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

Inaugural Meeting

Thursday, June 12, 2003

Vista Ballroom A
Wyndham Washington Hotel
1400 M Street, NW
Washington, DC

IN ATTENDANCE:

Chair

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C O N T E N T S

C O N T E N T S

P R O C E E D I N G S (8:36 a.m.)

DR. McCABE: Good morning, everyone. We're going to start off the morning with public comment. We're extremely pleased that we've had public comment both yesterday and today. If anyone wishes to sign up for public comment and is not signed up, please do so.

I have three individuals signed up at this time, Robin Bennett, David Sundwall, and Joann Boughman. If anyone else wishes to, please sign up at the registration desk.

So we'll lead off with Robin Bennett, representing the National Society of Genetic Counselors, and if you could come to the podium. Thank you, Robin, for doing that.

I should comment that just in terms of history and to maintain this, I think the National Society of Genetic Counselors made a commentary at every one of the Secretary's Advisory Committee on Genetic Testing meetings. So I'm glad that you're continuing that tradition.

MS. BENNETT: Thank you, and good morning.

The National Society of Genetic Counselors represents over 2,000 genetic counselors with specialized education, training and experience in medical genetics and counseling in an array of medical specialties. We are the leading voice, authority and advocate of the genetic counseling profession.

Genetic counselors are uniquely qualified to provide quality genetic services to the public throughout the life span, from newborns to elderly individuals. We have the skills and resources to answer research questions related to genetic services, counseling and testing.

The NSGC would like to assure that the following areas of concern are addressed by the SACGHS: prevention of harm from genetic testing, support for clinical genetics research, and access to genetic services.

Genetic testing is part of the process that must involve pre- and post-test counseling to enable consumers to make well-informed decisions. Consumers and their families have the right to expect that the correct genetic tests have been ordered, that specimens have been sent to a CLIA-certified laboratory, that costs of genetic testing are known, and that the test results have been interpreted correctly.

The nuances of reduced penetrance and variable expressivity complicate the clinical implications of positive or negative test results. Genetic test results are frequently used by patients and their family members to make complicated medical decisions about pregnancy outcome, medical treatment or disease prevention.

In the area of cancer predisposition testing, for example, a positive test result could lead to increased monitoring, chemoprevention, and in some cases prophylactic surgery. Genetic testing should be voluntary, accompanied by pre- and post-test counseling. The SACGHS should advocate for genetic counseling as an important component of genetic testing, including presymptomatic testing for conditions with no treatment.

When is a genetic test ready for application to the public? There should be oversight of a genetic test as it moves from research to clinical practice. There are even web-based companies offering genetic tests with no clinical validity or utility. It is as equally important for quality assurance measures to be enforced for laboratory analysis as it is to ensure high-quality pre- and post-analytical phase of testing.

The SACGHS should support education of health professionals, insurance companies, judicial systems, lawmakers and consumers about appropriate uses of genetic testing. This should include the differences between diagnostic, presymptomatic and pharmacogenomic profiling and the availability of genetic

services.

Strong antigenetic discrimination laws for insurance and employment must be enacted and enforced. Fear of genetic discrimination is pervasive in the community, despite limited documentation that genetic discrimination is indeed occurring. Fear of genetic discrimination causes underutilization of genetic services and inhibits individuals who would benefit from genetic testing from using this technology.

As a final area of protecting the public from harm from inappropriate uses of genetic testing, licensure of genetic counselors would help assure that consumers receive quality genetic services from individuals with appropriate training.

There needs to be increased support for clinical genetics research. Individuals found to carry germline mutations invariably ask their clinician what does this result mean? The only way to truly answer questions about genotype/phenotype correlations as well as appropriate management is through large-scale cooperative research studies. The NSGC strongly supports thorough large-scale cooperative research studies and believes that such studies will provide valuable information to our patients and their families. Genetic research projects should continue being encouraged and supported, including studies on ethical, legal, social and financial implications of testing.

The American public deserves access to quality genetic services, regardless of socioeconomic status, racial, ethnic or educational background, disability, ability to pay for services, method of payment, or geographic location. To meet the exponential demands of the public for genetic service providers, there needs to be an increased training of a culturally competent family-centered genetic workforce of genetic counselors and medical geneticists. Currently, there are 26 programs accredited by the American Board of Genetic Counseling. Each program receives 75 to 100 qualified applicants. There are about five qualified applicants for every available training slot. Genetic counseling should be recognized as a specific allied health profession with access to federal support for training programs similar to other allied health professions.

To assure uniform access to genetic services, there must be improved insurance reimbursement for genetic services. Many insurance companies do not cover the cost of genetic services or these services are limited to coverage during pregnancy. Coverage may only include genetic testing of a pregnant woman but not the necessary genetic testing of her partner. Few plans cover genetic counseling in the setting of prevention. For example, genetic risk assessment for individuals at high risk to develop cancer.

As President of the National Society of Genetic Counselors, as a genetic counselor with over 20 years of clinical experience, having personal experience with over 10,000 patients, and as a consumer of genetic services who has watched cancer tatter three generations of my family tapestry, I am here to extend the services of the NSGC to the efforts of the SACGHS. We look forward to working with you.

Thank you for your attention.

DR. McCABE: Thank you very much.

Any questions or comments from any of the Committee?

DR. LEONARD: Could I ask a question? As we move from a paradigm of genetics being involved in relatively rarer diseases to being involved in basically all of medical practice, how do you see the integration of genetic counseling services versus counseling by the physician themselves evolving?

MS. BENNETT: Well, I would advocate that just because genetic services become more common in the public, it doesn't become any less complicated, and actually when you move into common disorders, you have a much harder time sorting out the true meaning of a negative test result versus a positive test result. So instead of saying there's not enough genetic counselors, I would advocate that there needs to be an

increased workforce and a lot of effort put into funding for training and making sure that workforce is culturally diverse and cover geographic areas around the country.

DR. WINN-DEEN: I had one more question. You said that there's five qualified applicants for every slot. Do you have a specific proposal for how to increase the number of slots?

MS. BENNETT: I think there needs to be increased funding for genetic counseling training programs because there's definitely an interest in students who want to become genetic counselors that increases every year and there's very little funding. There are few scholarships for genetic counselors, no initiatives for minority development for genetic counseling students, and I think that has to go with medical geneticists also. It's not just genetic counselors. I'm just speaking because that's where my training is.

DR. LANDER: Can you tell us a bit more about the funding for genetic counselors? Do we need more resources at the universities to pay for these programs, more scholarships for people to make it affordable to do it?

MS. BENNETT: I think all of those things are true, and I think that having genetic counselors recognized by HRSA as a specific allied health profession would improve that because right now, it's not recognized as a separate allied health profession in any of those programs, so that the programs aren't eligible to apply for those grants.

DR. LANDER: So do you guys have a specific recommendation that would include the finances of it, to say this much money would be needed to launch the right number of programs, this much would go to scholarships, these are the HRSA recognition --

MS. BENNETT: We could certainly come up with that number. I'm not prepared to come up with it right now.

DR. LANDER: No, I wasn't meaning to put you on the spot, and I think I've heard it said many times that we don't have enough. We're clearly not going to be able to meet the demand, and I think it's time to come down to very specific proposals because my guess is that the amount of money needed to do it is really quite small relative to almost any of the other costs running around here.

MS. BENNETT: Right.

DR. LANDER: But having something concrete to get people to focus on it might help advance the cause some.

MS. BENNETT: I agree, yes, and I think there is the workforce to meet the demand. There just needs to be increased training programs.

DR. LANDER: So you'll come back to us with a proposal?

MS. BENNETT: Sure. I'd be happy to, and I think the other issue that comes along hand in hand with that is billing and reimbursement because that's an area that the demand is there but the hospitals can't fund the genetic counselors because they're not getting reimbursed.

DR. McCABE: Great. Robin, could you have something back to us in a month? I know one of the things that I've heard discussed is the need for diversity among genetic counselors and so the scholarship idea might play into that. So could you get something back to Sarah Carr within the next month, please?

MS. BENNETT: I'd be delighted to.

MR. MARGUS: So my quick question was how many men are among those 2,000?

MS. BENNETT: I think there's about 10 percent are men.

MR. MARGUS: Ten percent. Does that matter at all?

MS. BENNETT: Well, I am involved in social work training and there doesn't seem to be a problem with diversity in that field. I think that it is a problem.

MR. MARGUS: Are there ever counselees who would prefer to be counseled by a same sex or does it really matter in genetic counseling?

MS. BENNETT: I think that improving the number of men in the field would definitely be a service to the public. I would consider that part of the diversity of the profession.

DR. McCABE: And probably to some extent tied to the reimbursement, I would guess, because we've seen that in other disciplines as well.

MS. MASNY: I just wanted to mention as a comment and then a question, is that, we have an initiative from the Oncology Nursing Society Cancer Genetics Special Interest Group, along with the National Society of Genetic Counselors Cancer Genetics Special Interest Group, to do some collaborative work, so that each group could gain from the expertise of the other, and my question would be that in other areas of genetic counseling, could there be similar types of collaborations with other health professionals?

MS. BENNETT: I think absolutely.

DR. McCABE: Other comments or questions?

(No response.)

DR. McCABE: Thank you very much for speaking to us.

MS. BENNETT: Thank you.

DR. McCABE: And we look forward to receiving your written proposal to us, your recommendations. Thank you, Robin.

Our next speaker is Dr. David Sundwall, who is representing the American Clinical Laboratory Association.

DR. SUNDWALL: Good morning.

In order to be brief, I'm going to stick to a script here. I'm very pleased to be here to make a few comments on behalf of the American Clinical Laboratory Association, ACLA as we call it, a not-for-profit organization representing the nation's leading independent clinical laboratories.

ACLA member companies provide services in every state of the Union and provide the majority of lab testing done by commercial laboratories nationwide. I am pleased to have been invited here to make a statement at this inaugural meeting of the -- you know what you are. Secretary's blah, blah, blah.

(Laughter.)

DR. SUNDWALL: It's getting long. It's kind of hard to say. By the way, what is the acronym? It's not quite as easy as SACGT.

I want to compliment those of you who have accepted this invitation to serve on this important Committee. You now have the responsibility to carefully consider and wrestle with complex issues that will undoubtedly impact health care in many ways. Genetic testing is likely to be an increasingly important component of preventive medicine and enhance the ability of clinicians to make accurate diagnoses, to tailor treatments, and to make them more effective. Furthermore, the development of new genetic tests will likely be the focus of a significant amount of biomedical research.

Because the charter of this Committee is broader than its predecessor, the SACGT, you will also likely address more challenging issues related to ethics and social concerns. I personally commend each of you who have accepted this appointment for your time commitment to contribute to this very important advisory body. As you embark on your duties to advise the Secretary on genetic testing and its potential impact on the health of individuals in society, I want to leave you with just two points on behalf of ACLA.

Number 1 is the importance of appropriate and feasible regulation of diagnostic testing. ACLA understands that the government has to provide some oversight of genetic testing. Such tests have enormous potential to prevent and treat disease and the federal government has a role in ensuring that such tests are valid and appropriately used. However, we believe there is a significant risk to overregulation. For example, on January 9, 2001, Secretary Donna Shalala wrote Dr. Ed McCabe, then chair of the SACGT, and recommended that "oversight of clinical genetic testing services (so-called homebrews) as well as genetic testkits would be undertaken by HHS to fulfill their regulatory responsibilities."

ACLA, along with five other scientific medical professional organizations responded in a letter to Secretary Tommy G. Thompson on May 16th, 2001, expressing our concern that the January 9 directive may "hinder the development and dissemination of genetic testing advances by significantly expanding federal involvement in the clinical laboratory."

We remain of this opinion and believe that whatever regulatory mechanism is imposed by the department should be carefully considered and should take into account the well-acknowledged and medically accepted role of clinical laboratories in fostering genetic testing advances. We do not think such regulation should be generally applicable to clinical laboratory science and services. Having said that, we understand that it will be challenging for you to determine how best to focus federal regulatory efforts without risking public health by hindering access to new technology.

The second point I'd like to leave you is simply an offer that the considerable experience and expertise of representatives of ACLA member companies is at your disposal. Collectively, our member companies do considerable research, provide a very high volume of testing and have years of experience in complying with quality control and regulations currently imposed on laboratories. We would be pleased to work with you to help you in fulfilling your responsibilities.

Now let me just insert a paragraph here that isn't in my written statement, and I'm now speaking as a member of the Clinical Laboratory Improvement Act Advisory Committee, known as CLIAC. It wasn't mentioned yesterday, but the Centers for Disease Control also has a very important role in implementation of this law. At our September meeting, I'm given permission to announce that the CLIAC will revisit our role in the regulation or oversight of genetic testing, and I want you to be fully aware of that just because there would be a risk of overlap or redundancy, and they have done this before but nothing was forthcoming. So we're going to put that back clearly on our agenda and will be working to figure out what is the proper way for us to assure validity and reliability through the CLIA mechanism. I've already spoken with Steve Gutman at the FDA and we clearly want to work together to make sure that there's cooperation and not redundancy.

The second point is that the CLIAC will be following up on our presentation at our last meeting on direct access testing which was discussed yesterday. We also are very concerned about this. We don't know a

proper role of regulation and limiting access to patients to testing, but we are very concerned about what many would consider unethical promotion of such testing and their non-medical necessity use.

So with that, I'll just tell you again we look forward to working with you and compliment you on your efforts.

Thank you.

DR. McCABE: Thank you, David.

DR. TUCKSON: David, it's always a pleasure to hear from you, and I appreciate your comments.

I would sure appreciate maybe if you could supplement your comments back to us after this meeting with a sense of while we know that this Committee is not a direct pass-off of the earlier Committee to which you referred, there is a relationship, and you know that we struggled mightily in those last meetings around the appropriateness of defining criteria for appropriateness for release of tests.

A lot of time has gone by since then, and it would be important, I think, for us to know what progress has been made. How well does what exists today work? Do we feel like now that the private sector, through organizations such as yours and CLIA, have got this problem solved? Do we have anything to worry about? What's missing? Because at the end of the day, I think many of us might be well persuaded to say let the private sector do it. The government regulations are in place. The right balance. If it ain't broke, don't fix it.

But I think we would need to sort of know from you how well is the system regulated, how appropriate is it, and maybe others may have a different point of view, but I think that's a data point that I would sure like to see early on.

DR. SUNDWALL: Fine. I just need some help with you, Reed, on, do you mean the quality assurance efforts or are you talking about privacy of data?

DR. TUCKSON: The quality assurance effort, the appropriateness of the criteria that is used to ensure that the test before it's released to the public meets sensitivity and specificity.

DR. SUNDWALL: I see.

DR. TUCKSON: Clinical validity measures. I mean, all the technical stuff that says crap is not being sent out to be used on the American people.

DR. SUNDWALL: Right.

DR. TUCKSON: And what I think I hear you saying, I don't want to belabor it now because we don't have time, but what I hear you saying, I think, is the private sector feels that you all don't want government all over you with unreasonable regulations.

DR. SUNDWALL: They already are.

DR. TUCKSON: So at the end of the day, we need a sober, cold-blooded assessment from your point of view as to whether or not what the state of the situation is and then let's see how others may feel about it.

DR. SUNDWALL: I'd be happy to provide that.

DR. LANDER: I think that's just great. I very much agree that hearing that very concretely would be very valuable, and I wonder if you could also fill in a bit for me at least who sort of vaguely knows about

CLIA but doesn't quite understand all of the range of things that have been considered there.

I know that CLIA has a lot of experience in making sure that a well-defined test is practiced at a high standard, that when somebody says that person is Apo E4, CLIA has all the right kind of procedures in place to be sure that they are really Apo E4, that QC's been done, et cetera. But what I wonder about with respect to the genetic testing is it's less whether the genotype is done correctly than whether the interpretation is valid in a world where this is just radically changing all over the place.

Can you give us examples of the experience of the CLIA process in dealing with the validity of interpretation of testing as opposed to simply the laboratory process?

DR. SUNDWALL: Right. Well, I'm glad you make that distinction because at Reed's request, I think I can provide a wealth of data to give you confidence that the lab data result is good. I think that as CLIA does its job, we wrestle with the same thing. The most difficult component of lab testing, as I understand, is not getting an analyte you can trust, it's the pre- and post-analytic phase.

DR. LANDER: Yes.

DR. SUNDWALL: Was it ordered necessarily in the first place? Then is it interpreted or used appropriately? That goes hands-down for genetic testing even more so. So I think that as we meet in September, these would be the very issues that we're going to focus on, the pre- and post-analytic phase.

It's a challenge, and I think we're kind of pioneering in this effort, but I'm very pleased to hear this presentation on the genetic workforce and counselors because that's an important component of it, too. But let me just agree to provide for the Committee what CLIA does in their performance testing, their oversight of labs. I think you can be assured the labs that are CLIA-certified and CAP-certified are pretty good trustworthy operations, but that doesn't mean there isn't room for improvement.

DR. LANDER: But you would say this is relatively new ground with respect to the nature of the interpretation of information, not the analytes.

DR. SUNDWALL: Right.

DR. LANDER: Because I have that sense, too, and that's where I wonder whether -- I'm very much in favor of seeing the private sector take major responsibility in this.

DR. SUNDWALL: Right.

DR. LANDER: But I think it's probably fair to say there isn't that much experience anywhere, private sector or public sector, with regard to these questions, and so simply leaving it to the private sector may be difficult. So it would be interesting to hear from the private sector and from the laboratories what are the things you're most worried about? What are the failure scenarios you guys see? Because since I don't think there are very large numbers of existing failures, we're looking ahead. We're all on the same side on this, but hearing you say very specifically what could go on that would not currently be caught by the system, that would not currently be caught by existing practices would be helpful, too.

DR. TUCKSON: So less on the background, less on the background of CLIA and more on the interpretation of what does it all mean in the current time today?

DR. SUNDWALL: Well, let me just have a note of caution here. I think I can speak on behalf of the labs. One thing about independent commercial laboratories is they don't see the patient, and I am afraid that there may be some expectation put on labs they can't meet. In other words, if you don't have that relationship with the patient, you do the test on behalf of your physician client or sometimes the patient who requests the test to a lesser degree, but that's a significant minority of our business. It's primarily for

the physician, although I think we should foster this kind of improved follow-up and we have a role in interpretation.

I'm a primary care physician in my other life, and when I see patients, I'm grateful for the laboratories that provide for me not just reference ranges but some education at the bottom of the page, and I think that's very helpful.

DR. LANDER: So if I'm hearing you right, you're pointing out the need for or the potential need for thinking about regulations in two parts. One is of the laboratory.

DR. SUNDWALL: Right.

DR. LANDER: And the other may be a very different structure, not so much affecting the clinical laboratory but that interpretation and that's a very helpful distinction.

Thank you.

DR. SUNDWALL: I think you all need to pay attention to that. What is the role of the lab versus the clinicians or the counselors or the institution that's doing this testing?

DR. McCABE: So David, could you get back to us with that information?

Arden, and then Debra briefly, please, so we can move on.

DR. BEMENT: I think I may know the answer you've been searching for in the dialogue that took place, but I take it when you talk about regulation, you're talking about laboratory certification. You're talking about quality assurance. You're talking about instrument calibration. You're talking about chain of conformity to higher-level standards.

DR. SUNDWALL: Right.

DR. BEMENT: All those things.

DR. SUNDWALL: Indeed.

DR. LEONARD: I think that Eric has basically made a very important distinction, and I'd like to re-emphasize it, that the clinical laboratories are regulated by professional organizations, by the government, by a lot of different levels. Concern is raised by those laboratories doing testing that are not CLIA-certified, and I think that is an area that we need to think about and maybe those are the enhancement laboratories, but there may be clinical laboratories functioning out there, also, that aren't following appropriate regulations. So operating outside that window is of great concern, and then I think the whole health care professional education piece addresses this post-analytical use of test results. The test results may be done accurately, but the laboratorian -- I mean, I am a physician, and I do interpretation of results, but I can't control the physician-patient interaction if it fails, and I think that's where there's a great deal of concern.

DR. SUNDWALL: My last word, Ed, one suggestion is that you focus -- I base this on our last CLIAC meeting -- on grave concern about the inappropriate marketing of these tests, and I think, who knows, is that the FTC or who does that? I'm not sure. But I don't think it'd be necessarily CLIAC or customary bodies.

Thank you.

DR. McCABE: We took this up in the Secretary's Advisory Committee on Genetic Testing, and it does

get very complex because it's multiple agencies that are involved in that. So it's Commerce. It's FTC. So it's a number of areas.

And lastly, thank you very much, David, and we look forward to having your written responses to that discussion.

Dr. Boughman is Executive Vice President for the American Society of Human Genetics and was a member of the SACGT, and I want to thank you, Joann, for being flexible in allowing us to move you to your presentation today.

DR. BOUGHMAN: Well, thank you very much, Ed, and today, it will sound like refreshing the agenda rather than just hashing it over one more time yesterday.

The American Society of Human Genetics, for those of you who don't know, is the primary professional organization for human geneticists with nearly 8,000 members. It includes researchers, academicians, clinicians, laboratorians, genetic counselors, nurses, a variety of other people involved in or with special interest in genetics. Our mission is clearly stated to, one, promote and expand research; two, to apply the knowledge to enhance health care; three, to train the next generation of geneticist professionals; and four, to educate and inform other health professionals, the public, lawmakers, policymakers, and so forth.

We take our mission very seriously, and I'm pleased to say that sitting among you on the Committee are five members of our society, so that we understand that the viewpoint of the genetics community will clearly be heard here. But today, on behalf of the Board of Directors, I'm here to do two things, just point out a few of the issues that we see as the most urgent and most important, and secondly, to offer our services in any way of the 8,000 or so of us out there that are working in the area.

Let me make just a few points. In the area of research, we know that the basic science research is moving very rapidly, and we know that translational research is also moving rapidly but that we need in fact more focus on that translational research and the transition into clinical practice, and in fact those areas are broadening as we heard yesterday into the area of human genetics and bacterial genetics and the interactions between the two.

We also recognize that the more traditional academic model of publish or perish must be adapted in this rapidly changing environment to serve new and very exciting public/private partnership models without overreacting and making it simply a protect and profit model. We've got to come out somewhere in between. But in that vein, we have pride in the fact that we think the genetics community in general has demonstrated significant leadership in the sharing of research findings and the making of our research findings public.

Conversely, industry-based researchers as well as academicians who develop marketable intellectual property must also obtain and protect that intellectual property. Securing that balance between the protection of intellectual property and public access and patient access to the results of that is of great concern and we seek that balance. We encourage this group to further engage in substantive discussions around some of these issues, patenting, licensing. In fact, as suggested by Professor Sung yesterday, that there may be some ways, some new points that could be made and taken up by the Patent and Trademark Office in ways that biotechnology is different and encouraging some of those discussions may be extremely valuable.

We also are dealing with the new privacy rule, and we have found in the area of research that barriers, whether real or just perceived, to research participation by volunteers are also of some concern. We have new challenges in the interpretation of compliance with the HIPAA privacy rule and we have a new ad hoc Committee in ASHG that is looking at some of these issues directly related to genetics.

We obviously maintain the highest regard for privacy, but we are concerned about the lack of

understanding and interpretation and possible over-interpretation of the HIPAA rules. We also have some concern about consistent interpretation by IRBs and privacy boards and the inconsistencies that may arise institution-by-institution.

ASHG is the large umbrella organization, and in certain situations with regard to health care, in fact, it may be the American College of Medical Genetics or some of the boards that may be more specifically interested in that, but under our umbrella, I'll go ahead and make a few comments about health care issues as well.

As we have already heard, access and cost of testing and access for patients to services as well as cost recovery is, of course, of primary interest and a topic of importance and urgency. As has been mentioned several times, quality of service is a basic tenet of clinical practice, but in this emerging and expanding field of genetic testing and services, systematic and systemic quality control mechanisms are not yet fully in place as we know.

Because the term "genetic testing" covers such a wide variety of services, from newborn screening, diagnostic confirmation, and now more into the areas of predictive testing and risk assessment, as well as from identifying the common rare disorders to moving into the common disorders, we also urge you to think about the issues of the complexity of multiplex testing and now as we move forward into the concept of genome scanning. These will present us new challenges, we believe.

Adequate interpretation of the results is obviously essential, and we think that you've just reiterated the gap that may exist between the laboratory, the laboratorians, the oversight of their practices within the laboratories, and the translation and interpretation of that information to the patient and the possibility of information loss along that trail, if you will.

This is getting, of course, more complicated as the interpretation becomes probabilistic, not inevitable, in its interpretation, not just are you a carrier, are you not a carrier, but what is the probability that this allele or this gene may increase your risk for a predisposition to a disorder? We have concerns about that. Our collective concern is heightened when even the best lab reports go to unprepared clinicians to translate that information, and we move from concern to anxiety when we talk about some of the direct consumer testing, and I would remind us that while we in the professional community may clearly understand the difference between a CLIA-certified laboratory and some of the other end of the spectrum that we saw yesterday with the ego-genomics, that there is a very large area that's somewhat fuzzy in between and even if we clearly know the difference between those, the public may not know.

We also are interested in the protection of research subjects obviously, and we are very pleased with the Senate HELP Committee moving the Genetic Information Nondiscrimination Act from the Committee to the agenda of the Senate. We know that there may have been only a very few cases documented of discrimination in insurance eligibility and/or employment, but the perceived deterrent has been very important to us. We will be active in the support and interaction with members on the Hill, and I think if there was a message that I heard yesterday, certainly from Dr. Zerhouni, he gave a very clear message to this group that a comment from this Committee would be welcomed on the importance of the Genetic Nondiscrimination Act.

The last point that I'd like to make is about preparing future professionals. That's really a growing challenge for us as geneticists. We have a limited workforce of trained geneticists, and we are now in the area of trying not only to train the next generation, we're doing the research, we're providing the services, and now we have the challenge of sharing our information and training all of the other health professionals as well.

I think Robin Bennett made a very good point about the support of training programs in genetic counseling that has not been on the agenda of the nation recently.

We also are participating in and very supportive of many of the efforts of NCHPEG and other outreach efforts, but the resources remain limited. There have been some wonderful model programs, like the Genetics and Primary Care, that in fact are seeing fruits from their labors, but once again, unless we have more trained geneticists, we don't have enough of us to do all of that interacting.

We also have a long tradition of activities to inform the public and we're very active in that area now with the Mentor Network that we have developed with NHGRI. We have 900 of our members out there who have volunteered in their local communities to be mentors either in the public schools, K through 12, all the way through graduate school. In fact, in the week of the 50th anniversary celebration, we know of 60 activities in 27 states with almost 3,000 people in the audiences with geneticists in the community. I think this is a huge effort but once more, we can only be in one place at a time.

We would encourage this group to support those outreach activities in any way that they can and encourage these interactions between the professions to enhance this. We'll do our best as a group of geneticists to get the kids young and bring them up right, if you will, but in fact we need to have more focus and support for those activities.

And finally, with regard to what Dr. McCabe said yesterday, should this Committee be thinking very long term or immediate, I was immediately reminded of the think globally, act locally rule which I would encourage you to think long term but in fact focus initially on some actions that could make some immediate effect and in setting of the agenda for further conversations, and once again, the American Society of Human Genetics would be happy to help in any way we can. Please call on us for anything we can do to help.

DR. McCABE: Thank you very much.

DR. LEONARD: I would like to make a proposal or ask your opinion on an issue.

DR. BOUGHMAN: Sure.

DR. LEONARD: Which is it's my feeling that my 14-year-old probably knows more about genetics than does a 50-year-old physician, primary care physician. So focusing efforts on current training is not so much of an issue as figuring out how to reach the practicing physicians that are out there and this Genetics and Primary Care Initiative, those types of things, I think, need to be more the focus of initiatives because maybe in 30 years, we won't be having this problem anymore, but we have to get to that point.

So do you have any suggestions on how you get the practicing physicians trained?

DR. BOUGHMAN: Well, I would suggest that there are many of those activities going on and this interfaces with the Society of the American College of Genetics and the American Board of Medical Genetics and its relationship with the other boards and certification of physicians and the development of proposed curricula for a variety of current training programs in pediatrics, internal medicine, and so on, but even more so as the professional boards move to a climate of maintenance of certification, where in fact physicians are recertifying and the development of materials and curricula that can be moved into those other areas, that is one of the systematic ways that we can do it as well as doing it the best we can on a day-to-day basis with all of the grand rounds and other things that we might do in not only medical centers but general hospitals and such.

DR. LEONARD: I think your point is excellent, that we have moved to limited licenses for physicians, and so the recertification process may be a window of opportunity.

DR. BOUGHMAN: Yes.

DR. LEONARD: If those training materials or informational materials are out there, to at least get to them the basics of what does it mean if a disease is recessive or dominant or when are you in over your head and who should you refer to and things like that.

DR. BOUGHMAN: Yes, and Dr. Leonard, I think you just made a perfect point. I think as geneticists, we have not been good enough at defining who we are, what we do, and when you should refer to us, when in fact the information can be assimilated and truly used by the generalists and when the genetic counselor can be a part of the team that bridges some of those gaps in very important ways. But that's one of the systematic ways that we are trying to do it and developing curricula right now as quickly as we can in a variety of forms.

DR. LEONARD: Could we get information on their efforts?

DR. McCABE: Could you get us information on the efforts that you're putting together?

DR. BOUGHMAN: Certainly, and I will talk to Mike Watson over at the College and the folks at the American Board of Medical Genetics and get that list to you and also with NCHPEG and the development of some of their curricula in a variety of areas.

DR. McCABE: And NSGC also, because I think they're doing some efforts in this area.

I would just comment that I'm glad you're so optimistic about the physicians we're training now, Debra. Dr. Linda McCabe and I were asked by the Macy Foundation to put together a paper for a meeting they're having on how do we educate physicians for the future, for both the clinical future as well as educating them, and I think it goes back. It's not just problems in medical school but it's premedical education.

We're still requiring organic chemistry but we don't compound our drugs anymore. That's really just a memorization barrier that we put up for testing, and we still approach medical education as if we can teach a physician everything they're going to need to know for the rest of their career. I think we really need to revisit how we prepare for medical school as well as how we pursue medical school and really need to train people to be lifelong learners. We pay lip service to that, but we don't select for that. So there are a lot of issues here. Genetics is only one in terms of the technology.

DR. LEONARD: Well, that's why I went back to my 14-year-old because they just learned genetics and probably learned a lot of what medical students are currently learning.

DR. McCABE: Thank you.

DR. FEETHAM: As another source of information on the training of health professionals, again I have some information that I was going to distribute today, but HRSA in partnership with NIH, CDC, and other colleagues, we've done several programs focusing on the education of health professionals, and the Genetics and Primary Care that's been mentioned was under the initiative of Dr. Michele Puryear who's here who nudged us all to move forward in that. So we can add that to the information that you have. All the numbers of grants that have been funded in that area of education and those are often interdisciplinary education of health professionals.

DR. McCABE: This would be a big help to us since one of our issues really has to do with education and anyone else from any of the other agencies, anyone in the audience here, where there are activities going on in this area, if you could communicate with Dr. Boughman or, if you wish, directly with Sarah Carr, so we can begin to put these efforts together.

DR. BOUGHMAN: Yes, Dr. McCabe, if I could just make one more comment on taking advantage of every teachable moment, if you will. In one of the comments that Dr. Lander made earlier in part of this discussion about laboratory results and the difficulty in interpretation and the agencies involved, in the

SACGT and in other areas, one of the points of discussion is the transition from the trial process and the FDA approval process and in postmarket data follow-up that might come under the area of CDC and others, and in fact, what parts of those processes within the laboratory and beyond could be handled under the very broad category of labeling or information processes that go along with the test and the guidelines on the way the results should be delivered to the care provider.

This is something that we would not have concerns about with the large academically-based laboratories that do this on a routine basis but once again, as these genetic tests move out into the public sector and are being done by small hospitals or other small private laboratories that do not have this academic tradition, that in fact guidelines on the way those test results need to be communicated can in fact be a teaching process and a learning process if the guidelines are done correctly.

DR. McCABE: Thank you very much.

We're now prepared to move on. I don't think there are any other individuals who have registered for public comment. If there are none, then we will move on.

We're very pleased to have Kim Monk join us to present about the congressional efforts to prevent genetic discrimination. Kim is a senior health policy advisor for the U.S. Senate Committee on Health, Education, Labor, and Pensions, the HELP Committee. Her primary areas of responsibility for the Committee are in the private sector health benefits and insurance, ERISA, HIPAA, medical liability reform, the uninsured, genetic discrimination, medical records privacy, mental health parity, Patient's Bill of Rights, and long-term care. It appears that you're a generalist in this area.

Prior to joining the HELP Committee, Kim was senior health policy analyst with the U.S. Department of Labor's Pension and Welfare Benefits Administration, where she worked on policy and legislative analysis for the Department, Congress, and the employee benefit plan community. Kim was also manager of public policy for Washington Business Group on Health and worked for the health and welfare practice of William M. Mercer's Washington, D.C., consulting office.

Kim has a Master's degree in Health Services Management and Policy from George Washington University, and we're extremely pleased that you were willing to come and update this Committee on what's happening with the genetic discrimination legislation.

MS. MONK: Great. Thank you, and I apologize for being late. Unfortunately, that circle out there doesn't let you go all the way around the circle, so I found myself going north into traffic. How much time do I have?

DR. McCABE: You have half an hour.

MS. MONK: Well, what I'd like to do is describe for everyone the history of genetic nondiscrimination legislation as it's come through the Senate and the HELP Committee. We recently unanimously passed a bill out of the HELP Committee about three weeks ago, and we're very pleased about that, and I'd like to tell folks kind of how we got to that point and what's in the bill and where we might be going.

I've been on the Hill since 1999, but the genetic nondiscrimination bill actually predates me. I believe there were efforts made back in 1997 and bills drafted. Senator Daschle had a bill and Senator Snowe had a bill. Daschle's bill had both health and employment discrimination and the Snowe bill dealt only with health insurance, and those different approaches went on for a couple of years, and in the Senate, genetic nondiscrimination was included with Patient's Bill of Rights a couple of times and the Snowe bill was actually passed on the Senate floor three different times without the employment provision, but there were those who were repeatedly asking whether or not we didn't need to address the possibility of discrimination in the employment setting, and I used to work for Chairman Jeffords before he switched parties, and at that time, I think there was some concern and some questions about the degree to which

the current statutes, like the Americans With Disabilities Act, already covered genetics issues, and so back when Jeffords was chairman, we had several hearings, began to explore the reach of the existing statutes, looking at some of the more recent case laws and interpretations, and then after Jeffords switched and Senator Kennedy became chairman, we all came together at that point, Senator Snowe, and based on the hearings and what we had learned while Jeffords was chairman, decided that there was indeed some gaps or at least some questions about how the ADA could be interpreted or not be interpreted to include genetic discrimination.

So at that time, we went back and reworked the Snowe bill, updated some of the definitions, improved some of the provisions, redrafted the privacy provisions to reflect the comprehensive privacy regulation that had become final and come out of the Administration, and in addition, we added a whole new Title 2 which included discrimination in the employment setting which, I'll just point out for Republicans to draft a nondiscrimination bill for employment and allow employers to run the risk of being sued is a pretty big step. So it was not without risk on our side to come forward with that, but we did feel that there was a way to do it to address the gaps and a way to do it to not, if you will, open the floodgates so that you always are going to have a genetics suit every time you have any other kind of suit.

So we began down that path and while Kennedy was chairman, we got together with the Daschle-Kennedy camp and my boss was a sponsor of the Snowe bill to see if there was any way to iron out the differences in the two bills, if you will, kind of preconference them, within the Senate setting to see if we could merge them together for a single bill to bring that through the committee.

At that point, I think that the Snowe bill probably had the votes to pass but we did not control the committee agenda, and I think there was a strong desire on Senator Snowe's part and others to see if we could work out the differences and have a single bill because at the end of the day, even if you can get the votes in the committee or on the floor, what have you, those things most likely to become law are those that are bipartisan agreements and that everybody's on board. So the end game, I think, from Day 1 was always to draft a bill that could become law, that would be signed, and back when President Bush came out and made a very strong statement about supporting legislation, I think it was probably like June two years ago that was a really strong sign that everybody at least wanted the same goal and that's kind of a rare thing with legislation.

I mean, different people draft legislation for different reasons and some people want to put a marker out and some people do it as a defensive measure, some people do it to get involved and some people actually do it with the goal of seeing something enacted. So we were in a good place to start off, and we spent throughout the Kennedy chairmanship, I'd say on a part-time basis, we spent time with the other side attempting to primarily work out the differences on the definitions.

I see Kathy Hudson in the back. We worked a lot with her. She helped us out on that and NIH helped us out.

But what we did is we had two very different approaches on the bill. We felt that the Daschle bill was too expansive in that it included nongenetic information and therefore would get into the area of regulating insurance practices, insurance underwriting practices around current health status which is very heavily regulated by the states now and is also very tricky because states that have gone too far or have tried community rating have seen their premiums shoot up and so it's a very sensitive marketplace. So we were concerned about going too far and Democrats are concerned that our definitions potentially had loopholes where if we had an exception, that the exception was too broad and swallowed the rule or once information fell into the exception, it was forever then unprotected as genetic information. So we kind of put aside both approaches and started literally back from square one and over the course of almost a year, I think, redrafted a whole new definition section, and we completed that many months ago actually and vetted with everybody in the outside world and got really good feedback, and so we kind of got that done, put it aside, and then towards the end of the legislative year, we got busy with other things and really didn't get the opportunity to finish because even though we had come together on definitions,

we still had some really big issues to sort out, such as privacy and remedies and things like that, and so there just wasn't time at the end of the year.

So we kind of put it aside for a few months, and then Republicans won the election and my boss, Senator Gregg, became chairman of the Committee, and at the beginning of the year, when we sat down to figure out the agenda and what the priorities were for the Committee, Senator Gregg, he made his rounds. He went to NIH. He met with Dr. Collins and Dr. Collins said, "Well, this is really important to do this legislation because we will never realize the promise of the Human Genome Project if we don't have these protections in place." My boss agreed with that and made a commitment at that time to move this legislation forward, and in his view, there's not really a huge problem today. We haven't seen a lot of abuses. There's been a couple of cases that are troubling.

From what we can gather, there's only been one or two cases in employment, and from what we can tell in the insurance side, there might be some insurers in the individual market who are using family history which we do count as genetic information, but for the most part, these tests aren't out there. The underwriting process is not that sophisticated. It's not really being used. So on the one hand, we're getting a little pressure. Well, why do you need to write a bill and expose industry to lawsuits if there's not a problem? But I think over time, we really were convinced that now is the ideal time before this information is in the mainstream, before there's huge opposition against doing legislation, and more importantly, as the science continues to move forward, we're hearing stories and it's pretty well documented that people are afraid to get genetic tests. Doctors advise their patients, well, I don't know what to tell you. You could get the test, but you might want to pay for it out of pocket because I'm not sure what's going to happen with the information.

So the lack of a clear federal standard protecting this information has been an issue in terms of really being able to move forward with the science and the clinical trials and really, I think, getting insurance companies to come forward and pay for these things, and so this really is kind of a threshold opportunity. So we put genetics really at the top of the list for the Committee agenda and scheduled it for markup which really put the pressure on the two groups to negotiate very hard to try to work out the differences, and we did have it scheduled for a couple of markups and had to bump it back, but since the beginning of the year, we have been negotiating very rigorously, before it had been on a part-time basis, we really stepped it up, to try to sort out the remaining differences on the employment title and the health title.

I'm a health care person. So I don't really work on the employment title. My colleague David Thompson was the lead on that, but they had several big issues on that side to work out. One of the issues was what we call the "watercooler problem." Because family history is genetic information and can come up in casual conversation, oh, you know, sorry, I'm going to be late tomorrow. My mother's in the hospital. She has breast cancer. Well, that's a revelation of genetic information, and if you tell your immediate supervisor, that's a disclosure to the employer, and it might not be that the employer is acting on that, but we certainly had restrictions in the legislation not just for acting in a discriminatory manner but also for wrongful disclosure. So our concern on the disclosure front is somebody could read an obituary and, oh, don't send flowers, send a contribution to the American Cancer Society or something like that.

I mean, there's many opportunities to inadvertently acquire genetic information, particularly if you define it broadly to include family history. So we had the watercooler problem that we had to sort out which we eventually did by focusing in on, I think, key terms like "collect," "purchase," kind of steering away from the nebulous "acquire" term, and then we put a provision in there that specifically kind of creates the safe harbor for the inadvertent collection or acquisition of information like reading an obituary.

Another issue is the entire Title 2 is basically modeled after the Civil Rights Act and it kind of parallels ADA but it's the structure of the Civil Rights Act. So if you have a claim, you file it through the EEOC and they make an initial determination and weed out frivolous cases and then the rights and remedies that would flow for a genetic discrimination would be the same as those under the Civil Rights Act, and under the Americans With Disabilities Act, there's something called disparate impact which is even if an

employer doesn't intentionally or on its face act in a discriminatory manner, if the effect of their employment practice is to discriminate against a group or a class of people, then that would be discrimination, but the ADA includes sort of a protection for that, saying that if it's part of normal business practices, then that's a defense against a disparate impact claim. So it really gets into the details of really liabilities and lawsuits, but that was a big question as to whether or not we ought to have a disparate impact claim for genetics, and we really racked our brains trying to think about if there's some sort of fitness-for-duty test or requirement that might have some disparate impact on folks with a particular kind of gene, and I think right now, we couldn't identify anything but we were very aware that who knows what the future may hold.

So there are choices where to include disparate impact, have a business defense, include disparate impact but not have a business defense which I can tell you my boss was not comfortable with at all, or to not include it at all, and where we ended up was not including it at all but realizing that this is an area that could develop with the science. So we established a commission to study the issue to see if there was a need or a possibility for that type of protection in the law and those were the issues on the employment side.

On the health side, as I said before, it's really confidentiality or privacy and the rights and remedies, and as I said before, when we redrafted Senator Snowe's bill, all the bills were originally drafted before there were even draft privacy regulations, and since then, the Clinton Administration issued privacy regulations. The Bush Administration revised some elements and reissued final privacy regulations.

So we took a very careful look at the privacy regulations and they address the use and disclosure of all medical information, including genetic information, and we even had some hearings to see, well, do the privacy regulations adequately address genetic information, and our feeling was that, after studying the regulations and consulting with experts, was really did cover genetic information, except there was one place that we ran into a problem and that is, under the privacy rules, it allows for the use and disclosure of information, including genetic information, for purposes of treatment, payment and health care operations, and under health care operations, there's this one thing called underwriting, reinsurance, placing a policy with somebody, which, in insurance terminology, means basically discriminating. You're looking at somebody's medical information and deciding how much of a premium you're going to charge them and what have you and that's a practice right now that really only occurs in the individual market because HIPAA actually bans it for the group market and has since 1996. Then there's also reinsurance which isn't underwriting on the individual but is on the ground as a whole.

So we did realize that that was a problem in the privacy rules and attempted to address it in the Snowe draft but still didn't get it quite right. So throughout our negotiations, we refined that provision and we also had some in-depth discussions. The Daschle-Kennedy sponsors felt that the privacy regulations had an additional gap in that they only dealt with use and disclosure and they didn't deal with how an entity gets information up front. We never really came to any kind of agreement about that issue because I think that the privacy regulations do deal with that because in the use and disclosure, the regulations do lay out this consent process and an authorization process which really does get to how an entity gets information, and even if they get it and if they can't use or disclose it, our feeling was that there wasn't a gap. But there were still some who were concerned about ever letting an insurer even acquire that information and we were likewise concerned that, well, you know, there are bad uses of genetic information and good uses, and we want insurers to pay for genetic tests and genetic services, and if we create a whole different regime and set of rules around genetic information, that will be a disincentive. They'll never do it. We also want them to be able to continue with their disease management activities and health care improvement activities and genetic information can be a key part of that.

So we had to be very careful in our privacy provisions, and where we ended up, I'll just describe to you the privacy provisions, is we basically said the privacy rules apply to genetic information. That's kind of stating the obvious, but when it comes to use and disclosure of genetic information, the privacy rules govern, except that there is no allowance for underwriting because that's inherently contradictory to the

nondiscrimination bills. So we created an exception to say basically you can never use or disclose genetic information for purposes of underwriting and then we take it a step further and we say you can never even -- insurance companies are what we're talking about. Insurance company can never even collect genetic information for purposes of underwriting.

So we do have an upfront ban that they don't have with the privacy rules because there's a feeling if they couldn't use it or disclose it for underwriting, why do they ever need to even collect it? We also put an additional ban on insurers collecting genetic information prior to somebody being in their plan. So in other words, an insurance company can't go out and just make a database and collect information and solicit information for the purpose of having a database in case you're ever in their plan and then we have that information.

The marketplace is really changing. You're seeing a lot of Internet-based technologies and insurers like ehealthinsurance.com, where you get online and you do a bid. You may not sign up for insurance but it's an opportunity for somebody to collect information on you.

So we made it clear that there's no collecting of information, of genetic information, before somebody's actually in your plan, and then once they're in your plan, the privacy rules govern with respect to how they can get information and when they need to get authorization and then they can get it for treatment and payment and health care operations, except underwriting, so they can do things. Obviously they can protect themselves against fraud. They can conduct wellness programs, disease management programs and what have you. So that's what we did on the privacy side.

There are kind of two elements to this bill. There's the governing of the flow of information which I'm calling privacy, use, disclosure, collection, but then there's also the straight-up insurance ban on discriminating for underwriting, and what that does is say for group health plans and insurers in the individual market -- well, for group health plans. An insurance company can't set the premium or eligibility on the group as a whole based on any individual members of that group's genetic information.

That's kind of going beyond current law and HIPAA. HIPAA doesn't currently address premiums on the group as a whole, only addresses discrimination against an individual in the group, and then we further said because the HIPAA 1996 included discrimination provisions for group health plans, the individuals in group health plans with regard to premium rates and eligibility, you can't discriminate based on health status, current health status, including genetic information.

What we did was further flesh that out in terms of defining genetic information, making the rules a little more clear, breaking out the genetic piece and kind of clarifying some of the remedies around that, and the remedies are as follows. We really built on the framework of current law. So HIPAA's a really big statute and people talk about sometimes they mean privacy and sometimes they mean portability, but there are different pieces in HIPAA.

So Title 1 of HIPAA really addresses the traditional insurance things, portability, nondiscrimination. So for the nondiscrimination provisions of our ban, we just really build those into Title 1 of HIPAA which already addresses those issues, and then we do some additional things there. We say with respect to genetic information, we clarify that if somebody's in a group health plan and they're denied coverage and they think it's because of some genetic information or because they got a genetic test or something, we make it clear that if they're going through the appeals process and that's taking forever and in the meantime, their child needs to get well-baby care or somebody breaks their leg or what have you and there's health insurance claims, we make it clear that the person can kind of opt out of that administrative process and go to court and get injunctive relief and go to court and the court can say guess what, you're wrong, you have to let this person into your plan.

Then we think that's probably current law but it's not explicit in the statute, and so it's a bit of a gamble as to whether or not the courts would actually give you the right. They might say oh, you haven't exhausted

your administrative remedies. Go back and go through external review and all that. In the meantime, months could pass and you could be incurring pretty significant health care costs, and we've also clarified that if that should happen and you have to go to court and it turns out, you know, it took you three months, you were fighting with your health plan, turned out that the health plan was wrong, they should have let you in the plan, they were discriminating based on some genetic factor, and we make it clear that your coverage should be restored retroactively to that point where they actually denied you coverage.

Again, that's probably current law and these things usually get sorted out before somebody has to go to court, but we wanted to make it clear that if there was some gap in time and somebody didn't have coverage when they should have, that that individual is not on the hook then for paying for any health care costs that really should have been covered by the health plan. So we also clarify that.

We largely followed the remedy structure for the nondiscriminations that exist now under Title 1 of HIPAA, but we do make some, I think, important changes in terms of enforceability and also in terms of being a little more consumer-friendly than what's out there with HIPAA. The way HIPAA works now under Title 1 is kind of messy. It's three agencies are involved. States enforce for the individual market and if states fail to enforce, then HHS is the fallback enforcer. DOL enforces against the group market, in other words, employers who sponsor health plans, and then if there's any kind of violation that DOL finds, they don't have the ability to levy a penalty. They have to go to the IRS and bring them in to levy which is actually an excise tax under current law. So it's a lot of people involved, and as you can imagine, it's rather difficult to enforce that the way it is.

So we took it away from the Internal Revenue Code, the penalty here, and made it no longer a tax and we gave it to the Department of Labor, Secretary of Labor, and made the penalties civil penalties, civil monetary penalties. So now, when DOL goes and does plan audits, are you HIPAA compliant, they can also look at are you HIPAA genetic compliant, and if they find something wrong, they have the ability to go ahead and levy a penalty right then and there.

This is sort of the Secretary enforcing against the plan. The penalties would be the same as they are now for HIPAA Title 1, which is $100 a day per person per violation. So usually the way it works is if a plan is messing up, they're usually doing it on a planwide basis, so if you have 20,000 people in your plan, it's however many people you're treating this way times per day, and then they go up for more than de minimis violations. It goes up to $1,500, then to $2,500, and then for a really bad planwide violation, it's the lesser of $500,000 or two times whatever your total claims payments are, which could be a lot more than half a million dollars if it's like a General Motors plan where they have hundreds of thousands of people in their plan. So that's the change that we made to HIPAA.

We made one additional change which was a big concession on our part and made us somewhat uncomfortable but was really important for, I think, Senator Daschle to really give the consumers some ability or some leverage against the plans, because under ERISA now, you have a private right of action. We've also said in addition to the Secretary's ability to enforce a plan, the individual, if they are discriminated against and they end up suing and we give the court the discretion to award the HIPAA penalty to the individual -- so that $100 a day per violation and $1,500, $2,500, more than de minimis -- the court then would have the discretion to reward that to the individual.

So if I work for XYZ Corporation and I sign up for the plan and they don't let me in and there's no reason, except that I have a family history of breast cancer, and then I go and I sue and it turns out that they should have let me in, by the time I sue, say that's six months later and the court rules in my favor, that's $100 a day and they can direct it towards the individual.

I will say there are folks that feel like that's kind of opening a pandora's box because it's giving sort of like damages to the participant when heretofore most of the remedies have been on a planwide basis with the Secretarial enforcement, but there is also some precedent for it now in ERISA in a couple of places. Under COBRA, if your plan administrator fails to give you your adequate COBRA notice, you can go to

court and the court has the discretion to actually award the COBRA penalty to the individual, and there's also something in ERISA now that says if you request plan documents from your plan administrator and they fail to provide them within 30 days, then the plan administrator's on the hook for I think it's $1,000 penalty, and again the court has the discretion to award it to the individual rather than back into the health plan or to the general treasury.

So that's the framework, and I'm sorry it's so complicated, because it really does build in many ways on what's already out there and then there's some variations on what's out there, but that's the enforcement on the insurance discrimination provisions, and then on the privacy pieces, we kind of made a pretty big decision during our negotiations to split that out. The way we had originally drafted the Snowe bill is, it was all under that Title 1 framework, but the way the privacy regulations work now is very different from the rest of HIPAA. You don't have the multiagency enforcement. It is a federal standard. Unlike HIPAA Title 1 where states enforce against individual insurers and DOL enforces against group, with the privacy standard, it's a federal standard and that is the rule and states have the ability to do something more protective, but it's a federal standard that is enforced through the federal government, the HHS Office of Civil Rights, and we felt that HHS is going to be in the process of developing an incredible amount of expertise on privacy. They actually wrote the rules. They will build up their Office of Civil Rights and we didn't think it made sense to have pieces of privacy related to genetics enforced by DOL and IRS and kind of spread all over because they obviously do need to coordinate with the rest of the privacy rule.

So what we did for the enforcement of the privacy provisions is put it under the privacy enforcement framework under Title 2 of HIPAA, and the way that works is, it's a federal standard. It's enforced by the Office of Civil Rights, which we thought was important because we do feel it's a civil rights statute, and the remedies there are -- I apologize if I don't get this right off the top of my head. This is where I should have brought my summary information. But the penalties are similar to Title 1 but they're also a little bit heftier. It's $100 a day, but they graduate up for should have known, didn't know, should have known, knew but acted really badly, and it goes up to half a million dollars and also including criminal penalties for somebody who knowingly violates.

So that is the enforcement provision for the privacy pieces of genetics, and it seems kind of strange when we're describing the bill -- why did you split it up, it's hard to follow -- but I think from a consumer standpoint, actually it will make a lot more sense.

You know that all your privacy protections fall under the same kind of rules. You know you call the Office of Civil Rights and here are the remedies, and there's no interagency -- there's no question of, well, is it the state law or federal law or is it IRS or DOL or HHS? So we think that probably makes a lot more sense, and there's already on the nondiscrimination pieces, there's already nondiscrimination rules out there. So building on that framework makes sense because it doesn't make sense to have a discrimination rule for current health status under one framework and then a slightly different framework for genetic discrimination. So there is actually a rationale for how we ended up the way we did.

That is a lot of detail but that's the bill that we approved as a manager substitute in Committee, and I think everybody's very pleased. It was a unanimous agreement, and I think there's a very strong desire on both sides to move it forward quickly. We do have to report it out of Committee, which means we are going to draft a Committee report. We think it will help to have an English language explanation for some of these things, and our goal is to get that done as soon as possible and bring it to the floor, hopefully on a UC basis and not spend a lot of time having to do a floor debate, but as you all know, the beauty of the Senate is one person can object and then you do have to have a debate and schedule time. So we're hoping, given the amazing cast of characters who support this bill, from Kennedy and Gregg to Daschle and Frist and Snowe, that that kind of lineup really covers the political spectrum, that it would be really tough for anybody to object, but you never know in the Senate.

So our goal is to probably after the 4th of July recess because we're going to be turning to Medicare and that'll probably take up two weeks maybe -- I think there's a question about that -- bring it to the floor,

and hopefully pass it and then hopefully get your help and start working on the House.

DR. McCABE: Thank you very much.

I would point to everyone's attention to the last subsection under Tab 4 which is the summary of the bipartisan agreement that was prepared by staff. The bill is also there. I'm not sure how it's changed perhaps since.

MS. MONK: No, that's it.

DR. McCABE: Okay. So this is the way it is.

MS. MONK: That's what the Committee approved.

DR. McCABE: And I'd also point out that there is a letter, dated May 20th, from Secretary Thompson to Senator Gregg supporting this bill.

I think it's also important to recognize what was stated, and that is that everyone has anticipated that the real contest will be on the House side with this bill because things have come together so well on the Senate side and that's an area that we need to consider also as a group.

Questions, comments from the Committee?

MR. DANNENFELSER: Martin Dannenfelser from the Administration for Children and Families at HHS.

Could an insurer require certain tests, even if they did not acquire the information? For instance, could they have a policy that all women 35 years and older who are pregnant have to undergo amniocentesis, even if the insurance company does not acquire that information?

MS. MONK: That's a good question. It was my bad not to mention that provision.

We do have a ban in the bill that says an insurance company can never require you to take a test, request or require somebody to take a test. We do have an exception that says an insurance company can, however, as part of a bona fide wellness program, which is defined in statute now, can offer or make available information that, oh, you know, genetic tests are available for this or that through health care professionals.

So because we do know that insurers offer wellness services, quite a range of them, we don't want to curtail those types of activities, but the insurance company can never request or require somebody to take a genetic test. It's just a flat-out ban.

MS. MASNY: Thank you very much for such a comprehensive overview and the history that went into it and we can see all the work that went into it as well.

I have a question regarding, you mentioned that there will be a ban that genetic information cannot be used for underwriting purposes. Does that cover as well or apply to life insurance?

MS. MONK: No. The bill that we approved in Committee applies to employment and health insurance, and to my knowledge, nobody's really thought through the life insurance issue yet. It's a very different product than health insurance. The game of life insurance is figuring out what life expectancy is, and I think before anybody got into that, they'd have to have a better sense of what the implications are, but it's just health insurance.

DR. McCABE: Some of the agencies were mentioned, Labor, Civil Rights, EEOC. Anyone from those agencies wish to comment?

DR. ZURAWSKI: Hi, Kim. I have a question about the HIPAA changes.

Does the civil penalties only apply for violations regarding genetic information and not other HIPAA type of protections?

MS. MONK: That's correct. While we built on the existing HIPAA nondiscrimination provision, the changes that we made were -- and that is the clarification of your right to injunctive relief, clarification of reinstatement of your coverage, and I guess the discretion of the court to award the HIPAA penalty to the individual which doesn't exist now. We only did that for the genetic piece, and it was really a political reason for that, which was we didn't want to open up everything that was already out there and make it a much bigger bill, but that's obviously something that will come up and will be questions that you'll get in terms of enforcement.

MR. MARGUS: Just to clarify, I'm a neophyte in all this, but is there any difference at all in what the new law is as far as penalties for denying coverage versus just raising premiums?

MS. MONK: It treats them as the same violation.

MR. MARGUS: The same, regardless.

MS. MONK: Yes. Either way, it's discrimination.

MR. MARGUS: The second part of that is, I'm more familiar with what happens with individuals, but with the groups, my understanding is a company could be denied or charged atrocious premiums for group insurance if one of its employees has a preexisting condition of a serious disease, like even a genetic disease.

So in this case, it will now be impossible for that carrier to deny or overcharge for or to use genetic information to do that but not for a genetic disease, is that right? It's kind of like what you hope is that there are no symptoms yet. That's basically what happens, right?

MS. MONK: Well, this whole bill was drafted and we really actually did try to draw a line because the current law does deal with people who are currently sick, symptomatic, what have you. So we really did try to draft this bill so that it just deals with the genetic predisposition of healthy people, but even then you may have a family coverage and you may have dependents who are needing health care services here now. So you could get discrimination based on your family policy or what have you based on somebody in your policy, but I think you had several questions there.

To answer your question, yes, we do have a provision that prevents discrimination against the group as a whole. It's a little bit of a strange provision because you have self-funded group health plans out there. So to say you can't discriminate against a group is saying to the employer you can't discriminate against yourself. That doesn't really make sense.

But it could make sense where you have somebody buying a group health plan or employer, more likely to be a smaller employer buying a fully-insured product in which case you'd have an insurance company issuing a rate or issuing coverage, and in that case our prohibition does say you can't discriminate against the group as a whole on genetic information and genetic information is defined to include -- I'd just basically refer you to the summary, but it's not intended to get to current health status, but I think it is intended to get to the four corners of what would be considered genetic information.

MR. MARGUS: And this is new as far as protection?

MS. MONK: Well, under HIPAA, when HIPAA was passed in '96, there was a nondiscrimination provision, health status nondiscrimination provision, that said you can't discriminate against individuals in a group and that's premiums or eligibility under the plan based on health status, and health status was defined as like five different things -- evidence of insurability, claims history, receipt of care -- and genetic information was one of those.

So technically, genetic information has been out there for group health plans since 1996, but it hasn't been very fleshed out in terms of what is genetic information. So there are real questions about what it is and what it isn't and how it differs from health status items, and as I said before, that law only deals with discrimination against individuals in a group, and we took it a step further for genetics, just for genetics, and said you can't charge the whole group higher premiums because one person in there has the cystic fibrosis gene.

MR. MARGUS: Thanks, and thanks for your effort.

MS. MONK: Sure.

MR. MARGUS: I can only imagine what it takes to push something like this through.

MS. MONK: It's fun.

DR. FELIX-AARON: Kay Felix-Aaron, the Agency for Healthcare Research and Quality.

My question has to do with the use of genetic information when people leave the health plan. What happens to that information, and does the regulation address the use of that information, not people currently in the plan but what if people wanted to reenter that plan, can that information be used? One.

The second is insurance companies also engage in research activities, and does the genetic HIPAA regulation affect the use of genetic information in research by insurance companies?

MS. MONK: Two very good questions. The question regarding how do the protections work when you come and go from insurance plans and between insurance plans. The answer is they're meant to follow you and your information. So underwriting is a process that occurs when you first try to get into a health plan. It's most likely to occur when you first try to get into a health plan, particularly in the individual market. They may say what illnesses do you already have or what's your family history, now we're banning the family history, but that may occur also on an annual basis when they take a look at your rate and reassess you every year.

I think the important thing to remember is regardless of which health plan you're in, whether you're moving from one health plan to another, we basically apply the protection to the underwriting function. So it's protected at every possible point. Basically whenever somebody may try to use it for underwriting, we banned that. So whether you're coming or going or staying in the plan, it's covered.

Then to answer your other question on research, we were very careful and very concerned about not putting in restrictions that don't exist for other health care information that would inhibit research or quality activities or patient safety activities. So the short answer is the rules that apply, the privacy rules that apply to all health information really govern the genetic information, and I don't think we've done anything in our additional restrictions on insurers around their underwriting practices or business practices that would interfere with that.

Now, if the insurers say that the existing privacy rules interfere with research, then that would also be true for genetics, but I think there are still some questions about the research provisions under the privacy rules that are still being sorted out.

DR. FROHBOESE: And on that note, I'm Robinsue Frohboese from the Office for Civil Rights, and as you noted, we are the ones that are --

MS. MONK: I'm glad I didn't know you all were here when I was explaining this. I'd be very nervous.

DR. FROHBOESE: Well, we certainly, knowing what the Department went through in developing and then subsequently modifying the rule before it took effect, we are very pleased that this bill takes the approach of basically adopting the privacy rule and giving to the Office for Civil Rights the responsibility for enforcement.

The one thing that I did want to clarify, you had mentioned at the outset that one departure in this bill is giving an individual a private cause of action or being able to get penalties for violations under Title 1 if the health plan discriminates against the individual.

Under the privacy rule, there is no private right of action and individuals do not get monetary penalties, and I just wanted to clarify whether in this structure, are there any penalties that individuals can get under the privacy rule or is it adopting the same enforcement structure?

MS. MONK: It is the latter. It's adopting the same enforcement structure. The penalties that I was describing, the clarifications of current law and sort of the new rights, that is for the insurance nondiscrimination provisions. That's why we broke it up. So that is, if an insurance company says we're going to charge you more because you have the BRCA gene or we're just not letting you in the plan because your kid has spina bifida or the gene for it, that's the new or clarification of penalties are only for the nondiscrimination provisions.

For the privacy provisions, the use, disclosure, collection pieces, that does fall within the enforcement framework of the privacy rules, and obviously you would know better than I, but there's obviously, I think, a question under the privacy rules, which were regulations and did not create a new private right of action.

The states have passed laws. So some people may have the ability to enforce their state law privacy rights under a state law private right of action and this is something that's still very much, I think, in a state of gray in terms of who has what rights, depending on what kind of entity they're under and questions of when federal law preempts state law. That's very much a gray area, and we did include some language in the agreement that said we are not interfering with that framework. We're not taking away any of those rights and we're not interfering with those rights. So there's a general statement that we're respecting the preemption framework around remedies that goes with the privacy rules and not trying to put our thumb to tilt the balance one way or the other.

MS. MONK: You should look at that language.

MR. LESHAN: My name is Tim Leshan. I'm filling in for Dr. Collins as the representative for the NIH and NHGRI.

I wasn't going to say anything, but I know that I'd be remiss if I didn't just pass on his gratitude and thanks to you and the Senator for your extraordinarily hard work on this issue, and I know there's still more work to go, and we look forward to working with you on it, but we just want to express our gratitude for this piece of legislation because we think it's really a step in the right direction.

MS. MONK: Great. Thanks, Tim. You guys were a great help throughout the process and we appreciate it.

DR. ZURAWSKI: Just a follow-up to one of Brad's questions about preexisting conditions, that there is an existing HIPAA protection and sort of HIPAA rights around preexisting conditions that will no longer

be exactly like the genetic information type of violation, assuming this bill becomes law, but I think there are already some federal protections there.

DR. McCABE: Any other questions or comments for Kim? Yes, Martin?

MR. DANNENFELSER: Just a very quick follow-up. On the issue of requiring tests, is that current law now or would this be new, what you're doing?

MS. MONK: It's new. To my knowledge, Paul, there's nothing like that now. So this is new.

MR. DANNENFELSER: Thank you.

DR. WINN-DEEN: I just wondered if you might speculate a little bit on when this might move through the House and actually become law. I mean, you've made tremendous progress in getting it to the point that it's at, but it really doesn't help the community until it becomes law.

MS. MONK: Yes. Well, you know, usually people in the Senate are the last to know what's going on in the House. We have to hear it from you guys, but one complicating factor in the House is that there are two Committees of jurisdiction, and we had the beauty of having one and that was complicated enough. Actually, three. Well, technically, we had two in the Senate, but we only ever worked on it once. Technically, they have three in the House and it appears that two are interested and engaged and that's Ed and Workforce and Commerce. Ed and Workforce has the employment title and then they have the group health plan, ERISA stuff, and Commerce would have the all-important individual insurance market, and both Committees are interested in the issue and have looked at it.

Another kind of -- I don't know if it's a complicating factor, but it certainly is something that plays into how it's moved or not moved so far in the House is that in the Senate, we have always had a Republican bill and a Democratic bill, and so we've always had that tension between, on the one hand, both sides wanted to get it done, and we've had differences in agreement about policy about how to get it done, but I think that's really helped the momentum.

On the House side, they've only had the Slaughter bill. There hasn't been any competition, and I think that there are efforts underway to either negotiate with her and/or put a bill out that either looks like the Senate bill or the Republican alternative. It's a little hard for me to get a handle on, but I don't think there is the same kind of momentum over there, and so I am concerned. I'm primarily concerned that I know we spent 15 months on definitions alone, and when I talked to my House counterparts and they asked me questions, I'd think, wow, those are the kinds of naive questions I was asking a year and a half ago, and we don't have a year and a half for you to learn the way I had to learn.

So there really is a bit of a learning curve and just timing in that they're starting very late. So whenever I go talk about this, I urge people to really get in there and work with the House folks. I know many of you are not in lobbying positions, but certainly, hopefully when we pass it in the Senate, we'll get some really strong feedback from the White House and that will help create some momentum.

DR. McCABE: And just to comment, we had invited Christine Fitzgerald from the House Education and Labor Committee and she just could not come today, but we can try and continue to have contact with her.

DR. LEONARD: Does this blanketly apply to anyone with or obtaining health insurance or are there carveouts, like for groups with less than 50 members, that we still need to be concerned about?

MS. MONK: That's a good question. Again, ask an easy question, get a complicated answer.

On the discrimination provisions, there are no carveouts. The way HIPAA works now is the carveouts

are actually -- well, again I'm using HIPAA very broadly. There are different parts of HIPAA. You have the portability piece, you have the women's breast cancer rights, you have the mental health parity, COBRA. They all have different exceptions, but the nondiscrimination pieces of HIPAA out there today have an exception for what I call tiny group health plans, which are groups of one, and for retiree-onlys. So where you only have retirees in the plan, and we did not retain that exception. So that means there are no exceptions for the nondiscrimination ban, the ban on the testing and what have you.

Now, on the privacy side, we had to kind of rethink that because the privacy rules are obviously very comprehensive and there are some exceptions under the privacy rules and those exceptions are for groups of less than 50 that self-administer. There may be 10 or fewer of those in the country, but I'm sure there's a couple. Usually when they're small, they use a third-party administrator insurance company. What that means is those groups out there today don't have to put in that whole privacy framework. They don't have to have a privacy officer. They don't have to get their files and systems and investments in place to comply with the privacy rules. They don't have to sign business associate contacts and all that stuff.

So we felt that we really needed to pick up on that scope. Otherwise, we would be putting really tiny businesses, fewer than 50, who self-administer. So again, it's much narrower than the universe of 50 or fewer because it's only those who actually use their own employees to process claims, which is very rare. I don't know if you all might have data on that, but it's very rare. Because if we didn't have that exception for privacy, we'd be basically telling these really tiny employers in order to comply with these kind of several genetic use and disclosure restrictions which are really just icing on the cake on top of the base privacy rules, that now in order to do that, they'd have to have a privacy officer and they'd have to invest a lot of money in systems. So we'd kind of basically be back-in be forcing them to fall within the scope of the privacy rules, which we did not feel was fair.

So when it comes to privacy, it's the scope of the privacy rules. When it comes to discrimination, it applies to everybody.

DR. McCABE: Kim, I want to thank you very much for coming.

MS. MONK: Thank you.

DR. McCABE: And hopefully you will continue to remain in communication with us through the process and let us know if we can be of any assistance in moving this forward, though, of course, we work with the Administration side, so we'd have to work through the Secretary. But we do want to applaud you for making such progress.

I think, given the time and where we are in the agenda right now, we're going to take a 15-minute break. We will resume at 10:30 and proceed with the agenda from there.

Thank you.

MS. MONK: Thank you.

(Applause.)

(Recess.)

DR. McCABE: The next section is where we really begin to talk even in more concrete terms about the development of the SACGHS priority issues and work plan. I will be giving a presentation and will review the recommendations of the SACGT and the activities that were in progress at the time that the charter was not renewed for that Committee. Then Sarah Carr, who as we said yesterday also served as the Executive Secretary for the SACGT and now is fortunately for us our Executive Secretary, will provide a summary of the priority issues identified by SACGHS ex officio agencies and departments as

warranting the Committee's attention.

Since there are individuals who were involved with SACGT, both from the Committee and the ex officios, if going through, there's something you'd like to add to my presentation to clarify or in any way add to my discussion, please just let me know and I'll be happy to have you include your comments.

So the SACGT was chartered for four years but it took about a year to really get the group together. We functioned as a Committee from 1999 to 2002. The mandate was to identify policy issues raised by genetic testing and to make policy and procedural recommendations to the Secretary of Health and Human Services on how such issues should be addressed, including the safe and effective incorporation of genetic technologies into health care, the effectiveness of existing and future measures for oversight of genetic tests, and research needs related to the Committee's purview. So I think you'll see that the mandate for this Committee is substantially broader than before.

The policy recommendations of SACGT included reports, a report on oversight of genetic tests and methodology for classifying genetic tests into different scrutiny levels. We also had letters to the Secretary. Our first letter to Secretary Shalala and then with the new Administration, our first communication was Secretary Thompson, both had to do with genetic discrimination and health insurance and employment because as we had been asked by the Assistant Secretary for Health, Surgeon General Satcher, that we reach out to the American public, the American public was extremely concerned about genetic discrimination. We also sent letters to the Secretary about gene patenting and licensing and then secondary subjects and research.

So regarding the report on oversight, the oversight issue addressed was addressed at the request of the Assistant Secretary for Health, Dr. David Satcher. He charged the Committee in our first meeting with having a report to him as soon as possible. I think it may have even been like a three- or six-month deadline. We did not meet that but had it to him within the first year. That was a very specific charge which focused our energies over the first year.

It involved a comprehensive assessment of the adequacy of oversight of genetic tests, a broad multifaceted public consultation process, and a consideration of all options. It resulted in "Enhancing the Oversight of Genetic Tests: Recommendations of the SACGT" -- that was the title of the report -- and we recommended increased federal involvement in the oversight of new genetic tests through a flexible regulation by the FDA, augmentation of Clinical Laboratory Improvement Amendments, and development of a collaborative postmarket data collection effort by the CDC, and I think it's important to note that we did talk about flexible regulation by the FDA. There was concern that by putting this recommendation in place, we could shut down testing and we talked about new tests versus existing tests.

There were several iterations of developing a methodology for this, and as each of these was tested, there were certainly -- the first couple went by the wayside because of concerns that they did not really adequately address the needs and were difficult to implement when they were tried in specific testing examples.

I think many of us, and now I'll speak as an individual and not as the chair of that Committee, but I know that I was evolving to feel that what we really needed to deal with was labeling and that regulation could be very difficult to implement. I know the FDA has moved forward with this and perhaps they can bring us up to date with how that has continued, but we were beginning to talk about labeling as being extremely important and thinking in analogy with a Physician's Desk Reference or a PDR for genetic testing. That had come up in group discussions.

So as I mentioned, FDA is presently considering developing a proposed rule classifying analyte-specific reagents used in high-risk in-house tests, including genetic tests as Class II special controls or Class III premarket approval devices, depending on their intended use and risk profile. CDC and CMS are in the process of preparing a Notice of Proposed Rulemaking to develop a genetic testing specialty under CLIA,

and we heard this morning that that is moving forward and the CLIAC will take this up again. I guess actually what we heard was that this had not moved forward as I have here, but that the CLIAC will revisit it at your next meeting.

Is that correct, David?

DR. FEIGAL: That's correct.

DR. McCABE: Regarding the methodology for classifying genetic tests, SACGT considered several options for classifying genetic tests but concluded that classifying genetic tests based on a limited set of elements applied in a simple linear fashion for oversight purposes is infeasible, and part of the problem is that the same test used in different contexts, used when there's a positive family history, used when it's in a counseling process for a single family or used in a population-based screening process, may perform quite differently, has different demands, and may be ready for one context but not for others. So it's not the test, it's the test in addition to the context in which that genetic test is used.

SACGT's decision to defer further work on methodology was also based on significant progress made by FDA to develop an innovative regulatory process for genetic tests, including a template for facilitating and ensuring appropriate review of relevant data.

Letters to the HHS Secretaries included, as I mentioned, genetic discrimination, to place a high priority on the passage of federal legislation prohibiting genetic discrimination in health insurance and employment, and that that was a high priority.

I'll add parenthetically that one of our letters in earlier attempts at passage of these bills on the Hill, one of those letters to the Secretary from SACGT was blown up into a poster and used to try and move that forward. So we're very pleased that that is moving forward, at least in the Senate.

Gene patenting and licensing. We recommended that there be conduct of a study to determine whether certain licensing practices are adversely affecting access to beneficial genetic tests. This was a discussion that was had with parties representing a variety of perspectives on this. It was a quite heated discussion, as I recall, and we have heard some of where that has moved.

Secondary subjects to develop guidance to help define situations in which secondary subjects, meaning family members of primary research subjects, become human research subjects whose consent must be obtained or waived, and certainly we heard more about that from Kim this morning as well.

Reports that were in development. The HHS' effort to advance knowledge of genetic tests, genetic education of health professionals. We've heard again in this day and a portion of the importance of education of health professionals about genetics. Public understanding of genetic testing, informed consent and clinical and public health practice, reimbursement of genetic education and counseling services, development, translation, oversight, availability and accessibility of genetic tests for rare diseases.

Again in my role, along with the other two members of the SACGT who were appointed to this Committee, I think, and sort of under the subheading of lessons learned, that we worked very effectively when we were focused on, as a Committee, the original recommendations during that first year. I think we had some more difficulty finding our way and then, as we went out into work groups and became somewhat diffuse, I think it was much more difficult for us to make progress, and I would encourage this Committee to prioritize the needs and focus on one or at the most two efforts at any one time and to try and work as an entire Committee rather than diffusing out into work groups because that did not seem to work as effectively. I think it would be important. We can begin to serve as a forum for the broad range of topics but that in terms of products, we should focus on one or at the most two at any point in time.

In terms of efforts to advance knowledge of genetic tests by the Department, assessment of these efforts to advance knowledge of clinical validity and utility of genetic tests in both premarket and postmarket phases includes analysis of projects supported by relevant DHHS agencies and primary research, secondary analyses, summary information development, and summary information dissemination, and this was quite an effort that was undertaken by staff, develop case studies illustrating the development of three genetic tests to learn more about how the genetics work together and with the private sector to advance the validation and integration of genetic tests.

Regarding education, it involved an exploration of educational challenges posed by expansion of genetic testing and the adequacy of efforts to prepare health professionals to use these new technologies appropriately, and we've heard discussion of that today, and we held a policy conference in May 2002 that identified the need for teaching and faculty development, training of geneticists and collaborative teams, funding for translational research, outcomes research, meaning evidence-based medicine, development of pedigree tools and data on the application of lab guidelines, and reimbursement patterns and codes.

Regarding access to genetic testing services, that involved an exploration of issues and access to genetic testing services, including coverage, billing and reimbursement of genetic testing services, and disparities in access to genetic testing services, and we held a town meeting on the accessibility of genetic testing services and to hear perspectives on how the problem should be addressed.

Additional topics under study, the importance of public understanding and the need for information materials on specific tests and categories of tests to reach disparate communities. A generic information brochure targeted to the general public was to be developed as a model. Conceptual framework, correlating test characteristics and approaches to informed consent in order to improve decisionmaking and the consent process in clinical and public health practice, and assessment of the translation, oversight, availability, and accessibility of genetic tests for rare diseases.

So with that as a summary, I'd be happy to take any questions for a couple of minutes.

DR. LEONARD: Can you tell me what the products of this Committee are supposed to be? Are they simply supposed to be recommendations for work that needs to be done or does this Committee actually do work and produce products that are usable in certain formats by the public or professionals? I need to understand what we're supposed to be doing, producing.

DR. McCABE: Well, that's what we're going to decide during the rest of the day. Let me give you some examples, though, from the SACGT.

I would include among our products, there was a white paper that was done that was prepared under contract early on to bring the group up to a similar database from which we proceeded to make the recommendations. That ended up being very popular on the website and we heard was used in education of undergrads and others. So I think that's an important product, if it was used in the educational process, but it was really intended as a document for the benefit of our group.

The other products included letters, the recommendation for support by the Administrations of genetic nondiscrimination legislation, and those were received favorably by both Administrations as well as the other letters that I mentioned. That's a direct way that we can directly communicate with the Secretary and give recommendations to the Secretary, and then there were the recommendations which were longer papers that established sort of the foundation for the recommendations and then went into the specific recommendations.

I think all of these are the kinds of products that we can have. I think the other more nebulous product is serving as a forum for discussion, but until that dogma in medicine and until it's written down, it doesn't exist, I think that just discussing something is nebulous. If we can then move from that discussion to

something that is written and can be used in communication of the concerns, the issues, the priorities of this Committee, that I think that will have a lot more weight, but that's my own opinion and experience from before. I think it's important to have written products as well as the discussions.

DR. LEONARD: Thanks.

DR. McCABE: For example, Sarah was discussing with me this morning that a product that we could have would be the deliberations that we've had at this meeting, a laying out. It does not have to be a final recommendation, but it could be a discussion of the issues that have been laid before us and then a summarization of the discussions that will occur today because we've already begun to see some themes that have been brought up multiple times and just to put those down in written form and come to some document that we as a Committee could agree upon to move forward would be valuable.

DR. BEMENT: Did any of the products that came out of the previous Committee find its way into testimony or were they submitted as information for the record in testimony?

DR. McCABE: No, they did not, in terms of testimony before a legislative Committee, I assume you mean by that.

DR. BEMENT: That's what I mean, yes.

DR. McCABE: No, and we have to remember what our charter is, and we are a Committee of the Administration. We report to a cabinet-level Secretary. So that, our role is really to advise the Administration on these issues as laid out in our charter.

On one hand, in one case, however, I think it was the HELP Committee --

MS. CARR: The letter was used on the Senate floor, the poster.

DR. McCABE: Okay. The letter went to the Senate floor and was used there, but there was one time when, with the appropriate clearances, I think that some staff had some questions about some of the deliberations. So that, at one point, we did present to staff, and I think it was the HELP Committee but I could be wrong on that, but we had to have the appropriate clearances from the Secretary's office in order to do that.

DR. BEMENT: Right. No, I understood that and those caveats, but I'm just wondering if the Administration has seen the value of representing the work of the Committee to the Congress in that fashion, and I think you said in some cases, yes.

DR. McCABE: Yes. At least one of the products made its way to the floor and we're happy to provide guidance but that has to be with the appropriate clearance.

DR. BEMENT: Right.

DR. McCABE: And it's consultation. It's not really guidance. It's just our opinions.

Part of why that was requested was that we had been charged with public outreach, so we had also gotten a sense from the public, and we took that responsibility seriously, remembering that it was more than three years ago now, we were using Internet when it was in a much earlier stage to gather opinion.

Other questions or comments? Any comments from any of those of you who were involved with the SACGT?

DR.TUCKSON: I endorse what you said.

DR. McCABE: With that, thank you.

MS. MASNY: I know that you had already mentioned that the new Committee is not sort of a like a follow-up of the other Committee, but is there any way that we could make use of some of the information? Like for example, you have down that you were conducting the study on gene patents and had lots of discussion. Is there any way we could build on that as a foundation if we chose that as a priority area?

DR. McCABE: Yes. We didn't really conduct a study. We had a discussion at one of our meetings. We used that discussion. The Committee elected to use that discussion to recommend to the Secretary that there were health issues related to patents and licensure and recommend that those be considered. So I would assume that documents from the SACGT -- certainly the documents that were approved and moved forward are available.

What about the ones that were works in progress? Are they still available, Sarah?

MS. CARR: We can certainly share with the Committee the draft reports. In fact, we summarized the material in your briefing book and in the orientation book. I don't think in all cases we gave copies of the draft reports, more those that were farther along. I think the informed consent report is in there. But since they were draft and never approved by the Committee as a whole, they're not -- we don't post them on the SACGT archives, for example. But I think the whole point of Ed's presentation was to bring this Committee up to date on what that Committee did and as a foundation for you to consider as you go forward and define priorities.

DR. TUCKSON: Ed, I would say, in terms of the work that was done from the last Committee, one body of that that was detailed, I thought, very good was on the analysis of clinical utility, validity, specificity of tests, and the oversight process. That is a really, really complicated and, quite frankly, painful. Unless you do it every day, it is a painful area.

We went through that in exhaustive detail, and I would say if anyone wanted a very complete but I think a good primer on how the process works for oversight of tests, how you decide whether a test is useful to go forward or not go forward, you can certainly speed up your knowledge base if you would take a look at those sections. I think it's written at a level that -- I'm not very bright and I think I kind of got it after awhile.

MR. MARGUS: Can I just ask, so when you say that we don't need to focus on that too much because it was done so exhaustively before?

DR. TUCKSON: Sir, I would never make that kind of statement on behalf of the Committee. What I do take from your question which I think is useful is that that's why I was sort of asking Dr. Sundwall from his testimony earlier, is given all the work that has gone on and at a point in time we saw it, and I think, Brad, what I don't understand is what's happened in the two years since that analysis was done, and that's what I sort of was looking for, was whether or not the way the world actually implemented these things, the way in which the FDA and the CDC and the CLIA actually got their hands around this, was it effective? Is it working or is it not? Brad, I don't know the answer to that.

MR. MARGUS: Can I also ask about the very last bullet you had, which was the last Committee assessed development of tests for rare diseases, and what was the consensus on that? I've heard a lot of anecdotal evidence, but was it the general consensus that it was inadequate and there needed to be more or what?

DR. McCABE: Let me first give you the context in which that work group was operating. We had seen

data from GeneTests and there were other data as well. There was a paper published in JAMA by Peggy McGovern at Mount Sinai in New York, and the confluence of information was that if you looked at diseases for which there were genetic tests, the majority of diseases were actually rare diseases, which sort of fits with where we are in the history of genetics, that we're dealing with still the rarer "single gene" Mendelian traits, that a lot of those were being done in research laboratories.

This is a real problem for CLIA and for CMS as well. So there was a real concern about how to deal with this. How do you try and assure the public that they're having adequate oversight of rare disease testing without shutting it down and making it unavailable to a large group, a large population when you put it together collectively? So that's sort of where it was and it was a bit of a dilemma that the group was grappling with, but I don't think that work group had come to resolution.

Sarah's going to make this presentation. Lana Skirboll is Director of the Office of Science Policy for the NIH and Sarah is basically detailed to us out of that office. That's who Sarah works for, and Lana had some obligations at the NIH today that precluded her being available to us.

So Sarah, if you would?

MS. CARR: I also want to just address the point Reed made. If you'd like, the Committee can ask for a briefing, and I think even David Feigal might be prepared today to say a few words about where FDA is, but we could have a briefing, a more in-depth briefing, at the October meeting. Dr. Sundwall mentioned that the CLIAC is going to be looking at the CLIA regulation and how they want to move forward with whether to augment it for genetic testing. So that would be timely to hear from them in October about their deliberations. So that might be a way to bring you up to date on what's been happening in the time since the SACGT made its recommendations, if you'd like to do that.

Well, as Ed said, Lana couldn't make it today and she regrets it very much, and so do I because I think she would have made this presentation more fun than I'm going to. But anyway, we hope it'll be helpful to you.

What I'm going to present is the summary information about the perspectives of the ex officios, the 16 agencies that are represented on this Committee, and their perspectives on what issues they think are important for this Committee to consider.

I first want to tell you a little bit about the complicated process and how we harassed the ex officio agencies to request their help on this, but what we did was in March, we sent an email request to all of them asking them to identify the high-priority issues they thought, as I said, warranted this Committee's attention, and we used the seven areas of inquiry or functional categories that are set forth in your charter to organize the request. In each of the seven categories, we listed specific issues and we also suggested some other considerations that might be relevant in identifying priorities, and we indicated, though, that they were under no obligation to use this information. They were certainly welcome to think of the issues on their own.

As I said, we organized the request according to the seven functional categories, and I'm sure these are getting to be very familiar to you by now, but let me go through them. The first one is assessing the integration of genetic technologies into health care and public health; studying the clinical, ethical, legal, and societal implications of new medical applications and emerging technological approaches to clinical testing; identifying opportunities and gaps in research and data collection efforts; exploring the use of genetics in bioterrorism; examining the impact of patent policy and licensing practices on access to genetic technologies; analyzing uses of genetic information in education, employment, insurance, and the law; and serving as a public forum for the discussion of emerging ethical, legal and social issues raised by genetic technologies.

Now, I mentioned that we also suggested some other considerations that the ex officios might want to

think about as they were identifying priorities and these might also be relevant for the Committee to consider as you go forward in thinking about priorities. So what we suggested was to think about these questions.

Will the Committee's advice on the issue, on any given issue, significantly benefit society? Conversely, will failure to address the issue prolong any negative impacts it may be having? Is federal guidance or regulation on the issue warranted? Is there a governmental interest in receiving advice on the issue? Is there media attention or public concern about the issue? Is there a need for public discussion and understanding of the issue? Do sufficient data exist on the issue so that the Committee can develop informed policy advice? Is there another body addressing the issue or another body better equipped to address the issue? Does the Committee possess the expertise necessary to undertake a study of the issue?

Now, we had a very good response from the ex officios and I just want to commend all of them for the effort they made and the time they took to respond and they were very thoughtful in how they went about it.

Here you see the responses that we got, and I think it's important to note, though, that the ex officios used a variety of approaches in responding to the request. One of them ranked the seven functional categories in priority order. Some sent back priority issues that were on the list that we prepared. Others selected some of the issues from that list and then added some of their own and then some developed an entirely unique set of issues, and I have to say that this variety actually made the task of summarizing the data rather challenging.

Now, this slide shows the number of specific issues that the ex officios identified within each of the seven functional categories. So you can see that in the first one, integration into health care and public health, they identified 23 specific issues. With regard to uses of genetic information in education, employment, insurance, and law, they identified 19, 17 in the area of research and data collection. On emerging issues, they identified 11. On ELSI issues raised by new health applications and emerging technologies, they identified five, and on the use of genetics in bioterrorism, four, and finally three on patents and licensing practices.

Now, these final two slides that we're going to go through will show you which issues were most often selected by the agencies. As you can see, the use of genetic information in insurance, employment, education, and law was selected by eight agencies. The ethical, legal, and social implications associated with the use of genetic technologies to screen for traits as opposed to diseases was selected by six agencies. Then the following six issues -- standards for clinical readiness, the ELSI implications in new health applications, gene banking, the use of genetics in bioterrorism, impact of patenting and licensing on access, and genetic literacy of the public -- each were identified by five agencies. The last two top 10 pertained to the need for additional oversight and the impact of the privacy rule and they were selected by four agencies.

I just want to make one final point about this list of top 10, which is that they were distributed rather evenly through all seven functional categories.

Now, just one last thing. All of this information is in Tab 6 of your briefing book, including the individual by agency list, so you can see what DOD and Commerce identified in the first table in Tab 6, and then there's also another table in there that shows the priorities that were given, and we just hope this information might be helpful as you begin this important discussion of what you think the priorities are.

DR. McCABE: Questions for Sarah?

DR. BEMENT: I have an observation. As I looked at this list of top 10 issues, there seems to be a commonality in at least seven of the issues, Issues 1 through 4 and 8 through 10, in that they bear on the public trust and confidence, not so much concerning where it is at the present time but the extent to

which it could be a barrier or the means necessary to build public trust and confidence in order to move ahead, and so that crosscut might be worth also focusing on.

DR. McCABE: Yes, and that's one of the things we were going to talk about, is it's important for us to begin to identify those intersections because some of these do flow together. For instance, the one that was identified by eight agencies, is quite broad and that could be part of why it was identified by so many of the agencies.

But it's important, as you see these groupings, natural groupings, please help bring those to our attention because this is important for us to look at and may help us in identifying priority areas because we're not obligated to follow these specific areas. For instance, if we took that one that was identified by eight agencies, we'd probably have to focus on more than that because it itself would be too broad. So to the extent that we can redefine some of these categories in order to focus, that will be a valuable exercise for us.

DR. TUCKSON: For the record, I would like to indicate that on the slides, under a number of specific issues identified within seven functional categories, the next-to-last bullet, Use of Genetics in Bioterrorism, is not meant to be literal nor in Slide 9, Point 9, on the other one. So I just want to make sure. You never know who will read these things in the public record and decide that what we're here talking about is use of genetics in bioterrorism. It is a minor point, but knowing the way the world works today.

DR. McCABE: Thank you.

Other comments? Ideas of the Committee members or the ex officios on how we proceed to basically slice and dice to get to the priority issues? Reed?

DR. TUCKSON: I don't know how to get at it. Maybe it's in the material for our discussion today, is I think one of the questions that you asked, Sarah, and that Lana did, was what the relationship is between what's already being well addressed by the agencies, and I don't know how to tell from this summation -- can we assume that if it's on this list, that it says that this is an area that the agencies themselves feel like is not being addressed? Can we imply that?

Secondly, we need to be somewhat clear about which of these -- we are an advisory Committee to the Secretary of Health, and knowing that they all talk to each other, but our only portfolio is what can the Secretary of Health influence, I assume, or cause to be influenced, and so one of the things that's sort of a sense is that how much of that do we need to be thinking about in terms of looking at this list as, i.e., the agencies are saying we need more attention to this, therefore Secretary of Health, please, you need to be helped in getting this done.

MS. CARR: Well, your charter actually -- and there are 16 agencies, nine of whom are in the HHS but there are seven that are in other agencies. So the charter says that you're to advise the Secretary of Health and other agencies on request. So if there are issues in another realm that you think are important, it would be important to understand whether that department would be receptive to your advice, but I guess I don't want us to think that you can only speak to the Secretary, and I think copies of your reports and recommendations will be taken back by the other ex officios to their agencies because they may pertain to a health arena that relates to our Department but they may have some application, at least relevance, to the other agencies as well.

MR. MARGUS: So I also wonder about what's already being addressed, and then I realize also that people at these agencies are smart people who think about this all the time. So what are we supposed to add as a Committee, and I guess they have their fingers and tentacles out in the whole country. So it would be arrogant to say maybe we can bring our outside-of-the-Beltway perspective but maybe there is something else we could do, and one was maybe thinking looking forward, are there any trends coming

down the pike that we may be able to anticipate now rather than just thinking about what it is right now. So I'm not saying I can foresee those but maybe we should at least do a little ideating about what else could be coming that might affect it.

A couple of things we've heard about on the science side, we're going to shift now toward complex common diseases that are likely to be caused by multiple genes, either working in an additive way or a combinatorial way, and so genetic tests are going to get more complicated and then the utility of those tests might be a little more complicated, too, in the sense that you may not have as much of a certainty in the results, that there might still be use in them. The science would be one area to consider.

Another is what everybody eagerly bashed yesterday, which was the website things, but I didn't really hear the answer on who oversees marketing of genetic tests, but separate from all what seems like, as a layman, really rigorous review of clinical tests and how they're done and all that, then it feels like there's a lack of review on how they're marketed, and I know we don't like the bad science, junk science stuff, but there's no reason that there can't be other channels for it, but that's something kind of futuristic. The day may come sooner than we think when we'll have to care about that, and I don't know who's in charge of all the fine print that's mentioned when you see an ad for a drug company's product on TV these days. After the people run through the field, you have 50 sentences about all the side effects, but who's in charge of doing that for web marketing and everything else for genetic tests?

But those are just two areas, the science base or maybe on the consumer marketing base, but I think it would be a good exercise to go through just to think about anything else that's coming and maybe this Committee could come up with a statement on it, maybe not, but it wouldn't hurt for us to make a little effort in identifying those things.

DR. McCABE: I think certainly the direct-to-consumer marketing is complex and we did have discussions, but I think all that really showed me was how complex it was. Other than the fact that the regulation is fragmented, we didn't really get into that, but that's certainly something, given the audacity of some of these claims that we saw, that's certainly something we could explore, and so we can put that on the table as one of the things to look at as we move forward.

DR. WINN-DEEN: So I guess from my point of view in terms of time well spent, I would really like to see this Committee be focused on working on issues where we can make a difference and not just serving as a two-day discussion of stuff. Either it's not in our purview or there's really, no matter how much we talk about it, not too much to be done. So sort of in the category of things where we could make a difference or maybe some of the things like the genetic counseling group brought up this morning where there's some specific recommendations that they may come back to us with, which we can then debate and determine whether we as a Committee feel we want to make some very specific recommendations about how to advance the education of health professionals and the recognition of genetic counselors as health professionals.

I don't mean that as a single agenda item but that's the kind of thing where I think we could do something very concrete. We could have a Committee discussion and we could move ahead.

There are other areas, and again I'll just pick an example, where I think the issue of what the U.S. Patent Office deems to be patentable is probably outside of the charter of HHS or this Committee, and while there should be debate about that, I'm not sure that that's the kind of thing that we should be focusing our time and energy on. I think that the aspect of that that has been highlighted, which is, is there a barrier to access, that's where we should say that there's an issue there on access and health care provision to all, but I don't want to see us get sidetracked on things where we just are not going to be able to make any real progress and to have this Committee at the end look back and say what did we really do with all that time we spent together?

DR. FELIX-AARON: I was reviewing the third to the last and the second to the last slide, and it was not

apparent to me the connection between those two slides. So for example, the first bullet in the third to the last slide says integration into health care and public health care systems. I mean, that got 23 hits, but when I looked on the top 10 specific issues, none of those issues seemed to relate to the integration to care issue.

I make that point because a number of the things that have come up today focus and are directly related to the integration into health care. So the workforce issue, sort of the translation and implementation issue and access issue are clearly issues that yesterday and early today were brought up, and so I was just wondering how were we going to integrate sort of what we learned today and sort of the important themes that emerged into this priority list.

MS. CARR: I think you're identifying probably a part of the problem with summarizing that data in the way we did, but we also looked at it in another way, which was by the functional category, and when we did it that way, for example, seven agencies identified the integration into health care and public health. That's where most of their issues, if you look at them as a whole, fell, and so from the standpoint of the functional categories, that did seem to be the one that most agencies selected, and the seven were primarily the HHS agencies, but also DOD.

So I don't know if that helps at all, but I think you're pointing out something that we just cut it one way and there's a lot of ways to look at it, probably that data, or maybe not. Maybe we overinterpreted or beat it to death.

DR. FELIX-AARON: Right. I wasn't criticizing the methodology.

MS. CARR: Right. I know.

DR. FELIX-AARON: I was just trying to make the connection between sort of what is here and what we learned today and how what we learned today will help us in terms of determining sort of the two types of analyses and the way to move forward.

MS. CARR: Right. I think I'm revealing probably more of my misgivings with summarizing it.

DR. McCABE: So in follow up to what Kay said, what have we heard in the past day and a half that fits along with these priorities or other priorities that one might want to establish? I'll just throw one out there and that was from Kim Monk this morning. We heard the status of the genetic nondiscrimination. That's the kind of thing that if the Committee so desired, I think with Secretary Thompson's support of that legislation that we have here, dated in May, as well as prior indication of support that he gave us, it would be appropriate for this Committee to thank him for following through with that support and discussing what we understand to be the progress and offering our support for any future efforts in that area, but that's the kind of thing that we could do, based on what we've heard, but I'd like more discussion from you.

DR. LEONARD: Well, I agree with Emily completely, that we should focus on issues where we can have an impact, and the other point is when you look at something like integration into health care and public health, what are the problems? Are we anticipating that there's going to be an issue or have problems actually been identified that need to be addressed?

So I think if we focus on areas where there are issues, where we can have an impact, it would be better, and I don't know how much time we have, but I made a list integrating everything over yesterday and the first was discrimination protections at the national level, and I agree that we should send a letter from this Committee. I agree with you that we should support moving through the vote in the Senate and then moving it through the House so we can actually have a law in place. Like Emily said, until it's law, it doesn't do anybody much good.

The second issue is oversight of genetic testing. Since that was such a major issue with the SACGT, I don't know. There are recommendations that are in place that may or may not be being acted on. I don't feel that we got a lot of input from the FDA on where they're progressing with theirs or with CLIA, and so if FDA could be included in the October update, I think that that would be useful as to what they are doing. They seem to be focusing on ASRs rather than coming into laboratories, but from discussions here on oversight of genetic testing, there seems to be more concern not on the CLIA laboratories and the analytical testing that's being done but more on the postanalytical interpretation on the ordering of the tests.

So I don't know how everyone else feels, but with CLIA revising their regulations, I know I saw the draft of the original changes that they were proposing to include for genetic testing, and it extended beyond what the laboratory can do. So I don't know if this Committee can recommend to CLIA in their deliberations and in their new regulations to really focus on the laboratory rather than moving the laboratory as an intervenor into the doctor-patient relationship, which is very hard for laboratories to do, and then your suggestion that we focus on truth in labeling on genetic testing rather than actual FDA regulation.

DR. McCABE: Steve Gutman is here representing FDA for David right now.

DR. LEONARD: I know.

DR. McCABE: And was very involved with SACGT on the recommendations from SACGT. So perhaps you can comment, Steve.

DR. GUTMAN: I'd like to offer that the FDA certainly would be willing to come back at the next meeting and provide a more comprehensive briefing, and in view of the fact that some of the members are new and perhaps not familiar with our process, provide some background for what we do. We probably think it would be appropriate to share the stage with the folks from CLIA so that they can actually also clarify what they do, and I don't know where it'll be in terms of their revisit of their activity, but it might be highly timely.

We also might suggest, if you're really trying to look at the comprehensive selection of the options to get hold of this, it may be FTC might be brought in as well, so that there could be information on -- FDA does, particularly once it's classified something as a device, have some authority over the promotion, but obviously that's a major FTC effort, so they may be a player as well.

From our perspective, when the previous Committee, SACGT, the total product life cycle ran out and it was recycled into this new one, the Committee disappeared. The recommendations didn't, and as it's been publicly acknowledged, and I'll certainly publicly acknowledge it, that our agency is very hard at work on trying to move forward with the spirit of the recommendations. It's easier said than done. We've gotten queries from a variety of people, both in the manufacturing community and in the professional labs with notions that they'd like to leverage or partner, and we take those opportunities seriously and certainly input from this Committee on trying to provide us your insights in the direction we're going and how you view it would probably be quite welcome.

DR. WINN-DEEN: Steve, could we potentially also hear from somebody like Larry Lesko about the pharmacogenetics side?

DR. GUTMAN: Absolutely. Of course.

DR. McCABE: Just so we can really formalize this for our record, the people who we would want to invite would be FDA, CLIAC, FTC, and FTC is not represented among the ex officios but we can approach them, and perhaps among the ex officios we would know who would be the right person to approach in FTC. I don't know about that process, whether we'd have to go through the Secretary's

office. There's probably a protocol that we have to do when someone isn't represented here and we can explore that protocol, and then CLIA's responsibilities are shared between CDC and CMS, is that right, and certainly it's important to have their perspectives also.

DR. GUTMAN: It would really be important to have Judy Yost here, actually.

DR. McCABE: I'm sorry?

DR. GUTMAN: It would be very important to have Judy Yost here.

DR. McCABE: Yes, from CMS, Judy Yost. So that gives us a group of five.

I would suggest that we really focus on the status of regulation and regulatory progress at that meeting. I think we want to be cautious not to -- I was tempted to let's throw in the rare diseases and maybe we should get into the pharmacogenomics and those sorts of things, but I think we should focus on sort of where we are on the regulatory process because if we get too diffuse, I'm afraid we may miss where we are.

I think your suggestion, Steve, to give us a primer on the process -- because I know I get lost with some of the acronyms and everything. So I think that would be helpful, too, if you or one of your colleagues would be willing to do that.

DR. BEMENT: Trying to tie those two threads together and integrating health care with public health, one of the integrating elements, of course, is data and information exchange, and this gets into data formatting standards, privacy, encryption, networking, and I presume that would be part of the standards and regulatory discussion presumably.

DR. McCABE: I had some others listed here, but I think I want to follow up on this and then I'll come back to the others who might have taken us in a different direction, but let me just check.

Comments on this topic that we're discussing now? Agnes, was it a new topic or was it on this topic?

MS. MASNY: Somewhat on the same and somewhat different.

DR. McCABE: Well, why don't you go ahead?

MS. MASNY: Well, my first comment was that I wanted to just agree with your point about supporting the legislation and writing to Secretary Tommy Thompson to thank him for supporting the bill and that that would be something very specific we could do because I think that it came up by many speakers, the issue of genetic protections against discrimination as well as for privacy, and just to sort of reiterate what other speakers have said and from my perspective in my own clinical practice, that it is true that because of the fear that many patients have that they're not making use of the genetic tests or that fear is still there, so I think that would be a real priority area and something very specific and practical that we could do.

I just wanted to mention, as people sort of start talking about getting into specifics, is that because I think that the work that we were given to do and the charter that we were given to do looking at how genetics will impact health and society, that I think that maybe we should look at ways that we could also have sort of a broad focus as well.

One of the things that I was thinking of as we heard the speakers yesterday and today was looking at ways that we could actually see genetics be more integrated into health care. I mean, that's one of the priority issues, but I'm thinking that there was a statement from the Task Force on Genetics and Insurance that was a subcommittee of the National Human Genome Research Project ELSI Group stating that if we

looked at the genetic information as different from other medical data or other health information, it's conceptually confusing, practically infeasible, and ethically indefensible.

So I think that as we're approaching some of these things, if we look at genetic information totally in an exclusionary way and not integrative into how genetics will impact all of primary care and all of health, that I think we need to move in some way to help both the public and, as we mentioned about the workforce issues, of how genetics will be integrated and that we have to think of it as just another tool for medical and health care practice.

I talked yesterday evening at our dinner meeting to Mr. Tim Baker from the CDC, and I think one of the ways that that is already being done with the CDC is their way of incorporating and integrating genetic information into most of the bulletins that go out. So they now have a special section on how that's being integrated into all the CDC's information.

So that would be one way, and then as we integrate information, genetic information into health care and public health practice, I think that the issue of the validity of the interpretation of test is going to be critical. So that, if we have that briefing and we have more updates on the clinical utility and where we are with that, maybe the one thing that we could focus on would be as some of the criteria or a way to look at evaluating the validity of the interpretation of some of the information that we're going to be getting not only from single gene testing but the multiplex testing and the assays that will emerge from tumor or tissue in the future.

DR. McCABE: You know, we were headed down the path in terms of what we were going to do the next time. Debra, now you have affirmed the proposal that we basically send a thank you to Secretary Thompson.

DR. LEONARD: Well, not just thank you, but actually somehow supporting this further -- I mean, thanking him.

DR. McCABE: Right.

DR. LEONARD: But also, if there's anything that can be done to facilitate the process to law.

DR. McCABE: Okay. So could I have that as a motion, so that this would be something we could move forward?

DR. LEONARD: I so move.

DR. McCABE: And I'll take your comment as a second, Agnes.

Any further discussion on this? Clarification?

(No response.)

DR. McCABE: Given the need to move forward, if this is going to be dealt with after the July 4th recess, then it would certainly be something we'd need to move forward before our next meeting. So if we have a sense of what would be included in that, and if you could give us the go-ahead, meaning Sarah and her staff working with me, to move that forward. Would that be acceptable?

DR. LEONARD: Yes, and can the letter be sent out by email, so we can look at comments?

DR. McCABE: Yes, and we'll certainly have it out.

DR. LEONARD: But it should definitely go before our next meeting.

DR. McCABE: Yes.

DR. LEONARD: And in fact, as soon as possible.

DR. McCABE: Any further discussion of that letter?

DR. WINN-DEEN: Who's going to write it?

DR. McCABE: Who's going to write it? Staff will write it for my signature. But we will have it reviewed by the Committee.

MS. CARR: But if you would like to, you're welcome to.

(Laughter.)

MS. CARR: Yes, it will be signed and it will come out to you all for review before it's finalized.

DR. McCABE: And I could tell you the staff is extremely articulate, and then with the input that all of you will have to refine that.

Any further discussion?

MR. MARGUS: When you invite these people to the next meeting, besides this --

DR. McCABE: Why don't we deal with --

MR. MARGUS: All I want to say is we give them a primer on what they do. It would be great if they came just with some thoughts on what they thought was broken.

DR. McCABE: Okay. Motion we have on the floor. Any further discussion?

(No response.)

DR. McCABE: All in favor, say aye.

(Chorus of ayes.)

DR. McCABE: Any opposed?

(No response.)

DR. McCABE: Abstain?

(No response.)

DR. McCABE: So we will move forward on that letter to Secretary Thompson. Thank you very much.

So now, going back to the next meeting, the other group that -- I don't know if it should be in there, but if we're getting into the advertising side and with FTC, does Commerce then play a role?

MS. CARR: Well, FTC is an independent agency. So are you thinking other agencies within Commerce that would also be involved?

DR. McCABE: No, no, that was anyone.

MS. CARR: Anybody?

DR. BEMENT: I don't know. We could perhaps help with a workshop to get input from the private sector. It seems that a more appropriate role would be to look at it from a technology policy point of view.

DR. McCABE: Okay. Good.

MR. MILLER: Just to go in a little bit of a different direction in thinking about what this Committee might sort of intersect with an issue that's been raised a number of times, both this morning by Ms. Monk and throughout the day, dealing with the issue of genetic information and discrimination and insurance and employment and also added on here education and law, a number of people, both on the Committee and others, commented that there's really a dearth of information, of experiential information. There's not a whole lot of sort of cases coming forward complaining of discrimination and everybody is sort of foreseeing this as a problem sort of around the corner.

I wonder whether it might be an appropriate use of this time to sort of get to use the Committee and the various different points of view from the Committee to really get a sense of what is the concern out there, how deep does that concern go, how many people do feel that they're currently experiencing discrimination, to get a larger sense for the scope of the problem as it exists today because there's not a lot of information out there about that.

DR. McCABE: Now, these data are old. They're probably three plus years old and they were anecdotal.

MR. MILLER: Right.

DR. McCABE: But when we approached the public, this was a major concern, and as has been mentioned, while the number of cases are relatively small and tended to be among the self-insurance where the employer is also the insurer and you're certainly aware of that since you've been involved in those.

MR. MILLER: Right.

DR. McCABE: The public has extreme concern and is having the testing done anonymously or under pseudonyms because of their concerns and that creates certain problems as well.

This would be the kind of thing that we could certainly explore, though I wonder if perhaps you and your colleagues have already explored this and written about the cases that have come before the EEOC.

MR. MILLER: The issue is that there really haven't been any, I mean, with the exception of one case which got a lot of notoriety and which we prosecuted and resolved against a railroad. There really haven't been any cases that have been coming forward either to the EEOC complaining of employment discrimination on this basis or, quite frankly, in any of the state agencies, the state human rights commissions, that enforce any of these state statutes.

There has been anecdotal evidence and stories and some of the consumer groups talk about it and there's a lot of anecdotal talk about the existence of discrimination, but there isn't really a lot of cases that have been coming forward in any sense and there's a great deal of confusion and question about why that is.

One of the issues that has been sort of chewed through or put on the table in terms of some of the public discussions around the legislation and these other issues around discrimination is that it's really premature in that there are really very few cases coming forward. It may be worthwhile to go back and to explore some of those issues.

DR. McCABE: There was another one, which were the alpha-1-antitrypsin cases, though that one was under the ADA.

MR. MILLER: As was the railroad case, but the case with the alpha-1-antitrypsin never went forward into a lawsuit.

DR. McCABE: One of the things, also, that I've learned from you is the constraints on time on the reporting.

MR. MILLER: Yes.

DR. McCABE: And it might be good at some point -- Sarah was commenting that the Alliance has been exploring this, and it might be good to --

MR. MILLER: Or even to explore barriers, to the extent that there are anecdotal stories of evidence of discrimination that have occurred out there that people talk about and that groups know about but that they're not being translated into either enforcement of rights or what have you. If there's a sense of barriers out there or if there's a sense that people just don't believe that there are any laws currently protecting them or there's a lack of knowledge about sort of what their rights are or if there are other issues in play to try to better understand the issue of discrimination in this area as it appears that the sort of legislative train with respect to discrimination bill, whatever it may be, I mean, that's occurring on a separate track and there is little that this Committee can, I think, do to get involved in that process.

DR. McCABE: So that, perhaps we could have staff confer with the Alliance and with you to look at that.

MR. MILLER: Or the genetic counselor group or some of the other groups that are represented by your Committee.

DR. McCABE: Thank you.

DR. ZURAWSKI: Can I put a little context on that?

DR. McCABE: This is Paul Zurawski from Labor.

DR. ZURAWSKI: For the group health plan environment, we at Labor had taken an audit based on 2001 information of health plans, trying to examine whether they were violating preexisting conditions or HIPAA, including some health status terms, and the truth is that this was not based on complaints or cases. This was a statistically valid random sample type of audit, and there was very few, although, I mean, measurable, but under 1 percent type of evidence of health plans having terms within their plans that would violate HIPAA.

Now, the rules were not yet final and so it isn't in terms of a real-time subject matter. It would be interesting for this Committee to include the health insurance type of aspects of discrimination and we could provide, at least from the group health plan context -- the HHS has the individual market jurisdiction -- our findings, if required or asked for, regarding what we've seen and we have provided some of that information to the HELP Committee as they were deliberating because there was some question about whether or not we, in terms of -- I think last year, we had 184,000 consumer inquiries about their health plan and having almost none of those register as having a genetic information type of concern.

DR. McCABE: Others have had their hands up and these issues may have already been addressed, but let me run through them.

DR. HOOK: Since you're inquiring as to agenda items for the next meeting, I wanted to bring us back because I'm going to focus on the first three points of our seven enumerated functions and just to articulate support for speaking again with Ms. Bennett in terms of the counselor shortage and seeing if we can move along the process for encouraging the development of manpower. I think that would hopefully be able to be moved relatively expeditiously.

DR. McCABE: And we have asked for information within the next month, so that will be able to help guide us once we have that.

DR. HOOK: Yes. And in terms of our ELSI mandate, I think it would be very, very helpful as we are wanting to or are charged to explore some of these questions and PGD was one that came up frequently in the list from the supporting ex officio agencies, that if we can't fit it in in October, at least in the near future, we should have formal presentations from the President's Bioethics Commission and from the HGP ELSI Project on the status of where they are on some of the issues that they are discussing of this nature, such as the difference between treatment and enhancement and trait selection and things of that nature, so we're not reinventing the wheel but knowing where our colleague organizations are in their study of these issues.

DR. McCABE: Thank you.

DR. FELIX-AARON: I would also like us to consider in terms of the issues that we focus on a balance between macro policy issues and practice level issues, because I think a number of practice level issues, issues that patients and providers come up against every day have emerged here, and just to point out a couple of them, the issue of workforce development, the issue of providers having genetic competence and the number of providers, counseling providers available are some issues that came up.

The other thing that I heard in the area of practice is sort of the guidance that the field of patients and providers need in terms of oral translation from development, transitioning from the development of technologies to the application of technologies and what this group could offer as guidance to that whole area.

DR. McCABE: Thank you.

MR. BAKER: Tim Baker from CDC.

A couple of points were discussed earlier. The complexity of oversight of genetic testing that was well debated, richly debated in the previous Committees, include a couple dimensions, and Reed was addressing it earlier. There was a very complex but thoughtful report about the various elements of that.

So I want to bring forward that a consideration for it, if not at the next meeting, an additional meeting, would be the distinction between the regulatory to nonregulatory solutions for some of these questions. There is clearly a huge focus, and I heard you clearly speak to the emphasis on the regulatory process at the next meeting which is appropriate. The prior discussions have been this acknowledgement that there's a wide array of data collection challenges ahead that can be done through nonregulatory means as well. We have taken some steps forward in working with groups to try to characterize the data that is available through some disease-specific and test-specific considerations of what exists that would characterize clinical validity or clinical utility and how does this give us a potential for a model framework for collection, and then the question is how do you collect that? How do you work with laboratories and needing a stick or a carrot to do that?

So I've put that back on the table for discussion, and we do have a couple of projects that we're looking at, basically to characterize what is known and what is not known, and we think that's an important building block before we ever get to this notion of a large cohort study. It's sort of like what are these elements necessary? What's the potential framework, and how might we go about it?

The second point I'd like to put on the table is in response to Mr. Margus' question, and actually your question as well. We've struggled a lot with the notion of what does this integration into mean, and particularly for people whose day job is not genetics. It's like how do you make sense of that? Where's the traction point and where's the lens?

So what we find in working with our challenges in public health broadly and with our colleagues in public health and in the state and local settings is given what you're trying to do to understand and prevent asthma in this country, when and how and where will the developments in asthma fit into that, and what is it going to take to understand that context, so it's not driven so much out of genetics as it's driven from asthma as a broader challenge, and then understanding this science needs to be a tool that fills in gaps in knowledge, becomes an enabler toward that solution. What kind of training's necessary in guidelines?

Actually, Dr. Burke is leading a team within the Center for Genomics and Public Health at the University of Washington to look at asthma as a follow-on to a conference we had. We looked at some discreet diseases that asked the question, given these diseases and the pervasive challenge in this country, does it mean anything yet? If it does, who does it mean something for? And if it does, how do we further fill in the gaps of knowledge?

So that's the kind of lens that we're coming at that may be helpful to your deliberations in the future.

DR. McCABE: Yes, one topic has come up, and I just want to comment on it because I learned this with the SACGT, and that has to do with clinical utility. So there's analytical validity -- can you measure the same thing and get the same answer? -- clinical validity, which is how does the test function in a clinical environment, and then clinical utility, how useful is this information in clinical decisionmaking, and whereas the public in our discussions with them, they're ultimately interested in clinical utility. How helpful is this to them, and certainly with some of the website direct consumer marketing, this is clearly an issue because there'd be questionable clinical validity of doing a pH test on the urine for gender selection.

However, it became very clear from organizations like the AMA and others that they consider clinical utility the practice of medicine and staunchly defend the right of individual physicians to practice medicine independently without restriction and without regulation. So we need to be cautious as we use these terms, whereas it seemed fairly clear that people want to know how good is this in my health decisionmaking. That will lead us into some waters that are going to be very treacherous because there are people with major stakes invested in that distinction. So we just have to recognize that.

MS. ZELLMER: I just wanted to say, just like Emily, that I think, I hope that the ultimate outcome of this Committee is that we can focus on areas where we can do the most good, and while I think that protecting consumers against some of these tests, the ego-genomics test, is certainly a concern. I guess I hope that we don't focus all of our efforts in that area. I do think there are other ways of addressing those issues through state consumer protection statutes, things like that, that may be more effective.

I think if you want to help individuals who are dealing with genetic disorders, I think one huge area that I have personally experienced and my experience with other families dealing with genetic disorders is just lack of information of health care professionals, and I think that maybe that's an area that other agencies or other Committees are addressing, but I think that lack of information is a tremendous problem. I think most families I know get most of their information off of the Internet and rarely get information from their health care provider, and I think that as technology, genetic technology moves forward, I think there are a lot of health care providers who don't understand certainly the rare diseases but I think as we move forward with the sort of more common diseases where there are going to be genetic and environmental components, I think there are a lot of primary care physicians and even some specialists who don't really understand all of the implications and they don't know how to advise their patients to make informed decisions, and I see this as a big problem and I would hope that we would spend some of our efforts on

how we get the information out to health care professionals so that they can advise their patients of the best course of action for treatment.

I also think that informing the public would be very important. I think that would perhaps help some of the consumer protection issues that we're talking about, is if we can better inform the public, I think that maybe they'll make better decisions on whether to buy genetic face cream or not. But I think those are issues that I know that have been sort of uniformly discussed by each speaker as these education issues, and I certainly think that from my viewpoint, those would be the most helpful to the consumer.

DR. McCABE: Thank you.

DR. REEDE: This is a follow-up somewhat on what Kim has been talking about and others have mentioned in terms of this integration, this recurring theme of workforce and education and really understanding that much of what I'm hearing is very much in generalities in terms of there's a need for people or there's a lack of education or there's a lack of preparation without any real clear solid data in terms of what type of education is being provided, be it in medical education or continuing education. What are accrediting bodies or boards thinking about doing? What are certifying bodies thinking about doing? What is the status of this now, and what is planned for the future?

As I think or as I look at the top 10 areas that Sarah identified, much of what is included in there are topics that are going to end up being related to the future of education, and how is that being handled now? I think not for the October meeting but I think for a future meeting really being able to deal with the workforce education issues, the status, the need, getting a handle or an understanding of suggestions for what could or should be done in direction would be an important undertaking for the Committee.

DR. McCABE: Thank you.

DR. TUCKSON: I think where we are in this is I think we're just starting this discussion on prioritization and trying to figure out how we will lay that out so we'll know what things we want at the next meeting to start drilling down.

As I listen, I just want to keep also, and as people listen, as we listen to each other and learn from each other in this part of the discussion, there's one area that I just want to also keep in front of us and that is, genetics inevitably will not be something different from health care. It is health care. It has implications but so do so many other parts of health care today for all of the issues that are on our plate.

What I think here is important is to also keep in mind, is that, how we make decisions around the use of this information, the access to it, the affordability of it, and the integration in terms of how decisions are made or not made, I think, is exceedingly important, and I want to just keep the reality in front of us that affects all of the points that we're making, which is still that people can't afford what we have now. We've got 41 million uninsured people now. We've got lots of a context that shapes this new movement going forward, and I urge us to at least keep those in mind as we think about what our priorities are here as well.

I think that that sort of gets me towards beginning to think very carefully about the importance, as Kim has indicated, of how we educate the public to make appropriate choices and how the public can participate intelligently in the clinical decisions that are increasingly complex with their health care team and there are an array then of implications that result from that which we might be able to talk more about.

DR. McCABE: Thank you.

DR. FROHBOESE: Actually, I'm following up on Reed's comments and looking at the Committee's desire to identify both concrete issues as well as concrete ways in which to have a positive impact.

I agree that the Committee taking a position on issues, such as writing the Secretary, is very important and will have an impact, but one other thing that I'd urge the Committee to do, and this also gets to the issue of informing the public and I think following up on Paul's comment of lack of information about whether there really is discrimination, is to consider either holding hearings or town meetings.

I'd actually like to hear a little bit more from Dr. McCabe about how it worked with the Genetic Testing Committee because I see that you held a town hall meeting on the accessibility of genetic testing, and one thing that the Committee might want to think about is serving the role of either convening town halls on particular topics, to get information out to public and to get the public's views about issues, or stepping into areas, such as discrimination in use of genetic testing, to get expert and public testimony to then compile this information and step in and fill this informational void.

The National Committee on Vital and Health Statistics, I know, has sponsored both town halls and hearings on topics, including, for example, the impact of the privacy rule, and we in the Department really use this information to inform us about areas and directions that we need to go in terms of public education. So that might be something to consider in terms of both a method and then identifying issue areas that could be a concrete approach for this Committee.

DR. McCABE: Debra, you want to comment on that?

DR. LEONARD: I'm hearing from a number of sources that more information is needed about whether or not there is discrimination based on genetic testing.

My question is, is that something that this Committee wants to focus on since, in my mind at least, we have the discrimination bill that's moving towards law. I don't know at this point. I mean, the conditions are going to be changing under which there is or isn't discrimination if this bill becomes law, and I don't know if this is a real need and problem area and maybe you can comment or whether this is something that should be more on the back burner and us dealing with issues where we can have a real impact.

DR. McCABE: Yes. We heard from Kim, and I think it's likely, that it will move successfully through the Senate, given the support that it has had and the broad base of support that it has in the Senate. I think we also heard, I know she gave us the caveat that the Senate doesn't necessarily know what's going on in the House, I think from the read that many of us have, not much is happening in the House and the House has not seen this as the same priority that the Senate has.

There's hope that given the majority in the House and given the support of the Administration for this effort, that they might be able to bring it together, but I think many have been skeptical that this is going to make it through this year. I think it's been in process for at least three years. Seven years, seven years. So it's been in process for at least seven years.

So I think that while we're more optimistic than we have been in the past, that's just because it's making some progress this year, but I don't know that it's such a done deal.

MR. MILLER: If I can just jump in, one of the concerns that's been raised around the bill is or one of the issues around the bill is that there really isn't evidence or data that discrimination is currently occurring, and so I think that one of the issues that's going to be thrown into the mix into the House as the House begins to consider whatever bill they begin to consider is, in a sense, is this timely? Is this a problem going on out there in the world?

While there are some cases of great notoriety, including the one brought by my agency, there isn't sort of a landslide of these kinds of cases yet and that, I think, raises two questions. One is sort of to what extent beyond anecdotal stories here and there do you use to build a case or to understand the problem, and secondly, if there are a lot of anecdotal stories but nothing's turning into complaints in any formal kind of way, are there barriers out there that preclude complaints?

Well, either it's because genetic information is so amorphous and it's all over the place, people don't know that that's the reason why they're not getting hired or not getting promoted. People aren't saying we're not hiring you because of your predisposition to cancer. People just don't know why and maybe that's a barrier or maybe there are all sorts of other barriers. People may feel that, gee, this is such private information to come forward and make a complaint, that's going to reveal information about me and my family that I simply don't want to put out there in the public realm. Maybe that's a barrier.

So to get a more analytical understanding of sort of what's going on out there in the world I think would be a useful thing. As Robinsue said, sort of begins to fill the vacuum of sort of hard information that's out there, and I think that's why I'd raise the issue.

DR. McCABE: And I agree that I think that this could be very helpful, if it moves forward and it is successful. Still, frequently legislation isn't perfect and needs to be tweaked and this may help us with that process as well.

DR. LEONARD: So should this go under that first bullet of things that we're going to do to move toward nondiscrimination based on genetic testing? Is this something else under that category of work that this Committee would like to do, that we should try and facilitate this?

DR. McCABE: What I had as a note from the previous discussion, that the Committee staff would confer with the Alliance, EEOC, Labor, NSGC, and others regarding cases of genetic discrimination and any barriers to reporting.

Sarah? So I already had it as a note to myself to talk to staff about this, but we can --

MS. CARR: Well, I wonder, too, about something Paul Miller said about perhaps one of the barriers might be that people don't even realize that the reason they've had an adverse action or something that relates to genetic information, but Paul Zurawski said that you've had 184,000 complaints from people.

DR. ZURAWSKI: On all benefit issues.

MS. CARR: And would that be data to look at?

DR. ZURAWSKI: Yes.

MS. CARR: To scrub through it or work through it to see whether there are some underlying genetic things going on?

DR. ZURAWSKI: Well, we've tried to even break it down, that it would include a genetic classification of complaint. So when our field people who work for the agency receive those kind of complaints, we classify them for our own research and data needs, and so there is some scant evidence of some information that we'd be happy to share with you. It's not as specific as perhaps you would like, but it has to do with more of the timing of where this is going because we couldn't check on rule to see if a plan, a health plan was following the regulation, if that regulation wasn't in force or in final reg. So there was a little bit of that going on, but we do have some real information from 2001.

DR. McCABE: Barbara, would you comment on this topic? Okay.

Just following up with this topic then, Cindy?

MS. BERRY: Paul covered a lot of what I was going to raise, which was, that while in some respects, the train has left the station and so one might wonder, well, why are we going to even tackle this issue of discrimination and analyze it if there's already a bill and they've pretty much taken it as a given that there's a problem that needs to be addressed in some fashion, but on the other hand, if the arguments that

are being raised in opposition are really a primary obstacle to House action, then it sounds like there needs to be more than just a letter saying we think this is a good bill, but we could actually help the process along by providing the data.

Now, the old trial lawyer adage of you don't ask a question unless you know the answer could come up as well because if we don't have anything positive to offer, we just have a few anecdotal stories and that's it, in a way, it's reaffirming what the opponents of this legislation already believe. I'm not saying we shouldn't go down that road, but I raise that as something to consider.

My only other question on this topic is what are our capabilities? Is really the best thing that we can do to hold town meetings and gather anecdotal evidence and then talk to the agencies for what they have or is there some -- it almost sounds like there needs to be some in-depth research or a little bit more elaborate study, and I don't know that that's something that we're capable of doing.

DR. McCABE: We can. We can do that. I mean, we can't do in-depth studies. That's not what we have the ability to do, but we can certainly gather data from the agencies here.

Is that fair to say, Sarah?

MS. CARR: Well, sure, and I think we've had the offer of some, but we can't survey. We can't conduct surveys without OMB approval, and so we are limited to a degree in how we can go about -- we can certainly consult with the public and ask for their input on issues and so on, but I'm not sure that if we did that, that would qualify as quantitative data or also be considered anecdotal. So we are somewhat limited, and although I don't know whether the ELSI program has any grants currently that might be looking at this or that might be something to consider, too. I don't know.

DR. GUTTMACHER: If you're asking whether the current grants that are surveying cases of discrimination, none that I know of. I don't believe anybody's --

MS. CARR: Did you ever do that in the past, Alan?

DR. GUTTMACHER: I don't remember ever receiving any applications to do that.

DR. McCABE: Kim, you had a comment on this topic.

MS. ZELLMER: I mean, I would guess from what I'm hearing and sort of my gut feeling is that this is more of a perception problem maybe of people with genetic predispositions than maybe actually a problem, but just the perception, isn't that something that maybe could be addressed by the legislation?

I mean, I don't know. I'm going to guess that most employers are not sophisticated enough to even know not to hire someone because they have some genetic predisposition to some disease. I would guess the reality of it is that probably there aren't that many people that are discriminated against because of a genetic disorder, but I have a feeling that there are probably a lot of people out there who have genetic disorders and they're not disclosing them, certainly to their employers, because they have this perception that their employer may use that as a hiring decision or advancement or something like that.

Just by passing this legislation, perhaps that would give them a little more comfort in getting genetic testing done and getting treatment or whatever that they might need that they're not getting now, and I would guess it's probably more a perception than actual reality where we have a lot of people who are getting fired from their job or not getting a position or not getting advancement because of genetic disorders. I'm going to guess it's probably more a perception problem, but I also think that that's just as real, and if people actually believed that there's a problem, then I think this legislation could certainly help.

DR. McCABE: Someone from the audience, do you have a comment on this? If you could be brief, please?

DR. ROSE: I'm Ann Rose. I'm with Vicro, but what I'm identifying is a previous life. I was at OTA from '81 to '83 and one of the major topics that we worked on -- I don't know how many people know what OTA was, Office of Technology Assessment -- was genetic testing in the workplace, and it seems to me from these discussions that all the issues are still the same regarding discrimination, et cetera, anecdotal data that was there in 1981-83 has not changed. The only thing that's changed is the progress of where we are in genetic testing, and it was the House that commissioned that study to be done. Gore was there at that time, and as a result of not being able to have any more than anecdotal data about discrimination, it went nowhere, and my concern is that will the House again use this as an area to block it versus what you've got going seems to be a train that's running in the right direction and may be helping the House members and encouraging lobbying groups to do that to get the Senate bill through may be a way to consider it.

DR. McCABE: Thank you.

Barbara, and then I want to do some summation and some housekeeping.

MS. WILLIS: You know, as we sit here and wait to talk, everyone brings up what you're going to talk about in the first place. So I guess I'll try to really cut my comments short because so much of it has been touched upon already.

I think my main concern and one of the global topics that I hope that we'll be able to talk about in this Committee basically has to do with access, and again several people have touched on the different aspects under that. Just to enumerate some of them, I think obviously the workforce issues, and I think numbers are important. How many medical geneticists we have are out there, how many genetic counselors we have out there, and another layer on top of that is the diversity of the people that are providing these services because we know and it has been noted in the literature very clearly that people tend to be more comfortable receiving genetic services from people that look like them or they feel come from the same background and have the same interests, and so I'm not even talking from that strictly from an ethnicity standpoint but also religious standpoints, the levels of ability standpoint, and so I think all of those are issues that we really need to discuss and hopefully come up with some recommendations about.

Another aspect is education of the public and again not just education of certain aspects of the public but all of the public and so putting information on the Internet is not going to do it. There has to be other efforts in addition to that, and I don't know what agency may have already addressed that, and I would find it useful to find any information about efforts that have gone to that.

And then also the physician education again has been talked about very often. I can even tell you in my own practice of prenatal genetics, I still get referrals at 30 weeks for a woman that's 35 and older, that I know has been to that doctor since she was 10 weeks and just for some reason, that referral never got through, and it may be that the referral came through and for some reason, there was a barrier for her to come to me, and I think all of those things we need to try to work on and put some recommendations toward.

And then, I think a new, relatively new or different topic that we haven't talked too much about is minority participation in clinical research, and I think it was even mentioned yesterday that for some reason, minorities aren't interested in clinical research and why is this and trying to identify those barriers.

However, I challenge people that make those statements. At Howard recently, we are still participating in a study about hemochromatosis and iron overload called the HEIRS Study, and each of the clinical sites that were involved in this study were given the charge of recruiting 20,000 participants, and Howard

was able to do that along with the other sites with extra efforts that -- I think we learned about a little bit as we got into it and hopefully we'll be able to publish on some of those and get those efforts out there so other people can use them, but I think one of the points that Dr. Reede had made yesterday was to make those efforts on the front end and not just try to catch up later on, and so I think that's just another topic that we can hopefully talk about.

DR. McCABE: Well, thank you.

I want to begin to wrap up the morning and begin to have people prepare for the afternoon. One other area, though, that I want to bring up because Francis encouraged us to think strongly about it but then couldn't be with us today, and Tim has sorted of hinted at it, but Francis was really talking about large population studies and recommendations regarding funding, which is certainly something that we can take up and even though Francis isn't here, I think we need to address that. Is that an area that we want to explore?

We've had a lot of ideas that have been thrown out. We will do the letter to the Secretary. But I want you to be thinking over lunch of what one or two things are we going to pick up as a priority for a product. We're going to have presentations from FDA and the other agencies that we talked about at the next meeting. That's really to bring us up to speed with where things are.

We had talked about possibly another product being a summary by staff of the review with the ex officios and the issues that they brought to bear and certainly summarizing these data in a way that we could all agree on would identify a host of issues and might be helpful to the Secretary and to the other agencies as well in order to understand that. So I would suggest that that might be something we could do and is really based on work that's already been done up to this point and sort of help us perhaps give us direction for the future.

But I want you to be thinking about what you would like to do over lunch, and then, having said that and given the discussion that we've had, how long do people think that it's going to take? Do you think that we can come to this decision? Do you think it's going to take us till 5:00 or do you think we can come to a decision by a time earlier than that? Any thoughts on these?

DR. LEONARD: Well, when I started my list of priorities, we got through my first two, but my fourth one is the large patient cohort because one of our mandates is research and that is going to be absolutely essential for moving forward the complex disease understanding is having access to that kind of thing. So I know we're not supposed to discuss this now, and I would be all in favor for ending early.

(Laughter.)

DR. McCABE: Okay. I'm seeing body language indicating.

So why don't we aim for wrapping up some time between 3:00 and 4:00, more likely between 3:00 and 3:30, but what this will mean is for people to focus on priorities because we need to leave here with one or at the most two priorities where we can have, in addition to the letter to the Secretary, if I hear no objection, in addition to a summary of how we've gotten to where we are, then really begin to focus on one or two priorities for the future.

Thank you. Have lunch. We will be back at 1:30.

(Whereupon, at 12:28 p.m., the meeting was recessed for lunch, to reconvene at 1:30 p.m.)

AFTERNOON SESSION

(1:40 p.m.)

DR. McCABE: Let's go ahead and get started. I got sidetracked when somebody asked me about organic chemistry and premedical education, and I lost track of the time because it's one of my current soap boxes.

Well, I hope everybody had a good lunch, and now we're going to really knuckle down and identify specific priorities. A couple that have come up that I've heard about, one is to look at what expertise the agencies would have in genetics. For some, that may be obvious but for others may be less obvious. But what expertise do they need right now and anticipate they will need in the short run, and then what do they have on board, both at -- and then at what level, doctoral level, board-certified, and look at really where the human resources are in the various agencies, and it was suggested to me that this might help not only identify needs and where the needs were for future hires but also allow some sharing between the different agencies which would be good. So I just want to throw that one out as something that we should think about.

Another one was exploring a little bit more in terms of the large population studies but not just that there's a need for large population studies but exploring a little bit about what those needs are, and Alan, do you want to expound upon that a little bit?

DR. GUTTMACHER: I can maybe expound a bit about what the status of present thinking is, at least in several of the departmental agencies, about this because this is in fact a topic that in recent weeks particularly a number of them have been talking together about, particularly CDC, HRSA, and NIH, talking about what is the proper sort of need and scientific approach to a large population-based study that would really try to look at sort of genotype/phenotype correlations across a normal population.

As many of you will know, there have been a number of attempts to look using genotype/phenotype correlation around specific illnesses and those kinds of things which have been quite helpful in lots of ways, but this would be the idea and not a novel idea, an idea that's already been used in such efforts as BioBank in England, Estonia's launching something like this, Decode in Iceland.

So there have been a number of sort of national studies to do this, but for multiple reasons, including not just underrepresentation but the complete lack in some of those of some of the population groups in the U.S., especially if we're going to worry about issues of access and those kinds of things, so we really need to have knowledge about.

So there's some real scientific reasons why it might be worth thinking about having such a study done in the U.S. There are in fact in the U.S. a few efforts towards this. The Marshfield Clinic and others in the U.S. have started talking about doing studies like this. The rough back of the envelope kind of thinking, though, is you probably would need some place on the order of 500,000 to a million people enrolled in a such a study and following them over a decade or more to really get meaningful results.

So there have been some early thoughts about how this might be done. There's been, for instance, some sort of unofficial consideration about perhaps asking the IOM to help out with sort of an expedited review of this kind of thing to look at some of the options, to give some opinions about what exactly would be scientifically particularly useful in terms of approaching this.

I think it would be particularly helpful as this moves forward for this Committee to be thinking about, is that, if the science of this can be figured out and it's a somewhat daunting task, and I think it needs expertise of a lot of the agencies, the IOM and those kinds of groups might be helpful in this, I think, again looking as I've heard people correctly around this table thinking about what is it in particular this group can contribute that others might not be doing, would be advice to the Secretary if we can inform you a little bit more about some of the scientific bases, about the real usefulness of this kind of thing

because this is not a study that, even if the scientific utility looks great, that one could ever launch lightly.

It will cost hundreds of millions of dollars to do and in fact would require not just funding beyond the NIH but beyond the Department of HHS probably, would have to really be a kind of federal and perhaps a very good opportunity as well to have some public/private collaborations kind of thing, and would clearly need to be something that the Secretary understood well and felt was an important kind of item I think to move forward in the current budgetary climate.

So I think this Committee being informed somewhat more about somewhat of what the science might look like, what the need might look like, to consider that and to see whether, through the various expertises and eyes that this group has, to really consider that and see how important it seems would be very useful.

DR. McCABE: So the argument then would be to assess where we were, to work with Sarah and her staff to bring together a report to us, working with you and you mentioned CDC and HRSA, to look at the status, but then where it needed to go and what was required to really move it beyond where it is right now, just to put something concrete.

DR. GUTTMACHER: Yes.

DR. McCABE: And bring that back as a report to the next meeting. So we're not asking for a half a day of meeting to gather data but we could really do those among the ex officios working with staff to bring a report and then present the report, and then we would decide at that point whether it was something that we wanted to take to the Secretary.

DR. LEONARD: Could we get the report before the meeting, so that we have an opportunity to read it? Is that asking too much? It's just that it will make the discussions at the meeting more fruitful if we have an opportunity to look at that report.

DR. McCABE: Do you think that's doable, Alan?

DR. GUTTMACHER: As long as you don't want it two months before the meeting, yes, sure, I think that's doable.

DR. LEONARD: No, no, but I mean a week by email or whatever.

DR. McCABE: Yes, a week or two before.

DR. GUTTMACHER: I can do that. There's not much point in giving you a report the day you walk in. I agree.

DR. LEONARD: And I would just comment that regardless of how expensive you think this is, the Human Genome Project sequence was thought to be outrageously expensive and we did it, and if that is to be brought to the fruition of all the health care advantages which are one of the major reasons that that was done, then I think that this is one of the best ways to spend our money.

DR. McCABE: Yes, and this is something that I've been interested in because of this issue. How do you translate the fruits of the genome into improved health care, and without an evidence base, we're not going to be able to do that, and this is the way you're going to get the evidence base.

DR. LEONARD: Right, because you need such huge cohorts.

DR. McCABE: Well, we've been talking about common diseases and rare diseases, but in fact they're a continuum, and if you begin to think about the whole genome genotypes for common diseases, the groups

that are going to have the identical whole genome genotype are going to be relatively small groups, and so you're going to have to treat them somewhat similar as you treat rare diseases and if you're going to get genotype/phenotype correlations.

So it's going to take large populations, and I think that the study that I like to refer to, and it's a loose analogy, but if we look at the Children's Oncology Group, 95 percent of kids are enrolled in studies, 85 percent in treatment studies, and if people had said back in the '60s or whenever the precursors to the COG got started, oh, this is an impossible task, we're never going to be able to invest enough money in this to really understand this, then we would not have begun what has become an iterative approach which really has led to the successes in children's cancer. So at some point, we're going to have to do it and the question is do we decide just to do it now or 50 years from now.

MR. BAKER: Yes, I think the important thing that I hear from Alan is, it's not a question of if, it's a question of when, and then given that feedback from this group, the question then becomes how and whether this process.

I think it's important. One of the things we've constantly commented on is this is clearly a national study. It's not a federal study. It's not an agency study. It's a national study, and there's some lessons to be learned from the BioBank experience as mentioned as well as from NHANES and some large population-based studies that we do need to bring to bear, and there's some other related activities that are going on that we need to assess pretty clearly and take input from and perhaps include in the report that we're talking about having.

DR. GUTTMACHER: That's a piece. I think part of the report that we ought to give you is some reference to these other things, the National Children's Study and the other kinds of things, pieces of which are analogous to this and none of which is exactly the same.

DR. McCABE: As well as to the extent possible what's going on in the private sector because we know that activities are going on in the private sector.

MR. MARGUS: The best thing I heard was the idea of having the IOM or someone like that look at it, too.

My impression from talking to a lot of scientists on this is that it's nowhere near, decide that it's when, it's still an if, and there are definitely different sides as far as whether it's better to have one huge group that's kind of standardized on how you're going to phenotype them early on and it's quite difficult to change your mind later or, with the same billions of dollars, can you design a lot of different studies that are still very large, have all the power you need? So I think there's still a lot of scientific debate.

My impression also from Francis, to be fair, is that I think it's an idea being floated right now. I don't know that it's been explored that much yet.

DR. McCABE: Well, even if we come to the conclusion that it was a recommendation to the Secretary that the Secretary consider sponsoring an IOM study, the IOM only does studies that are funded for the IOM. Frequently it's the government going to the IOM asking for a study. So even that would be a benefit.

DR. LEONARD: I forgot what I was going to say.

DR. McCABE: Alan, any follow-up?

DR. GUTTMACHER: No, only that I would very much agree with what Brad is saying. I think that we're clearly not in a position where we would present some study for this group to endorse or something and that is the reason to go to the IOM. We think we have some expertise in the federal agencies, but I

think purposefully, one of the reasons to involve a group like the IOM would be that in some ways they can be even more dispassionate than we. We like to think we're dispassionate about it, but I think in some ways they can be even more dispassionate about it in terms of really weighing various kinds of options because I think even if you said you were going to endorse this study, there are many different ways this study could be constituted. It could be one large study. It could be actually sort of harnessing the studies that are already out there and somehow trying to get them so that they're using more consistent phenotyping or those kinds of things. So there are a number of different approaches one could use scientifically even beyond the question of the actual worth of it.

DR. FELIX-AARON: My question is a sort of a clarifying question. In terms of the study, are we saying that the study is to sort of understand the interactions between sort of the genotype, the environment, and health care, and how these all interact to produce a particular phenotype, i.e., disease? Is that what the study is about?

DR. McCABE: I don't know that we're talking about a study or the study. I think we're talking about a type of study.

DR. FELIX-AARON: But the idea, is that what we're talking about?

DR. McCABE: It could have several different, but several it could -- I mean, it could encompass genotype/phenotype. It could encompass variability. So it's a variety of these things, but I think that would be one of the things to explore.

DR. LEONARD: Kay, are you asking about the IOM study?

DR. FELIX-AARON: No, no.

DR. LEONARD: The data, this cohort?

DR. FELIX-AARON: Right. I mean, the data cohort in terms of what are the issues, sort of getting a sense of what are the issues. So is health care included and sort of health services included in that mix?

DR. GUTTMACHER: It depends on how ambitious one wants to be and how much money you want to spend essentially. I think clearly, I mean, the overall kind of thing, the devil is clearly in the details here, is it's what are the interactions between genes and environment that create health and/or disease?

Now, how do you define the environment? Health care, social status. There are lots of factors that are part of that environment that interact with the genotype. The easy part of this study, if we ever did it, would be the genotype. Everyone focuses on that. That would be the easy part. Defining what we mean by the environment, figuring out how to measure it, I mean, if you just looked at what exposures are we talking about, are we talking about industrial exposures at work and how do you measure the -- diet? Diet when? All these kinds of things are incredibly difficult scientific questions to do.

MR. BAKER: Yes, to qualify, when I say not if but when, I definitely mean the interpretation, and the question you're raising, there's going to be a series. There's going to be layering of studies, particularly if you look at these complex interactions of genes, gene-environment interaction, gene-behavioral interactions, gene-phenotype outcomes in different populations, enormous studies.

So I don't think anybody's thinking in terms of one study. I think we're talking about a variety of studies staged over time, shared by groups of people, public and private, that build the knowledge based on opportunities, based on maturity of science and what are priorities.

For example, there are priority diseases that have risen to attention. So do you start with those diseases? This is the kind of debate that you need to have.

DR. TUCKSON: Well, actually, I've gotten, like others, thoroughly confused here, and I think that there are so many studies, as you say, that need to be done to elucidate this and that is so much a part of the research enterprise ultimately of NIH and others.

If we are talking about, which I hope what this is trying to get to in some of the questions that we just heard, having an organization like the IOM try to help give some guidance around focus and interrelationship between the development of new knowledge and its practical translation into the health care delivery system and being able to provide clinicians and others with the relevant information that helps them to take this new knowledge and apply it in cost-effective ways, I would hope that that is part of what we really are talking about here.

If we're really essentially talking about just more of the NIH budget for this work, that's a different kind of conversation, and so I think that what we might want try to do is get a little more explicit about what's here.

I'm trying to preempt or presage this idea of focus. I continue to think that if we're going to as a Committee do right by the American people, there is an inevitable march of science going forward. We could and should be advocacy-oriented towards having the basic science research budget being robust in this field. I think that is in America's interests, and if we need to make statements about that, that's terrific, but that's moving on its own course.

I think the issue becomes how do we give affordability and access to these services to the American people which has to do, I think, fundamentally with clinical decisionmaking. It has to be how do people and how do their health care teams understand how to use these things in the most appropriate way in real life real time.

I believe that what that means to me and I become, as I listen to the conversation, more and more focused on patient education. How do you teach them about direct-to-consumer advertising, marketing, expectations? How do you help patients to have a conversation with their physicians and their other health care team so they can make rational, intelligent decisions about the use of this new technology?

I think it has a lot to do with counseling and the robustness of the counseling enterprise and how that will work, and I think it has a lot to do with technology assessment and which tests get included and paid for and funded and why, and I think it has a lot to do with the health services research agenda for determining how new knowledge and new testing and new capability in this area fits with the total disease treatment for disease and which things does it replace, which things do we get rid of? How does it fit in together so that at the end of the day, you don't have all this stuff bottlenecked because nobody can afford it, can't get access to it, and we continue to do the old medicine while we do the new medicine. You can't do both. No one can afford it. It is impossible.

So what we need is some way of giving the American people the chance for the rational use of this new stuff within the existing context of how we treat the entire disease spectrum for individual diseases, and I just hope that if that's what we're talking about trying to get at, that's at least my plea on this.

MS. BERRY: I think I got it, based on listening to everyone's comments, but to follow up just on Kay's original question, to clarify for myself on the study and the research issue, is it to have somebody, IOM or someone, do a literature review of what work has already been done and an agency review as to what studies have been funded and are ongoing and then an analysis of what gaps there are in the knowledge and the research so that you can move forward? Is it for a goal of recommending some funding, research funding level, in the President's budget or is it a more focused study with the particular objective? I think I get the answer but I still have a feeling there may be different ideas that are being articulated.

DR. McCABE: Well, let me tell you what I was scribbling as notes to myself here listening to the

discussion. The question that I would bring back, the purpose of doing this report which would be a combination of literature and agency review because there's a lot of this stuff that has gotten hung up and hasn't made it to the literature yet, but it would be the question and these are just thoughts but to begin to focus it, but the question to the IOM, the questions to the IOM would be: is the science and the phenotypic or clinical evaluation ready yet for large population studies to look at the effects of genes and environment on disease pathogenesis? If not, then where are the gaps in knowledge, and if it is ready, then how would you move it forward?

My guess is that the answers to both of those are yes, that there are gaps and yet there are parts that are ready to move forward. But it would really begin to try and identify because what one does with IOM studies is identify questions for them to answer.

DR. GUTTMACHER: Yes, let me just clarify one thing. I certainly don't want to mislead the Committee as to this. We've already begun to have initial conversations with the IOM, and because the Committee does not meet again till the fall and one of the -- those of you who've been participants in any way in IOM studies before, you will know they have much power behind them. As with anything with power, there are also some downsides. One potential downside is the length of time it takes sometimes to get the results. So that, if the agencies agree on what they're asking from the IOM, I think our plan would be potentially moving ahead with some kind of meeting with the IOM even before this Committee meets again, so that to some degree, that die may be somewhat cast.

However, it would only be cast in that we would have asked the IOM to do a study and the IOM then decides exactly how it's going to do the study. Nobody tells the IOM. That's part of the reason for getting IOM studies, is they figure out how to do it, you don't tell them how to do it, but I suspect they would be very interested in hearing from the Committee's views of various aspects of this as well and that also, again, I think for the Committee to sort of, for the Secretary, with the acumen of the Committee, to look at what is the general parameters of this kind of study, and then for the Committee, like the agencies involved, to digest the report from the IOM once it comes forward and figure out what does it say and what are the recommendations based upon that study might be very helpful.

DR. McCABE: So what would be your suggestions to this Committee then, Alan?

DR. GUTTMACHER: I think in terms of we've already in some ways brought the Committee somewhat up to speed but only, I think, a bit, and the idea of having this sort of white paper or background report or whatever for the Committee to take a look at before the fall meeting, and we can certainly update you at that point on what the status is of any IOM study or non-study, whatever, at that point and things could change, but if there were a study beginning at that point, even if it was laid out what the study's going to look like, have that be part of the background information for the Committee as well, for the Committee to have whatever length discussion at that point that you think is most appropriate and decide for yourselves how best to proceed, but it might be at that point to sort of proceed with interest in the area and say, well, this is something we're going to get back to once this IOM report, for instance, comes out and decide how best to shepherd it at that point. That might be one way to go anyway.

DR. McCABE: So does the Committee have an interest in this then? Debra?

DR. LEONARD: I have a distinct interest in it. I just feel a need to sort of clarify for the Committee members because there seems to be confusion. I think with this large patient cohort, basically what we're creating is a tool that would facilitate research. It's not the research funding. It's creating the information and the DNAs from lots of people correlated with medical information that cold be used for research to understand the genetics of asthma or hypertension or complex genetic diseases and that doesn't exist. Every physician or every researcher who wants to study asthma has to create their own cohort of asthma patients with the DNAs and do the genotype/phenotype correlations, collect the medical information, and sometimes one investigator has problems collecting enough patients to reach statistical significance when they go to do these genotype/phenotype correlations.

So it's basically creating a resource that would be the basis for all kinds of research. Correct me f I'm wrong.

DR. GUTTMACHER: No, Debra has it absolutely correct. I mean, an analogy is actually the human genome sequence itself.

DR. LEONARD: Right.

DR. GUTTMACHER: It's like that. It's something that a group of academic investigators get together and kind of get the data together and then make it freely available to anyone to mine in various kinds of ways.

DR. LEONARD: Right. So it's really creating a resource on which all this other research can be done and that resource is hard to achieve when you're not working on a national level. So that's what we're sort of out there. Should this be something that NHGRI creates or works toward as a next step to mining the human genome and understanding its variations for health care, and the IOM study, I don't know the purpose of the IOM study. I'm not clear as to what that IOM study is going to do, and I'm really distressed by hearing that it's going to take a long time because --

DR. GUTTMACHER: It's not going to take as long as some. We've already had discussions about that. So it might be --

DR. LEONARD: How long is --

DR. GUTTMACHER: One would hope that from beginning to end might take eight to ten months.

DR. LEONARD: That's a long time.

DR. McCABE: Yes, but if you look at some of the hang-ups in the U.K. BioBank study, they got hung up for years because of concerns about the open-ended nature of the study and ethical issues that were raised because of that. So investing eight to 10 months up front is probably worthwhile. Having multiple groups looking at this, as Alan is proposing, we'd be one of those groups.

For example, what's been brought up in this Committee meeting in the past day and a half is the need for diversity to assure that all of the populations are represented, so that this isn't a resource for majority populations. It's something that we've already talked about and certainly I'm sure that it's being discussed, but we could reemphasize.

DR. FELIX-AARON: I'm interested in this idea, and I think I find it intellectually stimulating and very exciting.

What I'm having difficulty with is sort of understanding how that process that you described, which seems to be on a particular track already, intersects with the work of this Committee and that's what I would like some help with in terms of seeing and sort of getting my head around what the intersection is and how the work of this Committee feeds into this process which clearly seems on track.

Again, I ask this question because in the background material, in the preparatory material that we were given, it really asks the Committee to really look at where it can make unique contributions, areas that are naive areas, areas that aren't already getting attention, and so in thinking about that and taking those instructions very literally, I'm thinking of trying to understand what we're proposing.

DR. GUTTMACHER: Yes. So I understand exactly what you're saying. Again, I'm not on the Committee, but if I were on the Committee, I wouldn't want to be spending 90 percent of my time looking at this study, as important as it might be. I don't think that makes sense.

However, for the Committee to weigh this thing, thinking about it ahead of time but then once there's a little bit more of a report to reflect upon, how important is this really in terms of -- I mean, this is the Committee that's supposed to give the Secretary advice about genetics, health, and society. This study would touch on at least two if not all three of those areas, I think, and so for this group to be able to say to the Secretary, yes, this really is important, it's worth spending both time, effort, and federal monies in terms of accomplishing it, that would be helpful.

If, however, this group says this is a great idea but it's just the NIH and CDC and HRSA trying to give themselves a bigger profile, this really isn't going to move the ball forward. As our advice to you, the Secretary, it's a nice thing. If you can find the money, go ahead, but we're not going to make a big push for it. I think that kind of thing would be helpful.

DR. McCABE: The other advantage that we have bringing this to this Committee is that our work is done in the sunshine. So that, as opposed to this being done in back room Committees and moving forward through the IOM and eventually emerging, at least the process has the sunshine on it during the process, not waiting till the very end.

Are people interested? And really, it would be the various agencies involved. It's not just NHGRI but also HRSA and CDC working with Sarah and her staff to put together a white paper on this with the status of this and some of the issues that are relevant to this Committee for presentation at the next meeting. Again, I don't see this as a half-day presentation. I think we're talking about a 30-minute presentation with time for questions, maybe a 20-minute presentation with time for questions, and we would try and get the paper to you at least a week or two ahead of time so that people could review it before the meeting. Is that acceptable?

I heard a motion. Do I hear a second?

PARTICIPANT: Second.

DR. McCABE: All in favor, say aye.

(Chorus of ayes.)

DR. McCABE: Any opposed?

(No response.)

DR. McCABE: Abstain?

(No response.)

DR. McCABE: Another thing for you to do, Sarah.

And what other things, other priorities that people are identifying, recognizing that we already have a significant agenda? So we probably have room for one more item to go forward.

DR. FEETHAM: A theme that I keep hearing is the access, education and workforce, and as you pursue these, and to me, they're all quite interdependent to the work of the Committee and the discussions, just reminding you as I briefly presented to you yesterday and in the handout you received today, that these are all areas again that HRSA can inform the work of this Committee, and one of the things that we do have is a genetic counseling report that was done in 2000 that, if it would be useful to you, it's a Genetic Counselors Workforce Study that we could make available to you. It is available on the web, but we could, if you wanted those copies, we could get those to you to help facilitate some of the issues that you brought up earlier today in the discussion, and as I mentioned yesterday, we are in the midst of the

genetic workforce study which is looking at both specialists and primary care and that's going into its second year but again an update on that at some point in time may be useful.

One of the things that we've also talked about among our agencies since we are complementary and interdependent in the work we do is that CDC, NIH, and HRSA and AHRQ has co-funded some of our projects in the education of health professionals, and again if it would be of interest to this group at some point, we could do a brief panel, pulling together what that work was but also at this point in time what we see as what has come out of that and how it may have influenced the community, whether it be the academic community or the professional communities, in moving forward rather than grappling with starting all over again.

DR. McCABE: This sounds much more focused than education more broadly. I think when SACGT, when we talked about the need for education, which many people have commented on here, certainly there's a need for education, but when you try and get your arms around what that really means, it's very difficult to be focused. So this sounds more like a workforce analysis.

DR. FEETHAM: Well, it's a combination. Again, we are doing a workforce analysis as part of the Center for Health Workforce Analysis at HRSA, but also again, my premise was that we have funded in combination across our agencies for several years now various grants for the education of health professionals, and since that seems to be something that keeps coming up, if that information, of the sense of where we are with that, if that would be useful at some point, that we did a presentation on that, that's what I'm saying we could do.

DR. McCABE: Discussion?

DR. LEONARD: Of the four issues that we've identified so far, the nondiscrimination, the oversight of genetic tests, this large patient cohort, and then the education workforce process, we have sort of action items for everything. We have no action items for this health care education workforce issue, and this seems to be one of the major issues if we're going to integrate into health care genetic information and yet we don't have anything we're going to do about it or on it. So maybe we need to brainstorm and figure out what we need. I mean, with the oversight of genetic testing, it's just presentations at the next meeting. I mean, maybe what we need is information on what's currently being done and wholes of ways that we could make recommendations that would facilitate additional education funding or programs or whatever.

DR. McCABE: An analysis of where it's going and whether we would agree that it's headed in the right direction.

MS. WILLIS: Just I think as a part of that, I don't know if you can give me the benefit of more information, but on the grants that were focused towards physician education, I guess I just feel like education is one step but action is a whole other one and even if you get at physicians all this information, if they don't go on to use it and really seem to change their practices, then it's all kind of for naught and I was just wondering, are any of those grants, do they try to go back and see --

DR. FEETHAM: Well, evaluation is a component of any funding that I know our agency does and NIH and the expectation with CDC, also, and that's why I did say about this was, again what is the effect? When I said community, I meant the academic and the professional practice community. How tangible those data are, I can't say at this point in time, but that is an expectation, and again our funding has been interdisciplinary. It's not just physician-based.

MS. MASNY: I would be very much in favor of that as well. In fact, I was going to make a suggestion that with the summary report that we would have from the ex officios, to actually list some of the initiatives that have been taken by the different agencies, especially in regards to some of the priority areas, and I was going to mention the handout that you gave because it does list all of the grants and the

education efforts. So I think that this would be an excellent way for us to see what initiatives have been taken and then where some of the gaps are that we could then make recommendations about.

DR. McCABE: Other questions? Other comments?

DR. HOOK: Just a question to follow up on that. I know a number of state legislatures for recertification of physicians do require ongoing education. Has there been data to show the utility of that type of mandated annual or biannual sorts of requirements for HIV and domestic violence and things, and should that be something that we would consider as a possibility for genetic education as well?

DR. FEETHAM: That's a good challenge, and the discussions that we're doing right now is that NIH sees their role in the research about education and at this point in time, that we are not a research agency and so that kind of question wouldn't be necessarily ours to answer, other than indirectly.

DR. REEDE: Just to follow up on the same sorts of things in terms of understanding better, in addition to the training, just what's the state of the art in terms of where are the accrediting bodies or the certifying bodies or others? What are they doing in this area, and if that could be incorporated in the report and not just for physicians, but I think in terms of physicians, nurses, if you could cut across the health professions and health professions training.

DR. FEETHAM: As I recall, that was an activity that NCHPEG was doing and Joann was part of that Committee at NCHPEG which I don't know how current that review was, but that was an effort to try to get a handle on that certification that you're talking about.

DR. REEDE: If that could be included in the report, that would be useful.

DR. FEETHAM: Again, coming from that.

DR. LEONARD: It seems that the education is happening on a lot of fronts, and we've already asked Joann for documentation from the -- I don't know if it's private but professional sector, and maybe we need information from agencies and then I don't know if certifying boards like Joan is saying, but is there a way to have someone take responsibility, staff or whoever, for consolidating these things into something that could come to the Committee before the next meeting for review?

DR. McCABE: Well, we've asked Joan to do this for the various professional organizations. If we want to do it, would HRSA be willing to take the lead among the agencies to put together what is being done by the agencies? You clearly have a handle on that for yours, but discuss with the other agencies on our behalf what is being done but also where the gaps are?

DR. FEETHAM: We do have that to the extent that you as the former Committee asked for it and that was broader than education, but it may be a matter of updating that profile that was done a few years ago.

DR. McCABE: Right. This is much more focused. This is much more focused than that was.

DR. FEETHAM: Only focused on education but those preliminary data were part of that initial profile.

DR. GUTTMACHER: And as a number of people referred to, NCHPEG, the National Coalition for Health Professional Education in Genetics, really has much of this information already gathered, so it should be pretty easy to provide.

DR. McCABE: And Joe was here yesterday. I don't see him today. But could you maybe work with Joe McInerney and NCHPEG and bring that data back to us? So that would be the action item, that we would have a report on the status of education in genetics. We would have, as Debra points to, these four actions items. We need to see if there are other ones that people feel we have slighted that are more

important than these priorities and don't be shy. Feel free to speak up.

MR. BAKER: I'll point out, if you're not aware of it, the recent Institute of Medicine report on who will keep the public healthy, which calls for the training of public health professionals across various crosscutting areas, genomics being one of those, and through the development and the focus with professional education and the majority of NCHPEG's focus has been around health care professionals, and we slip in and out of that language here. Health professionals or health care professionals. Most of the discussion we're having is about health care professionals, and the public health professionals are also health professional disciplines and health education and epidemiology, environmental health and so forth, all need a dose of training and need for certain competencies that we've tried to develop.

One of the questions for the Committee is where do you draw that line as far as defining this educational challenge for developing competency among health professionals, and to what extent do you want to get over into the areas of other health professionals, other than health care professionals? It's an important area of consideration, but as you hear, as I'm reflecting the views from public health, particularly public health practice, people that work in health policy and work in disease prevention programs throughout states don't seem themselves as geneticists, don't identify with genetics information or expect to have competency, but they have a clear, very, very important role in current disease prevention, understanding environmental factors in obesity and diabetes and analyzing those diseases, preventing those diseases, and need to own this to the extent it affects their job. That's one of the challenges that I think CDC and HRSA and the agencies here particularly are sharing, and it's really a continuum from all of the training needed from very, very professional credentialed training of health professionals all the way down to an awareness level among other health professionals that are in much more of an applied field in the states.

DR. WINN-DEEN: I mean, I think one of the things that we're trying to identify really is where are the gaps, and so, I mean, I think you're right. I think we have to think first very small, who are the people who have immediate contact with the patients and really need to be educated because if they're not educated, nothing's going to happen, but then we need to keep moving this out in whatever concentric circles till we really have basically every member of the general public educated to the point where they can at least be an informed consumer, if not a well-informed person who has the right tools with which to do their particular job, whatever role it is they play in sort of the world health care effort.

But I think what we need as a Committee is we need your help to try and identify what's already happening and where the gaps are and then we can make our best judgment about what recommendations to make about either endorsing the things that are already happening or making very specific recommendations about which gaps are the most urgently in need of being addressed.

DR. LEONARD: So as part of the reports, not only what's being done but if you have a sense of the holes, the gaps, that's also useful for us to know.

DR. McCABE: So for all of these, we need to identify gaps as well as what's being done.

Over lunch, Sarah, Suzanne, and Cindy put together some items, and I think it's probably good to run through these, to just make sure that there are not any areas -- we've identified four areas. We now have actions on each of these areas, and they would come back to the Committee at or before the next meeting. But we should check. I was suggesting that we look at what they had prepared and be sure that there's not something really big that we've missed in this discussion.

MS. WILLIS: Dr. McCabe? I've just now looked at this sheet that I probably should have looked at before, and there's also efforts under public education. I think we also talked a lot about patient education, that kind of thing. I know I would be interested. I just wasn't sure if the rest of the Committee would be interested in also getting a summary of those efforts as well as efforts for health care professional education.

DR. REEDE: I just want to say, while we're waiting to set up, in terms of the comments that came forward from CDC and Tim, that for me, even though I'm from a medical school, I don't make big

distinctions in terms of that health workforce that we're talking about, and I'm thinking both in terms of if you're talking about the practitioner, the one-on-one with the patient, all the way through the public health practitioner, and I think that when I'm speaking about workforce and where the efforts that are being made in terms of education, it's across the board. I don't make a distinction.

DR. McCABE: Sarah, you want to read those to us?

MS. CARR: We definitely agreed that the Committee would be writing a letter to the Secretary thanking him for his support of the bill that emerged from the HELP Committee and recommending that concerted efforts be made to continue legislative efforts on the House side, and staff's going to prepare that letter and circulate it among the Committee and hopefully get it out as soon as we can.

There was a request for an update at the next meeting in October from FDA, CMS, and CDC on the CLIA regulation, and I guess involving also perhaps Dr. Sundwall representing the CLIAC because they're going to be discussing the potential augmentation of the CLIA regulation to address genetic testing specifically, and also to involve FTC because we want to hear what FDA and FTC is doing in terms of monitoring or having oversight over direct-to-consumer marketing of genetic tests and websites and so forth. So that will be an agency update at the next meeting.

And then, the workforce issues are to get briefed on what activities are ongoing, what the status of efforts are in that area, and I think we heard that HRSA would take the lead in coordinating a status report on agency efforts in this arena and that Joann Boughman from ASHG would graciously take the lead in pulling together what the professional societies, including NCHPEG specifically, are doing, and will you be able, Joann, to gather information on what's going on in certification as well or is that -- you can do that, too. Okay.

And then, we were talking about the NIH/CDC/HRSA plan, I guess, to carry out or develop a plan to carry out a very large population study to understand genotype/phenotype correlations, to bring to fruition the work of the Human Genome Project, to make it more relevant to clinical care and so forth, and that NIH would take the lead in developing a report to the Committee that might update us on where the IOM report stands and also describe this study in more detail, describe other kinds of population cohort studies that have been going on, and the possibility, I guess, of the Committee perhaps taking up the question of whether to recommend to the Secretary that funding for this which would be significant be considered.

I mean, that would be the goal as I understand it. This would be a report, a written report, to the Committee in time for them to be prepared to discuss it at the October meeting, and it might lead to a recommendation from this Committee to support the funding of this large study. That was what I gathered.

DR. GUTTMACHER: Yes, but I think that such support would be months away, at some future meeting. This would be sort of the baseline and then assuming that the model that goes forward is the IOM study, that one would not expect the Committee to endorse spending lots of money on something that hadn't even really been --

MS. CARR: Well, I wasn't sure I understood what question the IOM would be addressing. Are they going to tell you how to do it or are they going to tell you whether it should be done?

DR. GUTTMACHER: Some of both.

MS. CARR: Okay.

DR. GUTTMACHER: Some of both.

MS. CARR: So it won't be ready then in October for the Committee?

DR. GUTTMACHER: So we have sort of preliminary report as to the status of what's been done so far, thoughts about the future, but I think it'd be premature to ask the Committee to do anything substantive in terms of action.

MS. CARR: All right. That's great then. So maybe at the following meeting or the following meeting, you'd actually be taking it up.

DR. FEETHAM: Can you say more about the training and education, what the status of the agency expertise is?

MS. CARR: Oh, no. Actually, that probably ought to be a separate item, I think, in a way. Ed, do you see it as part of this? This was actually what you initially -- sorry.

DR. McCABE: Yes. Sorry. I had decided that the other things had risen to the top. This had to do with identifying what the needs are for genetics in the various agencies and what the expertise is in the various agencies and then opportunities for collaboration between agencies. If there's expertise residing in an agency that's needed in another, can they utilize that which exists?

It felt to me like these other things were probably more important to that, so I had let that slide down on my own list.

DR. FEETHAM: I mean, from a different perspective, I mean, it may be useful to this Committee if again we did a brief panel for you at the next meeting to just clarify how we see our roles. As I said, we're complementary and interdependent, and, I mean, that might be a more informational kind of thing that may be more useful than anything beyond that.

DR. McCABE: If you were willing to do that, I think it would be helpful to us, because we've heard, as we did the round robin, that what's going on in genetics but that was very brief and it would be helpful to us to know why everyone is sitting at this table and why each of these agencies were identified, and then I think it would be also helpful to identify if there are agencies that you all think should be here that aren't here, like we've already talked about FTC and yet they're not sitting here. Is that something that should be here?

DR. FELIX-AARON: My comment relates to the patient and population cohorts and, I mean, I would just want to offer Alan, Suzanne, and Tim, the expertise in health services research at the Agency for Healthcare Research and Quality, seeing that the endeavor is anchored by practice and what goes on in clinical care. I think the process could be informed by sort of an understanding of health services and what goes on in clinical care. That's one point.

The second point is that I don't see on this list some of the issues that have come up in terms of again the application of the technology, sort of trying to help and sort of bring clarity and support to some of the issues around the application around technology in current practice.

I mean, did this fall to the bottom or I just wanted to get a sense from the Committee because I think that is a theme that has come up in a number of different ways and said differently in the sense of direct consumer advertising, but also understanding this technology and how it relates to current practice and the realities of changing practice.

DR. McCABE: Anybody wish to comment?

MS. CARR: Is that the health care issues here?

DR. FELIX-AARON: Okay. I guess you didn't get those.

DR. McCABE: There were other issues that we had not identified. This was why I wanted to put up the list to be sure that there weren't topics that we had forgotten about, but they're lower on the list, probably

not to be taken up at the next time, unless we move something that's in that top four down and move one of these up. So that's the purpose of the current exercise.

DR. FELIX-AARON: I see. So this is to identify the next four topics for the next meeting?

DR. McCABE: Well, yes. The topics that we can engage at the next meeting. We have action items for the next meeting, but the real goal is to then identify from that are there actions that we can take in terms of recommendations, consultation to the Secretary, or to the other agencies? So the papers themselves, while they're a product, they're really the foundation upon which we would make decisions and move things forward.

DR. LEONARD: Could we relabel that population patient cohort studies? Because it's really development of a population patient cohort tool and studies -- I mean, that is a tool that will be used to do large cohort genotype/phenotype complex disease studies, and then the IOM study is evaluating process and utility of developing that tool.

MS. CARR: So this would not include genotype/phenotype studies. This would be how do you go about putting --

DR. LEONARD: Developing a database.

DR. McCABE: Conducting.

DR. LEONARD: This is a database tool.

MS. CARR: So how to enroll, get a million people in this study? How to go about literally recruiting a million people?

DR. LEONARD: And how do you store that DNA so that for all the whatever, however many studies are going to be done, there's enough DNA to do it once you have it and all that stuff.

DR. McCABE: Alan, is that acceptable?

DR. GUTTMACHER: I don't think we can design the study right now.

(Laughter.)

DR. GUTTMACHER: So I don't know. I mean, again the IOM will not have us tell them look at 1, 2 and 3. They're going to want to study the whole area to some degree. So again, this is an information item. For now, I think maybe we'll bring to you sort of the status of what's been done out there in BioBank, Children's Health Study, et cetera, et cetera, what other kinds of things are out there and presently launched, and perhaps by October, we'd have a pretty good idea of exactly what the agenda would be for the IOM.

DR. McCABE: But from my perspective, this will not be a passive, just present it to us, and we'll sit there and nod. The purpose of presenting it to us --

DR. GUTTMACHER: Right. But I'm not sure whether there would be much -- eventually for the Committee to take some real action on it, but I don't know whether there'll be much action to be taken in October as opposed to a future meeting.

DR. McCABE: Well, but if we feel that --

DR. LEONARD: Information about what they're thinking.

DR. McCABE: I mean, if we are looking at this critically, not just passively, but are looking at it

critically, there might be opportunity for input to the IOM.

DR. GUTTMACHER: Oh, absolutely.

DR. McCABE: Because I don't think we're really interested in a passive receipt of the information.

DR. GUTTMACHER: And eventually, you're going to have an even more active role, but sure.

DR. REEDE: Just so that for what I thought would be coming forward is beginning information sharing so that we can start to articulate the need for such a study and that's what we would be moving forward. So that, we really would not be looking at how you would design a study or what would be the data elements of a study but really being able to articulate the need and maybe within that being able to raise some of the types of concerns we might have in designing a study.

MR. BAKER: The term we were using in earlier discussion was feasibility of and considerations for this type of activity.

DR. McCABE: Sarah, could you move so we could move the page so we can look at the ones that have fallen off?

MS. CARR: Sure.

MR. MARGUS: Ed, and one of these points includes the information we're going to receive from the woman from the genetic counselor organization about --

DR. McCABE: Yes. Yes, that was part of the workforce and I think was with the stuff that Joann was doing.

Yes?

DR. BOUGHMAN: Maybe I can try and tell you what I have conceptualized that I'm going to be able to bring to you before the October meeting and see if I really am on target here.

Certainly, from my position and working with NCHPEG and with the College and with a variety of other organizations, I will be able to tap them for some of the activities that are going on now.

I'm not sure whether you got the handout or not, but yesterday in the National Association of Social Worker News, the lead article was "Genetics Guidelines Issued." The Board of the NASW approved the set of guidelines that are now a part of the genetics issues that are included in social work curriculum.

So in fact, those are kinds of things that we have in our scan of the environment we know about and think it would be useful for you all to have, and it will be the professional aspects, the accreditation aspects and so on that we are in contact with through our interactions with NHGRI, HRSA, CDC as well, and NCHPEG, but also the American Board of Medical Specialties and the genetic counselors themselves, and I think that list will at least give you some feel for where people are, and that's the kind of scan I was hopefully going to bring to you.

DR. McCABE: And Robin, do you want to see how what we asked of you would fit into all of this? Do you feel that it fits in?

MS. BENNETT: Yes.

DR. McCABE: Why don't you come to the microphone? This is Robin Bennett from NSGC.

MS. CARR: And I just typed this, so if you want to make any edits in that, go ahead.

MS. BENNETT: Since I spoke this morning, I've had a chance to make a few phone calls, and I don't think within a month's time we can come up with a dollar amount for you, but I think that we can come up with a design for a study that may have outside funding that within a year's time we would have. We could even do it on the back of an envelope in a month or really do a quality job for you, and I think that we could come up with a recommendation in a month's time.

DR. McCABE: So what Sarah has typed here, would that work? Receive information from NSGC on initiatives needed to increase training.

MS. BENNETT: Absolutely.

DR. McCABE: Increase the numbers of genetic counselors in training. I think that's really the --

MS. BENNETT: And quality of training.

DR. McCABE: Okay.

MS. BENNETT: We can certainly provide the educational pieces.

DR. McCABE: Okay. Good. Thank you.

MS. CARR: Is quality of training -- did somebody say quality?

DR. McCABE: Yes, quality was in there as well as numbers. Quantity and quality.

MS. BENNETT: In terms of improving existing training programs, are they at risk of not being funded in the future?

DR. McCABE: Thank you.

MS. CARR: Can I just ask? Did you say that you were going to come forward with a proposal for a study that would take a year, and you would be doing the study?

MS. BENNETT: I think that we could come forward with the initiative. From my understanding, this group doesn't have money to fund the study.

MS. CARR: No, we don't.

MS. BENNETT: So that's not where we're at. What I think we could come up with is the plan for when we would have that data available and what type of data.

MS. CARR: But you would be gathering the data?

MS. BENNETT: In collaboration with other groups.

MS. CARR: Are we comfortable with Number 4, the wording of that now? Alan, genetic basis of common and rare, is that what --

DR. REEDE: We need advice about the stages. I don't think that's what we're --

MS. CARR: I'm sorry. Tell me what it's supposed to be.

DR. REEDE: I don't think, from my take on it, that the plan was not for NIH to come to us to provide information for our advice on how to design the study because it's more advice from us or thoughts from us in terms of you mentioned feasibility. I think in terms of is there a need for this type of study and documenting, right, right, and what would be some of the concerns that we might have for this type of

study.

MS. CARR: And some of the concerns?

DR. REEDE: That might be some of the legal or ethical or other types of concerns that we might have, if a study were to be conducted.

DR. LEONARD: I think it's also just including us in this process of deciding to do that, so that if funding is needed or letters are needed, if we can be convinced that this is something that is useful to do, we may be able to facilitate or effect the process somehow.

DR. GUTTMACHER: I think that's right, and also, as Ed brought up, the ability to do this in an open kind of way where you can all invite comments in some ways that the IOM may not, for instance. So there are other interested individuals, organizations, or whatever, might be able to talk about some of these same kinds of issues in a way that might not happen in any other setting, I suspect.

DR. REEDE: I think the other part, I see this as a start. This is a start of a conversation that will be ongoing.

DR. LEONARD: Can I bring up another issue?

DR. McCABE: What I'd like to do is look at the ones that are at the bottom that keep being off the bottom of the page, just because I want to be sure that everybody's comfortable that we have gotten the four that are really important that we're going to focus on and that we haven't missed something.

MS. CARR: This came up in the presentation from all the agencies on their mission and roles related to genetic technologies.

DR. McCABE: Yes. I think I would postpone this. I would not put this as something for the next time.

DR. LEONARD: But there could be individual agency input if there are other agencies that have a voice that we need to hear that aren't at the table, that the agencies may be aware of, to have input to Ed and Sarah, that maybe they should be invited to be at the table.

DR. McCABE: And at some point, I think it would be good to do this, but I think this is more of a catalog, and I'd rather take more actions where there's need than go into a catalog at this point in time.

I think this is the big one. We've talked a lot about access and the question is how does this one fit in? The integration, insurance coverage and reimbursement, affordability, disparities in access. A number of people have mentioned this.

MS. BERRY: Well, many people here have mentioned the health disparities issue, and I know it's a priority for the Secretary and I know there are health disparities grants and there may be a specific program. In the back of my mind, I feel like I've read a bit about it and maybe the folks from HHS or some of the agencies would be able to tell us, and I'm thinking maybe our action item is to make sure that genetics is incorporated into the larger initiative that the Secretary or some office or division is already undertaking, so that we don't necessarily have to do a separate thing, but that we integrate our thoughts and recommendations and study into the larger piece because genetics is only a piece of the health disparities issue.

DR. McCABE: Yes, that can be done in one of two ways. We could ask someone from the Secretary's office to describe what efforts were underway or we could write a letter to the Secretary, saying that we understand that these efforts are underway, we want to make sure that genetics is included. How would you like to proceed?

MS. BERRY: Is anyone familiar with what –

DR. WINN-DEEN: Well, it's hard to know how to proceed unless you know what's already ongoing. So I mean, I think at least we need some baseline of information.

DR. McCABE: So Sarah, could we ask you to make inquiries and maybe have a presentation?

MS. CARR: And would this be in October?

DR. McCABE: Well, the question is, where does it fit in? Does anyone feel that this is any more important than things above it? Just knowing how much we can fit into an agenda, it might have to wait till the meeting after, so Sarah could begin to work with the Secretary's office and warn them that this question was coming so that they could be prepared.

Would that be okay, Sarah?

MS. CARR: Sure.

MS. WILLIS: I just have a comment. I'm not against pushing it to a second meeting, but I just feel like before we can worry about even workforce issues, we need to have someone to serve. If there's nobody there to serve because their insurance isn't paying for it, then it just seems like that needs to be a little bit higher up on your list, but that's my own personal bias. I don't have a problem with it being pushed.

DR. McCABE: No, I think it's a big issue. I guess I'm worried that it's too big of an issue to really accomplish, but certainly access is a major problem in health care in general and in genetics in particular.

I think the first is a letter that is staff time, and you all will see that letter. It's not something that will occupy time other than to report that it was sent at the next meeting.

The laboratory and test regulation, I would guess that that's going to take two or two and a half hours. That's not trivial to get through that and then we'd want some discussion.

The workforce issues, probably somewhat similar in time allotment, and we'd want discussion. So we're pretty much almost to two half days right there.

We'd already talked that Number 4 is really probably more like a 45-minute to an hour presentation and discussion.

So we would have time to begin to address what the issues were in access, though I don't know that we'd have time to do a report before then.

MS. WILLIS: I was just thinking maybe if we just pick one aspect within that to address, like if we decide to just do insurance coverage and reimbursement or if we just decided to focus on health disparities or other -- we appreciate they're all interlinked, but maybe a report on one aspect of it.

DR. McCABE: Why don't I suggest that we focus on health disparities, and since we know that that's an interest of both the Secretary and the Surgeon General, why don't we try and identify what's ongoing in the DHHS regarding health disparities and any of the other agencies who may have some insight into that and use that as a springboard for an action item in terms of how could we look at that in more depth and assist the Administration.

Sarah, do you think that'll work?

MS. CARR: Yes.

DR. McCABE: Anything else that's down at the bottom there?

DR. FELIX-AARON: Ed, I agree with you that this access issue is a huge issue, but I think getting a

handle on it in terms of, again for our purposes, in terms of looking at the diffusion of innovation and genetic testing and all this therapy being a diffusion of innovation and how that relates to access, I think it's an area which is pretty specific. I mean, there's work being done in that area, technology over time, and how it relates to this field and what we can learn and what we may be able to do or to guide the further development of that, I think, would be worthwhile.

DR. McCABE: Maybe you could help Sarah in looking at future directions.

DR. FELIX-AARON: Sure.

DR. COOKSEY: If I could just make a comment on the access issue, and Suzanne is representing HRSA, but I'm the principal investigator on the workforce study, and as we've looked at that, we've had strong messages from one of the funders to be sure and look at the public health aspects of genetics services which cover a lot of, I think, basic access issues, and we've been looking at it in a little bit more depth. We should have information that can be incorporated into the report on the types of patients seen, the sponsors, insurance coverage of patients who are seen by geneticists. We have bits and pieces of information.

Plus, as we've looked in more depth in a few of the metropolitan areas, sort of what the public hospitals are able to do and what other sources are available, I think we can touch on that in the context of genetics services. It's only touching on a big, big issue, but we can contribute that.

DR. McCABE: Thank you.

DR. REEDE: Just one thought in terms of the health disparities issues is, I think sometimes issues around health disparities, diversity, et cetera, can get marginalized when they are pulled out as separate entities, sort of like if we're going to spend the next two hours looking at diversity or looking at health disparities, and I think that is an important thing to do. At the same time, I think that there are components of this that can be seen in any of the other areas, so that when we are addressing workforce or when we are addressing education or when we are addressing putting together this large database, et cetera, they get revisited. So it's just a concern that we not marginalize them into one component but rather understand that they're an integral component of much of the topics that we have.

DR. McCABE: So themes or threads that should run throughout are gaps, disparities, and diversity.

MR. BAKER: I'd suggest adding to the discussion, following Cynthia's suggestion, your comments, Ed. In fact, the Secretary has identified a commitment to particular diseases that are in the current disease prevention this year in cardiovascular, diabetes, and asthma, and that certainly includes the consideration of differential impact on populations. Health disparities is amplified to that lens. So you may want to walk that through your consideration here following the theme of what's already important to the Secretary. How does this help clarify and target those efforts that he's already got underway?

DR. McCABE: Can we see the bottom of the list again, Sarah, please?

DR. HOOK: To what degree do Items 6 and 7 need to be done in the form of presentations or simply could be collated in, as you say, a catalog that could then be distributed to us?

DR. McCABE: First of all, 7, I don't think we were going to take up at this time. Maybe ask at some time in the future, but I'd rather focus on the other things at this time. So 7 is for future consideration.

DR. FEETHAM: But the first part of 7, the way it's written, belongs, I think, with Number 6, and then the education and training, et cetera.

DR. McCABE: But again, I don't know that that's a presentation. That could be a written presentation.

We will have to communicate that in writing. So Sarah and I will work on that, but again, from my

perspective, it will have a lower priority than some of the other issues above it and we may not get to it until the next interval. So we have 5.

MS. BERRY: Do you think we need 6 at all?

PARTICIPANT: No.

MS. BERRY: I think we should just take it out.

PARTICIPANT: I thought we were going to just try and get some written background.

MS. BERRY: That would qualify as a priority issue. If we're talking about priority issues and action items for us, I don't --

DR. McCABE: So those are additional items but not priorities. The first five are priorities, but the goal of those is then to determine what actions we can take beyond the products that will be presented next time and advice we can give. The products themselves will not be the ultimate product but it's really our advice that will stem from those. So we will have adequate time to discuss each -- well, really four items because Item Number 1 will be at that time informational only.

Everybody comfortable with this? We've created the agenda for the next time. We de facto made a decision.

Just to let everybody know, the decision that's been made, because it hasn't been discussed, I suggested that we work as a Committee of the whole rather than splitting up into work groups. By creating this agenda for the Committee as a whole, not identifying work groups, you have gone along with my recommendation and I just want to acknowledge that so that if anyone disagrees with that, that you can speak now, but I really felt that that was a lesson we should -- I don't know if I'd call it a mistake, but certainly it didn't work as well in SACGT as when we worked as a Committee of the whole.

DR. REEDE: Just one last comment about that, the issue, in terms of on health disparities initiative. If there's a way to incorporate in that not just racial/ethnic health disparities but it may be geographic. It may be rural, urban, other kinds of things, sort of thinking broadly in terms of health disparities.

DR. McCABE: Debra, did you have something?

DR. LEONARD: Can I understand the process by accepting these five issues? Will there be time at the end of each meeting to figure out which ones may move up to the top and are no longer action items that need our attention and time and adding ones at the bottom?

Because I think the gene patent issue, even though Francis Collins says it's the horse out of the barn and there's nothing that can be done, the impact that this is having on health care and the ability to translate patented genetic information into tests, diagnostic tests, which are the first fruits of the Human Genome Project, has a real impact on access and this translation and integration -- I mean, we can't fit this in now, but I don't want that to fall off the bottom of our list because it impacts access and integration tremendously.

DR. McCABE: Right, and PTO is under Commerce, and Arden, he couldn't be here this afternoon, but he offered that if we wanted to have an update from the PTO. He suggested that we get it from one of the examiners, that by getting it at too high a level, we might miss it. I think there is a value in the high level thing, and I would remind everyone that my recollection is that the patents that were let before the requirement for function, based on sequence and homologies, were to have been revisited in the fall of 2001. Obviously we as a country have been distracted with many other things.

DR. LEONARD: Right.

DR. McCABE: Important issues, but certainly not concerned with gene patents at the same level as many other things. But I agree with you, that this is something that we probably should revisit. We might want to revisit it, though, at several levels and not just at one level or another level.

DR. LEONARD: Well, I think also, having the PTO perspective is one perspective, but they're in the business of approving things and if they raise the utility bar, it doesn't matter because those things that have usefulness are exactly those things that need to be translated into health care most urgently, and I think it's more important to hear it from the health care perspective of genetic counselors having to send tests only to one source or laboratories who can no longer do tests for their patients because of patent issues, and I think more the impact on health care and the translation into health care.

DR. McCABE: Yes, and so the answer to your question is yes, our goal will be to have time to discuss the agenda for the next meeting and as well as what actions we will take on the items presented at that meeting.

Any other items of business to bring before the Committee?

(No response.)

DR. McCABE: I want to commend everybody for really focusing on task and helping us to develop this agenda, and I look forward to the next meeting. We've set out a lot of work for a lot of you here, but thank you for taking this on and we'll look forward to the fruits of your labors at the next meeting.

MS. MASNY: I just wanted to, on behalf of all of my fellow Committee members, just to thank you, Dr. McCabe, for being such a wonderful chair and getting us through this process initially.

DR. McCABE: Thank you. Thank you.

(Applause.)

DR. McCABE: Travel safely. We'll see you at the next meeting.

(Whereupon, at 3:00 p.m., the meeting was adjourned.)