SECRETARY'S ADVISORY COMMITTEE ON GENETIC TESTING

Thirteenth Meeting

Tuesday,
May 14, 2002

Constellation Ballroom E-F
Hyatt Regency
300 Light Street
Baltimore, Maryland

IN ATTENDANCE:

Chair

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PROCEEDINGS (9:07 a.m.)

DR. McCABE: Good morning, everyone. I want to welcome everyone to the 13th meeting of the Secretary's Advisory Committee on Genetic Testing. The public has been notified about this meeting through an announcement in the Federal Register on April 15th, a listserv mailing, and a posting on SACGT's Website. We appreciate the public's interest in our work and as is our custom, we have provided an opportunity to hear from members of the public during this meeting. If you would like to make a public comment but have not yet signed up, please do so at the meeting registration desk outside of this room.

We have a full agenda over the course of the next day and a half. We will be discussing a number of important issues and reviewing reports and recommendations emerging or reemerging from our work groups. These include the status of the Department's implementation of the committee's oversight recommendations, outcomes of the Genetics Education Conference that was held yesterday, reimbursement for genetic education and counseling, informed consent for genetic tests using clinical and public health practice, public comments on an information brochure for the general public, technical assistance for non-CLIA-certified laboratories, and a white paper on rare disease testing.

As you know, at our meeting in February, we thanked four of our original SACGT members -- Pat Barr, Kate Beardsley, Ann Boldt, and Barbara Koenig -- for their excellent service. We are delighted today to be able to welcome four new members to the committee: Dr. Robert Baumiller, Ms. Cynthia Berry, Mr. Vence Bonham, and Dr. Daniel Robinson. I would like to extend a warm welcome as well as congratulations to our new colleagues on your appointments to this committee. We had an opportunity last night during our orientation dinner to begin to get to know one another.

At this point, for the benefit of the public, I would like to invite the four of you, beginning with Dr. Baumiller, to say a few words about your professional interests, your backgrounds, and issues in genetic testing that are of particular interest to you.

DR. BAUMILLER: Thank you. Well, I am obviously -- well, not obviously a Jesuit priest, but I am. And I have been interested in genetics throughout my career. I'm a trained geneticist, am boarded in clinical cytogenetics and in medical genetics, was head of the Department of Genetics and the Department of Obstetrics and Gynecology at Georgetown University for many years, and in recent years have switched out to Cincinnati at Xavier University. I teach bioethics there and philosophy and medical genetics and biology, chair several IRBs, two IRBs, sit on two DSMBs and an IACUC, and things with various other initials which I think they put older people on as time goes on.

So I'm very interested in this committee itself and its work and the people who are gathered here. It's a great honor to be part of this committee, Ed, and I look forward to working with each of you.

DR. McCABE: Thank you very much, Bob.

MS. BERRY: Good morning. Just briefly, my background is probably fairly different from the rest, and I really look forward to the opportunity to learn from all of you because I don't have a medical background per se, although I'm married to an obstetrician-gynecologist, and so say no more, we're informed by our personal experiences, and he enlightens me every day with all sorts of medical information and knowledge and that probably is what got me started in the field of health policy.

I practiced law for several years in Tennessee and did a lot of health care policy work there as

well as trial work in the field of health care and medical malpractice defense. From there, I went to Capitol Hill and worked on health policy issues for a congressman, and from there moved on to the American Medical Association where health policy issues and medical/legal issues were at the forefront of my work, and now at Wexler & Walker Public Policy Associates, I lead that firm's health practice, and we pay attention to issues such as access to health care services, coverage policies, Medicare/Medicaid as well as in the private sector, and work a lot in the area of privacy, and that's a keen interest of mine because it's implicated to a large extent in what this committee will do and the issues that we face in terms of access to genetic services.

It seems to be one of the barriers to accessing those services when people fear that their information will get into the wrong hands and be used inappropriately. So that is an interest of mine, access and privacy in particular, and I really do look forward to working with everyone. This is an illustrious group, and I am honored as well to be part of it and look forward to our work today and in the future.

DR. McCABE: Thank you, Cindy. Dr. Robinson?

DR. ROBINSON: I'm reminded of Victor Borges' "Pardon my back and pardon my front." I'm a Distinguished Research Professor at Georgetown University, where I've been for 30 years, and I'm on the philosophy faculty at Oxford, to which I return tonight. My original preparation is in neuropsychology. My Ph.D. was earned in that field, and as I said last night, when I was young and had a functioning brain, I used to publish a fair amount on brain function and particularly visual biophysics. Over the years, my interests have extended to aspects of law and moral philosophy and philosophy of mind. I do teach philosophy of science at Oxford. I have published in philosophy of law and intellectual history.

I have a special interest in issues of consent, the grounding of consent, the presuppositions that we must make about those whose consent we take to be valid, and also certain issues in the matter of heredity and the uses and abuses of the concept. I reminded myself that I think the first book I did was a book I did back in 1970 for Oxford titled "Heredity and Achievement." I almost forgot I had done it. So these are quite ancient interests that are ripening.

I'm deeply honored to be associated with the members of this committee, some of whose writings have influenced my own thinking over a course of years, and if I may say so and this will be the only occasion I think I can safely speak for the committee, the staff that has provided us with information and has arranged these meetings are unmatched in my experience for competency and sheer goodwill, and since I will be leaving this evening, I just want to thank Sarah Carr and her associates for the best organization I've seen for any kind of meeting like this in what is too many years at this sort of work. Thank you very much.

DR. McCABE: Thank you, Dan. And Mr. Bonham?

MR. BONHAM: Good morning. I'm pleased to be here and to be part of this important committee. I'm an Associate Professor at Michigan State University, Department of Medicine, in the Health Services Research Division. I've been a practicing lawyer for 20 years and involved in health care for the last 14 years.

My particular interest is related to issues of looking at some of the social, legal, and ethical implications of genetics related particularly to underserved populations and communities that in the past have historically had abuse and misuse with regards to issues of genetics.

I am currently involved in a project that looks at issues of engaging communities of color, both African American and Latino communities, in development of genetic policies, and so I look

forward to my involvement on this committee and participating with everyone here.

DR. McCABE: Thank you, Vence, and thank you all very much. On behalf of my fellow veteran committee members, welcome, and we look forward to working with you in fulfilling our commitment to advise the Secretary on all aspects of the development and use of genetic tests to help ensure the safe and effective incorporation of genetic technologies into our health care and public health systems.

We also have a new ex officio member from the Centers for Medicare and Medicaid Services, Dr. Sean Tunis. Dr. Tunis takes Dr. Jeff Kang's place, who is leaving government service to become a Senior Vice President and Medical Director at Cigna Healthcare. Dr. Tunis is Director of the Coverage and Analysis Group in the Office of Clinical Standards and Quality at CMS. On behalf of the committee, let me extend a warm welcome to you, Dr. Tunis, and ask you, if you would, to say a few words about your background.

DR. TUNIS: My background is in general internal medicine and emergency medicine. I continue to practice in emergency medicine, and my policy and academic field has been health services research and technology assessment. So I came to the Medicare Program about two years ago to lead the Coverage Group and trying to develop more explicit evidence-based approaches to adopting new technologies into the Medicare Program.

Since Jeff left, I've actually taken in an acting capacity his position as the chief clinical officer for Medicare and will hold that position until Jeff's replacement is found. I'm quite interested obviously in genetic technologies as an area in which Medicare has some particular challenges related to their statutory framework and some of the limitations around paying for screening technologies, and I think that those are obviously some of the issues I'm sure Jeff has discussed with this committee.

DR. McCABE: Thank you very much, Sean.

We're just commenting here that we feel much safer having your practical

clinical expertise on this committee as well as your broader expertise.

We'll now turn to Sarah for a review of the rules of conduct. We reviewed these last evening, but I think it's important that we have them on the record today as well.

MS. CARR: Thank you.

Being a member of this committee makes you a special government employee, as you all learned last night and were reminded last night, and thereby subject to the rules of conduct that apply to government employees.

The rules and regulations are explained in a report called "Standards of Ethical Conduct for Employees of the Executive Branch" and you each received a copy of this document when you were appointed to the committee. So I'm going to only just review three of those rules today.

The first one prohibits government employees from lobbying Congress. So if you lobby in your professional capacity or as a private citizen, you must be sure to keep that activity separate from the activities associated with this committee.

The second rule relates to confidential information. It doesn't occur often in the work of this committee but whenever we share confidential matters with you, you may not share that information with anyone outside the committee and you must take care to protect it from being disclosed.

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

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

ask you to recuse yourself from the meeting and leave the room.

If you have any questions about any of these rules, you can contact me, Mary Nuss, whom you heard from last night, or our ethics counselor as well, Holly Jaffe.

Thank you.

DR. McCABE: Thank you, Sarah.

We're now going to begin with an update on the status of the implementation of SACGT's oversight recommendations.

At most of our recent past meetings, we have heard progress reports from FDA, CDC, and CMS on how the effort to enhance oversight of genetic tests is progressing in those agencies. A good part of our meetings have been devoted in particular to following FDA's efforts to develop a premarket review process for genetic tests, including genetic tests provided as laboratory services or so-called home-brew tests. At our meeting in February, we learned that program-level efforts at FDA were in abeyance awaiting guidance from the Commissioner's Office on whether the agency had statutory authority to regulate home-brew tests.

Today, we are pleased to be joined by Dr. Sherrie Hans, Senior Advisor to Dr. Eve Slater, Assistant Secretary for Health. One of Dr. Hans' responsibilities within Dr. Slater's office is to facilitate communication and activities between SACGT and the Assistant Secretary for Health and, where appropriate, the Department.

Dr. Hans is well qualified to play this role with SACGT. She holds a Ph.D. in biochemistry from the University of California, San Francisco, and has more than seven years of experience working on biomedical policy issues in Washington, D.C., with the Pew Charitable Trust and the National Academy of Sciences.

Before we turn to Dr. Hans for a report on the efforts of her office in this regard, I want to take a moment to report on the debriefing I had with the Assistant Secretary for Health in March. You'll recall that at our February meeting, we were honored to have had an opportunity to meet with Dr. Eve Slater, who just days earlier had been sworn in as the

Assistant Secretary for Health. As the transmitter of our reports and recommendations to the Secretary, Dr. Slater has a critical role in relation to the work of this committee.

Since her visit with us in February was necessarily quite brief, given her already full calendar, we arranged to meet at a later time for a fuller review of the committee and its work. That briefing took place in March, and I want to summarize for you the main points I covered with Dr. Slater.

I reviewed the committee's reports and recommendations to date and the current activities of the full committee and within our work groups. I reiterated the committee's views on the need for federal legislation to prevent genetic discrimination and the steps we have taken to explore concerns we heard from the public about adverse impacts from gene patents and licensing. I talked in some detail about our current study of the agency's programmatic efforts to increase knowledge of the clinical validity and utility of genetic tests thereby enhancing their appropriate use.

In addition, we discussed our collaboration and communication among the agencies and discussed that this is key to the success of these efforts as well as the value of an overarching departmental vision for genetics and genetic testing. I reviewed the major elements of our oversight recommendations, why we thought enhanced oversight was needed, the acceptance of the recommendations by the previous Secretary and the steady progress we have been seeing the agencies making in implementing the recommendations.

I explained that we viewed FDA's role as central to assuring the safe administration of new genetic tests and expressed concern about recent developments at FDA that have called into question the agency's authority to carry out its part of enhanced oversight. We had a productive discussion of these issues. I made it clear that we understood that an increased role for FDA in regulating home-brew tests could have significant programmatic and resource implications for the agency and reiterated our desire to see the agency take an innovative and flexible approach to increased oversight.

I believe Dr. Slater appreciates very well the multifaceted policy and public

health challenges posed by the expansion of genetic testing and is committed to doing all that she can to ensure that genetic tests are used safely and appropriately and that efforts are made to enhance public understanding of the benefits, risks and the limitations of genetic tests. Dr. Slater also reported to us that FDA is working diligently to carry out the review of its statutory authorities and that these questions should be resolved soon.

Let's now turn to Dr. Hans for an update on these and other matters. Sherrie?

DR. HANS: Thank you, Dr. McCabe. Thanks for the opportunity this morning to address the committee on behalf of the Assistant Secretary for Health, who couldn't be here today. She's out of the country. I'd like to thank the committee for their attention and interest to this very important topic and one which the Assistant Secretary is quite interested in.

The past work of the committee has provided key guideposts, I think, to the Department's work and we look forward to the insights that all of you will provide in the future. In particular, I know the Assistant Secretary will be quite interested to learn of your suggestions and the outcomes of yesterday's meeting to enhance the knowledge and understanding of genetics and genetic testing among health professionals.

I'd also like to just take a moment and thank both Dr. McCabe and Sarah Carr, both for their work with this committee as well as their efforts to inform the Assistant Secretary for Health of the work that you're doing and to keep us informed on a very regular basis about the progress of your work, and I second Dr. Robinson's comments that I think this committee is extraordinarily well run, and thank you very much, Sarah.

As all of you are well aware and as Dr. McCabe just mentioned, in January of 2001, the former Secretary of Health and Human Services, Donna Shalala, sent a letter to this committee responding to the initial recommendations that all of you made regarding the oversight of genetic testing, and I know that that response indicated a variety of activities that the Department would pursue to address the issues you raised, and I understand and as Dr. McCabe just mentioned that you've all been receiving regular updates from the various

agencies and offices who are responsible for carrying out those activities, and I see some of the ex officio members here today, and I'm sure that they will continue to keep you informed about the details of their work.

What I'd like to do for just a very brief moment is to tell you the role of the Office of the Assistant Secretary for Health in this and our plans over the next couple of months. Because both the Assistant Secretary for Health is new and, of course, with the change of Administration that occurred shortly after that response came to this committee, the plans that we have at this time are to, working with our colleagues in the Office of the Assistant Secretary for Planning and Evaluation, we're in the process right now of polling, if you will, or collecting information and insights from the various offices and agencies that have responsibility for moving the recommendations forward.

We hope in the next month or two to hold a meeting of that group to discuss their work to date, to discern what issues may be going quite well and where there are areas where additional efforts may be needed and also at that time to formulate a briefing for the Secretary to bring him up to date on the efforts of both the Department and this committee.

As you can well imagine, the decision of FDA regarding the statutory authority that they may have to regulate so-called home-brewed/home-brew tests is an integral piece of the Department's response to the recommendations made by this committee and therefore we made the decision that we wouldn't go forward with briefing the Secretary and laying out the plans of the Department in this area in a formal way until we knew the results of the FDA's legal review. Obviously I think the work of this committee as well as the work of the Department will vary depending on what the answer to that question is.

Therefore, once the FDA has completed that review and informed our office and this committee of that result, we will bring the ex officio members together to formulate a briefing for the Secretary and at that time will inform the committee of the results of that and any recommendations that we've put forward. I hope that at your next meeting, all of that will have been completed and Dr. McCabe and Sarah can update you on that.

Once again, thanks for the opportunity to be here today, and we do look forward to the work that this committee is going forward with.

DR. McCABE: Thank you very much, Dr. Hans.

Are there any questions for Dr. Hans at this time? Yes, Wylie?

DR. BURKE: Is the question that's been raised and under review by FDA a question of whether FDA has regulatory authority versus another agency or whether there is simply a lack of regulatory authority over home-brews?

DR. HANS: The question is whether FDA has regulatory authority over home-brewed/home-brew tests.

DR. McCABE: Yes, Francis?

DR. COLLINS: In the process of doing that review, are there in fact written records of the review that occurred previously, because this was a topic this committee engaged the FDA on two or three years ago, and we were given assurances that at that point, FDA felt there was no question that they did have such legal authority, and I assume that was based upon some review of the statutes and arriving at that conclusion.

Is that material that was generated during that discussion previously available to the current reviewers in the FDA Commissioner's Office and the General Counsel's Office?

DR. HANS: That isn't a question that we've asked the office but we could certainly ask and see if that material is available.

DR. McCABE: Dr. Charache?

DR. CHARACHE: I want to thank Dr. Hans for this summary. I would just like to be certain that we will be addressing the fact that some agency needs to have that authority and what this committee was looking at is the fact that if it were CLIA, they are not as skilled in doing what the FDA has done which is to build a template that makes it easy for laboratories to follow what you need, that then CLIA could further observe and that, furthermore, it can be several years before the CLIA review gets around and then there's too

many tests that have been out there that may not have been shown to be either analytically valid or clinically interpretable.

But I think the main reason that I'm concerned is because what we're seeing is an explosion of such tests not only in the genetic field, but in others in which now two major manufacturers of molecular test instruments are no longer putting their kits through the FDA, but are rather selling the ingredients to laboratories which are used very extensively as home-brew tests in which each laboratory is supposed to have validated it in their own laboratory, but no one has ever reviewed the entire process or how it fits into a given piece of equipment.

So this is an issue in which we must have somebody skilled in test evaluation reviewing these products because the end-around has become explosive.

DR. McCABE: Dr. Hans?

DR. HANS: Thank you for those comments, and I think that your concerns are well understood by our office. Certainly as you did, we see FDA's potential role as a central one and certainly depending on the response or the result of their legal review may require some rethinking both by I think this committee as well as the Department on how we could achieve the goals that you're talking about.

DR. McCABE: Thank you very much, and thank you for coming and spending time with us today.

We're pleased that you're involved in these issues and in the facilitation of communication about this committee and our work. Please convey our thanks to Dr. Slater for your report today and also let her know how delighted we were that she was able to extend a warm welcome yesterday to the Education Conference participants by video. We know how much time that took out of her busy schedule to do that and very much appreciate it.

Before we move to our next agenda item, I want to report on another presentation that I made in March. The HHS Advisory Committee on Minority Health, which is advisory to the HHS Office of Minority Health, the Secretary invited us and a representative of the National Committee on Vital and Health Statistics to share lessons we've learned about

the role, function, productivity and impact of federal advisory committees.

It was a valuable briefing for us because we made connections with some of the leading thinkers in minority health. These contacts will be very helpful as we address issues of health disparities and in reaching out to diverse communities on all of our issues.

We also were asked to participate in the Secretary's National Leadership Summit on Eliminating Racial and Ethnic Disparities in Health in July of this year. The details of our participation in this meeting are still being worked out.

At our first meeting in June of 1999, we identified the genetics education of health professionals as a critical component of the oversight of genetic tests because we viewed a well-trained professional as the first line of defense against the inappropriate use of genetic tests.

The Education Work Group, under Dr. Joann Boughman's inspired leadership and with the dedicated support from Dr. Susanne Haga of the SACGT staff, has been exploring the critical questions about how well health professionals are prepared and are being prepared to respond to the substantial changes the expansion of genetic testing will bring about. Yesterday's Education Conference was the culmination of more than a year of information gathering, analysis and consultation with experts in the field.

We will now turn to Dr. Boughman for a report on the outcomes of the conference. After a break, Dr. Boughman will return to outline a proposed report to the Secretary on this important issue. So we'll have a summary from yesterday, followed by the break, followed by the recommendations.

Joann, if you'd please proceed.

DR. BOUGHMAN: Thank you very much, Ed, and I'd like to thank everybody here who attended yesterday and participated. We'll hear a little bit later some of the commentary and feedback that we got, but by the end of the day, I was very pleased with the amount of energy and commitment that was shown by not only those who were presenters but the level of activity in the work groups and discussion groups as well.

As a matter of review, you remember that our work groups were established back in August of 2000. During the spring and into the summer of 2001, our work group worked on a literature review and some workforce analysis, and if you'll remember, we presented that in the spring, in February of 2001. Through that spring and summer, we had also talked about planning for an Education Summit in November, but in fact as you all will remember, the questions and the challenges were so broad to our Education Work Group, that we found ourselves kind of mired down by the weight and breadth of these questions.

So in fact, at that time, we turned our focus to developing a roundtable where we would bring a group of invitees in to in fact help us focus our work and our questions. That roundtable was held in November and then that group helped us prepare for yesterday's meeting.

The goals of that roundtable in November of 2001 are listed here: exploring the integration of genetics into current and future practice, discussion of major curricular needs of various disciplines, including the approach of core competencies that now is being used not only in the medical and other professional undergraduate curricula but also in the postgraduate training programs, and issues around faculty development as well because one of the areas of focus at this point is that we have genetics professionals and then we have the practicing health professionals and the transition of that basic knowledge into the general practice areas is going to require a great deal of faculty development, and we also asked that Education Roundtable to help us identify specific obstacles or gaps that needed to be filled.

In the outcomes of that Education Roundtable, and I have to tell you at least for myself, this was one of those times in my career where there were a couple of very clear aha moments, light bulbs went on. It was a very useful meeting where those who were out in the practice areas came to the table and really stated what might have been very obvious to all of us but stated it in such a way that it really did help us crystallize some of the concepts we were working toward.

One of those was the very clear statement that we simply have to get past

the denial, the denial that some professionals out there have that this really doesn't apply to me. You haven't told me what it is or really why it is. You haven't convinced me yet that this genetics stuff really is important. Therefore, we need to increase awareness and that needs to start now and it needs to be on a daily basis.

We need evidence-based practice, that practicing clinicians out there are now turning to the need and recognizing the need for rock-solid data to demonstrate why these things are important. We also have to work on the team approach, that there are challenges, and I'll slip this in. This came back again yesterday in at least one of the focus groups, that the genetics community itself has to be better about defining the role of geneticists and the role of practitioners. Ergo, the referral system needs to be improved and in a team approach kind of way, and that we need to develop more clinical tools, especially for practicing physicians who need it and need it now.

On the level of content and curricula, we focused in on the patterns that a variety of professions are now using to demonstrate knowledge, skills, and attitudes that in fact affect practice. This pattern works extremely well and geneticists feel comfortable about this because of our interest and, I think, our demonstration of focus on attitudes and the importance of the social, ethical, and legal implications from a very long time ago. I think the genetics community in fact has led this focus area.

We wanted to look some more at courses and the integration. There was an affirmation at that point of the NCHPEG core competencies that had been developed, but we also talked a little bit about the appropriate review and revision processes for curricula in a variety of areas. Model programs were discussed, including the Genetics in Primary Care efforts that have been going on, and once again there was a discussion of the need for resource material.

There were in fact several gaps in needs that were described, and one of those aha moments was when -- actually, it was Dr. Caryl Heaton, who said very simply, "I am a family practitioner. Tell me what behavior change you want to see. Tell me what I'm

supposed to tell my colleagues they should be doing. Define those behavior changes and define them clearly."

Then we talked some about discipline-based guidelines and how those might be developed in such a way that they would focus on the desired behavior changes that would demonstrate the interest and practice of genetics in a variety of disciplines. Once again, faculty development was clearly seen as a need. Expansion of the genetics workforce was seen as a need in this transition phase, and once again we came back to evidence-based practice and the support and need for more translational research in the support of model programs.

That led us to in fact the Genetic Testing and Public Policy Program that we had yesterday, the goals of this conference. One, in seeking these desired behavior changes, the work group wanted to find some common ground. One of the themes that has emerged is that there are a lot of practitioners who are actually doing some practice of genetics but let's demonstrate to them what they're already doing and in fact we have focused in on the family history, that health professionals in their own ways do engage in family history-taking.

We wanted to better define the challenge, this integration of genetics into practice, identify the various roles of providers, including the geneticists. This was also a chance to promote discussion and debate, and we did in fact have lively conversations. As has been a very strong theme of the SACGT, we felt it extremely important that this be an open meeting and a public forum for all comers to come and tell us what they wanted, and our final goal was to obtain recommendations.

Ed has already commented. We had wonderful welcoming remarks from Secretary Slater. We also had the Maryland Secretary of Health and Mental Hygiene, Dr. Georges Benjamin. We then had a plenary session that I think did a very nice job of setting the stage for the day, including Dr. Gene Rich and Dr. James Evans, who talked about family histories, and Dr. David Mallott, who started us off.

We then had a lively panel discussion moderated by Reed Tuckson that represented several different representatives from disciplines, and we'll come back to that for a

moment later. We had a lunch with a speaker and then the very important focus groups and reports.

The four focus groups ended up interdigitating and overlapping to a great degree. Rather than overlapping, I think that it does show that this is a flow of information, but I think we ended up being pretty pleased with the way we had at least tried to focus, if not divide, the issues.

Group I focused on content and curriculum at the pre-degree level, the undergraduate medical education, but in fact that wasn't quite enough. They also talked about prerequisites for medical school and other professional schools as well. The inclusion of genetics in training, examination and accreditation criteria, the residency or postgraduate level, was the focus of the second group.

We had a third group that focused on the development of genetic tools and resources, and there were a variety of issues that were brought forward here, and then the fourth group talked about the implementation of new developments and this group used some of the model programs and some of the issues that are already starting their integration and implementation as a starting-off point, and when we come back to our discussion after the break, I think that it would be useful to have some discussion also that would help us focus on some of the other activities going on. For example, the CDC conference that was just held on family history was brought up a couple of times during the day, but I think that would be a helpful point of integration.

Last evening, a few of us sat down and Priscilla Short was very helpful in this as well. She's a member of our Education Work Group, and Susanne Haga and I and tried to merge across the groups and then kind of divide the other way, so that we would pick out the themes, rather than your hearing another report group by group.

Our charges and the summary, I think, there were some -- and I have these in capital letters because I think that was the level of energy that was brought forth here. These folks told us that we needed to find better ways to truly integrate genetics. There are several

issues that the work groups thought should be institutionalized, and I'll go through each one of these in a moment. They talked about some implementations. There were several specific ideas of translational research or evidence-based studies that needed to be done and then there were some identification of some needs.

On the institutionalizing, there was a request for more information on looking at the undergraduate curricula and professional school requirements. If you remember, this was actually mentioned in the very first talk of the morning, that while physics and calculus are still required as entry points for medical school, genetics is no where mentioned as a requirement. So in fact, and this came from an associate dean of education of a medical school, not a geneticist, and so it may be that the time might be ripe for some suggestions along those lines, and we did have AAMC representation at the conference yesterday and they heard these comments among others.

There was a call for the development of a common language. In other words, we need to make sure that we have our terminology straight, and we need to be utilizing some of this language in a more useful way, and we can talk about that a little bit later, if we'd like.

The institutionalization of genetics by getting it into those examinations and accreditation processes. Whether we like it or not, in undergraduate and postgraduate training, while we don't specifically teach to the test, if in fact the material is not tested in the process, it simply doesn't stand out the same way that it does if there is a designated score, for example. So that, and I know that from having attended and given an update on the Education Work Group to the Association of Professors of Human and Medical Genetics, that that is one area that they are working on very hard.

The institutionalization of the use of family history I think was a very important point, that we do need to have more standardized tools and need to be talking across disciplines more clearly about what family history really means, and yesterday, we had a request in one group for genetics vital signs, this idea back to what is it you want me to do

today, now, and what I can accomplish in my day-to-day practice as a pediatrician or a family practitioner. It would be wonderful if I could take three-generation pedigrees on everybody. It would be wonderful if I could spend sufficient time counseling. That's not what I need in my busy practice. Tell me what the vital signs are that would allow me to know that I needed to refer or needed to do more work-up.

The phrase came of "vital signs" yesterday, but this in fact goes back to the red flag system being developed by the Genetics in Primary Care groups and the use of the acronym SCREEN that the GPC group is working on, I believe, in their processes.

So once again, I think we are focusing in now. We're converging on some of these really important to-do points, and once again, we needed to capitalize on faculty development.

In this process, and we can talk again a little bit later about this, but in the implementation piece, we talked about enhancing models that work, the GPC and NCHPEG. I think this should not go unnoticed. Yet it may be stating the obvious, but I think all too often we don't reiterate what model programs and what kinds of organizations really work, and it became clear from both the roundtable and this open meeting yesterday that people believe very strongly in the work that NCHPEG has done and they believe that the GPC, the Genetics in Primary Care, model programs that are being funded federally, are indeed working, that the kinds of activities that they are taking on and work groups that they are pulling together are absolutely critical in making progress.

They would like the SACGT to actually explore the use of another model, and in the SACGT itself, we have made reference over a few meetings to the U.S. Preventive Services and the Community Services Task Forces activities and some of the ways that they operate which are a little bit different from SACGT, but yesterday, we were challenged to look into that and see if we might not take on some of their strategies, and I am simply not familiar enough with those activities to comment on that at this point but we would need to look at that.

The SACGT open process that we have had was also not only approved of

but applauded. So once again, we found out something that is working and I think that's worth saying. They also asked us to in fact go even further with the development of collaborative models. They were pleased with the fact that we had brought in a variety of groups, but in actually sitting down, putting the nose to the grindstone that SACGT should challenge a variety of groups, including strong representation from industry, consumers and payers.

More data, more investigation is needed on the application of laboratory guidelines. This is one of those translation or implementation pieces, especially into the industry sector, not only to talk about the big steps in the process but to get the right people together, to challenge the right groups, to lay down the true guidelines that need to be utilized in these processes, and this goes beyond the education piece into some of the other groups, but they figured and asked that the guidelines be there because those guidelines themselves would be educational tools.

We obviously need more data on reimbursement patterns and codes, and I hope that maybe some of the people who were asking for some of those things might be here to hear the reports that we will see later. SACGT has obviously been appropriately interested in these, but in fact what we heard from the educational perspective yesterday was that the reimbursement process and the coding process in its overlay with the actions and expectations from a general practitioner versus referral into and use by geneticists is going to be an important link because the behavior changes that we are expecting are going to be in part driven by the process of how we label things, how we code things. So in fact that ties back together.

Obviously the translational research or outcomes research reemphasis that we are working in a world of evidence-based medicine, of evidence-based practice and that we need to challenge ourselves and the genetics community to prove the efficacy of the activities that we think are so important, and then we also heard the challenge to see the development of more standardized pedigree tools that could be used as teaching tools as well.

Most specifically, there were some needs identified. Funding needs for the above kinds of investigations that we just went through which I thought coming out of a one-

day conference and we've got pages and pages, both regular-size pages and large sheets, that give us some specific ideas that will obviously be fleshed out in our report. More funding for teaching and faculty development. For example, question. Might the GPC model be expanded for inclusion in other interfaces of professions and develop faculty in other areas as well and then utilize that in a domino-type effect?

There was also the need for the training of geneticists and collaborative teams. This came out in a couple of different ways, including the fact that there are training funds for specialists to get trained in genetics, but we don't see training funds for geneticists to get trained in other specialties. So we're seeing a more one-way flow of funding, and also the question was raised about genetic workforce in and of itself and the availability of training grants or individual training funds for Ph.D. geneticists but not routinely training grants or training funds that might be available for competition for genetic counselors. That in fact came up in two or three different points of view.

It was very interesting that if you cut across all of the discussion sections, and I will correct my error of yesterday afternoon, in the development and discussion from the educational perspective, just as in each one of the discussions of SACGT over the last couple of years, we would always say and there needs to be more training and education and there needs to be more training and education. It was I think, I guess, rewarding to see from those who were attending an education meeting that we turned right back to focus on the policy issues coming out of each one of the other work groups here at SACGT.

It was clear that the informed consent process which in and of itself can be a training tool not only for those who are doing the testing but in fact for the public as well, that underserved populations and the representation of trained geneticists and the availability of services to underserved populations, that training of a variety of practitioners is absolutely critical to the ultimate goal of serving underserved populations.

Access issues. Access will be better available if in fact all disciplines are appropriately trained and referral patterns are more clearly delineated, and in fact, there was a

suggestion of an orphan drug kind of model which in fact we have discussed here in that some of the statistics that we've talked about have been based on some of these other models. Once again, the collaborative model was brought forward, and the data collection aspect that I missed yesterday actually was pervasive from several of the work groups.

There were a few comments and criticisms that I'd like to bring to the group from the green sheets, and I apologize that that was one of, I'm sure, many slip-ups yesterday that I did not announce from the podium, that we wanted everybody to fill out their green sheets with some ideas, but we did get several in. I think there were several of us that were very gratified that we also had many people come up to us with positive comments that I think was reflective of the audience and the degree of participation. There was a great deal of energy there yesterday, a great deal of focus. Everybody participated in almost every room.

There were some concerns and issues that were as specific as they didn't really like the title of the conference. Most of the issues and concerns had to do with the expectations that people brought to the conference about issues that may have been or should have been talked about. We couldn't talk about everything in a one-day conference, but reimbursement issues, access issues, pharmacogenetics and nutrigenetics, and gene therapy were some of the topics that people thought might have been included. These comments were more specific, I think, and I think we take them recognizing that we had people who were interested in not only the educational issues but the application issues as well and tucked those in the back of our mind.

One thing that I would like to comment on was that we had – and I got a couple of comments verbally as well. There was some concern that, for example, on the panel, we did not have a genetic counselor representative on the panel and that genetic counselor representation from the podium seemed to be less than it might have been. That's true, and certainly as I explained to those who talked to me about it, it was certainly not done purposefully as an oversight or a slight. It had to do as much with the fact that the panel was already bursting at the seams and that we had to make some what we thought were difficult

choices.

I did point out that Dr. Pyeritz in his two recommendations as a physician geneticist, one of his two recommendations had to do with genetic counselors and genetic counseling. So I think that the idea or the concept was not slighted.

One of the other issues, and this is my own personal bias, we also don't want to think in any way that genetic counselors, at least as we are defining their role now, can fix everything. Genetic counselors are obviously critical in this process, especially as it relates to informed consent and pre- and post-testing counseling and so on. This is an issue we will continue to talk about, but in fact, our focus was more an attempt to infuse/integrate genetics in other practices.

So I want to make sure that we collect that information and just as this committee has been very concerned about industry and consumer representation in its deliberation, I think we need to make sure that we continue to include the role of genetic counselors very clearly.

Our Education Work Group assignments at this point are to prioritize all of the comments and recommendations, go back through all those many pieces of paper and pull it together so that we can write a full report with possibly some streamlined and smoothed-out recommendations to the committee.

So the Education Work Group, the glory may be over and now the work really begins, but once again, I'd like to thank everybody for their help yesterday and their activities.

DR. McCABE: Thank you very much, Joann, and to Susanne Haga for staffing that.

I think I was particularly gratified that the recommendations got very specific, and I was concerned that they could have remained general, education is being done poorly, we need to do better, education is a good idea, which I think we had been at at some earlier stages in our deliberations, but I felt that the recommendations did get very specific and

the kind of specifics that we can really transmit to the Secretary as specific action items and recommendations for the Department.

Discussion of this? Yes, Wylie?

DR. BURKE: Yes, I wanted to say also that it was a wonderful conference, and I really appreciated the opportunity to be there and to listen to the discussion.

What I think is really useful and I just want to make a comment that I think that I'd like to suggest something that I think ought to be part of our continuing deliberations, is that the value of the very specific recommendations that came out is that they give us an opportunity to think critically as I think we need to do about what actions really are HHS actions versus actions that need to be taken on the part of other sectors involved in this process and even I think we're going to have some opportunity to talk about where in HHS some of these actions might be taken.

So I was just making notes as you were going through your recommendations and just for the purpose of making some examples, when we talk about medical school or other health professional admission requirements, I don't think that's an action for HHS, but I think it's something that is of value for us to note and acknowledge that there are organizations like, for example, AAMC, that might play a role. Putting genetics on exams clearly also is a role for professional organizations, not for HHS, and then we can look at other potential actions where we might be able to craft some fairly specific action and funding recommendations to HHS.

I mean, clearly, it's not going to be a surprise to anyone that we think translational research is important, but I think it's of value for us to add our voices to other voices. I think it will be particularly interesting to think about some of the actions that may be more specific in terms of thinking about which agencies should be involved. So a faculty development, is that an HRSA/NIH collaboration? You know, what are the relative roles? There may be some discussion there and development of things like the pedigree tools in context of CDC's willingness to take some action.

So I think this is where our discussion will be quite useful.

DR. McCABE: I have Judy, Michele, and Dan.

DR. LEWIS: Thank you.

I also thought one of the things I heard yesterday was the tremendous amount of synergy not just with the work of the Education Group, but I heard lots of recommendations that I thought were directly pointed at the Access Group, for example, issues around reimbursement which were things that we're talking about. So I think that not only did the Education Group get its agenda moved forward yesterday, I think there were lots of suggestions in areas, like informed consent and like data collection, that are going to inform all of us.

So I thank you for that.

DR. McCABE: Michele?

DR. LLOYD-PURYEAR: I appreciate the focus on GPC. I mean, I think NIH and HRSA and AHRQ should feel proud about that, and I didn't hear this, but was there anything in the discussion that pointed to the need for perhaps a continuum of skills and not just somebody trained in thinking genetically but some more skills?

The idea of a mini-fellowship in genetics for primary care providers or a bridge position. Not everyone's going to be able nor want to maybe have the kinds of skills that we think people need in genetics, but some may, and the GPC project really doesn't supply that, but did the – because I didn't see it on your list.

Also, I have two questions that also weren't on your list and seemed to be absent from the conference. Were recommendations directed towards public health and allied health education? The focus on this is very much for physicians and nurses, and I wanted to know if people stepped out of that box and looked at other professions also.

DR. McCABE: Joann?

DR. BOUGHMAN: Well, the answer to both of those is at least in part yes. In one of the discussion groups in the afternoon, there was some discussion about the fact that

the GPC model as good as it is, we have to make sure that we don't believe that we are making everybody into mini-geneticists was the term that was used and that's where the discussion in that group turned to the better clarification of the role of the professional geneticist and the red flag and referral system so that we could understand that there is a difference in these kinds of activities.

That group did not go further to talk about who and what might exactly bridge that gap, but I think your articulation of this and the articulation that we heard of the two different pieces could lead the Education Work Group into a useful discussion of that and that might be, I believe, a kind of recommendation that we could come up with based on this public discussion, even if that specific recommendation did not come forward.

To your second question, at least in two of the work groups, it was very clear that allied health professions were included and there were people who were bringing those issues to the table and certainly taking them away from the conference.

There was a nutritionist in one work group who was very articulate and talked about the NCHPEG competencies in several areas, and I think Dr. McNeilly on the panel, our speech pathologist, also made a couple of very important points that would be applicable in all of the allied health professions, that in fact I might be, an allied health professional might be the first person who really hears the family articulate the need or the readiness to hear and deal with the long-term genetic issues. It might not be the referring physician who hears that, and so certainly from the onset of that readiness, the teachable moments issue that Dr. Tuckson brought forward, I think was one of those. We also had at least one or two occupational therapists who were at the conference as well and those points were made.

So while I think the discussion was focused based on the experience of the people in the room, it certainly was not missed and the idea that it needs to be generalized was clear.

DR. McCABE: I think also Dr. McNeilly's perspective as a speech and

language therapist in discussions in the hallway, we talked about the opportunity that hers is a discipline that suddenly because of newborn hearing screening is going to have the opportunity to really insert genetic education very quickly and very effectively as sort of a teachable moment for the profession, and we have to think about other professionals and as these opportunistic moments arise, how are we going to respond in a timely and nimble fashion, so that we can introduce genetics into their professional education.

Dan?

DR. ROBINSON: Joann, many thanks to you and Susanne for an extremely informing morning session and the afternoon focus groups.

I attended two of the focus groups, and I was very much taken by a remark that -- was it Dr. Korrs who co-chaired one of those groups?

DR. BOUGHMAN: Dr. Korf.

DR. ROBINSON: Korf. When he made a distinction between those who look at the dashboard and those who look under the hood, the practicing clinician is looking at the dashboard, and you might recall I offered a distinction between the Old World and the New World practicing physician.

The Old World physician knows something about family histories and, let's say, knows nothing about molecular biology and has a very, very good clinical record, and the New World physician could instruct the Old World physician seven days a week on the intricacies of modern genetics.

The question that I thought was the sort of question our committee should focus on for purposes of advising the Secretary, how would one's practice differ just in case the Old World physician awakened in the morning with all the intelligence and knowledge of the New World physician?

Now, it's one thing to say that there's no point setting out to create a bunch of mini-geneticists, but with all due respect for the specificity that came in the form of these issues you presented today that really isn't specific enough, and I would think that in our

deliberations, we would come up with quite specific curricula recommendations so that very, very busy medical school faculties and students who have all they can do to keep up with what it takes to stop Smith from dying that afternoon don't think that they now have to reserve entire blocks of time for the intricacies of medical genetics.

Just how much does a clinician have to know to read the dashboard correctly, knowing full well that the only time that physician is going to look under the hood is with some sort of mid-career change of interests, I should think.

So the curriculum here I think is everything, and I think the only people who can guide us here are people who really have lived in both of those worlds, sort of the Old World physician who got converted to modern medical genetics and actually can answer the question what I now can do clinically that I couldn't do three years ago, and I don't think we did hear from that sort of schizophrenic, usefully schizophrenic, perspective.

DR. McCABE: I think we did hear a term yesterday which I had not heard before but that was "guerrilla genetics" and that doesn't mean like the 900-pound gorilla which is the usual geneticist approach, it's important, so therefore learn it, but more the guerrilla approach or the stealth approach which is to sneak it in when people don't even realize it's genetics and educate without people realizing that they're being educated, and I think that's part of how we insert it in a stealth fashion so that people aren't turned off because they're tired of hearing how important genetics is.

Bob?

DR. BAUMILLER: I sit on a Governor's Task Force in the State of Ohio looking for advice on public health department activity in the new genetic era, and I reflect how different the reflection of that group has been than what I've been hearing so far here, and there, there's a lot more emphasis on the people, the individuals, learning more about genetics as they go through grade school and high school and so forth in order to cope with the new genetics at the top.

Just about everything we've said here has been for professionals, and I think

we have to also advocate strongly, as strongly as we can, that this be built into the curricula at the lowest levels so that the ability to consent, the ability to understand what is being practiced on them is available to the populous at large. We need to look and recognize that.

DR. McCABE: Thank you.

I have Pat, Wylie, Judy, and Cindy, and then Vence, and then I'm going to cut it off so that we can have our break.

Thank you.

DR. CHARACHE: I'm on the trail that Wylie opened in terms of how much of the really important issues that were raised apply to the Secretary or Assistant Secretary of Health and how much involve medical schools.

Certainly the curriculum is a medical school issue. The issue of teaching discipline-associated genetics is the job of the chiefs of service, primarily medicine and pediatrics, to get their various disciplines, cardiology, GI and what have you, to include genetic teaching, and these are so core to needs that I'm wondering if it's also in the purview of this committee to perhaps send out some feelers to perhaps selected chiefs of service and ask their views on their interest in pursuing these methods and their suggestions of how to see it move forward.

DR. McCABE: Thank you.

Wylie?

DR. BURKE: I just wanted to follow up a little bit on the curriculum content issue, and I love the analogy of what's under the hood versus what's on the dashboard.

I think when we think about that in terms of what might be an HHS mission, it connects to me with the concept of translational research and also a point that was made by Dr. Evans and Dr. Rich in our plenary discussion yesterday. The translational research part is that I think even experts aren't all together sure what's under the hood. That is, there's still a lot of data that is needed, and so I think there's acknowledgement of simply the need for funding to get work done that pulls together the data needed to create guidelines, and in terms of looking

at the dashboard and knowing what to do, I think, for example, Dr. Rich's demonstration of the Cholesterol Treatment Guidelines on the Palm Pilot is a very good model.

You have to have good data. Once you've got good data, which I think is an HHS agenda, there is then the task of turning that data into practice guidelines. There, I don't think we're talking about an exclusively-HHS agenda but one where HHS might play a very important role in promoting not only the gathering of data but the collation of data that would lead to these tools.

DR. McCABE: Judy?

DR. LEWIS: Dan, I really liked your model of the Old World physician and the New World physician, but I think the health care scene has become even more complicated because it's not just physicians who are providing primary health care anymore, and I keep reminding folks that the largest workforce out there is 2.6 million nurses, and we have several hundred thousand nurse practitioners who are providing primary care in an educational model that's very different than a medical school model, and it's a much more integrative model where the two years of didactic training and the two years of clinical training aren't divorced but they're concurrent, and so I'd encourage us to look at the additional models besides the model of the Old World physician and the New World physician but to look at clinicians and clinician education broadly defined, and I think there are lots of things we can learn from each other and lots of ways if we could look at translating our models that we could start that interdisciplinary worked during the preclinical training, so that we could have folks who then really understand collaboration because they've been taught it.

You can't expect people to go through school in a discipline-specific model and then come out and practice collaborative care because it just doesn't work that way. Unless you learn to play nicely together as kids, you grow up like cats and dogs.

DR. McCABE: Cindy?

MS. BERRY: Joann, I was wondering if there were any specific discussions that came up in the focus groups about challenges in rural areas, in rural America.

For example, if a physician or a nurse practitioner or other provider gets the training, has the knowledge, is able to do some initial screening but then what? Is there a then what problem where you don't have -- and it overlaps, of course, into access.

Then secondly, again with the HHS goal in mind, with regard to community health centers where there are specific recommendations, I heard it discussed a little bit in the summary, but I know that that is a priority for the Secretary to try to do more to deliver care through those entities and maybe there are specific challenges or recommendations that we could work on to help them as well.

DR. BOUGHMAN: Those two areas were at least mentioned, and I think one of them goes back to what Judy was just speaking about in the training of nurses, nurse practitioners and others, and that would fit in with some of the community health service models.

I know in one group, I learned a new term. I didn't realize we had not only rural areas but we still have what are defined as frontier areas and that was new for me. So in fact, that was mentioned as an issue and a gap. The proposed solution to that was not brought up, but we did hear that.

DR. McCABE: Vence?

MR. BONHAM: I, too, thought it was an excellent day.

In Michigan, we are completing a HRSA-funded report on issues of literacy and genetics, and one of the things that I think is going to be really important as we put together our report for the Secretary is that when you integrate some of the data and the information that's going on within the states that is funded by various agencies within the Department, that will be helpful to the report.

So issues that are going on in HRSA and CDC around education I think are going to be very important as we put together our final report for the Secretary.

DR. McCABE: Thank you.

With that comment, we will take a 15-minute break. We will resume at

10:45 sharp.

I will ask the committee to be thinking as we continue the discussion after the break, to really focus on recommendations that we can make to Joann to take to the work group so that we can get very specific and consider whether we will after consideration of the work group's document then move forward with a letter to the Secretary to transmit these recommendations and when we feel that will be appropriate.

Thank you.

(Recess.)

DR. McCABE: Well, let's resume our discussion.

Joann, if you'd like to lead off with some specific comments.

DR. BOUGHMAN: I just thought I would fill in a few bits and pieces of information and conversations that I had over the break and then I have a couple of generic or, I guess, questions, hopefully provocative questions that we need some feedback on and then a suggestion for the outline of our report.

First of all, we don't have the final head count, but we did have 330 registrants yesterday when we count the preregistrants and the walk-ins. So it was not only an active group, it was a fairly large group.

Secondly, I would like to follow up on a couple of comments that were made in the earlier discussion for those individuals who have not been involved in the Education Work Group discussions over time. In fact, you are absolutely correct that, especially yesterday, our focus was not on the public, the consumer, education and elementary through high school kinds of advocacy and educational efforts. That was by design simply because we knew we couldn't cover the entire waterfront and we decided to start where we felt most comfortable in coming up with some very specific action recommendations, but you are absolutely correct that we are concerned about it and need to be, continue to need to be concerned about it, and I'll come back to that in just a moment.

Another point that was made to me by a couple of different people that I

just missed in putting together my summary was model systems that are working per their design include GeneTests and GeneReviews and that there were references to those resources and tools in at least three, and I'm going to guess the other session as well, and in fact it was almost to the point of a given, but I think that does need to be said, that it is so successful, it has essentially become a given and that people use it regularly.

That does bring up one of the questions on resource tools and developments and I did not see it in the notes that I went through last night, but one of the questions that I will ask is the issue of clearinghouse and/or central point of information gathering that may or may not be a focus at this point in time but it is an issue, and the relationship and how you would see the committee interfacing with the issue of electronic medical record, and also in tools and resources, the family history tool and the interface with the CDC issues and also how that family history and the family history tool might impact on reimbursement issues, but Dr. Tunis and I have already met each other now separately about some of the initiatives that are going forward in his division and how we might bring the experiences and value of past discussions to enlighten some of the work that they are engaging on.

One of the other things I heard is that there are other especially state-based models out there that did not come to light yesterday, so we may have some more requests for information and nosing around to do as we write our report and then rewrite our section on gaps and needs as they relate to model programs and then try and have the Education Group come down with very specific recommendations as they relate to activities within HHS and the agencies and in fact recommendations or requests coming from the Secretary's Advisory Committee or challenges, if you will, to the professional communities, whether that's AAMC, the Professors Group on Genetics, or medical specialties, or the deans of schools of nursing or allied health sciences. There may be some pretty specific challenges that we could come up with.

DR. McCABE: I would think it would be good to point out models that already exist, especially as a number of these are federally funded through DHHS, so that I

think that would be good. We don't always have to find that there are problems. If there are already solutions that have been begun to be developed, we should acknowledge those.

The term "clearinghouse" was mentioned, and I know that HRSA was involved in a clearinghouse for documents at one time, and I'm not sure. When you were at Georgetown, were you involved in that, Bob? So that perhaps Michele and Bob Baumiller could talk about the previous experience with clearinghouse.

DR. LLOYD-PURYEAR: Well, the clearinghouse still exists, and I assume every federal agency must have something similar. Ours is called the National Maternal and Child Health Clearinghouse, and I don't think it's located as of today at Georgetown anymore. A new contract. It was competitive, but the clearinghouse still exists, and they send out alerts, too, for any grantee that has published anything or produced any tool using the Title V funds. So it becomes a clearinghouse of storage and distribution for products from MCH grantees, and it's quite useful.

We also pulled together, and it's just going up now, all the old genetics educational and new materials that were produced through our networks, the regional genetics networks, and that's gone up on the National Newborn Screening and Genetic Resource Center Website. It's a GEM database. GPC curriculum is also stored on that Website, but I think every agency probably has a clearinghouse.

Do you want to talk more about that?

DR. McCABE: Bob, do you want to comment on your experience with that?

DR. BAUMILLER: Well, we went through a long evolution, beginning about 20 years ago, I suppose, when there was a genetics clearinghouse proposed and was given to a contractor, and I and several other people were consultants to that clearinghouse. They attempted to collect materials and distribute materials for anyone and this was well before the Alliance was in anyone's mind or other agencies really outside of the orphan drug people were operative then, and after a couple of years, they decided to put that into a grant which I was

successful in competing for, and the body of the grant was both the collection and there was another organization contracted with that did the distribution of materials, and there was a little bit of working together that had to be contended with which got done well, and for perhaps four years, the concentration was genetics and the funding was out of genetics.

In fact, most of the funds to the Genetics Division of MCH seemed to go through there and into, but the rest of MCH then found itself needing materials taken care of, such as the Surgeon General's report we got to rewrite and print and distribute because we had the basis, and when President Reagan came in and cut back a number of the people who used to work for MCH producing materials got thinner and all the other divisions of MCH started asking us to do things, then at the same time, we had the conference in which the Alliance evolved, and other directly genetic organizations came along, and so the National Center for Education and Maternal and Child Health, which is what the new organization was called, became much more concentrated on research and maternal and child health and only tangentially genetics, and we felt that genetics belonged to genetics groups that were serving that, and that separated, and now it seems that under your aegis turned back towards getting a genetic clearinghouse going again which would be in step with, I would hope, Alliance and other related organizations. But it was an interesting circle.

DR. McCABE: Yes. It sounds like important lessons in terms of evolution, also the concept that from 20 years ago when it was print-based to the fact that a lot of it could be digital these days and even more accessible.

Francis?

DR. COLLINS: Yes, that's actually a point that I thought I would bring up because many of the materials we're talking about are likely now to be electronic which has a certain advantage in terms of their ease of distribution, and many of you know and are part of this loosely-knit organization called GROW, Genetics Resources on the Web, which Alan Guttmacher has been leading for the last couple of years, and which is an effort to try to identify those organizations and individuals that have information of this sort, and it's a wide

swath of materials and come up with a mechanism by which those can be easily identified in a search and where there can be some degree of editorial oversight to be sure that things that are pointed to in a GROW search are in fact those that have been validated.

GROW has recently been merged with NCHPEG which has already been brought up a couple of times this morning, the National Coalition for Health Professional Education in Genetics, and will likely serve a very useful function for NCHPEG in terms of trying to collect together validated materials that health care professionals can find easily and benefit from, and so perhaps in this next iteration of a clearinghouse notion, this might be a useful player in all of that.

If I could say one other thing, I think many of the things we're talking about with regard to health professional education are going to be most successful if there is truly full engagement of the organizations that represent those health professionals and they don't feel they're being lectured to by those egghead geneticists which they're all too used to have happen, and in that regard, I think the model that NCHPEG has followed of engaging all of those specialties by encouraging them to be full-fledged members of the organization and there are now over a hundred professional societies that are part of NCHPEG is a useful mechanism to follow, although NCHPEG is still growing and developing and trying to come up with ways to implement a very ambitious agenda, much of which overlaps with the recommendations of yesterday's meeting.

DR. McCABE: So Francis, what would be your specific recommendations then, because this is an area where the recommendations may move outside of Department of Health and Human Services and where there may be other activities going on? So how does that fit with the model for our recommending authority?

DR. COLLINS: Well, again, I guess as we talked a little bit about last night, it's really, I think, helpful for SACGT in this topic and others to try to identify which parts of a challenging problem are specifically within the mission of this group and specifically that means therefore which parts would involve recommendations to the Secretary as opposed

to other possible listeners and also to identify whether we have the expertise and whether there are other groups that are better positioned to take on a particular issue than SACGT is, and if you sort of go down that checklist, it would be interesting to do that with the recommendations coming out of this, I think there are some that will fit better than others, and it would be good collectively if we could try to go through that exercise. I'm not sure I can quite distil it at this moment in terms of what would come out of this particular list. That's something we should do as a group.

DR. McCABE: I think that's something you also already commented on this morning, Joann, that you would look at those issues and try and determine what was general and what was specific to the Department of Health and Human Services.

DR. BOUGHMAN: Well, I think that we could delineate some tasks that would clearly fall within the purview of one or more agencies. There may be other recommendations for development of programs, I mean for example, expansion of certain programs or continuation of certain programs would be very specific but that certainly doesn't preclude our suggesting that the agencies consider funding for or support for or requests for proposals in certain areas and for then other organizations as they see fit to apply for those kinds of funds.

In fact, our goal would be to get the task done by the most highly qualified people and the agencies themselves could determine whether that would be inside or via a request for proposals kind of mechanism.

DR. McCABE: One comment that came up yesterday and you made again today and I'll just reaffirm, and that is the importance of reimbursement. Certainly that is something that falls within the purview of HHS through CMS. We know that people will do what they are paid for and will tend not to do things that they are not paid for.

I think we need to be cautious not to say that we need business as usual and if it takes 20 to 30 minutes for a trained genetics professional to take a three-generation pedigree that we need every family medicine physician in the country doing that. I think also,

the discussion yesterday about developing a common pedigree platform or genogram that could be utilized and transferred easily, updated and maintained is again something that one could interface with the reimbursement because it would be a huge cost savings.

As someone who just had to fill out one of these for my own physician, two pages of lists, not terribly interactive, in fact not interactive at all, certainly we ought to be able with modern technology to come up with something that would have layers so that when a positive came up, one could delve into that and should be able to reduce that then to a pedigree format. So I think we need to look at some of the specifics that came up and begin to think creatively about how the resources of the Department could be utilized.

Sean?

DR. TUNIS: Yes, I just wanted to link in on the reimbursement issues and kind of lay a perspective on the table that I'm sure Jeff Kang has talked a lot about as well, which is that particularly over the last several years, CMS and many of the payers have been trying to move more concertedly towards an evidence-based or empirically-based reimbursement coverage and payment policy, and so while I think there needs to be a focused dialogue on current reimbursement practices, it's also important to I think focus on what I would call, I guess, the translational research agenda or to see to what extent this committee feels like they would like to delve more deeply into really identifying key priorities for the empirical research or the translational research that would demonstrate the utility or the effectiveness or the value of the services, whether it's 20 minutes of an office visit to take a family history or whether it's actually using a specific genetic testing technology, but I would think it's quite critical to sort of highlight the importance of setting a research agenda that then will become the foundation for reimbursement policy that is evidence-based and defensible, et cetera.

DR. McCABE: Yes, I think it is. We've talked about the U.S. Preventive Services Task Force that was discussed, has been discussed throughout the history of this committee. It was brought up again yesterday.

One of the things as we delved into it before was the relative lack of evidence-based. If we were really going to get very critical on the evidence-based for genetic practices, we'd find ourselves in deep trouble to justify many of these. So I think it is very important that we develop the infrastructure to collect that evidence base so that we don't continue lacking in that area.

Muin?

DR. KHOURY: I just wanted to elaborate on that last concept and, I guess, mention the Family History Workshop which we had a couple of weeks and two people from this audience, Wylie and Joann, were present there and so many members from the GPC community, and it's very interesting when you approach family history as sort of an outside tool to be used in public health or disease prevention in general and then you subject it to all the criteria for evidence base, it's interesting, you go through that process and it boils down to evaluating these tools, similar to what you evaluate with genetic tests in general, meaning the analytic validity of the tool, meaning how good is it in capturing the information on your relatives, the clinical validity, what it means with respect to disease risks, and the clinical utility, which is sort of where the rubber meets the road.

So what can you do with that family history information, and I think a big research agenda will have to be developed around developing family history as potential tool that then can be tested out in the real world, and one additional wrinkle on this family history discussion which I don't know if it came up yesterday because I wasn't in the meeting, is that for most people, the red flag for family history is raised when you have the type of history that sort of multiple family members affected with a disease condition or early onset of a certain condition and those tend to be a much lower frequency in the general population.

Maybe 1 to 5 percent of all people have the kind of family history that would allow us to raise a red flag to be basically referred for further work-up, but there is a substantial chunk of the population, maybe somewhere between 30 and 50 percent, that have family history of something, primarily the chronic diseases, major killers for public health, like

heart disease, cancer, diabetes and asthma and obesity and other things, where the red flag is not raised but because you have one, let's say, first-degree relative not with an early onset but later onset disease, and therefore it doesn't matter a full genetic work-up but still that person is at increased risk of that same disease somewhere between two-fold, three-fold, four- and five-fold, depending on what the disease is, and for which the practitioner or the preventive medicine doc or whatever is left with the sort of the lack of knowledge what to do with that information in terms of real practice.

Do you use it to enhance your diagnosis or your prediction and therefore focus on the prevention messages that we all need to focus on with respect to diet, aspirin, smoking cessation, et cetera? So anyway, there is a big research agenda in developing the tools for family history that can identify people into, let's say, high-risk or moderate-risk and then test them out with respect to their validity and utility, and we should all I think embrace that idea that whatever tools come up will have to be validated, and I don't know how much that discussion occurred yesterday, Joann.

DR. McCABE: A little bit, but not in great detail.

On another topic, Muin, I was going to ask you to talk about NCCLS, some of the things that they're doing. Joe Boone had mentioned them to me.

DR. KHOURY: Joe, do you want to make that comment?

DR. McCABE: Or Steve.

DR. KHOURY: Because I'm not sure what --

DR. McCABE: Or maybe Steve could do it, but following up on the family history thing, is that what you wanted to talk about, Wylie?

DR. BURKE: Yes. I just wanted to say that it seems to me there was another issue on the table, and I think it's been raised to some extent in the access report that we'll discuss later as well, and that is we all agree that informed consent is appropriate, and we, I think, have had a lot of consensus around this table that that sometimes means that genetic counseling services that aren't currently paid for should be paid for.

I'm not sure we're ready to make an across-the-board recommendation that every time a genetic test is ordered, there should be an extra billable CPT code for genetic counseling and really that speaks to the evidence base. In other words, what kind of counseling and by whom makes a difference in terms of better uses of tests and better test outcomes.

It seems to me we're asking for that kind of data, if I was understanding correctly.

DR. TUNIS: Yes, absolutely, and I think what's sort of already coming up in this dialogue, and maybe this would fit in the sort of Education Work Group, but I would think that in terms of recommendations to the Secretary, trying to highlight as specifically as possible what are the key research questions, maybe that's one, which is what type of counseling for what kind of service. You know, provide some additional benefit.

But I would think, given that the Secretary has a fair amount of research dollars at their disposal, some of which are at AHRQ and some of which are at NIH, a little bit at CMS and some at CDC, and then there's, of course, setting an agenda also could potentially be the framework within which private sector funding for research on the testing side would come into play, but it seems like this committee would have a fairly important role in focusing on that as an area to make recommendations.

DR. McCABE: Judy?

DR. LEWIS: I just wanted to speak to the discussion we're going to have tomorrow afternoon, which is the second piece the Access Work Group has been working on, which is the Guiding Principles for Reimbursement and that document, we had a draft document and we had a lot of discussion, and now we're back to looking for some real advice from this group as to how we should proceed and Jeff was real involved in some of those discussions.

So I'm hoping that maybe we can get your involvement in that, too, because I think it really is a critical issue in terms of looking at responsible reimbursement and in a way that's not going to price health insurance out of the market. So I think that it was a really

important issue, and I look forward to your guidance on it.

DR. McCABE: Pat?

DR. CHARACHE: I think the issue of when is counseling appropriate and for which tests and at which level ties in to the issues we've been struggling with with informed consent, and we come back to the same issue we have to keep thinking about which is, who should be making recommendations as to which tests requires counseling and informed consent and which ones do not, and in the case of counseling, which ones require preanalytical counseling as well as postanalytical counseling capacity.

DR. McCABE: Any other comments on these issues?

(No response.)

DR. McCABE: Does somebody wish to talk about NCCLS? Does that relate to education?

DR. GUTMAN: NCCLS has an initiative. Actually, I think they had the premier meeting within the last two or three weeks to look specifically at the mundane issue of clinical utility, and Tim O'Leary, who was alleged to have been in the room a moment ago but has conveniently left, is actually the chair, but Joe was involved.

Tim, would you like to talk about your work?

DR. McCABE: Tim, you want to come up to the table and comment?

DR. O'LEARY: I'm a little informal to be talking to such an august group, but really in response to, I think, the concern throughout this committee and really throughout the community, NCCLS made a decision to try to work together with representatives from a large number of organizations to create a document, a set of guidelines for determining or assessing the clinical utility of genetic tests, to take into account not just the laboratory side which was NCCLS' long-term strength but to really put it in the context of the skeptical observer as well, and we have a very large and, I think, diverse group of individuals that are contributing to the development of draft guidelines. Something should be available in draft form by November or so of this year.

The consensus process for NCCLS is I think a very important part of this entire thing. Every part of what NCCLS does is open, publicly visible. The documents will be available for consensus review evaluation and revision. So anybody that would like to get an oar in is certainly welcomed to, and if anybody is interested in getting on the mailing list for what's in process, I have a set of cards here. You can email me and I'll make sure that the information gets widely disseminated.

DR. McCABE: Tim, could you give your full name and affiliation just so we have it on the record?

DR. O'LEARY: I'm Timothy Joseph O'Leary. I'm at the Armed Forces Institute of Pathology. So I'm a federal employee.

DR. McCABE: And again, just so we have it for the record for those who may not be familiar with what NCCLS stands for, please?

DR. O'LEARY: Well, it formerly stood for the National Committee for Clinical Laboratory Standards but now NCCLS is a stand-alone name because it, first of all, developed an international constituency and has developed a much stronger interest in health services generally.

DR. McCABE: So it's the rock star formerly known as the National Center.

DR. O'LEARY: Something like that.

DR. McCABE: Thank you.

Yes, Victor?

DR. PENCHASZADEH: I just want to make sure what is exactly the product that you will be originating. These will be general guidelines or will you look at specific tests?

DR. O'LEARY: These will be guidelines in general but the plan of the subcommittee currently is to illustrate it with specific tests. It will probably build to a large degree on some of the guidelines that have already come out, such as that from cystic fibrosis testing that have been broadly thought through by a broad constituency, but that's a statement of

probability. There are many people involved in bringing this together and the final guidelines end up being a consensus process.

I should point out that the constituency of the NCCLS is three-fold. It is industry taken in both forms, the health care industry as it's grown in managed care times, as well as the medical device and pharmaceutical industry, government as represented by FDA, CMS and others, and finally the professional community, again to be interpreted broadly, and the subcommittee and the area committee that reviews its work both attempt to achieve broad representation of all three groups to make sure that the diversity of viewpoints is well considered in hammering out this consensus.

DR. McCABE: We would appreciate your keeping the Secretary's Advisory Committee on Genetic Testing updated as you proceed, and also, what is the education? How will you educate these constituencies about the availability of these guidelines as they come forward?

DR. O'LEARY: Well, NCCLS is a membership organization. The members, depending on the nature of their membership, either receive all or may elect to receive some of the guidelines that are brought forth. There's also a Website for NCCLS in which it's possible to order the documents. Finally, we would anticipate making their availability known by one or more publications, appropriate professional journals, and again probably spanning a range of what would be looked at by different constituencies.

DR. McCABE: I think it's important to point out that since you will set standards, the community will seek this education so that they can be in compliance with standard of care.

DR. O'LEARY: I'd like to clarify that we will be issuing these as guidelines, rather than standards per se, because as the promulgation of the standard, of course, has some very potent meaning and it was the consensus of the area committee and the board of directors that we were not ready to attempt to promulgate a formal standard at this point, although many organizations and individuals will tend to use the guidelines that way.

DR. McCABE: I'm familiar with that but frequently, there are disclaimers on guidelines suggesting that anyone who deviates from these should document the reason for deviation, so that while they are strictly stated as guidelines, they do begin to be practice guidance in the strongest sense of that word.

Victor, did you have another comment? Thank you very much for that discussion.

Other points that the committee wishes to make to Joann to help guide Joann and her committee? Mary?

MS. DAVIDSON: I just want to articulate what's probably very obvious for everyone else here, but I just want to put it on the table, that we're looking at the education of health professionals as a way to kind of break down what is really a huge problem, and the end goal of all this is that the family or the individual sitting in that office receives quality, informed, up-to-date, et cetera, care, and I'm picking up, and I know this is kind of a slow rebound, Bob, on your comment because as a member of both the Education Committee as well as SACGT, I understand that it's important to break this down and look at health professional education as a piece of this.

At the same time, it is such an artificial separation of functions and so divorced from the reality of how care is delivered and how care is received and how resources, in particular information resources, are organized, that I just want to bring us back to remembering that there's a larger context to this and that to ensure that quality care is the end product is going to take educating children so that by the time they are studying to be health professionals, genetics has some meaning to them, and it takes understanding public education, which I think is a topic that all of us dodge because it's very, very difficult.

I've been on so many committees on education and inevitably they all prioritize health professional education, and I think that's because we just don't know how to quantify or get our hands around what kinds of resources or training or education is really necessary both prior to someone knowing that they have a problem as well as at that point of

service, and so I just want to bring our attention back to this much more important or equally important larger context, and sometimes the fastest way between two points is not the shortest but it's the longer, and I think that we have to remember that there are at least two people, usually a family, and the health professional in that office and that really making that a collaboration and a partnership because that's the way health services are happening now, and so some of this discussion I think is by intent and I understand the intent, but it's not looking at that piece of it, and as a committee, we need to in some way put that into our recommendations.

DR. McCABE: Thank you.

Francis, and then Judy.

DR. COLLINS: Yes, I appreciate Bob and Mary both bringing our attention to this because I think it is a crucial part of the ultimate outcome that we want to achieve which is information being presented by a professional who understands the nuances of that information and being received by someone who's about to make some decision about their health and needs to be able to somehow integrate information which is going to be difficult to integrate. It will be statistical. It will be relative risks. It will not be yes, this is the right thing to do and no, that's the wrong one. It'll be much more subtle than that, and I don't think health professionals alone being educated to be prepared for that is going to quite do it, although I think one can make the argument that in the early phases of this, the way that many members of the public will get this information is by going to their health care professional, and if they're not ready, then we know we're in trouble. At the same time, if the public is better prepared on their own grounds by having gotten useful information and begun to incorporate that into their thinking, then we're that many steps ahead.

I think the challenge for educating the public is multiple. Perhaps the easiest part of it is to look at the sort of captive part of the public that's in an organized school setting at the present time, and we should be doing a lot more in that regard with K through 12 and community colleges and colleges to try to get curricula that include modern concepts of genetics that are going to be relevant to individual health on the table and all too frequently

they're not. It's tragic to see how badly these topics can be taught and continue to be.

A more difficult one is how do you reach the general part of the public that's not currently in an organized school setting, and my favorite sort of hypothetical question is if somebody gave you several million dollars to buy a 30-second spot in the middle of the Super Bowl to tell the public what they need to know about genetics, what would it say? What would the message be? Do we yet know?

I've been asking myself and other people that question for five or six years, and I think it's beginning to get more focused, but it's still a little hard to say exactly how you would craft that zippy message that would actually have a long-term consequence and wouldn't just go in one ear and out the other, because clearly the information that you want people to have is information that they retain.

I do think, though, the time is right to really wrestle with this again and part of this little soliloquy is by way of an advertisement that we are going to have a workshop on June 10th that NHGRI is organizing on education and public engagement. This is part of a series of about a dozen workshops that we're holding this year to try to formulate what we hope will be a very ambitious and challenging next plan for genomics which will include both basic science, clinical science, educational challenges, legal and social issues, the whole gamut of what the next phase of genome research will be, and we're hoping out of that discussion, which is supposed to focus on public education and only on health professional education as sort of a sideline, that we might get a little further along in trying to define how to accomplish a goal that we all agree is laudable but which is in fact quite challenging to see the path forward that's actually going to have the impact that we're looking for.

Some of the cynics would say we're not really going to achieve that until there is for most members of the public a direct consequence for their own health that they can perceive is right there now and not something in the future but right there now, and the optimists will say, well, that's already the case. We have family history. We should be using it. We're not. If you could start there and build on that, you'd be ready for the next phase, and

somewhere in the midst of all that, I hope there is an educational agenda that we could try to formulate a little more clearly than it has been so far.

DR. McCABE: Thank you.

Judy?

DR. LEWIS: I just wanted to piggyback on to what Mary had to say and talk about the whole model of how health care is delivered, and if we look at it as a hierarchical model where people come to health care professionals for information and we're the guardians of the information or whether or not we have a model where it's two folks coming together with a common goal and working as a partnership and that involves a lot of empowerment of consumers, but it also involves a lot of demystifying of the role of what we do as health care professionals and says that we're working together because actually the ultimate stakeholder is the patient. It's not necessarily the health care professionals.

So looking at a much more collaborative much less hierarchical role and a way of blending all of this stuff together I think makes eminent sense, but it's going to come at a price in terms of looking at some of the issues of status because knowledge is power and if only one person has the knowledge, then the power differential is huge, and if everyone has the knowledge, the power differential is pretty flat, and I think that that's just sort of a basic paradigm shift that we have to deal with.

DR. McCABE: Any other comments before we move on in the program?

(No response.)

DR. McCABE: If not, thank you very much, Joann, for a very informative and important conference yesterday. I think you have some great direction that you've gotten from the input of the participants in that. I think it's also important that we heard the importance of that broad participation and how the public really valued the opportunity to do that. So thank you very much.

Let's move on to our public comment period. So we would ask the individuals as they comment to perhaps come to the podium, if you would, and first we have

Dr. Jean Jenkins representing the Oncology Nursing Society. The other individuals who I have, while Dr. Jenkins is coming to the mike, are Katherine Schneider from NSGC and Dr. Cooksey about a HRSA/NIH project.

So Dr. Jenkins?

DR. JENKINS: Good morning. I represent the Oncology Nursing Society, or ONS, today. ONS is a national organization of more than 30,000 registered nurses and other health care professionals dedicated to excellence in oncology care. We've also submitted a written report that expands upon the information I'll submit today.

ONS has a Cancer Genetics Special Interest Group that currently has more than 90 members providing cancer genetic counseling across the country, and we thank Chairman McCabe and the committee for the opportunity to testify today.

ONS maintains that all nurses must have an understanding of the relationship between genetics and health to appropriately identify and address genetic concerns in their clients. To fulfill these roles, nurses need to improve their knowledge base in genetics. Genetics education for the nursing profession is two-fold through academic settings for nurses in training and via continuing education for all others.

With the dawn of genomic medicine, the increase in patient numbers seeking genetic testing alone will require new models for genetic health care delivery. In the future, the integration of genetic information into the management of cancer will become an expectation of those practicing in oncology. This will include cancer genetic testing. All members of the multidisciplinary health care team in oncology will be required to have a baseline knowledge of cancer genetics.

To that end, nurses have an opportunity to become and must become involved in designing these services. Not every nurse needs to be an expert in genetics but basic genetics content is essential for the provision of competent nursing practice in the 21st Century. Examples of nursing roles in the application of genetics to health care are delineated by the Oncology Nursing Society for the generalist nurse, the advanced practice nurse and the

advanced practice nurse in genetics.

The competency of nurses to integrate genetics into practice is a challenge with limited resources, limited time for direct patient care and constantly expanding scientific advances and only a beginning perspective of what this information will mean for future health care. Currently, the nurse's limited understanding of the risks and benefits of genetic technology, such as genetic testing, impacts the availability and utilization of such services. Nurses serve on the front line of health care and are instrumental in educating patients and assisting them to identify appropriate resources.

In our written testimony, we include numerous recommendations about steps that should be taken to advance nursing education and practice with regards to genetics. Some of these include the development of strategies to enhance the professional recognition of the need to know about genetics, the marketing of genetics as applicable to practice to promote decisions to integrate genetics into care, and we heard some of those recommendations yesterday regarding the use of consumers or customers of care to inform us about their potential benefit for them, and the development of a focused effort to prepare educators utilizing successful models, such as modular information, summer courses or develop program curriculum.

On behalf of the Oncology Nursing Society and our members who are involved in the provision of cancer genetic counseling, I thank the committee for the opportunity to provide commentary. We continue to hold in high regard the comprehensive and challenging work that you all have and that you have done to date, and we remain available to offer our support and expertise as you continue your work.

Thank you.

DR. McCABE: Thank you very much.

Any questions for Dr. Jenkins?

DR. BOUGHMAN: I would just like to thank Dr. Jenkins. She was one of our moderators yesterday and did a wonderful job of collecting and capturing a great deal of the

information and asked a couple of very provocative questions at exactly the right time to reinfuse the enthusiasm in that group.

DR. JENKINS: Thank you.

DR. McCABE: Francis?

DR. COLLINS: And I would like to add my congratulations to Dr. Jenkins for the many important things she has done in this arena and particularly as the primary author of the Core Competencies that were put together by NCHPEG that so many people are now using. She's done a wonderful service for the field.

DR. JENKINS: I'd like to thank my committee that worked very hard and that was the disciplines of both genetics experts as well as lay consumers, such as ONS at the time when we first began this process. So thank you.

DR. McCABE: Thank you very much.

Our next presentation will be Katherine Schneider, who is President of the National Society of Genetic Counselors, a group that I think has spoken at every meeting, this being the 13th, of the SACGT.

MS. SCHNEIDER: We just don't run out of things to say to you.

Good morning. It is my pleasure to speak on behalf of the National Society of Genetic Counselors which represents nearly 2,000 genetic counselors in an array of medical specialties and is the leading voice authority and advocate for the genetic counseling profession.

At this point, I'd like to raise three specific points regarding supportive genetics training, better support and reimbursement for genetics services, and strategies for educating the public about genetic testing.

Point 1. Advanced genetics training and education needs to be a priority. The NSGC encourages the SACGT to recommend increased funding for the training of genetics specialists.

Yesterday's forum was an important opportunity to discuss the integration

of genetics education into health care. The increased use of genetic information in making health care decisions makes it crucial for all health professionals to have a basic understanding of genetics. Efforts to expand genetics curriculum and medical school programs and in the public domain are both important and laudable.

NSGC pledges its commitment to increasing awareness of genetics among non-genetics specialists, especially primary care providers. As one example, the NSGC is working together with the American College of Medical Genetics to sponsor an interactive genetics module at the 2002 annual meeting of endocrinologists.

However, the incorporation of genetics into general health care requires an infrastructure of support; i.e., expert specialists in genetics. Genetics specialists are needed to assist in the review of complex personal and family histories and genetic test results and to suggest appropriate follow-up.

Ensuring quality genetics services, a key charge of this advisory committee, requires the presence of knowledgeable providers with in-depth training and expertise in genetics. Thus recommendations about ensuring quality assurance need to include strategies for increasing the number of genetics specialists by increasing support of advanced genetics training and education. This includes supporting geneticist fellowship programs in genetics, genetic counselor master's-level training programs and nurse-credentialing programs in genetics.

Concerns have already been raised about the potential shortage of genetics professionals. Such a shortage could make it more difficult for individuals to have access to genetics services and could lead to the inappropriate use or interpretation of genetic tests. If the full benefits of genetic information are to be realized, funding is needed to support the training and education of genetics specialists.

Point 2. Better coverage and reimbursement for genetic testing services is needed.

The NSGC is committed to obtaining more appropriate coverage of genetic

counseling and testing services. One of the basic ethical principles of medicine is to assure access to services regardless of one's ethnicity, geographic location or ability to pay. The majority of individuals with health insurance are covered by group health plans. Coverage of genetic counseling and testing services varies greatly across these different insurers. Some plans have in-network genetics services or are willing to cover out-of-network referrals while others routinely deny such services even to families recognized to be at high risk for genetic conditions.

There also needs to be greater recognition that genetic testing services must encompass informational visits with a knowledgeable provider. In some cases, health insurers will pay for the cost of molecular testing, yet will not cover the fees for genetic counseling and education. This issue seems to be somewhat unique to genetics providers. In other medical specialties, it is expected practice to reimburse provider visits to discuss laboratory results and appropriate follow-up. Denying coverage of provider visits places an unfair burden on genetic testing programs as well as the individuals and families being seen.

The NSGC strongly supports the adoption of billing codes specific to genetic counseling and education, the licensure of genetic counselors in all 50 states and making it standard practice for health insurers to cover costs of genetic counseling and testing when medically indicated. To help resolve these issues, we have assembled an Ad Hoc Task Force on Billing and Reimbursement whose main objective is to identify strategies that result in better coverage of genetic counseling and testing services.

Then the final point. Educating the public about genetic testing requires effective strategies.

The NSGC commends this advisory committee for its continuing commitment to educating the general public about genetics. This effort needs to consider a range of strategies, from incorporating genetics units into public school curriculum to supporting the development of user-friendly resources on genetics. These resources might include Web-based interactive programs targeted on specific disorders or a series of public

service announcements discussing the potential benefits of genetic technologies.

Rather than spending your valuable time creating specific tools, we encourage this committee to focus on developing the vision and plan for increasing the public's awareness of genetic testing. Once that vision and plan are formulated, we, the genetics community, with expertise in translating complex medical issues and access to patient populations who can help guide us, we'd be happy to provide our expertise toward developing these resources.

In closing, we commend the SACGT on its accomplishments to date and appreciate the opportunity to comment on your continuing activities. Thank you for your time.

DR. McCABE: Thank you.

You may wish to comment on the upcoming Endocrine Society meeting because I think it's a very creative approach to education that really came out of the president of the Endocrine Society recognizing that the Human Genome Project was upon us, that there are a lot of genetic issues in clinical endocrinology, and the desire to educate his constituency about this.

MS. SCHNEIDER: I'd be happy to provide some detail. The annual endocrinology meeting has typically about 7,000 attendees, and it was recognized that raising the genetics awareness among endocrinology providers was not happening, despite frequent lectures, and after some brainstorming, we came up with, or the American College of Medical Genetics came up with, a very interactive approach by assigning everyone who attends the meeting one of three genetic conditions, either hemochromatosis, multiple endocrine neoplasia Type II, or BRCA1, breast/ovarian cancer syndrome. They are given a pedigree and a brief case history and then told that blood was drawn for genetic testing, and if they want to learn their genetic test result, they can sign up for a 15-minute counseling session with a genetic counselor.

There will be a few floating geneticists around to provide back-up support, and this is, we thought, a very personalized way to let people experience what kinds of issues

and what kinds of discussions go on in a genetic counseling interaction. So we're very excited, speaking on behalf of NSGC, to be part of this effort.

DR. McCABE: Someone was telling me about a similar exercise at another meeting they attended and people really personalized these data and they had to constantly remind people that these were artificial scenarios.

MS. SCHNEIDER: It's not real.

DR. McCABE: They were not real, but it is a way of engaging people. They will be bar coded so that people will basically give informed consent. If they wish to participate, they can have their bar code interpreted, otherwise they won't really know which scenario they would fit into, but it's a very interesting exercise that has been developed for the meeting.

A couple of other things. One is, you're talking about the need for increased training and getting more people interested in genetics, more dollars for training.

Is there any concern that with the current financial issues going on in medical care that there will be a market for these individuals?

MS. SCHNEIDER: I think at the moment, the market for genetic counselors specifically in areas of adult care, oncology in particular, the need outpaces the number of providers that we have, and I see that this trend may very well continue.

I constantly am aware of the growing need for genetics and genetics providers given the Human Genome Project and the fact that we are only scratching the surface in terms of common conditions and the importance of genetic information. I am very much in support of efforts to increase the information about genetics into the general practitioner's hands and to transfer some of what we do into their hands, and I see that as a very critical time to take that on and yet I also see a great need to have that back-up support available in the way of genetic counselors and geneticists, and I think that both need to happen hand-in-hand.

DR. McCABE: Francis?

DR. COLLINS: I appreciate your comments very much. In light of the

comments made earlier this morning by Dr. Tunis and by the Access Work Group which we're coming to in a bit and one of their recommendations, I think there probably will be a lot of interest in what kind of evidence has been collected with regards to the benefits that genetic counseling offers.

As a medical geneticist myself, I think there's no question that those benefits are quite real, but people will want more than my opinion or yours. They will be looking for documented research-based evidence of the fact that this model does in fact provide concrete benefits.

Has NSGC attempted in the process of what you're doing as far as influencing reimbursement to collect results of such studies and packaged them in a form that a CMS agency, for instance, would be interested in looking at and might actually compel them to take this as a serious argument that's already evidence-based as opposed to saying we need more research?

MS. SCHNEIDER: Dr. Collins, your point is very well taken, and I feel as though I have been saying data, data, data for the last five years and people are finally starting to hear that, and with our Ad Hoc Task Force on Billing and Reimbursement, that is one of the, I think, main charges that they will bring to us and that we will create a mechanism of funding to support a project that can look at that in a very objective way because you're absolutely right. We have small studies but it's not sufficient enough to utilize.

DR. McCABE: Yes, Dan?

DR. ROBINSON: Thank you very much for that presentation.

I think Dr. McCabe asked about the market, and I think your reply was in terms of the need. Now, there's a great, great need for men and women of virtue, but I'm not quite sure there's much of a market for them.

(Laughter.)

DR. ROBINSON: Is there in fact a market for persons pursuing training in this area?

MS. SCHNEIDER: Yes.

DR. ROBINSON: Is it a profession, unlike philosophy?

MS. SCHNEIDER: There is a market, not like poly sci, either. There is a market for genetic counselors. In the most recent professional status survey that we undertake every two years, the graduate of genetic counselors took an average of less than four months to find their first job, and again I think the largest area of jobs is happening in oncology and other adult specialties.

DR. McCABE: Wylie, and then Victor.

DR. BURKE: I just want to follow up on getting outcome data for value of genetic counseling. I agree obviously with remarks of both you and Francis that people involved in the field I think feel the palpable value but we need more data.

In that spirit, I think we do actually have data that at least to some extent defines the problem. That is, I think we have data that define people's misunderstanding of test results, people's misinterpretation of test results and tests being ordered without counseling. What I think we don't have is the evidence that the genetic counseling model that exists is the solution, is the optimal solution, is the only solution, and so I think it's that, sort of addressing those questions that we need to point research to.

MS. SCHNEIDER: Absolutely, and we've also been in conversation with the American College of Medical Genetics and thinking about collaborative projects where we can look at this issue very carefully. In addition to billing and reimbursement, it also becomes important to look at the time that's involved in providing services with recognition that there may be a difference between what is efficient and what is ideal.

DR. McCABE: Victor?

DR. PENCHASZADEH: Yes, my reflections on this subject are that there is no question in my mind that part of the health care that any patient requires for any condition is according to the condition, of course. It's a discussion of genetic factors involved in the condition that is bringing that patient to the health professional.

Whether we can put a name to that and call it genetic counseling and education is fine. Whether that is, as Wylie was saying earlier, a billable service, apart from the health care encounter in which that part is discussed, of course, will require a lot of assessments of outcomes but also of the degree or the threshold beyond which you really create a CPT code or a billable service.

My concern has to do essentially with the training that the medical geneticists and genetic counselors, you know, the genetic counselors as we know them, the master's-level genetic counselors, are having until now. Until now, most clinical geneticists and genetic counselors in my mind are not equipped properly to deal with the major portion of what we are talking here which is essentially common diseases and genetic factors in common diseases.

I think we need a different model for the interaction between health professionals and define exactly what is the training that those health professionals require according to the needs for what genomics is bringing up to the health care, and I think that we haven't yet been able to define exactly what is it that health professionals need in order to provide those services because we really don't know what those services are in my mind as of today.

We hear a lot of things about predictive testing, about things that are coming to the market, but still, I think we are in a process of where a lot of these things are being defined, and I think that as far as education is concerned, we really have to make sure that the skills, that we define actually the skills that people need to have and not so much focus on a particular specialty or a particular profession.

MS. SCHNEIDER: Absolutely right. This is an emerging field and to stay one step ahead of it is quite challenging, even for those of us within the genetics profession.

One of the things we spoke about at our small group yesterday afternoon was the importance of changing curriculum to meet the need of the testing and accreditation and also the need of the patient populations, recognizing that that is a circular effort and that a

change in any of those arenas will effect change elsewhere, and so that's a point very well taken.

I'd just like to end by saying that one of the other points that was made yesterday afternoon was that we really did need to resolve this ourselves before the courts do it for us, before liability is what drives us, and I would hate to see that happen. Genetic services may be considered expensive and yet the quality assurance aspects, ensuring proper informed consent, the importance of that cannot be overstated.

Thank you.

DR. McCABE: Thank you.

Sean?

DR. TUNIS: Yes, just to kind of round out on the conversation about outcomes data and gathering information about the effectiveness of the counseling services, I would encourage you to take advantage, if you're interested, of having your ad hoc group contact CMS, the Coverage Group particularly, although you can go through me, and we're quite willing to meet informally with your group to talk about what sorts of information we're interested in looking at, what areas of services there seems to be promising information of where we might be able to focus coverage and reimbursement policy potentially in a more focused area where the evidence is best, as opposed to a broad coverage and reimbursement for all genetic counseling services.

But we do make available staff to talk informally with people even absent a specific request for a national coverage or reimbursement policy change to see what information you have and to see where you might want to take it to sort of meet the standards for payment.

DR. McCABE: Thank you very much.

With that, let's move on to our last speaker from the public this morning, Dr. Judith Cooksey from the Department of Epidemiology and Preventive Medicine at the University of Maryland School of Medicine, and Dr. Cooksey is going to provide us with an

update on an important HRSA/NIH-funded project looking at genetics services and the health workforce.

Dr. Cooksey?

DR. COOKSEY: Thanks, Dr. McCabe, and committee and visitors. I'm delighted to be here.

I'm presenting a brief report on the project, Assessing Genetic Services in the Health Workforce, which was funded beginning the end of September 2001 for three years by the Bureau of Health Professions and Maternal and Child Health Bureau at HRSA and the ELSI Program of the NIH Human Genome Institute.

Just by background, I'm a board-certified internist and hematologist and have directed a health workforce research center at the University of Illinois at Chicago for the last three and a half years. This project is run from the University of Maryland where I'm also a faculty member.

The specific goal of the project is to improve our collective understanding of genetic services, the factors affecting the demand for services, and the roles of health professionals in providing services. We expect that this study will provide a baseline for building longitudinal analyses and will bring new approaches, research methodology to the study of genetic services and health workforce research.

The collaborators with the University of Maryland are with four academic medical centers that have HRSA-funded health workforce projects, one at Albany at SUNY, the University of Illinois at Chicago, the University of California, San Francisco, and the University of Washington, and we are expecting to add an affiliation with the University of Texas, San Antonio, Health Science Center which was the fifth HRSA-funded center just this year.

There are five specific aims to the project. The first, which I consider the core beginning aspect of the project, is to simply describe five broad categories of genetic services and the different ways or models that are used to deliver these services, including the

personnel involved, the supply and demand influences for these specific models and the patients served.

A follow-up to that which will begin in the second year is to develop a methodology for in-depth exploration in selected communities of the way the genetic services integrate with health care delivery within the services, looking at health care organizations consolidation, insurance, managed care coverage, safety net health delivery concerns issues and other issues.

A second aim of this study is to assess through separate survey studies current and emerging genetics-related practices of geneticists, of genetic counselors through the current 2002 Professional Survey done by National Society of Genetic Counselors, and the third, you'll be surveying nurse geneticists. In the second year, we'll be surveying primary care clinicians who are both physicians and nurse practitioners. We expect that these surveys will help and complement the core studies of understanding how services are delivered and issues facing practitioners.

The fourth aim is to develop a methodology to monitor and report on the volume and types of genetics testing offered and evaluate whether this measure can be used as a demand indicator for genetic services. We will in this second and third year look in depth at other potential demand factors.

The fifth aim is to develop working relationships and efficient communications with key public and private organizations planning for services, such as this committee, and to disseminate widely the study findings.

Let me take a minute to describe the staff on the project and then briefly touch on the first three projects for this year. The staff includes a talented group drawn across the country of social science researchers, survey researchers, social psychologists and others as well as clinicians from the field of genetics and non-geneticists. We have engaged as consultants an experienced genetic counselor. Just as a comment, I think about 12 or more people sitting in the room are directly involved with the project, either as staff to the project,

serving on our advisory group or funders. We have a nurse geneticist and a Ph.D. geneticist. We have a family practitioner from the state of Washington who is also involved with the Rural Health Research Center located there. We have a nurse health services researcher at the University of California, San Francisco, a survey researcher under Ed Salsberg's direction at SUNY who's done extensive surveys of physicians and other health professionals nationally and within their state, and within each of these areas, we have geographic breadth as well as disciplinary breadth.

The first-year studies. The first study that is beginning to look at genetic services models and the roles of health professions really begin in January of 2002 and is expected to extend through April of 2003. It's a qualitative exploratory study that will look at services in these categories: prenatal genetic services for children, adult genetic services, state-sponsored and public health genetic services and innovative models for service delivery.

So far, we've conducted a number of pilot interviews ranging from academic health center-based pediatric genetic services, a family practitioner in rural Maine, a physician who's a geneticist and directs funding coverage decisions for a group practice in an HMO in Central Wisconsin, prenatal genetics, an interesting model that is private practice, reproductive practice, in this area, genetic counselors working for a major laboratory offering genetic testing and counseling services, state-sponsored programs from the states of Washington and Iowa.

This is just the beginning. As we move through this, we intend to conduct about a hundred structured interviews with people from across the country from various settings, providing these services and trying to extract from this interview data emerging trends and themes, changes in the history of the organizations and the way their service model has developed and changes that are happening because of pushes and pulls within the market, shortage or availability of specific health professionals to provide services, referral arrangements back and forth to geneticists and a number of other issues.

In the second year, we will begin in-depth study of two or three communities and will follow an exciting research methodology that is known as the

Community Tracking Study. This is probably going on the eighth or ninth year, over $100 million investment that the Robert Wood Johnson Foundation has made in the Center for Health Systems Change. That's a Washington, D.C.-based organization that was begun in '93 after the Clinton reform when it was realized that policymakers needed more information about change that was occurring in health services delivery driven by insurance, managed care, other local market factors, and that study has looked at 12 communities in depth and we're hoping to overlay our genetic services studies on the extensive information that they've collected.

The second study this year is a survey of geneticists, M.D. and Ph.D. geneticists, using as a sampling frame the American Board of Medical Genetics to try to update some work that was done in the '90s and extend that to look at current issues, current practices, trends that are occurring among geneticists, referral relationships and other issues that they see.

In the second and third year, other groups will be studied, and the final study is our Assessing the Genetic Testing Volume, and this one has been the most difficult to develop a methodology for and it's still evolving. From working on a national pharmacist shortage study a couple of years ago with HRSA and with the research team, we found that as was indicated measurements of demand as opposed to need, both are very important but measurements of market demand for a service or a professional are very hard to get at.

For the pharmacist study, we found that one of the helpful measures was prescription volume which is a strong indicator of demand for pharmacists in the community or retail setting because every prescription is handled by a pharmacist in some way or another, and drawing that analogy to genetic services where we expect the breadth, the amount of services to be growing in ways that are somewhat unpredictable but expected to grow, we feel if we can identify some marker indicators that can be measurable, that this would be extremely helpful, realizing that there's lots of adjustment factors that one has to make.

This study would be population-based, getting estimates of population at risk. It would have information we would try to derive from other studies, uptake rates for testing services, involve repeat testing, lots of other issues, but beginning to develop a time

sequence monitoring of specific test volume, we feel, would be extremely important to help inform the studies that we're doing and inform the broader group.

We've had some exploratory contacts with genetic testing laboratories. We've had good relationships and comments and help with the GeneTests group and Roberta Pagon, who have taken us here, and with CDC, which is interested in looking at testing from the genetic counselor aspects. So I think there are multiple different strategies and ways to look at this, but I do think this will be a measure that will be very useful to the work of the committee and to others and it's been a tough nut to crack until now. So we're taking a shot at it and would welcome suggestions.

In our exclusiveness with the study, we have an external advisory group that we're having a meeting in September-October of 2002, and certainly we'd invite any members of the advisory committee or their delegates to attend that or to become involved with the project in any way that you desire, and we're grateful for the support and for the many opportunities in the year or so that our team was developing the studies, with opportunities to talk to many people around the room or your staff about the project.

Thank you.

DR. McCABE: I'd like to follow up on your last point, the issue about volume and the difficulty in getting volume. I'm glad that you're going to work with Dr. Pagon and GeneTests, but this has also been a difficulty for the committee to come up with the volumes which seems like a very simple and fundamental number that one needs to deal with to understand exactly what the impact is going to be.

I have a question, and Muin is away from the table now, so I'll address it to Joe Boone from CDC and CLIA. Does CLIA require as part of participation in CLIA documentation of the number of tests that laboratories perform? Can you come to the mike, please, Joe?

DR. BOONE: They have tried to capture that information and it's proven to be fairly unreliable because it's sort of voluntarily supplied and actually the fee that the

laboratory pays is based on the test volume that they report. So there's an incentive not to actually report a higher number, I think, in some cases.

But in general, we don't have a very accurate measure of the actual test volume that's being done, and we have been working with the National Society of Genetic Counselors to try to set up a survey that would allow us to get some information about that test volume.

DR. McCABE: It would seem that trying to get one's hands on a reliable measure of volume would be extremely important, and I would think while it might be considered proprietary information, it would be of value to the laboratories performing the test to show that there is demand.

DR. BOONE: I agree with you. New York State probably has a better handle on this than most of the other groups because they do base their licensing fee again on test volume, but it's on tax revenues, so taxable income.

DR. McCABE: Well, we appreciate your comments on your entire study, Dr. Cooksey, but this is certainly an informative point that you've made about one of the hurdles that you've faced.

Francis?

DR. COLLINS: Well, I'm a fan of this study, and I guess the fact that NHGRI is supporting it, it's a good thing. But I do think you have an extremely difficult task in front of you because not only do you have the challenge of trying to collect the data about today, which is not easy to come by, but the challenge of trying to predict tomorrow in a very rapidly changing environment where I don't think anybody imagines that we are riding anything like a linear curve, and so other models that may have been used in this kind of workforce analysis, sort of following, for instance, the volume of prescriptions, may not work so well in an unstable environment where I think all of us anticipate that the demand for genetic tests is going to rise pretty dramatically over the next five to 10 years.

In that regard, it's going to be very challenging to imagine then what the

possible models for delivering genetic services might be, and obviously we have a certain model that applies today and there will be arguments that that's the right model for tomorrow or that it needs to be completely redrawn for tomorrow, and it's going to be a real challenge, I would guess, for you all in this study to come up with some sort of possible proposal of what the right workforce might look like and especially so if you're trying to do this over a longer period of time than a couple of years.

So I guess to get to my question, I'm really trying to understand how far in the future is your study aiming to project in terms of what the workforce needs are going to be for genetics?

DR. COOKSEY: I think it's going to be extremely difficult to forecast that sort of thing, and I'm hesitant that I don't know how to do that, and I don't think good methods are out there.

Instead, I think we can cast a very wide web, and I think changes will be incremental in some areas. I don't think as we try to train the bulk of the providers there will be revolutionary change. I think it'll be incremental. We'll see adapters. We'll see others who are kind of ahead of the pack. So I think this continuing broad look at the many models for delivering services will be essential to do this.

The time frame is yours, 2010 is where we're trying to head for. There's a little disconnect between our funding in that.

(Laughter.)

DR. COLLINS: Lobby, lobby.

DR. COOKSEY: No. We anticipate, though, that our study and others that will be funded by your institute and others are extremely important to be aware of and to keep a very open sharing experience and that is something we're very interested in doing. I think this is one of the most exciting areas.

I may comment, though, that we learned from looking across workforce. Again with the pharmacist workforce, one of the critical professional issues for that profession

is being reimbursed, getting provider numbers and being reimbursed by Medicare and others for their cognitive services and delivering pharmaceutical counseling advice to patients, particularly the elderly, as a Medicare prescription drug benefit is being played out and again the quality of care. There is some data outcome study there. It's not great. It's tough work to do but again one can look at other professions on the realm of cognitive services.

So I think bringing together groups that have interests in health workforce research, but I think even more importantly health services research, where we look at how services are delivered and the constraints and the barriers and the opportunities in the market will be the way to go with this study.

DR. McCABE: Yes, Elliott?

MR. HILLBACK: I'd like to agree with where Francis was going. I think it is going to be very difficult to do.

Unfortunately, most of our examples of uptake are around relatively rare tests and rare diseases, but I do think the CF example, which has been quite dramatic since last summer and last fall, may be at least one chance to get a different look. I know in our lab at Genzyme, we're in the process now of moving the lab for the second time since last year just to keep up with the demand on CF testing, and I think we'd certainly be interested in working with you --

DR. COOKSEY: That's be great.

MR. HILLBACK: -- if you aren't already on how to do that.

DR. COOKSEY: That would be great. Again I think a policy switch or a reimbursement switch can make fairly dramatic changes in the system because we are highly intelligent and responsive individuals and organizations.

DR. McCABE: Wylie, Vence, and then we're going to break for lunch.

DR. BURKE: Elliott, I was a little disappointed that you didn't use the word "iterative."

(Laughter.)

DR. BURKE: I think there is going to be an iterative process between the development of workforce, the development of educational models, the potential changes in reimbursement, et cetera, and we probably just need to anticipate that as these kinds of data are collected. There may be intervention study opportunities that may arise.

The other point I would make is that although the CF model is an extremely powerful model, in fact, that represents a broad use of a test where one could anticipate a significant counseling component, a need either for many providers who aren't used to it to become accustomed to certain genetic counseling issues that arise around reproductive decisionmaking or enhancement of access to genetic counselors or both; whereas, I think that the big wave that's coming our way might be typified more by a test like Factor V Leiden, a genetic test that is a risk factor that if it has clinical utility has a kind of utility that is very comparable to other risk factor data that many clinicians are used to routinely dealing with, and so we may see a divide between the kinds of counseling services and special training required to deliver a test like CF carrier testing versus a test that is a risk factor for a common disease.

DR. COOKSEY: And I would add, one of the groups again simply because of funding limitations that we are not looking at in these first three years are the medical specialists, and I do think that a lot of those sorts of tests, the diffusion and the uptake among the professions will be in the highly-trained specialists who will see this as a tool to provide better care for their patients, rather than as a genetic tool per se. So to understand what's happening there and to track that over time would be extremely complementary to the work that we're doing.

DR. McCABE: And Vence, and Dr. Cooksey, if you can keep your question and your comment relatively brief because I was just informed we have another public commentator before lunch.

MR. BONHAM: Thank you.

This sounds like a very exciting study you have. I have two questions for you. First is related to the qualitative study that you've just started and how you plan to report

out some of the information you get with regards to barriers to access to genetic services for underserved populations.

Then my second question is related to your workforce survey, and will you be collecting data based on gender, race and ethnicity of the different types of providers?

DR. COOKSEY: To the second, yes, we will. To the first, the funders and, I think, the research team is very concerned about access to services for underserved populations, whether it be rural, inner city, low income, minorities and others, and that's a very complex issue around this. We are hoping as we capture models and hear about models to learn more there.

My colleagues in Texas are hoping to kind of join our study with a particular focus on border health issues among the Hispanic population, and I've heard you present and I think that your work and others is extremely important to inform us in asking the questions and to being good listeners to these hundred or so interviews that we conduct and be sure we have the right pool to understand those issues but certainly safety net providers and other groups are important to this.

DR. McCABE: Thank you, Dr. Cooksey, and I hope you will keep us informed as you progress.

DR. COOKSEY: Thank you.

DR. McCABE: Dr. Sundwall from the American Clinical Laboratory Association would like to make a comment.

DR. SUNDWALL: Thank you very much. I appreciate this opportunity. I promise to be brief. I don't want to keep you from lunch.

As you've heard, I'm President of the American Clinical Laboratory Association, but I'm going to wear another hat today because I've been so interested in your talk on education. I'm a former administrator of the Health Resources and Services Administration, HRSA, and I can't tell you how pleased I am to hear all this attention on HRSA. It's that unknown agency but very important in our public health efforts. Also, I'm a former chairman

of the Council on Graduate Medical Education and have a bit of an informed perspective on health workforce.

Let me just share with you that I just returned from the Sixth International Conference on Physician Health Workforce held in Ottawa, Canada. Australia, New Zealand, the U.K., Canada, the U.S., meet every now and then to talk about how do we measure the physician workforce.

Let me encourage you, please don't get paralyzed by trying to determine the right number or demand for a discipline in health care. You can't do it. No nation can. We share. What we do at these meetings is essentially commiserate with how poorly we do at determining future demand or the right mix of specialists and physicians, how do you get primary care doctors to practice where they're supposed to.

What I've heard this morning is that notwithstanding your inability to measure and all due respect to Dr. Cooksey and her committee, she won't be able to and her colleagues, the best and the brightest, because we live in a very dynamic health care world. What I've heard here is an enormous need for everyone to understand the promise of genetics, what it might do to improve patient care, and you need to do that through an educational process.

HRSA has, through the Bureau of Health Professions, a wonderful track record of providing carrots to educators to train, whether it be physicians, nurses, physician assistants, now genetic specialists, whatever, and they do that through a variety of mechanisms.

Also, don't be discouraged by the fact that this Administration has proposed zero funding for that agency or that bureau. Well, so did President Clinton. It's a bipartisan lack of regard for the importance of health education.

(Laughter.)

DR. SUNDWALL: But the good news is the Appropriations Committee has already approved, as I understand, full funding for these activities.

A friendly suggestion, and I know you can't lobby or you're not supposed to,

but is there some way this committee might pass a resolution, based on the work you've already done to date, about the importance of this?

The reason I say that is because Title 7 and Title 8 are up for reauthorization this year. That's the Health Professions Training Assistance Act, the Nurse Education and Training Act. These are of critical importance. Now, when I was administrator of HRSA, I would have so much welcomed, I did welcome directives on how to spend those funds because, trust me, there's a lot of competition from worthy health profession education interests, and I just wonder if you'll get the attention or get the money you need without a legislative directive or at least committee report language in the event they conference these bills. It's the House Commerce Committee and the Senate HELP Committee to do this.

So, anyhow, just a friendly suggestion. I think the time is important because if they are reauthorized this year, it won't come up again for three or four years. So that's something for you to deal with what you can and for those who are interested that aren't federal employees, you can lobby all you want for this kind of language.

The last point and I promise I will make more formal comments next time, I appreciate being here, it's been very educational for me. Speaking for ACLA, the American Clinical Laboratory Association, that's a small trade association but representing far and away the preponderance of the commercial labs in the country which come under these large national and regional companies that are members of my organization.

I just wanted to inform you if you weren't aware of it that we're very actively engaged in the coding issues and, of course, they are very important with reimbursement and the things Sean has to wrestle with at CMS. ACLA proposed five genetic CPT codes in February at the Phoenix meeting. We got one approved for cystic fibrosis, and then we were pleased about that and the fact that they listened to us, but on further reflection and collaboration with the College of American Pathologists and Academics, we realized it was probably premature to get a code for this particular test, even though there's quite a high volume of it already being used.

So a work group has been going to be convened. Dr. Michael Watson, American College of Medical Genetics, College of American Pathologists, ACLA, we're going to wrestle with how do you come up with a consensus on an approach to coding because it's so very important, as you know, to get access or get those tests paid for, get them ordered and paid for. So that's just something you should be aware of and as we work on our coding issues, we'd be glad to share that with this committee.

DR. McCABE: Thank you very much, Dr. Sundwall, for your comments and you're right, we cannot lobby but we can make recommendations to the Secretary. So thank you. We look forward to your continued involvement.

Very brief, Ms. Benkendorf.

MS. BENKENDORF: Just with regard to the Title 7, which is the Health Professions Act, both the Government Legislative Affairs Committee of the College and the Association of Genetic Counseling Training Program Directors have had their eye on that title for about a year. We do have some model language we're working on, and our understanding from speaking to members of the House and the Senate is that that's not going to be only in this session. But we do have our eye on it.

DR. McCABE: Thank you.

Let's break for lunch. We will resume sharply at 1:30. The lunch for the members of the committee will be in the Lombard Room upstairs where the breaks have been.

Thank you.

(Whereupon, at 12:34 p.m., the meeting was recessed for lunch, to reconvene at 1:30 p.m.)

AFTERNOON SESSION (1:36 p.m.)

DR. McCABE: A couple of things in follow up to this morning. I would like to ask Dr. Boughman and her work group to have the recommendations reviewed by the work group, which will require some conference calls finalized by the work group and back in that final draft form to the full Secretary's Advisory Committee on Genetic Testing for our August meeting. I know that'll be a relatively quick turnaround, but I think it's important that we move forward with those.

In keeping with that, also, Dr. Sundwall mentioned the importance of some of the laws that are being reconsidered, and do we have a recommendation that we should move forward with a recommendation to the Secretary on -- it was Title 7 and Title 8 which are up for reauthorization, is that correct? -- some indication that guidance might be appreciated regarding the role of genetics in those titles at the time of reauthorization.

Joann, do you wish to make a comment?

DR. BOUGHMAN: I would just suggest that the recommendations that we

had been talking about as a committee and as a work group were affirmed strongly enough and in fact specifically enough yesterday, I think that that would be one of the recommendations that would come forward and because of the timing here, I think it would be much more expeditious if we could write a letter recommending to the Secretary that Title 7 be supported, Title 7 and 8.

DR. McCABE: Title 7 and Title 8.

DR. BOUGHMAN: Be supported so that some of those funds might be sought for genetics professionals training.

DR. McCABE: Would this address genetic specialists per se or that there be some consideration of genetic services?

DR. BOUGHMAN: Maybe we can word it funding for the training of genetic service providers but in the context of the genetic services from this committee, so that it would not close the door to genetic specialists but would not be specifically limited in that way.

DR. McCABE: So would include the broader education of professionals regarding genetics and provision of genetic services.

Yes, Judy?

DR. LEWIS: My understanding of those titles is that they are relatively broad in terms of looking at health professions education and that they're not necessarily earmarked for a particular specialist. So I would think the letter that would talk about the fact that this was one of the important areas, but my understanding of those two titles is that one of them is medical education, I believe, and the other is nursing, if I'm not mistaken, and that they're really broad, so that if we were to put something in our letter about the importance of funds so that issues such as genetic education could be addressed.

DR. McCABE: Any further discussion on this?

(No response.)

DR. McCABE: Since we're talking about putting a letter together that

would go to the Secretary by way of the Assistant Secretary of Health in the interval between this meeting and the next meeting, is there any further clarification of language that anybody wishes to make sure that Sarah and the staff include?

Yes, Dr. Boughman?

DR. BOUGHMAN: This would not be in the letter, but I would just suggest that we defer to staff and their discussions with HRSA or anybody else that would make sure of the language of the letter but not to go in the letter but with that understanding.

DR. McCABE: Pat?

DR. CHARACHE: I just think that we can emphasize the key role of education that had been identified by this committee as a cornerstone of need for genetic testing and that with this in mind, we wish to urge that this be supported to address issues of changing medical needs, including genetics.

DR. McCABE: And we can certainly mention that the strong consensus at the workshop was that this was an area of need and that we will be following up with more specifics.

Since this is a letter to the Secretary, we need to be a little more formal than usual, and so do I hear a motion?

DR. BOUGHMAN: So moved.

DR. LEWIS: Second.

DR. McCABE: Further discussion of the motion?

(No response.)

DR. McCABE: If there is no further discussion of this motion, all in favor, say aye.

(Chorus of ayes.)

DR. McCABE: Any opposed?

(No response.)

DR. McCABE: Any abstain?

(No response.)

DR. McCABE: It carries unanimously. Thank you very much. So we will work on that and get that out as soon as possible following this meeting. Thank you.

We're now ready to move forward. We'll turn to Dr. Lewis, who is Chair of the Access Work Group, for her presentation on the Work Group Draft Report on Billing and Reimbursement for Genetic Education and Counseling Services.

The draft report is at Tab 4 in your briefing materials and a copy of the revised recommendations is in your green folder that was at your place today.

The mandate of the Access Work Group is to explore trend issues in access to genetic tests and genetic testing services, including coverage and reimbursement policy and disparities in access to these services. This is the first report to emerge from the Access Work Group and it addresses a very specific problem in current billing and reimbursement policies and practices and the significant impact that these policies and practices are having on access to genetic education and counseling services.

Tomorrow, Dr. Lewis will report on another report being developed by the Access Work Group on the broader topic of coverage and reimbursement for genetic testing services. She will review with us some of the challenging issues the work group has been exploring and seek our guidance on how the group should proceed with the paper's further development.

Before Dr. Lewis begins to review the work group's findings and proposed recommendations for enhancing coverage of genetic education and counseling services, I want to commend Dr. Lewis for her leadership in guiding the development of the report, the dedicated efforts of the work group, and the excellent writing and analytical support from SACGT staffer Suzanne Goodwin.

Dr. Lewis?

DR. LEWIS: Thank you, Dr. McCabe, and I, too, want to acknowledge on the part of the working group Suzanne's support and Sarah's leadership. Both of them have

worked very hard with our group, and we really appreciate your work, and Suzanne has done a wonderful job in terms of framing some of the issues and helping keep us on track.

This work group began in August 2000 and it came out of our report that we issued in July of that year, and the specific recommendation in the report that individuals and family members considering genetic testing should have access to appropriate genetic education and counseling resources to ensure their ability to make informed decisions led to this particular charge when the work group was tasked with addressing access issues, and one of the access issues includes being reimbursed for genetic services. We've got lots of other issues that we're looking at, but this is the first of many issues we hope to be bringing forward to this particular group.

Our committee has broad representation. Our work group has perspectives on it that come from the Centers for Medicare and Medicaid Services. We have private health insurers providing their perspective. We have labor union perspective. We have employer perspectives, the perspective of health care providers, and the perspective of patients and consumers.

In addition, back when the original report was issued, there was lots and lots of public comments. There are two books of comments on the table today. If you remember when we did our outreach meeting about the oversight, we ended up with four or five books of comments and many of those comments dealt with issues of access, so that those comments have informed our work as well.

Just to give you a sense of the overview of our report, the report that you've got behind Tab 4 tries to identify some of the gaps in the billing and reimbursement mechanism that currently exists, specifically areas like CPT codes, the Health Care Professionals Advisory Committee, various billing options, the Provider Identification Systems, Standards of Licensing and Credentialing, and some of the data issues, and we propose 10 recommendations to address some of these gaps.

Let's start with CPT codes. For those of you who don't know, this is the

system that's developed and it's maintained by the AMA. It's used by providers who bill both public and private health care plans, and every medical procedure that's done has an associated five-digit number, and the evaluation and management codes are the ones that are used for professional services. So that, if you don't have a code, you can't put something into the system. It's sort of like having your credit card numbers when you go to order something, that if you don't have them, there's nothing you can do, and there is no CPT code specifically for genetic education and counseling.

Currently, it's reported under some of the generic evaluation and management codes, and the codes that exist right now don't reflect the amount of time that has to be spent in terms of preparation time to meet with families, the extended duration of patient settings that are involved sometimes in genetic counseling, and the follow-up time after the genetic counseling visit, and the other thing is that ACMG has requested a new CPT code for "family history, risk assessment, pedigree analysis."

I don't know. Perhaps if Mike Watson joins us later, he can update us on the status of the request because that's something that's a work-in-progress, and we haven't heard back in terms of where that's at.

In terms of the AMA, there are several committees that are involved in the CPT process. There's an Editorial Panel that revises, modifies and updates the various CPT codes, and then HCPAC, the Health Care Professionals Advisory Committee, is limited to licensed practitioners and allied health professionals, so that they are another committee that works to advise the Editorial Committee, and then there is the CPT/HCPAC Committee which again has physicians and other advisory committee members and again they advise the panel, so that they're out there, too.

Mike, I don't know if you have any information about the request that went to CPT, if that's something you can share with us perhaps at the end of my presentation. You were out of the room when I took your name in vain. So just to be put on alert.

Okay. One of the things, though, is that the advisory committee doesn't

include certified genetic counselor representation. Now, so therefore, genetic counselors or NSGC members don't have a voice in any of the coding decisions, and NSGC has requested that genetic counselors be included, and I don't know what the status of that is either. So we're hoping that perhaps the NSGC can update us on what the status of that request is and whether or not they've heard back in terms of their request to be included on the HCPAC.

Now, billing terms. One of the things that happens is that as with many things, Medicare has certain people that are authorized to bill Medicare directly and those include physicians, nurse practitioners, physician assistants, certified nurse specialists, certified nurse midwives, clinical psychologists and clinical social workers. So those folks can go ahead and be direct billers to Medicare, and others who don't have that ability can bill incident to and their services have to be supervised. They're usually employed by and supervised by and bill services through their supervising physician or their supervising hospital and people who can authorize Medicare in an incident to fashion are non-advanced practice nurses, non-physician anesthetists, psychologists, technicians, therapists, and other folks in those categories.

There has been a request for this to go forward, but in terms of physician presence and physician supervision, there is a lot of discrepancy as to how close that supervision has to be to be incident to.

So the third gap we've identified is that certified genetic counselors aren't listed in Medicare's statute for direct billing or for incident to billing, and therefore along with the limited use of CPT codes, there's really not a whole lot they can do, and it's pretty much of a unique problem for genetic counselors. Other folks who do genetic counseling and patient education and counseling around genetics have the ability to bill either directly or incident to but genetic counselors aren't listed at all in either one of these categories.

Now, the other thing that has to happen, in addition to having to have a CPT code for the procedure, the provider has to have a number and there is a system of UPINs or unique provider identification numbers, that are assigned by CMS and they're assigned only to providers who are eligible to bill Medicare. Now that's fine, except that most of the private

insurance companies are also using these same numbers because it's a system that works and it's a system that numbers are available, so that even though they're issued by Medicare, some of the private health care insurers use those as the way to get in, and again if you don't have a number to enter in the box on your computer form, you can't just click submit because there's critical data that's missing.

There is new system that's been proposed that's in development that is a national provider identification or an NPI system, and these will be assigned to all providers who are licensed, certified or authorized to provide medical services and engage in standard electronic transactions with public and private health care firms, not just Medicare.

So the next gap that we identified is the fact that certified genetic counselors are not eligible for UPINs and that this is a barrier to their ability to obtain authorization to directly bill health care plans. We do expect when the NPI system comes forward that genetic counselors will be eligible for NPIs but that's not currently, that's in the future.

The fifth gap that we identified were licensing and credentialing standards for genetic education and counseling are lacking. Few states have licensing programs for certified genetic counselors and therefore even though licensing doesn't necessarily guarantee reimbursement, what it does do is it creates visibility, it does create awareness, and it does create recognition, and the other thing that I believe licensing and credentialing does is most of those come out of the Department of Consumer Affairs, so they do present some consumer safeguards.

The next gap that we identified and this has been spoken to already today is that data on coverage and reimbursement of genetic testing services are greatly lacking. There have been a couple of surveys and we do have an article that was written in Genetic Counseling, but there is not an awful lot of data on issues related to coverage and reimbursement.

So we tried to get some data. Now, this is not scientific and it was not done

through any great scientific process, but one of the things you're limited without going through agency clearance, which is sort of like the Federal Government's IRB review process, I would guess, which to me is always pretty awesome and intimidating, but in order to do something, that if you have more than nine folks in your sample, you have to go ahead and you have to go through agency clearance. So if you do a study with nine participants, you don't have to go through agency clearance. So we went ahead and surveyed nine participants, and you could basically call this pilot work, and we had a 36-question survey that we sent out to laboratories, private teaching hospitals, physicians' offices and groups and public health laboratories. So far, we've had six responses, and so what we've basically got is we've got some qualitative data on billing mechanisms used by genetic education and counseling providers and we have some sense, you know, at a few points that gave us some information.

Now, on their own, the California Department of Health conducted a similar survey at nine prenatal diagnostic centers. So we've got two points of information, one of which we did and one of which was done by the State of California. And just to give you a sense of what they found in terms of the barriers created by current reimbursement rates, we find that a majority or a huge number of people said that some of the reimbursement issues really have prevented them from hiring genetic education and counseling providers, and some people have actually noted that they've had to lay folks off. It's threatened the viability of programs. It's caused a limitation in access in terms of the number of patients, and it has prevented facilities from offering services at times that they were indicated, and a couple facilities said that they could only accept direct payment and that therefore they weren't able to bill insurance companies for some of the services that they offered. So we saw that this was, even though it's not scientific, it does give us some beginning evidence to support some of our assertions.

We think that part of what's happened in terms of the current mechanisms of billing and reimbursement is we really think that some of these issues have discouraged providers from entering professions or from specializing in genetic education and counseling

because there are other fields where it's just easier and that some providers have actually left the field because again it's been too hard. They've been laid off. They haven't been able to get reimbursed for their services.

We believe that it's discouraged medical facilities from hiring genetic education and counseling professionals, that it's threatened the viability of some genetics programs, and that at some level, it may dissuade patients from getting counseling or from being tested, especially when it forces people to pay out of pocket because their services aren't reimbursed, and I think we're in the era now where we just can't give it away anymore, where people's services have value and they deserve to be reimbursed.

So based on all of this and based on a lot of discussion, we have gone ahead and come up with some recommendations and you've got them in front of you. Please use the attachment, committee members, that's in your folder, not necessarily the one that was in your notebook, because there's been some revision to a couple of the recommendations and that's the most current list.

We want to recommend to the Secretary that the Department collect and assess data on the current state of coverage and reimbursement for genetic services, including genetic education and counseling services. We think it's a big enough problem that the Department needs to take the lead in terms of collecting the data.

That AHRQ or another appropriate agency should be charged with developing an evidence report that assesses the value of genetic education and counseling services and the qualifications of professionals providing these services, and I think this was addressed earlier this morning as well.

We want to recommend to CMS that you include certified genetic counselors in the list of auxiliary personnel eligible to bill Medicare under the incident to provisions and that CMS in its capacity in implementing and administering the NPI system should include genetic counselors in the list of providers eligible for those numbers.

Through its representation on the Advisory Council to the CPT Committee,

that the Department should support the expansion of the membership of HCPAC to include genetic counselors and that CMS as a member of the CPT Editorial Panel support the establishment of a CPT code that adequately reflects the time involved in providing genetic education and counseling services and case preparation and follow-up.

We also have some recommendations to some of the states and to some of the various and sundry professional associations, and we believe that states and professional organizations should establish licensing and credentialing standards for genetic education and counseling providers and that professional associations should work together to develop an overview and a justification for the service components needed for ensuring the appropriate use of genetic testing, including genetic education and counseling services.

We have some recommendations to health care insurers and to all health care plans. We believe that Medicare and other public and private health insurance programs should adequately reimburse genetic education and counseling services and that health care facilities and health plans should consider the various available strategies for billing and funding genetic education and counseling service.

We're able to come to consensus on all of those recommendations. There are a couple that are out there that we have yet to come to consensus on and we really need some guidance from the entire committee in terms of how we should move on these, and I'll discuss them one at a time, and the first was a recommendation, a proposed recommendation to the Secretary that the Secretary propose legislation to Congress that allows appropriately licensed and/or credentialed genetic education and counseling providers to directly bill Medicare for genetic education and counseling services.

One of the things that we've been educated by is that Medicare, the services that you can pay for are only those that are legislated. So if we want to make a change, this involves legislation. It can't just be a recommendation, and some of the issues that came up in our discussion was the issue of whether or not this should be looking at the service or at the personnel providing the service, and the issue was that there was some feeling that genetic

counselors were the only excluded group, so that therefore the recommendations needed to deal with them specifically, but the thought from others on the group was that the issue was the service and that we really needed to focus much more on the need of the humans out there, rather than on the lack in terms of health professionals getting reimbursed, that that might be a little self-serving, and that we might be better off if we phrased the recommendation in terms of unmet need in the population, but we never really came to consensus on that because there were people in our group who felt strongly in both directions. So we bring that to you for your wisdom and guidance.

The second one was public and private health plans allowing appropriately licensed and credentialed genetic education and counseling providers to directly bill for genetic education and counseling services, and some of the discussion around this issue were the issue of adequate versus direct reimbursement and should we be focusing on having services be reimbursed adequately, rather than having certain providers be reimbursed directly, and we were reminded that many employers are self-insured and that the self-insured employers account for about 60 percent of those covered lives in terms of health insurance and that the groups, these particular groups, actively fight against licensing and service mandates, so that we might end up not necessarily getting what we want if we focused on licensing and servicing mandates as opposed to focusing on adequate reimbursement for the kinds of services that were done, and that the other thing was that incident to billing might be more acceptable than direct reimbursement because of the strong opposition to this particular language from some of the Insurance Committee and the fact that the issue was again, is it something that we really need to be focusing on, whether or not we're looking at the individual provider or we're looking at the adequate reimbursement for the services.

So those are the two recommendations that we've had lots of discussion about, that we haven't necessarily come to consensus about, and so, you know, I'd like some discussion on that and then also to get a sense from all of you whether or not you believe that once we come to some conclusion on these recommendations, is this report ready for prime

time? Is it ready to go out for public comment or what tweaking do we need to do before it goes out for public comment? So that's where we're at, and I really would value your input.

DR. McCABE: Thank you very much, Judy, and thank you to your work group.

You had mentioned a couple of individuals who might wish to comment, and so Dr. Watson and Ms. Schneider, if you would be willing? Mike, you want to go first? If you could state your name and your position, your affiliation, please?

DR. WATSON: Mike Watson, Executive Director of the American College of Medical Genetics, and it was about the CPT code proposal.

DR. LEWIS: Yes, sir.

DR. WATSON: Yes, there you are. Yes, we submitted it for the early April deadline, a two-tiered code for pedigree development, analysis and genetic risk assessment as a package at two different levels. One was a base level where there wasn't much need for confirmation of medical records, confirmation of test results and such, and another which is more like the breast cancer situation in which you do often have to confirm a lot of information about other individuals within a pedigree in order to establish a genetic risk assessment for the individual sitting in front of you.

It goes to the committee in August. We're now in the process of discussing with various of the CPT advisors and interest groups who might or might not support that to better educate them about what it is that we're looking for in that code.

Interestingly, the payers have been among the most interested in the code because what they've seen happen in genetics is that when patients get good education and counseling as was evident in Huntington's disease when, you found, we found that before we had the gene, everybody said they'd get tested. Once you had the gene, it turned out that only 13 percent of people wanted to get tested when they were really told what that information meant and what they could do with it.

So many of the payers have seen a reduction in test utilization based on

good education and counseling about the use of tests. So they've been among the more supportive. The less supportive have been the urologist who says, oh, I get a family history and, you know, everybody else in primary care areas who says they get a family history, they don't need a new code for a genetics family history.

DR. LEWIS: If you could keep us informed as to how that's going, that would really be helpful. I don't know. Did you hear the question that we had? I don't know. Were you here?

MS. SCHNEIDER: Me?

DR. LEWIS: Yes.

MS. SCHNEIDER: Yes.

DR. LEWIS: Okay.

MS. SCHNEIDER: Yes. So Katherine Schneider, President of the National Society of Genetic Counselors, and in January, I sent a letter formally requesting representation on the HCPAC Committee. In March, I provided additional information and justification for such a request, and it is my understanding that their committee met last week and I have yet to hear the outcome of that. They promised to let me know one way or the other, and I will definitely keep you posted, Dr. Lewis.

DR. LEWIS: Thank you.

DR. McCABE: Wylie, you had a question for Mike Watson?

DR. BURKE: Well, actually, it's a question of clarification for either Mike or Judy or both, and that is, if you were successful and if the CPT code were granted, would there be a restriction on who could bill under that code or is that a code available to any provider?

DR. LEWIS: I think the issue is that the code becomes available, and Suzanne, you can help me if I'm not right on this. I think the issue is once the code is available, folks can submit under it and then the particular insurer chooses whether or not to reimburse, but you can't even submit at this point in time because there is no code and part of it would be

Medicare, but part of it is also that those are the same codes that all insurers use.

DR. BURKE: But part of my question is, would this enable a family practitioner who knew how to do it to get reimbursed for the same services?

DR. WATSON: Yes. It goes in outside of the E&M codes. So it could be stacked to an E&M code, rather than be an incident to that E&M service, and it is not restricted at this point in time as to who could bill for it. More restrictions tend to apply to who can get paid than to who can bill for something in those systems.

DR. McCABE: Sean, do you want to comment on this?

DR. TUNIS: Yes, just a couple of things.

One is, certainly you're right that on the issue of genetic counselors being able to directly bill Medicare, that would certainly require a statutory change. Medicare does not have the legal authority to directly pay genetic counselors, and I think your report reflects that understanding.

And just to illustrate how this happens, you know, probably the most recent example of Medicare beginning to directly reimburse a new provider type came through the medical nutrition therapy law that was passed, I think, two years ago that basically made dieticians eligible for payment by Medicare. It did require a law to do that and the law was really based in very large measure on an Institute of Medicine report on nutrition therapy services that did kind of an exhaustive systematic evidence-based review of the impact of nutrition services for all sorts of diseases, and the best evidence that there was in that IOM report was for diabetes and for renal disease. So the law actually now covers medical nutrition therapy for diabetes and renal disease based on the IOM report and created the ability for Medicare to directly pay those folks.

So what's missing from here is, you know, the evidence base. I mean, it's getting boring already with reiterating this point, but the point is you're not going to get statutory change unless you've got something other than a collection of testimonials or the firmly held belief that these services are valuable. It isn't going to happen, in which case if you

don't get legislation, you're not going to be able to get direct paid.

Now, on the issue of incident to, you know, being able to bill under the physician supervision, that is an area that's under the jurisdiction of service, a whole separate group within CMS. That's the Payment and Reimbursement Group. But Jackie Sheridan, who I think was part of your work group, I think recently talked to Prasha Patel, who's a senior policymaker in Payment, and I don't know.

Do you want to talk at all about that, Jackie?

DR. McCABE: Could you please state your name and affiliation for the record?

MS. SHERIDAN-MOORE: Jacqueline Sheridan-Moore. I'm with the Centers for Medicare and Medicaid Services on Sean Tunis' staff.

I have spoken with the staff in CMS regarding the incident to provision that came up during the Access Work Group, and I've been told that the rationale for the exclusion that currently exists in the incident to provision for nurse practitioners, physician assistants, clinical nurse specialists and nurse midwives is based on a statutory provision that provides those kind of practitioners to bill for their services at 85 percent reimbursement for the actual services that they are done.

So that, in order to do a similar kind of thing for genetic education counselors, a piece of legislation would be required similar to what exists for the other kind of practitioners.

DR. McCABE: Thank you.

Just before we move on, I think we appreciate your reminding us of this and helping us to keep our eye on the ball here, and I was going to look up our mission, but I remember in general terms, if not specific, and we can recommend research that's relevant to the rest of our charge, and so this is something you might want to include in your recommendations, Judy, that if we need an improved evidence base, that there be support given to that.

Cindy?

MS. BERRY: Sean, I appreciate your comments because I was actually intimately involved in the medical nutrition therapy case study, and Victor and I were talking earlier at lunch that it is something to keep in mind.

I can't speak and shouldn't speak at all for the Administration or HHS, but my general impression is that the Secretary might not or HHS might not necessarily want to propose specific legislation but rather would come up with concepts if it fits within the goals of the Administration but really it's going to be a congressional effort. It would really be something that someone, not us, would have to interest the Congress in the issue enough that someone would be willing to introduce a bill and then using the medical nutrition and therapy case study as a model, it took about five years to do. It was not something that happened very, very quickly, and in fact, the American Dietetic Association commissioned a budget analysis, similar to what the Congressional Budget Office would do, to analyze the cost-effectiveness of their service, which is something that we haven't really discussed either, and I don't know if the committee wants to talk about that, but that's often a necessary component to interest members of Congress, so they'll say, well, this service sounds great, but what would it cost, and do you think we might end up saving dollars in the long run if we provide this service now?

Then the next phase, of course, was a significant campaign to get the professionals to weigh in with their members of Congress, get co-sponsors on the bill, keep getting the bill reintroduced. Finally the model that was ultimately adopted by the Ways and Means Committee was we want to have an impartial scientific study done, and it wasn't just medical nutrition therapy. There were some other preventive-type services that were included in the package and that's when the National Academy of Sciences' Institute of Medicine was asked to do that work, and only after they made recommendations and after years and years of this effort did they see the actual legislation get signed into law.

So I'm wondering if maybe an alternative might be proposing to the Secretary areas in which genetics can fit in with existing HHS priorities, whether it's in the area

of prevention. I know that the Administration has talked a lot about that. There might be some other initiatives that this issue could come up and whether it's in remarks, whether it's in conferences, and that that might be a way to start the ball rolling and gain interest as opposed to proposing legislation.

DR. McCABE: Certainly, access, also, and disparities, health disparities is another area that has been discussed. Now, that's very helpful in terms of specific strategies. Thank you.

It is important, though, that we recognize that we need to make the recommendations to the Secretary. While our reports go to the Legislative Branch, that's informational and not really advisory.

Muin?

DR. KHOURY: Yes, there's a lot of work that has gone on here, and I just wanted to ask a question. I see all the roads lead to somehow evidence-based guidelines in my mind, and I think I just wanted to ask whether the group has considered in its discussion sort of the shape and form of what would genetic education/services look like in the new age of genetic tests, where you are putting together a bunch of polymorphisms to predict the future risk of disease, and then you can act upon that kind of information to reduce your -- you know, take more drugs, sort of in the context of pharmacogenomics, or change your behaviors or do more medical screening for early detection, outside the scope of the traditional sort of pedigree single-gene-type analysis that would allow us to get to a diagnosis of a single-gene disorder, sort of this futuristic practice of medicine, and for those, I would think the evidence-based guidelines would be even more pertinent than the traditional domain of genetics.

So has that been sort of factored in this discussion here?

DR. LEWIS: Actually, I don't think we even got quite that far. I think we're still trying to look at where we're at right now and get our arms around current lacks in the system, and I think we have to sort of get to today before we can move to tomorrow because I think that's just such a huge quantum leap. I mean, I think right now what we've been looking

at is the fact that the services we've got right now don't even reflect today's needs, and so I agree with you, it's just the tip of the iceberg, but I think that that's probably down the road once we solve past inequities or past problems.

DR. McCABE: Victor?

DR. PENCHASZADEH: I just wanted to make a follow-up question to Cynthia or to Sean regarding that bill, that law on medical nutrition, because you mentioned that it covers only gets credentialed and. So it was specific enough for those two conditions. I'm trying to educate myself as to the process that all this legislative process goes.

MS. BERRY: They tried to get broad coverage for medical nutrition therapy for any medical need as referred by a physician, and the cost data was most compelling in terms of savings in the health care system for renal disease and for those two conditions. So that's what led to Congress finally acquiescing and saying okay. Unfortunately, they make these decisions based on cost often, but we have some pretty concrete data not only on the value of the service in terms of health outcomes but also potential cost savings.

DR. PENCHASZADEH: I just think, because if you take a parallel with genetics, you know, one will have to come up with figures and concepts regarding particular degree of severity of a genetic condition or something that will qualify for the input of a different profession, in this case a genetic counselor.

DR. LEWIS: I think these --

DR. PENCHASZADEH: That's very complex.

DR. LEWIS: I'm sorry. I was going to say, I think these points are really important, and we keep trying to remember that the focus of the Access Working Group is looking at access of services to individuals, and I mean, even though the first thing we're talking about is reimbursement for health care professionals, really the focus of our discussions have been in the area of access. So that, I think this is really helpful.

DR. McCABE: Well, and as we've discussed before, without reimbursement there won't be access.

DR. LEWIS: Right. That's the point.

DR. McCABE: People will not provide services for which they are not paid or at least they won't provide them for very long.

Wylie, then Joann.

DR. BURKE: Well, this is actually a comment that refers to several comments that have been made and also acknowledges the conversation we're going to have this afternoon about a continuum of informed consent, and that is that clearly there will be a continuum of informed consent and therefore a continuum of counseling needs, depending upon the nature of the genetic test.

The Informed Consent Group I think gives us some good guidance on this. I'm guessing that the cost savings data that we would like to see to justify the kind of legislation that was used for nutrition services will be most readily obtainable. You can predict the value of counseling will be easiest to demonstrate where in fact we think the need is greatest. That is, where genetic tests are being used for reproductive decisionmaking or where genetic tests are going to be used for diagnosis of single-gene disorders, high-penetrance mutations, et cetera, and so it might be that the thinking should incorporate that.

The acknowledgement that the value of these codes, the value of these kinds of services is going to be greatest for exactly the services that, as you say, we have today.

The other thing we can predict is that even though there are going to be lots more genetic tests that are sort of genetic risk factors and may not engender the complex decisionmaking the single-gene disorder testing does, we are going to have an increasing number of tests for single-gene disorders. I mean that will go forward as well. That's a need that is going to continue to be with us. So I think we probably need to keep that framework of recognizing different kinds of tests, needing different kinds of counseling as we move forward on this.

DR. McCABE: Judy, do you have any comments?

DR. LEWIS: No.

DR. McCABE: Joann?

DR. BOUGHMAN: I would like to turn to Recommendation Number 6 from two different perspectives. First of all, since we serve as an advisory committee to the Secretary, I'm wondering about the wording in the term "recommendation to other organizations" or maybe "issues of importance" or "issues to be considered by" or "points to consider" or something, rather than straight-out recommendations to other groups, although I know in our heart of hearts, we want to recommend these things.

Under the recommendation that pools together states and professional organizations, I would simply urge the work group to reconsider the wording to separate those statements that we would like for the states to be looking at and other comments that we would like the professional organizations to consider. I say this as a result of some of the proposed state laws that have been coming forward on the licensing of genetic counselors and knowing how rapidly and how cursorily some of the language in some of these state bills gets glossed over. So I think we need to be very careful and in fact give the opportunity for a variety of state legislators to pick up exactly the language that would be the most useful to them by splitting out the licensing and credentialing pieces very carefully.

You've done such a good job in identifying the issues. I think we need to go that little step further and not confuse the issue for the audiences.

DR. McCABE: Joann, do you want to give us the specific wording that you'd recommend on Recommendation 6?

DR. BOUGHMAN: I can do that. There were three laws proposed this year and a couple of them were disastrous simply because they mixed and mingled.

DR. McCABE: I'm sorry. I meant on your Recommendation 6.

DR. BOUGHMAN: Right. In splitting them out.

DR. McCABE: No. This had to do with the fact that we should not be dictating to agencies outside of DHHS.

DR. BOUGHMAN: Rather than calling those recommendations, I thought

that we should call them issues to consider rather than straight-out recommendations.

DR. LEWIS: I hear what you're saying, and I think you're making a lot of sense in terms of 7 and 8. I hear what you're saying about 6. If you've got any specifics, if you could just zap them off by email, that would be great to Suzanne and to me.

DR. McCABE: Yes, I was just confused because 6 is really about CMS as a member of the CPT Editorial Panel should support the establishment of CPT codes that adequately reflect.

DR. BOUGHMAN: Seven and eight. I'm sorry. I was looking at the wrong one.

DR. McCABE: Okay. So it is Recommendations 7 and 8, and then if you could help us with model language because I agree with you, some of the language has been better than others in some of those state laws.

Other issues? Pat?

DR. CHARACHE: I'm struggling over here with the concept that if the rationale for this permitting of the billing is targeted towards the high-volume tests where you can get the data on its value, it seems to me that among the areas in which the genetic counseling is most needed are some of the rare diseases where the average practitioner doesn't have the information that they need to counsel the patient. So I'm considering that we should indicate not only that this is cost saving but also the medical value in the rare disease instance, even if they elect not to pay it.

We've already heard that they cherry picked for the dieticians, and they chose the two that had the dollar signs attached, but they didn't choose the overall dietary consultation which is of preventive value in terms of rare diseases. So I'm wondering how to address that issue in a way that can be maximally persuasive.

DR. McCABE: Wylie?

DR. BURKE: You know, you're raising actually what I think is an interesting methodologic problem, because it's possible, depending upon what kind of access

and what kind of data someone might have, it's possible that one could look at a host of different tests all for rare diseases and ask the question whether tests ordered in conjunction with counseling were more or less likely to be ordered appropriately.

I mean, I think what we predict if our hypothesis about counseling is correct, we predict that errors, choosing the wrong test for the particular issue or testing inappropriately, is much more likely to occur if counseling was not, and I think it might be possible to study rare disease testing that way.

DR. McCABE: And I think what Dr. Watson said was that, you know, it's to use the phrase that we've heard so often from Muin. If you tell people what we know and what we don't know about a test, they may not order it if they realize we don't know a lot or what we do know may not be what they were expecting to learn from that test.

Yes, Judy?

DR. LEWIS: What I'd like to do, and I hate to get directive, but in order to move us forward, if we could focus on the last two recommendations, the ones that are up there now where we didn't have any consensus, and it would be really helpful to me in terms of the committee, if we could have some specific guidance from this group in terms of the issues that I raised and moving those forward because I think that that's going to help us move our work forward because we really were pretty much split.

DR. McCABE: Michele, and then Wylie.

DR. LLOYD-PURYEAR: Does that mean I can't address the last point?

DR. McCABE: You can address the last point. Your hand was up before we were given that guidance but then we need to move forward to assist Dr. Lewis.

DR. LLOYD-PURYEAR: Because I agree we need to move forward, but actually picking up on what Ms. Berry and Pat suggested and one of the recommendations in here that's directing AHRQ to collect the evidence, I don't think the evidence is there one way or the other. I don't think the studies are there. So a word of caution is because you do not want the U.S. Preventive Services Task Force to actually look at evidence that doesn't exist

because they'll come up with a conclusion that you don't want, and I think what we'd need to divide, and I didn't think of that until now, I wish David was here, but divide the recommendation into a two-step process.

One is to propose that the Secretary or whoever fund the appropriate studies that Wylie's talking about and everybody's referring to, so you can get at the issue of counseling, and then have AHRQ analyze and prepare the evidence report based on those studies, but I think we need to be cautious because I don't think those studies exist.

MCHB probably would have been the one to have funded those, and just off the top of my head – well, not long ago. I mean recently, you know. Long ago, I think that was probably all MCHB, and I don't think the kind of quality studies are there that you need.

DR. McCABE: Wylie, and then Victor.

DR. BURKE: Yes, just following up on that, I'm going to address Judy's charge, but I will say it's very possible that there are good retrospective studies that could be done starting with laboratory data that identifies what tests were done and goes back and tries to track with counseling. So I think even if studies haven't been done, one might efficiently do some.

DR. LEWIS: And that fits in with the Data Group, doesn't it?

DR. BURKE: Yes, it does. Duly noted.

I would propose that the answer to the question about these recommendations is pretty straightforward and it's based on this discussion. These are premature. These recommendations are premature. Rather, I think we need to make a very forceful recommendation that this data needs to be gathered, that these analyses need to be done.

DR. McCABE: Victor, then Vence.

DR. PENCHASZADEH: I have a couple of concerns. One has to do with the original charge for the group which was an access group, and it ended up being a report or recommendations on billing and reimbursement. Granted, it's because, you know, I was part of

the group as well, so it's part of my doing it as well. In part, because we thought and we identified that billing and reimbursement is one of the major barriers to access. However, it's not the only one, and we are dealing here with Medicare, with insurance and so on and so forth, and we're forgetting that there are 40 million people without access to health care, private, public or whatever.

So I think that the moral strength of this report should include considerations about the access, access to services, and in particular we are dealing with genetic education and counseling services, but it goes beyond adjusting a CPT code or making sure that a particular profession gets credentialed and so on and so forth.

I think that I'm kind of uncomfortable by the level of discussion at the ultimate level that we are, you know, whether or not we should advocate for the genetic counselors' plight or about the particular service without the big picture of the lack of access in general.

Let me finish. That's one thing. The other thing is I, taking on what John mentioned earlier, am familiar with what's going on in the states in terms of language and so on and so forth. I think that one of the pieces that probably we should really make sure is to define what we mean by genetic counseling and education services.

If we don't define that, I'm concerned that everyone will understand what he or she thinks or may equate this to the work of a particular profession or a particular specialty, and I think that would be wrong because essentially I think of a service that can be rendered by a number of professionals and then a number of specialties or generalists provided they have the proper training and skills, and I'm not sure that this is conveyed here.

Probably this is one of the reasons we are ending up with Recommendation A. I mean, with the split or the conflict as to whether we should advocate for a particular service or for the needs of a particular group of professionals.

Now, let me give you my quick answer to your query of the one in which we didn't reach consensus, and as I say, you and we alternatively because I wasn't part of the

last set of discussions. I think we cannot advocate for any particular professional. I think we have to put ourselves in the other end, the end of the patients, the public, and the services the public needs and that's as simple as that.

Now, I would go beyond that and say I've tried to be more forceful or we should be advocating for more than simply being properly reimbursed. We should advocate for HHS to look at what's going on with genetic services in general and what are the main barriers to access and not only the fact that they we don't have a CPT code or a particular professional credential to bill for those services.

DR. LEWIS: Victor, I agree with you completely and that's why when I introduced the report, I think this was one of the issues that was easier to get our arms around, even though it's a hard issue to get our arms around than the overwhelming issue of the fact that there are 40 million uninsured folks who don't have access to basic health care services, never mind specialized health care services.

But we need to remember that this is all being done in the name of access, and as Ed said, if things don't get paid for, people aren't going to be willing to do it for very long, but I agree with you that we really do need to keep our eyes on the actual goal of the working group, which was looking at access of services to humans and that the reimbursement is what's going to get the services to the humans, rather than focusing on the reimbursement per se. So I agree with you completely and really appreciate those comments.

DR. TUCKSON: Ed, can you put me on the list? This is Reed.

DR. McCABE: Yes, Reed.

DR. TUCKSON: Put me on the list, please.

DR. McCABE: Well, we'll let you speak now.

DR. TUCKSON: Oh, no.

DR. McCABE: We know you're so shy that I don't want to risk not hearing from you.

DR. TUCKSON: That's good. I like that.

Two comments. One, Judy, I think your response is also good. I do, and as you know, as a member of the committee, I'm actually proud to be affiliated with this report, and I appreciate the process of deliberation that got us here.

I do think that there is that concern around access, also, because this is such a strong call for increasing reimbursement to certain providers and professionals that still I think the report doesn't indicate that it has to also recognize the context of the escalating cost of health care across the board.

So as you call for this, which I don't think is inappropriate, but still the report does not reflect that it's in the context that everything is inflationary in health care means that you diminish access to something else where you raise the number of people who will not choose to have health insurance and that as you increase the reimbursement to folk, anybody in the system, the co-pays that people have to pay still will have to be covered some way.

The report still has a little bit of an edge of naivete, I believe, in this regard, and it sort of seems like magically that there's a pot of money out there that can automatically be tapped to pay for this.

One of the recommendations in specific that gave me concern was the one for health plans which does say that health plans should find the likely areas of funding for this. Well, I'm not sure what those funding places are that allow this to occur.

My only other comment is on Recommendation Number 8 for professional organizations should work together to develop an overview of justification for the service components. I think the other thing that also that they need to be charged with is some sense of beginning the professional dialogue about who gets to bill for these services and under what conditions and when.

As I have expressed on more than one occasion, I am concerned that it's going to be hard to figure out if you have 10 different disciplines, all of whom are credentialed to do counseling and you have a patient that touches base with each of those disciplines, who gets to bill, and how many times do you bill for the same service? Does the physician ordering

the test bill for genetic counseling at the time of ordering the test then or then do they just simply pass the ball to a certified genetic counselor who will do it?

But at the end of the day, it's just some kind of some mechanism that we can recognize that will allow folks to know who should be doing the counseling in what case and who gets to bill or the first person that gets their bill submitted gets paid and the other person is left holding the bag. Those are the kind of questions I think the professional societies need to talk about among themselves.

Thank you.

DR. McCABE: Thank you, Reed.

DR. LEWIS: And Reed, tomorrow we're going to be talking about the other piece that you've got, and I think some of those issues may come out there, too, as well. So I don't know. Are you going to be joining us from the ceiling again tomorrow?

DR. TUCKSON: I will try my best to join you from the ceiling again.

DR. LEWIS: Okay. It's like you were in the balcony and looking down at us.

DR. TUCKSON: Oh, my God. It should be me looking up to you.

DR. McCABE: Vence?

MR. BONHAM: I just want to follow up on Dr. Burke's comment with regards to the research, and I think one of the things that's important is for us to survey our ex officio member agencies to find out what they have done or what they have in the pipeline with regards to research as well as any of the foundations that are funding research with health plans.

I think it's important for us to find out what's out there. There may not be much and that clearly gives us the direction of what we should be doing.

DR. McCABE: Thank you.

Sean?

DR. TUNIS: Similarly, just picking up on Wylie and Vence's comments, I

kind of share the view that the recommendations here in general on coding and billing seem a bit premature in the sense that they presuppose some knowledge about or general agreement that the services are worthwhile and should be paid for, et cetera. So that's, I guess, where the sense of the prematurity comes from and the need for supporting the research.

I guess what I'm also picking up, obviously there's a lot of believers around the table that it's kind of self-evident that these are useful services and obviously that the genetic testing is evolving quickly.

So I'm wondering if what should be reflected here in terms of sort of a sense of the committee to the Secretary is there's a really urgent need for more research here because we all believe that counseling is critically important and the tests are coming into play, and if we don't get started now, in a couple of years we're going to be in really bad shape because there's going to be this clash between what's paid for and what's known about its value is going to be even worse.

So I guess, you know, something of a reflection of the urgency to gather the information that's going to support this kind of recommendation in the near future, given that there clearly is a fairly serious consensus in the group that we know this stuff is good. We just don't have the evidence to prove it.

DR. McCABE: Is there anyone here who doesn't feel there's some urgent need for this kind of data?

DR. LEWIS: Well, I guess I hear what you're saying. I also hear what I heard loud and clear from the Education Work Group yesterday, which is the fact that we're looking at a workforce shortage and folks not going into a field, and if people aren't going to get paid, they're going to go do something else, and what we don't want to do while we're doing the research is lose the best and the brightest to computer technology or something else in terms of -- I mean, I heard that piece yesterday, too.

So I agree with you in terms of the evidence base, but what we're also dealing with and part of what we were trying to address in this report was the fact that there are

services that aren't going to be provided very quickly because there are no codes to charge for them, and so what we're going to end up doing is having people perhaps getting genetic tests with bad advice or with no advice because people are going to say, you know, here's the test, I can spend 10 minutes with you talking about it and you get to make up your mind, and that's not necessarily -- you know, we may end up having bad evidence before we have good evidence just in terms of the way some of these things happen.

I don't know if there's a way. The suggestion that Vence had about surveying the agencies, we did survey the agencies. I think it was our last meeting that we heard about that. I don't know if there's any way to pull out of that if there is any specific data in any of the studies that have already been funded, if we could ask the agencies to get to that level of specificity because some of them, we had more specificity than others and part of it was because NIH just had the volume that we couldn't get that level of specificity. We did do some review, but we didn't get, I don't think, to everything.

DR. McCABE: No, I think those data were categorized for different purposes.

DR. LEWIS: Right.

DR. McCABE: It had to do with primary research, education, a variety of different categories, but did not get to this level of specificity.

I think, though, that what Sean was saying was that if we feel there's an urgent need for these sorts of data, then we need to make that recommendation so that we don't continually be behind the tide on this. Is there anyone here who doesn't feel that there's some urgency in acquisition of these data?

DR. BOUGHMAN: Ed?

DR. McCABE: Yes?

DR. BOUGHMAN: Quite the opposite, I would say that as far as priorities go, even with the educational issues and so on, that these data would be supportive of the activities in general for which we were training. So certainly from the educational point of

view, this would be, I think, very wise advice heeded well by this group.

DR. McCABE: What would be the best approach to acquiring these data? Is it best to go through federal agencies? Is it best to go to make a proposal to the IOM? Do you have any recommendations, Sean?

DR. TUNIS: You know, just going back to an analogy of, in this case, the IOM doing a report on telemedicine and telehealth services, again kind of in potential support of broadening Medicare payment for telehealth services in an area where it's more similar to this, where the evidence base was weaker than for the nutrition therapy, and I think what that report did was did a nice survey of whatever all was out there, acknowledging that it had significant weaknesses and then actually did some very nice recommendations on research strategies, research methodologies and priorities. What kind of outcomes are you looking for in these studies? What sorts of methodologies are going to be adequate? So sort of set out a framework for actually gathering the data that then became something of a blueprint for Medicare demonstration projects that implemented that framework, et cetera. So it's one model to consider.

DR. McCABE: So since we're advisory to the Secretary, we're not advisory to the IOM, but we could recommend to the Secretary that this is an area that needs expert review of the kind that the Institute of Medicine provides and see if then the Secretary's Office was -- is that what you would recommend?

DR. TUNIS: Yes. You could probably directly recommend that the Secretary commission the Institute of Medicine to do such a report.

DR. McCABE: Right.

DR. TUNIS: Blah, blah, blah.

DR. McCABE: Right.

DR. TUNIS: I think.

DR. McCABE: So that's one. Is there anyone who disagrees with that approach?

Muin?

DR. KHOURY: Can I elaborate on this approach? Remember when SACGT recommended the three-prong attack to the oversight of genetic tests, an FDA process, a CLIA process, and then gathering more data on clinical validity and clinical utility, and that data stuff took us down the path of trying to get information from the agencies and what they're doing to enhance the clinical validity and clinical utility.

So in my mind, the primary objective for why we were asked the questions a few months ago is what are we doing as agencies both in primary research, secondary research, information dissemination, and synthesis, et cetera, to advance the collective knowledge on clinical validity and clinical utility.

So before we go back out and repoll the agencies for what they've done, I think some of the answers to your questions might be in that pile of stuff that was submitted by all the agencies, but if I have one hypothesis right now, I can tell you that you won't find that kind of research being sponsored by anyone or very few or there could be a couple of outlier projects that are designed by the nature of the beast to tackle specifically the issues around testing/counseling and the clinical utility of the test, because I know Sarah is shaking her head.

MS. CARR: I'm just trying to -- because my sense of what we collected was what are the agencies doing to advance knowledge of the clinical validity and utility of tests, not what is the current cost benefit of such tests and also the value of those services.

DR. KHOURY: Right.

MS. CARR: Susanne Haga's the one who knows that data better than anybody. Susanne, do you think that there were things? I mean, there may have been an ELSI study, I don't know, but might that have gotten close to it? I don't know. We can look again.

DR. KHOURY: The only caveat to what you just said is that the tests per se, the clinical utility of the genetic test is not the test but what you do with it in terms of improvement of health outcomes and sort of the outcomes research, and I've always assumed that this was part of the package.

Now, if we want to zero in on the value of the genetic services/education and counseling that accompanies the test and whether that makes a difference as far as outcomes or processes or appropriateness of testing, et cetera, maybe we can go back out, but before we go back to the agencies, let's see if we can find among the thousands of projects that were submitted, because I would think they would have submitted them anyway because they're usually intimately connected with a test of some sort. So there might be some treasures that are buried there, but I suspect that we may not find them.

MS. CARR: And I think the agencies are represented on the work group and if there had been such a study of the value of genetic counseling, education and counseling services generally, I think we would have heard about it. I hope we would have heard about it. But we can certainly look again just to be sure without taxing the agencies again.

MS. GOODWIN: One concern I have with that data, however, is when you're assessing the test and counseling, along with it you're assessing the outcomes of the tests and not necessarily counseling. So it's difficult to assess the value of genetic education and counseling separate from the test. So I don't know if that data that may have been provided earlier in the year would actually get to the value of counseling in and of itself.

DR. McCABE: Do I hear a motion that we proceed with an additional letter? We've already talked about one letter today, but another letter expressing concern to the Secretary about the absence of these data, that there is an urgent need for these data, and that we would recommend that an IOM study be commissioned.

Judy?

DR. LEWIS: I think that's very appropriate, given the fact that we really can't move forward with this issue without that. So I would so move on behalf of my work group.

DR. McCABE: Do I have a second to this motion? The motion is seconded. Further discussion?

Wylie?

DR. BURKE: No.

DR. McCABE: Pat, did you have some discussion relevant to this motion?

DR. CHARACHE: Only that I was thinking, again along Wylie's idea, that this may not be as difficult as we thought when you think about the access to the data, and I was thinking particularly of some of the tumor markers, like colon cancer, where if you get the wrong ethnic group getting the test, you've got people who are getting yearly examinations that are not necessary because of the false-positive rates.

DR. McCABE: Further discussion of this motion? Judy?

DR. LEWIS: My only concern is again, I think it's an important thing, but along with it in parallel, the timeliness of some of the other issues I think is something that we have to address as well, but that doesn't dilute the appropriateness of the letter.

DR. McCABE: Further discussion? Sean?

DR. TUNIS: Well, just one other comment. What the sort of scope of this, say, requested IOM study, if that's what it is, would be, whether it focused only on sort of the value of the education and counseling services or whether you'd want to extend it somewhat to other genetic tests themselves around which you also have reimbursement and coverage issues. So you'd probably want to catch it all at one time, unless you specifically wanted to focus just on education and counseling.

DR. McCABE: No, I guess I should have stated it more carefully. I thought it was the evidence base for genetic testing and counseling that we were talking about.

DR. CHARACHE: We should say including the financial value.

DR. McCABE: Right.

Joann?

DR. BOUGHMAN: I would merely urge that in the writing of this letter, that enough context and background be given to assure the Secretary that this request is coming out of a great deal of study and deliberation by the group, and the background could in some respects be a kind of interim report, but that this was a gap that we see that we've run into.

MS. CARR: But are there recommendations that the committee can agree to now going beyond the need for the data? I mean, what does the committee want to do with the report as it is, separate from the two recommendations where there was not consensus?

DR. LEWIS: I guess our point was that what we wanted to do was bring it here, and if people felt that we want to leave off these two recommendations that we didn't have consensus on, I can understand that, but is this ready for public comment, and will we be further informed if we hear from beyond those of us in this room at this point? Is that going to help us move forward? I'm not sure that I've heard enough here today that's going to inform further development that could bring this report back in three months.

DR. McCABE: My impression is that what we've identified is gaps, that we don't identify a ready source with which to fill those gaps, and so that we're trying to recognize that we aren't going to get anywhere by waiting for three months, and we need to move forward with the recommendation that data be acquired. Do we want to include the first six recommendations as part of the body?

Elliott?

MR. HILLBACK: I'd just like to go back a second to this question of the scope of what we were talking about.

It seems to me that it's two very different pieces of analysis, whether you're trying to ascertain the value of both the test and the educational process by which the test is communicated or whether you're just trying to do the latter, because you start to get into a whole set of issues, depending on which tests you pick, you know, and the value of a life, the value of quality of life, the value of all sorts of things, when you start to look at genetic tests and economic value, and I wonder if that scope doesn't create a project that's a much longer, much more complex, much more politicized request and to try to understand does genetic counseling and education make testing more effective and therefore more capable, and I don't know if you can split them just all of a sudden got to be a very big question that's going to bring a lot of different people in.

DR. McCABE: Well, it is a big question, but there's considerable absence of data. The advantage of doing it through the IOM is that it's an independent organization and should be less politicized there, and they will help identify gaps and the need for further research.

Wylie, Cindy, and then Judy.

DR. BURKE: I agree with Elliott's comment. I just had a few comments about how I think we should think about this, what kind of data we're now talking about, and it seems to me that if we're asking, if we're identifying an urgent need, what we're saying is in context of this report, is that there is generally a consensus amongst people who are involved in delivery of genetic services that genetic counseling has something to offer, and at the same time, we recognize not enough data to justify it at the level of Medicare/Medicaid service coverage.

If that is the issue, I think what we really are doing is narrowing the question fairly in a way that makes it quite doable. That is, the question we're saying is can we see a correlation between a patient having genetic counseling and there are probably a couple of things. One is, the test being used appropriately, quote unquote, which means that we have to limit the analysis to examples where there is a consensus about what the appropriate test use is, and we might consider, and I think this is a much softer issue, whether we want to also include some sort of patient satisfaction understanding kind of issues because those are certainly proposed as benefits of genetic counseling, but they'd be a lot harder to study.

We've heard a hypothesis from Mike Watson that I think it's worth stating, and this responds to Reed's concern, which is the hypothesis that good genetic counseling will actually save money in terms of genetic tests, and I think that is a reasonable hypothesis to test as part of this rubric of does genetic counseling lead to wiser use of genetic testing, particularly when we acknowledge that the genetic counseling for which there's likely to be the strongest evidence of benefit is that counseling that has to do with expensive, high-penetrant, single-gene disorders.

I think all of that leads to, as Pat has said, as I think I've said before, that we probably could put our arms around a study which identifies a limited number of tests or at least a limited category of tests, because it might include rare diseases, a variety of them, where there is a fair amount of consensus about what the appropriate use of the test is and what an inappropriate use of the test is and ask the question.

I would say that's what we should ask for rapid action on. I can say about the HHS review, we've done a little bit more work on looking at that data in terms of the case studies that we'll talk about tomorrow, and in particular Susanne and I have looked at that subset of studies identified by HHS that had to do with BRCA1, and we're not going to find the data we're talking about there. We will find some data comparing different counseling methods.

So that might be part of the review. That is, because some of those comparisons will look at more expensive versus less expensive counseling methods and seeing whether you get the same outcomes, but the outcomes will largely be psychosocial outcomes because the appropriate use of the test isn't the question or the study.

The other final point that I want to make, and I think it's extremely important, is that there's another urgent matter that this approach will not address, and that is that one of the issues in paying for genetic services and having a CPT code and having counselors be able to bill may have to do with access to services on the part of underserved or disadvantaged patients, and that may be particularly relevant when we're talking about Medicaid coverage, and if we believe that some people are not getting tested because they don't have access to counseling services because maybe Medicaid doesn't pay for it, the kind of analysis we're talking about isn't going to uncover that.

On the other hand, if we uncover strong rationalization for genetic counseling, we may be able to open up and we may be able to justify taking actions that would open up services to disadvantaged people. I think we just have to be clear about what we can find and what we can't find with data that's probably immediately accessible.

DR. McCABE: I'm going to ask that we keep the comments relatively brief. Cindy, Judy, Joann, and then we're going to vote on this and take our break. Cindy?

MS. BERRY: I would propose in terms of the actual report, we have the nuggets of it there in Recommendation Number 2, and I'm wondering if maybe we can't revive that a little bit and use that Number 2 and make it the first recommendation, move it up to Number 1 and reference IOM or however you want to frame the request for research and break it out in terms of evidence-based value of genetic counseling. You can talk about testing. You can talk about professionals, however you want to frame it, move that up to Number 1.

And then, I had just one other question, which was, there wasn't a specific recommendation, although I don't know what it would be, in terms of privacy concerns as a barrier to access. I know it has been discussed and part of reports and debate, and I didn't know if that was something that was worth putting in a recommendation form or not.

DR. LEWIS: Actually, it's gone in some of the previous reports, but it wasn't necessarily -- and it's not that it's not an important issue. It just wasn't an issue that was incredibly germane to this particular -- it's an overrider, but it wasn't anything that we felt was unique to this particular issue. So that's why it's not included in this particular report. It's not that it's not important.

DR. McCABE: So do you have any objection with it moving?

DR. LEWIS: No. No, that was around the privacy issue. I have no objection with the ones moving.

My question – I think I'm next on the list.

DR. McCABE: Yes.

DR. LEWIS: May I go ahead?

DR. McCABE: Yes.

DR. LEWIS: My comment right now would be I think that moving with the IOM report makes sense, but what would be important to me would be to know what we should

do with this in the interim, because it seems to me that by the time we get an IOM report commissioned and we get the results back, it's certainly not going to be within the next year or two. It's going to be a lengthy process, as you described. You know, you described a five-year process, but does that mean that the reimbursement issues get put on hold for this perspective or are there pieces of this that we can move forward in a different way?

I guess my question was, did it go out to public comment or do we get to move it forward before that? Because that's just a process question in terms of what happens to the rest of the recommendations in this report. Is it something we put on hold? I'm not sure that that's going to help the access issue.

DR. McCABE: Let's hold that because I want to take the vote on the motion that's on the floor and then we can come back to it.

Joann?

DR. BOUGHMAN: Well, I was actually going to make a combination of suggestions here because I think certainly Number 2 and maybe part of Number 1 are actually in this request for data to come from IOM, and I was going to suggest a two-step process here, that we put the specific request forward to the Secretary for the IOM-based report with some of the background information, and also say that we are putting newly minted Recommendations 1 and 2, and then 3 through 6 and possibly a slightly reworded 7 and 8, out for public comment, and then we could accept a final report in August after those few machinations, but in fact the letter to the Secretary would have already gone, and then we could send the final report in August. So it would be a one-two punch.

DR. McCABE: That's fine.

DR. LEWIS: That's what I was hoping for, was something like that, yes.

DR. McCABE: Then we have a motion on the floor to craft a letter to the Secretary recommending that an IOM study be commissioned, and we've specified the parameters of that study.

Any further discussion?

(No response.)

DR. McCABE: If not, all in favor say aye.

(Chorus of ayes.)

DR. McCABE: Any opposed?

(No response.)

DR. McCABE: Any abstain?

(No response.)

DR. McCABE: It was unanimous then, and Judy, I think we got the answer to your question, and so let's take a 10-minute break. We will resume at 3:15.

Thank you.

(Recess.)

DR. McCABE: I want to clarify what we voted and it's relevant to a discussion that we had at the break, and there was some discussion at the break regarding what it is that IOM can do, that IOM is good at taking data and generating policy from data, they're good at even doing secondary data sets and reanalysis, meta-analysis of data, but in the absence of data, which we think we're faced with here, that that may be difficult for them to do.

What I was thinking we would propose to them was that there is an absence of data, and could they help identify the kinds of data that they would need since they have experience with these kind of analyses and help direct how those data should be acquired and the analyses.

Cindy?

MS. BERRY: I was wondering if there are any private health plans that we know of that do a good job of covering these services for whatever reason, because I know what helped with the medical nutrition therapy situation is that there was a Group Health of Puget Sound which covered those services and that it provided a database for the researchers to look at to build off of.

DR. McCABE: There is a good group that I'm aware of because they

happen to be in California, and Kaiser of Northern California has a just stupendous database with very large numbers, and they have determined that it is cost-effective for them in their health plan to provide these services.

I saw other people. So I think that we could suggest that Kaiser of Northern California would be one resource that could be called upon. So that would be important for us to note that in our letter that they might be a resource for data and apparently Kaiser is on our work group, Sarah just told me, and has been affirmed by Judy Lewis. So that would be helpful also.

Is there any other discussion of that? I just wanted to clarify that point to be sure that there wasn't a misunderstanding. And should we include in that study the health benefits of cookies in the middle of the afternoon?

Okay. Well, thank you very much. That was a useful discussion. We ended up with two action items then, so that was very useful, and we will move forward with those action items then.

We will now turn to Dr. Penchaszadeh and Dr. Benjamin Wilfond, who are co-chairs of the Informed Consent/IRB Work Group, for a discussion of the revised recommendations in the work group's Draft Report on Improving Decisionmaking and Informed Consent for the Use of Genetic Tests in Clinical and Public Health Practice.

The draft report is at Tab 5 of your briefing book. At our meeting in February, Dr. Wilfond and the former member and co-chair, Dr. Barbara Koenig, presented the report and the recommendation that the work group believed were key to implementing change and effecting improvements in the informed consent.

After extensive discussion, the committee endorsed the report's conceptual framework that more than one approach to informed consent is appropriate for genetic tests, that the approaches can be viewed along a continuum from minimal to extensive, that the nature of the consent approach used will depend on certain test characteristics, and that if these concepts were applied in practice, informed consent for genetic tests could be enhanced.

However, the committee did not agree with all of the work group's proposed recommendations. Today, Victor and Ben will plan to rereview the key concepts in the report and present revised recommendations to us for our consideration.

So Victor and Ben, if you would please proceed? Thank you.

DR. PENCHASZADEH: Thank you. Thank you for the introduction which essentially sets the framework, so I won't say much more than simply to acknowledge the work of group members, and you will note that there are two people with asterisks, Joe Boone and Daniel Robinson. They are new members. Actually, Dan Robinson just joined our group kind of in absentia because he had leave, but he was very interested in the discussion.

So what we plan to do is for the benefit of the new committee members just review very quickly and Ben will do that for us, the conceptual framework, and we'll go immediately afterwards to the specific recommendations. So I will ask Ben. Ben was the co-chair with Barbara Koenig in the first couple of years of this work group, and I inherited the co-chair from Barbara when she left the committee last session.

So Ben will tell us a little bit about the framework and the first few recommendations, please.

DR. WILFOND: I thank you, Victor.

So what I will do is do a brief overview of the report, particularly for the new members of the committee, then talk about the first three of our revised recommendations and let Victor continue after that.

So the report describes five general characteristics of genetic tests that are relevant to decisionmaking and informed consent, and we identified five major characteristics of tests that are listed on the slide here, and we wanted to present the concept that these five characteristics, either individually or in sum, fall along a complexity continuum, such that sometimes tests are very straightforward, other times they're much more complex, and the last time in February, we showed you this slide here that sort of tried to give a graphic portrayal of that notion, that depending upon these five characteristics, different genetic testing situations

may be somewhat different in terms of their characteristics.

Additionally, when we think about consent, we tried to identify four specific aspects of consent that are important in trying to describe the consent process. These are not sort of the only aspects of informed consent that are important but the ones that we believed were useful in mapping to different strategies for informed consent as it relates to genetic testing and that had to do with information disclosure, assessment of comprehension, the provider input regarding test decisions and documentation, and similar to the prior slide, we had a continuum of consent approaches that, on one extreme, were a very minimal approach, and on the other side were much more extensive approach, based upon these four characteristics.

The key part to this part of the proposal is to note that there's a flexible framework that tries to correlate the complexity of the test with the extensiveness of the informed consent process, and it's really meant to answer the specific question and that specific question: is informed consent needed for genetic testing with the following answer, which is yes and no. By that, I mean, that we do think informed consent is important for genetic tests just as it's important for all sorts of clinical tests and clinical interventions, but that the nature of consent will vary on the circumstances of the situation itself.

This is another graphic slide again to show you how in a very general sense test characteristics will inform the informed consent approaches, such that a simple test will have a more minimal approach and a more complex test would have a more extensive approach. Complex in this regard does not just refer to information complexity but is a very broad sense of complexity that takes into account a wider range of issues than just information.

In February, the entire committee endorsed the importance of trying to develop guidelines for informed consent and endorsed the report's conceptual framework that I've just described to you but had several caveats. First, there was a very clear message that individual patient needs are paramount and must be decisive in determining the approach to consent, and that there's also some concern that the focus on high-complexity tests could

trivialize the consent for other tests.

After the meeting last February, the working group held another conference call. We discussed the suggestions of the committee and have made a new attempt at revising our recommendations based upon that reassessment and these concerns, and so what I will do is I'm going to start off presenting the first of three of our 10 recommendations and Victor will present the last seven.

For each recommendation, what I'll do is present sort of the basis for the recommendation, then the recommendation itself, which is in your -- is it Tab 5? Is that the number?

So the first recommendation just regards the encouragement of professional societies to actually just in general to use this approach, and the idea is that since there really is no current national guidance for informed consent for the use of genetic tests in clinical and public health practice, and we do anticipate that the availability of these tests will be increasing in the future, and that because informed consent is necessary but the approach will differ, that it's going to be important to have a systematic approach to thinking about informed consent.

So our first recommendation is that the framework outlined in the report should be used by policymakers, professional groups and third-party payers in establishing guidance for specific genetic tests. Each of the test characteristics must be evaluated in the context of the individual patient and his or her needs.

So the second recommendation regards the idea of a conference that you'll be hearing about a little bit later on after our formal presentation of the recommendations by Michele Puryear, but that given that there has been insufficient discussion of informed consent for clinical genetic tests in clinical and public health practice and that there is a need for a broader discussion about these issues, that the relevant DHHS agencies should convene a conference involving a broad spectrum of interests and parties to foster national dialogue on the types of informed consent processes currently in practice and to explore the methods that would be used in the consent process for different types of tests using clinical and public health

settings and that this could be a forum for gathering public perspectives on the SACGT report. Again, we'll be hearing from Michele Puryear of HRSA about their efforts in taking the lead in trying to convene this conference a little bit later today.

The last recommendation that I'll present, our third recommendation, has to do with the role of professional organizations in addressing consent issues, and professional organizations do have a critical role in developing recommendations, guidance and standards for things germane to their professional groups and therefore they can have a central role in improving the quality of informed consent, thereby promoting appropriate use of genetic tests.

So our third recommendation is that professional societies, in consultation with consumer organizations, should review their current guidelines for the informed consent process for specific genetic tests or test categories that are central to their field and educate providers about the responsibilities to assure the quality of the decisionmaking and consent process.

Additionally, the groups are encouraged to employ the considerations set forth in the report about the key characteristics of genetic tests that are relevant to the consent process and, if necessary, revise our current informed consent guidelines.

In some instances, it will be important for multiple societies with a mutual interest in a particular test to collaborate with the development of informed consent guidance for that test and that, finally, priority should be given to tests that warrant an extensive consent process.

This will become more important as we go into the recommendations regarding the FDA, but this should be the case both for tests that the FDA designates as requiring informed consent process as well as for those tests that are used for other purposes, but for which they are approved, but may also need a more extensive consent process because they're going to be used for different purposes.

So again, realizing that the task of professional societies will be challenging, we thought it would be best for them to focus on those tests where informed

consent process would be necessary.

With that, let me turn it over to Victor and let you finish up.

DR. PENCHASZADEH: So we continue this presentation, and the group also felt that there was a role for a number of agencies to support the work and complement the work of professional societies. In particular, the different HHS agencies should have a role and that funding would be needed for this purpose, not only to implement and supplement the work of professional societies, but also to support social science research on the consent process in general, which leads then to Recommendation Number 4, which is that DHHS agencies, such as HRSA, AHRQ, CDC, and NIH, should establish a program to support social science research to understand and improve the consent process and that the agencies fund also the development of informed consent guidance for specific genetic tests and develop companion information resources to enhance the decisionmaking and informed consent processes.

Where appropriate, these agencies also should encourage and support collaboration among professional societies and consumer organizations in the development of guidance for particular tests. As you see, you know, we are looking for multiple input here from professional societies, consumer organizations and federal agencies.

We also thought that since appropriate disclosure information is an essential part of the informed consent process and that patients and providers must have access to key information about the test, we, in line with what we discussed and the committee endorsed at the last session, thought that FDA should require developers of genetic tests to prepare and submit information about key features of a genetic test in a form that can be used by patients or consumers to facilitate their decisionmaking about genetic testing.

The type of information that would be required would include the purpose of the test, its intended use, the analytical validity, clinical validity, clinical utility, and the risks and benefits of the test results. After ensuring the accuracy, completeness, appropriateness and readability of this information through consultation with a broad-based panel of relevant experts, FDA should take steps in collaboration with the AHRQ, CDC,

HRSA, and the NIH, to make such information readily available to health professionals and patients.

Then we discussed the need for or the importance of consumer's prior knowledge and understanding of the test to determine effectiveness of the informed consent process, and we took notice of deceptive promotional and educational appeals that could undermine informed decisionmaking which leads to the next recommendation, which is that the FDA, in collaboration with the Federal Trade Commission, should screen and monitor advertisements for genetic tests, especially those marketed directly to consumers, and the FDA should pay particular attention to direct-to-consumer advertisements for genetic tests requiring an extensive decisionmaking and consent process.

Now, the extensive decisionmaking and consent process, as you see, is something that we consider of critical importance for those tests that pose the greatest risk to patients. When providers elect to do complex tests that need extensive consent, that can help assure the test's safe administration. We thought that there was a limited role for FDA in assuring that this indeed takes place, that there is appropriate consent for the high-risk genetic tests. The role of FDA that we recommend would not interfere with or serve as monitor of provider-patient relationships.

Furthermore, only a very small proportion of all genetic tests would fall into this category, and these are the ones that are defined as high-risk on a number of the characteristics and that would require extensive informed consent.

Furthermore, FDA has experience in categorizing products according to risk, and this occurs not only for devices but also for products and so on, and FDA involvement would increase the likelihood then that the informed consent process used is the appropriate one.

Therefore, our Recommendation Number 7 is that FDA, as part of its premarket review of genetic tests, should identify those that require an extensive decisionmaking and consent process to assure their safe administration and in consultation with

a broad-based group of relevant experts develop an appropriate requirement about the consent process that would be included in the label for that test.

In determining where an extensive decisionmaking and informed consent process is warranted, FDA should employ the considerations set forth in this report about the key characteristics of genetic tests that are relevant to consent.

We also discussed the role of laboratories and we know that laboratories can play an important role in assuring the appropriate informed consent process. They can require confirmation, for instance, that an extensive informed consent process has occurred, if and when appropriate, and that which would help integrate informed consent into the testing process, which leads to – well, we took notice of some practices, particularly in Massachusetts and New York, where some variances of that actually are occurring, which leads to the Recommendation Number 8, which is that CDC and CMS should augment CLIA regulations to require that the laboratory receive verification from the provider that a consent process has taken place and that informed consent has been provided before a laboratory conducts a genetic test determined by FDA to warrant an extensive decisionmaking and informed consent process. One way could be a check-off box on the laboratory requisition which could serve as a documentation that the consent process has occurred.

The need to tailor all of these processes to the individual patient-provider relationship is borne out in these two recommendations, and the basis for this is that health care providers and educators should be responsible for ensuring that the decisionmaking process is indeed tailored to the patient and the particular test, that reasonable reimbursement will help encourage providers to provide the appropriate level of informed consent for genetic testing, leading then to the Recommendation Number 9, that public and private health plans and public programs involved in the provision of health care services should take any necessary steps to ensure that individuals have access to a health professional with appropriate training in genetic education and counseling, especially when an extensive informed consent and decisionmaking process is warranted, and that public and private health plans should cover the cost of providing

informed consent associated with genetic testing and ensure that reimbursement rates accurately reflect the time and intensity of effort that can be involved in providing such informed consent.

In summary, our key revised recommendations include promoting and relying on the development of professional guidance, a role for HHS to support social science research and improved consent and the development of guidance for specific genetic tests, for FDA to require and disseminate test information to help facilitate enhanced consent and to identify the highest-complexity tests requiring extensive informed consent. CLIA should require laboratory verification of consent for highest-complexity tests.

Then we come to proposed next steps, which is to refine the content of the report to incorporate justifications for the accepted recommendations, to develop a points to consider based on the framework, and as an appendix to the report gather public comments and participate in a conference that was part of one of the recommendations and that we will hear after the discussion of this presentation later.

That's all.

DR. McCABE: Any questions or comments for Victor or Ben? Yes, Elliott?

MR. HILLBACK: Well, I still have some concerns. I believe there's a lot of these points that make a lot of sense. There's a lot of these things that really should be done, but I think there's a fundamental concern that I still have, and that is that we're trying to ask FDA to review an iterative, rapidly changing database.

If I understand correctly, this would be the only place in all of medicine where FDA would give an opinion about what needs in high level of informed consent, and the problem I have is that even once they've done that, there's no reach-through to the person that actually is giving the informed consent. So we're putting in relatively costly what I think would be a cumbersome process for the labs to jump through, so that we can hit the doctor with a 2x4 to say you ought to do an informed consent and, oh, by the way, would you please just check

off this box when you get done and tell us you did it, but we have no other way of making sure you did.

To me, it's a lot of work, it's a lot of cost, and it's another way to slow everything down where you don't know that you're going to get the impact anyway.

I think if we step back a step and say, you know, if you go back to Point 5, Recommendation 5, that says FDA should require, I think if you change that to say the laboratories must -- I mean, we need to put this data together. No one argues about that. The basic information has to be available, so that informed consent can be done, but I think to try to get FDA to get in the middle of that and then to opine on which ones are complex, I hope that, you know, all our work on education is going to start to yield some results, so that the physician can figure that out.

And then, the concern I have at the other end is making the labs the policemen in all this. I would ask what happens if the box isn't checked? Many of my lab directors sent me emails saying what do we do, not do the test on the sample and let the sample basically waste? We certainly wouldn't want to do that on an amnio where we put a fetus at risk to get a sample and now we're going to let the sample go while we're waiting to find a doctor so he can check a box off.

So I think we're adding a lot of what I would consider feel-goods that I don't think have any real impact on what we really want, which is we want physicians, primary care practitioners to do good informed consents, and we're again going back to let's manage the labs. We have to remember that every lab test is signed out by an M.D. or a Ph.D. who's board-certified, and when they sign that out, they are saying this is the test and this is what it can do and what it can't do, and I think we have to remember that and treat them with the same level of respect that we treat the M.D. in terms of making a decision.

So I think a lot of these are workable. I think five or six of them are workable exactly as they are, a couple of them are workable with minor changes, and I think a couple of them are very difficult to make work.

DR. McCABE: I have Wylie, Judy, Steve, and Pat.

DR. BURKE: I agree completely with Elliott's points. I'll actually be a little bit more specific, and I actually think most of these recommendations have problems, although some of them are wonderful and the framework is wonderful. So I want to wholeheartedly endorse the framework. I think it's extremely helpful, and it provides a framework in which to think about issues that we've been discussing all day.

Let me go through the recommendations I think are very problematic and then those that I think need some tweaking. I agree that Recommendations 5, 7 and 8 are very problematic. I don't think FDA should have responsibility for developing authoritative information sources. I do think the lab has a responsibility to provide some information, but I think authoritative information sources are a critical resource. I think HHS should support the development and maintenance of such resources. We in fact already have examples -- for example, GeneTests/GeneClinics, which we cite a lot, the HUGENet resource, the CancerNet resource -- and I think that recommendation should be very strongly revised to be a recommendation that HHS should continue and enhance its support of authoritative sources for information about genetic tests.

I can't support Recommendation Number 7. I think it's FDA dictating clinical practice. I think it's inconsistent with the first and third recommendations that say that professional organizations should make recommendations or develop guidelines, and I share Elliott's skepticism about labs being police persons.

The other recommendations that I want to comment on are Recommendation Number 3, the development of guidelines. I want to strongly endorse what I think is the intent of the recommendation, but I think the word "collaboration" should come up front and first. I think one of the problems when you look at guidelines that we have in this country, one of the problems that we run into is that different specialty organizations make their own guidelines. One is often inconsistent with another, and it creates unnecessary contention.

I think what we really want to do is strongly encourage professional

organizations to develop processes that get everybody that should be there around the table and back away from professional organization-branding of a guideline and really move toward a strong push for consensus and getting multiple organizations involved.

As far as Recommendations 9 and 10 are concerned, it seems to me they're very interactive with a long discussion we had with the Access Group, and it would be hard for us to make those recommendations without getting the data about the value of counseling which I think is largely the value of an informed consent and educational process. So I think we need to back off from those recommendations and really have this process endorse the same recommendation that we just made for gathering data.

I'll just end by saying with Recommendation Number 6, which has to do with FDA screening of ads, I need more information. Is this something that FDA considers within its purview?

DR. McCABE: Do you want to respond to that?

DR. GUTMAN: Sure. We would view it within our purview, and we would work with the FTC and we frankly do that now and the issue is one of resources. We're strapped, and then you go after stuff that's outrageous, you have to prioritize it, and there's plenty to choose from.

DR. McCABE: Judy?

DR. LEWIS: I just want to speak to the fact that I think this issue and the issue of it being informed is a real critical issue, and to me, there's the process and also the documentation which I don't always see as connected, and I see it as sometimes two very separate issues. One is signing a piece of paper, the other is the translation of information, and what I'd like to suggest is as we start to look at what it takes to be informed, we think we know because we're health care professionals, but, I mean, I think the only person who can decide whether they've got enough information to make an informed decision is the person who's making the decision, not necessarily the health care professional.

So I just want to make sure. I mean, you've got in here some consumer

groups, but I think it's broader than the consumer groups because I think basically the consumer groups right now, if you've got a kid with a condition, you're darn well educated on that condition, and you may well know as much if not more than the health care professional because you live with it every day, and if you're a health care professional that sees that kind of a condition once every 10 years, you know, you don't know. You don't have the same lived experience as to what it means to live with a kid with that problem.

But as we're going beyond some of the rare conditions, I think that getting non-affected consumers involved becomes even more important because those are the people who are going to be the new consumers of genetic education and they're not informed in the same way that somebody who's living with a condition and has spent their life on the Internet and is incredibly well educated.

So I think I'd want to make sure as you start to look at what it takes to really be informed to make a decision, that we involve the public very broadly defined because I think those are the people who are going to go home and live with the results of whatever it is that we do in our efforts to do good.

DR. McCABE: Steve, did you have something else you wanted to contribute?

DR. GUTMAN: Yes. The center, not just this review group that I work with, but the center has embarked on a strategic plan that is focused on a concept of total product life cycle and a component of that is knowledge management, and I have the particular view that not necessarily for genetics tests, since the whole issue of genetics tests is up for grabs, but for tests in general, we at the agency in general and perhaps we in the division in particular have not done as good a job as we could about making what we know about tests transparent and about making our process transparent and about making information about tests public, so that in fact your mother-in-law, who might have an interest in surfing the Net, can find out about a particular test and we can encourage consumers to ask more and different questions. That will probably drive some of the real docs in the room crazy, but, of course,

that's okay by me because I don't actually see patients.

I am a patient, and I like to drive my doc crazy.

So I find the whole intellectual notion, whether it ended up with FDA or for that matter we could so contract out to the Post Office for all I care, the idea of having information about this stuff, whether it's home-brew or home-brew/home-brew or FDA-reviewed tests or anything, having it public is a very sound and underutilized idea, and it's a direction we should push and shove and fight and bite and do everything we can to promote.

In terms of the informed consent piece and because I had a mole on it -- Dr. Michaud was our representative on it – we never try and lead any kind of advisory group or panel. We ask for honest intellectual responses, and we don't worry about whether they're legal or not or whether we can actually implement them or not. There's a certain truth to that even with this group, although it was a truth that I would not maybe have predicted the story. We still can't predict the story. To push us towards informed consent is admirable and as reassuring as that might be certainly set us in a new tradition.

Cynthia pointed out that there is a precedent recently in drugs for restrictions, but in devices, there's very little precedent. I don't wish to suggest it's impossible, but it would certainly be a new adventure. There is not a lot of review history to bring to it. It's not clear to me whether it would require new regs or new laws or simply innovative interpretations of old regs and old laws, and again you want to prioritize what you give us. So we're more than happy to listen to the dialogue and to listen to your recommendations but you are pushing us.

DR. McCABE: Victor, did you want to respond?

DR. PENCHASZADEH: Yes. Well, let me go back to the last meeting of the committee and remind you that indeed some of the recommendations that are here now were endorsed by the committee, specifically Recommendation Number 5. That is, that developers should be required by FDA for information regarding the key features of the test that will eventually serve the basis for the informed consent process.

Now, of course, I'm aware of all the discussion that ensued afterwards with the other recommendations that the group had made regarding FDA. Now, the discussions that we had in our group on conference calls, essentially they boiled down to whether we can find a mechanism that would have teeth, not only wishful thinking or good wishes regarding the practice of informed consent with tests that we think, and we are kind of in agreement that there are some tests based on that framework that would require extensive informed consent.

So the problem we face is with what are those teeth, because we do agree, of course, that I think our practice should be FDA is not the proper agency to regulate or to mingle into or to dictate the clinical practice. We do not think, however, that with this set of recommendations that we were actually infringing the actual practice of medicine. We are talking here about the role of professional organizations. I do certainly agree with Judy that we should talk about the public in general, not only the consumer organizations, but we have to talk about the public, talk about the professional organizations. We are talking about the labs, and everyone will have to have some responsibility.

We don't want to throw the ball entirely to any particular stakeholder here nor the lab nor the clinicians nor the public and so on, but we felt that any of these recommendations would have to be able to be enforced somehow, and that is, we don't think that we can just leave it to someone that has any kind of degree and for the very fact of having a degree. You can have a Ph.D., you can have an M.D. That per se is not something that will per se ensure safety or the proper use of informed consent, and there is plenty of data regarding that.

So if we decided to put our hands in the hot boil of the FDA, it's because until now, it appeared to us that it was the only agency that could carry out that task. Now, if, because of feeling of infringement of clinical practice or because of the review that is going on through FDA, they decide that that is not the task for them and so on and so forth, we'll have to find some other means in the same way that we were discussing earlier this morning regarding, you know, all the other recommendations of the committee regarding the safety, the safe and

effective use of tests. If FDA is not the right agency for that, well, we'll have to figure out somehow.

Now, I'm very interested then in hearing suggestions as to alternative ways of an enforceable avenue to ensure proper informed consent for tests that do require it.

DR. McCABE: Pat?

DR. CHARACHE: I wonder if I can share with the group the discussions on exactly this point of the Genetic Working Group that worked for a couple of years and reported to CLIAC which then passed on recommendations.

Now, the outflow track of that is being written into regulations now, so I have no idea what they'll look like after public comment and what have you, but I can tell you that this was an issue that was very sensitive not only for the Genetic Working Group but also for CLIAC as a whole which had on it several practitioners, including a general physician, who were very concerned about how this would all work as well as laboratory directors.

Bottom line is that it was decided that there should be a certain number of tests which required informed consent because they were of such complexity, that there was no other way of ensuring that the test would be appropriately ordered or appropriately interpreted by the average clinician, and as Ben pointed out, it was thought that the number of such events would be extremely low. It was likely to be just very rare diseases and a small number of them at that. So it was felt that it would not be burdensome because it wasn't every test, it was a small number of tests.

There was discussion about who should decide which those tests were. It was the consensus of that body and of CLIAC that it was probably not appropriate to ask the FDA to be the group that made those decisions but that rather it should be professional organizations who would advise the FDA, and we didn't go into exactly how that deemed status type of structure would be set up, but it was felt they could do it more effectively with better knowledge base and advise the FDA.

It was suggested that the FDA require a sponsor to provide information of

what they thought should happen as a guide for their perspective, for their particular test, but that the FDA was not required to consider that as the final answer unless they chose to do so and not referred on to this professional group. So putting it together, it was thought that it would not be a burdensome thing to make that decision.

In terms of carrying it out, we did address the questions that Elliott has put forward, such as what do you do if the box isn't checked, and the recommendation was that you stabilize the test, so that there's no loss of information. You do whatever's necessary for that particular test to be sure that the information would not be lost. Sometimes it would mean to free something, sometimes it would mean to stabilize a cell line that you're putting something in for cytogenetics or whatever, but that the test would be stabilized so there was no loss.

It was pointed out that one could assist the ordering physician in very simple ways to know what tests required this. Simply with the design of the requisition, you could put the things that needed the consent in one part in a great big box bolded or have a very simple way of flagging this small group that would need such an ordering practice.

So that was the bottom line of the decision, that it should be set up in a way in which it was easy to have those decisions made, stabilize the specimens, and for those physicians who didn't check the box know that the physician who hadn't checked it wouldn't do it twice, so that your number of people who failed to check it would go down and that it wasn't modifying the practice of medicine because a physician could decide what they wanted to do. We didn't say it had to be signed. They could say to the patient, "I want to send this off. Is that all right with you?" and check the box. I mean, that's the responsibility of the clinician, and all the laboratory was doing was being the only mechanism there was to monitoring the fact that this had been thought through.

Now, knowing that this was done then could be a review as CLIA or as -- yes, CLIA reviews, whether it's CAP or whoever it is who reviews that laboratory. So that was the outflow track of that group, not to burden the FDA with making those decisions, have it made by those who could it well, and we thought this was a very reasonable recommendation at

that time.

DR. PENCHASZADEH: If I may follow up on that. So essentially, if I understood what you said, Pat, regarding the labs, it's pretty much what we are suggesting here, right?

DR. CHARACHE: Yes, it is, although there is one other thing I will add that we have not considered, and that is the very high percentage of such tests that don't go through the FDA at all. These are laboratory-developed and if we want to bring that in, that's not in this at all.

DR. PENCHASZADEH: Okay, and that's regarding that. Regarding the role of the FDA, those recommendations of the Genetics Working Group, essentially what do they have less than what we have here? Instead of the FDA in conjunction with -- because we say here in conjunction with the professional agenda, essentially in the recommendations of that group is that the responsibility will be that of the professional organizations of setting up the level?

DR. CHARACHE: That was the recommendation, that the FDA be able to offload that to the professional groups who could then provide the data, and again it may well be that a lot of this could just be triaged below that, but it was thought that the number that would require this level of stringency would be very low when it comes out the outflow.

DR. McCABE: I have a question of clarification before we leave that, and that is, were you saying that the home-brews are left out of the CLIA?

DR. CHARACHE: The home-brews will not be left out of the CLIA, if the FDA's proposal for a template to be followed is effected. If there is no such policy that goes forward and we're going to hear further about this, then we're up the creek with that.

DR. McCABE: But isn't CLIA independent of FDA?

DR. CHARACHE: CLIA is independent of FDA, but they need the guidelines to follow in order to know what to look for.

DR. McCABE: Judy, did you have something to follow up on that?

MS. YOST: I just want to clarify that as far as CLIA, it covers any test, whether it's FDA-approved or not. The requirements are just different, depending on whether it is FDA-approved or not.

DR. McCABE: But I think what Pat is saying is that you need the template to pull the trigger.

MS. YOST: The template is the clinical validity piece that FDA does which is separate from CLIA, yes, but CLIA does address non-FDA-approved tests.

DR. McCABE: So CLIA could then include these tests. CLIA could identify which tests required this high level of informed consent?

DR. CHARACHE: I don't think CLIA is currently structured to do that.

MS. YOST: No.

DR. McCABE: That was what I was trying to get.

MS. YOST: No.

DR. McCABE: Elliott?

MR. HILLBACK: I guess there are a couple things. It sometimes feels to me, and I'm glad some days I'm not a lab director, because we're looking here at putting in a whole series of checks and balances for tests that are signed out by a board-certified lab director who's an M.D. or a Ph.D. that basically say you've got to prove to us all this, you've got to give it all to us, you've got to et cetera, and then we're asking this practicing physician who's going to use this data to check a box which no one's going to ask him did you really do what you said you were going to do, you just checked the box and sent the paper on.

It seems a little out of balance to me, but I think it goes back to where Wylie was awhile before. There is a recommendation that the professional societies need to think through what informed consent ought to be like. I think the other thing they ought to think through is how do they both train and then ensure that their members are doing that, so that the whole system becomes stronger, that the informed consent really is being done at the level that it needs to be done and not just a box check-off on a form to make sure that the lab will test my

sample.

It just seems to me to be a very different rate of balance. I do think that it is the responsibility of the lab and it always will be to provide enough information so that informed consent can be done, and Steve and I were kibitzing here earlier about how do you put it in a repository somewhere that doesn't require the debilitating review process that I always think is very time-constraining and will delay tests and will always be out of date, but how can you push the labs to put all this data in a place that's accessible to everybody, so it's visible, without subjecting it to sort of a long-term review by an organization that doesn't have the resources to review it all?

So there may be some ways to think out of the box where that might work, but I don't think the labs are trying to get away from their responsibility to make the data available or information available, it's not all data, but what we're trying to avoid is the complexity of multiple levels of review of that data which is just back to time, money and keeping things from the public.

DR. McCABE: Cindy?

MS. BERRY: Two points. One is it seems to me, as a lay person, though, so it's subject to your review and disagreement, but it seems that many or maybe most genetic tests don't touch on any more sensitive information than certain blood tests for very deadly diseases or other conditions, and so if we're not going to impose such a heightened level of informed consent for all of those, then it doesn't seem too necessary to me to do it in the genetic arena, except for the very special category that Pat was talking about and I can't identify what those are, but you all know better than I, and I would favor an approach like that so as to avoid adding undue burdens on the system and making sure that people get the access to the services that they need so that you do have a certain category, relatively small group, that triggers some of these more detailed recommendations and the rest are really left up to the discretion of the practitioner.

The second point, at the risk of provoking a Shakespearean revolt, and I

don't know that it needs to be in the recommendations formally, but we might add in something about medical-legal aspects of this because informed consent comes into play in lawsuits unfortunately, and you either involve them on the back end which is not so good or on the front end but probably the legal profession should be included among the collaborative entities in trying to figure out what would be the best practice or models for informed consent.

DR. McCABE: An example would be Huntington's disease, and we had learned of a case recently where a sample was just sent by a physician with no discussion with the patient and the results were brought back to the patient when that individual had never wanted to know the information in the first part but was told what the information was.

Elliott?

MR. HILLBACK: My reaction to that is whose problem is that? It's the physician's problem.

DR. McCABE: Well, ultimately it's the patient's.

MR. HILLBACK: There's nothing we're doing here today that's going to change that.

DR. McCABE: Well, it's the patient's problem.

MR. HILLBACK: It's the patient's problem, but it's the physician's error, and none of these proposals would have any impact on that. If a physician today doesn't know that Huntington's disease is a difficult to deal with disease, you know, where are we?

But you know what? I guess the next step I'd go is FDA, for example, doesn't give any advice to physicians on laser surgery devices and what informed consent to give to patients there, and so we're starting to create exceptionalism again which I don't think matters, and to me, and I said this on the phone at the last meeting, to me or to my mom or to my kids, the genetic tests that I'm having or they're having is just as important as a Huntington test to someone else, and so why do we want the physician to start differentiating? I want to know everything that there is to know, and I want to know all there is, and the physician can understand if teaching me all there is is five minutes or 15 minutes, but I think that's what we're

counting on the physicians to be able to do, and it bothers the heck out of me that we're going to create all these new rules that are really stifling, rather than get to the heart of the issue.

DR. McCABE: Wylie?

DR. BURKE: Yes, I would agree. I think the strongest reaction I have as I hear this discussion go forward is that I don't think the regulatory model, the what's enforceable, is really the right model here. The group has created a wonderful framework for thinking about a range of informed consent, and I think that's a major contribution to a process that needs to go forward, that is basically a process of developing standards of practice around what is acceptable in informed consent for different genetic tests, and I think that's why Recommendations 1, 2, and 4, professional organizations, Recommendation 3 with the collaboration put in, the meetings, the social science research to better accumulate data, so that we know better what different kinds of informed consent, all of those are extremely important.

Getting back to Victor's comments about why FDA was given "the teeth" for Recommendation Number 5, which is the creating of information sources, I think it's very important to separate out two separate pieces. One is what it is reasonable to expect from a test offeror, and I think what it is reasonable to expect from a test offeror is that the test offeror provides complete information about the test.

We've spent a lot of work previously developing a template and whatever happens with the premarket review process, I think the template stands as a model of what the lab offeror should provide, and I think that information should be made publicly available, but I don't think that we want to make it the responsibility of the lab to determine appropriate test use. I think the test offeror has an opinion about that and that should be respected, but it's professional organizations working collaboratively, making sure that they've got consumer input around the table. I think that's the way good practice guidelines get created.

So what I would see as HHS' responsibility is to make sure we've got the publicly available data and that involves resources. I mean, the requirement could be that if a test is commercially available, there's a public Website that has information, whatever there is

available. That public Website would include the test offeror's template but hopefully it would also include good quality guidelines developed by good collaborative process that involves the public and professional organizations.

I think there's one other responsibility that it's reasonable for us to ask of the lab and it would come under premarket review if that occurred but it ought to occur in any case, and that is I think we should strongly encourage labs to develop the kind of test result reporting that provides very clear guidance to practitioners about what a positive test result means and what a negative test result means. I think the provider needs to be reminded about possibilities for false-positives and false-negatives, and I think the provider needs to be reminded about patient characteristics that change the prior probability and therefore the test interpretation, that the test might look different if there's a positive family history versus a negative family history.

So I think labs should provide that template-based information about their test and they should provide very good test results, and I think the rest is up to a larger community in terms of good guidelines.

DR. McCABE: Wylie, who would maintain that database on the Internet? Would it be a federal agency? Would it be something analogous to the U.S. Pharmacopeia? A free-standing organization?

DR. BURKE: You know, the fundamental question is who's going to fund it. The easy answer would be it should be publicly funded. I mean, I could certainly argue that I think it would be good use of public funds, but there has to be a funding mechanism that funnels the appropriate resources into a body that everyone agrees is an authoritative and objective independent source of information, and it might be reasonable to think about a model in which test creators contribute to the funds, that it's not solely publicly supported, that professional organizations contribute, labs contribute and the public contributes, but I don't know. I mean, it's going to be easier to describe the product than it is to figure out how it gets funded, and so it's a complicated discussion that should go forward.

DR. McCABE: Bob?

DR. BAUMILLER: You know, this is a little weird in a sense because all tests demand informed consent and to suddenly say this is a different category of being entirely is -- really what you're doing is saying we need to define the standards by which certain tests ought to be run and we're going to devise these for the practicing doctor and that's kind of odd as well.

You restrict tests. You don't do it on any other level. You trust that what the doctor orders is in your best interests and his judgment the best thing for you, and it may or may not be, but that's the agreement and what you believe is happening, and there'll be an alteration, et cetera. The difficulty here is that the alternatives are much different than the alternatives generally are with particular disease situation and illness situation, and I'm wondering if just the inclusion of a warning that the results of this test are best interpreted by someone knowledgeable or an expert in genetics would be enough to alert the physician who just thinks that this is the next thing to do, that this is more complicated than he might or she might believe, without going through all these recommendations that try to teach at the bedside in a sense the involved physician, and it's just not going to happen that way effectively, I don't think.

DR. McCABE: Pat?

DR. CHARACHE: I think the challenge that we face is more global than genetic diseases, but I use genetic diseases as the hallmark to point out the issues, and that is that the advancements in laboratory diagnostics have gone well beyond the knowledge base of the users of laboratory testing, and what makes a real problem here is that there are few physicians who realize it.

The reason for that deficit is because most of what physicians order are numeric tests. They're metabolic panels and hematology and so on, in which you get back a numeric response and they know what to do with the numeric response that has normal ranges. But the disciplines in which you can predict a failure to understand what to do with the information include genetics, microbiology, coagulation studies, and a few others, such as

hepatitis testing and so on, quantitative testing for viral burden and so on.

There's a large number of tests in which the physicians don't know what to do with the answers, and in fact, we degrade the responses. We don't tell them numbers, if we know they're going to worry somebody about a small increment change when the small increment change has no meaning. So we'll just say whether it's reactive or non-reactive or degrade the data, so they can't get lost.

Now, I think it's in that setting that we have recognized this dichotomy in which Congress in their rules and so on and HCFA, CMS, have assumed that the clinician who orders the test is either going to know how to use it or is going to look it up on the Internet, and in fact, we've already decried the fact that he has 15 minutes to work up his patient and they don't.

Now, I personally have encountered situations where I had to explain results to somebody who should not have ordered the test in the first place, and I'm sure there are plenty of other people here who can because I get those calls.

We also in our institution screen people who request genetic tests that are very expensive and complicated and are sent out and at least 20 percent or more are tests that should not have been ordered. Now that's in an academic setting. So that, I think the example of the Huntington's diagnosis is unfortunately a very real problem. Physicians also sometimes just give in to a patient who wants to get screened for breast cancer or whatever, rather than knowing that they should be instructing the patient not to get it.

So I think there's a misconception that the physician who orders the test always knows what he's ordering, and I will point out that it is the responsibility of the laboratory director to help guide, but you can't guide if you just sign out the results and you don't know anything about the specimen that came in. All you're doing is saying does this result look right or not. So that's why they call it laboratory medicine because that's what it is, but I do think that we have to recognize the current state of knowledge of genetic tests which we've been talking about yesterday and today, and there is a deficit there that we were

stretching to figure out the best way to address.

DR. McCABE: Judy, and I would ask the members of the committee, we need to focus on helping Victor and Ben on how to proceed.

DR. LEWIS: You know, I think what we're trying to do is regulate something for which regulation is not necessarily the appropriate approach. As I was listening to Pat talk, I was thinking about the patient who goes to see their health care professional, be it physician or nurse, and doesn't feel satisfied if they don't leave with a prescription, and if you don't get pills, you don't think it was a worthwhile visit, and so that we've got a lot of patient-induced demand, and yet we're not setting up regulations that say unless you've got a Gram-positive whatever, you can't have an antibiotic, and as a result, we've created a whole bunch of bacteria that are now drug-resistant.

But the way to deal with that isn't necessarily by saying you can't prescribe antibiotics unless you've gone through an informed consent process. The process is educating both consumers and providers as to the danger of abuse of antibiotics, and so I think what you've got is you've got a process, and genetic tests are just another example, and we tend to think they're special and different, but I'm not sure they're any different than any other therapy or any other diagnostic procedure, other than the difference to me is the fact that more than one person is involved, that you can really do a lot of damage to a whole family, rather than just one individual, so that the damage control piece may be a little bigger.

But other than that, I think the principles are the same, and I think what we need to be doing is rather than going with a regulatory approach, looking at an educative approach, and I think some of your recommendations speak towards education, and I would like to support that we look at this as a way to educate professionals and consumers together to work in a partnership towards a goal that I think is a pretty mutual one rather than put up a bunch of regulations, and I think that any health care professional, if they don't think what they're being asked to do is good practice, could say I'm sorry, I won't do that, whether it's give out a test result or order a test or whatever, but I think that maybe what we need to be focusing

in on is education, rather than regulation, because I don't think a heavy-handed approach is necessarily the one that makes the most sense for whatever that's worth.

DR. McCABE: Ben?

DR. WILFOND: Certainly, I certainly do agree that education's important. We have I think been hearing a clearer sense from a range of places about the concerns about the regulatory approach.

I do have two specific questions I wanted to ask Wylie and Elliott to make sure I understood, two sort of peripheral parts of the arguments, because I think I buy their central concerns, and for Wylie, my question was, it sounded like to me that you actually agreed with the general tenor of Recommendation 5, that it is important to make this information available. It's just that you didn't think that the FDA was the appropriate place to do it, is that correct in terms of what I would heard you say?

DR. BURKE: Yes. I think that's correct. I think it should be a recommendation to HHS, rather than to FDA, but I think also it shouldn't be framed as a regulatory process in which the primary source of information is the lab because I actually think when we create authoritative sources, we want lab information but we want a lot more.

DR. LLOYD-PURYEAR: Can I ask a question? Because I was going to ask about that also. Then because this is very much the template idea, and I know FDA has been taken off sort of maybe the board as the gatherer of information, but how do you enforce or require the voluntary giving of that information to populate the template? Because we in fact as HHS tried to get information from several companies when we were first beginning and looking at data, looking at issues around clinical validity and utility, and except for a few companies, it was not given and not even voluntarily, and so that's the crux of the matter.

DR. BURKE: Yes, let me respond to that, and Steve may well have comments, too.

I would have no objection to a recommendation that any manufacturer of a commercially available test should provide a specified amount of information, and we've

already done the work to say what that information is. That information is the information in the template. I have no problem with that. I actually think that would be a very reasonable recommendation. Who decides whether they've done that, whether it's FDA or someone else, that may be for HHS to decide.

DR. WILFOND: Provide to whom, Wylie?

DR. BURKE: But what I was reading into your recommendation was addressing a far more important problem. In other words, if we have manufacturers fill out the template and we make sure that that template is available in a variety of modalities, it's part of the solution but it's by no means all of the solution, and it isn't generating the standard of practice.

So what I was reading into Recommendation 5 and maybe the way I'd like to see Recommendation 5 recast, perhaps what I'm saying now is there's two different pieces, one is a smaller piece that just says manufacturers of tests should be required to provide information according to what's in the template.

DR. WILFOND: To whom, Wylie?

DR. BURKE: Publicly. It should be publicly available.

The larger piece to me is that HHS should promote the development of authoritative sources of information about genetic tests which certainly include that template information but much more importantly include the information that comes out of your Recommendations 1 through 4.

DR. McCABE: So where would the muscle be in that proposal? Would it be medical-legal, that if it's done voluntarily but if in fact it's then not done in keeping with the standards that are recommended for the industry, that there is some responsibility to the laboratory for not providing that information?

I mean, the issue is if it's done – it could be done now. If it was purely voluntary, then there's no need for us, and it was working, there would be no need for us to be meeting. Obviously that isn't the case. So there has to be the question of where the muscle is.

DR. BURKE: And I think that this is where we're in limbo because, as I understood what we were trying to accomplish with the premarket review, it was mostly this step. It was mostly guaranteeing that tests would come with a certain kind of labeling that had reasonable completeness, and it wasn't, as far as I could tell, a whole lot more than that. So I still endorse the concept of premarket review, but I think we're in limbo until we hear more about whether that's really a possible thing to enforce, and I don't know that we can talk about teeth without that.

DR. McCABE: Well, I think that it's important that we're going down this road because let's assume that the FDA makes a decision that they will not deal with home-brews or that they continue to make no decision, either of which leaves us in the same position we are right now.

So to rely on the model we had before is probably unrealistic, at least at this point in time. So in point of fact, we can still press for the template because I think in fact the committee had evolved toward labeling being more important than regulation, and if we say that the labeling is important, then we need the template filled out. The question is where does the muscle come to get it filled out if it's not regulatory?

DR. BURKE: CLIA?

DR. McCABE: Steve, and then Elliott.

DR. GUTMAN: Yes, I think that you ought to throw that at HHS. I think it's their responsibility. If they don't wish to use FDA, maybe they could export some FDA tools. If they don't wish to use FDA tools, then they ought to talk to Judy about how far she can go with CLIA and they ought to be creative.

I absolutely agree. This shouldn't be voluntary. Maybe it should be lighthanded and maybe it should be like the IRS. You report it and only in competitive areas does the FTC or the FDA or whoever, CLIA, get involved because they are fighting and they're reporting each other. Maybe that's all you get. It ought not to be voluntary.

DR. McCABE: Elliott?

MR. HILLBACK: Yes, I think that to make sure that this phrase gets into this meeting, I think we've all said all along that a laboratory has to be able to say what we know and what we don't know at any point in time about a test.

There are, for all the nasty things that we've said about CLIA's abilities and inabilities -- I don't say them but some people do --

DR. McCABE: You only say the most positive things.

MR. HILLBACK: Of course. But I mean, there are all sorts of things that CLIA governs. CLIA governs the training of our lab techs, and yes, they're not there every day to check it, but when they show up and do an audit, if we've not done things the right way, we'll hear about it.

Now, it's not the same level of teeth as some people are looking for, but it seems to me that if the template idea and the concept of tell people what you know and what you don't know at every point in time are there and you have lab directors who are again board-certified M.D.s or Ph.D.s, who signing out the lab to that spec sheet, to that list of this is what we know about this test and what we don't know, it seems to me there are a number of built-in controls, checks, and balances that exist. They're not perfect, and some people will find ways to get around it, but it is a rapidly implementable and darn good system, not perfect but darn good, and then we can start to tune it once we get there.

I think, also, it puts the updating responsibility on the people with the data which is the laboratories to update their data and then hopefully we can marry it up with the professional society data in terms of the user end, but to me that's a much more workable solution, and yes, under CLIA, it's not going to be inspected before it goes up, but there are ways to audit, there are ways to check, there are ways to spot check that I'm sure CLIA could build in.

So to me, it's not a very big step from where we are today with CLIA and it would be a pain sometimes to implement but I think it's doable.

DR. McCABE: I'm going to let Ben comment on this, and then we've got to

come back to the issue of what direction we give to this working group.

DR. WILFOND: Well, in fact, what I want to do was ask a second question, which is my original question that I had a little while ago for Elliott before, which I think I understand what your central concern about requirements that involve regulation that may have modest effects.

The one that I was least convinced of was actually Recommendation 8 regarding the check-off box because, particularly after hearing Pat talk about it, it seemed that asking a laboratory to see that check-off box wouldn't necessarily impose a lot of burden to them but yet still would provide a fair amount of symbolic message to the physician they ought to be doing this, like they should be hearing from the professional organizations and such like that. So I'm still a little unclear from your perspective why you think that would create a burden for the laboratories to do that.

MR. HILLBACK: My reaction was sort of what was passed on to me by both our medical director and our lab directors, which was it puts them in conflict with the people we're in partnership with; i.e., that we become the police persons trying to chase them. We already chase them for some data. I mean, you can't do certain tests unless you know the gestational age of the fetus. So we have to get that data to do the test.

In this case, chasing someone for a checked box -- you know, let me give you the scenario I can envision. I'm on the phone. A customer service person or a person says we review every lab, the paperwork on every lab test, in the morning at 6:00 a.m. as it comes into the lab, so that we can see if there's something missing. So I'm the person that calls the doctor and says you didn't check the fact that you did informed consent. Oh, well, just check it, it's okay.

Now, has that gotten us anywhere? I don't think so. Has it hurt us a lot? Probably not. But I could tell you all my lab directors hate the concept because it becomes one more contentious thing that really has no value.

Now, if there's a symbolic value of having that there on our --

DR. WILFOND: Well, that's symbolic, but even the scenario you described of the phone call, I think somebody else made a comment that you would hope that after awhile those phone calls would stop. In other words, that they would remember to check off the box.

MR. HILLBACK: They don't stop on gestational age.

DR. WILFOND: Okay. Fair enough.

MR. HILLBACK: But you know, I understand where you're trying to go, which is, the doctor has to accept responsibility that they're doing this.

DR. WILFOND: Right.

MR. HILLBACK: I don't think the labs are the right police points and certainly none of my lab directors are excited about the prospect. Is it the end of the world? No, it's clearly not the end of the world. But it just puts -- we're not going to compromise the sample and even to stabilize the sample, many times you've got a time issue not with the sample but with the patient, and it's just not the way we really want to practice. So that's my position.

DR. WILFOND: I just have one more.

DR. McCABE: Sure. Ben, and then Michele, but we need to start focusing here.

DR. LLOYD-PURYEAR: Well, I am going to focus. I'm going to give a recommendation what to do.

DR. WILFOND: The one thing I wanted, we haven't discussed Recommendations 9 and 10 at all.

DR. BURKE: I did.

DR. WILFOND: You did, yes, Wylie, and I would suggest that we should probably table them based upon the conversation we had earlier today regarding genetic counseling, and I just wanted to raise that question one more time, Wylie, to you. I actually talked to Sean about this during the break before he left.

Clearly, there's a relationship between genetic counseling and informed consent. The conversation earlier today was mainly about counseling, that really we're talking

about pretest counseling which is like informed consent, but I asked Sean whether or not he thought that if the argument for informed consent was not one of empirical value but one of a moral justification, how would that fare in terms of the requirement of evidence towards value?

He thought that was an interesting question, and we got onto the issue of whether or not, for example, how the informed consent process for a kidney transplant is reimbursed because in fact that also involves a very elaborate consent process, and his comment there was, well, there's no specific reimbursement for that, but, of course, the surgeon gets paid a lot of money for doing the transplant.

So I think the difference between the transplant and the informed consent for genetic testing is that those activities are separated, so that the person who's doing the consent is not going to be reimbursed for the activity later on, but at least I got some sense that he was at least sympathetic to that notion of this being different.

DR. BURKE: I'll just respond by saying that there's a sense in which there's a mom and apple pie component that obviously I agree with and have no objection to. I think what might be reasonable is to fold them into a single recommendation, which would mostly be Recommendation 9, and I would change one element of wording.

"Public and private health plans and public programs involved in the provision of health care services should take any necessary steps to ensure," any necessary steps that's going to be dictated by standard of practice, "to ensure that individuals have access to a health professional with appropriate training," and I would take out "in genetic education and counseling," "especially when an extensive informed consent and decisionmaking process is warranted."

I think that's a reasonable general statement. I think it applies to all health care, and I think that is the moral statement that I would strongly endorse.

DR. McCABE: Michele, and then Judy.

DR. LLOYD-PURYEAR: My recommendation is, and I think you know probably, I agreed pretty much with Wylie and Elliott with their evaluation of the majority of

these, except for Number 5, recommendations that were put forth, but even given that, that I probably disagree with them. I think that they – and this sort of segues way into the conference idea. I think clearly there are people on both sides of the aisle here, and I think some of these recommendations should be let stand within the document and be heard by a wider audience, and I think that's some of what needs to be done, so that we're not just hearing from ourselves and we're encouraging a large public conversation about many of these recommendations.

DR. McCABE: Victor?

DR. PENCHASZADEH: Are you talking about public comment?

DR. LLOYD-PURYEAR: Public discussion.

DR. PENCHASZADEH: Public discussion in a conference?

DR. LLOYD-PURYEAR: Yes, because I do disagree. I don't think many of these recommendations are enforceable, but I'm willing to go with a wider audience and a different audience on the other side of the coast, too.

DR. McCABE: Judy?

DR. LEWIS: I just wanted to speak in support of what I heard Wylie say as I walked in in terms of that recommendation with one small change, that we change the word "training" to "education," because I think education is broader than training.

DR. BURKE: I'd accept that.

DR. McCABE: Is there further discussion or should we move on to the discussion of the conference and see where that takes us?

DR. BURKE: I'd like to hear some discussion of what Michele proposed, because it seems to me we have two options here. One is to recommend that some recommendations be dropped now and revise the discussion draft, which would then hopefully go for comment, and the other would be to pretty much keep the document as is, and in the spirit of Michele's suggestion, I'd be pretty open to keeping some of these in that I object to. Your point is well taken that a wider audience perhaps should respond, but I'd still think carefully about whether we want to include them all as is, and I actually think there's some

editing that might be beneficial.

DR. PENCHASZADEH: I think it is clear from the discussion that we have to revise them, because anyway, Michele, what you were proposing, if indeed this conference will occur at the end of November, I mean, it's the timing issue. I think it will be against time. So I think that we are getting a lot of feedback here. I think we can work on those. I sense what are the things that spur the reaction – and Elliott is laughing there -- and we can try to come up with the best consensus and compromise.

MR. HILLBACK: It's an iterative process.

(Laughter.)

DR. McCABE: Judy?

DR. LEWIS: Just to speak to that, I mean, I think that one of the issues that we have to pay attention to is managing expectations, and if something's going to come back that's unacceptable to us and, I mean, I don't think we should put out anything that's unacceptable to us because then we're setting up expectations in the community that we're not willing to meet.

So I think what we have to do, I mean, with all due respect, I think if there's something that we say no way to put that out for public comment, we're raising expectations that something could happen that we're not willing to support. So I do think that there's piece of managing expectations, and if there's some bottom line stuff, I would not support sending those out.

DR. McCABE: Vence?

MR. BONHAM: I'm interested in hearing about the conference and the timing of the conference, as a new member to the committee and there are three other new members.

DR. McCABE: Okay. Well, let's proceed then with discussion of the conference. It was the next item on the agenda. Michele will talk about this initiative from HRSA, about a conference on informed consent. We'll then have Sarah speak who'll discuss

SACGT's role in the conference and the forum it will provide for a broader discussion of the committee's draft report, but what I've heard is that the draft report should be redrafted or there should be consideration given to that, based on the input here.

So Michele, do you want to move forward?

DR. LLOYD-PURYEAR: Sure, and for the committee members, there's a handout called "Newborn Screening Task Force." It starts with that.

I began with, for those who aren't familiar with it, '98 through '99, HRSA asked the American Academy of Pediatrics to convene a task force on newborn screening. This ended up being sponsored by NIH, CDC, AHRQ, other health professional organizations, and the Genetic Alliance.

The task force came out with a report and the task force report looked at the issue of informed consent because not all genetic tests require informed consent. Four million infants are screened each year for genetic disorders and very few of them, very few of the parents, are offered informed consent or dissent.

Anyway, the task force report concluded, and I'm reading some of this, that the families should be educated about newborn screening and indicated that information may be provided prior to birth or following birth. They also indicated that information should be provided during the follow-up process if initial screening was positive. So their big focus was on educating and informing parents about and receiving permission for newborn screening, and they recognized that the process wasn't simple.

When they did their evaluation of state newborn screening programs, they said that the state policies regarding informed parents and parental refusal and consent varied widely from state to state. Forty-nine states had specific legislation that requires newborn screening, only three states at that time, and it's less now, have provisions for informed decisionmaking. The task force focused on the process and importance of education of and a conversation with the parent to achieve what they called shared decisionmaking. They felt that should be the end product in mind.

They then said that determining the best mechanism to inform parents and promote screening became the central issue as well as determining the content knowledge needed for the parent to participate in shared decisionmaking, and they made several recommendations about the educational process, the need for pilot studies to look at this, evaluation research to look at it, and they indicated a need for states to develop and provide educational materials for families about newborn screening. They also indicated that families should be involved in the development of those policies and procedures and educational materials.

The specific recommendations were that states should develop model -- actually, there was a recommendation to in general develop model legislation and/or regulations that articulate policies and procedures regarding utilization of unlinked and identifiable residual samples for research and public health surveillance and since any policies and procedures addressing informed consent must also address the use of and access to these residual blood spots, the development of policies and procedures must necessarily approach both arenas.

So we took the task force report and developed an implementation plan and part of that was developing a contract to specifically look at the issues of informed consent and that contract went out on the competitive basis. UCLA successfully won the contract last year, and I'm going to go through what that contract's supposed to do.

We're at the beginning of the contract right now, but the contract's supposed to examine current informed consent/dissent materials and processes for newborn genetic screening. It's supposed to develop model policies, procedures, materials for informed consent process based on that examination, and address model policies for storage and use of and access to newborn blood spot screening samples within the policies and procedures developed for informed consent.

Specifically, under the informed consent process, the tasks we've given UCLA are to analyze the ethical, legal and social issues relevant to informed consent for

newborn screening, prepare content for an educational program for parents on newborn screening, develop one or more options for an educational process for parents on newborn screening. With these options, organize and conduct pilot studies in three states and these three states are to serve as partners throughout the process, and analyze the impact of informed consent in terms of knowledge by providers and parents, response to false-positive results, participation in newborn screening policymaking by parents.

The specific tasks around residual blood spots and for those that don't know, every child when they get screened, the heel-stick blood is placed on a piece of filter paper. That's often stored and kept for varying times within state newborn screening programs.

So for residual blood spots, the tasks under that are for UCLA to define model review process for determining access and use of the stored newborn screening samples, define potential acceptable uses of these stored samples for quality assurance and research, develop standards for linked and unlinked specimens in this process, develop standards for retention for a potential national set of stored newborn screening specimens, develop a protocol for retention of the newborn screening samples in the state health department, define optimal preparation and storage methods for the tissue samples, define the data set to be linked to the samples, and prepare recommendations for maintaining privacy and confidentiality for specimens in linked databases.

Additionally, to define standards for research and quality assurance, access to tissue specimens and linked databases, to prepare materials for parent education, to recommend the informed consent content and process based on the above phase, and to prepare model legislation for implementation at the state level.

When these products are complete, the contractor's again supposed to partner with the three selected states in the project and to facilitate development and implementation of the pilot projects in the specific states.

So because it looked like the process that was going on at SACGT which was looking at the informed consent process in both the clinical setting and a public health

setting, and we were in particular looking at informed consent process within a public health setting, we thought it a logical move to combine these two efforts and volunteered to have UCLA host a national conference and to look at the SACGT recommendations and have further discussion on that day and then allow, also, to prepare for public comment on the second day, and then we would use that opportunity actually for ourselves for the newborn screening part of this conference that would -- part of the task of UCLA is to prepare a literature search and present that. So certainly the SACGT process, sort of the joint program that would go on during the first days of this conference, this effort would serve us well in educating both the advisory body for the public health and newborn screening part of this conference and our state partners.

So do you want me to go into more detail at all?

DR. McCABE: Actually, I'm turning the chair over to Wylie for this since I'm at UCLA and involved in this project.

DR. BURKE: So I think I would say at this point, maybe we should open it up to questions, but let's be really clear that what's being proposed is that because there's an existing process that includes the convening of a meeting at UCLA, it provides an opportunity. So what's being proposed is that we use that as the opportunity to pursue the conference that we've already identified as something we want to have happen and the result of that would be that a portion of the conference, as I understand it, would be --

DR. LLOYD-PURYEAR: I can go into some details right now about that.

DR. BURKE: Well, I think this is the specific question that you should clarify for us. I think you should go through the proposed sequence of events for the two days that SACGT would be involved and clarify for us that the portion of the conference about informed consent that would be joint, where SACGT would be participating, would be a larger conversation that is not just limited to newborn screening. Why don't you elaborate, and then we'll open it to questions.

DR. LLOYD-PURYEAR: Okay. The proposal is that we have the

conference on November 21st through 23rd. November 21st is a Thursday. We would begin with -- and I'm going to get real specific in here because I think you need some specificity -- a keynote speaker to lay out the issues of informed consent, many of the issues that this committee has grappled with, and have that laid out, not the specific SACGT document.

Then I think we could move at that point to some specific scenarios looking at informed consent, the informed consent process and public health, and the best example of that is its use in prenatal and newborn screening at this point and then also going on into more traditional clinical settings, having some examples of that, and then leading into the SACGT framework document, and at that point moving to time for some public comment.

In the afternoon, I think the SACGT document lends itself to four work groups where you could discuss -- and this is why I was thinking we could just leave many of the recommendations there, but if you wanted to, you could look at the clinical community, public health community, and I would also would bring in the laboratorians at this point, and we certainly have room for a fourth breakout group, but to use those groups, those professional groups, to discuss the implications within their perspective of the recommendations that were put forth by SACGT.

Then the following morning, Friday morning, again having time, and there's time set aside in the grand rounds at UCLA and/or at least what would have been the grand rounds, to have the work groups report on their discussion of the SACGT recommendations and a summary given. That's all Friday morning.

Friday afternoon would be SACGT's formal meeting. That's actually when also the newborn screening meeting would begin and all day Saturday would be newborn screening.

DR. BURKE: So you've heard a proposal. Why don't we open this for comment?

DR. McCABE: The one other thing that I would add about the grand rounds is that we have one of these audience response setups so that we could have very formal

input in terms of audience response. So we may in fact want to have the four work groups meet the afternoon before, summarize, so that we could come up with some specific questions to get feedback from the public in a fashion differently than we have done before.

DR. BURKE: And I think what's implied in your comments is that there's still some room for input on exactly how this meeting gets structured.

DR. LLOYD-PURYEAR: Oh, there's a lot of room for input.

DR. BURKE: Pat?

DR. CHARACHE: Two thoughts. First, it sounds like a marvelous opportunity to get thoughtful input in this area, and it's a wonderful subject matter because it covers the whole spectrum of stringency. I mean, obviously your need for sickle cell screening is very different from spectroscopy and when you get into the multiplexing.

My other thought, though, is that it probably would be very helpful if we could publish the document we've just been going over to get a better consensus maybe for the August meeting, so that the discussion would be more productive in November.

DR. BURKE: Vence?

MR. BONHAM: I concur that it would be valuable to let the new members of the committee and the working group really consider the document and to come back at the August meeting and finalize the recommendations that we will want to have for public comment, and I think it would be a great opportunity in November, but that would give us an opportunity to participate.

DR. BURKE: Thanks.

Joann?

DR. BOUGHMAN: I hear the differences, Michele, in the newborn screening public health generalized format versus the more private health provider-patient relationship, the dichotomy that you kind of set up.

I wonder if there is any consideration of the middle ground or group-specific testing and informed consent issues that we have grappled with here a little bit and

newborn sickle cell might kind of lean that way a little bit, but in fact that's been one of the issues, and I wonder if there's any place for us to address that group specificity anywhere in here.

DR. BURKE: What I'm hearing is that we'd like perhaps to have an opportunity to see more discussion about exactly what array of different settings and therefore informed consent challenges would be addressed, is that fair? We have five minutes.

So Judy?

DR. LEWIS: I think this is a really important topic. I've been concerned about the issue of informed consent around newborn screening or the fact that it's just sort of a standard of care, and I think that it's a really important issue, and I would welcome the opportunity to participate in this because I think it's probably the broadest use of genetic testing that we have with no informed consent at this point in time till you get test results.

DR. BURKE: So we've now had I think four comments on the basic decision which I think we need to make, which is, is this a good idea, and does the general structure sound good? So I want us to bring to closure on that and then we can talk just briefly about how we want to inform the process further, but first, are we agreed? Is there a consensus?

Bob?

DR. BAUMILLER: Just we mean that the tacit or implicit consent which we have relied on in the past is no longer valid because of the expansion of newborn screening to a broad, you know, much greater number of diseases?

DR. BURKE: My impression is that this is an open discussion, but why don't you comment on that, Ed?

DR. McCABE: Yes, there are various schools of thought on this. Newborn screening grew up in the public health model which was it's good for the public. It really came out of the vaccine, the public health, model, which did not involve informed consent. It has moved to what in most states is referred to as informed dissent.

One of the parts of this contract is that we're putting together -- we have a young M.D./J.D. who's actually a resident with us, but who is doing a law review article really looking carefully at the laws and regulations in the various states for newborn screening. So you can come at it from the autonomy issue and that is that parents have a right to know. We really refer to it as informed decisionmaking because it's less important whether it's dissent or assent as it is that it's informed.

In addition, then, because of the expansion and the rapid expansion of newborn screening, the fact that during the expansion phase, these are acquiring data and these are really pilots, at least in the early phases or, as we get additional data about efficacy, then it really is research or quasi-research, and individuals need to be informed that data are being collected and that we may not know what the outcome of a positive screening test means. So it's a complex issue that involves a variety of different axes in the decision.

DR. BURKE: And for my sake, to clarify, I think what we're saying is that in endorsing our participation in this conference, we are not signing up to any preconception about what the answers are. We're signing up to an open discussion, is that correct?

DR. McCABE: Well, the issue is that there really will be two parallel meetings going on.

DR. LLOYD-PURYEAR: The answer is yes.

DR. McCABE: Yes, yes. But that there will be an SACGT part, a HRSA contract part, and there will be some plenaries where the two come together.

DR. LLOYD-PURYEAR: No. No, and I think we need to articulate this clearly so there's consensus and agreement here.

There is a HRSA contract part on Saturday, and Friday afternoon's up for grabs what you'd call that, but Thursday all day and Friday morning are a joint conference that is sponsored by HRSA and UCLA with co-sponsorship by SACGT, CDC, and NIH, if they want to be added. Okay. Is that right?

MS. CARR: I think what we agreed might be the terminology and it's sort

of semantics, but I thought we thought we would be in association with.

DR. LLOYD-PURYEAR: In association with, yes.

MS. CARR: So that it was very clear that it was a HRSA-led initiative and to be sure that you all got the credit that you deserved for taking the initiative in developing this, and so I think it's the way --

DR. LLOYD-PURYEAR: Just keep reminding me that it's in association with.

MS. CARR: In association with, but that's language that the committee can consider, and if they want us to be full-fledged co-sponsors, that's a possibility, too, but I think we thought that would be a more appropriate way of demonstrating our role, which is not as prime sponsor per se.

DR. BURKE: Do you have any other comments you want to add about the meeting?

MS. CARR: No, only in that if the committee is in concurrence that this is a good idea, that we are prepared to devote -- in effect, the dates are the next dates of your meeting, the dates of your next meeting, and we are prepared to devote a day and a half of those two days to your participation in this conference, and then what we were thinking we would do is that we would spend half of the second day in a committee meeting at UCLA as well, and so there would be time for other committee business, but a day and a half would be devoted to helping foster this national discussion of informed consent on genetic tests used in clinical and public health settings.

DR. BURKE: Judy, and then we'll try and wrap this up.

DR. LEWIS: No, my only point was going to be that it sounded like this was preempting our meeting and was leaving us very little time for business. If that's within the plans, that's fine, but I just wanted to acknowledge that this was not in addition to but in lieu of the November meeting, because I just checked my dates and that's when we had signed up for, and it's the weekend. So we'd be traveling, and those of us who don't travel as far usually

would be traveling more the weekend before Thanksgiving, which I guess is only fair for our West Coast folks.

DR. BURKE: I guess so.

MR. HILLBACK: This is not the Wednesday before.

DR. LEWIS: Well, if it was the Wednesday before, we'd all be walking, right?

DR. BURKE: What I think I'm hearing is that there's a consensus that this is a good plan. I think I've also heard that we would like to recommend a revision of the informed consent document and rediscussion of that at the August meeting to make sure we've got a document we really are ready to put forward for public comment at this meeting.

Also, I think implicit in these comments is that we would like to have SACGT participation in the planning process. I know there's a steering committee and maybe we should just say that any members that have strong interest in participating in that should let Sarah know.

DR. LLOYD-PURYEAR: Actually, please let Sarah know very soon. Ben and Victor are on the steering committee, as is Sarah. So we wanted Wylie, too, but we wanted to keep it small, too, because this steering committee is going to be in essence the planning committee instead of duplicating things.

DR. BURKE: And whether or not people are involved in the steering committee, they are very welcome to let Sarah know.

DR. McCABE: And it's likely that people will be involved as participants, active participants who are not involved in the steering committee.

DR. LEWIS: I just thought it was important to volunteer since most of the samples are actually drawn and collected by nurses.

DR. McCABE: The other thing is that we have talked about having a meeting outside of the Baltimore-Washington metropolitan area. We've had two meetings outside the Beltway, but both over in Baltimore, and I think it is important to get perspective

from another part of the country on the activities of the SACGT.

I'll resume the chair then. Our final agenda item today is to review the public comments we received on SACGT's draft information brochure, some basic questions and answers about genetic testing. The brochure, the request for public comments and the summary of public comments are at Tab 5. The committee reviewed the draft brochure which was developed by the Informed Consent Work Group in November 2001 and approved the work group's recommendations that public comment on the content and utility of the document be gathered.

Sarah will now summarize the comments we received. After considering the comments, we will need to decide what next steps we should take. Sarah will propose a few options for our consideration.

So Sarah, if you would, proceed.

MS. CARR: Thank you.

As you recall, last November, the committee reviewed the draft information brochure for the general public, entitled "Genetic Testing: Some Basic Questions and Answers," which was produced by the Informed Consent/IRB Work Group.

I'll review the 287 public comments we received on the brochure, in addition to the summary of the comments that you have at Tab 5. We also have four copies or five copies of the full set of comments that came in and they're in these two spiral-bound volumes around the table, and there's also a set at the registration desk for the public to look at.

Now, the premise of the brochure is this: that genetic testing is expanding and that at some point in the future, many, many, many of us may face a decision about having a genetic test, and that brochure posits that prior awareness of the risks, benefits and limitations of genetic tests will be helpful to us if and when that time comes because we will be better prepared to ask some key questions.

The document was organized into a question and answer format with 12 major sections covering these items you see here which I won't walk through, but I also do want

to take a moment to review the origin and evolution of this project. The genesis of the brochure was SACGT's determination in 1999 actually that the education of health professionals and the public was an important facet of oversight of genetic tests.

In 2000, the Data Group began developing a template for information summaries for providers. The work group and the committee as a whole suggested that a similar effort should be undertaken to prepare information for patients and consumers and that the Informed Consent Work Group was the appropriate locus for that effort. That work group again drafting a document in February. Between February and November, the draft went through many iterations, and over the course of those months, its focus and target audience actually changed, shifting from a patient-centered document to one for the general public.

The central reason for this shift was the recognition that the brochure's basic content and broad scope would be more relevant to a general audience than to patients and consumers who would likely be in need of specific testing information.

In November 2001, as I said, the final draft was presented to the full committee for consideration. The committee agreed that public comments on the document should be sought on the utility of it but suggested that further work on improving the document's reading level be undertaken before the solicitation.

In January, we hired a contractor with experience in developing educational materials to review and edit the brochure. The contractor, using a scoring program called FROG, which was developed 20 years ago I'm told and is one of the three widely used readability indexes, assessed the reading level of the final draft of the revised document at between Grades 7 and 8.

We began the solicitation for public comments on March 19th and closed it off on April 22nd. The solicitation, there was a preamble and a "Dear Colleague" letter we used, but it described the brochure's purpose, goal, target audience, and it requested comments on the utility, content, readability, and strategies for dissemination, and it asked about these things in six questions and six areas.

We used three outreach mechanisms. We posted it on our Website, we used a "Dear Colleague," as I said, through both snail mail and email, a listserv that has about almost 3,000 people on it, and then we published it in the Federal Register as well. I think I saw responses actually from people who saw the brochure because it had been passed on by other listservs, including the Genetic Alliance and Save Babies Through Screening as well as the National Society of Genetic Counselors.

As I mentioned, we got a total of 287 comments which came in to us in these four different ways. We used email to me through my email address. Some people faxed it, some people sent snail mail, and other people did use the Website that we had set up.

This shows you the comments that came in by sector, and sometimes it wasn't always possible to determine what the background of the commenter was. So we have an other category and sometimes the comments came in anonymously.

I think it's notable that the health professionals submitted almost one-third of the comments, but we did have what I hope and think is a fair number of comments from patients and consumers, and unless they're in the other category, I guess we could say that we may not have reached our target audience, which was the general public.

Now, we did sort of an analysis of all the comments and categorized them in an overall sense to whether they were positive in an overall way or negative, and as you can see, the overwhelming number were positive, but there were a number of very specific changes suggested in the document in all sections of it. So I thought you might want to see actually where those negative comments came from by sector. You'll see here that most of them in terms of sheer number came from health professionals, but in terms of representatives, it seems industry had a problem with it and state government people.

Now, I'll walk through the six specific questions that we asked. First, we asked if the document was useful and most people though it was. We asked how people might use the document, and you can see that most thought – and these are sort of ranked according to how many people suggested these various uses. Patient education, and again this is despite

the target which is the general public, but it seemed to be a value for patient education, for health professions education actually, public education, and as a general information source that could be used for laboratory staff, IRBs, and general information resources.

Somebody said they would make it part of an exhibit at a science museum, and somebody mentioned policymakers might have a use for it, including staff in technology transfer areas, and then it was suggested it might be model language again for labs and clinicians in publishing their own brochures for informed consent documents and so forth.

I just thought you might want to see some of the reasons people cited for lack of utility of the document. There weren't that many, but there were some and they were important issues that were raised.

As you can see here, some people thought it was overly broad, too general, overly simplistic, too long, too much information, and some subjects weren't covered in sufficient detail. One person, I think, especially worried that it would be outdated too quickly and that it wasn't useful as a stand-alone document. Some said it wasn't different from existing brochures and wondered why we had actually even undertaken the effort to develop it.

I think many people said this won't be useful for patients asking about specific tests, but as you saw from the previous slide, people did think it was useful for patient education. So we did get very complex answers, I guess, and then the last point was that there's no clear target other than a general audience.

The second question was about content, and we asked whether people thought it was appropriate and complete. Most people thought it was appropriate. Most people thought it was complete.

We also asked about the reading level, and I think here most people thought we had hit the right reading level, but I would say a good -- well, more than half that or 50 percent of that thought we hadn't. So this I think is somewhere we might need to do more work.

These are some of the specific content problems that were mentioned, and I

won't go through all of these, but I do want to point out two. The first one about the explanation of genetic testing and the definitions. Some of the problems people identified here were that the definitions were either too broad or too narrow, that we failed to differentiate between DNA and biochemical tests. Some people thought that this was an important distinction to make and also that we hadn't distinguished between testing and screening and between predictive and presymptomatic testing.

Some said that our discussion of prenatal carrier and newborn testing wasn't adequate, and a couple people thought we ought to actually mentioned have preimplantation genetic diagnosis, and someone said that we hadn't made a good distinction between what's available now and what's coming in the future.

The other area that I want to point out is the section on similarities and differences in tests. This section where we had proposed that there were some -- and in fact, this is kind of a central thesis, I guess, of the brochure, that genetic testing, genetic tests, are very similar in very many ways to medical tests, but they do have some differences that are important for people to think about.

Then we sort of described some of those differences, and I think we emphasized the predictive nature of tests, and there was disagreement with that. People said, you know, cholesterol tests have predictive value. So this was controversial and I think in a way we weren't surprised that it was.

Question 2 asked whether there were other issues that should be addressed and here are some of them people mentioned. Some people thought we hadn't discussed the important role of genetic education and counseling, despite our lengthy discussion today about that, that our brochure hadn't described that well enough, and I think that gap or that omission may actually be because again we were targeting the general public and not someone who is on the cusp of having the test necessarily, but I think this is probably a problem that we need to emphasize, that maybe not for all genetic tests but for some it may be very important for comprehensive education and counseling to occur.

A lot of people thought we should really use examples, case studies and vignettes in the brochure, and they felt that this wouldn't take away from its general scope. Some people thought we should talk about research tests and they were surprised that we hadn't. Some thought we should discuss family history, and one person thought that it was an oversight that we hadn't talked about race and ethnicity, and then part of the document has some important resources and sources of information and some people thought we left off some important sources.

The next part of Question 2 was are there other questions that should be included, and a number were mentioned and this would be questions we are, I think, to actually include with answers in the brochure. As you can see, when should I consider genetic testing, where can I get a test, and so forth.

The third question asked about tone and whether the brochure was culturally appropriate. Most people who answered this question thought that it was appropriate, the tone, and fewer people addressed the question of its cultural appropriateness. Most thought it was, but some people made some important points about that and some were concerned that it wasn't and some thought its reading level was too high and geared toward an educated white male class audience. Some thought it was overly formal and some thought it was overdelicate, and one person thought or a couple people thought it was pedantic and condescending, actually.

On the other hand, some thought its tone was conversational and informative without being condescending, and another person thought it had a good balance of risks and benefits and an emphasis on values and personal choice in the questions and answers. So you can again see kind of a range in terms of the specific comments.

Question 4 was about translation of the brochure into other languages. We wondered whether people thought that should happen, and you can see of 287 comments, 250 thought we should, and you'll see here the languages. Spanish was mentioned the most, Chinese, including three dialects of the Chinese language, was mentioned, French, Vietnamese,

and then there were 17 other languages mentioned under nine times for a total of 70 comments. Seven people thought we should not translate it into any other language.

Question 5 was to whom and how should this brochure be disseminated, and this part of it, to whom, is a little bit overlapping with the part of Question 1 which asked how would you use this document. You can see here health professionals, and this is sort of ranked according to the number of comments. People thought health professionals and professional societies should receive it, specialty clinics, patients and patient-consumer organizations, public health facilities, clinics, hospitals, labs, the organizations listed in the brochure, and then some people said yes, we should get it to the lay public, all the lay public. One person said every taxpayer should have it. It should be in every doctor's office, every genetic counselor should have it.

In terms of how it should be disseminated, the most popular answer was the Website, through Internet Websites, ours, NIH's, HHS', and the organizations mentioned in the document, but people did feel, I think, almost as many thought we should definitely have a printed version of it in order to enhance its access to a wide audience. One person thought we ought to get on TV and start promoting it and that we ought to advertise it in Parade magazine and Reader's Digest. We ought to post it in airports, bus terminals, and subways, and we should use the approaches that pharmaceutical companies use to get it out there.

The last question was about whether this brochure should serve as a model for the development of more specific test information and brochures, and you'll see here that most people who answered the question said yes, 21 thought not, and 12 weren't so sure. Not that many people. You'll see kind of a decline in the number of people who answered this question. I don't know if people got tired by the sixth question or what or maybe they were too shy about answering the question and they didn't want to be negative or maybe they didn't want to be -- I don't know.

Then the other part of this question said who should be tasked with developing test-specific brochures? Is this an appropriate role for this committee? Well, I

didn't add up those first four, but I think it comes close to being -- maybe it's a little more than 61. Is it? One of the mathematicians on here? I'm sorry. I didn't do that.

But anyway, there are a lot of other people for whom it would be appropriate to do this, but I think the most popular answer was that it should be done collaboratively by professionals, and I think consumers were mentioned, professional societies, consumer organizations, and government through a partnership really.

Sixty-one people did think this was an appropriate role for the committee, half that many thought it wasn't and felt strongly, some of them, about that. I should point out that even though 61 people said it was an appropriate role for us, some of those same people said someone else should do it, and then we also asked whether we should recommend that HHS support the development of test-specific information brochures, and here again, not that many people, as many people, answered the question. Eighty-one thought we should, five said no.

Now, I just wanted to bring up -- that was sort of the quantitative data, but I wanted to give you an overall sense I think of the comments that came in, and I'm going to give you a negative one and then two positive ones.

This is from Professor Sorenson, who's at the Department of Health Behavior and Health Education at UNC. He said that "A general information brochure given to people in general probably will have little impact. People tend not to pay attention to such educational material unless the information is relevant to them at the time they receive it. This would argue in favor of disease-specific brochures, of which many currently exist."

On the other hand, Nancy Green, Acting Medical Director at the March of Dimes, said, "This brochure should help educate the public about the basic issues of the field of genetics and in so doing provide information which will prompt appropriate choice and action."

And Joan Weiss, the Founding Director of the Genetic Alliance, said, "This would fill a tremendous gap now existing in educating the public about what genetic testing involves."

Now, the last few slides are proposed next steps for the committee's consideration, but I just wanted to pause here to see if you wanted to talk about the comments in any way before going through those.

DR. LEWIS: Yes. It seems to me one of the suggestions I might have would be I was a little bit concerned about the cultural sensitivity piece. With so few people answering and not having the sense of the cultural backgrounds of the people who answered the question, I think it's real easy for somebody who is mainstream to be able to say yes, this is fine, it doesn't offend me, but it will be real important to me to have some people of diverse populations take a look at this and make sure it was sensitive, because those are the people that we want to make sure we're not doing anything that would not be appropriate. So I would think that maybe having a targeted subgroup of members of racial and ethnic minorities to take a look at the brochure might help us with the cultural sensitivity question.

DR. McCABE: And the group that we briefed back in March have volunteered to help us with any issue like this, so that we have better access to individuals, and they've offered to help.

DR. LEWIS: And the other people we still have on board are the folks who helped us with our outreach conference. I don't know if they're still available, but it's a group of people that were very helpful to us when we did the last Baltimore conference and really represented leadership in a variety of populations. So that's another group we might be able to tap.

DR. McCABE: Yes. That is a group that we have gone back to and probably received this mailing.

MS. CARR: Yes, they would have been.

DR. McCABE: So that is a group we have involved in the past. It would help to have some new individuals as well.

DR. LEWIS: And if the comments came from them, and you don't know that necessarily because you just kind of had comments -- if it came from that group, I'd be very

concerned. If it didn't, I might be less concerned.

DR. McCABE: No, I think it's an important point.

Elliott, and then Michele.

MR. HILLBACK: I think overall the feedback was positive. Sarah and I were talking about this last night. I lumped the comments into two sections. One are sort of mechanical things, level of reading, et cetera, et cetera, that are very easily fixable.

The other I think takes a little more work, but is doable as well, which are some of the comments. If you really look at the people that didn't like it, most of their comments to me seemed to be circling around some of the issues around the definition of what's a genetic test and whether this was inclusive enough or too inclusive or whatever, some hard thinking there.

But I guess all that's a function of what proposals we have about what we're going to do with this next and how much work we ought to put into it, but I think fundamentally it's an interesting response.

DR. McCABE: Michele?

DR. LLOYD-PURYEAR: I agree actually with the issue that Judy raised, the need for presenting this to more diverse groups, and I would like to volunteer. When this document is ready more for prime time, we're going to be doing through March of Dimes, a cooperative agreement we have with them, two community consultations, and in fact, this would be advantageous to us to actually present something like this. But it would need to be for one community translated into Spanish.

MS. CARR: For that.

DR. LLOYD-PURYEAR: For that.

DR. McCABE: Joann?

DR. BOUGHMAN: I just have a follow-up question on the comments that this was duplicative of other brochures. Did they send you or did they tell you what other brochures are out there that they thought this was duplicative of or was it disease-specific

brochures?

MS. CARR: I think people were thinking that other efforts to describe genetic testing had been made. I think the one publication in particular people mentioned was "Understanding Gene Testing," which is an NCI/NHGRI document.

It is somewhat different in the sense, even though ours is somewhat longer than perhaps people thought, that one's very long. It's Web-based, and so I think the work group had looked at that document and in fact we used it extensively, I will say, when we were putting our public consultation document together, and the very, very, very first draft of this brochure was in part based on that, but I also think that that brochure has a bit of a breast cancer focus, and there was another one that people mentioned that they liked that is also on breast cancer.

But I think it probably behooves us to look. I need to go through the comments probably and look for if there were specific examples mentioned.

Somebody, a colleague, really, from the Department of Education referred to a brochure that they had produced and that she thought was excellent. She also thought a cystic fibrosis brochure that NIH had produced would be a good example. So I think that answer probably got caught up in the specific versus general issue, too.

DR. McCABE: Do you want to move on to the proposed next steps, Sarah?

MS. CARR: Yes, I will, because I think they address some of the concerns that were mentioned.

So here are the proposed steps. First of all, in consultation with selected editors that we revise the brochure based on the public comments.

Secondly, that we conduct focus groups with diverse representatives of the general public and patients and consumers.

Then third, that we develop a report to the Secretary that highlights the importance of public understanding of genetic testing, calls for the development of and dissemination of information about genetic tests to the general public and to patients and

consumers considering genetic testing, calls attention to the need for informational materials tailored to particular communities, including groups linked by ethnicity, culture or language, and transmits the final brochure as an example of the type of information needed to enhance public understanding, and a model along with other such brochures for the preparation of additional brochures on specific tests and categories of tests.

Then our last suggested next step is that we post it on our Website in a downloadable form and make sure it gets reviewed periodically to ensure continued currency and accuracy.

DR. McCABE: So let me just highlight what the proposal is. It basically is that some more work be done on the brochure, that the brochure not be the report but the brochure be an example of the kind of activity, also discussing the hard work that goes into such an activity, and that it be a part of the report that would go to the Secretary then to address these issues.

Vence?

MR. BONHAM: I would be willing to work with the committee related to making sure we access diverse communities and we can work with the Community of Colors Project as well as the March of Dimes Project in accessing African American and Latino communities. So I'll be happy to do that.

DR. McCABE: Thank you.

Wylie?

DR. BURKE: I wanted to endorse the proposed next steps. I think they follow very clearly from the document and the public comment and will take us to the appropriate next step.

DR. McCABE: Joann?

DR. BOUGHMAN: I would also like to see some sort of call, if you will, to other organizations that would like to in fact put it on their Website as well. Certainly the ASHG, I'm sure, would like it in our educational materials, and I would expect that

GeneTests/GeneReviews and several of the other Websites that might be related to genetic testing of any sort would welcome one more piece of informational material.

DR. McCABE: Elliott?

MR. HILLBACK: Yes, I don't disagree with this process. I think it's sequential and probably leads us to the right direction. I wonder if you do want to raise the issue that's been mentioned which is, are we primarily doers of this level of detail or are we primarily trying to goad others into doing these kinds of things? Not that I disagree with the brochure or moving forward with it. It was an interesting conversation. It was offline last night, and I think maybe we want it online for a minute.

DR. BURKE: Yes, I'd like to follow up on that. I actually think it's good that you're raising that point. I think part of what I was taking from the conversation is we've provided at least the first version of what will probably be a useful model. The general document may have limited utility, but for the right patient it's the right document.

More importantly, it identifies and helps to flesh out the kind of resources that are involved, identifies what kind of content ought to be in a good model, and our job, I think at this point, would be to say to HHS, this is a good thing, and this is a good kind of model. I don't think it is our job at that point.

DR. McCABE: I'll tell you another model where I bet it gets used, and that is in middle school and high school science projects. I'm always getting emails asking for help with developing informational bits like this, and I see others saying the same thing. So having it on the Internet will then begin to educate in the pre-university level probably as well as in the university level as our folks pointed out.

Sarah, you wanted to make a comment? Mary?

MS. DAVIDSON: Yes, I just wanted to clarify next steps. So the plan is then to take this general longer document and then to spin out several --

MS. CARR: Yes.

MS. DAVIDSON: Okay. That's interesting, because I really agree, Wylie

and Elliott, with we already discussed the issue about products versus recommendations and issues. But I think we've come this far with this document that it would really -- and again, I'm thinking of the communities that I represent that use this to develop their -- take one more step, which isn't that big, to develop a specific template for a specific information, and this is linking back really I think to what concerns me somewhat about the shift in this working group from its original task to how to develop disease-specific information templates to looking at something that's public education.

DR. McCABE: Just if I could clarify that, and this is my own opinion, not the opinion of the working group, but once this is available on the Internet as a model and once we've discussed the model in a report to the Secretary, then I think it's freely available for anyone to use. So I think the beauty of it would be that it would be available. It would be freely downloadable and then others could take it as a model and tailor it to their own needs. I don't think there's going to be any proprietary – you know, in fact, I would think the more it got duplicated and utilized, the happier people would be.

MS. DAVIDSON: Right, and I'm just looking in terms of gaps. I mean, having that kind of specific model would just be a tremendous addition to the information system as it exists now.

DR. McCABE: Wylie?

DR. BURKE: Actually, following up on your comment, it seems to me that in the revision process, attention should be paid to its use as a model. In other words, it's a document that provides educational value, but it may be that either within the document or as a side-by-side document -- I'm not quite sure how this would go -- that there should be a list, a template, pointing to whatever the right word is, what needs to be in the document.

I mean, to some extent, it is that model, but the question that the revisers should perhaps think about is are there additional directions about information that one would specifically want to pursue for certain kinds of genetic tests, prenatal, you know.

DR. McCABE: Elliott, and then Michele.

MR. HILLBACK: Yes, I think this almost goes back to the idea of the report, which is to move the document forward but in parallel move the making of the document forward, so that part of the report we would provide is these are some of the steps. These are some of the things that are guidelines for others that might continue this process or adapt this and take it forward, and then I guess the next question we have to ask is whether we want to find a home for this document to keep it live and active long run or whether we will be that home, and I don't have a bias right now either way, and I just put it on the table.

But I think to say here's a document, here's all the reasons why people need a document, and in fact here's a number of the sort of points to consider as you update the document, modify the document for your own purposes, don't forget these kinds of things -- so it becomes a teachable moment as well as a useable moment or whatever you want to call it.

DR. LLOYD-PURYEAR: But I think there's a big caution here before it's put out, besides revising it based on public comments, that we have focus groups.

MS. CARR: Oh, yes. That would occur before.

DR. LLOYD-PURYEAR: Before you're going to put it up on the Web.

MS. CARR: Oh, Yes.

DR. LLOYD-PURYEAR: And then, I see it being not on anybody's one Website but it's public, and I think once it goes through several focus groups and reiterations, then it's for any of us to use as we want, but I like Wylie's idea of having not a side by side but, I mean, to really present it as a model with little --

MS. CARR: Annotated versions.

DR. LLOYD-PURYEAR: Yes. Sort of things to keep in mind.

MS. CARR: Yes.

DR. LLOYD-PURYEAR: I don't know.

MS. CARR: Could I just respond to the point Elliott was raising a few people ago, which was that, was this part of our role or is this an appropriate thing for this committee to do?

I think what we tried to do with these proposed next steps was to do justice to the public comments, especially because they're full of wonderful ideas. People went to a lot of trouble to look at the document and to take time to send in their ideas and suggestions. It's incredibly valuable.

But I think what it taught at least staff involved in reviewing it was that maybe we aren't the best group to do this. Perhaps what we've done here is stimulate some discussion, and what the proposed next steps try to do is make this more of a policy issue, a policy recommendation to the Secretary, so that what we're really saying, the most important thing we're saying, to the Secretary is that this is a very important thing, public education and public understanding.

We've tried to develop something that could be used as an example among others to help do that, and we think your agencies need to take a strong effort in making that happen and doing it in a way that will be sure that we reach all American communities, not just those who speak English or who are of a certain socioeconomic background.

So I think we were trying to acknowledge that maybe we drifted a little bit away from our central mission of policy advice.

MR. HILLBACK: Could I just follow up on that? I think that's a little bit, though, also, what I meant by see if we could find it a home. I don't think we should just hope that there's some takers out there in HHS land, but maybe we want to recruit some people who are excited by what we've done and are willing to take it to the next round and the next round as part of our process of doing this.

DR. McCABE: Pat?

DR. CHARACHE: This actually fits nicely with the educational report from yesterday's work in which we did not address the public information base, and this is a very good illustration of that approach which we were also concurrently exploring.

DR. McCABE: Well, Victor and Ben, I think you're both to be commended and your work group for putting these items together that you've presented to us this afternoon.

Thank you very much.

Do you have anything else you wish to add at this point in time?

DR. WILFOND: No.

DR. McCABE: Sarah does.

MS. CARR: I just want to give public credit to the main author of this document, who is Wendy Uhlmann, a genetic counselor. She did an enormous amount of work. She's part of the working group, as you saw before, and she did an enormous amount of work on it, and the work group members and I think the committee are grateful that she took the time to do that and made it a better document. Maybe not a perfect one, but certainly made it better than it was when we started.

DR. McCABE: Yes, and we should let Wendy know that. Linda and I were visiting professors at the University of Michigan a week or so ago, and we saw Wendy, and I know when she did this work. It was usually after midnight was when she finally had time to herself to get it done. So she put a lot of effort in on it.

I want to thank Dr. Sherrie Hans for spending the day with us. We really appreciate the input this morning and your willingness to spend the full day with us. Thank you very much.

For the committee members, we will meet in the lobby to go to dinner at 6:45. Those of you who've signed up on the sheet, please join us there at 6:45, and tomorrow, we will be meeting in this room. It's at 8:00 a.m. tomorrow, not the time we met today, but at 8:00 a.m. tomorrow morning is when we will begin.

Anything? Was there something else?

PARTICIPANT: No.

DR. McCABE: Okay. So we will see you tomorrow morning. Those of you who have to leave tonight, travel safely.

(Whereupon, at 5:56 p.m., the meeting was recessed, to reconvene at 8:00 a.m. on Wednesday, May 15, 2002.)