosocial issues and family issues that are involved with this type of testing, because, as you've heard, if you test one individual, that information is going to be applicable to their entire extended family.

If you look at the entire number of people that we see, 61 percent of patients have a family history strong enough to indicate the possibility of genetic testing. If an

individual is offered the possibility of genetic testing, we go through a rather lengthy informed consent process discussing the risk, benefits, and limitations of that testing and how it would apply to their medical care. Unfortunately, when I'm supposed to be talking about medical care and medical decisions, the bulk of this discussion I feel I need to be a lawyer, because it is strongly centered around the concerns our patients have regarding genetic discrimination, not only for themselves but for their siblings and, I find most importantly, for their children.

In our program's experience, 20 percent of those individuals who are eligible for genetic testing for presymptomatic cancer decisions declined having the test. Of those individuals, 22 percent did so because of a fear of discrimination. You must also realize that the people who see me are already motivated to learn more about their family history.

Of interest, the patients that did decline based on a fear of discrimination, 90 percent of those had a very significant increased chance for testing positive for a mutation that would increase their risk for cancer. Therefore, the people who would experience the greatest possible benefit from this testing are the most likely not to pursue it.

I'd like to give a few patient experiences because these numbers take on a more personal tone in the context of real people. I recently saw a woman who had a very strong family history of cancer, much like the women who have talked earlier today. She had just been diagnosed with a Stage 1 breast cancer at the age of 46. Her mother died of ovarian cancer at the age of 52. Two maternal aunts had breast cancer in their early 40s and have passed away, and her maternal grandmother died from breast cancer at the age of 41. Due to this strong family history and her own diagnosis, we determined she had at least a 43 percent chance of carrying an inherited mutation that would increase her risk for second primary cancers.

If she pursued the testing and was determined to carry one of these faulty genes, she would have up to a 60 percent increased risk for a second primary breast cancer, a brand new one, and up to a 44 percent chance of developing a primary ovarian cancer. Prophylactic surgical intervention could reduce her risk for these cancers 90 percent or greater. If she tested positive, each of her four daughters would have a 50 percent chance to inherit this faulty gene that could increase their risk for cancer.

She has declined testing, and this has been very emotional and very difficult for her, but she's done so because she is very concerned as to how this information could affect her children's chances of getting health insurance. She doesn't know what profession they're going to go into, she doesn't know if they're going to be group or self insured, she doesn't know what state they're going to end up living in. Because of that, without the genetic testing, it's unclear how to proceed with her medical care, especially the surgical ones that could reduce her risk for cancer developing again.

However, without documentation of a mutation that she would carry, her insurance company will not pay for any of these surgical interventions. Therefore, she remains in a state of anxiety, using imperfect breast and ovarian cancer screening methods and simply hoping that another cancer does not occur. Due to her current employment situation, she might have to change insurance companies. She's afraid that if she were to change insurance companies, she could be denied, and with her current diagnosis she simply cannot afford to be without health insurance.

Another patient we had had a strong family history of FAP. This is a dominantly inherited colon cancer predisposition characterized by early onset of colon polyps,

hundreds to thousands of these that can begin as early as the age of 10. Individuals with this condition basically have up to a 100 percent chance of developing colon cancer sometime in their life. He worked his whole life at a relatively small company with a small self-directed group insurance plan. He has been warned by his doctor not to have genetic testing for FAP as he would lose his job or his health insurance if they were to find out about the condition in the family.

At the age of 42 he had significant rectal bleeding and finally went in for evaluation. He was found to have over 400 polyps in his colon. It was so extensive that he needed to have his entire colon removed, a drastic but life-saving technique for these individuals. Luckily, he did not have an invasive colon cancer. Other family members were not as lucky. Most of those affected with FAP in his family died of colon cancer in their late 20s.

At the age of 46, he came to me for genetic counseling. He has two children, ages 22 and 24. He had not informed them of the condition prior to this time as he did not want the family history in their medical records due to a fear of genetic discrimination. Unfortunately, this meant that these early 20-year-old children were not having appropriate screening. With several genetic counseling sessions with him, he finally decided that he would do the testing, even with his fear of the discrimination, in order to have appropriate medical care for his kids. He was tested and the genetic mutation in the APC gene that was causing FAP in his family was identified.

His children decided to have testing. One child has tested positive, one child has tested negative. The 22-year-old that tested positive is now having appropriate screening, but also lives in fear that at some point she could lose her medical insurance.

The fear of genetic discrimination in this family could have caused the same early deaths in his immediate family as it did in his extended family.

I just saw a 24-year-old patient whose mother tested positive for a mutation in one of the breast/ovarian cancer suppressor genes. She did not have testing based on a fear of discrimination. She was diagnosed with a Stage 3 breast cancer and died in June. She finally had the testing done, and a mutation was identified. Therefore, we could cheaply test her three children to see if they indeed inherited this mutation or not. All three children decided they did want to be tested because they felt it was important for their care. All three children paid for this test out of pocket to keep this information as confidential as possible.

In fact, when we look at individuals when the cheaper test can be performed, this is about $350 when a mutation has been identified in a family, 74 percent of those individuals will pay out of pocket because of their fear of genetic discrimination. Unfortunately, if you're the first person being tested in the family, the test costs about $3,000, making that really not an option for the bulk of individuals that we see.

Although she has informed her family about this mutation, well over 50 percent have decided not to be tested based on a fear of discrimination. Although anecdotal, my experience with hundreds of families shows me that this is the case for many of these family members.

What about after testing? We conduct one-year follow-up surveys of all patients seen by our program. We're happy that we have a 72 percent response rate to these surveys. Of those patients who tested positive for an inherited cancer predisposition, 70 percent report having continued significant anxiety that they would experience genetic

discrimination at some point in the future. Fear of future genetic discrimination remains a real concern for our patients, especially those who have tested positive.

Then we were wondering about those people who don't even make it for genetic counseling, they don't even get to the point of being offered testing. In addition to a fear of discrimination from genetic testing, there's also fear of discrimination simply from participating in a genetic counseling session. In 2001, Geer, et al., studied factors that would influence an individual's decision not to come in for counseling. Of those declining genetic counseling, the biggest reason was a fear of genetic discrimination, accounting for 40 percent of those individuals surveyed.

Our program has had a significant number of physician-referred individuals who did not show up for their scheduled appointments. After seeing the Geer study, we wanted to see informally if this was a concern for the people referred to our program. We conducted an informal six-month survey of those patients not pursuing referral for genetic counseling by phone. In this time frame, we had 60 patients that did not show. Fifteen percent would not return our calls, and of those we could contact, 49 percent stated that they had changed their minds, that they had heard from their physicians and family that they should not have this documented in their medical chart due to genetic discrimination. I was unable to get the bulk of these people in for an appointment.

This informal survey supports the data seen in the Geer study. It shows us that a fear of genetic discrimination is a barrier for individuals that could benefit greatly from genetic counseling and possibly genetic testing for hereditary cancer predispositions.

When I graduated with my genetic counseling degree in 1994, there were but a handful of genetic tests available for inherited conditions. In 2004, just 10 years later, there are over 1,000 genetic tests available on a clinical or research basis. The number of genetic tests that will become available for single-gene and complex genetic disorders is expected to increase exponentially over the next decade. I fear that without strong federal protection, the appropriate use of these tests will continue to be under-utilized and we will not gain the benefit from the genetics revolution.

None of us are genetically perfect. Learning what genetic imperfections we have inherited and how they affect our risk for disease is difficult, sometimes frightening, and a life-changing experience. The decision to have presymptomatic genetic testing is multifaceted. It encompasses issues regarding one's sense of self, family relationships, anxiety, depression, and very complex decisions regarding future medical care. The citizens of our country need to be assured that when they are deciding whether or not to pursue genetic testing, a fear of genetic discrimination is not a factor.

Thank you for your kind attention, and I look forward to answering any questions.

MS. MASNY: Thank you, Mr. Shaw.

We'll hear next from Donald Hadley.

MR. HADLEY: Thank you. I'm an associate investigator and a genetic counselor within the National Human Genome Research Institute at the National Institutes of Health. I appreciate the opportunity to present our work to the committee.

My professional experience and work has focused primarily on providing education and counseling to families who are threatened with genetic and inherited diseases. I've had the privilege of working with these families for the last 23 years. In October of 1993, I

was invited to join the then newly established National Center for Human Genome Research. Our goal was to develop research that focused on identifying factors that influenced the interest in and uptake of genetic counseling and testing and the associated psychological, social, and behavioral outcomes. Our research agenda was inspired by the identification of a rapidly growing number of genes that predisposed or increased susceptibility to disease.

My research has specifically focused on families who are newly diagnosed with a hereditary cancer syndrome named hereditary non-polyposis colorectal cancer, or HNPCC, and families who specifically have deleterious mutations that have been identified. The identification of families with an HNPCC mutation allowed the opportunity to learn from them before, during and after the provision of genetic counseling and the offer of genetic testing. We felt that identifying the factors influencing decisions and their outcomes was necessary to plan for a future when genetic testing will be more routinely used to predict risk for rare as well as common diseases in the general population.

Within our study, once a family is identified to carry an HNPCC mutation, we sequentially offer participation to first-degree adult relatives who are at 50 percent risk of inheriting the mutation. This offer includes the provision of comprehensive genetic education and counseling about HNPCC and the option of genetic testing. For those electing to pursue genetic testing, the Clinical Center at the NIH pays for genetic studies, removing the issues of cost and insurance coverage from the decisionmaking process. However, prior to the education and counseling sessions, we ask participants to complete a questionnaire that collects demographic information, information about their knowledge of genetics and genetic testing, their perceived cancer risk, and standardized scales assessing mood, coping style, spirituality, their perception of their own control over their health issues, and family relationships.

Additionally, we also ask them to specifically identify what factors motivate them to consider genetic testing, such as the desire to clarify their risk or their children's risk for cancer, or to guide their cancer screening. Likewise, we also ask them to identify factors that concern them about undergoing genetic testing, such as emotional concerns about handling the results for themselves or for other family members, their level of confidence in the prevention strategies that exist, and their concerns about test results affecting insurability. These questions are asked individually so we might obtain a level of significance for each issue.

In addition, we ask the participants to identify the most important reason for them to consider testing, and likewise the greatest concern they have in considering testing. All the questions are validated measures developed and used by the Cancer Genetics Studies Consortium of the NHGRI Ethical, Legal, and Social Implications Program.

As we began to talk individually with each participant, describing the intent of the study and the process involved, one key issue was consistently identified. That issue was posed in the form of a question: How might participation in this study affect my insurance or that of my family? The question came unsolicited in the beginning of the informed consent process. This question and the associated worry seemed to persist even after we had provided each participant with information that reassured them of the confidential nature of the study, that the study had a certificate of confidentiality issued by the National Institutes of Health; that all participants are issued study ID codes that removes their personal identifiers from the data and the test results; and that the cost of the testing is paid for by the study, so insurers are not involved; and that records are protected by the Clinical Center and only released if written

permission is obtained from the participant.

Nevertheless, it was clear that there was an overwhelming concern and in some cases a palpable anxiety about the impact of genetic testing on health insurance. These concerns dominate our informed consent process and recur session after session with an intensity that opened our eyes to the level of concern that the public feels about genetic discrimination. I specifically recall one young woman and her mother, both of whom had experienced uterine and colon cancers at young ages within a family riddled with HNPCC cancers. Even though this young woman had experienced cancer twice and felt there was little residual discriminatory risk to her, she was immobilized by the concerns about the potential of genetic test results branding her family as uninsurable.

She opted to wait on testing but would periodically call to discuss the safeguards our study provided regarding test results and the information obtained. She was admittedly tortured by the concerns about insurance risk, which she felt was keeping her from protecting her family. Finally, after months of considering the implications of testing, she returned to pursue testing, knowing that her results may well prevent others from experiencing what so many in her family had already endured.

Sequencing efforts did identify a deleterious mutation, providing a tool for those within her family to clarify their cancer risk, to focus their cancer screening, and to consider preventive steps such as prophylactic surgery. We anticipated that within the months that followed we'd be hearing from at least a few of her family members, but there were no calls, there were no emails, and there were no letters.

Through follow-up we learned that she had shared the results with her four sisters, and those four sisters expressed that their concerns regarding discrimination were too great to safely allow them to participate in a genetic counseling study with the option of genetic testing. They were worried about being in small companies with limited insurance options and the associated risk that genetic testing posed for them, as well as their children.

In 2003, we published a paper in the Archives of Internal Medicine, which I brought along for your consideration. In the paper we reported on attitudes, intentions, and uptake of genetic testing of individuals within these families. Of particular relevance to this group were findings regarding the level of concern that exists within these families about participating in a genetic counseling and testing research study. The questions that we used to identify their concerns regarding genetic testing are also included in the packet before you.

In looking specifically at what factors influence decisions, we identified and published that 39 percent of participants reported that their most pressing concern was their worry about the potential of a genetic test result affecting their or their family's insurability. I recently looked at our data to see if the level of concern has held true from the earlier analysis, since we've added approximately 80 additional participants. I found that the current baseline data suggests a number slightly higher than originally published. It's now about 43 percent identifying that their greatest concern regarding genetic testing pertains to their concerns about discrimination.

Furthermore, on follow-up at six and twelve months, a greater proportion of them identified discrimination as the single most worrisome factor. Specifically at six and twelve months, 48 and 54 percent, respectively, identified concerns about genetic discrimination by their insurer as their principal concern. Obviously, this concern is not going away with time and adjustment to the outcome of testing. This seems surprising since research

from other studies focused on presymptomatic and susceptibility testing demonstrate that other variables such as anxiety, distress, and mood, seemed to return to pre-test levels by about a year out from testing.

What's different about the concerns regarding insurance? If people have not experienced what they perceive as discrimination, why are there increasing concerns? Participants often ask: "Has anyone in this study ever reported discrimination on the part of their insurance company or employer?" Our answer is always the same: "Not that has ever been reported to us." But apparently, just reassuring them that discrimination in general and within our study is not the norm doesn't help. There is a pervasive mistrust that seemingly worsens with time.

In summary, the prevalence of genetic discrimination by insurance companies does not appear to be the key issue. The real issue is that the public perceives that the potential for genetic discrimination by insurance companies is an overwhelming risk, and in my experience this fear provides a barrier to genetic research and clinical genetics care. This barrier limits our potential for research and basic sciences and social and behavior research. The greatest tragedy, however, is the missed opportunity to prevent cancer or diagnose it early in persons at high risk who are unwilling to risk the potential of discrimination. Providing federal legislation prohibiting genetic discrimination will reassure the public that genetic discrimination is not a risk, provide an increased opportunity for research to address other, more significant issues, and most importantly reduce mortality and morbidity associated with cancers diagnosed at later stages.

Thank you for the opportunity to present our work.

MS. MASNY: Thank you, Mr. Hadley.

The last of our health provider presenters will be Dr. Mark Brantly.

DR. BRANTLY: I'd like to thank the committee for inviting me to come and speak. My name is Mark Brantly. I'm a pulmonary physician and a physician scientist at the University of Florida. I've been involved in alpha-1 antitrypsin deficiency testing since approximately 1983 and have tested about 20,000 individuals, identified about 2,000 alpha-1 individuals over the last 20 years. In recent years I've been testing approximately 5,000 to 6,000 patients per year for alpha-1 antitrypsin deficiency.

I follow approximately 150 alpha-1 antitrypsin deficient individuals in my clinic at the University of Florida and have first-hand experience regarding the impact of this diagnosis on them personally and also their families.

Let me begin by giving a brief expose of alpha-1 antitrypsin deficiency. It's a very easy disease to diagnose. It requires simply an alpha-1 antitrypsin level and a PI type or a genotype. It's one of the more common genetic diseases, with a frequency of 1 in 2,500 to 1 in 4,000 individuals. The phenotype is primarily chronic obstructive pulmonary disease and liver disease. It's oftentimes associated with a rapid decline in lung function punctuated by lung infections. However, it's one of the classic genes in which there's an environment and gene interaction. That is, individuals who have alpha-1 antitrypsin deficiency lose lung function much faster when they smoke cigarettes. Indeed, they die 20 years prior to non-smoking individuals.

Importantly, in my clinic population I have individuals that are 80 years old with profound alpha-1 antitrypsin deficiency who are living active lives. Therefore, prevention of behaviors and interactions is a critical aspect of this disease. It is not all about having

expensive therapies. People can live their entire lives with not having disease or disability if they're identified early and we're able to protect them. That, I think, forms the basis of early diagnosis and preventive care being critical if we are to make a significant impact in this disorder.

In the State of Florida only, there are 900,000 individuals with COPD, and 9,000 die per year. Almost 1,000 of these individuals have at-risk alpha-1 antitrypsin deficiency alleles. In the State of Florida we've had a program in which we have done targeted detection. We first began by establishing a consensus among the community with the help of the Alpha-1 Foundation that testing exceeded the risk of testing. We established a high-throughput laboratory, and we provided professional and lay educational materials to deal with some of the educational issues that are associated with alpha-1 antitrypsin deficiency diagnosis. We developed an easy testing system where patients can prick their finger and send it to our central laboratory, yet we still have significant barriers to testing these individuals despite major recommendations from the major thoracic societies recommending a Category A recommendation for testing.

These barriers include genetic discrimination, and particularly fear of genetic discrimination, ignorance regarding the disease among the physician population. We've also established tertiary care referral systems to make sure that when physicians do identify these patients, that they have someplace to go with these patients.

So we have yet still an important job, and that is to be able to -- instead, right now, we have 5,000 individuals that are identified with alpha-1 antitrypsin deficiency, and there are approximately an estimated 95,000 that haven't been identified. If these patients were identified early on, they perhaps could be protected from developing disability.

One of the approaches that we've used is doing a coding testing trial through the Medical College of South Carolina and Charlie Strange. This is funded entirely by the Alpha-1 Foundation, and it's been a longitudinal study looking at the reasons why people do not wish to be tested through their physician. We've tested now more than 3,300 individuals in this testing program and have done some initial longitudinal follow-up. I've provided you with one manuscript that gives you some of the results, but I'd like to focus in on a couple of things most recently that we have done.

The first one is the risk and benefits of genetic testing. Thirty-three percent of individuals said that the reason why they chose the coded testing trial was because of fear for losing their health insurance or higher health insurance costs. The other thing is in the post-test, who would you give your results to? Well, not surprisingly, they would give the results to their children and their spouse, and not surprisingly they wouldn't give it to their ex-spouse.

(Laughter.)

DR. BRANTLY: In addition, they would not provide this information to their health insurance companies or their life insurance companies. Indeed, only about 16 percent would disclose that. Sadly, though, I have to say that only 80 percent of these individuals who were profoundly deficient would even tell their personal physician, and that's problematic as far as I'm concerned.

Finally, one of the things that this study I think brings up in close contrast is that when patients were diagnosed with alpha-1 antitrypsin deficiency, obviously one of the major therapies is to do smoking cessation. While there was a trend towards individuals who had alpha-1 antitrypsin deficiency quitting smoking, this was not significant. In actuality, it

was higher for alpha-1 antitrypsin deficient individuals, still there was a large portion, greater than 80 percent, who did not quit smoking.

In my clinic and in many of the physicians' clinics who take care of alpha-1 antitrypsin deficient individuals, I have a 95 percent quit rate for cigarette smoking. The national average is 10 percent. Why is that? That's because I hound these patients to death. I schedule them for appointments to see me every month, I have my nurses hassle them, because I know of all the things that I do for these individuals, getting them to quit smoking is clearly one of the most important things that I can do.

When we have to resort to coded testing and we leave out the physician and the health care provider in helping these individuals cope with and make these changes, we short-change them in a big way. We short-change them because they're afraid, because they can't trust our system to protect them and to give them the correct information. There's only one difference between my patients and me. We all as complex genetic organisms have five to fifteen "lethal mutations" that may be associated with our demise or our disability. The difference between me and my patients is I don't know about mine. My patients know about theirs and they have the ability to do risk prevention.

Thank you very much.

MS. MASNY: Thank you for all your testimony. It continues to clarify that genetic discrimination, and especially the fear of genetic discrimination, is very real.

We're going to open it up now again to the committee members to ask our health care provider panel any questions they may have.

Brad?

MR. MARGUS: It's astounding how people's behavior didn't change in that last story, even after they have their genotype and they know their risk. So along those lines of changing behavior, did any of you ask those participants who were wary of sharing information, participating in a study or being tested, if they would in fact do so if there was a law that protected them? I mean, is it clear to you that if tomorrow we announce there's a very solid law that says you can't discriminate, that these people would suddenly then all tip and all be willing to participate and take the risk, or would there still be quite a bit of paranoia?

MR. HADLEY: I think there are going to be people who won't participate for other reasons. It's not going to be 100 percent of those people who don't participate that have insurance concerns as their major focus. So some will still be out there not participating, not getting information, not considering testing. But it's clear that from a qualitative perspective from talking with these people in the clinic, as well as a quantitative perspective that this is a pervasive fear. If we could simply say there are federal laws that prevent discrimination by insurance companies, by employers, that that would be removed, that concern would be taken off the table, and then we would have better opportunities to focus our efforts towards helping them take better care of themselves through screening, through diet, whatever modifications exist.

So I'm quite convinced that if we could simply say there's federal protection that prevents that type of discrimination, we'd have much better cooperation and be able to attack the problems that really exist with some of these disorders.

MR. SHAW: I would second that. Our problem in Colorado, we actually do have a state law that says you cannot deny health insurance or adjust rates based on genetic information. The problem with our population is that they're highly mobile, so these

individuals don't know if they're going to be in Colorado a year from now. So I believe if we could say to those patients that their biggest concern is that they could move and be in a place where they're not covered, if we had federal legislation, that would take care of a lot of that worry.

MS. FUNK: I know I'm from the patient panel, but I have something to add to this. Arkansas has a state law that protects me, and that was part of the decision process that made me decide to go ahead and pursue genetic testing. Without that state law, I do not think that I would have pursued it. But there are holes in it. It doesn't help with individual insurance, and there is always the possibility that I could move. So although the law is incomplete, I'm still thankful for that law and it did really influence how I chose to pursue my health.

DR. BRANTLY: This is Mark Brantly. I think that laws will definitely help, but I think that there also needs to be a societal change, too. That is, we have to recognize that disability shouldn't be a scarlet letter, or the possibility of disability. I think until we eliminate those possibilities, we're not going to be able to remove fear from these individuals because trust has to be out there. It's not just a law, it's about trusting the system to do right by you. People won't stand up until they can see that other individuals have been able to go through this and did not have problems with it, because these things individually affect them, and not only do they affect themselves personally but also their families.

You don't want to take chances with your kids no matter what. You'd rather die than take chances with your kids being able to be employed properly, to be able to basically fulfill their potential. So I think that until we develop a track record of protecting individuals, I don't think we're going to see as much enthusiasm for genetic testing.

MS. MASNY: Dr. McCabe?

DR. McCABE: I just wanted to follow up on the comment that Dr. Brantly made in the closing part of his statement. I think it's important and I want to emphasize this, that we all have genetic predispositions to disorders. Those who have come to speak to us today recognize what their problems are, what their potential problems are, and I think that this is very important for all of us to recognize. It is discrimination because it is arbitrary and capricious. Simply because we can identify something about individuals because they are either fortunate or unfortunate to have their genes identified early in the genomic revolution, we discriminate against those individuals when we should be discriminating against all of us. Once we discriminate against all of us, hopefully then we would discriminate against none of us. So it is discriminatory because it is completely arbitrary.

The other thing we've been told is that the ADA, Paul Miller came and said the argument has been made, former commissioner for the EEOC -- Paul would come and say that one of the arguments is that all of this is covered under the ADA. Once we cover all of us under the ADA, we again cover no one under the ADA, and the ADA was put in place to protect vulnerable individuals in our population. So I am concerned if the ADA was extended. I don't think it would be. I think that happens to be just smoke. But should it be, then it would protect no one and it would not protect the vulnerable.

The other thing, Reed, I just want to point out is that we've been charged as a committee with identifying topics of importance within the agencies of the Department of Health and Human Services, and I think what we've heard today is not only genetic discrimination a problem, and we've had responses from the Secretary saying they understand this and they're supportive of that legislation, but what we've heard is perhaps even more

troubling, and I apologize to our panelists, but I heard the passion that you had for your children.

What we've heard is that this is a barrier to research. If it's a barrier to research, that certainly, since it's health research, falls within the purview of the Secretary. If it's a barrier to research, that means that we are not going to move forward to protect the children of our panelists, to protect our children and grandchildren into the future. In fact, that is even more troubling to me than the immediacy of the discrimination that we've heard about today. We are discriminating in ways that we can't even understand for our children and our grandchildren.

MS. MASNY: Any further questions?

MR. MARGUS: On a different subject, the thing about discrimination is that it sometimes allows people to discriminate without having any kind of standards for the information. So one thing that struck me that I wanted to ask you about is even if you have a genetic mutation or polymorphism that's been associated with some risk, is there really always a consensus about what that risk is?

I'll tell you my quick personal story. As many of you know, I'm a carrier for a mutation that completely wipes out a protein that plays a role in cell cycle control. When you marry someone, as I did, with another mutation like that, we've had two kids with a really serious disease. But along the lines of talking about people like me who are carriers who are supposed to have a three- to four-fold higher risk of cancer, there are actually a couple of New England Journal of Medicine articles that say that's true, and then subsequently there have been numerous other papers done where people challenge that, and there isn't a consensus out there yet.

We always dread the couple of times in our past when we've had insurance companies learn that we're carriers for this disease, are they going to pull out what we think are spurious New England Journal of Medicine articles, or are they going to pull out one of the more recent ones where it looks like the risk is not quite as great? So I'm just wondering, that's another serious downside to genetic discrimination, or even genetic testing where decisions like insurance are going to be made. It's bad enough if they're going to discriminate against you for something that's real, but what if they don't know what they're talking about?

So I don't know if my case is rare or if you run into that, too, but I think it's an important point that the epidemiological data -- I mean, you know how epidemiologists are. They disagree all the time. But in this case it's really important. Do you see that across a lot of your other situations?

DR. BRANTLY: Yes. We see it pretty often in our clinics. Again, the risk is very hard -- I mean, when you speak about risk in general in genetic diseases, is it really spread evenly across the whole population, for instance, of MZ individuals, or is there a small population of MZ individuals who have very, very high risk because of a second genetic hit? All these epidemiology studies have those types of risks. So the risk is X, with caveats. I think that again plays an important role in setting aside what is the risk.

The risk should have nothing to do with your interest, because we can't precisely fix risk for any of these genetic diseases. They're pretty broad confidence intervals, and they clearly are because of either other genes or environment, or both.

MR. HADLEY: I think in that regard we have the same situation in families with hereditary non-polyposis colorectal cancer, where the initial studies said that the

lifetime risk for colon cancer may be as high as 80 percent, but that may not be true for each and every family that we encounter. In fact, some may have significantly less risk than that, but still significantly elevated over the general population. So it is hard to use those numbers at this point in time when we're not exactly sure for the person who is sitting in front of us. But yet still, that information may be pulled and used to discriminate or set insurance rates.

MS. SHAW: I think in the genetics community our crystal ball is about as good as any other medical field. When you're talking with individuals about statistics and trying to apply them to an individual, it's very difficult. There are always variables that can come into play, especially with hereditary cancer predispositions. These are not 100 percent penetrant for the bulk of them. Therefore, when you meet with someone, you say you have an increased risk, but I can't guarantee you will ever get cancer. There are certainly many people that won't, and therefore it makes the genetic discrimination even more troubling because many of these people will not develop what they're at an increased risk for.

MS. MASNY: One more question, Muin.

DR. KHOURY: Actually, I wasn't going to have a question but more of a comment here, because I've been following the discussion and thinking about genetic information at the population level. We always seem to be stumbling as the difference between a genetic disease and the rest of the diseases. As basically the panel showed us this morning, even when you start with a genetic disease where you have, let's say, a mutation that has an abnormal protein product with a high penetrance for disease, it's really never a straightforward risk estimate, except in a few rare instances, whereas in most situations you have an interaction with other genes, and I come in is sort of at the environment at large.

I mean, we've seen it with hemochromatosis, we've seen it with alpha-1 antitrypsin, and we also see it with the carriers, people with various autosomal recessive diseases. But beyond that, to me what's the most troubling, and I guess we all have learned about sort of our lethal variance, that we carry between five and ten, and it's really much more than that because we all have genetic variants of different kinds, like our HLA and our blood groups and our ability to metabolize different things, like carcinogens. So we're all carriers of genetic information, and that information that produces different gene products doesn't have to be abnormal gene products but a variant of a gene product, like a variant HLA system.

I mean, when you think about it, we're all at increased risk of different diseases, even outside the scope of the lethal equivalence that we have learned in traditional genetics for genetic diseases. So I think if we kind of accept the fact that there is genetic variations that are going to put us at risk for various diseases, I think we can solve the puzzle a bit more. As long as we keep talking about genetic diseases, we seem to always come at it from the genetic exceptionalism angle.

Now, I'm not trying to negate or minimize the pain and suffering of people who have the labels of genetic diseases, but I'm trying to elevate that to suggest that we all have genetically driven information that puts us at a different set of diseases, and it's only a subset of those diseases which may be 5 percent of all human ailments, we arrive at the conclusion that this is a genetic disease. It doesn't mean that the 95 percent of all other diseases are not genetic diseases.

So it's a plea towards expanding the scope of genetics beyond the traditional purview of genetic diseases and having to deal with probabilistic information that hopefully will get us beyond discrimination.

DR. COLLINS: Well, very briefly, I can't help but point out that's a very nice connection with the topic that's going to come up tomorrow afternoon, which is the need to have a prospective population-based cohort study that would enable you to get a better fix on what the statistical risks are for particular variants and how they interact with the environment. Case/control studies have been our workhorse for making these various discoveries, but they're not necessarily good at giving you an unbiased assessment of risk, nor of identifying the environmental factors that may serve as important triggers.

If we really are going to get beyond this major barrier of genetic discrimination by passing effective legislation, and I sincerely hope that that will happen, the next step will be to try to implement this sort of individualized risk prediction, and I think we're going to need databases that contain unbiased information of the sort that are difficult to come by from case/control studies but which would derive quite nicely from a large-scale prospective population-based study that also does a very thorough job of collecting environmental exposure data.

MS. MASNY: Thank you. Thank you very much.

We do have to move on to our third panel. Now we're going to hear from this last panel that will be presenting more additional stakeholder perspectives. We'll be hearing from Kathy Hudson, who comes to us from the Center for Genetics and Public Policy here in Washington, D.C. As I mentioned earlier, it is her organization that has held town meetings across the country to discuss genetic issues. We'll hear then from Jane Massey Licata, who is an attorney and professor of law in New Jersey, who will help us to understand some of the gaps of the current legislation and policy that we currently have in the country.

We'll hear from Joanne Armstrong, a physician with Aetna, who will be representing America's Health Insurance Plans. We did invite the United States Chamber of Commerce to participate in the panel. However, they referred us to the Society for Human Resource Management, and we're pleased to have Michael Aitken, the director of government affairs, who is with us today.

Mr. Aitken, before we do begin, I don't know if you were here earlier but there was a question. Are you considered a spokesperson for the Chamber of Commerce?

MR. AITKEN: No, we're not.

MS. MASNY: You're not. Okay, thank you.

We'll begin, then, with Kathy Hudson.

DR. HUDSON: Thank you for inviting me to discuss with you what we've learned about the public's hopes and concerns about advances in genetic testing.

Amanda, are you going to be able to do my slides for me?

My name is Kathy Hudson. I'm the director of the Genetics and Public Policy Center, which is a part of Johns Hopkins University. The Center was funded by a grant from the Pew Charitable Trusts, and our mission is to provide information about genetic technologies and genetic policies to the public, to the press, and to policymakers.

Over the past two years, we have conducted fairly extensive qualitative and quantitative research to understand what the public who does not yet know about their genetic risks thinks about advances in genetic technologies. We've surveyed over 6,000 citizens in two separate surveys, the first in December 2002 with 1,200, a second one this past April with 4,800 citizens. In addition, we've done focus groups, 21 in five cities across the country, and most recently this summer we completed a series of public engagements called "The Genetic

Town Halls: Making Every Voice Count."

I'd like to share with you first results from our survey in which we ask respondents if a genetic test shows that a person has an increased risk for a genetic disease, does -- fill in the blank -- have the right to know, and you can see here that in 2002, 85 percent of those surveyed said that no, an employer should not have access to that information, and 68 percent thought that insurers should not have access to that information. Those numbers went up in 2004 to 92 percent for an employer and 80 percent for an insurer. I would note that if you look at those who have higher education levels or prior awareness of genetic testing, the percentage of those saying no goes up even higher.

We also conducted 21 focus groups across the country, and focus group participants were presented with a series of scenarios involving genetic testing in the reproductive context. In those focus groups and in the scenarios presented, we did not specifically ask about genetic discrimination, but participants spontaneously raised this as a major concern.

Focus group participants went on to speculate about the availability of reproductive genetic testing and if insurers had that information that it may possibly be used to coerce or influence their reproductive choices.

You can move the slides ahead, and one more.

One of the concerns about survey work in particular is that you're asking people for their off-the-cuff reaction to a question without having a lot of time to learn about it, to think about it, to ruminate on its possible implications. That's also true in focus groups. So what we did was we adopted a model of deliberative democracy which has been used to explore other central policy issues, although not in science policy previously, and tried to develop a program to find out what Americans think once they've had an opportunity to learn a little bit about genetic technologies, the issues they raise, and have an opportunity to talk with fellow citizens about these issues.

The next slide.

So this summer we did six cities in six weeks, town halls involving over 500 citizens, and we also did online town halls where citizens from across the country were engaged in discussions over a three-week period with other citizens through the miracle of Internet technology. Participants in both our online and in-person town halls were asked to consider three major issues in reproductive genetic testing: acceptable uses, safety and accuracy, and the impact on individual families and society. We provided background information about the technology, as well as a broad range of views from experts in medicine, policy, bioethics, and theology.

Participants were queried at various times during the town halls about their optimism and concern about these technologies. Two of the questions that were posed to the town hall participants were what do you think are the factors that should be considered in setting limits for the use of reproductive genetic testing, and what are some of the possible benefits and harms for individuals, families, and societies?

Participants were given an opportunities to talk with their table mates about these issues and then called out the issue of most concern to them. If a concern had already been called out by another table, that table was asked to move to the next concern on their list. Once all the concerns were expressed, they were projected up onto a screen and the entire group had an opportunity to talk about those concerns, and then with electronic keypads to vote

on those that were of most concern to them.

If you could put up the next slide.

That shows our people across America talking to one another about genetics.

Next slide, please.

The issue of genetic discrimination based on genetic test results came up as an issue in every single town hall. In fact, in Sacramento and New York, genetic discrimination ranked as the number one issue of concern when considering potential harm from reproductive genetic testing for individuals and families. In Seattle, Fort Worth, and Nashville, it ranked as the second concern. In Sacramento, every table listed it as a concern and said, as one participant did in Sacramento, will you have trouble getting a job because you have this gene that may cause cancer, whether or not you have cancer?

Like the focus group participants, the town hall participants feared that insurance coverage would be a factor in guiding reproductive choices.

The next slide.

So in conclusion, our research shows that an overwhelming majority of Americans do not want insurers or employers to have access to genetic test results, and that there's widespread concern. This concern is first and foremost on average Americans' minds. This high level of public concern makes it important to think about what we will do when, not if, we enact legislative protections, and this gets to Brad's point, that I think we will have to put some focus on making sure that we get public information out to both providers and to patients to let them know what their rights are so that they don't let concerns about newly unlawful practices influence their genetic testing decisionmaking.

Thank you.

MS. MASNY: Thank you, Dr. Hudson.

Next we'll hear from Jane Massey Licata.

DR. LICATA: Again, thank you very much for inviting me here to testify today. I've come at this issue over the past decade from a number of different perspectives. I'm a biotechnology patent and FDA lawyer, and I also teach at Rutgers School of Law in Camden, both patent and FDA law. I was trained as a scientist with a background in biology and biochemistry, and I hold a doctorate that I received in 1978. So that was back when we didn't know that much about genes. But over the past two decades I've been blessed to learn a lot about molecular biology and genetics from some of the greatest scientific minds in our country and the world.

My interest in genetic privacy is both academic and practical. I've served on and advised institutional review boards on genetic and biotechnology research issues. I've evaluated genetic technology for venture capital firms and pharmaceutical companies. I filed over 2,000 patent applications concerning biotechnology inventions, most involving the use of genetic information to develop new diagnostics and therapeutics. I've negotiated hundreds of research agreements concerning genetic research. So I've worked in both the non-profit and the private sector.

I come at this from a practical sense in that one message I've taken from all of my constituents is that there is a need for some certainty in the marketplace and in the academic research community as to how we're going to deal with both the ethics and the dissemination of this information. Now, I've been very fortunate, I guess, in that I've been

asked twice to testify before congressional committees on the legislation. I started working on this issue as an academic in the early '90s, and the first time I testified before a congressional committee was in September of 2001, and at that time there was quite a bit of hope that the legislation was going to be passed.

I recently testified again before a congressional committee in July of 2004 in an effort to try to encourage the House to follow suit on the compromise bill that had been passed by the Senate. There's been a lot of progress in definition in the issues that are pending and are being considered by Congress. There are three specific issues, after listening to the testimony today, that I would like to address because in each of the cases where I testified before Congress as an objective basically friend of Congress with a technical and legal background, a gentleman from the Chamber of Commerce was sitting on my left, and I think there are three issues that have been raised that I think are important to address.

The first is there seems to be a misperception in some parts, not on the part of Congress in general or the council to the Congress, because these people are incredibly knowledgeable and have, in fact, analyzed really where we stand with respect to all of our federal laws and genetic nondiscrimination. But basically, there is really very little federal protection for genetic information. We've heard discussion today about HIPAA, the Health Insurance Portability and Accountability Act, that was passed in 1996. HIPAA does some very good and important things.

It does prohibit group plans from using any health status-related factor, including genetic information, as a basis for denying or limiting eligibility for coverage, or for charging an individual more for coverage. So in the group health setting, not all group health insurance plans, as was so articulately explained by one of the other panelists, but in some group situations, it is helpful.

It does limit exclusions from group plans for preexisting conditions for 12 months, and prohibited exclusions for people who had been covered previously for a condition for 12 months or more, and specifically states that genetic information, in the absence of a current diagnosis, does not constitute a preexisting condition. So a good first step.

But HIPAA does not prevent insurers from collecting genetic information or limit the disclosure of genetic information about individuals to insurers. It does not prevent insurers from requiring applicants to undergo genetic testing, and it doesn't cover a lot of Americans because it doesn't apply to the individual market or many group plans that are exempt.

Another federal law that we've heard mentioned is the Americans with Disabilities Act, the ADA. Now, the ADA does protect individuals with symptomatic genetic disabilities. It does allow an employer to obtain extensive medical information about a person that is under a conditional offer of employment, including obtaining and storing genetic samples, requiring genetic screening as a condition of employment, to purchase genetic information about applicants from a genetic information databank, and once employed, the employer can request medical information that is job-related and consistent with business necessity. So there's a positive and a negative here in what's allowed under the ADA.

But the ADA does not explicitly address genetic information or deal with unaffected carriers of a disease who may never get the disease themselves, individuals with late-onset genetic disorders who may be identified through genetic testing as being at risk of developing a disease, or others identified through family history as being at high risk for

developing the disease. Those people just aren't covered. It does not protect workers from requirements or requests to provide genetic information to their employers.

Another federal law that's been mentioned is Title VII of the Civil Rights Act. Now, this is the stuff that law professors love. It does provide a basis for an argument that genetic discrimination based on racially or ethnically linked genetic diseases constitutes unlawful race or ethnicity discrimination, but it's an argument, and there's not a lot of cases where there's going to be that link, where it's going to be possible to establish that that link has a direct relationship to race or ethnicity. So although it's the stuff that we like to talk about in law school, on a practical level I don't think it's something that the American people can rely on to protect them.

Now, another issue that I've heard mentioned from the business community is a concern that if we do pass this legislation and we do have federal law and create a new right of action, that this is going to create all of this new litigation and this is going to be a huge problem for employers and health insurance companies, a huge cost to the community. I respectfully disagree with that for a couple of different reasons.

One is there's been an evolution. When I first started working with this legislation, there was in fact a private right of action in there for the individual who felt that they had experienced genetic discrimination. But even then, all that they were going to get was some reasonable attorneys fees, including costs of expert witnesses, in cases in which a plan sponsor, health insurer or any third party acting on behalf of the plan or insurer violated the law, and the civil penalties would not exceed $50,000 for a first violation or $100,000 for any subsequent violation. So it was always a fairly limited right of action, and to be fair I think it's been pointed out by the panelists that it's something that's not likely to happen. Folks aren't really so concerned about bringing a private right of action. It's a huge emotional and financial investment.

What they're concerned about is keeping the health insurance and keeping their job, and this is what the Senate compromise did. It listened to the concerns of business and basically it established enforcement for violations of the provisions, but in a way the business can deal with, because what they said is this is what it's going to cost you for not complying, this is what it's going to cost you if you don't respect this right and you don't figure out a way to build it into the way that you do business, like lots of other things that you do on a day to day basis.

Basically, the penalties are $100 per day for each day the group health plan is in violation, payable to the participant or the beneficiary, with a minimum fine of $2,500 or a maximum of $15,000, and even if you go the whole way and there's found to be a willful violation, you're talking about a cap of a half a million dollars. That's what you pay your lawyers to figure this out. I mean, let's be practical. The businesses that I deal with, they want to know what am I supposed to do to comply. This is what the health systems ask. Everybody figured out how to deal with HIPAA, and they found out how to do it in a cost-effective, respectful way because they were told you have to do it.

If we apply it across the board to all insurance plans, I really can't see, when you work out the economics, particularly when the penalties are encouraging compliance but not outrageous, there's not this huge specter of risk and cost that was originally suggested. It's simply not true. It's not in the compromise bill, and I think it's a respectful way to try to balance the concerns.

One thing that the compromise bill does that's absolutely awesome for the individual is that it allows them to keep their health insurance while this is getting worked out, and that's the big issue that people were concerned about. They don't want to lose their health insurance. So while this process is going on, they're not sitting in a gap wondering what's going to happen to me, what's going to happen to my family. What's been built into the compromise legislation is an assurance to the individual that they will be able to have their insurance protected while this issue is being evaluated, and I think that that's also very good for all parties. It's a fairly minimal cost to the business community, and it's a huge benefit to the worker.

Now, another issue that I've heard as an objection to this legislation is we don't need it because the states all pass these laws anyway, so let's just leave it to the states. Now, I have great respect for home rule. I'm from the State of New Jersey, and we're really big on that there. So far, 41 states have enacted legislation related to genetic discrimination in health insurance, and 31 states have adopted laws regarding genetic discrimination in the workplace. So the states have actually been very active and creative, but it's a patchwork, a patchwork of laws.

For example, in the State of Florida, it doesn't concern health insurance at all. The law that was passed in 1978 prohibits any person, firm, corporation, unincorporated association, state agency, unit of local government, or any public or private entity from denying or refusing employment to any person or to discharge any person from employment based on sickle cell.

That is the law in Florida. Here, Florida was one of the earliest ones to act, but that's the only coverage there is in the State of Florida for genetic discrimination under state law.

Now, in New Jersey, we actually enacted in 1981 and then brought in the law in 1996, and we actually have one of the toughest genetic privacy statutes in the nation. Under my state basically says, it covers situations of health insurance, life insurance, and employment. So it covers all of the issues, which is not the case, it's a very rare thing to have all of those covered. Basically, the problem is that these issues, as we've talked about, cross state boundaries and affect all of our citizens.

So if you're lucky enough to live in New Jersey and you have a problem in New Jersey, you're going to have a law that's going to say that genetic information is personal information that should not be collected, retained, or disclosed without the individual's authorization. The act prohibits discrimination by employers against employees carrying genetic markers of diseases or behavioral traits. It's unlawful for an employer to refuse to hire or employ or to discharge or require to retire an employee because of the employee's genetic information or a typical hereditary cellular or blood trait, or because the employee refused to submit to a genetic test or make available the results of a genetic test to an employer.

It also prohibits the use of genetic information in the fixing of rates or withholding of life insurance. It bans the use of genetic information to establish the amount of insurance premiums, policy fees or rates charged for health insurance contracts, whether it's an individual or group plan. The penalties for violation of the provisions in the act include fines and prison terms. Actual damages, including economic, bodily or emotional harm proximately caused may also be recovered.

MS. MASNY: You have one minute more.

DR. LICATA: Under New Jersey, you have a very strong law. Now, no case has ever been brought under this law, but people doing business in New Jersey understand what the rules are, and what they have done is they have managed the risk and they have built into their way of doing business, their way of making insurance decisions, their way of making workplace decisions consistent with the law because they know what the rules are.

So what I'm suggesting is that we need predictability. People need to understand where they stand. If we have at least a threshold level in this country, I think it would be extremely helpful to all of our citizens.

MS. MASNY: Thank you very much.

Next we're going to hear from Joanne Armstrong.

DR. ARMSTRONG: Thank you for inviting me to testify regarding this important issue. My name is Joanne Armstrong. I'm a senior medical director for Aetna, and I am testifying today on behalf of America's Health Insurance Plans and its nearly 1,300 member companies.

America's Health Insurance Plans, or AHIP, is a national trade association representing the private sector in health care. AHIP's member companies provide services for over 200 million Americans. Aetna serves approximately 14 million health care members through a national network of about 500,000 service providers, including laboratories, including over 300,000 primary care physicians and specialist physicians, and over 3,000 hospitals in its national network.

Genetic medicine is not new. DNA and non-DNA-based genetic testing has been in wide clinical use for many decades. Examples range from carrier type testing in reproductive health and pediatrics through serum testing for abnormal forms of hemoglobin and others. A promising new area of genetics utilizes genetic test results to guide the choice or the duration of pharmacogenetic therapies. These so-called selected or targeted therapies hold out the promise of directing medications to individuals who may benefit from them, while avoiding side effects and costs in others.

Examples are found in breast cancer and colon cancer treatment already in terms of duration of therapy. We know that pharmacogenetic principles are applied in hepatitis C management.

While genetic testing itself is not new, the rate of new genetic discoveries entering clinical practice is increasing at a dizzying rate. The speed of these new discoveries is challenging our health care system's ability to effectively integrate them into clinical practice and to optimize their benefits to prevent and to possibly cure disease.

Because of the complexity of genetic information, the optimal use of genetic technologies requires informed providers, informed members, and coordination of services across this complex array of delivery systems. Unfortunately, I think we know that much work needs to be done in all of these areas to get it right. For example, we know that 72 percent of physicians are not completely prepared; 72 percent of non-genetics physicians rate their knowledge of genetics as fair to poor, and we've heard some testimony about that. Patients are also not adequately prepared to navigate these waters. Fully 82 percent of consumers cannot correctly answer most genetic medicine knowledge questions in national surveys.

So as with the adoption of other medical services and technologies, health plans are and will continue to be instrumental in the coordination of this care. This process in

genetics has already started, and some of the benefits have already accrued. For example, health plans have demonstrated success in improving patient compliance in a number of pharmacogenetic areas, including hepatitis C management.

As the science of genetics advances, concerns over protecting genetic information from inappropriate uses have escalated. There is growing public awareness about the health benefits that can be derived from genetic information and concerns about the potential misuse of this information. We must, however, engage in responsible policymaking on these issues and not unnecessarily restrict the use of genetic information needed to promote appropriate health care decisionmaking and assist in the coordination of this care, which is already quite fractured.

I'd like to address my remarks today largely to the current use of genetic information by health insurance plans to give the panel an understanding of how it's used today, and also to briefly address some of the issues regarding the laws to protect this information.

So the first question is how do health plans currently use genetic information? Genetic information is just one of many types of medical information that is currently used by plans in all sorts of activities, including the promotion of risk assessment for its members, preventive screening efforts, disease management, pharmacy compliance management programs, quality assurance management programs, and the larger umbrella of just the coordination of these cares across this very complex array of delivery systems.

A key component of health care delivery is to make sure that patients and health care providers have the information they need to make informed decisions, and health plans are facilitating this exchange of information in genetics to encourage appropriate counseling and testing and decisionmaking that follows that. For example, health plans have had for a long time a very active role in promoting genetic counseling in the reproductive health arena to ensure the highest quality of information is provided to members to make the best decisions for themselves. We know that when genetic counseling is provided by formally trained genetic counselors, the amount of actionable and higher quality information that comes out of it is much higher than when it's delivered by non-genetics-trained clinicians. We know when this takes place, patients and members get better results.

As an early adopter of coverage for BRCA breast and ovarian cancer genetic susceptibility testing, Aetna incorporated genetic counseling services into the testing process to ensure that medical appropriateness of testing took place and to ensure that at-risk individuals receive accurate information to support their subsequent decisionmaking. Health plans also use genetic information to help enhance preventive screening and health promotion efforts for individuals who have a disease. Genetic data is used by plans to create deviations in standard coverage benefit packages to enhance the types of services that members get compared to what is available for the general population.

For example, screening tests that are available for hereditary non-polyposis colon cancer for affected individuals, they require earlier and increased frequency of screening. Access to this genetic information allows health plans to create deviations in their coverage processes to provide those services to these members. So obviously, this is an added value.

Similarly, for patients who are BRCA positive, these women need increased, more frequent screening at an earlier time and through different technologies than that which is recommended for the population at large. In order to administer these benefits to make it happen and deliver it across this complex system, this type of data sharing takes place.

Already discussed, obviously, in this meeting, genetic data is multigenerational and in some instances requires new testing paradigms from a health plan perspective to assess risk and deliver the most appropriate services to patients in the best possible way. The most efficient BRCA screening scenario for at-risk individuals may in fact involve the testing of an affected family member who is herself not a covered member. Aetna has led the industry in extending, on a voluntary basis, coverage benefits to non-covered members of the plan if those test results actually help the member of our plan. Again, to administer these types of benefits, data sharing is necessary.

Then on a very practical level, physicians and members call health plans on a daily basis, dozens and hundreds of times a day, with very specific questions about where can I get this genetic test done, what is my contract allow, how do I coordinate these services. So these are very basic operational issues where genetic information is shared on a daily basis.

I should also add that when claims are submitted, these claims come in with specific genetic markers that are on them, and that is a necessary part of getting these services paid for across a very complex health care system.

Finally, as scientists acquire a greater understanding of the role genes play in all disease states, especially chronic diseases, genetic information will be incorporated into disease management programs and pharmacy management programs. It sort of speaks to the comment that was made earlier that genetic information will be part of standard medical practice as we understand its contributions to chronic hypertension and other disease states.

There is every reason to believe that this will lead to improvements in health outcomes beyond that which have already been demonstrated and disease management and pharmacy management programs that take place within health plans today. Again, data sharing will be important to the success of these efforts.

Lastly, I would just like to briefly discuss some of the issues related to the inappropriate use of genetic information. AHIP believes that the importance of protecting genetic information from illegal and inappropriate use if critical. One of the unfortunate myths about genetic information is that health plans use this information to deny insurance at a global level or to disclose genetic information inappropriately. In fact, health plan companies have many years, decades now, of experience in sharing and using genetic information for their members with little empirical evidence that it is being misused.

As a matter of practice, health insurance plans do not use or disclose personal health information for purposes outside their necessary insurance coverage activity, and federal and state laws currently in place do provide some protection. As early as 2002, Aetna recognized the heightened sensitivity of genetic information and incorporated confidentiality protections into the everyday use of genetic information within our plan.

So protecting the confidentiality of all health information, including genetic information, is critical to preserving the open and honest communication between physicians, clinicians, and their patients. We also believe that consumers should be able to both benefit from coordinated and integrated health care delivery systems while being protected against unlawful disclosures of genetic information.

So in conclusion, AHIP and its member companies believe that genetic information can help providers and their patients make informed health care decisions. Health insurance plans have an important role to play in promoting the appropriate use of genetic tests by encouraging evidence-based counseling and testing and supporting consumer education and

patient awareness, and using genetic test results to enhance preventive screening and disease management. For many decades, health insurance companies have demonstrated responsible use and management of their insureds genetic data.

Health insurance plans strongly support protecting all patient identifiable health information, including genetic information, from unauthorized disclosures and other illegal uses, and as the science of genetics advances, we are committed to facilitating access to genetic services and guarding against misuse of this data.

Thank you.

MS. MASNY: Thank you, Dr. Armstrong.

Our last presenter is Mr. Michael Aitken.

MR. AITKEN: Good afternoon. My name is Mike Aitken. I'm the director of governmental affairs for the Society for Human Resource Management, and I appreciate the opportunity to provide commentary to the committee regarding genetic discrimination in the employment context.

I appear today on behalf of SHRM, which is the world's largest association devoted to human resource management. We represent more than 190,000 individual members, and our mission is to serve the need of the HR profession by providing the most essential and comprehensive resources available. SHRM believes that employment decisions should be based on an individual's qualifications and ability to perform a job, not on the basis of characteristics that have no bearing on job performance. Therefore, SHRM strongly opposed employment discrimination on the basis of an individual's genetic information.

The Society also believes, however, that any legislative remedy proposed must be carefully drafted so as not to be overly broad, thereby leading to unintended consequences with existing federal and state employment and benefits laws, as well as existing nondiscriminatory employer practices. In my commentary today I'll try to discuss the interplay with the proposed legislation that's been advanced previously may have on current federal and state laws, as well as existing nondiscriminatory employer practices.

Despite the fact that there hasn't been strong evidence to suggest widespread use of genetic information by employers, there is interest in enacting legislation that would codify current protections, as well as to fill the gaps left unaddressed by current law. Under the current federal framework, there are several statutes that could potentially -- and I stress that in particular with what Jane was talking about -- provide protection against genetic discrimination. However, these laws remain largely untested in the courts in this area.

For example, given that some genetic diseases have been found to be more prevalent in certain racial and ethnic groups, Title VII of the 1964 Civil Rights Act may serve to prohibit genetic discrimination against members of these groups. To date, at least one court case supports employment discrimination claims based on genetic information under Title VII. This was the U.S. Court of Appeals in the 9th Circuit held in Norman-Bloodsaw v. Lawrence Berkeley Laboratory that mandatory pre-employment genetic testing performed without consent may amount to an adverse impact under Title VII, since the claimants were tested for genetic markers based on their protected status.

Although it does not explicitly address the genetics issue, another federal statute that many argue offers protection against genetic discrimination is the Americans with Disabilities Act, or ADA. According to the EEOC interpretation for the 1995 guidance on disability, genetic discrimination is prohibited under the third part of the statutory definition of

the term "disability," which protects individuals who are regarded as having impairments that substantially limit one or more major life activities. This prong of the ADA reflects recognition by Congress that the reactions of others to impairment or perceived impairment should be prohibited in the same way as discrimination based on actual impairment.

In fact, the EEOC in 2001 filed a genetic discrimination suit against Burlington Northern Santa Fe Railroad in its genetic testing of employees who were filing claims for work-related carpal tunnel syndrome. Although the case was not decided on the application of the ADA to a genetic issue, the suit was quickly settled.

Should a new federal discrimination law be enacted, it is essential that it is developed to reflect the requirements and protections of existing employment statutes and that it is not in conflict with current laws or that it makes illegal existing nondiscriminatory employment practices. Let me briefly just touch on a couple of potentially challenging areas in current law, as well as employment practices where the use of medical and potentially genetic information is present in the workplace.

Under the ADA, medical records may be used to help determine if an employee has an impairment that substantially limits one or more major life activities or has a record of such a substantial limiting impairment. Moreover, medical information is often an integral part of determining a reasonable accommodation of a disabled employee. Since employers are required to determine whether or not an employee or an applicant has a disability within the meaning of the law, the employer of the applicant's medical information is often required. HR professionals and employers would face an insurmountable challenge in making proper decisions without this information.

The Family and Medical Leave Act creates a similar challenge. As you probably know, the FMLA allows an employee to take up to 12 weeks of unpaid leave for their own serious health condition or the serious health condition of a family member. In order for an employer to determine whether an employee qualifies for FMLA leave, that is whether the serious health condition is manifested by the employee him or herself, or the family member, the employer must collect relevant medical information on the nature of the condition. The medical information may very well indicate a genetic-based health condition.

For example, and many of you have probably heard of this before, an employee may request intermittent leave to assist her ailing mother who is receiving radiation treatment for a diagnosed breast cancer, a serious health condition under the Family and Medical Leave Act, and a disease with a known genetic component. In granting the leave request, the employer has just acquired genetic information. The interplay of legislation in various state worker's compensation laws will create more challenges for employers. Under state worker comp laws, medical information is necessary to file a claim and is used to determine whether or not the injury is work related.

In 1996, Congress addressed the issue of genetic information for group health insurance in the Health Insurance Portability and Accountability Act, or HIPAA. HIPAA currently permits a group health plan to disclose health information to an employer that sponsors a plan provided the information is only used for plan administrative purposes and the employer has put in place certain specified safeguards on medical privacy on its disclosures.

Employer-sponsored wellness programs is another instance where employers may uncover genetic information. Establishing a wellness program often involves a confidential individualized health risk assessment for the employee. However, in conducting

the risk assessment, information may be collected that would include family history, blood test results, and other potential genetic information.

Similar to that law, an employer may also inadvertently acquire potential genetic information through the water cooler scenario. For example, it's not uncommon for colleagues to share personal information about the health status of their family members with each other in the workplace. Proposals that include an overly broad definition of genetic information could turn that casual conversation about loved ones around the water cooler into a litany of costly litigation and workplace disputes.

In each of these instances, it's not the employer's intent to seek out the potential genetic information of the employees. Nevertheless, an employer that simply possesses this information, whether or not the employer ever acts on the information, could be exposed to future liability if legislative proposals to prohibit genetic discrimination focus on only controlling the information and not on the discriminatory intent of the employer.

As a result, SHRM makes the following recommendations to public policy decisionmakers considering crafting a legislative remedy. First, legislative proposals should differentiate between the mere possession of genetic information and the use of the information for discriminatory purposes. Any proposed statute should be directed at controlling discriminatory conduct rather than attempting to regulate the flow of information.

Second, we believe that genetic discrimination is wrong and if a company intentionally discriminates, remedies should be available. However, SHRM opposes legislation that would provide unlimited punitive and compensatory damages for victims of genetic discrimination. Many of the earlier versions of proposals that were out there did exactly that.

Third, legislative proposals should not impede employer efforts to protect the safety and well-being of their employees through workplace wellness programs and other services currently available under state and federal laws.

Fourth, duplicative efforts to guard against genetic discrimination are costly and confusing. Any legislative proposal regarding genetic discrimination should take into account the protections already available under federal and state laws.

With that, I would like to thank the committee again for the opportunity to appear before you today and will be pleased to answer any questions you may have.

MS. MASNY: Thank you, Mr. Aitken.

We are going to open it again for questions, 10 minutes, and then we will proceed with a larger roundtable discussion.

So any questions for the panel? Yes, Dr. Collins?

DR. COLLINS: So I'd like to ask Mr. Aitken, a number of the issues that you raised are certainly ones that have been discussed for several years as various versions of legislation have been debated, drafted, redrafted, and so on. I frankly think a number of the points you made have already been taken care of.

So specifically, what would your recommendation be, starting with S. 1053 as a template which, after all, did pass the Senate 95 to nothing, what additional changes would need to be made in that bill for SHRM and presumably the Chamber of Commerce to be comfortable with it? Can you be explicit about that? Because the general sense I think many of us have had is that there's a cloud of objections without really getting down to the kind of specificity that would imply an intent to work something out.

MR. AITKEN: Well, if I may, Dr. Collins, the employer community has

worked with the Senate and both Republicans and Democrats, as well as in the House, as well as with other stakeholders, and have been fairly consistent in where we've raised our concerns. I would concur with you, I think the Senate did a very good job in trying to meet a lot of the issues that have been raised by the employer community with regard to that.

There have been some continuing lingering concerns with the genetics bill, and we've seen this -- particularly a state law, and Jane mentioned, as well as Joanne did, about some of the state law efforts. One of the concerns with S. 1053 is that there isn't a sunset provision, and that is because we've seen -- our experience has been with the 30-some state laws that have been enacted that the states have had to go back and revisit those statutes because they found problems with how they've interacted either with the health care plans or with the employer state laws, and it's caused problems and conflicts.

One of the things that's been advocated is a sunset provision to give an opportunity to review the implications, and as genetic advances are made and more discoveries are made, that there may be things that need to be done to modify that legislation to look at that issue.

Second, there's no federal preemption in S. 1053, meaning that the federal law does not trump the various different state laws that are out there, and there is a concern that you could have situations not just within the employment side but also with regards to the health care plans where plans might be complying not only with the federal statute in addition to the state statute and have conflicting laws with regards to that. That's another aspect that's been raised.

It's certainly not my area of expertise, but in talking with others, there's the definition of "family member." Many have felt that it's an overly broad term as described within the setup bill, although it's better than earlier versions. But there is a concern that it's not narrow enough. I understand there are different trains of thought out there within the geneticist population, but those are some of the main concerns that have been raised.

I will say that we have raised these concerns with supporters of 1053 and we've asked them to try and go back and address those, and as yet we've not heard back. EEOC, in fact, shepherded through an informal meeting with stakeholders on all sides back in April, and it was specifically asked of the others to come back and try to address some of those issues, and we're still waiting. The employer community has not put its foot down and tried to destroy the genetic discrimination bill. We've worked very closely with all on this.

DR. COLLINS: So can I then ask you directly, is it a goal of the SHRM to see effective federal legislation pass to prevent genetic discrimination in the workplace?

MR. AITKEN: I think that we would certainly be supportive of that. The concern becomes -- you remember who we represent, as well. We're giving our employers another cause of action to have to deal with, and you're never going to have employer groups standing around the table supporting that type of effort. I think, frankly, that the least that you would see is that we wouldn't oppose that effort, and we didn't oppose S. 1053. We continue to work with the Senate in crafting that in that effort.

MS. MASNY: I have Cindy, and then Ed.

MS. BERRY: Perhaps Dr. Licata can help with this. A point of clarification, because I was wondering about the plans that are apparently exempt from HIPAA. I was wondering if you could help us clarify that. Are ERISA plans subject to HIPAA, or are they not? I thought they were but have heard that perhaps some may fall under some sort of

exemption.

DR. LICATA: Right. There is an exemption, and it's basically these smaller self-insured plans that fall out. I think this is kind of relevant to understanding what ERISA does, because ERISA does have some federal preemption. In addressing the issue of this concern about having to deal with lots of different state rules or different rules even within federal guidelines, what is intended by S. 1053 is it creates a threshold. So basically, if you have a state law that is more strict, like New Jersey, those are enforceable. But everybody has to come up to at least one consistent national level, and what that national level is is that these same standards that are currently applied would generally now apply to all individual plans and to all group plans.

The thought is that if you're covered by ERISA, once you're in the federal legislation, you do have preemption, and this is one of the issues that was discussed before Congress as well. Another aspect of this is understanding who is covered and who is not covered. There was a great debate about the concept of the family members and so forth, and the compromise that was made in the Senate there was to focus on a definition of the use of genetic tests instead of getting all hung up again on this definition of genetic information, and also to include people that might not necessarily have a blood relationship because they're covered by the plan, such as the gentleman was explaining with his adopted children.

When a claim gets made against a plan, against a particular member's plan, then you have to go into the explanation of whether this is a relevant risk to this person or not. So the burden has now been shifted. That's part of the compromise to protect the individual member or beneficiary so that they can maintain their insurance and not feel threatened while that's being sorted out and that there's not a limitation on the blood relationship there.

I think everybody took very seriously the concerns of the employer and was trying to make it simpler and to address these concerns by saying we're going to do some risk shifting here. We're going to be concerned about protecting the individual's rights, but at the same time we're going to give more certainty to the business in knowing that they're going to apply to a generalized federal standard that is a threshold, and it will call now all group plans in and all individual plans in.

DR. McCABE: Thank you, and thank you for appearing before us. First I have a comment and then a question.

I just want to follow up on Agnes' question to Mr. Aitken, where he said that he was not a spokesperson for the National Chambers of Commerce. I would just like to point out, then, that the National Chambers of Commerce basically gave this committee a response of no comment when we asked them to appear before us, and any of us who have had media training know that no comment implies a concern about guilt.

(Laughter.)

DR. McCABE: So I would just like to point out that I'm not judging the National Chambers of Commerce, but they did respond no comment to us. Perhaps they will wish to continue that in the future, perhaps not. That will be their choice.

I would be insulted, by the way, except for my interpretation of their response.

The other is a question for Mr. Aitken. First, perhaps you weren't in the audience this morning for the initial panel. If not, then I would encourage you to read the testimony of those individuals who appeared before us in the initial panel today, because it was

really quite powerful.

You said that there was no evidence of widespread use of genetic information in employment discrimination, and yet in your presentation you raised a number of examples of potential for genetic discrimination, like the water cooler scenario and those sorts of things. You also said, or I have a question. If there's no evidence of genetic discrimination, then why is there the fear of widespread litigation around an issue which doesn't exist? Your testimony seems somewhat internally inconsistent to me, and I'd like you to clarify it for me.

MR. AITKEN: Sure. I think there's a difference between the actuality of cases being brought and fear, and I did not hear the first panel. But even hearing my colleagues on the second panel talk about was the fear of the discrimination. I do not discount that, nor does the SHRM. We certainly heard that, but we're not hearing from employers even wanting to have this information. I know that the EEOC has had very few cases brought under the ADA in terms of a discriminatory content, and I don't have the exact figures, but I know on the 30-some state laws that are out there, there have been very few cases that have actually been brought.

I guess our concern about the litigation has to go back to the Title VII, the Americans with Disabilities Act, the Family and Medical Leave Act, and the other litany of employment legislation that employers are required to comply with right now. What we've seen sometimes is even the laws that are well intended, we don't take exception to what the intent of folks' efforts have been to try and craft legislation to prohibit genetic discrimination. But our HR professionals deal in the land of Murphy's Law and the law of unintended consequences. Often what happens is it's not the employees -- the employers get faced with frivolous lawsuits. If a law is not carefully crafted to address some of these issues that we've raised, they end up in court. The burden shifts to the employer to prove that just because they had the knowledge that somebody may have a genetic marker, that they in fact discriminated against that individual.

What we've been concerned about is most of the earlier versions of the legislation focused principally on trying to control the information, and our concern is that it's already out there. It's already been required to be out there for compliance with a variety of different federal and state laws. If discriminatory content, that's something entirely different. If somebody is discriminating on this, on genetics, then that's a different issue, as opposed to the information itself.

MS. MASNY: Thank you.

We do have to move on because we're already behind on our roundtable discussion. So I have to move us on to that because we have only until 1:15 for our particular discussion.

So what we actually need to do as a committee now is to look at where do we want to go, what steps do we want to take after having heard some of this information. We have heard already some particulars that have been brought up, some particular recommendations, one being that we have another letter from our new chair to the Secretary describing some of the information that we've heard today, broadening the aspect of discrimination.

The second is, again, do we want to specifically invite again the Chamber of Commerce to address some of the questions that our committee has?

Third is to make a specific request to the Department of Justice to review

some of the gaps in the legislation as we currently have as to whether we need to go any further with this. Would new legislation then cover those gaps?

The other that was actually one of the purposes of this particular committee was to look not only at the current genetic tests and genetic diseases as we know them today, but the broader aspect of genetics and genomics as to where we're going to be going in terms of research and medical services. So I think that's another area to look at.

We'll begin first with Debra, and then Reed.

DR. LEONARD: I think from the presentations today, it's clear that fear of genetic discrimination is real and it's prevalent. It is not rare. This is fear of loss of insurance as well as employment, and not only for themselves but down to the next generations. Real discrimination probably has occurred but may not be realized to the full extent because of this fear. People are not having the testing, so there may not be as much actual discrimination occurring as might occur if people were having the testing.

This fear is compromising medical care, diagnosis, defining risk, treatment after testing, and realizing possible benefits for other family members. It's also compromising research participation, and it's most disturbingly compromising life and death decisions by patients and people. Health insurers look like they are supporting laws to protect individuals from genetic discrimination, and I guess employers are not going to impede laws for genetic discrimination, but I think it's also clear from Jane's presentation that federal laws protecting against genetic discrimination do not exist, not that provide adequate protection. So I don't know that asking for a reiteration of that from the Department of Justice is really worthwhile.

SACGHS has already agreed that legislation is needed. We've sent two letters to the Secretary, and so far the House is not moving forward. I don't know if they are deliberating on, but they are certainly not passing legislation. I am loathe to continue our previous pattern of sending letters to the Secretary since this appears to have had little impact on achieving passage of genetic discrimination protection laws.

So my question is what else can we do? Can we send information from this public forum to members of the House or to the Secretary to ask him to pass on? I guess the only thing we can do is address the Secretary. Can we request members of the House to address SACGHS, chairs of the committees that are deliberating this process? Is there anything else we can do other than simply sending letters? Because that has not been very effective, in my opinion, to date.

DR. TUCKSON: Let's just get a quick response in terms of some of the technical issues there. On the first part of your comment, we are not allowed -- I'll get corrected, I'm sure, here in a second, but we're not allowed to directly write to the Congress. What we can do is to ask the Secretary to take our information and send that to Congress. So instead of sending the letter to the Secretary, you can ask the Secretary to send stuff on our behalf to Congress.

As regards inviting members of Congress to come here and be a part of a discussion, that's one I'm not sure about.

MS. CARR: Well, you know, we have invited congressional staff in the past, and I don't think there's a prohibition in inviting a member of Congress to come and talk to us. We could certainly check with the Secretary's office about that and convey a letter of invitation.

DR. McCABE: My point in asking the Secretary to communicate with the

Justice was to broaden the discussion, perhaps, within the administrative branch. So that was really the intent, because I agree with you, it's been very frustrating the efforts that we've made and have not moved forward. So that would be also, perhaps, when I asked Vahid Majidi the question before, and I apologize for not providing some pre-question information to him, but that was really the intent. So perhaps we could discuss are there other things that we could do to increase the visibility of this issue within the administration?

The President is on record as being supportive of this, both before he became President and during his presidency. Secretary Thompson similarly is on record, both as a governor and now as a Secretary, being supportive of this. Yet there has been no movement. Part of my reason to challenge the Chamber of Commerce to come before us is that the scuttlebutt that I have heard is that the Chamber has been very active in holding this at the desk, that they are really the people -- and I appreciate Mr. Aitken being sent to represent that segment of our economy, when the people sending him didn't have the guts to come before us themselves.

So I appreciate your willingness to do that, Mr. Aitken. I think you should take back the message, though, that we see what was done. We're not stupid, and you were sent here so that we could not question the people who are holding this at the desk. That's what we have to do is figure out how we can mobilize all of what we've heard today. Dr. Hudson said that this is a major issue throughout the United States. How do we mobilize the people of this country to counteract the Chambers' efforts to subvert what is right for the American people?

DR. TUCKSON: The next person on the list is Emily. But just for factual information, Mr. Aitken, can we be clear from you whether or not -- you represent through our association, or the coalition -- and I need you to clarify whether it's an association or a coalition, of which the Chamber is a part. Can you give us a sense of how many other organizations are there that have the same opinion or philosophy that you've expressed so we'll know whether or not, as Ed really lays it out here, how much of this is the Chamber, but how much are also other interests here?

MR. AITKEN: The Chamber is a member of the Gene Coalition. SHRM is a member of the Gene Coalition. I would guess there are some 40 other associations that are members of the Gene Coalition. The principles that I laid out are principles that have been adopted by the Gene Coalition, which the Chamber is a part of. I don't recall, and I certainly don't want to speak on behalf of the Chamber, that the Chamber has ever said no, they're opposed to a bill prohibiting genetic discrimination. They have raised concerns within 1053 and have shared those concerns with public policy decisionmakers on both sides of the aisle, as has SHRM. Again, I don't want to speak for SHRM, but the Coalition has been very consistent on those set of principles, and that's looking at any legislation that's moving through, whether it was advanced in the House or the Senate.

DR. TUCKSON: As we turn to Emily, Mike, you've been very forthcoming here. I wonder whether -- and I know you've got staff support here as well, or other colleagues -- if we might get a list of those other 40, and our team can try to, during the lunch break or sometime today, get that back so at least we'll understand the range and depth of the interests that you are so articulately representing today.

Emily?

DR. WINN-DEEN: So I guess what I'd like to understand while we have you on the hot seat here right now is what exactly it is that employers are concerned about?

Because it seems to me that from an administrative point of view, for a company that has employees in more than one state, that having a federal piece of legislation covering this would actually simplify their lives in many ways, rather than having to worry about the sales rep in New Jersey having a different situation than the sales rep in Tennessee or Texas or California or whatever. So even if you don't have multiple headquarters in different states, many companies have employees distributed over all the states. So is it just the particular wording? What is the issue with having a sort of unified floor?

MR. AITKEN: Well, S. 1053 is not a preemption. It's in addition to whatever the state laws are, so there's a difference. If it was a preemption, I don't think there'd be as big a concern with 1053.

DR. WINN-DEEN: I guess my understanding was that if it was in a state where it was a higher level than the state, that the federal law would then apply. So the only places where the federal law wouldn't apply would be in states that have even higher levels of protection. Is that correct?

MR. AITKEN: Correct. But that could be quite a few. I don't have the information in front of me, but there could be several, and that could --

DR. WINN-DEEN: Do you know? If 1053 went to becoming law, how many states would have stricter regulations?

DR. LICATA: If you give me about five minutes, I can tell you, because I have a complete analysis of all the states. So I'll work on that right now.

MS. MASNY: Heidi Williams.

MS. WILLIAMS: Yes, thank you. There are a couple of points that I wanted to make personally. First off, my children are the ultimate irony as far as health care insurance is concerned, because Humana, Inc. is headquartered in Kentucky, in Louisville, Kentucky. My children were born in the State of Kentucky, and yet they were denied twice in writing, and the Genetic Alliance has copies of these letters, due to their carrier status. That was the only reason they were denied.

Also, I want to make this point, that alone in my family history -- and there is a history of obesity, diabetes, heart disease, stroke, prostate cancer, breast cancer, and my personal favorite, male pattern baldness. These are all considered genetic disorders under the genetic disorders website at NIH, every single one of them. What I am afraid of is that once employers and insurance companies get tired of picking at the more exotic disorders, that they're going to come after people's family histories, where these things have been showing up year after year after year, generation after generation.

I'm all for education, too, as far as these disorders are concerned. I think this is just as important as the legislation, because I know that somewhere down the line my children are going to have to have their spouses genetically tested to see whether or not they carry the AAT gene, because this will go on to their children possibly, ending up as a symptomatic patient just like myself if they are not tested.

So this could come down to even their children as they are born, or even before they're born, in the womb, and they could be prevented from employment or insurance.

DR. TUCKSON: Thank you very much for that. That's very helpful.

We have Debra, and then Cindy, and then Francis.

Let me just try to caution everybody and just do a time check here. We've got a tough challenge of a 1:05 time we were supposed to stop, break at 1:05, but we can maybe

move it a little bit. Did we change the schedule? We've got a new schedule, but even then we've got a tough challenge. This is great, between yesterday and today. This is terrific. But it's 1:15.

So the bottom line of what I'm trying to get at is we're trying now to take this and zero in on what this committee can do. So I want to just keep people on point to what can we do and what should we do.

Debra, let's go there.

DR. LEONARD: Just a quick follow-up question to Mr. Aitken. I might have missed it. What is the Gene Coalition? What is their mission? What actions have they taken to date?

MR. AITKEN: The Gene Coalition is a group of organizations, principally employer organizations that have been involved -- there are some health care organizations that are active, as I recall -- that have worked with both the House and Senate on legislative proposals dealing with genetic discrimination.

DR. TUCKSON: Cynthia, and then Francis.

MS. BERRY: It probably goes without saying, but I'll say it again anyway. I think it's unanimous on this committee that we all strongly support federal legislation to prevent genetic discrimination. But I've also been in Washington long enough to know that sometimes we have a tendency to say to someone, well, if you disagree with me or you have a different approach, you're bad or you're evil or you have nefarious purposes, when really that's not the case.

I think everyone in this room has similar purposes, and the people that we heard from earlier on the first panel were telling us compelling stories about their personal lives, their health, their livelihoods, their families. Folks on the other side of the issue, though, are probably doing similar things and they're focused on their livelihood and their ability to stay in business. I don't want to demonize one side or another, except what I'm thinking in terms of what we can do as a committee to help facilitate some sort of positive action is see their common ground.

I know that individuals have been trying and working on this issue, but just for the sake of throwing something somewhat controversial out there, I haven't seen, Mike, the specific proposals that the Coalition may be putting forward or is advocating with regard to changes that need to be made to the Senate bill, but if you've got actual legislative language, it might be helpful for us to look at that.

To throw out the little controversial piece is to say to the rest of the folks here, and I'm not saying I agree with them because I haven't seen the proposals, but just for the sake of argument, if we had the recommendations of that coalition, is a bill that incorporates those changes better than no bill at all? In other words, if we were to say to the employers who were expressing some concern, or maybe some of them even oppose the legislation, if we were to adopt their recommendations, is the feeling that it would gut the legislation and not be worthwhile, or is it something that people could live with as a compromise? In other words, is it better to have a bill that's less than perfect, or do we fight for something that we feel is absolutely critical? Each and every one of the provisions as crafted by the Senate is integral to the actual legislation?

I throw that out there as just sort of a -- and it's hard probably for you folks to answer because you don't have the draft proposals or recommended changes in front of you

either. But I wonder if that could be a starting point for what we might do as a committee.

DR. TUCKSON: Excellent point. We'll come back and nail that one.

Francis?

DR. COLLINS: So a suggestion, and then I want to ask a question.

The suggestion in terms of trying to influence the political process beyond what the committee has done already in terms of writing letters to the Secretary. I like the idea of encouraging the Secretary to follow up by contacting the relevant members of the legislative branch that are in the best position to actually get this unstuck, and in my view that would include the speaker, that would include the majority leader, Tom Delay, and it would very much include Joe Barton, who is the chairperson of the Energy and Commerce Committee, which is the committee that would have the primary jurisdiction over this bill. If Mr. Barton were to take an interest in holding a hearing on this, I'm sure that would be something that would happen.

So those are the three sort of pressure points that might be included in such an appeal to the Secretary to make good on what I believe is a very sincere effort on the part of the Secretary to see something happen here. We could perhaps push this just a little further by that explicit suggestion.

The question I wanted to ask, I didn't want to let the health insurance side of this completely off the hook. Somebody a little bit ago said, well, the health insurers are in favor of a bill that would prevent genetic discrimination. I'm not quite sure that that's the way I've heard it. So can I ask Dr. Armstrong, obviously your arguments are that health insurers have not been using genetic information and will not be interested in using it, yet we heard an example from Heidi Williams where her children almost lost their health insurance. But for the intervention of a reporter might very well have had that happen.

I think, frankly, all of the information we hear from the surveys that Dr. Hudson reported and from all of our distinguished panelists is that the public basically doesn't believe that. The public does not believe that health insurance companies are going to pass up the opportunity to use this information if they had the chance to do so. That's just not an argument that seems to be carrying the day.

In that situation, if it's true that health insurance companies are not interested in using the information, then I would say they probably shouldn't care too much if there's some legislation that says they can't because, well, they weren't going to anyway. So does AHIP support S. 1053?

DR. ARMSTRONG: One, I would refer you to AHIP. I can speak from an Aetna perspective, only because I don't know the evolution of AHIP's thinking on it, although somebody in the audience is here from them, so they might address that. From an Aetna perspective, we have supported the Senate bill. Our perspective on the use of genetic data is that, in fact, it's already occurring. This activity has been happening in health plans for decades. We handle hundreds of thousands of genetic claims a year. In the health plan there's a tremendous amount of management programs that go around that involve disease management, pharmacy management. Some of it may not be identifiable as genomic activity now. In the future, it may be.

It may plug into chronic hypertension management, disease management, a whole host of other management conditions that are being actively managed, perhaps not under the rubric of genomics, but a few years down the line that will probably be the case. Our

interest here is in having access to the data for our operations in our management programs but not to use in a discriminatory manner, and then I would further question about the evolution of AHIP's position on it, too.

DR. TUCKSON: Can we ask if there is a person from AHIP, can they come and let us know what the thinking is? I think, Francis, it's important to underscore what Joanne just said in the sense of the use of the information. I think for the record I'd like us to be precise. If I heard Joanne, using the information for the purpose of -- her whole testimony was around using information for appropriate coordination of care is something that you are very clearly interested in.

I think Francis, the thesis of his question, if I understood it, is the use of this information in ways that would be negative to the individual from an underwriting or insuring point of view. I think that Joanne's answer stands on its own. But I think this is a very subtle point that needs to be distinguished.

MR. WILDER: Thank you. My name is Tom Wilder. I'm the vice president for private market regulation with America's Health Insurance Plans.

We did not oppose the Senate bill. We have expressed to Congress on various occasions our concerns that legislation be very carefully drafted for some of the reasons that Joanne pointed out. There are a number of reasons why health plans use health information, as part of disease management, as part of other programs in communicating with a provider, in communicating with patients. We want to make very sure that if there is legislation that passes Congress, that it doesn't jeopardize those functions.

We would agree that you should not be able to use genetic information to unfairly discriminate against somebody, but we want to make sure that the information that the health plan has -- in fact, the health information the plans have been using for decades is not jeopardized.

DR. TUCKSON: Could you then extend that a little bit towards the real issue now, then, which is the bill in the House? Do you find that AHIP's position is one that is supportive of -- and again, you've probably got a better finger on this than I will, so I apologize for the general nature of the question. But is AHIP in support of trying to reach a consensus that gets this now through, or would it find itself more aligned perhaps with some of the positions that people are sort of associating with the Gene Coalition?

MR. WILDER: We are members of the Gene Coalition. Again, what we've tried to do in dealing with this issue is educate members of Congress about how health plans use health information. The bill that's on the House side, the Slaughter bill, to be honest with you I'm not familiar with all the details of that bill. I do know it's very different from what came over from the Senate in a number of respects. But again, if there is going to be federal legislation, our only concern is that some of the issues that Mike and others have raised are addressed in the bill.

DR. TUCKSON: If you can hang there for a second, let me turn to the other side of this, the equation, for a minute. NBCC -- and I think we sort of heard, I think in testimony earlier, that they sort of didn't like the Senate bill because it wasn't -- and maybe this is not a fair way to say it, but wasn't tough enough on some of the sanctions. So on one end we've got this Senate bill which is -- it's kind of strange to say it this way, but a middle ground sort of deal. Some people feel like the Senate bill has too much in terms of risk on the part of employers in terms of legislative sanctions and unintended legal exposure, which makes them

nervous.

On the other hand, you've got some folks who are saying it isn't tough enough, and then you've got this sort of middle ground somewhere.

Do we understand anything or can we learn anything right now on the potential of fractionization on the side of the people that sort of want it more? I mean, are we going to see any bringing together of that part of the community, or are we going to see the NBCC staying way out and not coming into any chance of a grammar of coexistence on this?

MS. FISHER: Can I speak to that? I know something about the NBCC's position, and they are generally people who say that things like breast cancer would be cured if it were a man's disease. I think they've politicized the issue so much that they really are off to one side. I agree with Ms. Berry that this should not be a situation where someone becomes sinister and malevolent.

I do think that the stakeholders that you're asking about, those of us who do have a vested interest in getting this moved along, are not championed any longer because the National Partnership for Women and Families has basically lost our charge leader. She has since gone to a different organization. So there really isn't anyone who is still out there consolidating, still leading the charge for those of us who are still in it. I think that's a big problem.

I also think that it's difficult for your committee to approach Congress because of enjoinders against your lobbying activities. However, I would say, in echoing something Dr. Collins said, Secretary Thompson's wife, his mother-in-law and his daughter have all had pre-menopausal breast cancer. I think there's BRCA1 afoot. I think that those of us who are out there are waiting for him to speak formally on this issue, and I think he would go far toward getting it done.

DR. TUCKSON: One last question. So as we try, then, to see where the leverage points are for us to be able to do something meaningful, Mr. Aitken mentions a meeting that occurred a few months ago where it sounds like there was some sense of trying to get a consensus on the part of differing parties, and to date no response. I'm sure the other folks who didn't respond yet may have some view about that. But at the end of the day, is there anything that anyone here can tell us that is another meeting that is planned, any behind the scenes brokering to try to get this done, or from all available information at this point is this committee left to assume that this thing is basically fractionated, that parties are in separate camps and there is no work to try to pull this together? Can we know anything about the current status?

Please state your name, please, and come to the mike.

MS. HOWES: My name is Joanne Howes, H-O-W-E-S. I'm with the firm DDB Bass & Howes, and have been working with the Coalition for Genetic Fairness that Becky Fisher referenced, half of the National Partnership for Women and Families.

I think the reality that many of us feel is that we worked very hard together. The bill that passed the Senate, they were changes made outside of the National Breast Cancer Coalition. Groups saw it. It was a compromise. It wasn't what everybody wanted, but we felt in good faith that that was the right way to go. We had been assured at that point in time that it had the support of the Chamber of Commerce, that it had the support of the health insurance industry, that they had been at the table, frankly, more than we had been at the table when those negotiations got made to move S. 1053.

So where we are now is that we've worked also very hard to get the attention of -- Phaedra here is in the speaker's district, and she has been working very hard to get the attention of the speaker. We've been working very hard to get the attention, as Dr. Collins said, of the people in Texas. I think that we feel very stymied.

DR. TUCKSON: Thank you for that. I think I can understand what you're saying there. But as of right now, I think what your answer is, though, is that we're unaware of any particular activity that is on the docket where everybody is sort of sitting down and trying to work this out, that there seems to be a great deal of frustration.

DR. COLLINS: There's an election in two weeks.

(Laughter.)

DR. COLLINS: Nothing is happening in this town if not election-related.

MS. HOWES: The message that we have heard from the Republican leadership is that they're not going to do it this year. They've said it publicly, they've said it privately at the House. They're not going to do it.

DR. TUCKSON: Thank you. Thank you very much.

We're going to start now mailing this through to solutions. But, Ed, you wanted to comment?

DR. McCABE: I just wanted to comment, first of all, about the election. But then I'm concerned that in the next session, that it will still be stuck at the desk. So the issue, and maybe Cindy, I think the issue is how do we pry it loose so that there can be the deliberation? As long as it's not under deliberation, then there is no discussion and it basically doesn't exist. So the issue is how do we pry it loose so there can be public discussion?

DR. TUCKSON: Phaedra has a comment.

MS. MALATEK: I just want to quickly comment on my conversations with the speaker's office. They had a ready answer. It's clearly something that is on his plate and something they have reviewed, and this was with the senior health policy advisor. So I felt like it was somebody who was in the know in terms of where the speaker sat on this.

His comment was that we are not going to legislate something that's not happening. So they're looking for bodies, they're looking for casualties, and until they have those casualties, they're not willing to move forward on it. We're casualties, but I'm the only one here from the speaker's district. Perhaps it needs to come from the constituent level, I don't know. But they're not going to do it if it's not a problem at a constituent level is the message that I heard from the speaker.

DR. TUCKSON: Thank you.

Ed?

DR. McCABE: Well, that was the purpose of today's panels, because we've been hearing that it doesn't exist. But I can tell you as a medical genetics professional, it does exist, because I hear it over and over.

I think what we do is that we perhaps, as a way of preparing ourselves for the next round, I don't know if anybody has looked through this, but the testimony we heard this morning in the first panel was incredibly powerful. The documentation that we have in the spiral-bound book is incredibly powerful and suggests that it's still anecdotal, but it's an accumulating number of anecdotes. It's not something that can easily continue to be swept under the rug.

I would then propose, Reed, some discussion perhaps with you and Sarah

over lunch about what it would take and whether there is staff time available to take this spiral-bound notebook and summarize it to send it forward.

DR. TUCKSON: That's a terrific suggestion. Let me try this, then, and I really have to see now what the committee wants to do. Let me propose -- and I'm going to miss something -- what I think I hear at least as next steps.

Number one, and this is the most minimal step, that we ask for a meeting with the Secretary of Health to tell to him about how serious an issue we think this is, that we would present to him a summary with detail of the testimony we heard here today, particularly the overwhelmingly powerful statements we heard from our public panel, as well as the scientific studies that we've heard from our health professional colleagues, so that we bang that somehow together in a document, as Ed I think has just suggested, that we can hand to him, and at the same time perhaps ask him if he might broker at that conversation a meeting with the Secretaries of Labor and of Commerce? Justice. Excuse me, I forgot. Justice.

We'd sort of ask them all to come together, and/or senior people, so that we can sort of give it all to them at one time to try to extend the nature of the conversation within the federal government. Of course, all this depended upon Francis' very important reality check about what happens in the next couple of weeks and so forth and so on.

That means, by the way, that whatever we come up with, we're going to want a conference call discussing next steps based on some of those realities.

DR. McCABE: I would point out that a very important thing has occurred. You mentioned it. Our charter has been continued to 2006. So no matter what happens, we continue.

DR. TUCKSON: Good point.

DR. McCABE: This issue will continue.

DR. TUCKSON: So maybe the strategy could be the same.

DR. McCABE: Yes. I think it's important for us to begin the strategy so that we can address this problem in the next session of Congress.

DR. TUCKSON: Number two, that as part of our asking for that meeting with the Secretary, that we would take the same document that we've already got bound together and we're going to work on, Sarah -- thank you -- would be that we would ask that that testimony be transmitted to the Congress as a function of what we do. Since we can't do it directly, we have to sort of say that we're so concerned about this that we've asked the Congress to -- we want to get it to them, and you're the only way we can do that, so please send it.

Number three. Francis indicates that whatever we do with Congress should be focused in on three major foci, number one being the speaker, number two being the majority leader, and number three being Joe Barton. What's that committee again?

DR. COLLINS: Energy and Commerce.

DR. TUCKSON: Energy and Commerce.

MS. BERRY: Also add Education in the Workforce. I believe that bill had been referred to three different committees in the House, so you'd want to --

DR. COLLINS: I think it's pretty clear that Energy and Commerce is the one everybody is looking at to take the lead. If you want to sort of make your effort most likely to yield fruit, instead of being too diffuse, I would go for Energy and Commerce.

MS. BERRY: Well, it's not a matter of being diffuse. You don't want the other chairmen to feel like they've been ignored or to give them some sort of excuse for saying,

hey, I wasn't part of that deal, or I didn't get that stuff. So I would send it to the chairmen and ranking members of all of the three committees that have jurisdiction.

DR. COLLINS: Fine, but prioritize.

DR. TUCKSON: So friendly amendment, and I think that sounds like a winner.

You had a comment on this?

DR. WINN-DEEN: In looking through this book, what I don't see is actual copies of everyone's testimony that was given today. So it would be really good if we put all that testimony in, and also where people are from, what congressional district. City and state is extremely helpful because it brings home the fact that this is not just one Congressman's issue. This is an issue for all.

DR. TUCKSON: Good, and I think I see Ed's hand.

DR. McCABE: I just would say that as powerful as the comments were from the first panel, I would include the comments from all panels this morning. I think that would be important.

DR. TUCKSON: Yes, that's terrific.

The next thing I think that we hear is that we will send a letter to the Gene Coalition asking -- Cynthia's point, and you'll have to help me with the language on this later, Cynthia. But ultimately I think it is would you please be specific about what it is that is wrong with this legislation that you object to and what is the solution to this problem. I think it was Francis' point as well that was pretty clear on it. So if you guys are perceived as being the holdup here, then help us to solve the problem or declare for one and all, if that's how you feel, that this isn't a problem and we're going to stick to that and we're not changing. So just what is it? So we don't have to keep arguing with you about it, just tell us what it is.

Similarly, AHIP I think has a responsibility here because it's been asked specifically in the meeting to state where you are on it and how you view it. I think the issue specifically for AHIP is to be very clear about whether or not it is a problem of use of the information for coordinating care, that stuff which is -- and I'm going to, by the way, ask the people from the advocacy coalitions. I think our thinking on this would be greatly advanced if you have strong feelings about the use of the information for coordination of care, if you have a problem with even that part of it, we really all need to understand that.

But if it's your issue is really the discrimination issue around underwriting and insurance and so forth, that's one thing. But if somebody says, hey, you're not supposed to know it to even help coordinate care, which is all the testimony we heard from the Aetna representative, and that's what AHIP's point is, we need to know whether that's an issue or not. But I really think it's probably the other, and maybe I'm generalizing too much.

Yes, Ed?

DR. McCABE: Reed, I would ask also, I don't think that we can approach or you can approach the Chamber directly, but I would ask the colleagues at the Chamber of Commerce, given that I have demonized them here today, to respond because, as Cindy said, people in the room all understand that we have issues, but not being in the room raises serious questions. So I would ask friends of the Chamber to take back the message that perhaps they might want to respond in this packet as well.

DR. TUCKSON: Well, Ed, you have more experience at the helm than I do, will ever have I hope, but I think we are able to write a letter given that testimony has come

up and offer them the opportunity to clarify. I think you're good. So we will also send a letter to the Chamber. But I would probably say, though, that given the number of organizations that I assume are in that coalition and the clout, the Chamber may be an issue but everybody in there is going to be important to our committee, and so we'd like to hear.

Lastly, I think that's my list, and I'm wondering what did we miss?

DR. FEETHAM: I would just recommend Dr. McCabe's point about we have cumulative anecdote, but we also, as you indicated, have all the speakers' testimony, but they also have publications with hard data, and I would strongly recommend that those data be part of this packet that you're compiling.

DR. TUCKSON: Thank you for that.

I guess the last point would be this, and that is I think what the committee is pretty clear about is that this issue has been prioritized. We as a committee have prioritized this. I would be surprised if there's anybody on this committee who feels less strongly today about this than they did before. I think we're probably pretty well jazzed up about this. So what I wonder, and I have no idea whether this is going to be appropriate or not, but we need to assess, and that is maybe at our next meeting consider bringing together those people that are determined, and if there's no negotiation or conversation happening outside of the room, put a little circle and invite people to it, and if you don't come, you get Ed McCabe.

(Laughter.)

DR. TUCKSON: So that should strike fear in everybody's heart. So we create a circle and we say, look, we're going to lock the door and we're going to have everybody work it out. I mean, if there's nobody else working it out, then maybe we have to try to work it out. I mean, that's not usually the kind of job that we would have, but in a vacuum maybe that's something we have to fill.

I want to conclude this with that clearly we need to talk about this again as a committee. It is very clear that we're going to need to probably have a conference call on this if we're going to get anything. So we're going to summarize all this, get it back to folks, probably do a quick call, not take a lot of your time but make sure that we're locked in on this as we go forward.

MS. DOMINGUEZ: I just wanted to offer an assistance to the Department of Justice, to have the Commission independently analyze the gaps there may be legislatively. We have a lot of experience in this area.

DR. TUCKSON: Great.

MS. DOMINGUEZ: So we'd like to do that.

DR. TUCKSON: That would be a very, very good addition.

We've got to go to lunch, but let me say one thing about the lunch. You all are not going to be happy, either the members of the committee or our guests, about lunch. It's short and it's a no-fun lunch break. It is not because we are rude. It is not because we don't love you or care about you. There are more laws and regulations that tell us whether or not you can have four people in a room with a sandwich that it will make your hair hurt.

I just have to tell you why there is a reason why this thing is set up the way it is, and it's not because we are rude. So if you don't like the fact that you all have to run and try to find a sandwich and be back here in 15 minutes and that it's a working lunch and that it's all scrunched up like this, it's the only way we can feed these people, to make them work during the time that they're actually eating. Otherwise, we couldn't even give them a hamburger. So

it's not our fault, and I apologize for it, but that's just the way bureaucracy is.

Thank you all very much. See you in 15 minutes.

(Recess.)

AFTERNOON SESSION (1:45 p.m.)

DR. TUCKSON: Well, listen, let me get everybody back and thank you. For the record, the ex officios had to buy their own food. I get all these new challenges when I come into the job. It's incredible.

We want to get started on the next session. We're a little behind time. The committee remembers that at the March priority-setting meeting, we identified coverage and reimbursement of genetic technologies and services as a high priority issue requiring in-depth study. At the June meeting, the committee considered a preliminary draft report on coverage and reimbursement which was revised over the last couple of months.

Let me just say I want to really, really thank the staff for these revisions. While the report still will be revised further, there has been a considerable enhancement of an already very good piece. But they have really, really changed the report. I can see a significant improvement as they have listened to the inputs from lots of different stakeholders in this. It now includes revisions that they have worked with that they have received since June. The revised report is in Tab 5 of the briefing book.

Cindy Berry really needs to be thanked for her leadership on this, as well as Debra Leonard, Emily Winn-Deen, Muin Khoury, and Sean Tunis from CMS. I really want to thank Suzanne as well as Amanda for their staff support.

So to lead us through this discussion, I'm really pleased to ask Cindy to take it away.

MS. BERRY: Thanks, Reed.

I want to echo Reed's word of thanks to the staff, because this was an enormous undertaking. Reports of this magnitude don't just appear mysteriously. To my knowledge, they haven't invented a machine that will take down everybody's comments and then synthesize it and make sure everything is grammatically correct and organized and insightful. That is something that the staff did, and we owe them a debt of gratitude because there is no way that any committee can draft a document such as this in the amount of time that it has taken and produce the results that these folks have produced.

So Sarah, Suzanne, Amanda, Fay and others, incredible job.

I'm going to switch these two, the report purpose and goal. I'll start with the goal, really to improve access to genetic technologies, genetic services, genetic tests in particular. We have in there the word "appropriate," and that is key because no one here is suggesting that all genetic tests and services should be paid for and covered no matter what. We're talking about appropriate coverage and appropriate reimbursement, and that is the overarching goal, one of the goals of this committee.

So to that end we said, as you'll recall from previous meetings, how can we do that? How can we accomplish that? What are the barriers to achieving that goal? Two of the largest barriers really have to do with coverage and reimbursement. In many cases there is inadequate, insufficient, or non-existent coverage of genetic tests and services, and to the extent that some are covered, the reimbursement is not adequate and is a further barrier to access.

So the goal of the report or the purpose of the report is to sort of do a comprehensive analysis of the state of play with regard to coverage and reimbursement, and then offer recommendations to address the discrete barriers to access and the problems that we identify with regard to coverage and reimbursement.

As you'll recall, at the June meeting we did take a look at a first draft of the report, and we had a robust discussion, but we didn't finish the discussion because there are so many issues involved with coverage and reimbursement. We didn't get too far, so the suggestion was made, and we implemented it, to form a coverage and reimbursement task force which would be tasked with further examining the report, the objectives, the barriers, revise the report based on some of the recommendations we heard at the meeting, based on input from members of the public.

We held a task force meeting on September the 8th to further delve into the issues that we identified and recommended some additional deliberations. This is the task force. Then our September 8th meeting consisted of the following participants. We had some outside folks because we had a lot of questions in terms of how operationally genetic tests are actually provided, the reimbursement issues, what are the challenges there. So we had a lot of questions. Based on that input, further revisions were made to the draft report that you already saw.

These were the goals of the meeting, develop some concrete recommendations for the full committee to consider. The task force did not want to and did not attempt to say we're going to just come up with recommendations and everyone is going to bless them. Not at all. What we were doing is developing a list of possible recommendations for the full committee's consideration, and you'll see as we go through the report, and if you already read the report you'll see that there are numerous recommendations under certain sections. Some are additive, some are somewhat mutually exclusive, and in some cases we really didn't have any recommendations.

The purpose of today's session and tomorrow's session is to really get everyone's input on the committee, ex officio members, and members of the public so that we can come up with very thoughtful, concrete, and productive recommendations to addressing some of the barriers that we identify.

A final goal of the September meeting was to plan this session.

I next want to turn to Dr. Linda Bradley, who is going to give us a presentation. It was done really at the request of the committee from the last meeting. We discussed the need for some sort of mechanism for assessing when the evidence base is sufficient for establishing clinical utility and making coverage decisions, as well as a mechanism for addressing gaps when there is no real evidence to support coverage necessarily for genetic technologies. It was brought to our attention that the EGAPP project, Evaluation of Genomic Applications in Practice and Prevention, which is really the next step in the ACCE process Muin had identified for us the last time, really would be a potential model for us to consider.

So at our last meeting we requested a presentation on the EGAPP project so that we can assess whether it's something that we want to incorporate, either some of its work or all of its work in our coverage and reimbursement report.

So with that, I'd turn to Dr. Linda Bradley for her to brief us on this project.

DR. BRADLEY: Mr. Chairman and members of the committee, we really want to thank you for allowing us this opportunity to provide a brief review of a recently launched model project by the CDC.

It's important to point out that this project does not represent a new concept as much as an evolution of ideas and methods dating back to the 1997 report of the Task Force on Genetic Testing. As most of you know, this report emphasized the need for evidence-based review of new tests during transition from research to practice, and for a coordinated process to collect data in pre- and post-market periods. It also described assessment criteria, analytic validity, clinical validity, and clinical utility in the context of genetic tests.

SACGT, through its subsequent deliberations, affirmed the task force assessment criteria and added emphasis on ethical and social issues as a component of evaluation. SACGT also encouraged collaboration for data collection and education and made other specific recommendations, among them that CDC play a coordinating role in data gathering and analysis.

In 2000, CDC took a step in addressing the need for pre-market review of data by funding a cooperative agreement with the Foundation for Blood Research to evaluate the ACCE model system, which has been referred to here. ACCE, as most of you know, simply reflects the components of evaluation laid out by the task force and SACGT. The ACCE process is based on the premise that you first define the specific disorder or phenotype to be tested for, then the test, and the setting in which the test is to be performed -- for example, diagnosis or predictive testing -- then an analytic framework -- in this case, sets of targeted questions can be used to systematically review evidence on each component -- analytic validity, clinical validity, clinical utility, and related ethical, legal, and social implications.

This process was designed to assess availability, quality and usefulness of data on DNA-based tests for disorders with a genetic component. ACCE differed from some other standard methods, and some characteristics of the ACCE process included a broad focus. ACCE aims to provide a first look at all the available data, not just the published literature, and

also reviewed all evaluation components, including analytic validity.

ACCE used an ad hoc approach to grading quality of evidence versus a more structured approach in order to extract maximum information. ACCE reports included review, analysis and integration of data, and identification of gaps in knowledge and the data needed to resolve them. An objective of ACCE was not to suggest policy or make recommendations but to provide complete, accurate, up to date summaries in formats useful to a range of audiences.

Five ACCE reviews have been posted for comment, and you can see the titles here. We've learned a great deal from this process. I think two points that I would raise is that we learned, as I expected, that this information is eagerly received. We've had rapid uptake and application of the evidence developed. For instance, data developed for the CF report was utilized in preparing the 2004 revision of ACMG's mutation panel recently published in Genetics and Medicine.

We've also learned that in moving beyond the published data, we can uncover new and useful information. For example, in the absence of any other data, new estimates of analytic validity in the CF, HFE, and Factor V reports were derived from external proficiency testing data. Though not an ideal approach, it provided a reassuring snapshot of U.S. performance as not perfect but really pretty good. It also highlighted situations in which understanding of analytic performance could impact how a test is implemented in some settings.

So as we considered the next step from a public health perspective, the question really was what might a non-regulatory process for evaluation of genetic tests look like? Well, certainly evaluation needs to occur at two key points. The first is transition from research and development to clinical practice, ideally before a test enters widespread use. It should include systematic review of evidence on clinical validity, and with the rapid development of new complex technologies with very little performance data, systematic review of analytic validity needs to be part of this process, too. In some cases, certainly a first step.

Assessment of risks and benefits, while focused on outcomes, needs to include more, including consideration of resources for the testing process, counseling and education, the need for pilot trials, and cost-effectiveness analysis. We need to identify more effective approaches for assessing ethical, legal, and social implications of testing, and there needs to be a plan for dissemination of the information developed to all relevant target audiences.

The second key point for evaluation is in the post-market period to assess performance in practice and public health impact, beginning with very basic information that we currently lack on utilization and access. We need to be able to document problems and successes with implementation and fit with health care delivery systems, and we need to be able to update the knowledge base after the test has been in practice.

Evaluation of Genomic Applications in Practice and Prevention, or EGAPP for short, is a three-year model project. The goal is to establish and evaluate a systematic mechanism for pre- and post-market assessment of genetic tests and other genomic applications in the U.S., hopefully one that can be sustained in some form beyond this model project. EGAPP is a public health initiative with a population focus, and like ACCE the objective is a first or early look at new tests and technologies to determine what is known and to identify important gaps in knowledge.

The project plans to utilize information and recommendations developed through this and other advisory processes, as well as the knowledge gained from the ACCE project. Partnerships and collaborations are vital to the success of this project. Examples include existing evidence-based processes -- for example, the Agency for Healthcare Research and Quality, the U.S. Preventive Services Task Force, evidence-based practice centers, and the CDC's Task Force on Community Preventive Services. We began talking with these groups early in the planning process, and ongoing discussions have been helpful as we sought to define the scope of the project. These and other agencies and organizations, many represented here today, will have a continuing role in advising the project.

We're also interested in collaborations with the international health technology assessment community, and we are developing very productive contacts with groups in Canada, the United Kingdom and the Netherlands who are involved in similar projects.

We're also interested in relationships with other projects and initiatives. There are certainly too many to cover here, but just to mention a few, with regard to quality assurance and improving laboratory practice, CDC's Division of Laboratory Services is currently developing a process to obtain and distribute quality control materials for genetic testing to labs, researchers, and the diagnostic industry. CDC Division of Laboratory Services, in collaboration with Emory University and the National Institutes of Health Office of Rare Disease, held a May meeting on promoting quality laboratory testing for rare diseases. Outcomes included the formation of this laboratory network and planning for a second conference to move into implementation of the recommendations developed. There are also a number of other initiatives related to policy, programs and services, and research.

The process aims to provide a clear linkage between the evidence developed and the recommendations made, minimizing conflicts of interest in the review process, but keeping in mind some very good advice from Al Berg of the University of Washington and the U.S. Preventive Services Task Force, that evidence-based requires that the linkage be transparent, explicit, and publicly accountable, not that it be objective. The project will develop a plan for effective dissemination of information to target audiences.

The question of are genetic tests different or exceptional comes up here again, and whether or not genetic tests are exceptional or different in other ways, methods for assessment of genetic tests have basic similarities to those used for other tests. However, there does seem to be an increased awareness and concern about genetic testing and a public perception that it is different, and I think the compelling testimony you heard this morning about potential harms makes that very clear.

EGAPP is focused on genetic tests and other genomic applications, responding to the demand from health care professionals, policymakers, and the public for a source of reliable and reasonably objective information about appropriate use of genetic tests. However, the knowledge gained about successful evaluation approaches, methodologies and infrastructure should certainly be applicable to assessment of other tests or emerging health care technologies.

Technical and logistic support for the project will be provided by RTI International, a non-profit contract research organization, and this contract was awarded in late August. RTI brings to the project a wide range of scientific expertise, but one resource we feel is very relevant to this process is the RTI University of North Carolina Evidence-Based Practice

Center.

So the central element of the project is the working group, independent, non-federal, multidisciplinary, made up of 10 to 12 experts from fields such as health care, genomics, epidemiology, health technology assessment evidence-based review, public health, health economics, and potentially others. Two in-person meetings are planned in year 1, three meetings in each of years 2 and 3.

Proposed roles of the working group -- and this is the concept -- is that the working group would first develop an organizational plan defining protocols for evidence-based review and development of recommendations. The working group will certainly consider input from stakeholders, develop criteria for selecting topics, and then select and prioritize topics for review. When a topic is selected, the working group will request that RTI commission or conduct an evidence-based review. The working group will ensure appropriate review of reports and develop recommendations based on the evidence. They will consider needs and strategies for post-implementation monitoring and data collection studies and will take part in evaluation of the project.

Stakeholders, very important. The project will identify and engage a wide range of stakeholders. The primary focus for this project is health care providers and consumers. A secondary focus is policymakers and health care payers and purchasers. RTI will conduct needs assessments and set up a process for ongoing dialogue with stakeholders. Basic information that will be sought from stakeholders in the early parts of the process include recommendations on specific topics for immediate consideration by the working group, an also on the content and format of the information needed by the stakeholders and useful from their perspectives. Stakeholders will also be a source of content experts, and their roles will include technical assistance, review of reports, and involvement in development of informational messages for key target audiences.

This just provides sort of a very simplistic overview of how the process might work, beginning with the working group and a large group of stakeholders. The stakeholders provide input on topics and priorities. The working group makes decisions on their criteria, selects a topic, and requests an evidence-based review. I put the little RTI on there just to remind you that as we go through this, RTI underlies this process in terms of making these things happen. The request will go to an evidence practice center where systematic review will be done that identifies the gaps and the data needed to fill them. Then that information comes back to the working group for appropriate review and comment, and then they will prepare recommendations, reports, and disseminate these materials to the target audiences of consumers, providers, policymakers, and purchasers and payers of health care, with an opportunity again for stakeholder input on the development of these targeted informational messages.

Under certain circumstances, the working group may decide to refer a topic out to other groups for further appraisal, for instance the U.S. Preventive Services Task Force, and they will be involved in developing collaborative projects for pilot data collection with stakeholders.

The first year of EGAPP really begins, obviously, with process development, recruitment of the EGAPP working group, two organizational meetings to follow, and the development of the working protocols by the working group. There will be a methodology conference which I'll come back to in a moment, preliminary needs assessment

activities, two small pilot data collection studies, and an evaluation that's based mainly in this first year on process.

In years 2 and 3 we'll see continuing support of the working group, commissioning and oversight of evidence-based reviews, four full reviews in these two years, and three what we're calling fast track reviews. There will be dissemination of reports, working group recommendations, and informational messages. There will also be ongoing dialogue with stakeholders who will be involved in the development of these informational messages for target audiences, as I mentioned, and who will provide feedback on the value of the process and the products.

There will be two pilot data collection studies in each of those years, and there is a comprehensive plan to evaluate the success of the process, the quality and usefulness of the products, and the impact or value of the project overall. At the end I think we really want to be able to consider mechanisms for sustaining whatever we can validate from this process for evaluating genetic tests.

So why talk about methodology when there are standard and sometimes gold standard methodologies that are being used? Well, it's been pointed out, and I think many people have seen this, that the standard process and methodologies -- for instance, the U.S. Preventive Services Task Force -- may not be as effective when we're dealing with conditions that are uncommon to rare, where interventions and clinical outcomes are not well defined, where the evidence base is limited, and that obviously is going to be the case very often here, and where there are problems with study design or quality of data. We have already noted that ethical, legal, and social issues are less amenable to the evidence-based approach. We need to think about that. We need to consider the influence of advocacy, which I'll talk about in a moment.

So the plan that we're working very hard on right now is to have a methodology meeting in July of 2005 to bring together a relatively small group of U.S. and some international experts in evidence-based review, health technology assessment, epidemiology, genomics, and health economics, particularly those having experience in evaluation of genetic tests, for a working meeting that focuses on elements of evaluation, selecting and defining topics, developing an analytic framework for evidence-based review, literature searches, grading quality of evidence, and translating evidence to recommendations.

We will also address questions about the use of unpublished data sets and the gray literature. We'll consider how to deal with proprietary data. We'll seek information and agreement, in some cases we hope, on minimum standards for determining when a test is ready to move into clinical practice, how much information is enough, what is a reasonable threshold for quality of evidence for genetic tests, is the threshold different for different evaluation components such as analytic validity and clinical validity, and how do we optimize the quality of data to be collected in the future. The results will be used to inform the EGAPP working group deliberations and will be published, we hope.

We feel that the timing is right to use what's been learned and to move forward from ACCE. Certainly the situation is not going to become simpler. More tests are certainly coming, testing will move into primary care, and health care providers and the public need a source of reasonably objective advice about appropriate use of tests. In the short term, whatever is learned will be useful and is likely to provide information to address questions posed by this committee related to oversight of genetic technologies, coverage and

reimbursement, access, potentially public awareness and understanding.

In the long term, we hope to create an expectation that a certain level of review will occur prior to acceptance into routine practice. We hope to facilitate standardization of data collection formats. We hope to identify specific gaps that may stimulate research, and we hope that what we learn will support the need for postmarket review of testing practices, clinical guidelines, and recommendations based on new information.

Thank you for your attention, and I'd be happy to answer any questions you might have.

DR. TUCKSON: Any questions?

MS. MASNY: My question is, is your process for review the same process as the methodology meetings that you're going to have, or are they two separate types?

DR. BRADLEY: Yes, that's a good question. We know that the standard methodologies that are being used in many evidence-based review practices are going to be problematic with genetic tests because we have a lack of quality evidence in many cases. So what we're hoping to do is to have this methodology conference in order to look at different ways that we might approach some of these issues. What comes out of that methodology conference or meeting in January will certainly inform the working group's deliberations on how they want to proceed, but it will be separate.

DR. TUCKSON: Have the requests gone out, the letters inviting people to the meeting in January? Do we have a sense of who you're inviting?

DR. BRADLEY: We're certainly working on a list, and I think we're about to make final decisions in the next week or so. It's been a tortuous process trying to decide on who are the right people to invite to the meeting, as you can imagine.

DR. LEONARD: This may sound like I'm self-serving, but it's a little disturbing to me to see in the stakeholders list laboratories. Since you're talking about genetic testing, I liken this to investigators who want to study breast cancer or lung cancer and they have no knowledge of what those tumors are, and so they treat them all the same. You really need an intimate knowledge that breast cancer comes in lobular and adenocarcinoma forms and stuff, what you're actually talking about.

So to see the laboratories that are doing this testing as a stakeholder rather than an active participant in this process informing the group as to the details of the testing and what it means to do it one way versus another way and applications is a little disturbing to me.

DR. BRADLEY: Well, in fact, they will be filling just that role. As I told you, we see a number of roles for stakeholders. It's a very broad group in there, and I appreciate what you're saying. But, in fact, they will have a very important role in this process as experts and will be involved in providing technical assistance to the working group, doing review of reports that come out in their preliminary stages so that the working group can be sure that the evidence is accurate and complete and there aren't issues that they don't understand. I think, very importantly, we're hoping to get guidance from this group about what priority topics are and why, and also I think a very, very important role, especially for laboratorians and people in the genomics and genetics communities, is at the other end of the process. Once you have an evidence-based review, the evidence has been collected and it's been presented, helping to develop appropriate informational messages to go out to these different target audiences.

So I think, in fact, that it's a very big role of stakeholders, and I think

obviously laboratorians are going to be a big part of that.

DR. TUCKSON: Thank you very much.

Cindy, I think we can move on.

MS. BERRY: What we'll do now is go through the version of the report that you have seen. The intent is to go through section by section, outline the report as it is currently drafted, discuss the barriers that have been identified, then discuss the recommendations that have been suggested. This discussion, which will continue into tomorrow, will provide input for additional revisions. We will also solicit further input from members of the public. There will be a notice that goes out so that we can get formal written comments from anyone who would like to comment on the draft report. So it is by no means final. In fact, today is by no means a final discussion of the report. This is just the next phase in this process.

So to start out with the structure of the report, it will give you an overview of how it is laid out. Of course, we have a preface, introduction. These are the basic chapters, I suppose, if you will, and you'll see the background section goes into fairly good detail of how health care is structured in the United States, both public programs as well as the private programs, private insurance. It goes through coverage decisions, payment decisions, how they are handled operationally, and the billing process. Then the final section outlines the barriers to access for genetic technologies, coverage and reimbursement, and then potential recommendations under each section.

We probably don't need to discuss the preface too much, and the introduction as well. It gives some general background on what we're talking about with regard to this report, genetic tests and services. What are genetic tests and services? What is it that we're trying to improve coverage of? It talks about, again, how the health care system is structured and what are the constraints under our current system, and it outlines the purpose, the goal, and the objectives of the report.

This section, of course, as I mentioned, outlines precisely what it is we're talking about when we talk about coverage and reimbursement for genetic tests and services. It describes them in a sort of a general sense. It talks about how they are different from other tests and other health care services, and it explains some of the challenges that they pose to the health care system.

We've got the overview of the U.S. health care financing system, Medicare, Medicaid and SCHIP, the public programs, private insurance programs, a discussion of managed care, and there is a bit of a discussion on the uninsured and the underinsured.

Feel free to stop me as I'm going through this. I intend to go through this part of it pretty quickly. You all have seen the report and we're sort of going through how it's structured. But if there's something that you want to comment on and you feel needs to be addressed in this aspect of the report that I'm just glossing over, just raise your hand and we'll stop.

Coverage decisions. This section of the report --

DR. LEONARD: Cindy?

MS. BERRY: Yes?

DR. LEONARD: Can I ask a question about something that still remains in here and is inconsistent with another draft document that we're working on and we'll discuss tomorrow? We define genetic tests where we're doing the overall thing -- it's not our vision

statement but whatever that thing is that Emily did. We defined genetic tests or genetics as inheritable and only inheritable, and genomics is defined as the broader inheritable and acquired. Yet on page 19, when it says "What are Genetic Tests?," we persist in including the acquired testing in the definition of a genetic test. While I agree that would go in the definition of a genomic test, I still strongly disagree that that goes into the definition of a genetic test and, in fact, is inconsistent with the definition of "genetics" that we have in our other document. So we're not even being internally consistent between the two documents.

MS. BERRY: Suzanne, I'm trying to recall where we pulled that definition from.

MS. GOODWIN: The current definition in the report as it stands was SACGT's definition. So if there is modification that this committee would like to make to that definition, you certainly can do so. If there's discussion amongst the committee members about whether they want to do that, I suppose now may be a good time to do so.

MS. BERRY: I couldn't remember where it came from, but it seemed to me that it was from some prior committee work and that it was some sort of a consensus. I am no expert in this field, so I defer to others in terms of modification to that definition. We should be consistent. We shouldn't have different definitions in different documents that are the work product of the full committee.

DR. LEONARD: It does come from SACGT?

MS. GOODWIN: Right, the oversight report, and there was a lot of deliberation around that at the time. But certainly we want for the different reports to be consistent.

DR. LEONARD: Right, and I could refer you to the other report if you want, or whoever. But there's a clear definition of genetics and genomics. So if we're referring to a genetic test, then I would think that's a test to look at genetic diseases, and that genetic definition does not include acquired. That's in genomics.

MS. GOODWIN: If you look on page 19, there's a definition for genetic and genomic technologies, but there is no definition of a genomic test.

DR. LEONARD: Right.

MS. GOODWIN: So you would like changed, then, the definition of genetic test that --

DR. LEONARD: I would, but I don't know what other committee members think about this. Ed and Reed, you were part of the previous SACGT deliberations that included acquired, but it is concerning that looking at the overview document of our deliberations on how to come up with our priority setting, that we do define genetics without including acquired, and that's included in genomics, which I agree, acquired does fall into genomic testing if you want to define that kind of testing.

DR. McCABE: I'm trying to read this through quickly. I see the sidebar on 19. Is that what you're referring to?

DR. LEONARD: Yes. It's the first sentence. It says, "A genetic test analysis performed on DNA/RNA genes and/or chromosomes to detect heritable or acquired genotype mutations, phenotypes or carrier types." It's the "or acquired" that bothers me, and I can refer you to the other document.

DR. McCABE: Okay. So I think if you then look down to what are genetic/genomic technologies, I think that's where it talks about acquired there. So it looks like

if we went back to the vision report and brought page 2 of a roadmap for the integration of genetics and genomics into health and society -- is that what you're referring to? Yes. So I think one could go back to those definitions.

DR. TUCKSON: The road map is in Tab 3, and we're talking about page 2.

DR. McCABE: I think it's important for us to be internally consistent, and I don't see any need for us to be consistent with the work of a previous committee, recognizing that some years have elapsed since that. I think the SACGT definition was taken actually from an earlier committee. So there's been quite a bit of time between when that was crafted. So I would suggest that we just bring it into consistency with the road map. That would seem to make sense.

DR. TUCKSON: Is everybody on the same page? So bottom line is we're talking about trying to take the definition in the reimbursement document and substitute that one with the box on page 2 so that we're consistent in terms of how we do this.

Sarah?

MS. CARR: I just wanted to point out that when SACGT was deliberating, perhaps it was an oversight on our part, but the issue about the term "genomics" didn't really -- we didn't have deep deliberations about that the way we did here in the last meeting. So we weren't trying to make that distinction. That's what the definition in the road map report does, tries to clarify what we mean by both those terms and that they're both important. But if we were going to use that as the basis for what we have in the coverage and reimbursement report, it still seems like we need to do a little bit of work to translate that concept, those concepts into what a test is, or what a genetic test is and what a genomic test is. Is that what you're saying, Debra?

It might be helpful to have, maybe not right now, but you or Emily or others and Ed involved in actually helping us do that, because I'm not sure it's a straightforward translation.

DR. McCABE: Right. As I look at it, the definition on page 2 is really a definition of genetics and genomics. It doesn't translate into the testing. I think if you actually look at what are genetic/genomic technologies, that would probably be acceptable to translate that for testing, for genomic testing, and just separate them out. I think it could be done by a small group.

DR. TUCKSON: Great, and I think maybe what we might also do is, one of the challenges and jobs that we have -- and Sarah, I want to make sure here -- is to educate the public. So we ought to take every opportunity to remind people about the fundamental blocking and tackling terms. So I think what we should do perhaps, or consider as a small group, is state the definition first in the vision statement and then say what this means in terms of genetic tests and technologies, so you can see the logical progression of the ideas. But I think we've got to take every chance we can to educate.

DR. LEONARD: I know this sounds like a small point, but in the mindset it's not, because genetics, where it is inheritable, has different ethical, legal, and social implications than acquired mutations. While they're done using the same technologies in the laboratory, the surrounding issues are quite different.

MR. MARGUS: But Debra, for coverage in this case, is the intent to include genomic somatic changes and all that, or is the intent to focus only on germ line inherited stuff?

DR. LEONARD: To include.
MR. MARGUS: To include everything.
DR. LEONARD: Yes. It's just that it's confusing to have a definition of a genetic test where we define genetics elsewhere --
MR. MARGUS: We just need to make it consistent, but (inaudible) include it all.
DR. LEONARD: Right. I definitely want genomics included.
DR. TUCKSON: So we'll have Ed and Debra work on this with you, Suzanne. So Ed and Debra will work on this. Thank you for doing that.
MS. BERRY: The next section of the report focuses on how coverage decisions are made in both the public sector and the private sector. It outlines the Medicare coverage decisionmaking process, national coverage decisions as well as local coverage decisions. It reviews in general how these decisions are made in the private sector, and then reviews the state of play in terms of coverage for genetic tests and services in both the public and private sector and the role of economic evaluations.
Then we move on to payment decisions, the clinical laboratory fee schedule, and the existing payment rates for genetic tests and services, and then of course the billing process, how are codes developed, how are these tests and services coded, what are the billing practices in Medicare as well as in the private sector.
DR. LEONARD: So I will provide those CPT codes, but a question. Do you want them as genetic CPT codes, or there are genetic ones and genomic? The same CPT codes cover both.
DR. McCABE: Well, I would think if we're going to be inclusive, we need to clarify that so that those reading it will understand.
MS. BERRY: Here's where we get to the meat, barriers and recommendations. The report outlines specific barriers to coverage and reimbursement in the Medicare program and other public programs, as well as in the private sector. Then in the blue boxes in the report -- and if you have simply a xerox copy, I suppose it's gray. But anyway, in the boxes is where we outline some potential recommendations for the committee to consider to address each of the barriers or most of the barriers that have been identified.
Now, how we propose going through this is really a systematic analysis of the report in that part of the report called "Barriers and Recommendations." We want to focus on that section, pages 49 to 73 of the report, and go through each one in a systematic fashion, and then discuss whether we should make a recommendation, first of all. If we should, if there is a recommendation outlined in the blue/gray box, whether it's something that the committee would like to put forward, and are there alternative recommendations that the committee feels we should include in the report.
Of course, our objective here is to reach a consensus. If there is no consensus or we can't come up with a recommendation to address a particular barrier, then it won't go in the report. Our idea, though, is to, at the end of the day or the end of tomorrow, produce something concrete that can be utilized by the Secretary and by others as a helpful guide to improve policy in this area.
DR. LEONARD: Cindy, can I ask a question?
MS. BERRY: Yes, Debra.
DR. LEONARD: Unfortunately, I just took a bite of brownie.

The question is, is this going to go, after we make our recommendations, directly to the Secretary, or is this something that goes out for public comment first?

MS. BERRY: Public comment.

DR. LEONARD: Oh, great. Okay. I missed that step.

MS. BERRY: Turning to Medicare, one of the first things that was identified, and we talked about it at our last meeting, and it's in the report here, is the screening exclusion in Medicare. CMS policy is such that tests that are performed in the absence of signs, symptoms and complaints or personal history are not covered unless explicitly authorized by statute. So a predictive test or presymptomatic genetic test is not covered by Medicare because of this policy. Preventive services that have been covered by Medicare have been specifically authorized by Congress, and we don't have that here. So this exclusion is one of the critical barriers to coverage of genetic tests and services in the Medicare program.

Emily, yes?

DR. WINN-DEEN: I think it's important for us to say major barrier for some genetic tests, because this paragraph the way it's written, it sort of implies that all genetic tests are screening tests and that this is a barrier for everything. This is a barrier for just a subset of genetic tests.

MS. BERRY: That's right.

DR. WINN-DEEN: Carrier screening, presymptomatic screening, those kinds of things. So I think we should just make that clear in the opening sentence, particularly since, for whatever reason, this, which I think is a subset of all genetic tests, is the first thing that appears. I might suggest that we put the order in a little bit different way so we deal with the things that affect all genetic tests first and then some of these subset things later just in terms of organization of this section.

DR. LEONARD: Especially since in the Medicare community it's less likely to be a presymptomatic test.

DR. WINN-DEEN: So from the point of view of presymptomatic screening, and probably even the majority of carrier screening, Medicare is not the carrier that's going to be paying for that. So this becomes one of those minor extra points instead of a major point, and I think we should start with major points.

MS. BERRY: Ed?

DR. McCABE: Just before we leave that, I'd also point out on page 61 is preventive nature of genetic services. It would just seem like as we're reshuffling and reorganizing, this is a nice lead-in to why screening is important and why it's important to eliminate the screening exclusion, and there is no recommendation that goes with that, so it would fit nicely together.

MS. GOODWIN: The way this chapter is organized is the Medicare-specific barriers are listed first, and then the preventive nature of genetic services section is listed under a barrier that's applicable to both public and private insurers. So in combining the two, there would need to be some reorganization of each section. So do you have any suggestions on how best to do that? We tried to keep separate the barriers that were specific to Medicare and the ones that were applicable across the board.

DR. WINN-DEEN: Again, you might want to do it the other way around, the ones that are done across the board first and then do the Medicare-specific things, because Medicare is a subset of all insurance.

MS. BERRY: I think probably the reason it was initially done this way, and it certainly isn't critical that we leave it in this order at all, but I think, if I recall correctly, that the thinking was that Medicare oftentimes is the model that many in the private sector follow. So how Medicare goes, the private sector goes as well, but not always, because I think as we've discovered, in many cases the private insurers are taking more of a lead and are being a little bit more progressive in the area of genetics and genetic tests and services. But I have no objection at all, unless anyone else does, to reordering it so that we do the barriers that are applicable to both public and private sector first and then do the Medicare section after that. I think that was really the only reason that it was done the way it is.

DR. WINN-DEEN: And I think in part it's because of data on what's going on in Medicare is easily and publicly available, and what's actually going on in private insurance is not always as easy to get at, exactly what they're reimbursing, exactly how they're making their decisions, since they're not obligated to be public about that.

MS. BERRY: Right. Does anyone have any objection to changing the order?

(No response.)

MS. BERRY: We'll do it. We'll still go through the report the way it's currently configured, but we'll change the order in the next draft.

The screening exclusion barrier, the recommendations that are outlined, there is sort of an order to this. CMS had informed us that number 4, which is for them to unilaterally change their interpretation of the law to allow them to cover screening services without any legislative action, is very unlikely to occur and is not particularly feasible. I think some of the folks at CMS thought number 1, which would be for Congress to change the law, would be a better approach. Having said that, we're all aware of how difficult it is to get Congress to change the Medicare statute by adding a benefit category for preventive services.

We don't want to give the impression that we think that's just a cake walk and we'll just put that as number 1 and everybody likes that and let's just be done with it, because that has its own challenges as well. These four recommendations are not necessarily mutually exclusive in any way. It's just that the original order was what might be the most effective recommendation, not necessarily the easiest or the most feasible.

I don't know if anyone has any suggestions, additional recommendations they want to put up for consideration before we go through each one.

Ed?

DR. McCABE: I'm sorry. I don't have any additional ones, but I think, especially given the visibility that family history is going to have, and perhaps it's in here, but we need to make sure we take the information that Alan Guttmacher presented, which represents a coalition among the HHS agencies, and really play that up since that's something that the Secretary is going to be quite aware of. I would just fill that in here with the website and all of that information.

I think number 1 is going to require a culture shift in American medicine, really, and culture is always hard to change, but it's essential that this change. So while it may not happen overnight, perhaps at least we can begin hammering that we need to move from acute intervention to prevention.

DR. LEONARD: How difficult is number 2 to do, though, in the context of this family history tool that's being rolled out? It would be a nice thing to coordinate that

family history that we're emphasizing constitutes medical justification for a test being done. So how does 2 happen? Can somebody inform us?

MS. BERRY: Terrence?

MR. KAY: Hi. This is Terrence Kay. I'm from CMS. Actually for number 2, we have a fairly straightforward process. We have it on our webpage of how anyone, the committee or anyone can ask for a national coverage decision and what kind of information we would look for. So process-wise, I think it's fairly straightforward.

I think the major issue is going to be -- in general, when I've listened to the discussions today, whether the issues are legislative or regulatory, I think for communication purposes -- for example, I heard today that there are over 1,000 genetic tests. So I would think -- I obviously couldn't speak for Congress, but I would think both Congress and the agency would be thinking, gee, what really is going on here? What are we walking into? It almost seems like, if you look at the history of preventive services as they've gotten added to the Medicare benefit, whether it's mammograms or colon cancer screening or whatever, that there was a case made, a specific case made for those services, and in a way that everyone could understand.

For right now for genetic tests, just sort of listening to the conversation and reviewing the material, I think a lot of folks, frankly, would be like me. I'm not a clinician, but it would be sort of hard to understand the implications. Earlier today we were talking about a thousand genetic tests. Would you want to cover all of those? Which ones are really most beneficial for Medicare? It's a long answer to your question.

The process to do number 2 is straightforward, but I think the comment that somebody made here a little while ago that CMS had indicated that it might be unlikely that we would do number 2, all I would say is that we take the committee very seriously and we're very interested in your recommendations, and we have tried to attend all the meetings. I also would put in a plug, a lot of appreciation for the involvement of the staff for this committee and how cooperative and responsive they've been to comments we've made that have been reflected in the draft report. I think it's been a very productive relationship, and we understand a lot more than when we started. Likewise, you've gained appreciation for that.

We can do number 2. We'll certainly give it serious consideration. But we're very much looking for an evidence basis for decision-making on national coverage decisions, and I think a fair amount of thought would need to be given to how best to craft the argument so that the agency would be persuaded to make a change like that.

DR. LEONARD: So if I'm understanding you correctly, I don't understand how national coverage decisions are made, and I haven't read the website. But you're implying that someone has to bring this to you as a proposal? It's not something CMS does actively themselves. And who would that somebody be who would bring this national coverage decision proposal to whatever committee it is that would say yes or no?

MR. KAY: Again, because you're the Secretary's committee, I would have to defer to whatever your process is. In general, our process is that anyone can ask for a national coverage decision. At the agency, we have our Office of Clinical Standards and Quality. We have a Coverage Analysis Group. Dr. Steve Phurrough, who has attended a number of the meetings in the past, heads up that group. We'd be happy to provide further details on exactly how one would do this, but basically if you look at some of the examples, and they're wide-ranging, basically folks come and provide the specific request and why they think

we should make the request and what data/studies are available to support that request.

MS. BERRY: Muin?

DR. KHOURY: Just some quick comments about family history. Two years ago we had an expert panel at CDC to talk about family history as a genetic test, and at that time the thinking was, and it sort of propelled us in the next phase, that if you approach family history like a genetic test that deserves its own evaluation for its utility and whether or not there is a genetic test that goes along with it -- in other words, how good is it analytically, sort of the ACCE paradigm -- it's very obvious that family history per se is sort of a mixed bag. When you take family history, even if it's reliable, sometimes it leads you to genetic testing, sometimes it doesn't. Just to elaborate on the evidence-based process, I think family history deserves its own evidence-based process.

Now, we all think that family history is good, that we need to collect it, but in terms of whether or not it will be associated with genetic testing, it's not always obvious. I mean, a simple case in point would be BRCA1 testing. There are a lot more women that have a family history of breast cancer, maybe 10 to 1, as to the small fraction of women that are in the range of being even considered for BRCA1 testing.

So I think we can use family history as a launching pad for coverage decisions, but I guess following the same rules of engagement for genetic tests. I'm not suggesting any change to the wording here, but at least the good thing is that you put family history as somewhat up there for the decisionmaking process.

MS. BERRY: I should point out as well that the first recommendation talking about legislative change, we had discussed a bill, the Medicare Preventive Services Coverage Act, S. 2535, H.R. 4898. That legislation would add preventive services as a Medicare benefit category and enable CMS to determine through its national coverage decisionmaking process and after assessing the evidence so that they could determine whether an item or service is reasonable or necessary. So even with the congressional authorization option, there still would have to be an assessment of the evidence in order to lead to a national coverage decision.

So in many respects, 1 and 2 are closely related. In fact, I don't think you could -- number 1 isn't going to do you any good at all if there is no evidence to support coverage, so they're closely aligned. The question is whether we can get to number 2 on its own in the absence of any congressional activity.

Emily? Oh, I'm sorry. Barbara was first, and then Emily.

MS. HARRISON: I was just going to say that number 3 makes me cringe a little bit. We haven't talked about it very much, but I just didn't know if we were definitely talking about 2, and my feelings were going toward 2 and a combination of 1, if we can do that. But just to kind of put out there that these presymptomatic and predictive tests are diagnostic for the mutation that may be found but not diagnostic of a disease per se, and the limited genetic literacy sometimes that we're dealing with out there, just to be wary of that. So as I kind of get our conversations off of 3 if we ever do go there, to just be careful about that.

MS. BERRY: Emily?

DR. WINN-DEEN: So I think in response to Terrence's comment that one thing that we might think about is really what tests do we know about today that we would want to see Medicare apply this to, and I'm having a hard time really thinking about anything where you would screen someone at a genetics level where you would wait until they were 65 to do

that. So I think we need to think as a committee about are there some specific examples that we might put forward instead of a more broad strategy? Is this really just a few smaller things that we should put forward specifically? It's different than the concept of cholesterol screening or mammograms, where it's something that you test for every year and you continue to be tested as you age. It's really only a question of what age do you start screening.

Genetics is a once in a lifetime test, and the question I would ask is, under Medicare, how often would we want to be doing this once in a lifetime test in that population? Having participated in this program, I feel sort of stupid bringing that up at this point in time, but it helps I think to be specific, and then you can work to a specific example of what you'd like to see and why you'd like to see it.

MS. BERRY: Agnes, and then Kimberly.

MS. MASNY: Just as a follow-up to your comments, Emily, I know that in what Terrence had mentioned about the case by case basis for looking at some of these screening tests, that the testing for breast and ovarian cancer is now approved by Medicare, and they approve it under a specific code that states hereditary breast/ovarian cancer. So maybe in view of looking at the use of the family history as a tool, one of the things that we do in a cancer setting is actually give a diagnosis to the family history as to whether it looks sporadic, which would be more the acquired mutations, something familial that we're not really sure of, we're seeing something going on in the family or something that truly fits a hereditary pattern where you're actually then providing a specific, sort of almost diagnostic category to that, sort of giving credence to why someone would go on then for a screening test, and that even in many of the situations when Medicare first approved the coverage for the testing for breast and ovarian cancer, the woman had to have had breast cancer. So she already had the disease, but then it was actually to find whether a gene was present, most probably to help other members of the family.

DR. WINN-DEEN: That's not screening. That's in the presence of signs and symptoms, which would already be normally covered.

MS. MASNY: They did expand it now to cover people without the disease. So that would be screening.

MS. BERRY: This would be a small percentage of the population, but Medicare is also for the disabled. If they're on Medicare because of disability, they obviously have signs and symptoms of some problem, but theoretically I suppose there could be a screening component where they have other problems going on. But then that's probably a very small percentage of the population that we're dealing with.

Kimberly? Barbara?

MS. HARRISON: I was just saying that we even here had an example of that with Mr. Hardt, who had hemophilia and then also has a risk for Huntington's disease. It can happen. You may have people who are on Medicaid who need a screening test for something that may be unrelated, but they have a positive family history.

MS. BERRY: Barbara had raised some angst about number 3. Does anyone want to comment on that? Do they share her concerns about that? Does someone feel strongly that we need to keep number 3 on the table as a possible recommendation? For those who maybe can't see it there in the back, number 3 is for CMS to redefine predisposition and predictive genetic tests as diagnostic laboratory tests through a rulemaking process or a national coverage decision. So in the presence of a strong family history of disease, these tests would be

considered diagnostic and not subject to the screening exclusion.

Brad?

MR. MARGUS: Can you just clarify what we're trying to do here? From a very, very, very big picture, we're trying to get coverage for tests that sometimes don't get covered, right? So we have four shots at it. You told us that number 4 is a really, really, pretty much long shot. Number 1 is certainly not trivial. That leaves us with 2 and 3, and 2 says that you make tests that are, as long as there's family history involved, reasonable and necessary, and I take it that makes it qualified to be reimbursed. And 3, instead of being reasonable and necessary, that makes it the same as a diagnostic test, in which case it gets covered.

But do you have to pick one of these, or can you give them both 2 and 3, ask them to pick one, and it gives us multiple shots on goal?

MS. BERRY: Correct me if I'm wrong, but my impression is that these are not mutually exclusive. There may be some on here that people just don't want to pursue and don't feel as a committee that we should put that forward as a recommendation to the Secretary. On the other hand, if we think, hey, why not, all of these seem viable and it's, of course, possible, perhaps even likely that the Secretary would reject, for example, number 4 and just say, look, that's just not feasible, that's his decision to make and we are at least offering that up as something to consider.

MR. MARGUS: Did the task force or the staff see any negatives to any of these four? I mean, we're just hoping one of those could come to pass, right? Is there any negative?

MS. GOODWIN: If you turn to page 53, we tried to list some of the limitations of each one. Certainly, there's a point to that for the first one in terms of the legislative process, which isn't always the simplest. In terms of doing a national coverage decision, my understanding of that process is it's generally done on a technology by technology basis. So doing an across the board change or making a change to the family history definition isn't something that's generally done. Certainly correct me if I'm wrong, Terrence, on that, but it's probably something that the Medicare Coverage Advisory Committee doesn't typically proceed in that manner. It's usually done on a case by case basis with different technologies.

But maybe you can clarify whether 2 and 3 are approaches that could be taken even though they are not your typical process.

MR. KAY: I guess my comment would be that I could see it would be within the realm of possibilities. It's clearly not typical. Believe me, the agency is very interested in making sure that our beneficiaries have access to proper medical care and new technology and all that kind of thing, so clearly I would want to be in a position to rule in or out anything that you would recommend. But again, just sort of reemphasizing what I said earlier, I've been at the agency since the mid-1980s, so I've had the advantage of kind of seeing -- I mean, Medicare is usually described as not having a lot of preventive services, but it turns out that if you take a look at all the preventive services that the U.S. Preventive Services Task Force has recommended, I think Medicare has just about gotten to the point where we cover just about every one of them.

So we're very interested in making sure beneficiaries have proper care. Congress has clearly made changes, and it just seems strategy-wise, whether you're going to make recommendations for congressional change or for the agency to make a change, still folks need a comfort level of what really would it mean to make a reg change like that, because at the

agency we get the Department's clearance, of course, we have the Office of Management and Budget to deal with, and in all of these clearances we would need specifics about what would likely happen if we would do this change.

That's why I'm not saying that you should only focus on individual services, but if you want to go broader, I think it still is helpful to identify some list of specific examples to show there is a big problem here, Medicare doesn't cover these services as an example, and why care would be better if Medicare covered them. Then I think it's easier to have the more theoretical conversation about some of these things. But I think regardless of whether you go general or specific, it's helpful to have a few crisp examples of why there's a problem in Medicare and what changes should be made to improve the program.

MS. BERRY: And that gets to the point that Emily had raised earlier, which is is the committee aware of specific tests that are needed by the Medicare population that are not currently covered because of the screening exclusion? And if there are, should we perhaps identify them in this report and list them as partial justification for this change in policy? Because you're right, Terrence, I can't imagine a federal agency would just up and change their policy without any real demonstration of need based on concrete practice.

I defer to others on this. Suzanne?

MS. GOODWIN: Also, another important consideration, actually for all of them. Here you're actually talking about changing how CMS deals with preventive services in general, and certainly that's one aspect of a preventive service. But from CMS' perspective, you also have to take into consideration that in making this change for genetic tests, you're essentially opening a can of worms for other tests that might not necessarily be genetic tests but that would fall under the rubric of a preventive service.

MR. MARGUS: Would pharmacogenomic tests be considered preventive? Because that would be one you might foresee an elderly population participating in a lot more than disease screening. So if you had tests that predicted whether someone was going to have an adverse reaction to a drug or have efficacy, would that be a test that would also fall in this category of maybe not being covered?

MR. KAY: That type of service, as you describe it to me, sounds like the kind of service that Medicare could potentially cover, and we probably already do. I think we'll probably get increasingly more attention on that kind of an issue, because in 2006 Medicare will have the prescription drug benefit. So to date, we've had a limited list of drugs we cover, but as you describe it, I don't see why those kind of services would not already be potentially coverable.

MS. BERRY: There was someone in the audience. Yes, sir. Over there; does that microphone work?

State your name, please. Is it on?

DR. ROLLINS: My name is Jim Rollins, and I'm a medical officer for CMS. Some of the questions that you've been asking, Terrence has given you a lot of information that I basically want to reiterate.

One of the questions that was just asked a minute ago about pharmacogenetics, that is probably something that would be covered because it's considered diagnostic as opposed to a screening test. If a person already has the illness and tests are being done to determine whether or not an appropriate medication level or medication is being applied, yes, that would fall under what we would consider as a diagnostic test. So that's

something that would be covered.

In terms of the four recommendations, 1 and 4 would be very difficult for the agency to pursue simply because of the current mandates which are in place. As far as number 2 goes, a national coverage decision, that is something that can be initiated by an individual, an organization, a group, and we would take into consideration the evidence that's in the literature to determine whether or not there is sufficient evidence to support its use. So that would be an option to try to address this particular issue.

MR. MARGUS: Am I right in detecting that your message to us is that you'd prefer to have it case by case or test by test rather than this is being so broad stroke that it's likely to be too scary to them?

DR. ROLLINS: I think that it would have to be on an individual case per case basis, just like we look at all technologies, the same way.

MS. BERRY: Ed?

DR. McCABE: I'd just comment, though, that if there are a thousand tests and we had to do this test by test, I think part of the concern of the committee is that it could take a decade or two to work through. So I think part of the goal was to try and get, while more difficult, a broader discussion of these issues and see if more than one test at a time could be rolled into this. Is that not possible?

DR. ROLLINS: It's possible. I would also hope that in submitting those tests, perhaps the frequency or the prevalence of the particular disease within a population might be something that might be used as a means of determining which ones you may want to pursue first.

MS. BERRY: Suzanne?

MS. GOODWIN: I had a follow-up question regarding the pharmacogenetic tests. I understand that CMS is currently considering developing some sort of guidance document specific to pharmacogenetic testing, and I wonder if you could elaborate on that.

MR. KAY: We had an open-door meeting recently on a requirement in the Medicare Modernization Act to provide guidance documents, and we asked for public comment and suggestions. That was maybe two or three weeks ago. So we're still sort of in the process of evaluating our comments and we have not decided on specific documents. Clearly this is an example of the kind of guidance we might provide in the future depending on the review of our public comments.

MS. GOODWIN: But could you articulate what would be the need for that document? Would it simply clarify what pharmacogenetic tests would and would not be covered under current CMS statutes and policies?

MR. KAY: Right. Our guidance documents basically would be to provide the information to the public about our current coverage and coverage processes. It would not be the mechanism to make refinements.

MS. BERRY: Debra?

DR. LEONARD: I'd like to go back to the point that Barbara raised. Having had Barbara make that statement about being concerned about redefining predisposition of predictive genetic tests as diagnostic laboratory tests, I'm worried about number 3 because when you're doing predisposition predictive, it is not diagnostic because diagnostic by definition means there are symptoms present, and I don't want to create worries for the patient,

and confusion. So I don't know if there's a different way to word that other than redefining them as diagnostic laboratory tests. Maybe you could say redefining them as I don't know what, but that worries me. And if 2 is doing the same thing, maybe we can just get rid of 3, because they're both basically asking for --

MS. BERRY: I think 3 is just a creative way to get around statutory language. I mean, it's wordsmithing and creative argument. I think it's nothing more than that, and I agree because it certainly could open up doors that we don't want opened. If we can accomplish the same goal with recommendations, we might want to go that route. Unless anyone feels very wedded to number 3, it sounds like there's some concern about that approach, and perhaps we should focus our attention on one of the other recommendations, or a few of the other recommendations.

Does anyone have any strong support and want to argue in favor of 3?

(No response.)

MS. BERRY: Let's get rid of it. Nuke it.

Any other suggestions or comments?

Emily?

DR. WINN-DEEN: Can I just ask on the asymptomatic testing for BRCA1 and 2, was that national coverage decision that was basically brought up and put through on a national coverage decision basis, is that the way that got to be reimbursable?

MR. KAY: Unless Dr. Rollins happens to know, I would have to look it up. Historically, what's happened is that our current coverage process has evolved, frankly, over the last five years, and without knowing the exact timing of each coverage decision, I don't know what process was followed.

DR. ROLLINS: I also can't give you specifics on that, but what I can say is I think that that was one of the preventive tests which was mandated, and for that reason it was covered. In terms of having a national coverage decision for that, I don't think we have one for that. I'm not sure.

DR. WINN-DEEN: So you think it may have gotten through on the breast cancer congressional mandate where mammograms got through as reimbursable?

MS. GOODWIN: I've actually looked at this recently. There is no national coverage decision for BRCA1 and 2 screening. There are, I believe, a couple of local coverage decisions regarding that, but there may be just one or two, and those are limited to the specific geographic areas that those local carriers cover.

MR. KAY: Unfortunately, again, without looking it up in our coverage book, which we can do and get you that for tomorrow morning if you'd like. No problem. But I just don't offhand recall the history or exactly what the policy was.

DR. WINN-DEEN: I'm just trying to think about, as we go for recommendations here, if there's any test that we would put in this category that has successfully become reimbursable and covered under Medicare, then it would be helpful to know how that process happened and if it's something we should recommend as a replication process, or if there's nothing that's ever been approved, then we have to consider everything from a de novo point of view.

MR. KAY: I was going to say I'd be happy to take a minute now to try to make some calls to the agency, if that would help the committee, just so you have those facts. I'll be happy to do that now.

MS. BERRY: Before you go, can I ask you a question? Number 4, just in the process of elimination here, would this cause a great angst at CMS if we included a recommendation such as this in this report knowing that it's a very unlikely option to occur? Is there a real downside to including it in there, or is CMS recommending that it's so unlikely and worse than unlikely that it just shouldn't be included in the report? Is there a downside to that?

MR. KAY: I guess, again, as sort of CMS staff, we're not really wanting to rule anything in or out. If the committee wants to go in a certain direction, we'll give it a look. I'd say that the advice you've received in the past, it would be a tough hurdle to get the agency to agree to number 4, I would agree. That would be tough to do. 1862, the medical necessity portion of the law, is just a major issue for Medicare in determining what it does or doesn't cover. I think it could be very difficult to craft a policy change in some way that just didn't have wide-ranging implications for just about anything else. As you can imagine, at the agency we get a lot of requests for a lot of services, and that portion of the law is one of the major defining elements of what Medicare does or doesn't cover.

DR. TUCKSON: Just remember also, Terrence, that anything that we send you will come through the Secretary, not straight from us. So you've got a little immunization, if that's what you're worried about. You don't have to craft a deal for your agency right here on the spot.

MS. BERRY: Ed?

DR. McCABE: I just wanted to comment that if there has not been a national coverage decision on BRCA1 and BRCA2, I think that illustrates some of the problems that we face. The Preventive Services Task Force is a very good mechanism, but it's also quite conservative in what it considers evidence. So I think that's sort of an example and point of what the problems are. We heard this morning from individuals who have utilized this test or are afraid to utilize this test in their family. It's available, it's recognized to be beneficial to the health of individuals. So I think that would be one that I'd be very concerned if it hasn't had a national coverage decision.

MS. BERRY: Does anyone have a proposal in terms of what we would like to put forward in the report?

I'm sorry, Ellen. Did you have a comment?

DR. FOX: Just on page 34 of the report, it says, "Of the approximately 274 national coverage decisions issued by CMS, only one relates to genetic tests and services, cytogenetic analyses for monitoring acute leukemia, myelodysplasia, and congenital abnormalities." That seems to answer the question about what national coverage decisions have been.

MS. GOODWIN: And just as an addition to that, that's not a screening test that's done in symptomatic individuals.

DR. FOX: Right, so it should be genomic, I guess, instead of genetic.

DR. CHESLEY: An observation?

MS. BERRY: Yes.

DR. CHESLEY: Just in follow-up to the comment about the U.S. Preventive Services Task Force, an observation. That group also considers the strength of the evidence, as well as the strength of recommendations that it makes. I would encourage, as the group moves forward with considering these four recommendations, to underpin them as they move forward with the strength of evidence that would support them. It might allow the group

to think about levels of recommendation. If you feel strongly, for example, amongst the four in terms of how you would position them as you move them forward towards the Secretary, you might use the evidence that would underpin, for example, recommendation 2 or 3 in order to more solidify the strength of that recommendation that you move forward to the Secretary. I think, from the perspective of a federal agency, that's going to be a critical component of making the case.

DR. TUCKSON: In that regard, Muin, if you could update us just real quickly on the family history deal. Apparently, I think I'm somehow aware that CDC is doing a review of the validity of family history as a clinical tool. Given that we've already a couple of times touched on that, can we find out where it is now and whether we can be updated on that progress as well?

DR. KHOURY: Sure. Actually, two years ago -- I tried to update the group earlier, but I guess I failed to do that properly. Two years ago, we convened an expert panel to take a look at family history as a tool for disease prevention. It became very quickly obvious to the group that family history, while it's good and great and it should be in everybody's medical record, and it's the ultimate genomic test, that its validity and utility for most prevention efforts have not really been evaluated.

So we embarked on a process to evaluate family history utility and validity for six common chronic diseases, three cancers, breast, ovarian and colorectal cancer, diabetes, heart disease, and stroke. We are actually in the midst of funding a controlled clinical trial as we speak that hopefully will have some results two years from now.

Now, the discussions around family history here, especially vis-a-vis genetic testing, should underscore that these two things, while they seem to be independent from each other, they both should follow evidence-based processes. I think family history may be the easiest of the two because we all have it, we just have to remember it somehow and move forward with it. But in terms of making recommendations for reimbursement, I think the underscoring principle here is evidence-based guidelines.

To echo what Francis just said earlier, the U.S. Preventive Services Task Force, as a matter of fact, is taking on BRCA1, which they never took over something like that before. They're struggling with it because the evidence base is not in the traditional strength of something that can be brought to the U.S. Preventive Services Task Force. In a way, that's one of the impetus for the EGAPP project, because the EGAPP initiative takes into account all available evidence as a first look, as Linda Bradley suggested to us earlier, and then having sort of a pre-U.S. Preventive Services Task Force recommendations, if you will, that would put these tests and practices in play while further data are being collected and further gaps are being plugged.

Inevitably, we have great knowledge gaps in both family history and genetic tests. So, in other words, if you ask today the question how many genetic tests fulfill the rigorous clinical utility look from a U.S. Preventive Services Task Force, I would say that number is between zero and minus 1, or maybe plus 1 or plus 2. There is really nothing that comes to mind that meets those rigorous criteria of the U.S. Preventive Services Task Force.

That's obviously not good enough, not good enough for the consumers, not good enough for the researchers, not good enough for access, and what we need is a process that can do this ongoing evaluation while data are continuously collected so that gaps are plugged, and we're doing that collaboratively with all the agencies both for family history and

genetic tests, and occasionally for both. For example, with BRCA1, you can't do it independently of family history, and for most of the single-gene disorders, family history goes hand in hand with genetic tests.

MS. BERRY: Should we maybe consider amending 1 and 2 to bring home the point about evidence base? Because I don't think anyone is suggesting that we'll just add a benefit category for preventive services, boom, things are automatically covered, and the same thing with number 2, have an NCD family history, boom, it's justified, it's covered. I think what's implied there, what we probably need to just affirmatively state, is when the evidence supports coverage, CMS would have the flexibility, which they may not feel they currently have because of the screening exclusion, to cover these tests and services. I don't know if that does the trick or not.

DR. TUCKSON: By the way, as you all consider that, what we want to do just from a process check is let's resolve this issue, bring this little piece to closure, and then we haven't forgotten about the break. Then we'll take a break and do the rest. But I just wanted to give you that as an incentive to drive this point to closure, nail it right now.

MS. BERRY: Debra?

DR. LEONARD: I would think we would be sufficient in recommending a combination of 1 and 2 based on evidence. I'm uncomfortable recommending 4. It would be kind of like somebody recommending to this committee that we go talk to Congress. You know, it's just not something that's in our purview to do. So I feel with 4, we're making a recommendation for CMS to do something that really isn't part of their ability to do. So why do it? It makes our recommendations weaker. Whereas 1 and 2, a combination of those based on evidence that's out there, and that comes back to Emily's point, I'm not quite sure what's out there, and I don't know that we want to go making a blanket recommendation that all 30,000 genes worth of genetic tests be presymptomatically covered.

So I would say combining 1 and 2 with the evidence base into maybe even a single recommendation, because I don't think there's a lot of urgency with this. I think if maybe there are genetic tests in the future that predict late-onset Alzheimer's disease and late-onset osteoporosis and things that are degenerative in older people so that you could then prevent those, it might be useful, but right now I'm struggling to come up with specific tests that I would urgently want covered right now in the Medicare population.

MS. BERRY: Francis?

DR. CHESLEY: I would second that comment, and given the work that's gone into this document to date, I think it would be worth the advisory group's effort to investigate an example or a case in the Medicare population in which this would be relevant. It would certainly make the case for CMS to then do the work that you're asking CMS to do.

DR. ROLLINS: I'm not against recommendation number 2, but I think earlier someone said that the U.S. Preventive Task Force would be the underpinning of the recommendation. I have some concerns about that only because a number of their recommendations are consensus based as opposed to evidence based.

DR. CHESLEY: Just a point of fact. They're evidence-based recommendations, but what I was trying to say is rather than use the U.S. Preventive Services Task Force, simply that this group consider the strength of the evidence that supports any recommendation that they would make, not using the U.S. Preventive Services Task Force rubric, although you could use it. The beauty of that rubric, though, is that it allows you to

make a recommendation in the absence of evidence, which is different than making a recommendation when there's evidence to the contrary. So it allows you to walk through a grid in terms of the amount of evidence that exists as you try to get to a recommendation such as this.

DR. ROLLINS: I'm in agreement. I think currently the methodology which was explained in the previous lecture is something that we could use in terms of some evidence-based model for making these recommendations.

MS. BERRY: I might suggest, to go along with what Debra has offered up, actually I don't think we necessarily have to merge 1 and 2. We could leave them separate and discrete but amend number 1 to track what the legislation that has been introduced last session of Congress would do, because it does essentially merge those two things. It sets up a preventive services category but then also enables CMS to move forward with an NCD and assess the evidence and make a coverage determination.

So number 1 is a legislative fix, but it also would incorporate in that authority for CMS and weighing the evidence and whatnot. So I think number 1 amended could be that merger that you talked about, and number 2 could just be a freestanding thing that, if they choose to do so, and should we upon further reflection and after getting testimony and other public input find that there are specific examples that urgently require CMS' action, number 2 would be a separate and discrete option that could be implemented in the absence of congressional authorization.

So I'll put forward, just for the sake of nailing this down and so Reed won't get mad at me for prolonging this, amending 1 to flesh out a little bit more what the legislation would do, and making sure that evidence-based decisionmaking is part of that, that we're not just automatically proposing automatic coverage. Then number 2, the same thing, making sure that any coverage decision is based on an analysis or an assessment of the evidence clearly supporting coverage in that circumstance, and getting rid of number 3 and number 4 and just having amended versions of 1 and 2 be part of our recommendation.

Any opposed? Suggestion? In favor?

All those in favor, say aye.

(Chorus of ayes.)

MS. BERRY: No?

(No response.)

MS. BERRY: Okay, 1 and 2.

DR. TUCKSON: Great. We're going to take a 10-minute break. You all have worked hard today and you deserve the whole, full 10 minutes, so you're going to get a whole 10 doggone minutes. We stop this session at 4:30, and then we have public testimony, so that 10-minute break will be important because it only gives us a little bit of time to come back.

One thing I don't want to lose from Debra's point, and I alluded to it in my earlier comments --

PARTICIPANT: I thought we were going on break.

DR. TUCKSON: You are, you are, you are.

(Laughter.)

DR. TUCKSON: But the point that Debra made, which is key, is I think we're going to start evaluating our success as a committee, and the way in which we're going to

look at how we evaluate is how many of the things we recommend got done. So if we have a whole range of 99 things, some of which nobody can do anything about, our scorecard is going to look terrible. So Debra was right on on that. Thank you.

(Recess.)

DR. TUCKSON: By the way, I will let you know that we are quite happily aware that there are people out there on the webcast who are listening and sending emails in. They like Ed's tie, apparently.

(Laughter.)

DR. TUCKSON: But there are people out there, so be aware that you are being watched.

(Laughter.)

DR. TUCKSON: Thousands and trillions of people whom you cannot see are hanging on to your every word. So, Brad, behave.

(Laughter.)

DR. TUCKSON: With that, we're going to go ahead and march through. Apparently, we've actually only gotten through two pages.

MS. BERRY: We'll motor through this.

The next section of the report deals with the national and local coverage decisionmaking processes that CMS undertakes. The local coverage decision process obviously allows a certain amount of flexibility, taking into account local practices, it's more rapid. The national coverage decisionmaking policy does preempt local policies and has broad applicability across the country.

This one, I don't want to jinx it or anything, but I really do think we might be able to get through, Reed, pretty quickly. We don't really have too much in the way of a formal recommendation. The task force and in our previous report, we recognized that really there probably always will be and probably always should be a healthy mix between local and national coverage decisionmaking processes. No one is advocating eliminating one or the other.

But in the new Medicare law that passed, there is a section, Section 731, which requires the development of a plan to evaluate new local coverage decisions to determine which of those should be adopted nationally, the idea being to provide greater consistency in Medicare coverage policy where appropriate and where possible.

So this pseudo-recommendation, I suppose, would be simply to encourage CMS to move forward with that plan as outlined in the Medicare law, to have the ultimate goal of trying where appropriate and where possible to have more consistency in Medicare coverage policy but retaining the local/national mix.

Ed?

DR. McCABE: It's not stated in here. I think it's implicit. But given that somebody might look at the recommendation in isolation, do we want to say as it applies to genetic and genomic testing or something to that effect? Because as it reads, it really is all LCDs and NCDs, and I just think we ought to -- given the context of the report, it would obviously relate to genetics and genomics, but I think it would be good to state that in the recommendation.

MS. BERRY: Kimberly?

MS. ZELLMER: I have a question, and Terry, maybe you can answer this.

Do the Medicare policies as far as decisionmaking on these testing decisions have any influence at all over Medicaid? I realize Medicaid is largely state by state, but do they have any influence over Medicaid testing policies? Because obviously that would have a greater impact certainly on rare genetic testing, but that would involve children.

MR. KAY: At Central Office at CMS, we actually don't have a lot of detailed information on each state Medicaid program. They're basically state run. I've certainly heard that raised, that someone said that some Medicaid programs adopt Medicare policies, but I don't really have any information on that. I also just note as an example that maybe two or three years ago there were issues related to flu shots, completely different from today's issues, and there were concerns that Medicaid would be adopting some policies that Medicare had, and it turned out that was not the case at all, that the Medicaid programs maintained their own policies. So I don't think there's a direct correlation, but that's not to say that there couldn't be some states that adopt Medicare policies.

MS. BERRY: Any other comments or questions, suggestions?

(No response.)

MS. BERRY: How about if we include a revised version of this recommendation along the lines of what Ed had suggested where we do reference the genetic component. Any objection to that?

(No response.)

MS. BERRY: We're done.

The next one's going to be harder, genetic counseling. As we all know, the Medicare law, the Medicare statute, does not permit genetic counselors to directly bill Medicare, and the thinking is that that is, or certainly potentially is, a barrier to access. Reimbursement obviously, even if they could directly bill, would be limited by the other restrictions that we talked about earlier on screening tests.

We did, however, come up with a list of five potential recommendations to consider addressing that barrier with regard to genetic counseling. I'll just go through them very quickly.

Increase state licensure of certified genetic counselors. Adding genetic counselors to the list of non-physician providers eligible to bill Medicare directly. A demonstration project conducted by CMS that would examine genetic counseling, its value, and effectiveness. An Institute of Medicine study to assess the effectiveness of genetic counselors.

Then the fifth recommendation is not so much a recommendation, but more a statement of the need for consensus on which health disciplines should be providing these services, what would be the appropriate level of supervision for each, and under what conditions should they be reimbursed and should they be allowed to bill Medicare.

I'll just start off, and then jump in. The first recommendation dealing with professional licensure, I think that's a piece of a solution. I don't know if they are excluded from billing directly under Medicare. You can be licensed in the states all you want and it's not going to change that. So it's not a complete solution to that problem, but the thinking would be that it may reduce or lower the barrier to direct reimbursement, but it certainly will not guarantee that.

The question I would pose is what would be the nature of our recommendation? How do you go about increasing state licensure? There may be recommendations that would be needed, subrecommendations, under that recommendation.

The second one obviously requires a legislative fix. It would require Congress to pass a law to specifically add genetic counselors to the list of non-physician providers, and we've discussed previously that that isn't easy to do. It's not that we can't recommend it. It's not that we shouldn't. But it's not something that we should expect would happen in short order.

My only other comment would be that perhaps we might consider merging number 4 and number 5, and it doesn't have to be the Institute of Medicine, but that's something that we came up with. It could be somebody else, but the idea would be when you study and examine genetic counselors and the services they provide and their effectiveness and their value, you might as well go ahead and do an assessment of all the health professions that provide these types of services if that's the route that we want to go.

The question is do we feel that that is necessary or is that make-work? I mean, is there any question that genetic counselors provide valuable services? I think we've spoken quite a bit in past meetings about access to genetic tests and technologies as long as people have access to the appropriate counseling, that a genetic test by itself, a consumer having direct access without the requisite counseling and medical guidance, isn't the best scenario and could do more harm than good.

So I throw that out. Those are the comments that I had on those recommendations and throw that out for discussion.

Emily, Reed, Ed.

DR. WINN-DEEN: So again, I question why this is limited to Medicare. Is there something unique about Medicare that we only want to talk about genetic counseling reimbursement under Medicare or should we put this under sort of the all insurers umbrella and talk about it as a generic problem of getting reimbursement for genetic counseling? Because I think the only thing that's under the public and private thing is UPINs, which I don't think is necessarily the only issue that's coming up with genetic counseling in the private insurance sector.

So I almost think that the whole genetic counseling section should be under the umbrella of public and private, and then we can talk about if there's anything specifically unique to Medicare that we would need to make a recommendation specific for Medicare separate from a generic recommendation that counseling should be recognized and reimbursed for the value that it brings to the care.

MS. BERRY: I had the same reaction, but I thought maybe this bounced back and forth. I could be wrong, but I thought it was under the other section and there was some consideration.

I agree with you. It makes sense except to the extent that we're looking at number 2. Medicare not allowing them to bill directly is a Medicare-specific barrier, but there's nothing that precludes us, if we decided to include the genetic counseling section in the part of the report that covers both public and private, identifying what are the Medicare-specific barriers and then a potential recommendation that applies only to Medicare. I don't think we're precluded from that at all.

DR. LEONARD: My question is do we have the ability to influence private insurance companies and what they pay for and what they cover? I thought that we were directing it at Medicare because if we can change what Medicare does, then other insurers are likely to follow, but we can't mandate what they -- I mean, I don't know that we have any

influence in that arena. We can make a statement that genetic counseling services are useful and will be increasingly useful and need to be covered as medical services, but I don't think we can influence that other than through Medicare.

DR. WINN-DEEN: I agree with that, but what we have under public and private insurers is just this UPIN issue, and I don't think that that's really the only issue under public and private, that the whole issue of genetic counselors' current status seems to vary by state, some have licensure, some don't -- you know, we need to get a more unified, maybe national, level of approach to it.

DR. TUCKSON: That's a great question, Debra. I think that we've got testimony and knowledge that says that one of the barriers for all payers in this area, whether it's public or private, is the concern around who is actually qualified to do genetic counseling and what is the scope of practice for them and what is the licensure.

So to the extent that we can say that we recognize this is a generic issue for all payers, and then be able to say that while there will be some specific Medicare recommendations, but that at some point this licensure issue and definition of scope of practice issue are germane to the whole field, we could solve it that way, I think.

Which sort of leads me to my comments on this. I think that what the section doesn't get at adequately enough is what are in fact the qualifications for these genetic counselors to be able to say that this is a real genetic counselor and this is what their scope of practice is.

I think there is a lot of language around distinctiveness or effectiveness of genetic counselors, and I think that we need to be a little more precise. I think that we all understand that there is, just on the face of it, a valuable role. So that's mother, God, and country in a way, but the question is who is a legitimate counselor and what should we expect? I would urge that we spend a little more attention to that.

The fifth recommendation is also I think good in that what I think it tries to get to is also this sense of what happens if you have the genetic counselor bills for the genetic counseling and then the doc decides to bill for genetic counseling and then the advanced practice nurse bills for the genetic counseling all on the same patient? Who is the accountable entity? I mean, how do you work that out? So you've got all three of them arguing with each other around who did what for the patient. So just some thought there.

But I think the real core here is if we could maybe at least be able to describe where we are with the national standards for what is a genetic counselor, and then hope that that gets translated down to the state level, maybe that's one of them.

MS. BERRY: Ed?

DR. McCABE: These are both more really more technical editorial comments, but first of all, I like your bullets as opposed to these long -- I think it would be good, perhaps, to use those as headings, the kind of things you have on the board here, so that they're almost like the executive summary of the recommendation. They are the action item, as opposed to the whole paragraph for some of these.

Secondly, I would suggest that these be collapsed into one recommendation with parts A, B, C, D, E, perhaps, because by my count, so far we have two recommendations, or if we included these we'd have seven, five of which would be related to genetic counseling, and I think they're intertwined. So you could say something like "Because SACGHS recognizes that genetic counselors are critically important to the effective delivery of genetic

services, therefore we make the following recommendation," A, B, et cetera, to tie them together but have them really be one recommendation with subparts.

MS. BERRY: Barbara, and then Debra.

MS. HARRISON: Just to respond to Reed's comment, I think a jumping off point that we can use as to who is qualified to do genetic counseling, I think a logical answer to that would be certified genetic counselors certified by the American Board of Genetic Counseling to do such work. That could be a jumping off point.

Now, whether we identify some auxiliary people who can do that as well if they have particular qualifications, that's a discussion point, but I think at its very core we can identify the qualified people as being certified genetic counselors certified by the American Board.

DR. LEONARD: A question about that and then I have a different question. ISONG has genetic counselors that do cancer-related genetic counseling, and I can imagine that if there's a nurse that works in a particular type of genetic clinic, she or he may be qualified to do genetic counseling for a specific disease. How do you link those in without being a broadly trained genetic counselor? Because they actually may know more about that specific disease and the genetic counseling than a generally trained genetic counselor, and so their medical services are equally valid, and they wouldn't, I don't think, be certified by the American Board of Genetic Counseling.

MS. HARRISON: I certainly agree with that. I think we do have some kind of sister allied health people like, especially, nurses in oncology I think really provide quite a bit of information that we're not even always trained to give.

So in that, you know, I don't think particularly I have an answer for you, except that I can definitely see how we would need to make allowances for that, and I don't know if maybe with conversations between the genetics nurses and the American Board, maybe we can come to some agreement about that. I'm not sure.

DR. McCABE: I saw that as being covered under 3 and 4. I think that's where that issue would be considered and evaluated.

DR. LEONARD: Right. I was just concerned about the definition of -- I mean, my sense of genetic counselor is you think of a genetic counselor that does all kinds of genetic counseling, as opposed to these other allied health professionals that can do very focused types of genetic counseling, and I don't know how you bring them in, but I don't think they should be left out.

MS. MASNY: Well, just sort of as a clarification, even to Reed's point and your questions regarding nurses and the qualifications issue, is that the International Society of Nurses in Genetics does have a certification for nurses working in this whole area, both at an advanced practice level and at a basic level, trying to incorporate some of the recommendations of NCHPEG that all health professionals should be at least trained how to take a good family history. So there's a whole scope and standards of practice that are put out by the International Society sort of giving recommendations for what is the scope of practice for a nurse at the basic level and at the advanced level, and that a nurse who's actually practicing in this field could go on for certification. So then they would meet specific qualifications for certification, and the International Society of Nurses in Genetics has their scope and standards of practice approved through the American Nurses Association.

So I think that the other aspect or the first comment here about increasing

the licensure is that nurses already are licensed in their own state, so that it gives them specific qualifications. They already have scope and standards of practice in which to work, and counseling and health promotion fall under the purview of the nurses' field. So in many instances, nurses are being trained to provide some genetic counseling services.

So I think we would have to look at all of these aspects before we start to kind of specify just one particular discipline, and I think as Barbara pointed out, there is lots of work already underfoot looking at the collaborations between specifically nurses and genetic counselors.

PARTICIPANT: Are nurses reimbursed?

MS. MASNY: Nurse practitioners and advanced practice nurses in some states can be reimbursed for their services, but unfortunately, in the field of genetics, the question just comes that there's not reimbursement for the genetic counseling, is what the problem is.

DR. TUCKSON: But then that's the key thing here, I think, is that we're just basically saying, then, that how to get -- and I think this is a narrower part of the debate, and that is simply we're not so much interested in who can do the counseling as who can bill independently for their services, and I just don't see, quite frankly, any way that anybody is going to allow somebody to pay a bill for a professional service if there is not sufficient evidence that that person is qualified to act independently of anyone else and provide that service. I think that it's going to be a tough row to hoe.

So I think our decision that we have to make here is whether we call for this certification, as it were, to become clarified and acknowledge that that's the rate-limiting step and then sort of call for that to get fixed, or whether we can domino over that, jump over that, and say, okay, in the absence of that certification, we recommend that you just pay people. That's what I see the ultimate argument boiling down to here.

MS. BERRY: Suzanne?

DR. FEETHAM: My comments build off of everything that you said, and that's what struck me, Reed, building off of what you just said, as I read this, is the beginning of the document focuses on access and we've talked about primary care as a point of entry and that genetic counseling and genetic services are fundamental to what is needed and the reimbursement is a major piece of it. So I would encourage, and the theme I'm now hearing, is that there's the genetic issue of access to genetic services, of which genetic counseling and bottom-line reimbursement is the basic line that, rather than focusing on just a discipline, it seems like we've jumped a whole lot and lost some of the basic essence we've been talking about in our other meetings.

MS. BERRY: Emily?

DR. WINN-DEEN: So I guess the question is do we want to encourage that there is some mechanism put in place that recognizes that certain licensed health professionals -- let's just leave it open for right now -- do provide genetic counseling, that this is valued, and should be reimbursed, because certainly when you go for a genetic counseling session, this is not an office visit brief. This is generally an extended interaction and maybe even multiple interactions by the time you get through a couple of counseling sessions pre- and post-testing.

So we want to make sure that that's valued and that those people are being properly compensated. So the question I guess is do we need them to be separately billable or is the umbrella, you know, a physician supervising them the same way that the nurses in the

physician offices are typically part of the "overhead" rate that physician charges? The nurses aren't billing you for the time that you spent with them getting your height and weight taken and your blood pressure and all that.

I think we need to be very concrete about what it is that we think as a committee we want to recommend, keeping in mind that we all understand and I think there's consensus that genetic counseling is a valuable service and that it needs to be part of the continuum as appropriate for the severity of a specific condition.

MS. BERRY: Brad?

MR. MARGUS: So these all seem like no-brainers to me. I think we all agree. I don't think we disagree. It's absurd that after all these sessions we've sat through that we would think that genetic counselors can't even find a box on that thing representing them.

That's a given, and then there's this more complicated thing. We've heard testimony from people who have come and talked about physicians or nurses that couldn't bill for their time. So they were running people out of their offices instead of providing counseling.

But all that being said, I appreciate whoever's idea it was to stick the couple of last bullets on where you're going to build a case for it because, to play the greedy businessman for a second, I still think you're going to get pushback where someone's going to say tomorrow we're reimbursing this whole new class of people called genetic counselors, a different species, and then we've also got a whole new category by which nurses and physicians can bill, and even if they did only the same amount of counseling that they're doing today -- a paltry amount that isn't enough, but even if they are, how much more does it cost, and then now, if the floodgates are open, how much could it really amount to that this could end up costing the system?

It's pretty daunting. I can see people justifying needing, yes, days with patients to walk them through genetics, and there's a real concern about that kind of cost and people are going to just push back completely.

So I'm 100 percent in favor of the first three, but the last two, and I don't know if the IOM -- and the big question there is, and those of you with more history on these committees can maybe say it, but maybe this has already been done numerous times, where economic modeling has been done and people have figured out what the impact would be, but if hasn't been or hasn't been done recently, it seems if you don't build a better case for it from a very cold economic point of view and quantity of care point of view, you're not going to ever sell it.

That's the scary part. I always try to think about what could the other side be thinking, as you did earlier with Congress, and why would anyone be opposed to genetic counseling if someone's got a devastating possible diagnosis or risk suddenly and why can't that be part of the treatment? The scary thing must be that there's this whole new area that people are afraid might end up costing an awful lot and it might not be properly regulated or there aren't enough standards. You know, have standards been set up that if you have this and this possible genetic risk now, you're entitled to 1.7 hours of counseling? I don't know how it works, but have those things been put in place?

People are going to be concerned that you're putting the cart in front of the horse if you haven't figured all that out, and so I'm putting a plug in for the last two, which seem to be premised on building a case for it.

MS. BERRY: Related to that, I wanted to ask and get some input on

number 3, which calls upon CMS to conduct a demonstration project, and theoretically that could be part of, Brad, the idea of building a case. I question whether that's the most effective way to build the case or whether an assessment, a comprehensive study, is the most effective way to build the case.

MR. MARGUS: That one just felt really slow to me.

MS. BERRY: Sorry?

MR. MARGUS: That one felt really slow to me.

MS. BERRY: Which one?

MR. MARGUS: Number 3, compared to number 4.

MS. BERRY: So that's why I asked. Is that something that we feel is effective? They're not necessarily mutually exclusive, but if one is more effective than the other and if the goal in the end is the same, which is to build a case and demonstrate that there is this service that is being provided or should be provided, that it's a value to people, it helps improve access to care, and improves the quality of care, what is the best way to achieve that result? Is it through a limited demonstration project in a few cities or is it through a more comprehensive look, a la an IOM assessment?

Yes?

DR. FEETHAM: Just as a reminder, I forgot to mention, remember Dr. Judith Cooksey presented last year on the study being funded through HRSA on genetic services and with counselors and other genetic specialists, and part of that study is winding down or is about at its end, but just a reminder that here we're moving forward suggesting additional activities when you may be having data from that that would help address some of the things that you're talking about.

MS. BERRY: Francis, did you have a comment?

DR. CHESLEY: Yes, and reflecting on your question, it occurs to me that what the IOM would do is look at extant evidence. So if there is evidence to demonstrate the effectiveness of such services, they will be able to put that in a neat package with a bow and present it to you. If the question is demonstrating the effectiveness through a demonstration study, that comes at it from a different perspective. Those data don't exist. We need to do a study to generate those data, which is slower.

So I think the fundamental question for the group is whether or not those data exist already -- a quick query on that and that's easy to do -- and if those data exist, that points a direction in terms of getting the underpinning for such recommendations.

MS. BERRY: Emily?

DR. WINN-DEEN: So I see sort of two things. One is about Recommendation 1, which sort of deals with the whole who's qualified issue, and I think we could make a very specific recommendation that's sort of no-brainer. Anyone who is certified with some bullet point list of organizations should be qualified to deliver genetic counseling. This doesn't say get paid for it, but at least that is our endorsement, if you will, of those programs as providing some specific level of training that gives people the right qualifications.

The next question is this whole direct billing, and I think it's almost going to be impossible to justify direct billing without having a specific set of data to support that.

So I'm less inclined to try and go for number 2 or put that as sort of the third level. So the first is identify what we believe are legitimate qualification programs, if you will. The second is where's the data that shows that this service has value, and the third then is

what's the best mechanism for reimbursement? Is it still under the umbrella of the physician or should they be allowed to separately bill for some specific services?

I don't think anyone who sits down and chats with you and gives you informed consent forms to sign should necessarily be billing, but I do think that people who spend their lives working on counseling people should be able to get recognition and compensation for that.

MS. BERRY: Debra?

DR. LEONARD: That was one of my concerns, is where does this go when we move toward genomic medicine? Because we think of genetic counselors now more with single-gene disease counseling and as we move more toward genomic medicine, where you can do genetic counseling for asthma and hypertension and congestive heart failure and who knows what, will that require the same level of genetic counseling and we're imagining that that will be done by primary care physicians potentially? And then we get back into the education issue, and if people aren't educated, then can they -- but in the current health care economic situation, anything else you can bill for would be highly valuable because you could get more money for the undercompensated services that we currently provide.

DR. WINN-DEEN: Right.

DR. LEONARD: So would this end up being a mechanism that would be used?

DR. WINN-DEEN: Correct me if I'm wrong. I thought PCPs really have a mechanism, although it may still be inadequate, because they can bill for office visits of varying duration. So if they are providing that discussion with the patient -- about anything, about cancer, about heart disease, about a highly-penetrant genetic disorder -- that they have a mechanism to be recognized as qualified and to get paid for the time they spend with someone. That would be really sort of focusing on the counseling part, where the counselors right now are not either officially recognized or officially able to bill and directly get compensated.

So I don't want to just miss the fact that PCPs are hopefully going to provide a lot of this counseling and certainly, in some situations, and I'd say probably in the cancer scenarios, the referring oncologist is probably providing a lot of the counseling.

DR. TUCKSON: Well, there's a question that we're really entering a whole new area, and I'm sure primary care physicians are frustrated with how much they are able to get reimbursed for cognitive, as opposed to procedural, oriented services. That's one of the tough challenges about being a primary care doc. You don't get to do stuff and the more stuff you do, the more you make, and the more you talk, the less you make.

So there's a certain imbalance in there, but I think that where I sort of hear Debra and Emily going here, and also actually Brad's point, was if you sort of fast forward this thing, when genetics is the essence of medicine, basically then it means almost every encounter ultimately is going to have a genetic component. If this family history deal is a terrific success, then by the day after Thanksgiving, everybody is into their doc's office talking about Cousin Sue and then what do I do? And then, well, does the doc sit there and chat with you or do you refer that out like you would do a nutritional consult and expect that somebody would need to be compensated for those services?

You've just opened up a heck of box, which I don't think we're going to solve, and so I think what this really boils back down to again is, and I'm just putting something out there as a way of giving you something to shoot at for the report, do we acknowledge that

this a big issue, this independent reimbursement?

Well, first of all, as you went through, Emily, that first we do support and recognize that it is important to have genetic counseling being done? Not only for all the good reasons that are mother, God, and country, but Brad, for the business reason, is in the absence of it you're going to get a lot of inappropriate testing, which costs a whole lot of money to people.

So genetic counseling, done properly, ought to decrease health care costs, not actually increase it in the aggregate. That would be the hypothesis. So we ought to encourage that.

Number 2, though, is the independent billing, we might have to acknowledge we cannot solve that problem in the absence of certain things that must be in place, and therefore, given that this is important, we would urge the logical steps to get these things in place.

I think finally, to be more complete than we are here, would be to acknowledge those efforts that are ongoing now to resolve this matter. I still have to go back and reread, I guess, the testimony from the counseling community. I just don't remember what else is going on to get national standards and get them implemented. I can't imagine that it's just laying dormant as an issue. Somebody's got to be working on this.

We might need to reference the work that's going on and then finally add to that the call for some of these demonstrative studies that we think need to be done to finally get that done. Then when that body of work is complete, then the issue of independent billing may be able to be addressed and we will take that up as events progress. That's one way to get at it.

MS. BERRY: Barbara?

MS. HARRISON: I just want to at least acknowledge that two states, Utah and California, have put through their state legislature processes for licensure for genetic counselors, and also there is an effort -- you're right, it's not laying dormant -- in order for genetic counselors to be able to be assigned UPIN numbers in I think 2007 when that process goes through.

But importantly, I guess I'd just be very curious to see what this final wording is because I think licensure without a strong argument for billing behind it doesn't mean very much. You know, if we're licensed, then fine, but I think it's been made very clear, as in previous testimony, that counselors are doing this job now and they're not getting reimbursed, and that's a threat to the genetic counseling community, to the patients, et cetera. So as self-serving as it may be, I think it's very important that we don't just say, well, we can't do anything about the billing, so we'll just make sure licensure is in place.

DR. TUCKSON: Well, I think that's really important. I think we need to acknowledge that explicitly as part of our narrative, that without reimbursement -- it's almost like which comes first, the chicken or the egg? One thing, what I've been saying is I guess without the licensure, it's going to be hard to get reimbursed. I think we ought to give equal credence to the fact that without reimbursement, who cares about licensure, is basically I guess what it comes down to, and we don't want to kill off or stifle the growth in the number of counselors, given that we have too few now.

I think also your point about some of the examples that are going on, hopefully we could appendix this in the report because I think probably very few people are aware of these groundbreaking initiatives in a couple of states and we ought to appendicize

those to try to move this forward.

So I guess, Barbara, where I would agree with you, and at least as one person's opinion about this, is we do want to move this faster and I don't want it look like we're putting a lot of steps in place that look like we're not seeing the urgency of it. So I think we ought to be urgent and give examples of concrete things that we ought to build upon.

MS. BERRY: Brad? Brad, and then Debra.

MR. MARGUS: Just as the complaints right now about the discrimination bill are that not enough people are being harmed, I think it would be great if we could also point to things that aren't right because genetic counselors aren't reimbursed.

I mean, you keep having Congress up there. Congress needs to respond to a problem that won't go away, and as long as it's not a very noisy problem right now, I don't think they're going to react. If we could point to serious problems and damage that's being done because people aren't getting counseling, and I'm sure there are as many good anecdotes for that as there are for discrimination and maybe more, it would be probably very helpful in stating our case.

DR. TUCKSON: Cindy, could you try to just give us, then, given that we've got to stop here and -- not got to. We're excited to stop and turn to public testimony in the last half-hour. We've given them each of them their amount of time and we're supposed to stop at 5:00. Can we challenge you right now to sort of frame where you think we might be and maybe just start right at this point tomorrow morning bright and early? But give us something to sort of help to see whether we need to noodle over this a little bit more tonight and then come right in and attack this or do you want to put this part of the report to closure right now?

MS. BERRY: I think maybe we should rework based on this discussion. I think we can merge some recommendations. I also think that what I heard, and if I did hear it correctly I agree with it, that number 2, the congressional component, is probably premature, given that we're calling for these other things as sort of a prerequisite. So we could maybe take that one out. Maybe we could noodle around and come up with some revised recommendations and put them up on the screen first thing in the morning. I'll just get with Suzanne and others and we'll try to come up with something, and then hopefully, with the limited amount of discussion tomorrow morning, we can close that out.

DR. LEONARD: But while you're doing that, I think the recommendation on page 61 about the UPIN, I mean, if we're going to talk about reimbursement, that's kind of stuck off there by itself. Can that be incorporated into these genetic counseling recommendations as a whole?

And I really don't agree with dropping number 2 because I really think that if there is licensure, there should be payment and they have to be recognized and they have to get UPIN numbers in order to get paid. So I don't think we can take 2 off, but maybe make that that basically once there's licensure, that's the state saying these people are qualified to do this medical service that they're doing. They should get paid for it.

DR. TUCKSON: All right. Well, Cindy has got some stuff to look at tonight, and we'll watch and see how many glasses of wine she has at dinner and see what she's able to do.

So thank you for that, and Cindy, thank you for volunteering to take even more responsibility.

Let me thank the committee for your hard work so far today, and now let's

turn with great attentiveness to the public comment. One of our critical functions is to be able to receive input from the public, and we appreciate the views that they're sharing with us. We also have received written comments that can be found in your table folders. In the interest of time, commentators are to keep their remarks to five minutes, and as I said, we do have your written testimony which we will be looking at very carefully.

Let me say that I've got right now on my list first Kelly Ormond from the National Society of Genetic Counselors -- well timed -- Sharon Terry from the Genetic Alliance, Miriam O'Day from the Alpha-1 Association, Gary Martucci from Myriad Genetics, Christine Broderick from the National Partnership for Women and Families, and Donald Horton, director of public policy and advocacy for Laboratory Corporation of America. I'm sure that Sarah will get me if I've missed anybody else.

Let's start with Kelly Ormond from the National Society of Genetic Counselors.

MS. ORMOND: Thank you. It's a pleasure to be speaking here today. I'm Kelly Ormond, president of the National Society of Genetic Counselors.

As you are aware, the NSGC is the leading voice, authority, and advocate for the genetic counseling profession and represents over 2,000 members. Together, our members provide genetic counseling for prenatal, pediatric, and adult genetic indications, as well as work in academia, research, and biotechnology companies. A high percentage of our clinically practicing members offer some form of predispositional genetic testing on a regular basis, whether carrier testing or presymptomatic testing, for adult-onset disorders.

Today, we would like to primarily address two issues related to the provision of genetic services, genetic discrimination, and coverage and reimbursement of genetic counseling services.

NSGC would first like to address the issue of genetic discrimination by employers and insurers and the related topic of genetic nondiscrimination legislation. We have testified on this issue at past SACGHS and SACGT meetings. NSGC has also provided testimony to other organizations, including the National Conference of Insurance Legislatures in February and July of 2004, and is an active member of the Coalition for Genetic Fairness. We've also recently collaborated with FORCE, a cancer advocacy organization, to develop an educational brochure on genetic discrimination.

Our organization is disappointed that Senate bill S. 1053 was not taken up by the House for discussion in the past year and we are committed to working with all stakeholders to develop policies that are equitable and fair to the American public.

We would like to address three points with regards to genetic discrimination, beginning by reflecting upon the current status of documented genetic discrimination. It is clear that there are few documented cases of genetic discrimination in either the insurance or employment setting, but the oral testimonies this morning, written testimonies, and cases presented in other resources, including the "Faces of Genetic Discrimination" booklet published by the Coalition for Genetic Fairness, have reinforced that it is clearly an ongoing problem for at least a small percentage of families with inherited disorders.

In a paper that is currently in press, 7 percent of survey respondents at risk for colon cancer perceived that they or a healthy family member had experienced genetic discrimination based on genetic testing or family history. These reports were primarily around

difficulty or denial in obtaining health or life insurance coverage or in denial of screening coverage. It remains unclear, since the bulk of these anecdotes remain unpublished, whether the individuals are experiencing discrimination due to a specific disability as compared to discrimination occurring solely based on genetic status and the extent to which discrimination may or may not be occurring.

Second, regardless of the rate at which genetic discrimination occurs, data suggests that individuals want to keep their genetic information private, as they do all health information, and that individuals are afraid that they will be discriminated against on the basis of genetic information. As a result, the topic of potential genetic discrimination is frequently discussed in genetic counseling sessions. As was reinforced by the health professionals panel earlier, this is usually brought up by the client, rather than the genetic counselor.

We have also heard that data, further backed up by published studies, suggests that a proportion of individuals who are candidates for genetic testing and for whom medical management may be changed based on test results, declined testing based on this fear of genetic discrimination. Specifically, two recent studies document that nearly half of surveyed individuals are highly concerned about genetic discrimination. This fear may result in at-risk individuals declining genetic counseling as well as genetic testing, undergoing testing using an alias or in an anonymous manner, not billing health insurance for genetic testing, or obtaining life insurance or other policies prior to undergoing genetic testing.

There are also studies that document that a high percentage of individuals at risk for breast or colon cancer do not tell their physicians or insurers about these risks or that they ask that the information not be recorded in their medical records. Such behavior certainly has personal and public health implications on medical management if individuals do not undergo early screening or if they choose not to share genetic test results with health care providers.

While education through the media and health professionals will be useful in minimizing the perception that genetic information is different than other personal health information, fear related to genetic discrimination appears to be pervasive.

As we discussed this morning, it seems clear that both state laws specific to genetic discrimination around health insurance and/or employment discrimination and the federal ADA, HIPAA, and civil rights statutes may not be comprehensive and that there are gaps between state legislation which become relevant in our highly mobile society. One critical point that was not discussed earlier is that research data suggests that neither primary care providers nor the general public are aware of the potential protections these bills provide.

As was noted in our 2002 physician statement, "The NSGC opposes discrimination against an individual with regards to eligibility for or maintenance of employment, insurance coverage, or medical benefits on the basis of genetic information. Genetic information includes the results of genetic testing, other tests which reveal genetic information, and information gathered upon review of family history. Consideration of this information is appropriate only when used to protect the individual's best interest."

While the NSGC does not support a position of genetic exceptionalism, we strongly support the passage of federal genetic nondiscrimination legislation. Such legislation would likely alleviate the majority of concerns regarding genetic discrimination and allow members of our society to use genetic information to help clients make informed medical and personal decisions. To quote Paul Miller from a publication several years ago, "Whether it,"

genetic discrimination, "is a huge problem or a small problem, it should be prohibited."

DR. TUCKSON: Terrific. Kelly, let me ask you to do this.

MS. ORMOND: Can I have one more minute to just finish really quickly?

DR. TUCKSON: Can you do it in a minute?

MS. ORMOND: Yes.

DR. TUCKSON: Go for it.

MS. ORMOND: In summary, NSGC supports federal legislation for genetic nondiscrimination and we are available to work with SACGHS to further this matter until such legislation is passed. We are also committed to working with SACGHS and other medical and public policy organizations to educate the members of our society regarding the key issues around genetic information and privacy and to address the misconceptions which have unfortunately become prevalent.

Finally, I would like to state that in our quest to improve the access of our American society to high-quality genomic medicine, it is critical that this committee consider not only the need to decrease risk of genetic discrimination, but also ways to increase access to both high-quality and affordable genetic services. As such, I would like to conclude by addressing the issue of coverage and reimbursement of genetic counseling services, primarily addressing services provided by masters-trained genetic counselors.

Issues of billing and reimbursement are among the most pressing that face members of the NSGC and it is one of the three areas prioritized in our recent strategic plan. Through our past testimonies, this committee is already aware that coverage and reimbursement for genetic counseling services are limited by the lack of CPT codes and ineligibility for non-physician provider identification. While some payers contract directly with the health plans to include genetic counseling as a covered service and some services are covered by Medicaid and Medicare when provided to individuals with disabilities, the bulk of genetic counseling services are not currently reimbursed.

While we have only preliminarily reviewed these newest draft recommendations, NSGC is pleased to see that SACGHS and the Secretary's Office consider ways to address these two points. In particular, we are heartened to see that SACGHS is promoting the development and funding of evidence-based studies around clinical genetic services through any agencies. NSGC offers its strong support in developing and conducting such studies and have repeatedly been told that studies documenting such value will be critical. We are also pleased that SACGHS is continuing to advocate for the inclusion of masters-trained genetic counselors as recognized providers in both private health plans and national provider identification systems.

If NSGC can be of additional help as SACGHS works on these issues, including offering formal testimony on our efforts towards licensure or documentation of the value of genetic counseling, please do not hesitate to contact us.

DR. TUCKSON: Are you going to be around tomorrow, by the way?

MS. ORMOND: I'm going to be here until 1 o'clock, and I'm happy to address any of these issues at that time.

DR. TUCKSON: Great. Well, I really want to thank you, first of all, for coming and also for getting your stuff on the record and reading as quickly as you did.

MS. ORMOND: Well, I try.

DR. TUCKSON: You're very good.

MS. ORMOND: I'm sorry. It was long.

DR. TUCKSON: That's all right. I'm the one that's in trouble. But thank you for that, and actually, you heard the discussion earlier and we can't stop for questions, but if you all could amend your testimony based on what you've just heard us go through, and if you want to really underscore that, we would really much appreciate even more specificity based on what you just heard. Thank you so much.

MS. ORMOND: Thank you.

DR. TUCKSON: Sharon Terry from the Genetic Alliance.

MS. TERRY: So I'm going to help you out, Reed, and I told Kelly I'd give over some of my time to her. I'm only going to be three minutes.

I'm Sharon Terry, president and CEO of the Genetic Alliance, which is an alliance of 600 genetic disease advocacy organizations that represent 14 million individuals. The Alliance is a founding member of the Coalition for Genetic Fairness, a coalition of organizations working together against genetic discrimination.

We understand the promise of basic and medical research and are appalled that many families and individuals experience genetic discrimination and fear both knowing their own risk and participating in research as well. You have heard some of their stories here today. The Coalition's "Faces of Discrimination" provides you with more stories. We believe that all genetic information, including family history, deserves strong protections against misuse in health insurance and employment. Such safeguards will protect the rights, privacy, and confidentiality of the individual and their family.

This is an exciting and hopeful time for medicine. It is imperative, however, that we the public take full advantage of new medical advances that could help prevent disease before it develops. Genetic nondiscrimination legislation will reduce the likelihood of genetic information being misused in health insurance or employment decisionmaking. As you well know, simply having a positive genetic test does not mean one will develop a disease. Thus, this information should not be used to make decisions about insurance coverage or employment.

You have heard here from both consumers and professionals that as biomedical research advances, genetic testing is a critical tool in the provision of health care. As a result, many people know their own genetic makeup, putting them at risk of genetic discrimination. People who would like to avail themselves of genetic testing already have enough to worry about. They should not have the additional burden of genetic discrimination.

In addition, it is important that we who carry mutations for diseases are encouraged to participate in genetic research. A fear of discrimination discourages that participation, adding another hurdle to the pathway from basic science to health care services.

The Genetic Alliance and the Coalition for Genetic Fairness have worked for years on this issue. In the past year, we presented a letter to Speaker Hastert signed by hundreds of organizations and hundreds of individuals. We held a press conference with Heidi Williams and Dr. Collins on Capitol Hill. We continue to work together on this issue and plan to go forward until legislation is passed in a spirit of cooperation and compromise.

Therefore, on behalf of millions of consumers and advocacy organizations, I convey to you our strong support of genetic information nondiscrimination legislation. The Senate passed 1053 95 to 0, as we have heard, and President Bush has said he will sign it, as we have also heard. We've come here today to ask that you be bold and clear in your

communication with the Secretary. Please ask the Secretary to ask Speaker Hastert and Representatives DeLay and Barton to move this legislation. Public policy must keep pace with scientific advances and provide those advances with a climate conducive to their translation into health benefits for all.

Thank you for the opportunity to bring these voices to the table. Thank you also for your leadership. We need you in this fight.

I will submit a much longer document and a copy of our letter and the signers to you in writing.

Thank you.

DR. TUCKSON: Well, that's just terrific, Sharon. Thank you.

You did so well and you only did two and a half minutes, so let's take a couple of questions.

DR. McCABE: Sharon, you said I think 14 -- one, four -- million people? I couldn't tell whether that was 14 or 40.

MS. TERRY: Fourteen. One, four.

DR. McCABE: Fourteen million. So 14 million. That seems like a lot of people.

We've been told that genetic discrimination doesn't occur. We had limited resources in terms of time to hear anecdotes. I'm sure we'll be told that this was just seven anecdotes or it doesn't occur. I would ask you to please document and help assist us as we accumulate this documentation. Among those 14 million, I hope we could find more than seven anecdotes, so that we could document that this is in fact a serious problem that does occur.

DR. TUCKSON: Terrific.

Anyone else? We're at three and a half minutes.

(No response.)

DR. TUCKSON: Great.

Would you also, in terms of what you submit back, because you had prepared your comments, because you're always prepared, that you heard our discussion this morning and I think you hit one of them for sure in terms of what we're going to do, and that's the three members of the House that we need to prioritize. If you have any other things that come up as a result of our conversation, I'd ask you to be a little more flexible. Take what you've already sent in and add to it. That would help us out.

Thank you so much. Really appreciate your being here.

Miriam O'Day from the Alpha-1 Association.

MS. O'DAY: Yes. Good afternoon and thank you for your time today. I have submitted to you a copy of the Alpha-1 Association and Alpha-1 Foundation statement. You have heard from a patient. She has talked about her children's carrier status. You heard from a provider today. My testimony I hope will sort of round it out from the organizational perspective of organizations that serve individuals with alpha-1.

Certainly, we believe that alpha-1 is a very good model for discussion on genetics, health, and societal issues. As you know, it's a pediatric liver disease. It's pediatric cirrhosis. It is the second leading cause of transplantation in the pediatric population. It's an adult-onset lung disease. In fact, it's genetic emphysema. It is treated in end stage with lung transplantation. Lungs are now going through an allocation process and we feel that

individuals with alpha-1 are being disadvantaged in that allocation process. So we have a huge battle and barrier there.

It also has, as you heard Dr. Brantly discuss, a very large environmental component, and in fact alpha-1 is related to the fourth leading cause of death, chronic obstructive pulmonary disease, and there is a belief that COPD is going to have very strong genetic components and alpha-1 is the first identified genetic component.

The organizations that I'm here representing have taken responsibility for finding their way around the labyrinth of the patchwork that exists in state legislation, and in absence of federal protective legislation, what we have done is we use an ELSI Working Group. We have invested heavily in bioethics, and in fact we've had our ELSI Working Group take a look at the question of neonatal screening and population screening, which has been suggested by some of the physicians who treat these patients. In fact, our ELSI has felt that it's not ethical to do so in absence of protective legislation, and so what we do is we do targeted screening and detection. So this has seriously impacted our ability to identify those 95 percent of patients with alpha-1 that are unidentified.

In addition to that, and I've shared with you a copy of our ACT trial brochure and it's been handed out to committee members, our ACT trial, which is funded by the Alpha-1 Foundation and conducted at the Medical University of South Carolina, offers a free and confidential finger-stick test that can be completed at home. The results are mailed directly to the individual participant. Since 2001, the ACT trial has done 2,400 test kits. We have shared this protocol with NIH at their request. We've been very pleased to make it as public as possible.

The test is administered through a research study that evaluates the perceived risks and benefits of genetic testing. It does that through sending out a follow-up questionnaire. In that questionnaire of the people who have responded to our survey, we found that over 30 percent report fear of losing insurance as a reason for seeking confidential testing, 34 percent report concern about facing higher health care costs if the results were made public, and 85 percent seek testing for the genetic knowledge. In fact, that was the most popular response.

We would be very pleased to provide you with any additional information that you'd like to have as a committee and we strongly endorse the need for federal protective legislation.

Thank you very much for your time.

DR. TUCKSON: Terrific. Thank you.

We have about 20 seconds for a question.

(No response.)

DR. TUCKSON: The one thing I want to add from what you've done that we didn't explicitly have in our earlier testimony, as I wrote down a little list of all the issues that come out the concern around discrimination, we had the HIPAA issue come up, we had the chilling effect it has on research, but the just overall screening policy was not a category that I recall was explicitly stated. So I would urge staff to add that to the list of chilling consequences of the absence of this, screening policy, and here we've got an organization that feels ethically it cannot recommend screening in the absence of this kind of thing, and I think that's a very important category that you've brought to us.

MS. O'DAY: Thank you.

DR. McCABE: And I would ask, because that's something different than we heard this morning, that perhaps it be added to this packet of material that we will be forwarding also.

DR. TUCKSON: Yes, yes, a very, very nice contribution.

MS. O'DAY: Thank you.

DR. TUCKSON: Gary Martucci, and I hope I'm saying that right.

MR. MARTUCCI: It's Martucci. Correct, yes.

DR. TUCKSON: And you are, by the way, with Myriad Genetics.

MR. MARTUCCI: I'm with Myriad Genetic Laboratories, correct.

Good afternoon. I think what we've heard today is that it's clear that the fear of genetic discrimination is preventing high-risk patients from accessing appropriate care.

Myriad began providing clinical and genetic testing for common hereditary cancer syndromes eight years ago. In 1996, two of the greatest barriers to genetic testing were the fear of discrimination and the unknown rate of reimbursement from insurance carriers. Since 1997, I have been responsible for securing coverage and reimbursement for genetic testing from health insurers and managed care organizations nationwide. Myriad's experience is such that genetic testing for common hereditary cancer syndromes is paid by insurers 90 percent of the time at an average of 90 percent coverage. Therefore, the insurance coverage barrier has effectively been eliminated.

However, the fear of discrimination has not been. For almost eight years, I've had the opportunity to discuss genetic services with hundreds of medical directors, physicians, and patients across the United States. The concern and fear about discrimination arises in virtually every discussion.

To reduce anxiety around genetic discrimination, Myriad has implemented a policy that patient test results are not released to anyone except the ordering health care provider or designee without the patient's express written consent. Insurance plans representing approximately 200 million covered lives comply with this policy because they recognize the clinical value of cancer genetic testing, which leads to the most effective medical interventions.

Our policy, along with numerous state and federal laws that prohibit employment and health insurance discrimination, results in numerous protections for consumers of cancer genetic tests. Yet there still remain gaps. The fear of genetic discrimination remains the most commonly cited reason for both patient and provider not to utilize genetic services to prevent life-threatening cancer.

We find ourselves in an awkward place. A large body of literature demonstrates the benefits, both clinical and psychological, of cancer testing. While peer-reviewed literature suggests that actual genetic discrimination is not a significant problem, the media continue to portray genetic discrimination as a common risk to individuals poised to take advantage of the health benefits offered by genetic services.

Roth, et al., echo many experts' opinion when they state "Unless these people believe that they and their families will be adequately protected from discrimination and from the possibility of losing or being denied health insurance, many will choose not to be tested for genetic conditions or predisposition to disease." Therefore, comprehensive legislation is a necessity or the media and other uninformed stakeholders will continue to use the fear of discrimination to dissuade patients from appropriate health care.

In the arena of hereditary cancers, genetic services and testing offer the

hope to reduce the burden of disease that many families suffer. Fortunately, tens of thousands of individuals have benefitted from the power of genetic tests to guide their providers in the most appropriate medical management. While this number may seem impressive, there are over a million in the United States who carry mutations predisposing them to cancer, yet fewer than 2 percent know it.

While we know there are several key issues that underlie these statistics, such as a lack of awareness of genetic tests and the need for additional education and clinical support to health care providers, we consistently found that the fear of discrimination is a top reason for refusing genetic services and testing.

To integrate the promise of the Human Genome Project into clinical care, patients, clinicians, and insurers need the best available information to coordinate medical management. Without the information available from genetic risk assessment, patients and health care providers are left with only limited knowledge of how best to manage the risk of disease. Not only does this dilute the benefit of medical management for the patient, it often results in poor allocation of resources. Truly high-risk patients may not pursue risk-reducing options, while the very low-risk individuals may in fact overutilize the medical system due to their fear of disease.

It is our experience that patients interested in obtaining potentially life-altering genetic services sit idle in fear of discrimination. The science and technology to positively influence a patient's outcome are with us today. It is our responsibility to make sure that patients are confident that there will be no negative consequences in insurance or employment for pursuing this important information.

Perception is reality, and the public's perception is that genetic discrimination is a serious threat. People have allowed an essentially nonexistent or limited risk for discrimination to prevent them from managing a very real risk of developing cancer. We must eliminate the fear of genetic discrimination to allow the public to participate in the benefits of genetic medicine. Comprehensive legislation will reassure the public and get media coverage to spread the word. Comprehensive legislation will eliminate the confusion and mixed messages sent to individuals who need these technologies the most.

Ladies and gentlemen of the committee, comprehensive legislation prohibiting and banning genetic discrimination is the answer.

DR. TUCKSON: Thank you very much. Right on the money. Thank you so much.

Christine Broderick from the National Partnership for Women and Families.

MS. BRODERICK: Good afternoon. On behalf of the National Partnership for Women and Families and also the Coalition for Genetic Fairness, thank you for holding this hearing to gather information about the scope and nature of genetic discrimination.

The Senate has taken an important step in advancing genetic testing and research by passing the Genetic Information Nondiscrimination Act. This is legislation that would provide much needed protection for all Americans from genetic discrimination in health insurance and in the workplace. We join with you in the hope that the evidence presented today and also in the data that was noted in the accompanying articles will encourage the House of Representatives to take action to enact this legislation.

With the completion of the Human Genome Project last June, the possibility for genetic testing and research is expanding rapidly. There are now genetic tests for hundreds

of disorders and some of the most widely available tests are for women. Women and families stand to benefit from improved prevention, detection, and treatment of diseases like breast and ovarian cancer.

However, all the advances in the world will not help women and families if, by participating in genetic research or taking a genetic test, they can or fear they can be denied job opportunities, health care, or both based on their genetic information.

In addition to being long-time proponents of genetic nondiscrimination legislation, the National Partnership for Women and Families leads the Coalition for Genetic Fairness, a diverse group of disability, women's advocacy, and civil rights groups that recognize the need for meaningful protections against genetic discrimination. The scope of this group reflects the impact that this issue has on all Americans.

To illustrate the impact of genetic discrimination and the fear of genetic discrimination, the Coalition evolved a report, "Faces of Genetic Discrimination," which is included in your briefing book. The report notes telling statistics about the overwhelming opposition of individuals to allowing employers and insurers to access their genetic information, but also shares the stories of individuals like some of those you've heard today, like Heidi, Kim, and Mary.

Heidi was denied health insurance for her children because they were carriers of a gene for a liver condition. Medical professionals knew that the children would never develop this disease themselves, but the insurance company denied coverage because they carried the genetic marker.

Kim, a social worker for a human services agency, was fired because of her employer's fears about her family history of Huntington's disease, which she revealed during a staff workshop on caring for people with chronic illnesses.

Mary has a family history of breast cancer, but decided against being tested for the genetic mutations that make women more susceptible to breast cancer because she feared a positive result would jeopardize her chances for promotion at her law firm.

To allow individuals like these three to realize the full benefits of genetic testing and keep genetic discrimination from standing in the way of improvements in public health, strong, meaningful federal protections must be enacted. The Coalition has developed four core principles that we believe must be part of any legislation.

All genetic information that predicts future health risks, including family history, must be protected. Health insurers and employers must not be allowed to collect predictive genetic information and use it to discriminate in the health care system and the workplace. Individuals who experience genetic discrimination must have the right to seek redress through legal action with access to meaningful remedies, and entities holding genetic information about individuals must be prohibited from disclosing it to third parties without the individual's permission.

As science progresses ever more swiftly, it becomes more critical that Congress act to ensure that Americans are protected from genetic discrimination.

Thank you.

DR. TUCKSON: First of all, thank you very much, and I would urge the committee to refer to Tab 4 of your briefing books for a very important document called "Faces of Genetic Discrimination: How Genetic Discrimination Affects Real People," which I believe you all had a lot to do with and I think is a very important addition to our material.

I also hope that, again, if there are any specific things that you want to send forward based on the discussion this morning about specific things that the committee can do to try to be a part of this solution, part of the leverage, the coalition building, and all that sort of thing, please, would you send that forward?

MS. BRODERICK: Certainly.

DR. TUCKSON: All right. Thank you very much for being here and congratulations on this really excellent document.

MS. BRODERICK: Thank you.

DR. TUCKSON: Terrific.

DR. LEONARD: This document will be included in the information we send? Because it's right now in our book and not in this.

DR. TUCKSON: So the point is that the Tab 4 material is not in the bound material, and Debra's sort of urging, requesting, that we augment the material we send forward with this report.

DR. LEONARD: Yes.

MS. CARR: Are you amenable to that, to make this part of our record as well?

MS. BRODERICK: We would be, yes.

DR. TUCKSON: Terrific. That's great.

Our last speaker today is Donald Horton, director of public policy and advocacy for Laboratory Corporation of America.

MR. HORTON: Mr. Chairman and members of the committee, thank you for the opportunity to speak with you today. As a national leader in genomic and genetic testing, LabCorp views genetic discrimination and the coverage and reimbursement of genetic tests and services as being very serious, highly important issues, the resolution of which will have very long-lasting, long-reaching effects on the future of genomics, and therefore on the future of medical care in general.

I have to salute the individuals who very bravely came here today and told their personal stories of genetic discrimination. They were very powerful and very compelling.

Like those individuals and like the groups who have spoken here today, LabCorp supports federal legislation to prohibit discrimination on the basis of genetic information in health care and health insurance and employment matters. We believe that's the only way that the full benefits of genetics and genomics can be fully realized because that's the only way, we believe, that fear of genetic discrimination and its actual practice are going to be banned from the health care system entirely.

We support that for many of the reasons that you've heard today. Existing federal law is simply inadequate. When you put together HIPAA, when you put that together with the ADA as well as Title VII of the Civil Rights Act of 1964, all of those together do not create a comprehensive federal framework to protect against genetic discrimination.

We do have some good state laws, but they vary significantly in their depth and scope of protection, and even when you have a state that does have a good state law, you have ERISA that jumps in and preempts that, and ERISA does nothing to protect that information.

So it's critically important that we move forward. I think your next steps that you've identified today are right on target. I would just throw out a couple of things.

First of all, don't give up. This is an issue that's not going away regardless of what happens in this session of Congress. It's extremely important that this remain on the agenda and that we be persistent and consistent in moving this idea forward that we need comprehensive federal protection.

Secondly, I would say that let's not let the perfect be the enemy of the good. There probably is a little bit of room for negotiation here and there to get the parties together. Let's get something on the books and then we can do what we need to do down the road to polish it up a little bit. But we're very, very close. We need to take this opportunity now that we have it.

And just a moment if I could just to speak to coverage and reimbursement issues. I'd just like to thank you for considering the real-world examples that our own Dr. Paul Billings and Tammy Karnes offered to your Coverage and Reimbursement Task Force last month. We believe that those observations will be very helpful to you in putting together the final draft of the report.

Thank you.

DR. TUCKSON: Well, you all are terrific.

Let me just stop first and thank you, and make sure, is there a question on this issue? Yes?

DR. McCABE: Actually, not a question, but while you were speaking and referring to Tab 4, I was looking through other materials that should be included. I would think all the correspondence both to and from the various Secretaries of both committees might be included.

I know that we were told that the U.S. Chamber of Commerce was not adamantly opposing this, but I would refer people, after the "Faces" document, to the testimony of Mr. Lorber before the hearing of the House Committee on Education and the Workforce Subcommittee on Employer-Employee Relations. Read that testimony and read his questions under the end.

When we were told by their mouthpiece who was sent here this morning for them that they were not blocking this legislation, it is very clear that they blocked the legislation single-handedly in that subcommittee, and I challenge again, include this. Let's name names. Let's recognize who is doing this to the people of the United States.

If this continues to go forward, Mr. Chairman, and I know that we should not give up, and I'm glad to hear you say that, then let's request yet again -- you'll do it in a much more diplomatic fashion than I would sitting as a member, I'm sure, but I do not think they should be able to get away with this and then obfuscate by sending a mouthpiece to deny.

DR. TUCKSON: Yes. Me, diplomatic. I like that.

(Laughter.)

DR. COLLINS: I wonder if I might ask a question of the LabCorp folks. You, I assume, employ folks all over this country. So you're a large employer who has to deal with these laws, on the other hand, as well, and I'm wondering if you have experienced any difficulty in complying with the various pieces of legislation in many states in terms of genetic discrimination, and if so, do you think it would be easier to have a federal piece of legislation to comply with rather the many different state laws as a large employer?

MR. HORTON: Well, it would certainly simplify things to have a single comprehensive federal law. You know, we're in all 50 states. Quest Diagnostics has the same

situation and a number of other labs face that very same situation. So yes, it would simplify things greatly.

DR. TUCKSON: Terrific.

DR. COLLINS: Maybe you can talk with your colleagues in the Chamber of Commerce, as you're a large employer, and maybe be a voice within that community as well.

MR. HORTON: I don't think we're members, actually.

DR. TUCKSON: One other request as we close out here, and that's for the public. Again, if we could also ask you, each time we develop materials, if you could make sure they got to your constituencies, we sure would appreciate it. I know you keep them well aware of what we're doing as a committee, but I would really want to explicitly get from you a sense that you can distribute the summaries or whatever it is that we're doing to your constituencies in whatever way. Whatever it is, through your newsletter, but something that keeps this pipeline going back and forth between you and us I think would be just terrific.

As far as Ed's challenge to us, I think we'll probably have to have a little staff meeting to figure out, because this is important. We've got quite a lot of material that we want to transmit. So we're going to have to figure out how do we put a summary document on the frontispiece and then have the appendices because we're going to be sending out a telephone book, but that's because I think we have a lot of things to say. I think that's the challenge. I think Ed really appropriately challenges us to think about that.

DR. McCABE: I think it's important to send a telephone book, given that we have been told that this is a problem that does not exist.

DR. TUCKSON: No, I think you're right on the money. I think we'll work with him and others on trying to get the summary document and then the additive material.

We are starting tomorrow at 8:30, on time, and you all have put in a heck of a hard day. As a result, you get this reward that Sarah is going to tell us about.

MS. CARR: All the committee members and the ex officios who are attending dinner should meet in the lobby at 6:40 or meet us at the restaurant if you're coming independently. There's a little purple sheet in the table folder that has the address and so forth. So see you in the lobby at 6:40.

DR. TUCKSON: Thank you all. Good day's work.

(Whereupon, at 5:12 p.m., the meeting was recessed, to reconvene at 8:30 a.m. on Tuesday, October 19, 2004.)