U.S. DEPARTMENT OF HEALTH AND HUMAN SERVICES

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

Fourth Meeting

Monday,
June 14, 2004

Grand Ballroom Salons A-D
Bethesda Marriott
5151 Pooks Hill Road
Bethesda, Maryland

IN ATTENDANCE:

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C O N T E N T S

Welcome and Opening Remarks

Edward R.B. McCabe, M.D., Ph.D.

Update on Genetic Nondiscrimination Legislation and the Activities of the Coalition on Genetic Fairness

Joann Boughman, Ph.D.
Executive Vice President
American Society of Human Genetics

Discussion and Next Steps

Presentation on Information Gathered on Efforts in Genetics Education and Training

Joan Y. Reede, M.D., M.P.H., M.S.
Chair, Education Task Force

Roundtable with Selected Organizations on Their Efforts in Genetics Education and Training

Joan Y. Reede, M.D., M.P.H., M.S.
Facilitator

Discussion of Draft Education Resolution

Report on the Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children

Reed V. Tuckson, M.D.
Liaison Member, ACHD

Discussion

C O N T E N T S

Public Comment

Kathleen Rand Reed, B.A., M.A.A.
The Rand Reed Group

Andrew Fawcett
American Board of Genetic Counseling

Barbara Handelin, Ph.D.
Handelin Associates

Overview of Draft Coverage and
Reimbursement Report

Cynthia E. Berry, J.D.

Discussion of Draft Report and Development
of Recommendations

P R O C E E D I N G S (8:39 a.m.)

DR. McCABE: Well, good morning, everyone, and welcome to the fourth meeting of the Secretary's Advisory Committee on Genetics, Health, and Society. The public was made aware of this meeting through notices in the Federal Register, as well as announcements on the SACGHS website and listserv.

First, I want to begin by taking note of some changes in our membership. We are very pleased to welcome a new ex officio member, Dr. Howard Zucker, who is the Deputy Assistant Secretary for Health and is representing the Assistant Secretary for Health in the Department of Health and Human Services.

Dr. Zucker trained in pediatrics and has held faculty appointments at Yale University School of Medicine, Columbia University College of Physicians and Surgeons, Cornell University Wyle College of Medicine, and the National Institutes of Health. Dr. Zucker also served as a White House fellow and worked at the Center for Space Research at MIT. Welcome.

Dr. Steven Gutman, Director of the Office for In Vitro Diagnostics Evaluation and Safety, has been named as the new ex officio for FDA, replacing David Feigal, who recently accepted a position at NDA Partners, a product development consultancy to the biopharmaceutical and medical device industries.

In addition, we welcome Joe Hackett from the Food and Drug Administration, filling in for Dr. Gutman this morning, and Dr. Hratch Semerjian, Acting Director for the National Institute of Standards and Technology, will be representing the Department of Commerce for Arden Bement, who is currently Acting Director of the National Science Foundation.

Also, Dr. Steve Phurrough will be substituting for Sean Tunis today. Dr. Phurrough is Director of the Coverage Analysis Group at the Centers for Medicare and Medicaid Services. Judy Yost will be here tomorrow representing CMS.

Dr. Sherrie Hans will represent the Department of Veteran Affairs for Ellen Fox. Ms. Amy Turner is here representing the Department of Labor for Tom Alexander.

Welcome to everyone.

I also want to explain a couple of absences in the membership. Christopher Hook will be joining us later today. Hunt Willard will join us tomorrow, and may be able to participate for part of today's meeting by phone. Reed Tuckson will be joining us by phone this morning for the remainder of the day.

Eric Lander is also not present. Last week, with great regret, Eric resigned from the committee, due to extensive commitments and responsibilities as Director of the new Brod Institute of MIT and Harvard that prevents him from having a more active role in the committee's activities. Eric told me that he very much wishes to stay involved in the committee's work, and I promised him that he would become a distinguished member emeritus and that we would feel free to tap his expertise as needed. The Secretary will be appointing a new member to take Eric's place in the very near future.

Before I review today's agenda, I would like to remind everyone that at the end of the March meeting, we set some extremely ambitious goals in preparation for this meeting. We planned a number of short-term actions, including sending a second letter to the Secretary in support of federal genetic nondiscrimination legislation.

We decided that education and training of health professionals in genetics

was a high priority issue, and that after additional fact-finding on genetics education efforts, we would be equipped to consider a resolution regarding these efforts at this meeting.

An Education Task Force chaired by Joan Reede has been hard at work gathering data and drafting a resolution for our consideration today. The committee also decided to prepare a report describing SACGHS' mandate and priorities, and providing background information policy considerations on priority issues.

Sarah Carr and her staff, with the assistance of Emily Winn-Deen, prepared a draft vision report for our consideration. In March, we also identified high priority issues that require more in-depth study. Coverage and reimbursement of genetic technologies and services was identified as the first topic on which we would focus our efforts.

Staff, with the assistance of Cindy Berry, carried out additional data and information gathering, and prepared a draft report for our consideration. This afternoon, we will be discussing the draft, and developing recommendations for the Secretary.

Three other issues were also identified as warranting in-depth study. Large population studies, pharmacogenomics, and direct-to-consumer marketing. Our in-depth work on these three issues will begin at future meetings. With regard to the direct-to-consumer marketing issue, we also decided to take a short-term step by drafting a resolution expressing concern about the proliferation of direct-to-consumer advertising of genetic tests.

A task force chaired by Chris Hook accomplished that short-term goal, and we will be considering the draft resolution tomorrow. For this meeting, we hope to finalize the two resolutions on education, direct-to-consumer marketing, and the report on our priority-setting process. Given the length and complexity of the coverage and reimbursement report, and the work that needs to be done to develop consensus recommendations, Cindy and I expect that we will want to continue refining the report and gathering additional public comment after this meeting.

We will begin the meeting with an update on the status of federal genetic nondiscrimination legislation by Dr. Joann Boughman. Following our discussions on genetic nondiscrimination, we will engage in a roundtable discussion on educational issues with involved professional organizations, and consider the draft resolution on education. The afternoon is dedicated to discussions of coverage and reimbursement of genetic technologies and services. Tomorrow we will hear a presentation from Dr. Muin Khoury about CDC's public health approaches to genomics, and deliberate on the vision report and direct-to-consumer marketing resolution.

Finally, we have allowed time to discuss the other issues we prioritized at the March meeting, and identify next steps. Let me also point out that we have public comment sessions scheduled for both days. We will hear comments immediately following lunch today, and tomorrow morning. Thus far, five individuals have registered to provide testimony. If there are others who wish to give public comments today or tomorrow, please sign up at the registration desk.

We have received a considerable number of written public comments on the draft documents as well, including requests that we provide additional time for the public to comment. The comments can be found in your table folders. As our deliberations on the draft documents proceed throughout the next few days, we will take careful account of the comments on each document.

At this time, we'll hear from Sarah Carr regarding conflicts of interest.

MS. CARR: Good morning. Being a member of this committee makes you a special government employee, and thereby subject to 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." You each received a copy of this document when you were appointed to the committee, and I'm going to review one of the rules in that document.

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.

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

Let me add a sincere thank you to all of you for being so attentive to these rules. We know that the reporting process is time consuming, and we very much appreciate how conscientious all of you are about fulfilling your duties.

Thank you.

DR. McCABE: Thank you, Sarah.

Now we're going to hear an update on genetic nondiscrimination legislation, and the activities of the Coalition on Genetic Fairness from Joann Boughman. Dr. Boughman is Executive Vice President of the American Society of Human Genetics.

Protection against genetic discrimination is this committee's top priority, as we determined in March, and by previous correspondence with the Secretary. We were encouraged when Senate Bill S.1053 unanimously passed in the Senate last October, 95 to 0. Following the March meeting, we wrote a second letter to the Secretary urging that continued pressure be applied to facilitate passage of this bill in the House.

Dr. Joann Boughman is here this morning to provide an update on the status of S.1053 in the House, and the related activities of the Coalition on Genetic Fairness. Dr. Boughman?

DR. BOUGHMAN: Thank you very much.

Each of you in the table folders, and outside, have the list of slides, and we'll go through these slides very quickly, and then I think just get down to conversation and a couple of ideas that people might have.

Don't mistake the bad news slide being blank for the fact that there is no bad news. The bad news is that absolutely nothing has happened in any real kind of way. That doesn't mean there has not been activity, but there has been no real noticeable movement forward.

The good news since the March meeting, is that the session isn't over yet. We still have some time, and we will continue working folks on the Hill in every way that we can. The other part of the good news is that the members will be going back to their home

districts, and we may be able to call on people from home to contact and discuss issues with their members.

As Dr. McCabe just pointed out, 1053 passed the Senate last fall, 95 to 0. There were three bills that were brought over to the House, 1910 and 3636 actually formally introduced S.1053, not yet having been formally introduced. Once introduced, HR 1910 was sent to two committees, and HR 3636 sent to one committee. Now, there are appropriations committees that do eventually see these bills on the House side, but the committees listed on this slide are the committees of real action and deliberation for this kind of bill that involves both work force, and insurance issues.

Our plans, or our hopes, have been to in fact get 1053 to committee, and then to the floor, or around the committees and directly to the floor. Another opportunity might have been to get 1019 through some committees so that 1053 could end up, or the wording in 1053, the concepts in 1053, could be the goal of compromise. We continue to believe that it is not useful to have 3636 on the agenda. If you remember, House Bill 3636 is very much a shell bill, and this is not really a Christmas tree bill as it is referred to on the Hill, but this is just a skeletal framework so that as it would go through the process, members would get more and more wording on it, and it would be more and more complicated to in fact negotiate this through the process.

We have had several meetings by advocates. We have had multiple meetings with the Chamber of Commerce, and the Chamber of Commerce has not been terribly vocal on this in a negative way. They have made some statements about some definitions in the wording.

We were very pleased, for example, to see a wonderful editorial in the Washington Post by the Insurance Association that said this genetic nondiscrimination stuff is out there. We don't believe that the insurance companies are discriminating, but we don't have a problem with the Congress telling us not to, because we think we don't do it anyway.

So in fact, they basically said in a very positive way, go ahead and pass the bill, we'll deal with the details in some sort of way. We thought that might be helpful and make some movement, but it has not. In the meantime, many of us continue with meetings on the Hill, and with members of a variety of agencies. These meetings are now even broadening. We are not just meeting with staff members of key committee members, we are meeting with staff members, and with members of Congress who are good friends of the staff members, and point blank asking them to nudge their friends who are in the right positions to get some of these things going.

We also continue to work on the leadership in the same kind of way, every opportunity any of us has to make that comment to any of the House leadership or members who have influence with the House leadership. Let me give you the kind of example beyond the formal meetings that we have.

I had been at a couple of events where Representative Chris Van Hollen from the District here in Maryland that represents a significant number of scientists, and FASEB is in his district as well. My conversation centered around our frustration on genetic nondiscrimination. He agreed with me, and basically I said, let's just not get frustrated anymore, it is time to get angry, it is time to really get upset about this and move forward. So those are the kinds of changes that we are trying to make with our friends that may have meaningful positions.

Since the March meeting, we have had a letter from distinguished scientists sent to the leadership. There have been a series of blast emails that have gone out via CapWiz and some other venues, to allow letters or emails to go to members. For example, I know from the American Society of Human Genetics, when we sent out our blast email encouraging all of our members to email their members, there were 1,100 emails that were received within about a 96-hour, or 92-hour period of time.

So in fact, we've had some of these waves of activity, and those were only the ones that I could count, and other organizations had some of those activities as well. When 1,000 or 2,000 emails show up on the Hill in a two or three day period, what basically happens is the staff members must take notice of this. When that level of activity happens, they must report that to their Congressman, and then the discussion goes on in the halls, which may be another way.

We have also prepared a one-pager of information from geneticists that we have given to the Coalition for Genetic Fairness. We are still working on some more good stories to get out there as well. We are also asking for more follow-up contacts by individual constituents.

Additional strategies that we would encourage everybody in this room to put forward are additional personal discussions with members. Maybe we'll just be able to wear them down. If we can't get them excited about it, maybe we can at least wear them down. I know that you can hear the frustration in my voice on this.

We are going to be continuously requesting personal contact by constituents, especially when the members are at home during breaks, and encourage strategic contact between members and leadership. What we have got to do is keep the Genetic Information Nondiscrimination Act on the agenda. As you all are keenly aware, there are many other things on the agenda, and the strategy that we just have to keep putting forward is that this issue, while maybe not as big, or as front page as many of the international, or even other national events, it is still the right thing to do, and this is something that they can do for the American public.

Hopefully by coming today and venting some of my frustration, at least among friends and allies, we can state a renewed commitment and gain encouragement from each other to try and move this forward. At that point, I'll be happy to answer questions. There are other members in the audience who have been at some of these meetings and have met with some of the members of Congress.

I would ask if any of them have specific comments, just reiterate the fact that while on the one hand, it is appropriate for us to continuously recognize that this is not the most important item on the agenda, and that we do have a special interest in this, and it is not self-interest. It really is a bill for the people out there, and to pass this bill, everybody could go home with a win.

DR. McCABE: Thank you very much, Dr. Boughman, and if you could join us at the table perhaps for this discussion.

While you are doing that, I'll just remind everyone that government employees and special government employees, and all of us on the committee, while we are acting for the committee, are special government employees, so we are precluded from lobbying. Therefore, it is just important to note that this is an update from ASHG, and represents the views of ASHG and the coalition. Our discussion here is really on how we can

provide advice to the Department of Health and Human Services and the Secretary.

DR. BOUGHMAN: Dr. McCabe?

DR. McCABE: Yes?

DR. BOUGHMAN: I would say that I am well aware of that, and hopefully people understood that this was the activity of an organization here in town, and the update on those activities, and recognize that we all wear a variety of hats, and that information and education to ourselves and to others that we work with about the importance of individual contact.

I think one of the roles that this committee can continuously have is educating individuals about the situation, just where things are in fact.

DR. McCABE: I appreciate that very much. We were just clarifying that the "us" that you were using was referring to the actions of the ASHG and the coalition, not the actions of this committee. We discussed the possibility in March that further action by our committee may be required in the event that legislation is not passed in this Congress, and I hate to be pessimistic by stating that, but I think it is reality.

As a reminder, we discussed organizing briefings from individuals who have faced genetic discrimination, who paid out of pocket to keep their genetic information out of their medical records, and who chose to forego treatment due to fear of genetic discrimination.

I think it is important to remind ourselves that those were activities we considered, and that we might consider for the future, should this legislation not pass. We now have some time to discuss how we should proceed. I would take questions or comments from the committee for discussion, and particularly since we have Dr. Boughman at the table for her comment.

Dr. Winn-Deen?

DR. WINN-DEEN: Yes. I just wanted to ask if you had had any success in gathering up some of these patient stories. I know you had mentioned at your last presentation for the committee that you were going to be working on that, and I think that would be extremely helpful, at whatever point you have those.

DR. BOUGHMAN: Yes. In fact, and I'm sorry, I failed to put this on, and it was a very important event. The Genetic Alliance is a very active member of the Coalition of Genetic Fairness, and it had a press briefing on the Hill. In fact, there was a very impressive woman who told of her case. At that time, we were reenergized to gather some cases.

We have not created other forums for the release of that information, but there has been some volleying back and forth. Coalitions of organizations, and coalitions of coalitions tend to be a little bit slower in getting some things done than we might like sometimes. But in fact, we have also worked at getting some of these cases, encouraging them to talk directly as constituents to talk to their congressional representatives, too.

I mean, this woman and her two children who were carriers and denied insurance, made a very impressive presentation. It clearly indicated what the challenge was before us.

DR. McCABE: And we know also of cases where individuals have lost employment because, presumably at least in one case that I know of, because of concerns regarding insurance. I know that case appeared before the EEOC. I don't know if you'd wish to comment, Paul.

MR. MILLER: No, I think you articulated the issue.

DR. McCABE: Other comments? I would remind everyone that the first letter of the Secretary's Advisory Committee on Genetic Testing, wrote to the Secretary was about that, this issue, that our first letter, and the first letter of SACGT to the new administration, to the current administration, was about this issue.

Our first letter to the Secretary of the Secretary's Advisory Committee on Genetics, Health, and Society was on this issue, and we followed it up with another letter. So clearly this has been a top priority for these two committees, and I would guess continues to be. Yes?

MR. LESHAN: I'm Tim Leshan, sitting in for Dr. Collins today, with the Genome Institute, representing the NIH. I just feel like I'd be remiss if I didn't say that Dr. Collins has been providing technical assistance and working on this issue, both within the administration, and in responding to requests from folks on Capital Hill about this issue, and really thanks the coalition for its efforts in this area. NIH feels it is very important that we see if we can get this legislation passed, as you know. We're very encouraged by some of the press activity this year, but also concerned that we haven't seen some action, but still hopeful that we may in the future, and we'll continue to provide assistance and work on the issue as much as possible.

DR. McCABE: Tim, thank you for introducing yourself. I apologize for not recognizing you in my opening remarks. You are representing Alan Guttmacher, who will be sitting in for Francis Collins when he arrives.

Also, I apologize to Suzanne Feetham for not recognizing you sitting in for Sam Shekar.

Yes, Joann?

DR. BOUGHMAN: I also just wanted to remind the members of the committee that the situation that we're in is that there has been written documentation, the White House, the administration support of 1053, and it is actually in writing. If 1053 came to the desk of the President, he would sign it, basically, is the wording in this SAP, as it is referred to.

So that any comments that would be made, or could be made to the Secretary, would be made in a context of already having documentation from the Executive Branch in support of the legislation that has passed the Senate.

DR. McCABE: Thank you.

Other comments? Yes, Emily?

DR. WINN-DEEN: Are there some specific issues? Because the last time we discussed this, it seemed like the issue was, is this really an issue. Have we gotten past that? Is it just a prioritization versus other more pressing congressional matters at this point? Or what is the barrier that we need to overcome?

DR. BOUGHMAN: I think it is both height and width of the barrier. There are just so many very big issues going on, budgetary issues being one of them, along with everything else that is going on, that it is difficult to get the more focused issues onto a series of agendas. As you are well aware, several things that have happened, in fact, Congress spent almost all of its time last week on the issues and commentary on former President Reagan. So what issues were going to be debated during the week last week, slid another week. So that the challenge is both height and width, I think.

DR. McCABE: Paul?

MR. MILLER: I would just add that it is always in the context of these discussions, as I have heard and participated in them, it is always useful to reinforce, and to have more information about the reality of this problem, about the concerns, and to develop the issue of the concerns of real people out there, and the impact on the research and science. It is not the kind of issue that is sort of addressed, and then move onto something else. I think that that is constantly an issue and concern in the background underlying a lot of these discussions.

DR. McCABE: Debra?

DR. LEONARD: So beyond writing letters, which we've done now twice to the Secretary, and we clearly have the Executive Branch's support, what are specific steps that this committee could take to assist with getting this through the House?

DR. BOUGHMAN: My hope would be, and we will utilize the fact that this issue was on the agenda of this, a federally appointed committee, to look at these issues, that this was the top priority and the first topic of discussion. That fact will allow us, and anybody here in the room, to comment on the importance of this issue to the committee, responding in part to what Paul said.

It doesn't enumerate individual issues. But in any way that we can utilize this support to bring forward more individuals who need to be supported to go see their Congressman or whatever, and we will be writing additional letters to the leadership and Congress once again, to try and do this.

But we are using just about every venue that we know to use at this point, I must say. I don't know that there are additional specific actions that we could do at this point.

DR. McCABE: Well, I'd just remind everyone that we have discussed the possibility of having briefings where we would have individuals appear before us who had been the subject of genetic discrimination. I would ask if that is something that the committee would like to consider for our October meeting, should the House bill not pass by that time. Again, I don't wish to be pessimistic, but I think it is important for us to plan if the actions of the coalition and others were successful, then there would be no need for that, and we could change the plans for that briefing at that time. Is that something that the committee would wish to consider? Debra?

DR. LEONARD: I think that we should do that. I don't know, five or 10 minutes for each person with a significant number, not just two or three people, so we can have an impact.

DR. McCABE: Okay. Is that something, Sarah, that the staff could work on for October?

MS. CARR: We can certainly try to do that. I guess I would also, if it is appropriate, to reach out to the coalition, because you've done so much of this already. If there is a way we can augment that work, or complement it in some way by bringing it to the attention of this committee, and thereby to the Secretary, we'd certainly want to, and would do whatever we can.

DR. McCABE: Is there anyone on the committee, among the members or the ad hoc representatives, who would disagree with that plan for October?

(No response.)

DR. McCABE: Okay. Seeing no one wish to comment, then I'll assume that we will move forward and begin planning for that at the end of this meeting.

MS. CARR: Maybe it would be helpful if some of the members, we could form a little task force to help organize these briefings, and have your input on how much time to devote to it and so forth.

DR. McCABE: Does anyone wish to volunteer? Barbara, Debra, Agnes, Emily. Yes, please. We'll call on the ad hocs probably as well, but thank you to the two of you for volunteering, and Paul. You have been a leader in this in the EEOC. We may call on others of you as needed, especially among the ad hocs. If anybody has been shy and wishes to volunteer, at the break, please let Sarah know.

Dr. Boughman, thank you very much for briefing us, and thank you for your efforts. They clearly fit with those expressed by this committee in our correspondence in the past.

So next, we're actually running just a little bit ahead of time, but we're going to have a presentation on information gathered on efforts in genetics education and training by Dr. Joan Reede. Dr. Reede will review the education task force's information gathering efforts over the last three months.

I'd like to thank Joan for your chairing the task force, as well as Kim Zellmer, Hunt Willard, Barbara Harrison, and Agnes Masny for your service on the task force. We appreciate the time and effort that all of you have put into gathering the data and preparing the draft resolution.

Joan, please proceed.

DR. REEDE: Thank you very much, and thank you for the opportunity to present the work of the task force.

You just mentioned the names of the members of that task force, but I also want to acknowledge the staff who supported us. Amanda Sarata did a wonderful job of helping to facilitate our meetings, moving this forward, and helping to put this presentation together.

During the discussions of genetic education and training at the March meeting, the committee decided to draft a resolution to the Secretary on the issue of genetics education and training, outlining key recommendations in this area. Toward this end, they decided that a task force should be established with these charges to collect information on the activities of health professional organizations that relate to genetics education and training, to organize and facilitate a roundtable discussion to be held during this June meeting, and to draft a resolution to the Secretary on genetics education and training.

The committee's request to hear formally from professional organizations in the private sector on their activities in genetics education and training was meant to serve as a follow up to, or addition to the survey of federal agencies on their activities, as well as the presentation and information from Joann Boughman at the October meeting.

It was felt that this information would be used to inform our resolution, and should ensure that any recommendations made to the federal government would be complementary to activities already ongoing in the private sector. We solicited information from 26 organizations.

The organizations can be divided into three categories. Genetic-specific organizations, health professional educational organizations, organizations involved in the education of health professionals, and health profession organizations. The latter really targeting three groups, those that represent generalists such as AMA, those representing

specialities such as family practitioners, pediatricians, OB/GYN, and those representing specific constituencies, such as the National Medical Association, AAIP, and NHMA.

Of note, we had only a maximum of nine organizations in each category, and this was really to be consistent with guidelines so that we would not have to turn to OMB for review of our survey of the organizations. Fifteen of the 26 organizations responded, which is a fairly good response for a three-week period from when the surveys went out. Of note, however, we shouldn't assume that the groups that we did not hear from do not have important activities in these areas.

Here is a list of the organizations that responded. You'll be hearing from some of these organizations during the roundtable discussion, as well as some additional organizations that did not get their responses in in time for this.

In addition to having information that represents organizations, it is also important to note that this information represents multiple specialties or disciplines. There is information on nurses, geneticists, allied health professionals, dentists, pharmacists, and physicians.

With regard to the three categories, I will go through them in sequence. The first is on genetic-specific organizations. The committee felt that it was important to solicit information from organizations who are focused on the issues of genetics. This provided the committee with the information about the "state of the state," which is currently going on within organizations that have a high awareness of and focus on genetics and genomics.

We have already benefitted greatly from their input on many issues, and appreciate their willingness to continue providing their input. We highlighted three important areas of potential gaps in our understanding. These were diversity, life insurance certification, and curricular development.

We wanted to know about current initiatives and activities that related to diversity in the genetics workforce, that enhanced genetics and genomics curricula, and that promoted the incorporation of genetics and genomics content into licensure and certification.

Diversity in the genetics work force is an important issue to the committee. Genetics is a field where both cultural diversity and cultural competency are particularly relevant, the impact of culture on perceptions of attitudes about genetics, and also in the area of the debate about the scientific basis of race and its place in medicine and genetics.

What we found was that these organizations were involved in many activities that targeted both the pipeline, such as those involving K through 12 education, as well as the recruitment of individuals from diverse backgrounds into genetic counseling.

These efforts also targeted minority health professionals, minorities who attended colleges, high schools, colleges and universities, and also at the organizational level brought diversity in through structural forms, such as the organization of a diversity standing subcommittee.

With regard to curricula, it appears that genetic-specific groups are leaders in the development of genetic curriculum, and are actively engaged in dissemination and outreach efforts to other health professionals. They are engaged in activities such as speakers bureaus, short courses, conferences, as well as the dissemination of position papers and practice guidelines.

The core competencies provide universal guidance, and the core competencies have been a major effort on the part of these genetic-specific organizations. It

was felt that specialty-specific competencies could be determined by professional societies, and the use of the Internet to disseminate and share information was also felt to be critical.

Increasingly, medicine will become more electronic, and genetics, being a field that changes rapidly and involves large amounts of information, benefits from this change. It was felt that educational materials should conform to this trend.

Courses to train faculty to teach genetics content is a key element. This matches up with a barrier that was noted by another category, the Health Professional Educational Organizations, feeling that there was a lack of appropriately prepared faculty for genetics training and education.

Licensure, certification, and accreditation have the potential to change behavior, and increase integration of genetics and genomics knowledge throughout the health care system. Although many of the agencies did not report a great deal of activity in these areas, some of the types of activities that they are engaged in included credentialing programs in genetics, use of core competencies to help inform discussions or programs related to licensure and certification, and biannual genetics review courses.

The issue of licensure certification and accreditation can require several different types of specific policy solutions. Part of this is related to the fact that there are different factors across the board here. In licensure, oftentimes we're dealing with state certifications, we're dealing with specialty professional societies in the federal government such as with CLIA, and accreditation, and we're dealing with private organizations, such as JCAHO.

The overall recommendation from this group was that "we should be strong in our appeal to helping the HHS Secretary to actively support a wide variety of endeavors based in or funded by any of the HHS-based agencies, as well as seeking partnerships with other relevant federal agencies."

The second category. Those organizations that are involved in the education of health professionals. For this category, these questions deal with curricula development for education of health professionals. With these questions, we are trying to gauge where genetics/genomics stands in health education organizations that have a general, rather than a specific, genetics focus.

The question for this group included those about the need for the integration of genetics and genomics in their curriculum, barriers to this integration, and current ongoing activities and initiatives.

With regard to their perceived needs, there is a need to acknowledge that genetic science is for both generalists and specialists. This points out the tension between educating everyone, and maintaining a niche for specialists. General practitioners have an important role to play in integrating genetics and genomics in health care, and specialists will also play a necessary role in that new paradigm.

Due to the complexity of and the speed at which the genetics field is evolving, there is also a need for improved access to knowledge, and this was cited by many organizations. There is a need to be able to evaluate product claims. This comment ties to the committee's interest in and focus on direct-to-consumer marketing and advertising. Education will facilitate provider's ability to assess the validity, efficacy, and safety of various new products for their patients.

There is a need to redefine and recast genetics as an inherent and overarching part of health and to improve communication between all health professionals and

the public about how genetics affects health. In keeping with the theme that genetics is relevant to all specialties in medicine, and will be an important part of all aspects of clinical management of the patient, from prevention, diagnosis, and treatment perspectives.

There was a perceived need to determine the level of knowledge that is needed, who needs to know what, and who should provide that, and also the need to provide tools for lifelong learning. This last comment highlights the fact that genetics and genomics knowledge will be rapidly changing, and therefore, education should be a lifelong pursuit.

With regard to barriers, it was difficult to find current case examples or models. This comment represents an important theme that was raised by several organizations in response to many different questions. In order to teach genetics and genomics, models using clinically relevant examples are needed. There is a lack of trained faculty broadly competent in genetics and genomics, and it is expected that there will be a lack of uniformity in the rate of integration of genetics into the various specialties in medicine. This, in turn, will make it difficult to determine who should be learning what, and when.

Other barriers include difficulty in motivating students to learn something based on the promise of its "future importance," an overcrowded curricula that is already struggling with issues of basic science and clinical practice, and the fact that genetics by many is still considered to be an esoteric field.

Current activities from health professional educational organizations include membership in NCHPEG, integrating those core competencies that have been identified by NCHPEG into their entry-level competencies for their specialties, or their special disciplines, and the serving of faculty competency in and understanding of genetics/genomics.

With regard to current activities, one organization is sponsoring relevant legislation of the allied health professions that would help support curricular development in the area of genetics and genomics. Overall, it was felt that cultural competency was addressed broadly within the context of outreach to underserved populations, and there was no specific focus on the area of genetics.

Recommendations from these organizations included that schools and professional organizations must provide leadership in preparing the next generation of health professionals in genetics. The primary role of preparing health professionals lies with the schools and professional organizations, not with the federal government.

Continuing education is needed to train the trainer, and this specifically refers to helping clinicians to determine when to refer, how to obtain information about genetics research studies, and ways to discuss research options with patients. More funding is needed to support training and education in genetic technologies, and to facilitate the incorporation of new knowledge and skills.

With regard to the last set of organizations, the health professional organization responses, the committee reasoned that it was also important to gather information from umbrella organizations that focused on specific health professional disciplines. These responses provide the committee with useful information on the relative importance of genetics to organizations grappling with many other equally important issues.

The information may also be an indicator of how genetics and genomics is perceived within the health care system generally. The types of questions that were asked of this group include characterizing the need for integration of genetics and genomics, what types of activities or initiatives they have currently ongoing, and in here, we ask specifically about

partnerships, about interdisciplinary efforts, about educational products, and about outcomes, what has been evaluated, and the impact.

We asked them specifically about steps they were taking with regard to diversity, and about their particular concerns and recommendations that they would like our committee to make. With regards to their needs, they felt that all health processionals need a strong, knowledge-base in genetics and genetics testing.

Identifying clinically-relevant examples of genetics and genomics would help address the need to understand how genetics interfaces with practice. You hear a recurring theme of the need for applications, and there is a need to understand how this relates to practice.

There is a need to help professionals with up-to-date advances in genetics. This is brought by many health professional organizations in terms of the need for access to new knowledge. Again, we have the issue of lifelong learning, keeping up to date. In terms of current activities, the health professional organizations are undertaking a broad array of activities related to genetics education and training. Some are using traditional tools for consensus building and dissemination. You see a wide range from CME educational sessions, to web-based educational tools, to newspaper articles, journal articles, symposia, and across the board.

With regards to their current activities and their interdisciplinary nature or partnerships, several of the organizations are partnering with others, federal agencies, genetic specialty societies, medical societies, non-profit organizations, and private companies. The majority of the organizations report that their activities are interdisciplinary in nature.

With regards to outcome and evaluation, it was found that most of the organizations measured their outcomes or evaluation based on increasing interest, or continued interest, and in issues related to genetics and genomics. This was gathered by looking at numbers who attended genetic sessions at national meetings, hits to genetics websites, CME certificates, and distribution of educational materials.

Current activities related to diversity take many forms, from community outreach, career development, education, research, advocacy, and organizational position statements. Concerns of this last group of organizations. The science underlying issues of race in medicine needs to be examined and integrated into genetics education and training.

Issues of race in genetics necessitate special consideration and treatment in the educational setting. Awareness of these issues is an important part of a health practitioner's cultural competency. For many physicians, genetics does not have immediate, daily, and clinical applicability.

This comment relates to the need to understand how genetics interfaces with practice, and the barriers caused by the difficulty in finding case examples or models in genetics and genomics cited by health professional organizations previously.

To quote one person, "Genomics is spinach. Everybody knows it is good for them, but nobody likes it." Genetics education must be represented throughout the entire continuum of medical education. There is a need for tools that facilitate this lifelong learning, and a need to keep professionals up to date.

Additional concerns. Educational programs must have a focus on pediatrics and genetics, and physicians prefer interactive learning with case studies. The effect of the nursing shortage on patient education and informed consent was also raised. It was felt that

this point might raise the broader question of the impact with the emphasis on efficacy in our system might have on integration of a rather time intensive and complex issue, like genomics.

Recommendations from this third set of organizations. Ongoing, continuing education should be the responsibility of the licensing agencies and professional organizations. The development of profession-specific materials should be left to the health professions. The support for genetic education programs would be an appropriate role for the government.

Further recommendations. Funding is needed for new programs that feature educational practices known to change physician behavior. This includes areas such as learning based on perceived need, and interactive learning. Efforts in genetics education must extend to related areas of molecular medicine, and benchmarks should be instituted to inform and assess the bidirectional impact of translational research, i.e. from bench to bedside, and bedside to bench.

Recommendations. Education and training should address population-based genetic variation, and its utility in the emerging era of individualized medicine. The impact of genetic polymorphisms on the determination of "what is normal" in the era of molecular medicine, the scientific relevance and significance of minority participation in clinical trials, to the quality of health care in a merging era of genomic medicine, and the diagnostic importance of obtaining a good and complete family history on all patients.

Turning now to our second charge, which relates to the educational roundtable. The purpose and goals of this were to discuss in detail, the organization's efforts in and attitudes about genetics education and training, and to add to the information already provided in the surveys, to identify key concerns and barriers with respect to these issues on the organizational level, and to help inform any resolution coming from this committee to the Secretary.

The organizations that will be involved in this roundtable are listed here. Our representatives will be introduced to you formally later.

In the third area, we were asked to develop a draft resolution that could go to the Secretary with regards to education and training. The purpose is to arrive at a consensus of the committee on the issue of education and training, to convey this consensus to the Secretary, and to make recommendations or possible steps to address this important issue.

The conclusions that we had were that genomics can improve health, that adequate education and training in genetics and genomics is essential to integrating genetics into the health care system, and that access is contingent upon effective integration.

Additionally, the education professional organizations identified the following needs. There is a need for inventoried, widely-relevant clinical applications, a recurring theme, educational models that use such applications, and a broadened focus from genetics to genomics. There is also a need for appropriately trained faculty, and training programs that address genetics/genomics, and public policy.

With regard to our task force education resolution recommendations, there were six. We decided that while genetics is important and special, in that it is relevant to all areas of medicine and health care, it would be inappropriate to single it out in the educational setting. Rather, it should be integrated throughout all stages of learning, in all settings, and throughout all disciplines.

There is a need to support programs that enhance diversity among cultural competency of health professions. We need to engage other stakeholders in the process of

cataloging genomics applications to clinical medicine and public health. This third recommendation addresses the concern about the lack of clinically relevant genomics and genetics applications.

Fourth, we need to support programs that "train the trainers" in genomics and genetics education, addressing the identification of barriers to the lack of professionals trained to teach genetics. Fifth, we need to promote communication between faculty to enhance use of genomics educational models. And last, to encourage incorporation of genetics and genomics into the certification and licensure process.

Thank you.

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

At this time, let's take a 15-minute break. The members of the committee and our ex officios are invited to enjoy the refreshments here at the front of the room. For members of the public, refreshments and beverages are available at the gift shop near the hotel lobby.

We will reconvene in 15 minutes. That will be at about five of the hour. Thank you.

(Recess.)

DR. McCABE: If everybody could take your seats, please. We're now going to proceed with a roundtable discussion with the eight organizations on their efforts in educating and training health professionals in genetics.

The purpose of this roundtable discussion is to explore in greater depth the efforts of key professional societies and educational organizations to enhance knowledge of health professionals in genetics and genetic technologies, and to identify what steps, if any, need to be taken to advance these efforts.

At this time, I'd like to welcome our roundtable of participants. You've already joined us at the table, so I don't need to extend that invitation.

I'll now turn over the gavel to Dr. Reede, who will lead the discussion and introduce the roundtable members, or have them introduce themselves.

Joan?

DR. REEDE: Thank you, and thank you very much to those who are participating in this roundtable discussion.

I'm going to introduce them, and they're not in order. They're Joann Boughman, who is the Executive Vice President of the American Society of Human Genetics, a medical geneticist and adjunct professor in the Departments of Pediatrics, Obstetrics, Gynecology, and Reproductive Sciences at the University of Maryland.

Joe McInerney, who is the Director of the National Coalition for Health Professional Education in Genetics. He was also, in the past, Director of the Foundation for Genetics Education and Counseling.

Felissa Lashley, and Dr. Lashley is Dean of the College of Nursing at Rutgers, the State University of New Jersey. Previously, she was Dean and Professor at Southern Illinois University School of Nursing, and a clinical Professor of Pediatrics at the University's School of Medicine.

Dawn Allain, who is a genetic counselor for the Children's Hospital of Wisconsin in Milwaukee. She is a genetic counselor and clinic coordinator for the Cancer Genetic Screening Program at Froedtert Hospital in Milwaukee, and is the current President of

the National Society of Genetic Counselors.

Judith Lewis. Dr. Lewis is a professor in the Maternal Child Nursing Department at Virginia Commonwealth University, and also Director of Information Technology for the School of Nursing.

Georgia Dunston. Dr. Dunston is professor and chair of the Department of Microbiology at Howard University College of Medicine and founding director of the newly formed National Human Genome Center at Howard University. She is here representing the National Medical Association.

Toby Citrin, who is Director of the Office of Community-Based Public Health at the University of Michigan School of Public Health and Director of the Michigan Center for Genomics and Public Health. He is here representing the Association of Schools of Public Health.

Michael Whitcomb. Dr. Whitcomb is Senior Vice President of the Association of American Medical Colleges, and in the past served two terms as Dean at one, the University of Missouri Columbia, and the other at the University of Washington.

Thank you all for being here.

I'd like to open it up with a question for all of you. If you could speak a little bit to your key issues, concerns, and problems with respect to genetics education and training from the perspective of your organizations. Here, if you could in particular refer to gaps in current genetics education of health professionals.

Would one of you like to take it? Mr. Citrin?

MR. CITRIN: I'll take a stab at it. I should say initially at the outset that to my knowledge, the Association for Schools of Public Health, who I'm here today representing, has not a current survey of what the schools in fact are doing in incorporating genomics into education. But we did have a graduate student of ours do a survey based on websites of all 35 schools of Public Health to see the extent to which genomics has found its way into programs or curricula. I shared that survey with Amanda Sarata, for whatever usefulness it might have to your work.

Interestingly enough, of the 35 schools, ten of the schools have been stating that they now have genetics programs, so most of these are very research-related, as opposed to teaching related. Twelve schools are offering courses. Very few schools are identifying ethical/legal/social issues among what they are teaching. So the current scene, if that is an indication, suggests that the large majority of schools of public health really have not incorporated genetics or genomics into the content of the education of people going into public health.

This, in spite of the fact, or maybe leading to the fact that the Institute of Medicine's report on the teaching of public health that came out in 2003, "Who Will Keep the Public Healthy?" makes a strong plea for genomics being one of the eight content areas that all schools of public health should teach.

Now, with specific reference to your question of one of the barriers. The very fact that the recommendation of the IOM report is that these eight content areas should now be incorporated into the teaching of public health, rather than replacing what is now being taught, suggests an add-on to what is already seen by many in the schools as an overload of information to be taught to students.

Now, the answer to that question, of course, is integration. In fact, the

"Who Will Keep the Public Healthy?" report suggests that genomics needs to be integrated into an overall ecological view of causation of health and disease. But this suggests that people who teach epidemiology, biostatistics, health policy, and environmental health and health behavior, should all incorporate genomics in what they teach. Most of the people who teach these subjects do not understand the role that genomics plays in these areas, and the field has been so fast moving, that it is hard to have people who specialize in these, what are now identified areas of public health, to keep in touch.

So most of the schools that say they are teaching genetics, these 10 or 12, have specific, discreet courses that are typically electives, so that by far, students going through schools of public health today are receiving little or no genetics education. So the ability to have people who know how to teach incorporating genomics into the broader view of causation of health and disease, this would be a very major barrier.

The issue of time to incorporate this into what is already a very busy curriculum is yet another. Of course, related to that is the fact that there is no requirement to incorporate genetics, and it was good to see that accrediting bodies are incorporated into the draft resolution that you have put together.

So these, it seems to me, are some of the barriers. I want to add another barrier with respect to the one aspect of teaching, which is the ethical/legal/social issues. The very teaching of ethics in schools of public health is a hot topic these days. Most schools do not require training in ethics as a condition of getting a masters degree in public health. So if ethical/legal/social issues related to genetics are to be part of the required curriculum, which many of us who are in this field feel it should be, this then presupposes that ethics itself is a part of the curriculum of people going through schools of public health, it since becomes part of a broader issue. So that is at least one summary of some of the issues that colleagues of ours, and people in our school are seeing.

DR. REEDE: Thank you very much.

Dr. Lashley?

DR. LASHLEY: I'm here representing AACN, which is the national voice for baccalaureate and higher degree in nursing programs across the country. The membership consists of the deans and directors of those programs throughout the country.

AACN has had a few efforts in recognizing genetics as an essential part of nursing curricula. One of the first was to incorporate some knowledge of genetics in the 1998 "Essentials of Baccalaureate Education" document that was put out by the group. What this group does is put out essentials for the different levels of education that includes core content and core competencies for nursing at the baccalaureate and at the higher degree levels.

At the masters level, there was no such content included when that document came out originally, but that is going to be revised, and provides an opportunity for getting those essentials into those curricula as well, and that would be advanced practice nursing and nurse practitioner programs, and the like.

They have also had some conferences on incorporating genetics, one in which Dr. Collins spoke to the group, and I did as well, as far as applying genetics education into nursing curricula. So the group is moving on the need for that. What some of the barriers are are similar, I think, to what all the health professions are experiencing, and those are the intensity and density of the curriculum. There are a lot of competing topics for all of us in terms of what we put into our programs, and how much time we spend on them.

The second is that there has been traditionally a lot that relates to genetics in nursing programs throughout the years. But they tend to be more in the specialized, discreet disease-type areas, rather than a broad look at the influence of genetic and genomics across the curriculum, where most of us believe that that needs to be in terms of patient education, counseling, and that those things are not all that different for people with genetic disorders, but that the knowledge has to be integrated throughout the nursing curriculum.

Another barrier has been the lack of qualified faculty to be able to do that, and there have been efforts to address that. Summer institutes, NINR programs, in terms of building that out, but it hasn't suffused the programs, if you will, nationally. So I think those are some of our major challenges, is getting genetics through the life span in nursing education.

DR. REEDE: Thank you.

Dr. Lewis?

DR. LEWIS: Probably a nice segue from the same discipline, and I'm here today as President of the International Society of Nurses in Genetics. It is also interesting to me, having been a member of the former Secretary's Advisory Committee on Genetic Testing, to watch the continued growth and development, and I want to applaud you all for continuing along some of the lines that we felt were so important.

One of the things that I really think is important is when you talked about the lack of looking at genetics as exceptional. I think that becomes really important. If it becomes part of base knowledge, rather than considered specialty knowledge, that is very critical. I think of myself when I worked as an advanced practice nurse, and if every time I saw a patient with an elevated temperature, I called in the infectious disease specialist, we wouldn't get very far.

The infectious disease specialist certainly has a place in looking at patients who have complicated problems, but when I think we get to the point where every generalist is able to incorporate basic genetic knowledge, and those of us with specialty knowledge become like the infectious disease specialist, or the subspecialist, that that is when we'll know we've arrived. So part of what ISONG is working really hard to do is to ensure that all clinicians and all health care professionals, and in our case, we can only deal with nurses, but that people have a certain base knowledge.

ISONG has published in collaboration with the American Nurse's Association, scope and standards of practice of looking at what kind of knowledge is based on the NCHPEG competencies. But looking at what kind of knowledge we would expect a generalist to have, and what kind of knowledge we would expect a specialist to have, that model makes an awful lot of sense. And because we are a relatively small group of people who are specialists, part of our goal for ISONG is to work with our 2.7 million colleagues who are not members of ISONG, to ensure that people have basic knowledge, and that we're available as consultants and collaborators, but power is only powerful when you share it. So the idea of being able to share it is so that the knowledge base trickles down.

What are the barriers to that? The barriers are, I think the same ones that have been recognized in terms of competing priorities for all of us. But I also am old enough to remember when the new disease called HIV was discovered in the early '80s, and we got mandates to include that in the curriculum, and we sat there and argued over what was going to stay in and what was going to go out, so we could include information on HIV and AIDS. Somehow we have managed to do that, and I don't know of a health professions education that

doesn't include content on HIV at this point.

So I think it can be done, it is just a matter of making sure that everybody has the will. So I'm really pleased to see the certification and the credentialing bodies at the table, because I think that is where the push is going to come. Good will only goes so far, and mandates work much better.

DR. REEDE: Thank you.

Dr. Whitcomb?

DR. WHITCOMB: Well, from the perspective of medicine, it may surprise some of you when I say that I really don't think that the issue, in terms of being a barrier, really relates to how crowded the curriculum is. I think that the issues, as they relate to medicine, are two fundamental problems that sort of transcend all education, at least at the level of thinking about the continuum of education, including medical school, and then onto residency training.

One of those is simply how do you integrate content that should be integrated really across the curriculum in a way that makes that content relevant, important, and learnable? We are perhaps fortunate in one sense, because the movement in medical education reform, as it relates to the undergraduate, medical school curriculum for the past decade has really been to evolve towards an integrated curriculum, as opposed to the discipline-specific sort of departmentally-controlled courses that were typical of most medical school curricula.

This is an ongoing process with many schools involved, and it takes time to do it, but I think the challenge is simply then how do you integrate within the framework of that curriculum? I think perhaps the biggest challenge as we look down the road, and I think this will apply not only to medical students, but I think it will be an issue for residency education as well, is the opportunity for the learners to have exposure to patients that will make real for them what it is they are expected to learn, and how they will then apply their learning in the patient care environment.

That is an extraordinary challenge of an increasing severity simply for education of medical students at all levels. I think that with the changes that are occurring within the delivery system, it will continue to be a major challenge as we try to figure out how to get access to patients, the kinds of patients that students and residents need to be exposed to, since more and more of that care is being conducted outside of the traditional teaching site, which is the major teaching hospitals. So I really think those are the two major barriers right now for medicine.

DR. REEDE: Thank you.

Mr. McInerney?

MR. McINERNEY: Thank you. The barriers that everyone has discussed so far, apply across the board for NCHPEG's efforts. We have more than 145 member organizations now here in the United States, and abroad as well. Certainly we have a fair number of organizations that represent physicians, but the vast majority of our members are non-physician allied health groups, commercial organizations, consumer groups, and so on.

So one of our great challenges is to address the needs of this extraordinarily diverse membership. I should say that across the board, again, the greatest gap we face is the availability of evidence and educational materials that demonstrate to our constituents that genetics makes a difference in patient outcome, and can change effectively what they do now, rather than five years from now. We need to make that case more dramatically, we need the

evidence to do that, and we need the educational materials that can do that.

Another gap is providing more guidance about what to teach. You have heard referenced a number of times this morning for the core competencies, and I think they have been very effective. Lots of organizations have begun to incorporate the core competencies into the development of their own curricula, whether it is undergraduate curriculum, or continuing education.

Increasingly, we are getting questions about what should we teach then? What is the content that we should teach? I think the core competencies do a very nice job of saying this is where we would like your constituents to end up when they're finished, but what do we teach to help get them there?

This week, we're posting on the NCHPEG website a set of core principles in genetics that really are, we believe, core concepts, that most health processionals should be able to understand with respect to basic genetics. So that's another gap.

Another serious gap for us, and we're trying, believe me, we're trying, is the issue of diversity and cultural competence. How do we represent issues related to diversity and cultural competence more effectively in our programs, and how do we involve organizations more effectively that can help us do that? So any suggestions that anybody in this room has to help us do that more effectively, we would certainly appreciate.

The last gap for us, I think, is related to what I have just mentioned. That is, there may be some significant gaps in our membership. There may be entire constituencies within the health professions that we are not reaching, that we need to be reaching, and that we need to have involved in the development of our materials.

We have a membership committee that is looking at that issue now, taking a look at our membership and how it sorts out with respect to disciplines, and trying to figure out where we should place our efforts in that regard.

DR. REEDE: Thank you very much.

One of the recurring themes for many of you related to faculty, and the lack of trained faculty in genetics as a barrier. Do you have suggestions on how we could address this issue of training our faculty? Dr. Lewis?

DR. LEWIS: There are several programs that I know of that are available that might serve as models in terms of nursing. I believe one of the ELSI-funded projects was the project that Cindy Prows has out of Cincinnati Children's Medical Center that runs in two formats. One is web-based for 16 or 18 weeks, and the other is a summer, two-week intensive program that is basically designed to provide nursing faculty who have no previous knowledge, or have minimal knowledge in genetics, with what they need to infuse the curricula with genetics.

I took that a couple of years ago, and every year I get a follow-up server that said so, what did you do this year? You took this course and you used the government's money, and what have you done this year? That is one program that I know of that exists that was ELSI-funded, I believe. I may be wrong in that, but I'm pretty sure that is where the original funding came from.

Dale Lea at the Foundation for Blood Research has put out a group of modules that are available that you can order by mail that give faculty information that they can then use and adapt to their curricula. Then there are other programs like the National Institute of Nursing Research has the summer genetics institute that is an

8-week residential program for advanced practice nurses, and nurse faculty that deal with both education and research training. That one is really to help faculty with the knowledge development, as well as the knowledge transmission role.

I think there is some really good programs out there. The problem is that at least in nursing, they are relatively small. My class last summer at SGI was 20, and we were the fourth or fifth class. So in five years, they have trained 100 nurse educators. The Cincinnati program probably has groups of 25 twice a year, so I think there are programs there. Part of it is volume, part of it is taking people and finding people who are willing to do this, and then get back to their home institutions, and then have to convince curriculum committees that this material becomes important.

So I think there are models out there, but the question is what is the best model? I think a mixed model is best. Some people learn best in web-based, other people learn best when you take them out of their environment for a couple of weeks and turn off their pagers and their cell phones. So I think it is just a matter of what works best for individuals, but there are models out there, at least that I know of in my discipline.

DR. REEDE: Mr. McInerney?

MR. McINERNEY: Yes, thank you. I wanted to comment on one of the slides you showed during your presentation, and the comment that physicians prefer interactive learning with case studies. I can tell you from our experience with the NCHPEG membership that that applies across the board to all health professionals, not just physicians. We found that that is probably the best way to go, to engage people in the genetics content.

With respect to models, we have just finished developing in conjunction with a number of colleagues from the dental community, a program to train faculty in dentistry and dental hygienics. It is creatively titled "Genetics, Dentistry, and Health" on our website. Go to nchpeg.org and you can find that.

We have developed that around case studies with the intent of trying to get dentists and dental hygienists ultimately to change practice, to think a little bit differently about genetics when their patients come to them. But again, it is focused on case studies, with some core basic genetics.

DR. REEDE: Dr. Dunston?

DR. DUNSTON: First, just to set the record straight, I am a former chair of the Department of Microbiology as of March of this year. Also, I'm sitting in for Dr. Randall Maxey, the President of the National Medical Association. I just wanted to make a comment to your first point, and then the comment on education.

The National Medical Association promotes the collective interests of physicians and patients of African descent. It is the largest and oldest national organization representing the interests of African Americans, representing more than 25,000 physicians, and the patients that they serve.

The NMA is dedicated to keeping its members abreast of the many rapidly occurring advances across the various medical specialties, and the other changes affecting medical practice. So from that general perspective, certainly the organization considers genetics and genomics of paramount importance.

The National Medical Association was founded more than 100 years ago, and had its formation in the history of the American community. From that history, the subject of genetics becomes particularly important, because the subject concerns the inclusion of

African Americans, not just as a social imperative, but the subject matter forces the inclusion of African Americans as a scientific imperative, recognizing that the subject of genetics itself is challenging to our whole concept, construct, as well as methods of teaching.

Biology. We are moving from group-based kind of analyses, to individualized. Genetics as a discipline is often equated with the prospects of individualizing medicine. So there is a particular challenge that the NMA recognizes in a science that has the potential of individualizing medicine in a society that tends to group in its approach and methodology. So genetics as a subject that deals with biological identification, and classification of groups at all levels, is particularly challenging.

The issue, as was stated on one of your slides, is what does the science of genetics have to say about our biological thinking about race, race medicine, profiling in medicine, and developing drugs for groups when you are using a science that is potentially distinct in its capacity to individualize?

The NMA is sensitive to genetics as a subject that is very destabilizing in its whole concept with regard to the community. In the evolution of the science, the organization is concerned about the apprehensions of the community to become engaged in the potential benefits because of the historical context, in which the community, as well as the particular science, has been applied.

So a major barrier is how do we engage the community in such a way that the prospects to benefit become the driving force? To be proactive for engagement, recognizing that exclusion is not an option in the science of genetics? Because the science itself deals with the fundamentals of disease diagnosis, treatment, and progression in ways that not being included at every level of the science and its application in society is critical to how help will be administered to the group as long as we are part of this American community.

So the education of the community, as well as the professionals in medicine is of paramount importance, as reflected in the meetings at the local, regional, and national level that the organization sponsors in its continuing medical education, and its outreach to community. It is outreach to the community in ways that are complemented by current focus on faith-based organizations. Again, the history of the population is one where the engagement of faith-based organizations become perhaps a proactive means of addressing a subject that tends to be approached from a fear-based perspective.

So the challenge of how do you ensure that as new knowledge is coming forth, that you are represented, that your perspective is reflected, is paramount, and is certainly underscored. I would just say as one who is in academia, the role of education about the science, as well as the application of the science, and helping the community to appreciate that, that this is a science that demands participation.

It is a science that is challenging the reference point, and is challenging definition of what is norm. Inclusion of African people brings with it the challenge of variation and diversity in biology, and needs to recognize that what one sees cannot be separated from the context and the reference point from which you are viewing. How do we begin to incorporate that in our medical practice? That we have diversity as a strength, as an instructive commodity, not just a pathological or a basis of disease. That is my last point.

In trying to engage the community in terms of genetics and its impact, recognizing that the genome project not only led us to techniques and technologies for identifying single genes that have major effects in causing disease, but it now has brought us to

an appreciation of haplotype variation, and many common genes that vary, but are not pathological, just different in their functional capacities, and that they work together to accomplish functions.

So how do we begin to change the concept of medicine when the gene is not the pathological agent, but it is a means by which we can analyze, process, and understand disease, not always as a structural defect, but a regulatory one, and understand what is regulating the expression of genes that are associated with various diseases?

Thank you.

DR. REEDE: Thank you very much.

DR. BOUGHMAN: It is because of this change in science in general that the American Society of Human Genetics has been asking the question, as if we didn't have enough questions at the professional level and the health professional level, we have asked the questions kind of on the front end.

How can we work together better to improve on our answers to several of these questions? We asked ourselves, what resources do we have, and I'm speaking now as the umbrella organization of genetics, knowing that the American College of Medical Genetics, and the other medical genetics organizations would be here and be able to say some of the same things slightly more focused, but I'll speak in the broader terms.

What we bring to the table, if you will, is human genetics resources, in the form of individuals who have a knowledge base. What we have with the other organizations here, and as Dr. Dunston has pointed out, in the general public is a set of needs and questions that are being asked. So it seems to me, anyway, that we are now at almost the point of what I call genetics readiness across the board.

People are beginning to identify genetics as interesting, as important, it is appearing in the newspaper and so on, and now they want to ask the more practical question. We, the geneticists, are saying we have some information, and it is the gap in between. The geneticists need to become more teaching ready, and the organizations and communities need to tell us more specifically what they need.

At that interface, I believe is the concept of models or materials, as Joe McInerney pointed out before. And now we have had two tries at DNA Day, and each one has been better than the former one. This year, we got a couple of very interesting responses to our survey, and some terrific ideas on how to engage second and third graders in some movement, and some cutting and pasting kinds of activities that talk about differences and similarities, and so on, in a very practical kind of way. So some new kinds of things that we think can be put out there.

Exactly the same thing would apply in public health in our work with the American Academy of Family Practice in their genetics initiative and so on where the needs and the human resources are coming together in the form of models to tell us how we can be more helpful in the development of certain models, although we don't have the expertise to actually translate and put those onto the
CD-ROMs. We are going to need the resources together to get that activity and bring the other professionals in that need to focus on that. But I think we have the major portions covered, and now we just need the impetus to put it together and build the rest of the model together.

DR. REEDE: Thank you.

Ms. Allain?

MS. ALLAIN: Jo actually very eloquently said much of what I think NSGC's perspective would be on this. I think we are bringing to the table the human resources, we are at a number of tables now, along with ASHG and NCHPEG, and other organizations. Primarily NSGC has been focusing on in the last year, the allied health professions, by joining the Health Profession Network, which is an organization of allied health provider professional organizations, as well as joining the Association of Schools of Allied Health Providers. So trying to be the resource to integrate genetics curriculum into allied health provider education, as well as ongoing education for those professionals.

But as Jo pointed out, we have the resources, they have the interest, but it is getting the
evidence-based activities, developing case studies based around those specific disciplines, in order to integrate those into their curriculums and daily activities.

DR. REEDE: Mr. Citrin?

MR. CITRIN: For the schools of public health, I should note in terms of your question of what actions are necessary in order to train faculty, I should note the role that the Centers for Disease Control and Prevention has been playing in moving genomics more into the forefront of both research, particularly teaching, and practice connections with the schools of public health.

The Center, and of course, Dr. Khoury, is the resident expert on this program, and your committee, and nationally, has been funding three centers for genomics and public health, of which Michigan is one, and North Carolina and Washington are the other two. Part of our role has been, up to now, to advance knowledge of genomics in public health, and to further the interaction between the schools and the world of public health practice around genomics, and the integration of genomics into public health practice.

I should say one of the facilitating factors here is that at the same time that the schools of public health are increasingly trying to develop closer relationships with public health practice, genomics being in an early stage of expanding in public health practice, presents a wonderful model for the schools on how to connect with practice. Because the schools and practitioners can be learning about integration and expanding knowledge in the field at the same time, and can be keeping pace with each other, and performing the usual loop of research applied into practice, practice being studied by researchers. Genomics is just a wonderful model of how to do that.

It seems to me that the associations, and here in our case, our own association of schools of public health, can do a lot to facilitate the linking up of people within each of the schools who are carrying out these roles within the schools of expanding the teaching of genomics.

CDC and their centers program have certainly been supporting this, and they are about to expand their program of centers. They may not all be in schools of public health, but they're all going to be involved in public health teaching and practice, and sharing education.

All of what has been said about the increasing resources that are available for teaching needs to be paralleled, it seems to me, by advocates within the institutions of learning who not only carry out the roles of spreading out the need for teaching in their own schools, but need to be able to connect with each other. Here is where the associations can do an awful lot to form committees that connect people across, in our case, the schools of public

health who are trying to advance teaching of genetics in their schools, so they can be sharing lessons learned, the barriers, and how to overcome them.

DR. REEDE: Thank you.

I'd like to open the questions up to the committee now.

MS. ZELLMER: Does anyone have any comments on ways that practitioners, not students, that we can get the word out to them? I mean, I'm the parent of a child with a genetic disorder, and the frustrations in dealing with medical professionals, it seems like medical students, nursing students, you've got a captive audience, and that's probably the easier question.

You can teach them, and my experience was I've got a husband who is a physician, and I have got a father who is a physician. We saw four different physicians. The first time we were recommended genetic testing was by a resident. I think the problem I see is how do you get the people that are already out there practicing to realize the importance of genetics?

At least I would guess with continuing education, it is strictly voluntary. I mean, they are going to go to the things that they want to go to, and I think probably most physicians don't understand the importance of genetics. How do you encourage them, or force them to learn what they need to know about genetics?

It seems we have the materials, but how do you get them to use them?

DR. REEDE: Dr. Lashley?

DR. LASHLEY: I would make a comment in regards to nursing in that regard. In some states, continuing education is in fact a part of relicensure, or for recertification for specialists, depending on your specialty. But I think the aspect that we briefly touched on before, which his requiring a certain amount of genetic knowledge, much in the same way that HIV knowledge is part of mandated, continuing education in many states would be one way to unfortunately, one has to look at driving curriculum sometimes by licensure and certification aspects. That is one way to do it.

Another way is to go through the specialty of continuing organization's genetics in each of their programs, whether it be as a feature by itself, or integrated within such things as MI, if it is a cardiology conference, updates in that, or whichever way that goes.

DR. REEDE: I'd just like to acknowledge that Dr. Reed Tuckson, a member of the committee, has now joined us. Welcome, Reed.

DR. TUCKSON: I'm listening carefully, and I will chime in if I get a chance. Thank you.

DR. REEDE: Dr. Boughman?

DR. BOUGHMAN: Let me put on a slightly different hat for just a moment. For the last 14 years, I have been a representative from the American Board of Medical Genetics to the assembly of the American Board of Medical Specialties. There have been two or three things that have happened in the last five years or so in that organization. Coming back to the comment that Dr. Lewis made earlier about statements and accrediting bodies being extremely important.

One of the things that has happened is the shift in continuing medical education to required maintenance of certification. This is a formal shift, this is a difficult shift for many of the specialties to make that once had the attitude, once I pass my boards, that's really it.

So we do have a window of opportunity here to in fact include genetics as at least recognizably new information. Our challenge is to make it the most exciting among the various kinds of continuing medical education courses, or whatever, that individuals can take. So it is a two-way street there, certainly.

One of the other comments that I would like to make is that as the genetics representative to the process of creating the principles of practice in training and in maintenance of certification, I think it was the approach of the geneticist and our tradition in number one, looking at family units, and number two, fully recognizing and addressing cultural-specific issues and the need for cultural competence in every clinical situation.

I think our experience in that helped bring that to the fore, and I think that may be the results of the way, if you will, geneticists kind of look at the world at the same time we're looking at individual patients. But we do have a window of opportunity here in maintenance and certification for physicians.

DR. REEDE: Dr. Lewis was next.

DR. LEWIS: One of the things that we've been working at really hard with ISONG is to make people with specialized knowledge available for professional meetings. For example, I'm speaking with three colleagues, one of whom I believe became interested in genetics because she has a child with a genetic condition, and the nurse from NIH who is her research nurse, and another colleague and myself, the four of us are presenting at the American Nurse Association, which is probably the biggest generalist nursing organization.

Dr. Lashley is presenting there too this year, and so we're working really hard to present at non-genetics conferences, and to be able to work with our colleagues. Now obviously when I'm speaking, I'm one of 27 break-out sessions, and how many people are going to choose to go there versus retirement planning, versus how to take care of the patient in the intensive care unit? That becomes a hard issue.

I think part of what happens is that as people are exposed and learn from people like you, because I learn most of what I learn not just from books, but from the patients that I see every day. So as I interact with people like you and your family, that hopefully will pique my interest in that this is an area where I need more education. So when I go to my professional meeting, I'm going to choose the genetic session over the retirement planning session, or whatever.

So that I think it is a two-way street. But to forget that patients are probably our most valuable source of education, and that is not helpful as a patient when you're going in to realize that you're as responsible for educating the clinician as the clinician. So if you look at it as a partnership, I think sometimes that becomes real helpful, too, and that each of us has an opportunity to help educate folks.

I'm sure that the folks that you and your family dealt with learned as much as you did, it sounds like, but that becomes an important partnership, I think, too.

DR. REEDE: Mr. McInerney?

MR. McINERNEY: Yes, thank you. Professional societies are an extremely important vehicle for reaching out to health professionals. I'll give you four concrete examples, one of which Ms. Allain can describe better than I. But the National Society of Genetic Counselors worked a couple of years ago with the Endocrinology Society to integrate genetics thoroughly into its annual meeting. I don't remember how many members of NSGC were there, but I think there were at least 20 members of the society who were at that meeting.

They actually constructed genetic counseling sessions around a particular genetic test for the endocrinologists, so they could come in and do hypothetical genetic counseling sessions. The point here, and you'll hear it again, is to work with the professional societies so that you are meeting their needs.

NCHPEG will be working with the National Black Nurses Association to develop a half-day workshop on genetics for its annual meeting in 2005. Beginning this year in October with its annual meeting in Orlando, the American Academy of Family Physicians will begin a
year-long clinical focus on genetics, and there are lots of organizations involved in this.

The Genome Institute, I think CDC is involved in one way or another, and that will continue for the year. But again, we worked with the society, the Academy, to determine what their needs were. I thought it was very interesting, when the group of geneticists sat down to develop 12 modules, one per month for the clinical focus, when we looked at it, it was what geneticists would come up with.

When we looked at what the interest of the family physicians were, there was almost no connection, except for the fact that each of the diseases, and they were all disease oriented, each of the diseases has a genetic component. But the importance is to work with the professional societies to build those programs. So there will now be one module a month rolled out by the American Academy of Family Physicians in conjunction with a number of us around this table, to bring genetics to family physicians once a month over the year.

We have also just finished working with the American College of Clinical Pharmacy to develop a new continuing education program on pharmacogenomics. And again, they came to us as a genetics community, and we have been working with them now to bring the genetics that they need to that program for their members. So professional societies are extremely important.

DR. REEDE: Thank you very much.

I have a general question. As I'm hearing about these various forms of curricula, the cases, the different materials that are being developed, is there a mechanism in which those can be shared? Part of my concern is everyone is starting from scratch every time.

MR. McINERNEY: If I may, that is why NCHPEG exists. If you come to our website, you'll see a list now of about 55 different educational resources in our database, about ten of which we've had reviewed. Two reviews usually, one by a member of the profession for whom the materials are intended, and the other review by a practicing geneticist.

There are also lots of other resources on our website, but that's why we exist. If you have materials that you would like to bring to the notice of the community, please let us know, and we'll link to them, or we'll put them on our website.

DR. REEDE: Thank you.

Dr. Felix-Aaron, you had a question?

DR. FELIX-AARON: I didn't have a question, I had more a comment on what Kimberly said in terms of trying to reach practicing providers. Somebody said earlier in terms of the window being open, I think that is right. But I think in addition to the window being open, sort of being strategic about the types of providers you engage, would be something that would be important for this group to consider, if indeed that was the direction that it wanted to go forward.

So I think it is important that the family physicians are coming to this issue,

and they have that particular focus. But I think there is also a role for the committee in terms of understanding the science, and understanding the progression of the science, and with the progression of the science, which types of providers, whether they be physicians or nurses, are at the forefront of the evolution of that science? Which organizations we need to target as a committee, and which professional groups that need to be targeted as a committee.

So just to summarize my main point, is that clearly there is a need for targeting in terms of the providers that are on the forefront. I would imagine that the cardiology is important to engage the cardiologist, that the pediatricians, I mean, the state of the science is such that engaging the pediatric community would be much more of a priority than say engaging someone with subspecialties.

DR. REEDE: Thank you.

Dr. Leonard?

DR. LEONARD: So on a national level, CME is, I think, directed on a state level. Is there a national mechanism to mandate CME in, or continuing education? I call it CME because I happen to be a physician, but continuing education, professional education, for all groups at a national level, rather than doing it state by state, or organization by organization?

DR. REEDE: Dr. Whitcomb?

DR. WHITCOMB: The simple answer is no. Licensure is granted by states in medicine. As a matter of fact, not all states even have requirements for any continuing medical education. About one-third of the licensing authorities do not, and of the states that do have requirements for licensure, or relicensure I guess I should say, very few of the states mandate any specific content that needs to be covered as a part of the relicensure.

There are some that do, but as a general rule, the answer is no. But if one wanted to think about this across the country as a whole, the answer is there is no mechanism in place to accomplish that.

DR. REEDE: Brad?

MR. MARGUS: Thanks. So the people in this room obviously, I think, all concur that genetic education is important. I feel a little bit like in several other meetings, too, everyone is preaching to the choir, there are all these reasons why science is changing, and people need to be kept up to date and all that.

I'm a little unclear about what genetic education means. If it is how genetics works, so that a physician, nurse, or counselor has to be able to explain Mendelian inheritance, or what the risks are of a particular test they're going to have, or if they are supposed to, as in Kimberly's case, know the catalog of all the rare disorders and be able to help in diagnosis, that is a completely different and much more challenging thing. Or is it the ELSI part that we always hear about?

But either way, it is pretty clear, and our resolution is going to say that education is really necessary, it has to be coordinated, and all that. I think, though, because if this committee is really going to have any impact, it is kind of like the Genetic Discrimination Act, which is we're going to have to tell the Secretary that something is definitely broken.

I'm not really hearing that something is broken, so maybe things aren't broken, and certainly if you look at diversity among genetic counselors and things like that, we can go down there. But is there something that you can point to that it is really broken? Can you bring a large number of patients or families in front of us, or in front of the world and say, these patients were mistreated because their physician was clueless about genetics, or got it

wrong?

I haven't heard any of those stories. I have only heard that kind of like we're anticipating that this is all coming, and we'd better be ready, and we only have one genetic counselor for millions of potential patients who are all going to be screwed up if we don't have more educated people out there. But today, is anything really broken? If it isn't, it is going to be challenging to get more funds in the things that we're asking for?

So if the people who are going to continue to make comments could just highlight for me anything you can point to that is really, really broken, it would be really helpful. Again, if part of the genetic education and continued education is things like knowing everything about rare diseases, I know from my experience as an advocate, I could bring forward thousands of families who, like my family, spent a year and $60,000 of needless tests to find out what the real disease was that my kids had, because the physicians at world class medical centers and the geneticists didn't think of the right disease to test for.

But other than that, other than knowing that kind of information, which I'm afraid maybe it is always going to be a challenge with rare disorders, all the other things that we're talking about is part of genetic education. Is it so bad right now that things are really broken?

MS. ZELLMER: I just want to make one comment, though, Brad. I certainly don't expect every physician to know every rare disorder, and I think you're right, that would just be a useless resource. What I was looking at more is just the fact that a physician should recognize that there could potentially be some genetic basis for the problems that the child is having.

Certainly my child's disorder is rare, but if you look at the symptoms that she was having, in looking back on it now, I think it should have been something very obvious to a physician that genetic testing should be recommended to us, that there were potentially genetic bases for the problems, and certainly not that she had this specific disorder.

I think that it was very clear to a resident who saw her for one time, well, you know, have you ever had genetic testing? I think something is broken when you have several doctors, and it takes them two years to even recommend genetic testing for a potential rare disorder. Not that they know the specific rare disorder, but that genetic testing is even recommended as an option. That would be my recommendation. I think something is broken if it takes two years to even have genetic testing recommended.

DR. REEDE: Mr. Citrin?

MR. CITRIN: Let me suggest, and I'm delighted that question got put on the table, because one of the comments when we got around to any specific comments on the draft resolution, one of the comments that I was going to make was that the resolution could carry more of a sense of urgency to get on with this activity.

Let me suggest three areas where it is not so much that things are broken, but that things are, at this point, moving towards what could be very significant crashes. One is in the area of costs, that when you look, for instance, at genetic testing, here is an expanding technology as to which the corporate sector, the
profit-making sector has an incentive to get more and more tests used, far beyond what the scientific sector is able to demonstrate is effective, valid, and cost-effective.

So the ability to teach people who are professionals on how to assess and evaluate the worthwhileness, the validity of this expanding technology, is absolutely critical. If

people, whether they are medical professionals or public health professionals, don't have this ability to assess the worthwhileness of this vast array of exploding technology, what we'll end up with is an enormous wasteful use of health care resources.

Secondly, there is this whole matter of distortions that are occurring in the public's view of genetics, as the result of where the public is getting most of its information from. The public, of course, is getting most of its information on genetics, as well as it does other subjects, from TV, from media generally, relatively little from health care professionals, and almost none from public health.

The messages coming to the public now are both deterministic in terms of media hype on genetics controlling her conditions, which it really doesn't, as well as an item which is on your agenda, fortunately, and that is the direct-to-consumer advertising, which will further confound the public's view of what it means to have this or that gene.

And so here again, it seems to me we need a cadre, an increasing cadre of professionals who understand how to inform the public about genetics. This is a particularly critical role for public health people, because we see ourselves as having a role of public education, and here again, this happens to be one of the major things that CDC has been emphasizing in their new announcement of funding of genomics centers, this role in trying to correct distortions in the public's mind.

The third area where we could be heading toward a serious crash is one that is directly relevant to what Dr. Dunston, at my left, was talking about. That is the fact that we have here an expanding science that has the potential to exacerbate health disparities, at the same time that it has the potential of reducing health disparities. It is uncertain at this point as to which direction it is going to travel.

How our professionals learn about genetics, how they use that learning informing practitioners how to practice genetics, and how, again, they inform the public about what is the meaning of genomics, will have a lot to do with whether this new technology is going to create further gaps between the haves and have nots, further stigmatization of groups that have already been stigmatized too much, or whether genomics as a new, powerful tool to address some of the diseases that are responsible for which genetics has a component, and diseases that are responsible for health disparities, whether this very powerful tool will be used to reduce disparities.

So this, it seems to me, would suggest at least three areas where action is absolutely essential if we are not going to see this science lead to crashes in the future.

DR. REEDE: Dr. McCabe?

DR. McCABE: I want to make several comments about several of these points. First is one that I don't think I have heard here, and if I did, I missed it. That is in preprofessional education, and I'll focus on premedical education, since that is what I have more experience with, and the whole concept that we should really equip our professional students when they arrive in professional school with some background in this area.

The topic that I find particularly appalling in premedical education is the fact that we still require organic chemistry. I quite honestly have not formulated a medication once in my career. It is simply there as an energy barrier to premedical students, and yet, something as important as genetics, or one could pick another topic that has relevance to 21st Century medicine, is not required.

I would urge that in each of these areas, that we begin to look at what the

prerequisites are, so that our students come understanding the importance of these areas. With respect to the integration of content that was mentioned, I think it is important to note that some of our revamping of medical school curricula are treating genetics as a threat that runs throughout the medical curriculum, not as a specific topic.

I think it is important that we make it fundamental and exciting, and case-based on all of those topics. The degree with which we are successful does have to do with the politics of curricula, and arguments about crowding of the curriculum. So again, to the extent that organizations such as the AAMC, or other professional organizations, can encourage that this be a part of the curriculum, that will help those of us who do have to deal with the realities of fighting for space in the curriculum.

I think it is also important to note the cost benefits of genetic education. It does prevent the diagnostic odysseys that have been mentioned, which in fact are cost-beneficial. Off site, several examples with which I'm familiar where these diagnostic odysseys have occurred with delay and treatment of management of patients.

PKU, the classical newborn screening test, screening of screening, not every child will be identified, and those that are missed frequently go on for years before somebody suggests that the appropriate test be done. There are numerous examples of hemoglobinopathies, sickle cell disease, and thalassemia.

Again, relatively common disorders, and certainly quite common in specific communities in this country, that are not understood really by practitioners. Cystic fibrosis is another case where individuals can go on for years and years before they are identified.

Finally, we need to make sure that all of our professional students understand that this knowledge will prevent medical/legal mistakes, and medical misadventures. There is a big focus on medical errors these days. We talk about drug errors and all of those things, but errors of omission, errors of lack of diagnosis, or misdiagnosis, are also extremely important, and we have to recognize that not only are those cost inefficient for the families, they lead to tragedies where diagnoses are not made, or made too late. But ultimately, they also cost society, both in productivity, as well as in real dollars for those diagnostic odysseys.

So I think that there are a number of ways that we can approach this. If we don't do this in the health care professions, then our colleagues, some of whom are sitting around this table in the legal profession, will force us to bring this to the fore in health profession education. They will point out the misadventures, and the costs of those misadventures will become even higher with medical/legal actions.

Thank you.

DR. TUCKSON: Can I get put on the list, also? This is Reed. Thanks.

DR. REEDE: Reed? Go ahead.

DR. TUCKSON: Oh, okay. One of the things I also found valuable, the phrasing of the question -- can you hear me okay?

DR. REEDE: Yes, we can.

DR. TUCKSON: I thought it was valuable, the phrasing of the question around what was the problem, what was broken, what do we fix? I think two things. One, based on what Ed McCabe just said, that the Secretary of Health has a new initiative around health information infrastructures. I would wonder whether or not we might be able to have some relationship, or propose some initiative with that new health information infrastructure

task force to find ways in which we can support the availability of the best evidence-based science, as it is continuously updated, in an easy and accessible way for a clinician.

Obviously, nobody could possibly keep up with all of those permutations of information and knowledge in this field. So what I'm hoping is that somehow, one of the solutions may be that we could find a way for the Secretary to use this information task force as a potential solution to the availability at the point of care for people as these information systems are standardized, for both the outpatient, as well as the inpatient environment. That might be an idea.

Secondly, I think that the point made earlier about the maintenance of certification is exceedingly important, because at the end of the day, it is what you hold clinicians accountable for in terms of their criteria for certification. I think they, like the MS, is an essential group of people that we need to bring in.

Lastly, I would hope that maybe we would discuss later in this meeting, some things on HRSA. We will get to the idea of performance assessment. When you hold people accountable for their performance, that becomes perhaps that national unifying standard that someone asked earlier about, for getting their attention. So that as we start to get to performance assessment and it starts to get into the area of the use of technologies for genetics, then I think you provide a fertile environment for people to want to access the best appropriate evidence-based information, and then apply it. That provides, I think, a stimulus to go forward.

Thank you.

DR. REEDE: Thank you very much, Reed.

Sarah, do you have some comments on the committee that Reed was referring to?

MS. CARR: It's just to point out that this would be a very opportune time to make a suggestion to the Secretary about that. The President has actually asked for a strategic plan on the use of the improved use of health information technologies. So it would be an opportune time.

I do think that the more specific the committee can be about how that technology and genetics would dovetail, I think the better. That would be very helpful to the Secretary, I think.

DR. REEDE: Thank you.

Ms. Berry?

MS. BERRY: I'm not trying to stir anything up here, but I was curious in reading the different comments on this issue of education and training of health professionals, I was wondering whether there is any disagreement about who does what?

The reason it is a concern to me is because in the area of coverage and reimbursement, which we're going to talk about later, I know who does what will have some impact on whether an insurer, or whether a federal health program, will cover and reimburse for a particular service.

So I didn't know if we needed to go down this path or not, or whether everyone just prefers that we ignore it and have everyone sort it out amongst themselves. But that was an issue that popped into my mind, and I would love to hear your thoughts.

DR. LEWIS: I just want to speak briefly. Back before when we were talking about credentialing, and you basically said there were no national standards in medicine, that's not necessarily true in all of the disciplines.

Any advanced practice nurse is required to recertify every three years, and the recertification usually has both an educational and a clinical practice component. While I may not be required to do mandatory continuing education for the State of Virginia to keep my license, one of the things I have to do as an advanced practice nurse is I have to maintain credentials as a certified nurse, and that requires me to have 45 hours of continuing education every three years, and a practice component that is at least equivalent to a half a day a week of patient care, depending on my particular discipline.

Some states, Massachusetts, for example, requires 15 hours every three years for all nurses to maintain their licenses. So I think that there is variability among the professions, but to look at the fact that there are windows of opportunities, at least for some disciplines to start to look at the fact at, at this point, for example, as a women's health nurse practitioner, of my 45 hours, 30 of those have to be core, and 15 of those have to be supportive, but 30 of the 45 hours have to be clinically based.

So that there are opportunities in some disciplines to produce programs that would be attractive to people, and have them out there, and it may well be variable. Even though people are licensed by the state, in nursing, the credentialing is national.

DR. REEDE: Thank you very much.

Dr. Khoury?

DR. KHOURY: Thank you very much for all your presentations this morning. I may be a bit out of sequence, I have been gathering my thoughts for a little while here. I have a couple of comments, and maybe a question of the group.

We all talk about education and training, and I think many of the issues I heard this morning are right on target. There is sort of a two-prong approach to this, at least in my own mind, and I may be saying those same things tomorrow morning when I talk about the public health approach to genomics.

The first thing to keep in mind is the issue of genetic diseases, and the rare conditions that individually may be rare, but in aggregate, may affect about 5 to 10 percent of the population. We have heard about the diagnostic odysseys that cost money, cause anxiety in families, and destruction of the social and familial fabric, but also sometimes leads to medical issues.

Dr. McCabe mentioned cystic fibrosis, and I can count a number of other conditions where there are interventions. One example comes to mind, that is familial hypercholesterolemia, which is an LDL receptor defect that leads to premature heart disease, and people die in their 30s and 40s from heart attacks. That is about 1 in 500 disease, about a million people in this country may have familial hypercholesterolemia, and there are data from population surveys that about half or more of these patients are missed by the medical system, because there are so many people with high cholesterol levels due to other more polygenic or multifactorial causes of high cholesterol levels, that people with LDL receptor defects may be missed completely.

So if not for anything else, we need the kind of red flag raising. We are not trying to get everyone to become a geneticist obviously, but it sort of raises a red flag so that appropriate follow up can be made.

But coming back to what I heard also throughout the committee, and I think Dr. Whitcomb and others mentioned the issue of relevance today. Because for most practitioners, these kinds of incidents may be too far and too few in-between, I guess, they may

not be seen by one single practitioner on any given day, week, or month. So what else is going on? I want to paraphrase Judith Lewis' comment about the fever.

What is the equivalent of fever, a febrile episode in genetics? I mean, you made the analogy of the infectious disease specialist, so okay, you've got the patient with fever, and you're not going to refer everyone who shows up with a fever to an infectious disease specialist. Fever is a very common occurrence in the population. So what is the equivalent of that in genetics? To me, the equivalent of that in genetics is the occurrence of something in your family.

We have plenty of data that shows that at least the major killers, heart disease, diabetes, and cancer run in families. If you have at least one affected relative, you are at increased risk of these conditions, and they may not be due to a single-gene disease, and you may not have to refer every single patient with a family history to a geneticist. And yet, it is the equivalent of a fever, because half the population may have a family history of something, and that becomes sort of the run of the daily practitioner. Therefore, the relevance of genetics/genomics/family history becomes of urgency to the general practitioner, in addition to the issues of rare single-gene diseases.

I'll talk tomorrow about the public health approach to family history we started, but I'm curious to see what the various organizations are. I know some of them have done a number of things in this area. I'd like them to expand on the equivalent of fever in genetics, things like family history and the development of tools that everyone can use today, and we don't have to wait 10 years to show relevance of genetics in practice.

DR. REEDE: Would someone like to respond to that question? Mr. McInerney?

MR. McINERNEY: Yes. NCHPEG develops, three times each year, a family history newsletter that is devoted to exactly the issues that Dr. Khoury is referring to. This actually was a bit of a retrenchment for us. Originally, we were supposed to develop a generic family history tool for use by all health professionals, and it simply proved to be impossible with the time and the resources we had available, so we backed off on that a bit. Our family history working group spent a fair amount of time on it, but concluded that it was an impossible task.

What we do now, however, is develop this newsletter that we put online three times a year, devoted entirely to the issue of the family history in health care. It has articles about family history, but it also refers to tools that other people are developing, such as the tool being developed by the American Society, the Genetic Alliance, and NSGC, and the tool that CDC is working on now.

So in fact when we talk with our colleagues in the health professions, we refer to the family history as the first genetic test. It is inexpensive, and it is relatively easy to do. We are also building an extensive section on family history into our new CD-ROM on the genetics of common, chronic disease, which will be out later this year, and that is intended for primary care providers and public health professionals.

At some point, I would like to respond to Ms. Berry's question about is there an agreement about who does what, and who is going to pay for it. But I'll come back to that, perhaps.

DR. REEDE: Thank you.

Dr. McCabe?

DR. McCABE: Well, I just wanted to respond with an anecdote about the family history. In retooling our medical school curriculum, we had a laboratory in how to take, and how to interpret a family history. We were told by the organizers of the curriculum that that was not interesting, and was something that we really needed to completely rethink the next year.

On the other hand, I would point out that I'm Chair of a Pediatrics Department, I attend morning report whenever I can when I'm in town, and I'd like to think that our residents are more sophisticated in a family history now than a few years ago. They used to say, is there any family history? And now if one of them says that, the others will chime in with specific questions about what that is, rather than just do you have a family history of anything, which used to be the question, which the usual answer is no, or something completely irrelevant.

So I think that it is somewhat analogous to taking the temperature, but it requires a little more sophistication than simply taking a temperature. And yet, to many non-geneticists, it is considered boring, trivial, and uninteresting.

DR.. REEDE: Thank you.

Dr. Felix-Aaron?

DR. FELIX-AARON: I just wanted to elaborate a little bit on the Department's health information technology. AHRQ, the Agency for Healthcare Research and Quality, is leading part of this effort. We funded this year, $60 million dollars in grants, specifically to do two things. One, to build the infrastructure on IT in small community hospitals, as well as in rural settings, and the other is to demonstrate the value of health information technology to health care.

So I think we have the opportunity to look at this really closely and to see where the opportunities are between what is going on in genetics/genomics, and what is going on in IT. If this committee was interested in that, I'd be happy to go back to AHRQ and look at the portfolios we have, and whether there were any grant projects that specifically looked at genomics, and I'd be happy to report to the committee on whether they were interesting programs, or interesting projects that was directly relevant to the work of this committee.

DR. REEDE: Thank you very much.

Dr. Guttmacher?

DR. GUTTMACHER: I'm just going to state the obvious. I think in terms of linking the number of the comments that we've heard, one, the range of comments about the importance of focusing on those areas of genomics that are actually usable and useful today to the provider. Two, this calls for thoughts about how we might feed into the Secretary's Committee on Health Information Infrastructure, and three, the importance of family history.

That really is the genetics tool that we can offer today that doesn't add cost, but in fact we think it is of some benefit, that reminds the practitioner of why genetic factor is important in health, and can be used as the basis for a lot of things. There is work that CDC is specifically doing, but others are doing as well to really further the idea of coming up with instruments and other kinds of things.

It seems to me it would be useful for the committee to call attention of the Secretary and his other committee to the potential use of family history, and the electronic medical record particularly. If we could establish the idea that everyone deserves a good, relevant family history and/or electronic medical record, we will have done a good thing for

patients, but we will also have established a template for providers to continue to think about other more sophisticated genetic testing and those kinds of things once they do become more broadly available and useful. So I would think that would be an obvious sort of first step to take.

DR. REEDE: Thank you very much.

Dr. Winn-Deen?

DR. WINN-DEEN: I think I greatly agree with the comments that Alan just made. I think one of the issues that keeps coming back to me about genetic testing, and I'm not talking about diagnostic genetic testing in the context of signs and symptoms, as much as presymptomatic testing, or maintenance of information about carrier status.

A lot of this information might be lost between the time a test is done, and the time the information is actually needed at some future date. I think it is very important that we, as we start to consider moving into the electronic age and doing electronic medical records, and having an IT infrastructure for that, that we have family history, we have a mechanism for recording any once in a lifetime genetic test that someone is given, so that that information is there. I'll say that extends to something as simple as blood type, and to things as complicated as pharmacogenetic drug metabolism enzymes that might be useful at multiple times in a person's lifetime.

So I would be very interested as a committee in hearing from this other group what they are doing, and how we could integrate these things together, because I think the combination of those two activities will help us to integrate genetics into the practice of medicine.

The third point that we have to lay on top of that is that there still are concerns, and Agnes mentioned this to me at the break, about having genetic information in your medical records, that some institutions still maintain your genetics record as a separate parallel file from your actual patient record.

So we need to deal with that issue. Why is that done still? Is it still an issue of fear of discrimination in some way in insurance, health, life, or employment? Or what is the reason that that is still happening? That is clearly a barrier to having a really fully integrated lifetime electronic medical record that can bring you the full benefits of a genetic test, rather than having to sort of hide that information, and then have it redone every time you need it.

So that won't deal with the situations where there is no family history, and we still have to train medical professionals to recognize in the absence of family history, which is what happens in a lot of rare genetic disorders, the probability of two carriers coming together is low, and so quite often they don't have any family history, but then they end up with affected children in recessive disorders. So I think both of those things are very important for us to keep in mind.

DR. REEDE: Just a question as you talk about this information, and are there questions with regard to confidentiality and sharing of information, it reminded me somewhat of some of the comments from Dr. Dunston, and the public's view of this information, how it gets shared, and concerns from special populations in terms of how this information gets shared. Could someone speak to that, please? Dr. Lewis?

DR. LEWIS: It's real interesting in terms of what people want in their medical records or not. I know when I deal with the women that I work with, for example, lots of folks don't want their sexual orientation in their official medical record.

So I think when we start to look at information that is different information that is potentially labeling, it is genetics information, and it is a whole lot of other information, too. I'm not sure all of the privacy protections that we have in law that are so challenging to implement, necessarily help people in terms of the fear of what is in their record, or what is not in their record.

I guess I worry about the fact that we have a system that has created the need for people to want information to be excluded from their medical record. I know several years ago when I went to Iceland and was sitting and talking to the President of Iceland about the issue of privacy and confidentiality, he basically looked at me and said, you know, that is not an issue in our country, because we are a democracy, and people trust the government.

I thought, wow, how unique and how wonderful. For whatever reason, we have created a climate of distrust in this country. Not us sitting around the table, but the system has created a climate of distrust, and of people's need to keep private information private. So I think the problem is even more than genetic information, I think it is an information of the fact that people have been treated badly.

Once people have been treated badly, or groups of people have been treated badly, then we have a lot of work to do, because once trust is lost, it becomes twice as hard to regain. So I do worry about people who have concerns about keeping information out of their medical record, and why.

DR. REEDE: Thank you.

Dr. Hans?

DR. HANS: Thank you. I just wanted to extend some of the ideas that have been put forward around where this committee may want to consider interfacing with the Secretary's Health Information Technology Initiative. From the lessons of a department that has an integrated health information system, electronic health records available at the bedside for patients, some of the issues that you may want to consider along the following lines.

One is certainly proposing what elements of genetic knowledge should be part of the basic components of the electronic health record. That is one of the decisions going on across the agencies at this time. Family histories could certainly be something that is considered, if you are out there as a technology developer, you should certainly put that capability into whatever system you are developing.

I think it would also be helpful to look at the other kinds of tests and knowledge that should be part of the basic record, and part of the recommendations that this Department and others are looking at at this time.

In addition, in a more future looking perspective, to think about once folks have records at the bedside and have the interactive capacity with those programs, what sorts of prompts and guidelines on a daily basis would be helpful for the practitioners gets a little bit at how much knowledge does every practitioner need to have?

If you can build in some of that knowledge and reminder to the automated system, then you don't have to make all the effort to make sure that every practitioner knows every bit of information. But if certain diagnoses and conditions come up as they are put into the record, if there is an electronic prompt that then says what about this, or consult a genetics expert, you may want to consider this, that would be helpful in helping this entire enterprise around health information technology begin to map out where they may need to put that kind of information into the records over the next five to 10 years I think would be extremely helpful,

particularly from the professional societies.

Certainly that is the role many other professional societies are playing at this time in that arena, as providing information and guidelines that then can be incorporated into these electronics systems.

And finally, an area that I think both for genetics/genomics, as well as the rest of the fabulous knowledge that is coming out of NIH at this time is how do you incorporate knowledge management systems into a desktop electronic medical records system?

What do I mean by that? Well, you can't overwhelm a practitioner with all sorts of prompts and guidelines. We certainly found within the VA that after sort of a certain number of prompts, that physicians start to turn off the prompts. So in any one visit, you can't have too many things coming up on the screen at the same time. But you can think about a way where when you have a particularly difficult case and you're not able to diagnose what is wrong with the patient who is in front of you, if there is access on that desktop to information, ability to query based on the diagnosis in front of you, it can help physicians and other practitioners seek out information, and perhaps assist them in understanding sort of the conditions of the patient that is there in front of them.

So those are just some ideas that the committee may want to consider in this interface with the Secretary's committee.

DR. REEDE: Thank you very much.

Dr. Feetham?

DR. FEETHAM: Thank you. I have a number of things, and some of it goes back to the broader issues brought up in the original presentation, which I'd like to commend that you looked at what is common across the disciplines, because this is truly an interdisciplinary issue, and that we're really talking about quality of care and access. The way your report was framed, I think that comes forward as very important.

Many of the things that you brought forward are consistent with a report with recommendations we'll be taking to Dr. Duke, the head of HRSA, in just a few weeks, after doing an analysis across all of our agency about genetics activities.

But what I'm hearing is a key issue of communicating what is available to support the integration of the genetics to practice research and education. Again, part of what I see in the role of HRSA in working with our other federal and non-federal partners, is that we can facilitate, and we do have some mechanisms that we can help in moving forward with that.

Art even mentioned several activities, including we do fund the GeneTools with a contract with the University of Washington and Dr. Wylie Burke. This does emphasize current clinical labacobility as one example. Also, the Genetics in Primary Care Program we have had for several years is the focus of that, is getting that into practice, and I can give you several other examples. But the issue is how do we disseminate that? How does that get brought forward across all the disciplines, whether you are an active clinician or an educator? I think that is a key issue that perhaps this committee can look at in the future as an issue of the next steps of where we go.

Also in stepping back, some of the activities and current mechanisms we have within HRSA are we have programs that can look at cultural competency, and we have funding streams and programs in the Office of Minority Health, and other parts of HRSA, that that may be a help in what you're talking about.

We also have responsibility in workforce diversity, and have programs

within our Bureau of Health Professions that again, can be tapped into perhaps, and informed to move in this direction. Also we have programs looking at the pipeline for the workforce, whether it is a genetic specialist, or the generalist.

So again, just as a reminder, we do have some structures within our agency in working with our federal and non-federal partners that perhaps we can address some of the issues that you brought up in your original report.

We just had a meeting that Dr. Hans was at on Thursday, looking across information system capacity in our health centers. As you may know, we served 12.5 million patients in 3,500 sites in our federally-supported health centers, although we're only on average 25 percent of the funding for the health centers. We can influence some of the direction, and one of the things we're working on within our health centers is the integration of the genomics into the latest science, which we do through our Health Disparity Collaboratives, which again, that has the patient registry with some of the prompts and activities that you have been talking about. So again, I think we can help facilitate in a variety of ways, some of the issues that you have brought forward with the review earlier.

DR. REEDE: Thank you very much.

Dr. Boughman?

DR. BOUGHMAN: I urge the committee in its deliberations to, and I'm quoting from a conference I was at recently, "don't let the perfect get in the way of good."

As far as Cynthia Berry's question on who is going to do this, and are people getting in each others' way, and are we stepping on toes. At this point in time, it seems to me that there is so much to be done, and there are in fact from the genetics point of view, so few of us to continue to do it, workforce issues being one of our issues, that from the educational point of view, that is not our primary issue.

It will be an issue if the question is, who is responsible for all this education without dedicated resources to get it accomplished? So as long as we can continue to generate possible resources from federal agencies, or from within institutions, then I think exactly who is going to do what is not our basic question. I think we have generated several models where we're working in concert with each other, and I hope that certainly continues.

The other point that I would make, and we have gotten some very sophisticated comments back about family history in the medical record, and specific prompts and all of those things, but I would simply remind folks that the approach that the Genetic Alliance, the National Society of Genetic Counselors, and the American Society of Human Genetics have taken in the development of a generic family tool as to remind people that it is the consumer's information, and it is the consumer's responsibility to gather that information to provide it to the provider, no matter who it is.

There is no blood test, there is no fingerprint, there is no way that any provider can get the information without engaging the consumer themselves. So in fact, the approach that we have kind of built our strategy on is let's get the consumer, let's get the general public excited about their own family history and information, some of which at least may be medically relevant. As they bring that to their provider, whatever the provider's credentials are, then in fact they will initiate and engage the provider.

If the provider, once again, has gone through their genetics readiness training, they will be much more able to accept and interpret that information correctly. But we have in fact not directly addressed this most perfect genetic or family history tool, and we are

working with other organizations to try and get down to the basics.

In many situations, we would take almost any information that we could get as a starting point, and I only need refer or remind people that many of the best genetic studies have gone back to family bibles, and the information that the family had to begin with to in fact garner our information, which would be considered the geneticist information.

DR. REEDE: Thank you very much.

Mr. McInerney, you had mentioned earlier wanting to make a comment in regards to who should be doing what. If you could speak to that, and also in some of your written comments with regards to the recommendations, you had mentioned promoting public education that provides knowledge and skills to consumers, with requirements to participate effectively with health professionals in decisions that informed genetic prospectus, which I think follows very nicely from the comments just made by Dr. Boughman.

If you could speak to both of those, please?

MR. McINERNEY: Yes, thank you. Well, Dr. Boughman really covered the first point that I was going to make, but I will tell you just a quick story. That is when we first published the core competencies in January of 2001, we heard a lot of complaints from people in the genetics community, that these competencies were trying to turn all health care professionals into geneticists.

I suspected that that was not likely to happen, to begin with. And in fact, it hasn't played out that way. I will say that the competencies are challenging, and we ask a lot of health professionals in those competencies. But what we've heard in the subsequent three and a half years roughly, is that health care professionals are quite sanguine about their own limitations, not only their own knowledge, but their own limitations in practice. They want to know how much they need to do to incorporate genetics effectively into their own practice, and when it is necessary, then hand that off to somebody else.

What we found is that people are using the competencies in that way. They are thinking carefully about how genetics manifests itself in their own practice, and then picking and choosing the competencies appropriately. So I think that issue has resolved itself, perhaps only for the moment, but I think it has resolved itself now. The issue of who pays for what, who delivers genetic services, that's an entirely different issue, and one that I'm not even remotely qualified to address. But from an educational standpoint, the other stories should serve, I think, as a helpful guide.

With respect to my proposed recommendation, or proposed addition to the recommendation you want to take to the Secretary, it just strikes me, as I said to the committee when I testified here at the last meeting, that we can't think about education of health professionals in the absence of education of the public, particularly if the assumption is that genetics ultimately is going to move health care more and more towards a prevention-based paradigm. That is something we have all been hoping for for a very long time, and for me, the notion of prevention always has implied a partnership between patient and provider.

If we are going to have an effective partnership, both of the partners have to be well educated. The education, I think, for the public, has to proceed from the same conceptual base, the same set of assumptions about genetics that we use for health care professionals. The details will differ, but I think the set of conceptual assumptions has to be congruent with that for health care professionals.

DR. REEDE: Thank you very much.

Dr. Dunston?

DR. DUNSTON: Yes. I wanted to underscore a point made earlier, too, about the importance of getting the consumer involved as part of this transition. But I wanted to preface my comments by saying that I thought it was very instructive that this committee is genetics, health, and society, and not genetics, disease, and medicine.

Instructive in the sense that I think that engagement of the consumer in why they need to be engaged in understanding the knowledge, participating in the research is a challenge that is part of what I like to call, the very positive potential of genomics. In that this seems to be a time where the genomic technologies offer an opportunity to understand disease at the biological level that requires the participation of the consumer, participation of the non-patient, and family histories must engage those who are not coming to the attention of the physician because of the disease.

The absolute necessity of public education, so that we can get participation in research to really utilize the power of genomics, and understand the biology of disease, is a challenge that I think is putting public health in the forefront. The necessity of the consumer, and the population, both those affected and non-affected with disease, are integral to understanding the biology of disease in ways that the challenge to me would be to take everything that we have learned on the negative side, if you will, from genetics with disease and medicine, and ensure that laws are put into place now that would at best, minimize the exploitative and manipulative, and the issues that would keep the public from participating in the research that is absolutely necessary to really use the power of genomics today to understand biology.

So on the one hand, we can take what we have learned during the era of genetics that may serve as the basis for the public being fearful, and look at the laws. I loved the beginning of this meeting with the emphasis on the Nondiscriminatory Act. We can ask, what are the things that were wrong with the way we used genetics before? And what do we need to put in place to minimize that occurring so that the science can move forward in a way that the public will want to participate?

I really underscore the importance of research that engages the public, engages the non-affected, as well as the affected as essential to getting the benefits of how we understand our genetics today.

I think, lastly, that for the public to understand that the power of genetics now, or genomics, large scale, high-throughput, information technology driven science, to tease out the underlying biology that is related to disease, requires public participation. So the question becomes what do we need to put in place with our laws, policies, and education, that will ensure that we do a PR job for what the potential of genomics is is one that we are not concerned about a whole battery of things to present discrimination. But what do we need to do to protect our privacy? To protect confidentiality?

Those are issues driven more by how do we get the public excited about the power of science today to help us understand disease in a way that gives us whole new approaches to prevention and promoting health.

DR. REEDE: Thank you.

Dr. McCabe?

DR. McCABE: I think that's a terribly important point, because if we are to really utilize the power of genetics, then we will need large population studies, and we'll be

talking about that more. But given that the unique genotypes, whole genome genotypes, that individual patients will have will be relatively rare, if we're really to develop the correlations, we will need those large population-based studies.

I think it is also important that you tie it to the Genetic Nondiscrimination Act that was discussed at the beginning of this by Dr. Boughman, because until our patients feel that they can be safe with genetic knowledge in their medical care, they're not going to feel safe in the research arena.

If they understand that the research is to improve their care, and not to be used in a discriminatory fashion, because that is outlawed, then I think that they will feel far more secure in gaining that knowledge about themselves. So I agree wholeheartedly with your points, Dr. Dunston.

DR. TUCKSON: This is Reed Tuckson. I'd like to get on the list also.

DR. REEDE: Reed, go ahead.

DR. TUCKSON: Thank you. Two things. I think it was very helpful to hear those last comments. One of the things I have been impressed by, particularly by certain people on our committee, and many of the people that have testified, is how specific they are of families who are going through the experience of genetic disorders and diseases, how quickly their learning curve reaches fairly impressive levels.

With that as a hopeful sign, I am also, though, faced every day with the reality of just how tough it is to get any level of understanding about health and disease in the general American population. If you even just take an example like obesity, it is a devil of a time trying to educate people in our country about the etiology and other therapeutic issues regarding something that is commonplace as that disease and disorder.

If you think even of the number of people who are currently appropriately getting diagnosed for hypertension, my point is that I think that while this is an important area that we have underscored, I think this committee is going to be challenged by trying to figure out what the priorities are in this area.

We obviously cannot reorder all science literacy in the country. In fact, if you look at the science literacy of the American population, it is staggeringly low. And so I wonder whether we might start to, as we continue to discuss, drill down to the key priority areas that we think we can meaningfully through the power of the Secretary, actually do something about.

Secondly and finally, I just wanted to briefly get at this idea of who does what, and what those competencies are that Joann talked about, and others. I hope that we can also get to this idea of defining what needs to be done, and then start to figure out the whole range of comprehensive tasks, and then start to determine what is the interrelationship between different parts of the health care system, and the various professional, and maybe even non-professional disciplines that have an opportunity then to play a role in that. I think if we talk about it, we can get more specific.

Thanks.

DR. REEDE: Thank you very much.

In the interest of time, if there are any comments or questions with regard to the recommendations for resolutions? Additions? Mr. Citrin?

MR. CITRIN: Well, the recommendations that I had as comments relate to a couple of my earlier comments, but just to be quick. One is that while public health is

mentioned in page two, there are references in page one to clinical medicine that could be broadened out to make clear that they relate to public health as well. There may be some more generic terminology. I still get the sense that there is more of a clinical/medical focus than there is a broader focus on all health professions and education, including public health.

Secondly, it seems to me that the ethical/legal/social implications, the ELSI, ought to be referred to specifically somewhere in the resolution. It is interesting that in the Institute of Medicine's report that I referred to on the teaching of public health, a good deal of the language in the recommendations on the genomic competencies that all public health professionals need, related to the ELSI dimension.

It isn't automatically evident in reading the resolution that this is a component of the education, that this committee particularly ought to see as essential since it is so much at the heart of what this committee is all about.

Then in terms of the preamble, and I guess this relates to the question that was put on the table earlier about what is broke, that it would be helpful, it seems to me, in gaining attention to the resolution if some content were put into the preamble to suggest some of the really compelling reasons why it is urgent that increased attention be given to the education of health professionals in genomics because of concerns of what will happen if the pace of development of technology goes far beyond the competence of professionals to make use of the technology, or to inform the public adequately about the implications of the technology.

Thank you.

DR. REEDE: Thank you.

Dr. Lewis?

DR. LEWIS: My only comment is on your points in terms of the recommendations on number five. I'm not sure who is going to decide what is adequate knowledge in human genetics and genomics, because that might depend on where you sit.

So to use something that is either to take out the adequate, or to determine how it is going to be measured would be helpful, because I think that having that be left to interpretation may be problematic.

DR. REEDE: Dr. McCabe?

DR. McCABE: I just wanted to follow up on Mr. Citrin's comments about the ELSI. Not only are they important, but also they are engaging to the students. Perhaps even more importantly, however, while the technologies will change, the ethical, legal, and social implications of those technologies will remain somewhat constant. So I think it is important to cast a lot of the teaching in those, because they are engaging, and they are important. But they also will be somewhat longer term issues than whatever is the technique de jure.

DR. LEONARD: This is a rather specific point, but in recommendation number five, "Encourage accrediting licensure and certification bodies to condition accreditation licensure and certification among demonstration of taking out adequate knowledge in human genetics and genomics." Can this be specified to refer to health care professionals?

Laboratories that do testing are also accredited and licensed, and I don't think you were referring to laboratory or testing accreditation and licensure. You are talking about accreditation and licensure of health care professionals. It raises a measure of concern

among those of us who do laboratory testing, because there aren't a lot of people out there who are accredited and licensed in genetics, such that if we are limited to hiring only people who are accredited and licensed, we won't have a workforce.

So while I agree that we should encourage accrediting licensure and certification of health professionals, it shouldn't include laboratories.

DR. REEDE: Any other comments? Dr. Felix-Aaron?

DR. FELIX-AARON: Yes, a couple of points that came up in the conversation today. I don't see them represented in the points in these documents. I want to build on Dr. Whitcomb's point, the question around building the evidence. So that not only relates to Dr. Whitcomb's point, but also to what Dr. Tuckson said about the what to do in terms of building the evidence base for genomics and its implication, not only for practitioners, but for medical education. So I don't see that reflected in the resolution.

The other point I would like to make is also related to this issue, but how to integrate content. I'm not sure what I'm about to say has implications. Well, it is something you would try to implement in this document, but I think it should be captured, and the committee may want to consider it in terms of how to integrate genomics into medical education.

I think we may want to study what HRSA has done with the disparities collaboratives, and how they have used a collaborative and improvement model to get changes in practices. So I offer this as a model that we could use, or at least study to see how you get change in medical curriculum. So it is an area which I think is right for a pilot.

You have best evidence, you have tools, you have people in small pockets doing best practices, and you could use that, and develop a model for getting that kind of curriculum changed into medical schools, schools of public health, and nursing schools. So I think it is something that I would like to put on the table that if the committee was interested in looking at.

It is something you could comment on sort of what the health centers have done, and how they have used a collaborative learning model, coupled with an improved model to change practice. I come from the Agency for Healthcare Research and Quality, and we think a lot about integration and trying to change practice.

We struggle with the issue of overburdening not only teachers and practitioners, but really trying to give people the tools, give them the tools, give them the setting they need to support and foster change in their practices.

DR. FEETHAM: I mainly would like to say I had written that down earlier that they, again, the collaborative or breakthrough series type of model may be something that, again, various disciplines could come together and make application to the Bureau of Health Professions to look at this as -- because system changes really are a major piece of what we're talking about. Not only within the academic institutions, but in the practice and providers.

So again, I would just encourage our community to be thinking of something innovative that you might look at that type of a model for coming in for funding from the various federal agencies to build off of that idea. Again, I can give people information about our website if you want to know more about the breakthrough series, and that type of learning model. But I do think we need to look at this within the context of system change.

DR. REEDE: Mr. Gray?

MR. GRAY: Yes, thank you. I'm sitting in for Commissioner Miller, who wanted me just to raise this one point, and I guess it relates to point number six. That is that we're talking about issues relating to diversity that we keep in mind diversity within the disability community. Cultural diversity should include that segment of the population as well, and we should just bear in mind that within the disability community, there is a wide range of interests, concerns, and differences among persons with disabilities. So that should be something that we should focus on as well.

DR. REEDE: Thank you.

Ms. Masny?

MS. MASNY: Yes, this is a comment, and then just a suggestion for one of the resolutions. In reviewing the papers from the responses from all the professional organizations, I do believe that almost every single one of the professional organizations did mention NCHPEG, and what the core competencies meant for them, and how that was one of the pieces that they were able to integrate into the work that they were doing to integrate genetics into practice, in genomics into practice.

One of the comments that we received from Dr. Jean Jenkins was to actually recognize NCHPEG and the work that they have done in part of the resolution. I think that it would fit very well into the second recommendation regarding the partnerships and the cataloging of information, and sharing information to actually mention NCHPEG there, both to hopefully get more visibility for NCHPEG, and maybe future members, as well as to see it as one of the models for partnership.

DR. REEDE: Thank you very much.

I want to thank all the members of the roundtable and the members of the task force for their work on this, and we'll use this in further deliberation. Thank you.

DR. McCABE: Thank you very much, Dr. Reede, and all of our invited guests for sharing with us the work of your organizations in advancing the education and training of health professionals in genetics and the insight on how to encourage more of these efforts.

Your responses, and all of the responses we've received to our information request and this roundtable discussion, are immensely helpful to our exploration of private sector efforts in genetics education and training, and certainly it will be very useful to us as we further develop our resolution to the Secretary on this important topic.

With that, we will be discussing the draft resolution during a working lunch, and so to all committee members and ex officio members, box lunches are available for you out in the hallway. Please take a few minutes -- literally a few minutes -- to gather your lunches.

For members of the public, lunch is available in the hotel restaurant, which is on the way to the lobby.

We will reconvene in about 10 minutes.

(Whereupon, at 12:15 p.m., the meeting was recessed for lunch, to reconvene at 12:25 p.m.)

AFTERNOON SESSION (12:36 p.m.)

DR. McCABE: We're going to consider the draft resolution on genetics education and training of health professionals. This can be found at Tab 4 of your briefing book, and staff has also put it up on the screen here. Joan, are you ready? I didn't check with you. I'm going to turn it over to you for this discussion of the education resolution, or we can do a tag team so we can both eat, if you'd like to do that.

DR. REEDE: Why don't we tag while I chew?

DR. McCABE: Okay. So I'll start off, and then Joan will take over. You have the draft resolution up before you. Somebody commented that it sounded a bit stilted to have all the whereas', but unfortunately, that is the nature of resolutions, to have all those whereas', so I think we're stuck with those.

So, "Whereas the Secretary's Advisory Committee on Genetics, Health, and Society was established to advise the Secretary of Health and Human Services on the range of complex and sensitive medical, ethical, legal, and social issues raised by new technological developments in human genetics."

I know it has been brought up about family history being the first genetic test. I think that a couple of the ex officios were looking at where we might fit family history into this. Any comments? Alan, were you one of the ones working on that?

DR. GUTTMACHER: I wasn't working specifically on a place to put it, but I think there were a number of us that thought it should be put somewhere.

DR. McCABE: Okay.

DR. GUTTMACHER: But I will pay attention now while we work over lunch as to where it should be put, unless somebody else has a good idea.

DR. McCABE: We could certainly work on including it in the recommendations, if we didn't want to insert it in the whereas.

DR. GUTTMACHER: And I would think it would make more sense as a recommendation, rather than a whereas.

DR. McCABE: Robinsue?

DR. FROHBOESE: I do have a specific recommendation. I think in keeping with the very good recommendation that Reed first came up with, and then others supported in linking with the Secretary's efforts on health information technology, and the fact that that could be one place where we can include family information, I think in recommendation Number 1 which already addresses departmental policies and programs about genetic information, that there we specifically include family history, and a recommendation that the CAHIT effort in the
Department on health information technology include looking at genetic information and family history as part of its overall recommendations in formulating an action plan.

DR. McCABE: Okay. Joan, are you ready to take over?

DR. REEDE: Sure. Moving forward, any other comments on incorporation of something related to family history and looking at inclusion of information technology, looking at genetics and family history within recommendation one?

DR. KHOURY: Maybe this was said earlier. But I guess anytime we use the word "genetic information," we can put slash, family history to it, because family history is much more than genetics. It represents shared environment, shared cultures, and shared behavior.

So the philosophy that genetic information and family history should not be exceptional and so on and so forth, you know, search for the word genetic information in family history.

DR. REEDE: Martin?

MR. DANNENFELSER: I guess I just wondered, in the context of the Genetic Information Nondiscrimination Act, is this covered information within that, or not? Obviously if you have information in a medical record relating to preexisting conditions, that can be problematic, I imagine, to a person in terms of health insurance coverage, or potentially employment. Does putting family history in there have any impact on whether or not that is protected information within the context of the Genetic Information Nondiscrimination Act?

DR. REEDE: Any comments?

MR. GRAY: Well, the bill, as passed by the Senate, defines genetic information to include family medical history. It is specifically included in that, and I think it is important that we recognize family medical history is a part of genetic information. I would certainly make the point.

DR. REEDE: Any other comments with regard to this family history and information technology in recommendation one?

DR. FROHBOESE: I was just going to add that by including it in that first recommendation, and in the context of the Secretary's Health Information Technology effort, a critical part of what that group is doing is looking at privacy, confidentiality, and nondiscrimination. The Office for Civil Rights is an active participant in that, and is ensuring that the privacy rule under HIPAA and nondiscrimination considerations are part of the formulation.

So I think by putting it in that first recommendation, we'll have some assurances that those considerations will be part of formulating any recommendations.

MR. GRAY: Can I just make one suggestion? The way it is written now, it says, "genetic information, including family medical history information." I would just change that to say, "which includes family history information."

The reason for that is that I want to make sure it is clear that family medical history is a part of genetic information, that is not something that is separate.

DR. REEDE: Moving on, the next whereas is, "Advances in genomics have the potential to greatly improve health status and outcomes," and "Appropriate and adequate training and education in genomics is crucial for all health professionals to assure the successful integration of genomic concepts and genetic technologies, and services throughout the entire health care system."

DR. LEONARD: Maybe we should add to that, "Whereas health professionals and the public." Because we have been bringing up over and over again that the public is an active part of this process, and so their education is just as important as the health professionals, in a different way.

DR. REEDE: Any other comments?

MS. HARRISON: Joan? I would advocate that we make that a separate whereas, because I think the adequate training and education in genomics, obviously we're meaning a different type of training and education. So I very much advocate for the public education piece to be in the whereas, but I think it should be separate.

DR. REEDE: Any other specific comments with regard to the public

education piece? I think there was actually a very rich discussion at the end about the need for the public education. It seems clear that it should be incorporated here somewhere in terms of a separate whereas, or an integration into these current ones. Any thoughts?

DR. WINN-DEEN: I think it is important to include sort of the public education and capture this concept as an informed consumer, that we need them to be educated so that they can be informed consumers, and active participants in their health care.

DR. FEETHAM: For the previous one, to be consistent with the language from Muin's comments, and Toby Citrin's comments of saying health care and public health professionals in your prior whereas, and look at that throughout the document.

DR. REEDE: So in this area, and any other area?

DR. FEETHAM: Pardon?

DR. REEDE: So in this section, but in any other areas, making it more broad?

DR. FEETHAM: Right. Saying health care and public health professionals, I think you'll cover a broader scope.

DR. REEDE: The recommendation here was to, "Engage other health professionals, the private sector and colleagues at the federal and state levels to facilitate the cataloging and dissemination of genomics applications to clinical medicine and public health, and models based upon these applications to ensure genomics has impact now, as opposed to far-off in the future." Emily?

DR. WINN-DEEN: I just want to say I think before we move down to number two, that we should probably split one into two different things. One is sort of including genetic stuff and Department policies, and then the second part of that is talking about education. Then I think two becomes three then, if we do that. It just seems like one now has sort of two different thoughts in it.

DR. REEDE: So one relates to departmental policy, and the other one relates to overall education and training. Okay.

MS. CARR: And where would you see the point about family history? Would that be part of the second one, then? Or a separate one altogether?

DR. WINN-DEEN: I think the family history, it needs to be integrated as part of the training and education of health professionals to take family history, and to make it part of medical records. I think it is appropriate, sort of the way you have split it there.

MS. CARR: Okay.

DR. REEDE: Look at recommendation Number 2.

DR. LEONARD: And so two should start with "Integrate genomics into the education." So it is like the other ones.

DR. TUCKSON: (Inaudible.)

DR. McCABE: Reed, we're not hearing you. You'll need to speak up.

DR. TUCKSON: The third whereas. Can you hear me now?

DR. REEDE: Yes.

DR. McCABE: Reed? It sounds like you are a little bit close to the mike.

DR. TUCKSON: How about now?

DR. McCABE: That seems a little bit better.

DR. TUCKSON: (Inaudible.)

DR. McCABE: You're breaking up.

DR. TUCKSON: I'm breaking up now?
DR. McCABE: And we aren't trying to ignore you by saying that.
DR. TUCKSON: All right. I'll try to call on a different line.
DR. McCABE: No, that's better.
DR. TUCKSON: Okay. The third whereas --
DR. REEDE: I beg your pardon?
DR. TUCKSON: I heard you a moment ago about the third whereas (inaudible.)
DR. REEDE: Right.
DR. TUCKSON: Have you completed that discussion?
DR. REEDE: No, I don't believe we have. We talked about the addition of the public, and putting it within this whereas, or creating a separate whereas that refers to the need for public education and training.
DR. TUCKSON: I'd like to urge a little bit of (inaudible) integration, add a little bit of that to the discussion.
DR. REEDE: No, it hasn't. So you're speaking specifically to the word "successful" and another word that might be more suitable?
DR. TUCKSON: Yes, and I'm not sure what successful means. And so I'm thinking of appropriate, effective, and efficient integration.
DR. REEDE: Appropriate is what we have up there now.
DR. TUCKSON: Appropriate, effective, and efficient.
DR. REEDE: Is there any further discussion around incorporation of the public within this, or setting up a separate whereas for public education?
DR. FELIX-AARON: I think as a standalone whereas, I think it is very effective. The only other thing you may want to consider is to add a phrase at the end. It could say, "Whereas appropriate education and genomics is crucial for the general public to reap the benefits of, or to take advantage of the power of this." So that it clearly specifies that not only the public needs to be engaged to be better consumers, but also for them to get the benefits of this vast technology.
DR. REEDE: Okay. The next whereas was "Whereas such integration is a necessary component of access."
MS. CARR: Dr. Reede, can I just go back to the one prior to the general public? The last part of that read, "the entire health care system," and we had a suggestion that it just read "the entire health system," that that might broaden it a bit. Is that okay?
DR. LEONARD: In the second whereas, I guess it is, the third whereas, we were going to add public health and health professionals, or we were going to broaden that to be public health and health professionals.
DR. FEETHAM: The language that we used in a lot of our documents through HRSA and NIH is health care and public health professionals.
DR. REEDE: Brad?
MR. MARGUS: I have another whereas to talk about. Are we ready for another whereas, or do we have to finish that whereas? Going to my question about what was broken, and Mr. Citrin's response to it, I was thinking that this might sound a little negative, but it would still underscore maybe an urgency to do something.
Something along the lines of, "Whereas insufficient training and education

can lead to excessive costs incurred on inappropriate tests," since he brought that up, "inaccurate, or at least illusive disease diagnoses," which is what we just talked about, "and in many cases, misguided disease management and family planning." Those are all negative things that can come about if the Secretary doesn't do anything about this.

DR. REEDE: Sort of in this area, the broken things, the issues that created urgency. Are there others that you would want to add to that list?

DR. WINN-DEEN: Yes. When Sarah gets done typing this list, and I'm just going to comment that I don't think the excessive costs should be the first thing. I think the first thing should be the patient management issue. Obviously costs are important, but I think the key thing is that patients are not being managed as they should be for their medical condition.

DR. FEETHAM: Another concept of that lack of education is access, in addition to the patient management.

MS. BERRY: We'll probably have to reorder them, won't we? Because when it says, "such integration," it refers to the two other whereas' above.

DR. REEDE: What you said is education is a necessary component of access. I was wondering if you wanted to say something more specific about genetics.

MS. ZELLMER: On the whereas as far as where it says, "Such integration is a necessary component of access," which should refer to the third one, I think actually access is affected by education of not only the health care professional, but the public. So maybe we could just say, where education is a necessary component of access, rather than integration.

DR. REEDE: Or possibly mention both the public and the health professionals, sort of public health mentions specifically the education of this body as an integral component.

MS. CARR: Tell me what to do. I was so engrossed in that.

MS. ZELLMER: Under the next whereas, instead of "such integration," say "education of health care and public health professionals, and of the public is a necessary component of access." Does that sound right?

DR. REEDE: I'm sorry. Either one.

DR. FELIX-AARON: The fourth whereas, or Brad's whereas. I think one of the points that Mr. Citrin made that wasn't reflected in this whereas is you name cost, sort of the media distortion, and the role of expanding technology that can either reduce disparities, or exacerbate disparities. I'm not sure that whereas, that third point, was captured in that whereas.

DR. REEDE: You want to offer some wording?

DR. FELIX-AARON: Exacerbate disparities, health disparities.

MS. CARR: This is a --

DR. FELIX-AARON: I'm not asking for there to be a separate whereas.

DR. REEDE: It is just another item to be mentioned.

DR. FELIX-AARON: Sort of the urgent need for this.

MR. MARGUS: It sort of belongs maybe in the next whereas, related to access to.

DR. FELIX-AARON: Where is education?

MR. MARGUS: Isn't there a place where we talk about new technologies?

DR. REEDE: No, actually, I think the issue around exacerbation of health disparities belongs up with the other urgent areas.

DR. FELIX-AARON: Yes. Sort of where we talk about the impending crashes.
DR. REEDE: Down? Where insufficient and training can lead to?
DR. FELIX-AARON: Right. So whereas insufficient education and training can lead to inaccurate disease diagnosis, and misguided management.
MS. CARR: Do you want it before costs, or after?
DR. REEDE: Before cost. Last, okay.
Muin?
DR. KHOURY: The idea captured here is evidence-based health care prevention. When we talk about access and education of public health professionals (inaudible), we're saying education is very important for the evidence-based practice of medicine. So maybe we can repackage that paragraph in a positive light or a negative light, because we're talking about sufficient education can lead to that stuff. (Inaudible.)
DR. REEDE: That was going to be my question. How would you massage this?
DR. KHOURY: I mean, just the same thing, a necessary component of access, a necessary component of evidence-based practice of medicine and health care, whatever you want to say. Really, education is a component of for various reasons, not only access.
MS. CARR: Muin, are you saying that should replace the "Whereas, insufficient education"?
DR. KHOURY: Yes, or put it somewhere in there.
DR. WINN-DEEN: Sarah, why don't you just put education and training prevent inaccurate or (inaudible) disease, so it is not a negative.
DR. KHOURY: If I could use the word "evidence-based," that's all. If we can find a place for it, that would be nice.
MS. CARR: Well, and I guess positives are always better than negatives, but I think Brad was sort of -- well, it is in response to your own concern that we need to call attention to a problem, I guess.
DR. WINN-DEEN: Well, what about if we just add the evidence-based medicine thing in the one that you were working on that ends in "of access," evidence-based medicine improved prevention and public health, and whatever buzz words we want to put in there.
MS. MASNY: Or we could put it in the higher up with the, "Whereas advances in genomics have the potential to move us to a preventive model of health and evidence-based practice to greatly improve the health status." Because that is going to be the mechanism by which we're going to improve the health status and outcomes.
DR. HACKETT: If you're talking about evidence-based medicine, isn't that entirely a third new topic that might confuse things? We're talking about genomics, genetics, and then evidence-based medicine.
MS. MASNY: I would think that the implication there would be that now that we understand the biology of the disease, or conditions, that that is going to be the evidence base. So I think it goes hand in hand with the genetics and genomics.
DR. HACKETT: But that is such a big topic by itself, is the only concern.
DR. REEDE: I think it is written where it is the application of evidence-

based medicine. I have no problem with it being printed that way. Do others want to weigh in?
MS. CARR: What about what Agnes was suggesting? Were you saying that we should modify the second whereas in some way? Or does this take care of it?
MS. MASNY: That is what I was suggesting for the second one, because I think that the issue of the preventive model that was suggested, as well as evidence-based is the actual cause, or the thing that precedes the health status and outcomes, much more so than related to access.
MS. CARR: What would you suggest, then?
MS. MASNY: Just that whereas advances in genomics have the potential to provide evidence-based practice, and greatly improve health status and outcomes.
DR. REEDE: Any other comments on the whereas' so far?
DR. WINN-DEEN: Do we want to say evidence-based practice in medicine, or something, instead of just evidence-based practice? Practice of what?
DR. TUCKSON: (Inaudible.)
DR. REEDE: Reed, we can't hear you.
DR. TUCKSON: Can you hear me?
DR. REEDE: Yes.
DR. TUCKSON: (Inaudible.) How does that read now?
DR. REEDE: How does it read now? I'll read it for you. "Whereas advances in genomics have the potential to facilitate evidence-based practice of medicine and greatly improve health status and outcomes."
DR. TUCKSON: The difficulty here is needing the word evidence-based practice. The genetics doesn't facilitate evidence-based practice. What is the evidence for the (inaudible) knowledge of whatever the discipline is. So advanced genetics (inaudible) into the guidelines and the actual practice of (inaudible).
DR. KHOURY: I agree with Reed. I agree with what Reed Tuckson just said.
DR. REEDE: Reed, would you like to make a suggestion for how we might rephrase this?
DR. TUCKSON: I think if I understand (inaudible) advances in genetics will lead to more (inaudible) understanding of disease process issues, and will (inaudible) better guidelines around what interventions genetic (inaudible) effective, and appropriate.
MS. CARR: Reed, we have modified it along the lines you suggested. So now it reads, and just make sure it is what you suggested. "Whereas advances in genomics will lead to more precise understanding of disease processes, and will provide better guidance on the application of therapeutic and preventive strategies," and I guess I lost you after that.
DR. TUCKSON: No, that's much better.
MS. CARR: Okay.
DR. LEONARD: But then can it just read, "to greatly improve health status and outcomes?" So take out, "have the potential to facilitate," right?
DR. REEDE: We have incorporated evidence-based practice in another section. Do you want to read that one again, also?
MS. CARR: Okay.
DR. McCABE: Before we leave that one, the editor in me sees a split

infinitive. Can we take the "greatly" out, please?

MS. CARR: Okay. The second reference to evidence-based medicine reads, "Whereas education of health care and public health professionals and the public is a necessary component of access and the application of evidence-based medicine."

DR. McCABE: Reed, could you mute the phone in between, please? We're getting some feedback and echo.

DR. LEONARD: That's like two concepts in one whereas. I don't see what access has to do with the application of evidence-based medicine. Those are two different concepts, and they need to either be split or --

DR. REEDE: Dr. Khoury?

DR. KHOURY: Why don't you split them? Have two whereas'.

DR. REEDE: Is there agreement on this? Okay. The next whereas reads as, "Whereas, through a survey of Federal agencies on their role and activities in genetics education, training, and health workforce analysis, it was found that Federal efforts are focused on enhancing access through facilitating the translation and appropriate integration of new genetic technologies into health care and public health."

DR. KHOURY: Is that all the federal survey found was related only to enhancing access? Or everything else? It seems to me that there was more than that, and maybe I missed that in the last meeting when you guys discussed this. But it seems like the focus of the federal efforts is not only on enhancing access, but a whole bunch of other things as well.

MS. CARR: Well, I think this is probably a distillation and sort of boiling it all down. But if in doing this, we have neglected to highlight something important, we can just add it here, I think, if the committee agrees.

DR. KHOURY: Well, the whole concept of translation and appropriate integration includes access. So if you just say that the federal efforts are focused on translation and appropriate integration, because without just highlighting enhancing access, it seems to me that it is too focused. Just take away "enhanced access." That's it.

DR. McCABE: Do the other federal agencies agree with that? Anyone who disagrees?

MS. CARR: Well, the access agency isn't in the room at the moment, so we might want to come back to that.

DR. HACKETT: A quick question. Would you want to change that to genomics, or just leave it as genetics? Both the first and last sentences.

DR. REEDE: Genetics and genomics?

DR. McCABE: I would make it genetics and genomics, I think.

DR. REEDE: I would suggest that when HRSA returns, we revisit this. The next whereas is, "A solicitation of information from educational and professional organizations, identified the following needs in genetics/genomics education and training." Those needs were identified as, "Inventoried, widely relevant clinical applications stemming from advances in genomics; educational models that use such applications to clarify how genetics/genomics should be integrated into practice; a broadening of the focus of education and training from genetics to genomics; appropriately trained faculty; and training programs that address the interface of, and interaction between, genomics and public policy." Khoury?

DR. KHOURY: I would suggest to revisit family history here as under its

own bullet, or one of these bullets. But it could be its own bullet.

MS. CARR: Well, I think we'd have to make sure that the information that came back from these organizations, that is what this bullet is about. Amanda or Joan, was the family history highlighted in the survey response?

DR. REEDE: No, it wasn't mentioned there. But one of my questions would be, this is a solicitation of information. If you consider it a solicitation that included the survey and the roundtable, in that case, it could be included.

MS. CARR: And the two changes that are here are reflective of comments made by the roundtable, too. So how would you like to add it, and where?

DR. KHOURY: It could be under the third bullet, "A broadening of the focus of education and training from genetics to genomics, including."

MS. CARR: Which includes the point Peter --

DR. KHOURY: Which includes family history tools.

DR. REEDE: One of the other areas that was mentioned, I think, by Dr. Whitcomb, in addition to the clinical applications, he talked about the need to access patients, and this isn't reflected here. Do we want it to reflect that?

MS. BERRY: I don't know if it's accurate to say, but this gets to the point about how significant is the problem. Is it accurate to put in the beginning part before the bullets that these are urgent needs? Use that modifier? I don't know if everyone would agree that every one of those is urgent, but that would sort of add some oomph to the resolution, if you think that is accurate.

DR. LEONARD: Can I make a suggestion in the bullets? The family history tool is really more a part of the second bullet, which is integrating genetics and genomics into practice. The way that you're going to do that in general practice is by use of family history tools, more than a broadening of the focus of education and training from genetics to genomics.

MS. CARR: And maybe in this case, it could read "including," rather than "which includes."

DR. LEONARD: Or "through the use of." Well, yeah, you could just say "including," or "through the use of." Whatever.

DR. REEDE: One of the areas that was mentioned in the comments related to the use of the Internet technology and those types of things, and I'm wondering if there is not a way to integrate some of that, along with the information technology that we're talking about here. The importance of this is sort of the future area.

DR. LEONARD: Well, it could be through the use of family history tools, information technologies, and
web-based practice tools, or things like that. That is basically what we were talking about, as a way to get it into the hands of practicing physicians, which I think is what this second bullet is really getting at.

DR. REEDE: I think it's fine. I think the more we can link the concepts across, and since information technology came out so much today, and it was mentioned in another way, but it was mentioned in the responses, to have it incorporated here would be useful.

Agnes?

MS. MASNY: Under these bulleted items, during the roundtables, it was

mentioned several times about the importance of the certification, licensure requirements, and things like that. Someone had mentioned that it should be included in the whereas as to why education should be mandated.

So since the certification question was one that we had with all the survey participants, as well as in the roundtable, if the group thinks that a separate bullet item about the importance of mandating --

DR. LEONARD: Well, I think the point was made that unless you mandate it in the certification or licensure, whatever, by those bodies, it is not going to change in the curriculum.

DR. REEDE: I'm wondering if a way, since so much of that is occurring at the state level, or through private organizations and not really the government level, for part of the certification licensing, so the licensing is occurring at the state level, I'm wondering if there --

DR. LEONARD: But accreditation is more at a national level, so there are accreditation bodies. The licensing is more state by state.

DR. REEDE: Right. So I'm wondering, just being very specific, and not sort of putting the licensing, credentialing, and accreditation all together, because it is difficult to mandate that from a level that does not really control that area. So are you speaking specifically to the accreditation here? A bullet specific to accreditation?

DR. LEONARD: Well, I actually found it very frustrating to know that for physicians, that there is nothing that can be done nationally. That is pretty pathetic, frankly.

DR. McCABE: Well, I think what can be done nationally is to work with our boards, our professional boards, and those are the organizations that set standards nationally.

DR. LEONARD: And that's more accreditation, rather than licensure. So maybe if we want to put something in about national accreditation standards that, or the need for national accreditation standards to drive the incorporation of genetics into the curricula of educational bodies, or whatever. I don't know. I can't say that again, though.

DR. McCABE: Then make sure we put accrediting and reaccrediting, because in fact it is the reaccrediting that is required these days that is even more powerful in keeping the standards up.

DR. REEDE: The reaccrediting also addresses parts of the issues that you raised about what do you do about physicians and health providers that are already in practice.

MS. CARR: I'm sorry, but I didn't get that.

DR. LEONARD: It would be need for --

MS. CARR: I'll just say "need" at the end, but we have national -- remember, this is the urgent needs of the --

DR. LEONARD: Okay. National accreditation standards. Accreditation and reaccreditation standards, sorry.

PARTICIPANT: "Accreditation" is a facility term. I think you're meaning "certification."

DR. McCABE: That's right. It's certification, and then recertification.

MS. CARR: So are we changing this?

DR. LEONARD: Yes. "Certification and recertification." "Standards for genetic education, or genetic and genomic education to drive" -- or is it education or

proficiencies? Or competencies? "Genetic and genomic competencies to drive the" -- I don't know what you want to call drive.

MS. CARR: Influence?

DR. LEONARD: "Influence the incorporation of genetics and genomics in educational curricula."

DR. REEDE: I have a question. Is this better under a whereas, or should this be one of the recommendations that should be put forth?

DR. LEONARD: Well, this is something that they identified, at least in the discussion here, at least one person said that when there are mandates to provide certain types of education, that is what drives the curriculum changes.

So we may want to sort of take that as something that we could resolve to do also, but that was something that they were saying is a need. If you're going to change the curricula for many health professionals, then it helps to have accreditation standards that include genetics.

DR. WINN-DEEN: I think the other point was that we're not looking for people to become certified in genetics. We are looking for them to have standards for genetic and genomic competencies in order to influence the incorporation of genetics into genomics in educational criteria across disease areas, or across practice areas, sort of that concept that we're not just looking for some specialty in genetics, but that this crosses all medical disciplines.

DR. REEDE: So across disciplines?

DR. WINN-DEEN: Yes.

DR. LEONARD: Or could you say genetics and genomics in general, health care education curricula, or something?

DR. WINN-DEEN: Yes, just so that we're clear that --

DR. LEONARD: You're not creating specialists. We're creating competent generalists.

DR. WINN-DEEN: Right, or whatever, competent subspecialists.

DR. REEDE: Any other whereas? Robinsue?

DR. FROHBOESE: Yes, I have another suggestion for an addition. I think another central theme that came out of the survey of organizations, and that we've heard this morning which is also reflected in the recommendations, is the cultural diversity within, and the cultural competency of health professionals in genetics or genomics. So I would suggest adding that as a bullet.

DR. LEONARD: But that is something that genetics health professionals do. Do you hear that being stated as something that needed to be done in general for all health care providers?

DR. REEDE: I think it was felt by all. It wasn't just for the specifics of genetics, it was across the board.

MS. CARR: Would that be incorporated into the appropriately trained faculty? Or that would apply to all practitioners as well?

DR. REEDE: All practitioners.

DR. McCABE: While Sarah is working on that, let me just do a little bit of housekeeping. That is that we're running over the allotted time for this, but I think that it is quite appropriate that we do that. The coverage and reimbursement, we probably are not going to complete discussion of this afternoon, and that will continue, I'm sure, into the next meeting.

So I'm going to take the Chair's prerogative to extend this session. I just want to check, Reed, are you able to be with us for awhile?

DR. TUCKSON: (Inaudible.)

DR. McCABE: It sounds like you are commenting from outer space somewhere, Reed.

DR. TUCKSON: (Inaudible.)

DR. McCABE: Sorry. We really can't hear you.

DR. TUCKSON: Can you hear me now?

DR. McCABE: Not really. Is there any chance you can get to a land line, Reed?

DR. TUCKSON: (Inaudible.)

DR. McCABE: You sound really cool, but we can't understand a word of it. (Laughter.)

DR. TUCKSON: I'll call back on a land line.

DR. McCABE: Thank you.

DR. REEDE: Since HRSA is back in the room, there was one of the whereas' where we changed the wording and removed "enhancing access," and wanted you to weigh in on that, to read it, and give any comments.

DR. FEETHAM: I was here for that part of the discussion, and I think that clarifies and strengthens it.

DR. REEDE: Okay. Is that it for the whereas'? Can we move to the --

MS. HARRISON: I have one comment. If we can just go back to Brad's whereas. I wanted to try to make it even a little bit stronger. We had put "training can lead to," and I was wondering if you could change it to "is leading," because we know that it is happening now from the two stories that people have said.

MR. MARGUS: It could be "has led."

MS. HARRISON: "Has led," or something like that. You know it is happening.

DR. REEDE: If we're in agreement, do you want to go through these again just briefly?

MS. HARRISON: I see you're scanning through the rest of them. I don't know if we can change other verbs in there.

DR. McCABE: We're probably going to have to have this edited. What we have done typically in the past, because it is hard to get all the grammar right while we're doing this. So Sarah, can I volunteer you and your staff to work on this this evening?

MS. CARR: Yes.

DR. McCABE: Okay. And then bring it back tomorrow. I think that will be a more efficient way to go to get as detailed as we can, but not to worry too much about the grammar, having tried to unsplit an infinitive myself.

DR. REEDE: It's more are there any other concepts that we should include within this section?

DR. McCABE: What I would suggest is that we go to the resolutions per se, and then do a quick readover at the end to see if there is anything we pick up, because the two may inform each other.

DR. REEDE: Resolutions. "As such, and in light of the importance of

ensuring that the benefits of the genetics/genomics revolution are accessible to all Americans, the committee urges the Secretary to take the following steps to ensure that genetics education and training of all health professionals is adequate." I think it would be, "All health and public health professionals," that would be changed.

The first as currently written. Let me turn, because these have been changing. Actually, Sarah, it is easier for you to read than for me to try to do this.

MS. CARR: Okay. Let me read the first one. "Promote and actively incorporate into departmental policies and programs the philosophy that genetic information, which includes family history information, should not be treated as exceptional, but rather as part of the spectrum of all health information."

DR. REEDE: One of the comments that was returned with regard to this is the question of talking about exceptionalism, and this language, to the extent to which it is compatible with the language that we're using when we talked about direct-to-consumer marketing and wanted to know if there was any comments.

MS. CARR: I think what you are saying is someone pointed out the fact that what we are suggesting in the direct-to-consumer marketing resolution is not compatible with this recommendation. I think in Reed Tuckson's comments on the coverage and reimbursement report, he also took note of a couple of places where it appears to recommend a more exceptionalist approach.

I think the committee in March, when addressing the issue of genetic exceptionalism, concluded that it might be necessary to assess the need for exceptionalism, or the problems associated with it on a case by case basis. So I think you have left yourselves room to be inconsistent, I guess.

DR. WINN-DEEN: I think in this case, what we're talking about is really sort of not so much that genetics is exceptional but that it happens to be the new thing that we have to integrate into medical practice today, and so it is just the latest thing which we need to deal with.

DR. LEONARD: I think also you're talking about genetics as a subspecialty, and moving it from subspecialty practice to influencing every health care and public health professional. So I think it is more that it is not a subspecialty, so we may not even want to use the word "exceptionalism," and talk about it as specialized as opposed to general practice. Genetics is going to influence all of medical practice, so it has got to be part of every health care professional's training.

DR. WINN-DEEN: So this should not be treated as exceptional, it should not be treated as a subspecialty, or as a specialty.

MS. CARR: Or what if you just said, "should be treated as part of the spectrum of all health information?"

DR. WINN-DEEN: Yes, that's fine. Just take the "not" and the "exceptional" out.

DR. LEONARD: Sarah, in the preamble paragraph, in the last sentence, can you change "all health professionals," to "all health care and public health professionals?"

DR. REEDE: Reed, are you back again?

(No response.)

MS. CARR: And the second one isn't really integrated yet completely, but this part was part of the prior one. "Promote the integration of genomics into the education and

training of all health professionals," and this is a fragment, I think, from the first one, "so it should be treated and viewed." Actually, I think it belongs up there.

So the second one is, "promote the integration of genomics into the education and training of all health professionals." Here is where we wanted to build in the suggestion that the Secretary incorporate in the health information technology initiative, some reference to family history, or incorporate some electronic family history tools into that initiative.

DR. McCABE: Sarah, I would suggest that we separate out "add" into another resolution. It seems like we're mixing education and the Secretary's initiative, plus it will make it more obvious to the Secretary if it is not buried as a second part of the second resolution.

DR. REEDE: Wherever we have "health professionals," if we could sort of in the wordsmithing later, change it to "health care" and "public health professionals."

DR. McCABE: Reed, are you back with us yet?

(No response.)

DR. HANS: Just on this one, I guess I have some concerns about, because the discussion about CAHIT was broader than what actually the topic of this resolution was of education and training for health professionals. It is a much broader topic than just that.

So I would suggest one of two things. One, to narrow this down to just talk about the educational components to be considered in CAHIT for genetics information. And then put the question to the committee, if you want to take up the broader issues of CAHIT in the interface of this committee, sort of as a separate something, whether it is a letter, or some other separate topic.

I'd just be concerned if you'd try and capture everything in this resolution, because it is a more narrow topic than this opportunity offers. So this, if you want to narrow it down, could say something, "incorporate family history, tools, and practitioner educational tools, support." Instead of "tools," use "support," Sarah. And then I'm not quite sure where we're going with privacy and confidentiality, but there are a whole bunch of issues that may not fit with education and training.

DR. LEONARD: Can I suggest that practitioner educational support, really what you're looking at is point of care educational support? Because practitioner may be misconstrued as CME, or continuing education, which you really want it at the point at which you're seeing the patient.

DR. REEDE: I think I would also change the order. So I'd have as what you have as Number 2, I'd move down.

MS. CARR: So it would be Number 4?

DR. REEDE: Well, it would become 3, and 3 would become 2. While you're doing that, I'm going to read the next recommendation. "Engage other health professionals, private sector, colleagues at the Federal and State level to facilitate the cataloging and dissemination of genomics applications to clinical medicine and public health, and models based upon these applications to ensure genomics has impact now, as opposed to far off in the future." Any comments on this?

DR. LEONARD: There was a suggestion during this discussion to recognize the efforts of NCHPEG in this point, and I don't know how we do that.

DR. WINN-DEEN: Well, we could just say engage health professionals,

such as NCHPEG. Use it as an example.

MS. MASNY: But I think here, it is even stronger than just to engage them. I think some of the resources we heard about, both through HRSA and through NCHPEG, that what we want to make sure is that these resources are utilized, and that the public, and even the professional organizations and academic institutions know about them, so they don't have to reinvent the wheel. So somehow if we could make it stronger in terms of utilizing existing resources that have been developed by state and federal organizations.

DR. REEDE: Giving NCHPEG as an example?

MS. MASNY: Yes. We could give NCHPEG and HRSA as an example.

DR. McCABE: I would just argue private, state, and federal organizations.

DR. REEDE: And not use an example?

DR. McCABE: It's fine to use an example, I just think we need to recognize the private sector.

DR. REEDE: Kay?

DR. FELIX-AARON: In reading this point, I agree with it in principle and what it says, but I was just struggling with sort of what the federal rule is here in terms of if we have an audience, and I imagine this is directed to the Secretary, how do we want to use that space that we have with the Secretary?

So I would think like for matters of just being effective as a document, as well as being efficient, I think this needs to be done, but sort of what the Secretary's role here is, isn't readily apparent to me. So I'd just like somebody around the committee to just clarify for me what the Secretary's role is here.

DR. WINN-DEEN: Well, it seems like it is sort of covered in the next point after this one, where we talk about specifically federal.

DR. FELIX-AARON: Right. I mean, that is one of the rules that the federal government has provided technical assistance, and I think the federal role is to do things that cannot be ordinarily done in the private sector because there is not the will, or there is not adequate incentive to do it. It is not clear to me that that recommendation captures the unique federal role.

DR. WINN-DEEN: So maybe what we want to say is partner with the private sector and state organizations, and encourage --

DR. FELIX-AARON: I think the one below says to support efforts. The point below, provide adequate program and technical support, but it is not clear to me why the federal government should be engaged in inventory and the cataloging process.

DR. LEONARD: Well, actually, the CDC is doing that, aren't you, Muin? You're cataloging the most common genes, and the most common diseases, and so in a sense, you are providing some of that information that would take genetics and genomics to the family practitioner.

DR. KHOURY: I think what I'm hearing Kay say here is that you need to give the Secretary sort of the best possible chance for integrating genomics, genetics, and family history into whatever Department-wide initiatives are going on. I think under Number 2, the Secretary's Health Information Technology Initiative is something that he can take and say okay, I'm going to integrate family history tools into that. But there are a number of other initiatives that the Department has. For example, the STEPS initiative, which is a big initiative for the prevention of common chronic diseases, and health promotion. That is really prevention

oriented.

So by encouraging the Secretary to look across the board and say okay, these are the initiatives that I own, that I've started, but so far I haven't seen the role of genetics and family history in it. But now, there are more legs I could stand on and use these as forums to empower big initiatives that use and integrate genetics, rather than be a standalone activity for the Department.

And so I think singling out the Health Information Technology Initiative is one thing, but I don't think we should focus on just one activity from coming out from HHS, we should give them a broad range, a number of activities that could touch the lives of all the agencies by using the concept of integrating family history tools and genetics into whatever we do, because that will affect the practice of health care, disease prevention, and health promotion.

That would include that last point, which is what Debra was talking about, which was what was the point about the cataloging function? It is only a minor tool, or a minor service that the Department can provide, but I think you should give the Department the broadest possible advice that affects most of the agencies, rather than one or two. That would be my advice.

DR. FELIX-AARON: I mean, I agree with that, but I also think that the point that I was trying to make in terms of to me, this seems like a small, narrow, small, function, and not necessarily something that the Secretary in his unique role would necessarily engage in. So CDC is doing it out of the programs, and I understand that.

But I agree with you in terms of providing vehicles. So to the extent possible, to provide vehicles where those types of recommendations can be tapped on, I think is extremely important.

DR. REEDE: Can I ask a question there? So is there a role sort of somewhere in between these, as we look at these multiple federal efforts? And having some place or some mechanism for understanding what is being learned within those efforts, and to link that back to the private sector, and to the educational sectors? So that as this work is being done, it doesn't end up being done in isolation, but the federal government is actually cataloging the work that it is doing in a way that it can be disseminated easily across these groups. Slightly different, but I think if you're not sitting in this room, you may not know about the work that is being done across the multiple agencies.

DR. FELIX-AARON: I mean, one of the things that come out of this Department and the Secretary is the issue of coordination, and coordination around the departments, and speaking with one voice. So I think, if what we have here could be framed in terms of speaking with one voice, or one Department and really understanding to make our Department more efficient and effective in that area, that resonates, at least in my mind, with what I hear coming out of the Department.

DR. McCABE: Well, if it read, "To coordinate and disseminate genomics information," would that be better?

DR. FELIX-AARON: I think so. I mean, I don't know what other people around the table think, but I would that at least at a minimum, it would. It sort of captures some of the conversations that have been going on here, and it resonates with what I hear coming out of the Department, and at least the goals of the Department.

DR. TURNER: I was just sort of agreeing that to determine what the

Secretary's role is is important, so that we can phrase these in ways that he is able to respond. In addition to what she is saying, I'm struggling with the softness of the language. Someone earlier suggested that we stand up a little taller and say, we may not want to go as far as direct, the integration of genomics into the education, because I don't think he can direct things.

But to look at where we are, and then sort of envision how would he carry this out, I think the words she just mentioned are the operative words, if you will, that would get the job done. So I would suggest that we think in terms of, do we want him to make a phone call? Or do we want him to take this to some meeting? Or how do we want this to play out?

DR. REEDE: So coordination, dissemination, and pulling it together.

Debra?

DR. LEONARD: From this discussion, can we kind of take Number 2 up there, and I think what I'm hearing is that we pulled out one initiative, but there are steps, and there are other things, other initiatives. So maybe we should say incorporate genetics, family history tools, point of care, and educational support in the initiatives of the Department of Health and Human Services. For example, CAHIT, STEPS, that type of thing, and coordinate the dissemination of this into practice through private, state, and federal efforts.

But get in there that you want these things developed through the Department of Health and Human Services, and then you want that information that's developed there disseminated into practice. That is going to involve coordination and use of this information by professional organizations.

DR. REEDE: All right. I hear what you're saying. One of my questions for the agencies is the extent to which they can coordinate how the professional societies actually use this, as opposed to a coordination of how they disseminate the information that they have gathered. So I sort of see sort of a collecting what is going on, and being able to disseminate that, and to work with other organizations, but not really being able to coordinate after that, the activities of those professional organizations.

DR. FEETHAM: I think moving the discussion that we're having now about showing and giving the Secretary the guidance to look at this within all those key initiatives is really critical, and is a broader scope than adding it into some of these more specific processes. You're closer to that in Number 1, and promote and actively incorporate, and I think if you bring this overall concept of how this fits within many, or probably all of the Secretary's initiatives, I see as higher up the list, and more of an overriding. From then on, you're showing him some examples.

But I think you are losing it if you try to add all of those things, such as, such as, but I would bring it up, and I think it is closer to your current number one of incorporating interdepartment policies, but you're really talking about Department initiatives.

DR. REEDE: Ed?

DR. McCABE: I would ask that we try and get the big points down over the next 10 minutes, and then wrap this up. We're going to then take like a three to five minute break to try and make sure that we can get back to Reed and have him hear us, as well as us hearing him, and then proceed on. But if we can get the big ideas, we'll try and rough them out this evening.

DR. REEDE: Okay.

DR. LEONARD: Another big idea that needed to be added as a separate

bullet point was the education of the public. So that's a whole separate point that needs to be added.

DR. HANS: I'll wait for Sarah to get back down. Where I thought you were going, there are sort of two directions. One is to encourage, promote, and raise with the Secretary and the departmental leaders to incorporate the sort of concepts of genomics and genetics into a variety of ongoing activities.

The other direction which I thought you were going there for awhile, and now I have lost track of the numbers there, but is to say something like recasting one of the numbers that is up there, direct the HHS agencies to work together, and with federal, state, and private partners to, and I don't remember what words we used, collect all of the genomics education programs, and I don't remember where we were. And then work again with these same partners to promote the use of those materials through the various avenues that they have.

If you wanted to encourage HHS to go farther, even then just within HHS agencies, you could even ask that it be done in an interagency setting, like through the Quality Interagency Task Force that exists, that is a forum for interagency cooperation.

The second step is once you have sort of the collection in place, and you have sort of made the federal statement that this is what we should be doing, you then ask all the agencies to go back and look within their own programs, within their own regs, within their own activities, how do you appropriately incorporate it into what they're doing? Which certainly for VA and DOD, for instance, who run health programs, you have a more direct impact than you do just putting out recommendations to health professionals. So I'm sorry, Sarah, you probably weren't able to capture all of that.

MS. CARR: Sum that up.

DR. HANS: I was really looking down there where you have the red Number 1. So instead of framing it, because I thought that is where the discussion really was, instead of framing it as engage other health professionals, but rather to say direct HHS agencies, or direct the HHS to work among themselves, and with the state, federal, and private organizations that you have listed further down, to not facilitate, but actually catalog and disseminate genomics applications, and then work together to implement, where appropriate, within ongoing activities, or ongoing programs. We can work on wordsmithing that, but I don't know if you want to go that far, to be that direct.

DR. LEONARD: My concern is that I think we're losing the spirit of what is now the red Number 1. What the educational groups were asking for were cases that are relevant to everyday practice, so that they could do
case-based education. This has now morphed into something that is not getting at, because it wasn't clearly stated, I think, in this one to begin with, that what you were looking for were cases that you could use for educational purposes, for case-based teaching.

So now that whole case-based teaching, the need to get the cases that are out there in everyday practice today, has been lost from this list.

DR. REEDE: I'm wondering if that is not a separate issue in terms of support for programs that will develop and help disseminate these specific cases, or interactive learning modules, or whatever that might be.

DR. LEONARD: Right, but it is now not in this list anymore.

DR. REEDE: I'm wondering if that shouldn't just be listed as separate. This list is going off in another direction, so I'm wondering if this is a separate entity.

Two other things, and given our 10-minute time frame that is just about used up, if we look at some of the other recommendations, there was one with regard to what was Number 5, "encourage accrediting, licensure, and certification of health professionals," that that be changed. That was one of Debra's recommendations.

DR. LEONARD: It would be of health care and public health professionals.

DR. REEDE: Professionals.

DR. LEONARD: So certification bodies for health care. So up in the first line, Sarah, bodies for health care and public health professionals.

DR. REEDE: With regard to what was Number 6, a specific mention was made for us to keep in mind that cultural diversity and competency include the disability community. So I would question whether or not you'd like to add specific wording with reference to the disability community for Number 6.

Another item that we actually don't have time to word smith right now, but it was a strong point, was to have a recommendation that related to the ethical, legal, and social issues in training.

DR. McCABE: Any other big issues?

MS. HARRISON: I just had one about Debra's point about the public. I see that Sarah is trying to word smith something there to represent the thought. I just wanted to point out that Joe McInerney in his public comments did provide some wording for that, which I thought was pretty strong. So maybe that should at least be considered. One thing I wanted to add to it was to say that the initiatives should be done in a culturally sensitive and appropriate manner, so that we know that the initiatives that are put out there, that it is encouraged that some of them can be focused on certain communities that may otherwise not respond to other types of education.

DR. REEDE: Thank you.

DR. McCABE: Any other big issues?

(No response.)

DR. McCABE: If not, Sarah, we can try and work on these tonight and bring them back. But I think we do want to move on, so that we don't shortchange the public. What we're going to do now is take a three to five-minute break. Please don't go far. We are really just trying to link back up with Reed Tuckson. So please try not to go far.

(Recess.)

DR. McCABE: Before we proceed to the public comment portion of the meeting, I'd like to turn to Dr. Reed Tuckson, who is with us by phone -- that is sort of a hopeful statement -- to provide a brief summary of the inaugural meeting of the Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children, in which he participated last week.

This congressionally-mandated committee is charged with providing advice and recommendations to the Secretary of Health and Human Services concerning grants and projects authorized under Section 1109 of the Public Health Service Act and technical information to develop policies and priorities for this program that will enhance the ability of the state and local health agencies to provide for newborn and child screening, counseling, and health care services for newborns and children having, or at risk, for heritable disorders.

Given the mutual interest in genetics, and the relevance of our committees to one another, Reed serves as a non-voting liaison to the advisory committee.

I would remind everyone that the largest body of genetic testing that is done in the United States is through newborn screening with 4 million babies born each year, and each one of them tested for between 3 and 30 disorders.

So with that as a preamble, Reed, I hope we can proceed.

DR. TUCKSON: Well, I hope that you can hear me.

DR. McCABE: Yes.

DR. TUCKSON: I appreciate the opportunity to report on the Advisory Committee on Heritable Disorders and Genetic Diseases. It was ably chaired by Dr. Rodney Howell, who is well known to many of us, and Michele Puryear, who has been involved with our committee, was one of the principal staff people, and she has done a great job.

You have introduced the mission of that committee, which is that the advisory committee is charged with providing technical information to the Secretary for the development of policies and priorities for the administration of grants for newborn screening and related programs and providing recommendations, advice, or information as may be necessary to enhance, expand, or improve the ability of the Secretary to reduce mortality or morbidity in newborns and children from heritable disorders.

During its first meeting last week, after much testimony from federal agencies and others, we heard a lot about the state of the art and the challenges that are involved in the genetic basis of newborn screening. As a result, the committee decided to focus on a few key areas in which we could make a significant impact.

First, the preliminary goals include making recommendations for a uniform panel of conditions for newborn screening, assessing the capacity needs of states' newborn screening programs and how they should be able to implement those, and making recommendations on parent education, notification, and informed decisionmaking.

This recommendation was also because there is an extraordinary amount of variability in the way individual states either require, mandate, or conduct newborn screening activities. It is not based upon an understanding of the best science in genetic screening, it is not based on a consistent application of that science, and it is differential in terms of the ability of states to collect information for long-term follow-up.

The committee also plans to study newborn screening tests and use the product of their work to develop recommendations on a nationwide core set of tests. This work will refine and extend a study that HRSA commissioned with Mike Watson and his team at the American College of Medical Genetics to carry out, which is sort of a landmark work in this area. The committee also plans to look at the costs of individual genetic tests and the costs of tests at the state level, including administrative costs. Finally in this area, the committee will carefully consider state-specific information such as workforce needs, the ability to assess and translate technologies into practice, and public education.

In addition, recognizing that the addition of new screening tests is likely to accelerate, the committee plans to develop a system or a process by which it will consider whether and how these new tests should be integrated and incorporated into the diagnostic panoply available for newborn screening.

Finally, the committee is interested in research needs to improve newborn screening and will use the American College of Medical Genetics study to assist them in recommending gaps in knowledge that would then focus on priority research initiatives and needs. They're also going to work carefully on creating a working group on interagency

coordination.

The only other point that I would make is that the committee was very impressed by our committee's strategic planning effort where we engaged in that process of identifying priority initiatives that require immediate and long-term action. They were interested in how we accomplished that task, and they have been given information regarding the process and lessons learned that we have from our experience. So I thought that was very useful that our committee's innovations have now spread to others.

With that, let me stop. It was a very good first meeting, and I am encouraged by the direction that this committee is setting off on.

DR. McCABE: Thank you, Reed.

Any comments on the report? Emily?

DR. WINN-DEEN: I just want to know if there is a written summary that either you have, or will be provided by the people who ran the meeting, so that we could get that sort of for our background reading?

DR. TUCKSON: Terrific idea. There will be a summary, and I'm sure that that will be easily made available to you.

DR. McCABE: Any other questions or comments?

(No response.)

DR. McCABE: If not, Reed, we know we are ably represented on that committee, and thank you for doing that.

DR. TUCKSON: Well, I also must say that, as always, I had terrific support from our staff. Our staff was there and is always great.

DR. McCABE: Thank you very much. Will you be able to stay with us?

DR. TUCKSON: Yes.

DR. McCABE: Thank you.

With that, we now move onto the public comment. One of our critical functions is to serve as a public forum for deliberations on the broad range of human health and social issues raised by the development and use of genetic technologies. So we greatly value the input we receive from the public. We set aside time each day of our meeting to hear from the public, and we welcome and appreciate the views that you all share with us.

We also have received written comments that can be found in everyone's table folders. I would especially like to call your attention to several requests that additional time be provided to allow the public to comment on the draft documents being considered at this meeting.

In the interest of time, I ask our commentors to please keep your remarks to five minutes, if at all possible. Today we will be hearing from -- and I'll give this in order so you'll know when you're up -- Kathleen Rand Reed from the Rand Reed Group, Andrew Fawcett from the American Board of Genetic Counseling, and Barbara Handelin from Handelin Associates.

First, Kathleen. So please come up to the table, as Kathleen is doing.

MS. REED: Good afternoon. First of all, let me say thank you very much to Dr. McCabe for just a couple of more extra minutes, and I want to bring two issues to the record, and to the table.

I followed the bouncing ball, and that is I stuck to the priorities, and to the determinations such as overarching issues, and the priority short term, and the highest priority

requiring in-depth study. The two comments that I would like to bring to the table, the first one would be continuing on genetic education and training of health professionals short term.

I am an anthropologist and an ethnomarketer, and I wanted to look at genetic issues that typically come into communities of color, especially those where the communities are what is known as hypersegregated. In other words, they are more than 95 percent of that particular group, because oftentimes in those particular communities, it is a little bit more difficult to get information and ask some of the questions. And yet, if they are to be served as well, then those are the kinds of communities to look at.

First of all, let me also say thank you to Joe McInerney, who was gracious enough in January to allow me to present a poster for the NCHPEG meeting. The title was a little bit controversial, but it was called "U.S. Prison Policies, the Baby Daddy, and Genetics in the Hood." Let me tell you the key components of the poster, and I think you'll understand why I said it is controversial, but it is critical that these issues are addressed.

The key components for the poster were one, the removal and incarceration of young black males from hypersegregated intercity neighborhoods and some rural towns, the high incidence of teenage mortality from violence, poverty, and poor health, the resulting skewing of the male to female ratio, the phenomena of multiple matings, which often result in high consanguinity levels, i.e. the baby daddy.

A review of cohorts born during the height of the war on drugs in 1990 to 1995, are currently on average reaching puberty in the years 2003 to 2008. The matings in many of these cases, because we have a member of the community that is often missing now, which is the gatekeeper, the genealogical gatekeeper, which is usually little old ladies that used to know who everybody was, and who belonged to whom.

Some of these matings may be between half-sibs, whose familial relationships may not be known within the general community. The potential is for possible epidemics of an increase in autosomal recessive disorders in this population, as well as a "blame the victim" mentality, and possible genetic redlining.

The reason I bring it to this particular group is because part of the comments that I got from the head of some of the genetic organizations, especially state-run, were chilling. They were chilling in this respect. As much as I was told that I was right, and in fact, I even had a deep conversation with Barbara Willis Harrison to get a reality check, to see if what I was thinking was absolutely true.

Ms. Harrison told me yes, we see this when I'm out doing pedigrees, this is a reality. And yet, one of the problems that we run into is this issue of genetics and the whole coming to terms with the community, and PC. What I found when I presented this poster, and I've had conversations with Mr. McInerney as well, is that many of the genetic counselors said yes, no, we see this in the pedigrees. The problem is because we are white, we can't do the PC part, because some people might think it is eugenics.

Now, I'm bringing these controversial issues. Anyone that knows me knows that I'm pretty much the one that always tries to bring up these kinds of issues, because some people don't, and I'm very comfortable doing this. But I would say that one of the things that we need to really work on, and this is an emergent situation, because I also went to Chicago, Detroit, Oakland, and Philadelphia and checked in and found that this is also a real big issue there, in many of the urban areas, and it is emerging.

I would say that we need to get on this and take a look. But one of the

things that we need to do is create these bridges where we have deep conversations and issues around who is white, eugenics, and PC issues. As they say, get over it. We need to have deep conversations so that we can protect these communities. Let me move quickly, because I don't want to run out of time, but I throw that out there as an issue.

The second one is, and very quickly, you have already identified the diverse representation of the population in clinical trials has particular importance in genetic research, since genetic variation among populations may account for differences in disease prevalence, drug reactions, and susceptibility to environmental triggers, among others.

There is a problem. The problem is I just came back from California, spending time going back and forth and being an expert witness in several cases where minorities are now suing. They are becoming more sophisticated about genetics, and suing because they are having serious adverse events with regard to their pharmacogenetic reactions to various prescription drugs and their respective diseases.

They are beginning to sue both the institutions, the hospitals, the HMOs, their physicians, and the pharmaceutical manufacturers. In the case of minorities, often their primary care physicians are also minority physicians, and often these physicians rely on the details and warrants issued to them from the pharmaceutical manufacturers for their prescribing guidelines.

There is now a scientific gap, and a legal gap, that causes injury to both the minority patient, and in some cases, the minority physician. When a lawsuit of this type is filed, all parties are sued. The hospital, the HMO, the patients, physicians, and the pharmaceutical manufacturer. But the pharmaceutical manufacturers, however, have an automatic defense, namely the learned intermediary doctrine, or the LID.

The LID can best be described as the manufacturer owes no duty to warrant to the patient, but to the learned intermediary, i.e. the prescribing physician. The liability, if anything, rests with the physician, or the institution, for the prescription to the patient.

However, this learned intermediary doctrine has several caveats. Namely, the information given to the physician must be "adequate." You as a committee have already identified the the flaw in this process, where you say pharmacogenomics, page 2, paragraph 3. "The pharmaceutical industry has very little incentive to do pharmacogenomic studies on "already marketed drugs or generic drugs." Such studies are expensive, appear to offer no market advantage for the sponsor of the studies, and the identification of persons for whom the drugs would be ineffective, thereby creating a stratified market for their products."

This disconnect causes several downstream consequences. Number one, it relieves the drug manufacturers of a look back, and the action with current information on pharmacogenomic information to offer not only adequate information to the learned intermediary, or the physician and the institution, but it also does not allow for the development of new information where minority participants were not included, and need to be included, and they need to have that look back.

From an ELSI perspective, this disconnect between the marketed drugs and revisiting old data, increases the discriminatory aspects which already face minority physicians, i.e. HMO's, and managed care. But it further reduces their numbers in the ranks, and to sum it up, this may wind up giving a look see also into some health disparities.

I would ask that you review these relationships, because the same relationships that occur with the drugs are going to be the same kind of relationships that you're

going to experience when you look at pharmacogenomics and the genetic testing.

Thank you very much, and I hope that has been helpful.

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

Are there questions or comments?

MS. REED: That's Ms. Reed. Not quite doctor yet.

DR. McCABE: Questions or comments? Debra?

DR. LEONARD: So this type of look back is being done in Japan, where they are asking all pharmacogenetic analysis for drugs on the market be done for the Japanese population. Would you suggest that the federal government ask for this type of thing to be done in all the ethnic populations? Because it is not just African Americans, but Hispanics, and all the different ethnic groups in the U.S., and that would be a major effort, because we are so ethnically diverse.

MS. REED: I think it is the old expression, how do you eat an elephant? One bite at a time. I would think to go up against this, as you say, straight up, would be a problem. One of the things that I'm making a recommendation on, I sat on an IRB for Heart, Lung, and Blood at NIH, and one of the recommendations is that there is an easier way to do this.

When Hopkins had a problem with the literature search, and now people do real deep literature searches, because no one wants to be shut down anymore. There is so much of a proliferation of information now, until one of the things that I'm asking is that when you do your literature search, that you run out as one of your key words, the ethnic groups, just to get the information back and see if there is anything out there, and then you can incorporate that into your research design.

But I do think that there is a responsibility for some of the older drugs that have been out there. One on the new end to put the responsibility on the pharmaceutical manufacturer to do that kind of inclusive work, which they should be doing anyway, but to also on the older drugs, to begin to revamp and take a slow approach, but still take the responsibility of a look back.

DR. LEONARD: Maybe this could come, Joe Hackett, through the FDA with some sort of truth in labeling, such that for different medications, it has to be stated what ethnic populations it has been studied in, so that at least as part of what comes with a drug, you know that it has only been studied in Caucasians, or Caucasians and African Americans, or something like that, so that physicians have some ability to know the adverse reactions. They may see adverse reactions in other populations in which the drug has not been tested.

MS. REED: And you are getting some of that in the DTC, in the direct-to-consumer advertising.

DR. HACKETT: My own perception is when you say that drug companies are very reluctant to do this, and we are trying to work with them on the newer drugs, it is all voluntary at this point. I can say again this year that there won't be any new diagnostic tests in using genomics this year, so it is slow.

MS. REED: At least it is on the table for you to examine. Thank you again.

DR. McCABE: Thank you, and I would point out that as we think about different ethnic communities, we need to think about genders, too, because that has been a problem in testing drugs as well. Thank you very much, Kathleen, for bringing that to our attention.

Next is Andy Fawcett from the American Board of Genetic Counseling. Andy?

MR. FAWCETT: I'm here today as a member of the Board of Directors of the American Board of Genetic Counseling. I'd like to begin by thanking the committee for allowing us this opportunity to provide comment.

The ABGC is a national accrediting and credentialing body of the profession of genetic counseling. The ABGC establishes minimum requirements for graduate programs in genetic counseling, and develops a criteria by which individuals become eligible to sit for the certification examination.

As this committee is well aware, genetics is one of the most rapidly advancing areas of scientific research, with clinical applications in practically all areas of medicine. The advances in medical genetics are forcing fundamental changes in the way health care providers practice medicine and think about health and disease.

Knowledge about genetics and its social and ethical implications is becoming increasingly essential for many health care professionals, and must become an integral part of their curriculum. The draft resolution on genetics education and training of health care professionals developed by this committee clearly recognizes this need, and makes several solid recommendations related to integrating genomic concepts into our health care system.

The ABGC, however, feels that it is critical that a distinct and separate focus on the education and training of genetics health care professionals be included in this resolution. As certified genetics professionals, we recognize that the demand for genetic counseling services will continue to increase, and unless more genetic counselors are trained, there will not be enough counselors to provide care to all patients and their families.

Many certified genetic counselors devote significant time and effort educating other health care providers. The primary objective of these endeavors is to teach these providers to recognize a genetic condition in a patient, handle more straightforward genetic issues, and develop a relationship with genetic professionals in their community as a resource upon which they may draw to provide optimal care to the patient.

This goal is not to teach other health care professionals to provide comprehensive genetic services. The issues surrounding genetic conditions are frequently complex, and as a result, a team of genetics professionals representing multiple genetic specialties may be required to provide this type of service.

For example, genetic counselors have unique and extensive training in human genetics and counseling skills. They take didactic course work in human, medical, and clinical genetics, counseling theories and techniques, bereavement, crisis intervention, cultural competency, social, ethical and legal issues related to the delivery of genetic services, health care delivery systems, principles of public health, teaching skills, and research methodology.

Genetic counseling programs accredited by the ABGC also provide extensive clinical training involving over 800 hours of field work, teaching, laboratory experience, as well as research. Genetic counselors are taught to prepare, deliver, and evaluate educational programs as they apply to various groups of learners. They are specifically qualified to deal not only with the complicated genetic and technical issues that often arise in the context of genetic evaluation counseling, but also the complex psychosocial, ethical, and legal issues with which patients and their families struggle.

Adequate financial resources must be in place so that an adequate number of individuals can be trained as genetic specialists. While the ABGC recognizes that educational efforts related to genetics must be aimed at our entire health care system, we encourage this committee to acknowledge that equally important is support for the continued existence of health care professionals who have specialized training in genetics.

Genetics health care professionals, such as genetic counselors, have been, and will continue to be, the ones who train and educate other health care professionals about the many complexities of genomic medicine.

Seventy-seven percent of all genetic counselors are currently involved in the genetics education of physicians and medical students, according to a professional status survey administered by the National Society of Genetic Counselors in 2002.

One-third reported teaching genetics to other health care professionals. Many genetic counselors have developed and implemented innovative educational models that facilitate the genetics education of other health care professionals and students. They are also involved in forming public policy related to genetics, and actively participate in genetics education programs for the public.

The recommendations made by this committee to ensure that genetics education and training of all health care professionals is adequate, will only be successful if there is an adequate genetics workforce to implement these recommendations.

Currently, there are 1,811 certified genetic counselors, and 25 accredited genetic counseling programs, and graduate programs in the United States, and three programs in Canada. Of those who responded to the professional status survey referred to earlier, only 6 percent indicated they were non-Caucasian.

If genomic medicine is going to be equally accessible and practice in a culturally sensitive manner, individuals from minority populations must be recruited into the genetic counseling profession, and programs must be established in less populated areas of the United States.

In October of 2003, Robin Bennett, past president of the NSGC, presented data to this committee related to increasing the number and diversity of genetic counselors. To promote the training of genetic counselors, Ms. Bennett recommended that funding and granting opportunities be made available to support students and faculty in current genetic counseling programs, as well as to promote the establishment of new training programs.

The ABGC strongly supports the allocations of funds for this purpose if we are going to be successful in increasing the number and diversity of genetic counselors. Genetics health care professionals must reach out to other health care professionals to help them learn about the great promise and potential pitfalls of genomic medicine.

For this to be accomplished, we must ensure that there are competently trained genetics professionals. We strongly urge this committee to make specific and distinct recommendations to support the continued training of individuals in the field of genetic counseling, encourage increased diversity in our profession, and encourage those uniquely trained individuals to demonstrate their competency through certification and licensure.

Thank you.

DR. McCABE: Thank you, Andy.

Questions or comments for Mr. Fawcett? Yes, Debra?

DR. LEONARD: I think that this is an excellent point. We have often

discussed in this committee the paucity of genetic counselors, and so I think this is sort of an oversight in our education resolution, and we may want to consider addressing this in some way.

DR. McCABE: Thank you.

Any other comments or questions?

(No response.)

DR. McCABE: Next we have Dr. Barbara Handelin from Handelin Associates.

DR. HANDELIN: Thank you, Dr. McCabe and members of the committee for allowing this additional opportunity to provide some comments on your excellent deliberations, resolutions, and recommendations.

Let me just begin by saying that I'm here today principally representing myself as a Ph.D. geneticist who has been a technology developer and early practitioner of the provision of laboratory services and genetics in the commercial sector, as a consultant to large and small companies, and academic centers in the development of new genetic technologies and their implementation and practice.

I also speak from some 15 years of working with the IRB community in improving the oversight of genetics research involving human subjects, especially those that were sponsored by private industry.

I provided some written comments. I'm not going to reread those to you here today, but rather to move on to some additional thoughts that I have had in the meantime, in particular influenced by your conversations this morning.

I wanted to respond to the question that Brad Margus raised earlier about what may be broken, or is there something broken in the practice of medicine today with regards to genetics inclusion? I had the following thoughts.

While I'm routinely amazed by how often the genetic basis of a set of symptoms is not considered in a differential diagnosis, I also have to admit that I have an equal number of stories of people who have spent years of suffering and expense, failing to get a correct diagnoses, even when genetics is not involved. I know that all of us do.

I think that this is a reflection of the way we are perhaps not teaching physicians to think about a constellation of symptoms, about how to ask enough questions and hear enough answers about a patient's condition. But most importantly, how to think through a complex problem, considering many alternative diagnoses, as well as the physician being able to think about what is all the data that I'm going to need to collect in order to weave amongst those possible alternatives.

I would posit that perhaps this also says, most importantly, that we have far exceeded the capacity of the majority of physicians to keep a reasonably complete fund of knowledge of human disease in mind as they are seeing patients. Genetics are not.

In this vein, I would strongly support the idea that arose earlier today, I think originally by Dr. Tuckson, and then echoed by Sarah Carr about the timeliness of promoting the development and implementation of information technology systems that can support routine clinical practice, including the suggestion of genetic etiologies or contributions to common disease in everyday practice.

But I make this suggestion with a caution, and that being that these sort of practice tools have been developed previously and marketed by private companies, only to be

rejected by many physicians because they don't like the feeling that a computer may appear to be doing my job. That kind of sense of pride in providing knowledge and experience is a barrier to the acceptance for such tools, and it causes me to wonder if perhaps we need to see some kind of requirement for such tools in large medical practices, in large institutions, or the requirement for such tools to be incorporated into new training programs.

So I'd also like to speak to several other points that were contained in the various briefs that the committee provides as background. In particular, I'd like to speak to the brief on large population studies. Genetics as a discipline, I learned when I was a Ph.D. student, is not the study of inherited traits, nor the study of inheritance itself, but rather more fundamentally, the study of variability. Variability in populations is sourced through inborn traits of a species. Variability in populations is the source of our biochemical patterns, our physiology, our response to infectious disease, to nutrition, in the form of allergies, for example, to exposure to xenobiologicals, plant toxins and venoms, to daily life stress, and to administer treatments, as in the case of pharmacogenetics.

Variability in populations is the source of our racial and other group traits, as we share specific variants with family members, and choose parenting partners within our close at hand community groups. Variability in populations allows us to be at once a part of a group or sector, and to be individuals.

I would posit to the committee that the basic goals of our pursuit of knowledge about the human genome is fundamentally about understanding the variability in populations. Therefore, ensuring the potential to undertake such large population studies all across the United States through the various strata of our diverse population seems to me to be among the most important missions of all persons and committees who are charged with overseeing the exploration and application of genetic technologies in research and medicine.

So I hope that you here, and those at the NIH, and the NHGRI, for example, will focus on finding ways to list the main barriers to the ethical and productive conduct of large studies. I agree wholeheartedly with the observation made in the committee brief, that "the lack of the universal health care system, and the lack of a uniform electronic medical record system here in the U.S. are likely two very important barriers today."

The simple promise of guaranteed health insurance and health coverage, as are seen in other countries in the world, such as Sweden and Iceland, go a long way to encouraging people to participate in such large research protocols.

Other safety concerns for us is subjects, of course, about privacy and confidentiality. I would suggest that it has been now my conclusion I guess that we cannot ever guarantee absolute privacy, or that we could hold out all individual details completely confidential. Therefore, it seems paramount to me that we must feel confident that those who may choose to attempt to harm us through use of our private information will not be tolerated, that there will be severe consequences for such abuse of privacy.

I have an uncle who is an attorney who used to say that no one would cheat on their taxes if the death penalty were the required sentence. And so to be clear, while providing clear requirements for utilizing all the available methods for shielding private information from wrongful use, I believe it is equally important to develop a system of clear consequences for such wrongful acts, to send a message that as a society, we will not tolerate such violations of respect for persons.

In conclusion, I just want to say that I am using this language of respect for

persons, which I take from the Belmont Report, one of the three general principles laid out by the Belmont Committee in the Protection for Research Subjects as a way to remind ourselves, again, that the reason that we have rules and regulations about the ethical use of human subjects arises from the misuse of genetic and other kinds of information here in the United States, and in Germany decades ago.

So I urge us to return to those simple principles, respect for persons, beneficence, and justice, as you continue in your important work here. The final point that I would like to take a minute to make is to applaud you on your thorough and thoughtful treatment on the reimbursement and coverage issues for new technologies, including those in genetics.

Since the first application of our knowledge of genes and performing carrier tests and prenatal predictive tests for single-gene disorders, we have been practicing a new kind of preventive medicine. I suggest that today, our nearest benefit from the genome knowledge base remains primarily in preventive medicine. As we see the very first of new cancer screening tests, first tests to identify patients who will or will not benefit from toxic chemotherapies, these are the kinds of new products that I think are going to typify the implementation of genetics in the next decade.

Therefore, I would especially encourage you to drive home the importance of creating a sea change in our coverage for preventive medical services, so that we can encourage to participate in the research necessary to get there, as well as in the enjoyment of those benefits.

Thank you very much.

DR. McCABE: Thank you, Dr. Handelin.

Any questions or comments, anyone?

(No response.)

DR. McCABE: Thank you very much.

I'm sorry. Emily.

DR. WINN-DEEN: I just had a question. How much do you think of the reason that genetics isn't considered is because physicians in their time constrained, by the clock office visits, are basically applying the 80/20 rule, where you take the most common diagnoses and sort of farm everybody through those, and just don't really have the time to consider the less likely scenarios, or the more infrequent scenarios?

DR. HANDELIN: Well, I guess I would question whether genetic etiologies, or genetic components of many common diseases are typically the uncommon explanations for many presenting symptoms. But I agree with you that certainly a reluctance to consider many other alternative diagnoses clearly falls principally from a lack of time right there in the office, as well as a lack of time to incorporate new ideas, new concepts, and new knowledge into your fund of knowledge. Again, I think it is pointing to the need for some assistance in doing that.

DR. McCABE: Any other questions or comments?

(No response.)

DR. McCABE: Thank you, Dr. Handelin.

The way we will proceed now is we're going to have Cindy Berry give her presentation, and then we'll take a break before the discussion of coverage and reimbursement.

At the March meeting, the committee decided that this topic, coverage and

reimbursement, would be the subject of its first major report. A draft report was prepared by staff, and can be found at Tab 5 of your briefing book.

I'd like to take this opportunity to thank Cindy Berry for her leadership on this issue, and Suzanne Goodwin, and Amanda Sarata for preparing the draft report. I think they have done a remarkable job in a very short amount of time. I think we should acknowledge, however, that this is a draft, and neither Cindy nor the staff have given it any "final blessing."

Also, as I indicated before, I doubt that we will complete the discussion of this topic here, but that it will proceed into the next meeting. So now at this time, Cindy is going to give us an overview of the scope and content of the issues covered.

Cindy?

MS. BERRY: Thank you, Ed.

In the interest of time, I'm not going to go into enormous detail on each of the topics that are covered under the report, but rather give a general overview of how the report is structured.

Many of you, Reed in particular, have looked at this in great detail and have provided some comments, and this afternoon's efforts are going to be focused on really taking a good, hard look at the report. So at this time, I'm not going to go over specific language. We all are going to have edits, comprehensive edits, I suspect, on some things that have been overlooked that we will want to make sure are included in the report, and there are things that may not be addressed adequately, and we'll need to flesh them out in greater detail. But I'll just give you a general feeling for how this report is structured, and then we can proceed to the discussion phase where we can get into some of the details.

As you may remember from the last meeting, we did have a pretty elaborate priority-setting process. Out of that process came coverage and reimbursement as one of the priority issues that this committee should look into, and coverage and reimbursement being one of the key barriers to access the genetic technologies.

The purpose and goals of the report really are to give an overview of the current state of play. What is going on right now in the area of coverage and reimbursement for genetic technologies and services, and how can we improve that?

The sections of the report are as follows. We have an introductory section of course. Genetic technologies and services, what are we talking about when we talk about genetic technologies? Reed has brought this up in the past, and I think we may need to do a little bit more work there as in particular, let's define this. What is the scope of genetic technologies and services that we're talking about? General principles.

Then in the background section, the report delves into our health care financing system in the United States. How are coverage decisions made in federal health care programs, and in the private sector? How are services and technologies being reimbursed? What is the billing process? And then after that general overview, the report delves into the barriers to coverage and reimbursement, and of course the recommendations section is something for this committee to come up with.

Basically the introduction sets out what the problem is that genetic technologies can help enhance clinical care and improve outcomes, but there are limitations in terms of what is covered, and what is reimbursed, and those are barriers.

This section on genetic technology, as I mentioned, goes into what are we

talking about when we talk about genetic technologies and services? What distinguishes them from other health care services and technologies?

The general principle section really has sort of an outline of some things that we have discussed in this committee in the past, and that undoubtedly we will delve into in greater detail in the future. It is not a comprehensive or extensive list, you will see others there at the bottom as we go through the discussion this afternoon, I think folks will come up with additional principles that probably need to be incorporated in this report.

As I mentioned, this is the general overview of our U.S. Health Care System, the different federal programs, the private sector programs, managed care, and then the issue of the uninsured, and underinsured. The section on coverage decisions reviews how Medicare makes coverage determinations, and then it goes into the private sector determinations, which really are varied. Every health plan is different, every health plan has different guidelines, and the results and outcomes are different.

The situation is really the same in terms of payment. In Medicare, you do have a system for determining what is reimbursed, and the report does go into what the current payment rates for genetic technologies and services are, and we'll undoubtedly have a discussion about whether those are sufficient. I suspect based on previous testimony and comments that we've heard, that payment is not sufficient for many of these services.

The report also goes into billing processes. Coding, CPT codes, and who is entitled to bill for genetic services and technologies. The next step, of course, is to then examine the specific barriers, barriers that are particular to the Medicare program, barriers to access that are particular to Medicaid and state programs, and then of course there are barriers that are applicable to all insurers, whether they are in the public or private sector.

We'll just run through a few of these very quickly. In Medicare, for example, the screening exclusion is one of the barriers that the report identifies. If there are no signs, symptoms, complaints, or personal history of a disease, and there is not an injury, it is going to be difficult, if not impossible, for Medicare to pay for a screening test. That, of course, is based on the Medicare law itself. It is not something that you can necessarily fault CMS for. They are going by what the statute dictates, and so this is a matter that we'll discuss later, is a change in the statute warranted.

The report goes into detail about local medical review policies and national coverage decisions. There is sort of a tension in some respects between local decisions as to what is covered, and then a national coverage decision. How do those interact, and how do they affect genetic services and genetic technologies?

Genetic counselors, are they able to submit bills to Medicare for their services? The answer is no, according to the statute. There are issues with regard to private health plans undoubtedly as well. Medicare is a national leader in health care. This is an interesting point, because in many, many cases, we see that Medicare really sets the stage and serves as a guide for other insurers.

So if Medicare covers something or doesn't cover something, often that is replicated in the private sector, but not always. My personal experience, and others will have experience here as well, when it comes to preventive services, oftentimes we see the private sector taking a more proactive role simply because the Medicare program is really bound by statute. As we all know, Congress doesn't move too quickly, and it is very difficult for a legislative body to keep up with advances in health care.

In this case, sometimes the private sector actually is able to move more quickly. So this will be a discussion in the report that explains why the Medicare program is so important, not only for the Medicare beneficiaries, but as a model for other insurers, but conversely, maybe there are some lessons that we can learn from the private sector in terms of their ability to incorporate some of these technologies in health care.

The report goes, as I mentioned, into state programs, and then we have barriers that are applicable to all insurers, and all health care programs. Medical necessity criterion, and of course one of the underlying issues, threshold issues that we will undoubtedly discuss in comments today has to do with the informational value of genetic tests, and genetic test results.

If it is just great for someone to know the result of the test, does that warrant coverage under a federal health plan, or private health plan. There is the issue of the future benefit, testing someone for a genetic disorder. If there is no treatment available right now, is that, again, an informational benefit? And also, if someone is not experiencing any signs or symptoms, if you test now, that will produce some benefit at some point in the future, is that a barrier to coverage, whether it is in the federal health programs, Medicare, or whether it is in the private sector?

Of course, the role of cost and coverage decisions. It is not explicitly considered in the coverage decisionmaking process, but undoubtedly it is a factor, and we'll have to be mindful of that.

Experimental exclusions. Some of these technologies fall under that category, and that's another barrier to coverage and reimbursement. CPT code modifiers, that's an important part of the report, because it outlines really the process for getting a CPT code, and what that means for coverage and reimbursement, and for payment. There are no CPT codes specific to genetic counseling, and that could be a barrier as well.

Evidence-based coverage decisions, we talked a little bit about that this morning. It is a current theme in our comments today, as well as what is going on in the report. Obviously a lot of insurers, rightly so, feel the need to have some kind of data with which to make a decision, and on which to make a decision to whether a treatment, test, or some technology or service should be covered.

We have talked a little bit about how important it is to have this kind of research available so that we can help make the case if certain technologies should be covered. Right now, there is a little bit of, and I think actually not a little bit, a pretty significant need for that kind of information.

The report goes into payment rates, and the issue of low reimbursement. It is not necessarily unique to genetic technologies and services. It is a common complaint, I think, from many providers of health care services. But the question is if the reimbursement is so low, and oftentimes is below the cost of delivering the service, or delivering the product or the test, what impact does that have on access?

There are broader issues that the report discusses. It doesn't attempt to resolve all of them, because these issues transcend genetics. They really go throughout the health care system, but we have talked a lot today about, and we will continue to discuss the issue of health disparities.

Education and training, we have spent a lot of time on that today. I think probably I should close with this, topics for discussion this afternoon. The overall structure of

the report, the content, the tone of the report, the general principles, and then we have to come up with some recommendations.

Now, I'll bring up a point. In one section of the report if you have read it, there are some boxes on policy options. My view, it might be a little bit confusing to have policy options there, and then have recommendations someplace else. We might want to figure out how we want to structure that. I don't know if others agree or disagree, but I think the charge for this committee is to finalize the report, make sure that we've captured all of the basics, and that the language is something that we all feel comfortable with, but really hone in on some of the recommendations, concrete recommendations, that this report can make to improve coverage and reimbursement for genetic technologies, thereby improving access to these services.

Here are some of the topics, and I'll leave this slide up for our discussion. These are topics that staff have identified that we could go into all of them, some of them, a few of them, and it would be important to get the thoughts from the folks on the committee as to priorities, issues that you want to focus on, and that you think should be the basis for our discussion today for purposes of putting together some concrete recommendations for the last part of the report.

DR. McCABE: Thank you, Cindy.

I think with that, why don't we take a 15-minute break. Again, we have the refreshments up here for the ex officios, the members of the committee. We'll reassemble here in 15 minutes, so around a quarter after.

(Recess.)

DR. McCABE: Let's begin again, please. I'd like to remind everyone that our goals for the next roughly two hours are to discuss the draft and develop recommendations to be included in this report to the Secretary. Again, I doubt that we will finalize this over the next two hours.

We have written public comments and have heard public testimony regarding the report. Cindy has identified some areas where we might want to focus our attention, so I will turn it over to you now, Cindy, to lead our discussion for the next two hours. Our plan is to finish up here at 5:30 tonight.

In case I forget, let me remind everyone that we are starting earlier tomorrow. We will start at 8:00 tomorrow morning. Thank you.

Cindy?

MS. BERRY: Based on the amount of time that it took us to go over the education and training resolution, it seems like, and here is just a suggestion, but let me know if anyone disagrees. That for specific edits, because the report is so extensive, perhaps we should give a deadline to ourselves to hand write, or email specific wording changes and things like that, and get them to staff, to Suzanne. I don't know what deadline you want to put on there, and focus our discussion today more on the bigger picture.

Well, the structure of this is wrong, or we need to add a section on such or such, or here are what our recommendations should be, that kind of a thing. I don't want to unilaterally impose that, so tell me if you disagree. But it seems like that might be a more efficient way to deal with this today.

DR. McCABE: Our next meeting is October 18th and 19th. We'd want to have time to get those well incorporated. When would be a reasonable time, Cindy and

Suzanne? A month, six weeks before that? Six weeks?

MS. BERRY: Where does that take us?

DR. McCABE: If it was six weeks, that would be the beginning of September.

MS. GOODWIN: What does this relate to?

DR. McCABE: To get in feedback on the document even sooner than that. Mid-August? Do I hear mid-August? Even earlier. Staff is much more reasonable about time than I am. So early August? Mid-July?

MS. GOODWIN: Mid-July, or even early July, depending on how extensive the editing process needs to be. Part of that relates to when, if we plan to go out for public comments, but in time for the October meeting. So there will have to be significant time to prepare for the Federal Register notice for public comment, and also to give the public sufficient time to respond. Also in the Federal Register notice to make sure that we publish, or have available, a revised draft, based on your edits and today's discussion.

MR. MILLER: How about Thursday?

(Laughter.)

DR. McCABE: Well, if we give people a month, that would be roughly the 16th of July. If we want to do it even earlier than that, we could go for the 1st of July, which is a Thursday, as per Paul Miller. Okay. So July 1. So two weeks would be July 1. Would that work for staff?

MS. GOODWIN: That's good. Thank you.

DR. McCABE: Okay. So July 1 it is.

DR. PHURROUGH: For those of us who are representing other people, for those people that we're representing, do they get an electronic version of this?

MS. GOODWIN: There is a copy available, a PDF copy on our website.

DR. PHURROUGH: Okay.

MS. GOODWIN: There is also one that was sent electronically to you before the meeting, but we can certainly send it out again after the meeting to make sure that you have a copy.

MS. BERRY: Emily?

DR. WINN-DEEN: I was going to suggest that perhaps what we should do is rather than focusing, as you mention, on edits on sort of the background information where I think we've gotten some comments in that some of the background information needs updating, but I think it is very important for us to discuss the policy recommendations. I certainly would like to see, because this is going to be a topic that I think already has evoked a lot of public comment and is likely to evoke more public comment, that we try and discuss those policy recommendations in this committee.

But also I would like to make a suggestion that it be circulated one more time for review to the committee before it gets posted on any site anywhere, so that if we have additional comments on each others' comments, we can sort of get that all worked out ahead of time, if it is possible to do that.

I know I got a whole bunch of calls the day it went up on the website. What are you guys thinking about? How did you get to this, that, and the other thing? I'm like, gee, I haven't even read it yet, I don't know what we're thinking about. I think when we have subcommittees, we need to somehow share the subcommittee work with the whole committee,

at least a little bit of time frame before it is shared with the --

MS. BERRY: Well, in this case, we didn't have a subcommittee at all, this was a staff prepared document. Now, is there a requirement that it be posted? That's the question I have, is are we able to circulate it amongst ourselves and comment on it without it being posted until it is in final form? Or is there some sort of requirement for our committee that it needs to be posted?

MS. GOODWIN: The reason why this draft was posted before the meeting is, well, first of all, staff was working on it up until right when the briefing books were sent out. Part of the reason we opted to post it before the meeting was part of the FACA requirement, the Federal Advisory Committee Act, requires us to make documents that will be discussed in detail during the meeting, available to the public, so that they can participate in the discussion.

DR. WINN-DEEN: Yes. So I didn't have any problem with it being publicly posted. I just felt like we were caught a little bit flat without having something that went out, representing this was a work product of the committee, and the committee hadn't even seen it. That was all that my comment was directed at, that I'd just like to have a little bit of a time window for the committee to actually have read it and thoughtfully gone through it before we start getting a lot of feedback.

DR. McCABE: Yes, it does have to do with FACA, and our practice with respect to FACA. That was actually my decision, that we would do that, because we wanted the public to be aware of this before we came here, so that was my decision, and I'll take responsibility for that.

DR. REEDE: I think part of it is if you look at the top, on each one of them it says it is a staff draft, however, and that it didn't represent the views of this committee, or of the government. I don't think anybody reads the fine print, but the fine print did say that.

MS. GOODWIN: But certainly before the next meeting, there will be an opportunity to go back and forth as a committee by email to revise the next draft.

DR. WINN-DEEN: Thank you.

MS. BERRY: Does anyone have any suggestions for kicking off the discussion on policy recommendations? Are there issue areas that you want to prioritize for us? Steve?

DR. PHURROUGH: I'll make a few comments from the CMS viewpoint, since we are seeing it for part of the discussion. First of all, on the draft in general, a big concern in your first principle is we need to collect data, and yet that was very minimally discussed throughout the report, the importance of a coverage decision being evidence-based.

We sort of tied coverage and reimbursement together, and you really need to separate that concept. Certainly speaking for CMS, and I suspect for most private insurers, at least in reading Allan Korn's comments, and Reed's also, when we as payers make decisions about the kinds of things we're going to cover, we want to make sure that there is a good evidentiary base for it.

That evidentiary base, at least in the Medicare world, is there are good quality controlled trials that demonstrate that the application of this technology improves the outcomes of beneficiaries that we have responsibilities for.

It brings up a whole host of questions in genetic testing that we, as a society, and we at CMS, need society to have this answer. One is in some cases, genetic testing will alter treatment, but in many cases, it won't, it will just be information. I think that

something perhaps even broader than us is a question of whether health care dollars should be expended for information when it will not alter outcomes. We've had that discussion internally, and that is certainly a topic for further discussion.

More specific, policy issues, specifically around the screening exclusion. The draft is correct that it has been a longstanding Medicare policy that Congress prohibits us in its language from paying for tests that are screening in nature, or in most cases, paying for preventive services.

We have had a lot of discussion in the last several months as to whether in fact it was Congress' intention. There are some viewpoints that say that CMS could change its mind without congressional input. Even those who think that it could only done through rulemaking, but even those who think that may be possible strongly suggest that we should let Congress tell us to do that.

Since Congress has been very involved in adding screening and preventive benefits over the last decade, and they seem to enjoy that, and they have yet to give us a broad mandate to do screening and prevention, preferring to, how shall I say, receive the accolades of adding a new benefit that they may get from their constituents, which is in fact their job.

The agency could, based on our legal authority, perhaps change our screening exclusion, but we're not sure that that would be well received in Congress, and believe that the congressional direction is probably the preferable solution. And in fact, there is legislation in front of Congress now sponsored by a whole host of members on both sides of the aisle that I believe has been encouraged by the Partnership for Prevention, that would allow us to do coverage determinations on screening and preventive services, if they were in fact recognized as beneficial by a national organization.

So there is some legislation there. We are not a lobbying organization, but as we discussed on the education and training earlier, that may be a role for this committee to encourage.

On the other policy of who can and can't be paid, that is very clearly a congressional action. We can only pay people that Congress says we can pay. Congress has been very hesitant over the almost 40 years of Medicare to expand that list, having only expanded it a couple of times in that 40 years from physicians to one other occasion, nurse practitioners, CNSs and PAs and in very narrow instances, physical therapists, and most recently, a very narrow indication for dieticians.

That is going to be an extremely difficult barrier, I suspect. Congress is not very receptive to adding new providers. So that is the only way we could pay genetic counselors, and that is going to require some significant support from others outside this committee to encourage them to do that.

MS. BERRY: Steve, do you happen to know the bill, the legislation that you referred to? The sponsor of the bill number?

DR. PHURROUGH: I don't have that with me. I can see if I can locate that, and provide that to the staff.

MS. BERRY: Terrific. Does anyone else have any comments? Emily?

DR. WINN-DEEN: Well, so, in the great minds think alike kind of thing, one of my comments back was why can't we encourage Congress to enable groups like the preventive task force when those recommendations come by a "deemed body" that any recommendation that is coming from certain institute medicine or USPSTF, that those would be

automatically then covered prevention services.

I guess what I'd like to ask is so which way do you think the wind is blowing? And is this something, because CMS and CDC that is sort of running the prevention side of things, both report to our guy, who we are supposed to make recommendations to, Tommy Thompson, is it possible for us to send something up to Secretary Thompson that basically says, we think this would be a good thing? Or are there other political things that you should make us aware of?

DR. PHURROUGH: I'm not an expert in how the legislative process works. But as I understand it, Secretary Thompson provides to Congress every year what is called the legislative budget, which has nothing to do with money. It has to do with these are things that we recommend that you consider for enactment.

Someone will have to check and see if I'm correct, but I think that is the way the system works. We could encourage the Secretary to add that to that particular recommendation to Congress, which is not then us as government employees lobbying Congress directly, but using the vehicle that Congress has established, this legislative budget. I think that's the terminology.

DR. McCABE: Yes, I think that's an important point. The process is that we advise the Secretary. We can be specific as we have with genetic nondiscrimination with respect to influence on the legislative branch, but that's the mechanism. It is through Secretary Thompson.

DR. WINN-DEEN: Yes, I think my point was that since sort of both parts of this equation report to him, that it is definitely sort of 100 percent under his umbrella to be able to do something like that.

DR. TUCKSON: This is Reed. Can you all hear me?

MS. BERRY: Yes.

DR. TUCKSON: (Inaudible.)

DR. FELIX-AARON: That's right. AHRQ does do prevention as it applies to clinical practice, and what goes on between doctors and patients as distinct from sort of the community preventive services, which is under the purview of CDC.

Reed, are you finished? Because if you aren't, I'd like to make a point here, but I'll wait.

DR. TUCKSON: Go right ahead.

DR. FELIX-AARON: Right. I mean, in listening to both Steve and the conversation which went earlier, somebody mentioned the partnerships for prevention. The question, I hear us going around, is sort of what are the levers for prevention?

I think one we can do is sort of say, this is genetics, how do we push this forward in terms of looking at coverage for these specific services? What I hear is that genetic services are part of a larger category of preventive services. And so the question I think we're faced with is what are the levers for prevention, and who are the major stakeholders?

So I was very struck by your point, Cynthia, where you said that some of the innovators in this area is not necessarily the Medicare program, but actually private insurance companies, in that they are much more advanced in terms of prevention than Medicare. And so those groups are ahead of the pack, so to speak.

So the question for, or not the question, but a question for us is really trying to understand what is going on in prevention, and who is pushing prevention in this country,

and what can we as a group do to sort of piggyback our efforts either to supplement what they are doing, or to augment what is going on in those circles, and what advice for the Secretary, how can we provide the Secretary with guidance which strengthens what is going on in that arena?

MS. BERRY: Reed, you're probably the best person to address it. Just to put in context, my comment wasn't at all that CMS doesn't want to go down the road of prevention, but simply their hands are tied by the structure of the Medicare program, legislation in Congress, and all of that.

The private sector doesn't have as many of those constraints, and so they're a little bit more at liberty to be innovative, and to respond more quickly to innovations. But Reed, I'd defer to you. Do you care to address that point?

DR. TUCKSON: (Inaudible) how do we give the Secretary a reasonable body of work to get around? I think that the implicit guidelines are that we do want to see appropriate genetic prevention diagnostic services to be incorporated into the Medicaid/Medicare rules.

The issue is what does that mean? I think that (inaudible) automatically anytime there is a partnership or prevention (inaudible) there are some other issues here. I think what our challenge is, without having to (inaudible) that is the issue.

MS. BERRY: Reed, you cut out a little bit at the end where you were making the big point.

DR. TUCKSON: Well, (inaudible) responsible way, and those issues and those questions are no different for the public sector than they are for the private sector.

DR. WINN-DEEN: Reed, can I ask you a question from the private insurance point of view? How far do recommendations from things like the U.S. Preventive Services Task Force go in the private sector to encouraging coverage for services, just sort of in general?

DR. TUCKSON: They are exceedingly important. Because again, they provide the kind of very rigorous evidence-based analysis for (inaudible).

DR. PHURROUGH: I'm Steve Phurrough from CMS. One of the difficulties in waiting for the USPSTF is that their process is a very detailed, slow, arduous process, as it should be. I think based upon the amounts of evidences that are out there today, the benefits of genetic testing, that they're a long way from coming up with recommendations that if we were to be told to follow them, and aside, our preference would be we're given permission to use their recommendations versus told to follow them, but that's a different subject. I think they are a long way from coming up with those particular recommendations.

I think the real emphasis that we need to be encouraging is there are some policy changes that need to occur, but we need an evidentiary basis that's just not there, or is there to a very small extent, particularly in the populations that we are concerned with in the Medicare population.

There is a whole host of evidentiary basis for the younger populations, but I'm not sure that I need to worry about ordering a PKU for my population I'm concerned with. We need an evidentiary basis if Medicare is going to cover genetic testing, for that particular population, and the benefits of that testing in that population.

So I strongly agree with Reed's comments and his written comments, that that is the key factor that needs to be encouraged, and that's the development of an evidentiary

base for these various technologies.

MS. BERRY: Muin?

DR. KHOURY: I think this is where the rubber hits the road. We have been talking about these issues now for about five to 10 years, dating back to Tony Holtzman's sort of NIH/DOE task force on genetic testing. I guess as a public health agency, and I'll present some of this more tomorrow morning, we have been looking for ways to supplement the oversight mechanisms that FDA and CLIA have in terms of assuring the quality of services and using evidentiary-based models like U.S. Preventive Services Task Force.

We have played around with different kind of model processes that I'll present tomorrow morning, but I think there may be another option for developing the sort of evidence-based approach as we move forward. Let me give you an example. We have been working very closely recently with the U.S. Preventive Services Task Force and AHRQ using the experiment of BRCA1 testing as an example, using one of their evidence-based centers, and looking at the evidence.

We have been meeting with various folks about this. It is very obvious that for most of genetics testing and technologies, what the U.S. Preventive Services Task Force would do is return insufficient evidence as their criteria. So this is a problem, because in the meantime, these tests are widely used. There are obviously direct-to-consumer campaigns about them, and so what we need to do is develop a process that can guide and help bodies like the U.S. Preventive Services Task Force, and other technology assessment processes, to do a first look at genetic technologies when they come out of the research oven, identify the gaps, and then use the processes of data collection that are a bit more targeted using public/private partnerships as models. Some of you may have heard me sing this tune now for five years --

DR. McCABE: Yes, we've been listening to this for 10 years, Muin.

DR. KHOURY: No, actually you're absolutely right. You've been listening to me for 10 years, so bear with me. I think there will be another process that I will present to you tomorrow, another experiment, a three-year experiment to see how we can do this.

But to me, that's the most important missing link between research and practice, is the sort of how do we implement an orderly process of transition that uses both the regulatory oversight mechanisms that already exist, along with processes of public and private partners coming together to weigh in on what is ready for prime time, and what is not ready for prime time. I think I'll just end here.

MS. BERRY: I have a suggestion. I don't know if this lends itself to it or not, but I sort of like the process we went through in terms of prioritizing our larger issues. I wanted to throw this out there for the group, because there are so many on this list here of possible topics for a recommendation, whether we should undertake a similar effort here where we determine what should be the focus of our efforts, and kind of prioritize amongst ourselves and see what that comes up.

It is really a daunting list. Now, others may say you know what? There is no reason why we can't look at each and every one of those issues and develop recommendations for each. But I suspect that that wouldn't be quite as helpful to the Secretary or anyone else if we were to cast that broad a net, and perhaps we should be a little bit more focused on some of the recommendations that have the greatest likelihood of producing an impact.

I throw that out for discussion, but I'm trying to figure out how do we move from kind of the theoretical, we kind of know what our goal is, but to get to the nitty gritty of where should we focus our deliberations.

DR. McCABE: I think that is probably a very good idea, because I think with this list, nobody can keep in mind a list this long, let alone the Secretary and his office, with all the things that he has to deal with. So I think that would be a very good idea.

MS. BERRY: Yes, Sherrie?

DR. HANS: I also think that is a terrific idea. I just wondered whether we wanted to sort of finish with any general comments first before moving onto that. I had just a couple of ones I wanted to get on the floor, and I don't know if there were others.

Just really quickly, and the reason I wanted to sort of make them publicly is just so people could think about them, particularly during the recommendations. I have more detailed written comments that I can give to the staff. One is to just pick up on an earlier theme.

I think that Toby Citrin in particular framed well, staff did a fabulous job of putting this document together and covering a very broad area. The one area where I would encourage them to work a little bit more on is making a stronger statement of need. In particular, framing out some of the cost issues.

Why is this something important to consider now? What kinds of dollars are we talking about? How quickly is this an emerging issue? If I'm the Secretary, why should I care about this today? I thought that there needed to be more of that in the front material in the report.

The other piece, which may come up in the specific recommendations, and I apologize that I wasn't at the previous meeting, and perhaps this was covered at that time. I was a little bit concerned about some of the statements about the sizable portion of costs going toward royalty fees. I wasn't sure whether there was actual documentation such as a survey that documented that as a statement, and I don't know whether that was simply through testimony. I just wanted to sort of put that marker in there that if it is supported by really good evidence, that should be included in the report and be considered for folks making recommendations. If it is, then it needs to be noted in more couched language, that it was brought up in testimony and through comment, but I'm not sure that we have a handle on exactly how big that problem is.

MS. BERRY: Did staff want to comment?

MS. GOODWIN: The evidence provided in the report was primarily given by Andrea Ferreira-Gonzalez at our last meeting. And actually Debra might be able to comment. Mildred Cho has published a few papers, and I think it goes into this topic a little bit. Is that right?

DR. LEONARD: Well, Mildred Cho, John Mertz, and I have worked mostly on the availability of testing, and the impact on gene patents. I don't know that the question has directly been addressed as to the cost. But it certainly has had a significant impact on availability of testing, because 25 to 50 percent of laboratory directors report that they have not done, or have had to stop doing testing, because of gene patent enforcement.

That is shutting down laboratories, but then when you can license patented technologies, the costs of those are exorbitant, relative to what we get paid for doing the testing. That is data that is hard to collect, because usually there is a confidentiality, or like a keep your mouth shut about what the conditions of the license are, so that most people who

have a license can't talk about how much it costs them, and what the royalty fees are, because part of the agreement is that you can't say.

So that would be information that is very hard to collect. But the up-front licensing fees are anywhere from $10,000 to over $100,000, and then a per test fee of anywhere from I think I've heard up to $60 per test, for a range of different genetic patents.

DR. HANS: And I would just encourage the staff to put in more nuance statements that capture some of what we just heard here, that isn't sort of a definitive statement of you can point to a particular study or collection of information.

DR. LEONARD: But hopefully part of this data could be collected through the NAS study that is being done on gene patent's impact on research and health care services. But I don't know if they're going to collect that information.

MS. BERRY: Debra, did you have anything additional beyond responding to that? I had written down that you had --

DR. LEONARD: Well, I was going to suggest that staff has come up with various policy options, and maybe one way to do this is to go through the policy options, and then go back and see if there are other areas where policy options have not been recommended where we may be able to address some of the issues. But that may be a way of working through the various issues that we have to address.

MS. BERRY: Joan?

DR. REEDE: I just had a question in terms of this issue around the royalties and reimbursement. Is there anything, or would it be useful to have something that required transparency in terms of that information for reimbursement to occur?

DR. LEONARD: It would be great if there was a CPT code that could capture that, but the problem is that then a reimbursement level would be set for that CPT code that probably would not reflect the actual cost of the royalties, because they would vary.

A number of discussions have looked at ways of having like mass bargaining for different gene patents so that it wasn't laboratory by laboratory, but there was more standard policy on how much is a reasonable amount to charge per test for royalty payments. I'm not aware of anything moving forward on that front, but it would be great if there could be transparency.

The problem is that all of the licenses that I have seen, and contracts, specify that you cannot discuss any of the terms of the license agreement once you sign. So I don't know how you would achieve that transparency. Maybe a lawyer in the group could say whether those two things are contradictory, I don't know, but it would seem to me that they are. But for health care coverage, it would be very nice to have those out in the open, so more people were aware of what they were paying for, and what the cost of gene patenting and enforcement is on health care costs.

MS. BERRY: Emily?

DR. WINN-DEEN: So I had a couple of things that I thought were important for us to talk about that aren't on the topics list. They were actually addressed in the body of the text, but somehow didn't make it onto the pullout list. One is time dilemma in terms of real preventive medicine where the cost of the test would be borne probably by younger people on private insurance, and many of the benefits would be reaped by older people on Medicare.

So you have different parts of the payer organization bearing the costs and

getting the benefits. So this deals with sort of common complex disease, so when you get into cardiovascular preventive medicine, diabetes preventive medicine, and those kind of diseases, we get away from monogenic kind of disease.

I think that is something that in the U.S. system, we are going to have to grapple with. The systems that have nationalized health care don't have to worry about that, because they very clearly see the benefit.

The other thing is we have evidence-based coverage decisions, but we haven't really talked about the health economics of that. So in terms of making a case for preventive medicine, do we need to just make a medical case? Do we need to make a health economic case? And are we only going to get coverage if we make a health economic case, and not just a medical case?

So I think that is all part of that whole preventive medicine shift from thinking about genetics just in terms of pediatric, neonatal screening, monogenic disease, and getting into sort of the next wave of the genetic component of most major diseases. And somehow we have to deal with that, or at least highlight it as an issue that needs to be dealt with in the future, if not immediately.

MS. BERRY: How about the threshold issue? It is a threshold issue to me, but maybe it is clear to everybody else. Do we need, or want, to be specific as to what types of technologies we're talking about?

We use these terms very loosely, genetic technologies, genetic tests and screenings, and I guess this gets to the evidence-based discussion. Are we at the point where we can establish guidelines or guidance, or help the Secretary come up with, and this gets to things that Reed was talking about and others, ways to assess whether something has value and should be incorporated in the whole health care diagnosis and treatment system? I don't know. Do we need to go down that path, or do we purposely leave it vague? We just keep talking in very general terms, but we all know from previous discussions that some tests are better than others, and some have validity, and some don't. Or has that bridge already been crossed, and we don't need to worry about that?

DR. TUCKSON: This is Reed. I would hope that we would be able to start, at least for our discussion of these issues, to be specific about examples. For example, if you take the DNA tool assay exams, those are very professionally checked. They have a meaning for what it will or will not be in terms of diagnostic colonoscopies going forward.

It has something to do with whether or not we will (inaudible) regular fecal occult blood. So that is a very concrete example of a kind of test. As we have alluded to, there are things that have to do with is your kid going to have blue eyes sort of thing, which is a different kind of category.

I think that it would be useful for us to have some specific categories of these things as we try to dig a little deeper into (inaudible) as finding examples that highlight and illustrate the real policy interventions that we're trying to concern ourselves with.

MS. BERRY: Debra?

DR. LEONARD: Well, in reading this, I went back to SACGT's definition of genetic test. In reading this, particularly in Policy Option 1, a lot of it is based on family history, and focuses on truly inheritable germline variations or mutations that cause disease risk.

The SACGT definition was much broader, and went beyond true germline

genetic tests, and included somatic mutations, and potentially even infectious disease applications. And so how those are treated is very different than those genetic tests and services that are related to inheritable germline variations and mutations that correlate with disease risk.

I don't know if we need to go back and revisit the SACGT definition of what genetic testing or genetic services were, but it still remains. That was one point that was very controversial at the time that SACGT developed that definition.

MS. GOODWIN: Can I actually draw your attention, Debra, to page 13 and 14, where we propose the definition for genetic technologies and genetic services to kind of frame the discussion in this report, and certainly taken from that SACGT definition, it is altered a bit to better reflect the different discussions. So rather than taking the definition from a more oversight perspective, we are trying to broaden it for the purposes of this topic.

DR. LEONARD: But then when you put such an emphasis on family history, somatic mutations are not included in family history, unless they are genetic cancer syndromes. And so I'm concerned about putting somatic mutations into the same category as germline mutations and variations, since you will not pick up somatic changes by family history.

DR. WINN-DEEN: Yes, I just want to concur with Debra, that I think we have to be really clear when we're talking about cancer genetics, about whether we're talking about cancer syndromes, or tumor profiling. If we're going to expand genetic testing to include tumor profiling, which has been suggested, and a lot of the --

DR. LEONARD: Well, it is part of the definition.

DR. WINN-DEEN: Right, right.

DR. LEONARD: The existing SACGT one.

DR. WINN-DEEN: Right. But we just need to be very clear about when we're talking about these things, which scenario we're talking about, either somatic genetics, or germline genetics.

MS. BERRY: Muin?

DR. KHOURY: Actually, if we take colorectal cancer as an example, you can have the whole spectrum or continuum of genetic applications, from the rare to the common. We need to be very careful, because at the end of the day, what we're trying to do here is figure out the value added of either a genetic test on the blood to determine germ mutations, or a genetic test on the stool to figure out whether there are DNA signs of the cancer, or a simple family history, too, that you administer and you find out that the person has a first-degree relative with early onset colorectal cancer.

We need to be able to use whatever SACGT has done, because they spent a long, long time figuring out the definition of genetic and genomic tests. And second, the process of validation of genetic tests. I mean, they use analytic validity, clinical validity, clinical utility, and the ELSI issues in a very detailed fashion, and they kind of laid it out like cold supper for us to implement.

So here we are talking about reimbursement of services, so you have to come back to what is it we are being reimbursed for? Is it a somatics test on this tool, or a genetic test combined with a family history, pedigree taken to find the people with HNPCC, or the APC syndromes for colorectal cancer, or somebody who has just a first-degree relative with colorectal cancer at the age of 65, and therefore, may need a

colonoscopy earlier than the general recommendation, which is after age 50.

So the bottom line in all of this is what is the value added? I think the health economics discussion is very apropos here, because in a time of limited resources where you are comparing options A and B, you have to kind of compare a genetic-based practice of medicine with a general practice of medicine that doesn't use genetics, and do a decision analysis, like they did with the TMP example in 6MP with treatment of acute leukemia that David Veenstra represented to you last time. So what are the pros and cons? And then at the end of the day, how much money are we going to spend if we use a family history/genetic test tool on some people, and a DNA-based assay on other people?

So the main recommendation here from me is take this high level document, and then begin to apply it to very specific case studies, perhaps guided by what SACGT has done for us over the last three years, because they lay out very clearly the types of genetic tests for diagnosis, prevention, prediction, and then the process of validation on how to do this.

DR. TUCKSON: Could I ask Muin to take it one step further? I wonder that after we've got all this analysis, what do you see as the level of detail coming out of our committee after we have done that? I think that is what I was trying to get at.

The question is, obviously we're not going to see ourselves then recommending a specific test for the Secretary to get access to, but this is leading to some sort of an algorithm for the next step of policy or process development in the Secretary's office. What do you see?

DR. KHOURY: I think, Reed, you should be able to determine this as a committee, because I don't see you getting into the real specific test by test discussion, but more of a high level guiding principle for HHS on how to go about doing this. You could be guided by the case examples as you go through the deliberations, but I see kind of an intermediate menu or cookbook in a way, but not very detailed, because that has to be worked out by the agencies themselves, CMS, and what they cover for.

But moreover, general guidelines, perhaps of the kinds that SACGT started doing during the tenure, and then leave it for implementation. But you can monitor the progress of HHS agencies.

DR. McCABE: I wanted to go back to the point of definition, and bring people back to page 13, where I think the implication, with a sidebar from staff, is that it was their intent that these be germline mutations here. It certainly says somatic genotypes, but if you look at all the examples, they are all germline.

DR. LEONARD: So can that somatic be taken out?

DR. McCABE: That's the purpose of this discussion, would be to get the feeling of the committee. We need to move forward on this, as Muin says. We have been debating this issue of definition also for 10 years.

If you look at the examples, they are all clearly germline.

DR. LEONARD: In a way, this is kind of like the genetic exceptionalism issue. Sometimes it is good to lump it in, and sometimes it is good to exclude it. It is the same sort of thing with somatic mutations. When you are talking about genomics, medical advances, and improving health care, it is probably good to include the somatic mutations.

But when you are talking about the kind of regulatory issues of appropriate counseling, appropriate ordering of the tests, and getting the testing through family history, those only apply to germline mutations. So I guess there are times when you would want the

definition to include all types of genomics beyond just inheritable, and there are other times when you really need to distinguish between somatic and germline, because the implications are very different.

Somatic mutations have no implications for the rest of the family members, whereas germline mutations, you are dealing with an entire family.

DR. McCABE: And so that if we took it out of the definition in the sidebar here, since in fact all of the rest of that sidebar relates to inherited, then you could put some sort of a thing at the end. The title here is "What are Genetic Technologies?" as opposed to genomic technologies. Either in the text or somewhere else, you could put that similar approaches may be used for looking for somatic mutations using genomic technologies.

DR. LEONARD: And even infectious disease, because part of our charge is bioterrorism, although we don't look at that that much, but infectious disease and emerging infectious organisms, that type of thing also benefits from genomics, but they don't have the same familial component.

DR. McCABE: I guess it is a judgment that you all have to make, what to include or not include. I'm just reacting to what Ed said about similar approaches can be developed for genomic tests.

I think what comes to my mind immediately is that those approaches are not similar. I mean, what applies to germ cell is different from what applies to somatic cells. I think we are going to be faced increasingly with the mixing and matching of both technologies. So if you have a first-degree relative with colorectal cancer, you may use a somatic cell to find a somatic diagnostic test on the stool to find early evidence of cancer, rather than colonoscopies on the proband.

So we are going to be mixing and matching somatic and germ cell mutations, so that's number one. Number two, even among germ cell allelic variation, we do have to clearly separate the high penetrance rare alleles, the PKU, the Huntingtons of the world type diseases from the day to day SNP variation that is associated with low disease risks and complex gene/gene and gene/environment interaction. Because those kinds of genes do not lead to significant familial aggregation, and their use as a technology is likely to be bundled up together in genomic profiles that have very different kind of a more sturdy, I guess, validation process, if you will, than the simple single-gene genetic disorders.

So as we go through this process for reimbursement and coverage of different tests, I guess you start by asking the high level questions. Are the tests analytically valid? Do they predict any clinical outcomes? More importantly, can they change the outcome in reducing morbidity and mortality? And what are the ethical implications?

And then as you go through this armed with the processes of different groups like the U.S. Preventive Services Task Force, each one of these types of tests may have a different threshold for crossing between what is ready for prime time, and what is not ready for prime time. We spent quite a bit of discussion in SACGT about rare diseases, because those rare diseases, and I guess this committee has taken it on too, may have a different kind of a threshold, because of the difficulty in collecting data because diseases are rare, and the validation process therefore may be either delayed, or non-existent to begin with.

So we have the whole gamut here of tests from somatic to germ cell, and within the germ cell you have the highly penetrant single-gene diseases to a bunch of SNPs, polymorphisms, and gene expression profiles that you use in future bundling, either for

pharmacogenomics, or just genomic profiles of the kinds that some companies are sending us right now prematurely for preventive medicine.

MS. BERRY: Ed, and then Agnes?

DR. McCABE: My point was purely practical. Having heard these discussions for the past decade and been a party to them for the previous three years, recognizing that the division between somatic and germline can get a little bit fuzzy where sometimes somatic is superimposed upon germline, as opposed to as far as we understand, purely somatic at this time, though there may be some predisposition. It was just a way of moving on, getting beyond that, and at least getting somewhere by limiting the definition to germline, and taking it out of the debate that has to do with these definitions that are used in the clinical laboratories.

So that was my purpose. I think we could say let's focus on germline for this document, let's recognize that the lessons we learned here may be applicable to somatic, but let's at least move forward at this point. That was the purpose of my comments.

MS. MASNY: I was thinking more along what you were saying, Dr. McCabe, that if you divide it that way, then I think we may be defeating the purpose of what we were trying to even do earlier looking at the education of the general health force. If you are only looking then at the germline mutations, then the focus on the genomics and where most of the actual practice in genomics is going to be done in the primary care, if we don't focus on that and address some of the reimbursement issues, we know that reimbursement very often drives practice, as well as what people implement.

So I think that we should continue, if we do that, we're looking specifically at the genetic technologies, and focus on the germline mutations. I think maybe if we had not just at the end of the document, but somewhere right after this, another sidebar looking at what are genomic technologies, so that they sort of are dealt hand in hand the way we did in our previous documents where we were putting the slash between genetics and then genomics.

I think it has to be addressed. I do think even from the level of somatic mutations, it will involve family history. Because, for example, in the ovarian cancer testing that they are proposing, the use of proteomics, they are already talking about looking at guidelines for who would be best to use this test based on family history. So although the proteomic may be picking up as certain somatic patterns or tumor profiles of those protein patterns in an ovarian cancer, they still would be using family history to determine who would make use of that test.

DR. McCABE: Yes, I was just talking to Suzanne, and certainly we could add a sidebar that would deal with the somatic and other genomic technologies, even getting into some of the issues about genomic technologies applied to microbiological organisms. I understand that a lot of what we do are really the applicability to the general population, that are going to go beyond this. I think that it is important that we move forward after a decade, and not get hung up again here.

MS. BERRY: Debra?

DR. LEONARD: Well, in the context of coverage and reimbursement, the genetic counseling only applies really to the germline mutations. When you talk about the testing services, laboratory tests are all coded with the same CPT codes, regardless of whether you are talking about germline, looking for germline mutations, or looking for somatic mutations.

So in the context of coverage and reimbursement, it is all the same codes anyway, regardless of whether you're doing somatic or germline, or whatever, and they are all inadequate.

DR. McCABE: So by including them as sidebars within here, but separating them out, perhaps we could try to make those distinctions, and not have them continue to be blurred.

MS. BERRY: It seems to me that that is really a threshold question. What are we talking about when we're talking about coverage and reimbursements? Coverage and reimbursement of what? And then we seem to go and we take a leap, and we all do in our discussions, the report, and public comments. Genetic technologies need to be covered and reimbursed.

We have to back up a little bit, and this gets to Reed's point and Muin's point, and others, we are not really saying that. We are not saying that all technologies need to be covered. So perhaps there is something that we need to do in the way of recommendations pertaining to what scientific evidence is necessary, where are the gaps that would help decisionmakers, be they federal government programs, be they private insurers, make the right decision in terms of coverage, and subsequently, reimbursement.

DR. WINN-DEEN: So I think one of the things that is important to think about when you come to predisposition or risk assessment testing, is in much the way that family history has been used in the past, this testing is quite likely to be used as some kind of a gatekeeper for more intensive monitoring.

So either you start earlier, you do it more frequently, and to Agnes' point, you identify people who would benefit from expensive testing, or you never would recommend it for general population screening, but for a subset of people who are "at risk."

And so I think we need to think about also the trickle down effect of a risk assessment test as a gatekeeper for other more traditional monitoring tests, you know, BRCA1-positive individuals start mammograms earlier, or start mammograms a whole lot earlier than general population screening.

There are good monitoring things. Not only do we need to worry about whether the primary gatekeeper test is covered, but also once people are put into a risk category based on that gatekeeper test, is their monitoring also covered? Because that then would be they are not symptomatic, so it is still presymptomatic testing, but it is absolutely medically indicated, or we all believe it is, anyway. So I think that is another wrinkle that we somehow have to consider at sort of the risk assessment test as a gatekeeper for more standard monitoring analysis.

DR. LEONARD: And not only the more expensive monitoring, but also the follow up things that may be done, like prophylactic mastectomies. Will they be covered? The real sort of treatment choices that people who are identified then as being at high risk may choose to have done.

DR. WINN-DEEN: Yes. So the whole preventive medicine strategy, whether it is a colostomy, a mastectomy, an oophorectomy, whatever.

DR. TUCKSON: This is Reed. I tried to write a lot of my comments down, and I think you all have copies of it, so I won't waste your time rehearsing any of that. But what I think I like about what just got stated is it is that level of detail that I think we need to, and I think this may be what Muin was getting at a little earlier, is that as we start to work some

of those out, we then by delving in that kind of depth of the downstream consequences of some of these things, that we start then to think about what our policy recommendations might need to look like.

What I'm sort of getting at here is that things have consequences. One of the things I hope is that we don't wind up putting ourselves in a position wherein advocating for a good thing, that it means that automatically a new test, because it is available and exciting and good, becomes the subject of something that is automatically reimbursable, when it may result in five other things needing to occur that are associated with it.

Maybe those things are appropriate, maybe not. But then that one new thing causes maybe four other diagnostic restudies to confirm or whatever, and then the economics of that get to the point where it becomes crazy. At some point in this, I just want to make sure that we don't position ourselves as being irresponsible to the notion that things occur in context to other things, and how do you think about that in a world of real constraints is, I think, something that we could add. How do you approach those kinds of issues, or do you simply say let whatever happens, happen, and that is not our responsibility to think about.

MS. BERRY: Ed?

DR. McCABE: You know, I think it would be worthwhile to look at the recommendations now to give staff some assistance with how to couch those. And Cindy before said maybe we should prioritize those. I went through and began to categorize them, but I should have looked over my shoulder and seen that staff had done a much better job than I did on the fly here.

So that if you look at the PowerPoint slide there, it really falls into four categories. Even if we take the broader issues and separate those out, we're down to five categories. So that it is a way to organize by Medicare, and what the issues are there for Medicaid/SCHIP, then what the issues are for all insurers, and then the broader issue.

So what we might do is look at how we could prioritize within each of those categories perhaps if we feel the need to do that, but at least it is an organizing principle that allows us to go from whatever the number was before, down to four or five categories. So perhaps there could be some discussion within each of these categories now.

MS. BERRY: Emily?

DR. WINN-DEEN: So can I just ask sort of a point of order? Because if I had to look at those and say, should we talk about all insurers, or just Medicare, because all insurers encompasses Medicare and includes a whole lot more people, I would say, do that. But in terms of what HHS is directly responsible for, it is only Medicare.

So could you just give us a little committee guidance? Should we focus on issues that HHS can control, rather than the whole global insurance industry? Or how do we do that?

DR. McCABE: I certainly think we could comment on all, but in terms of what the Secretary has control over, I would think it would be those things that the Secretary has control over, so that we could perhaps have some guidance from the ex officios with respect to what they think, where they think this committee could have the greatest impact. It is Medicare and Medicaid, and also then the other agencies sitting around this table. So could we have some help from the ex officios, perhaps, with respect to where you see the biggest bang for this committee's efforts?

MS. BERRY: And if I can just sort of add to that. Are there studies, either

going on right now, or possibly being contemplated or studied, that perhaps we could suggest that the Secretary commission either through one of the agencies, or from the outside, I don't know the most appropriate way to do it, that would get to this issue of evidence-based decisionmaking that could be while not directly influencing private health plans, certainly if there were a government-sponsored study, that could influence private health plans indirectly, in addition to informing Medicare program. I don't know the answer to that.

DR. KHOURY: Is Reed still here?

DR. TUCKSON: Yes, I'm here.

DR. KHOURY: I guess this committee can make an influence in a number of areas. Obviously HHS direct jurisdiction is Medicare/Medicaid. But HHS presides over NIH, CDC, FDA, and AHRQ. As you said, Reed, earlier, that recommendations from the U.S. Preventive Services Task Force count a lot with respect to the private world, isn't that correct?

DR. TUCKSON: Yes, very much.

DR. KHOURY: So basically what you are recommending to the Secretary is not only something that would influence Medicare and Medicaid, but sort of this convening role that hopefully by doing other things, like evidence-based coverage decision, will influence the whole practice of health, health care, and preventive medicine in this country for the private sector as well.

So I don't feel like you need to be constrained necessarily by this hierarchy of Medicare/Medicaid, nor do I think that a recommendation should be different for different groups, because what you want to do is lay down the basic principles and guidelines for what should be covered and not covered, and reimbursed or not reimbursed, and then it will play out through the various processes of the public and private sector.

I mean, you are tackling here issues that are relevant to the practice of medicine and health in this country that are way beyond genetics. I don't think we're trying to fix medicine here through the lens of genetics, but to try to see how those new technologies fit in the underlying scheme of the practice of health care in this country. But that is my opinion.

MS. BERRY: Yes, Kaytura?

DR. FELIX-AARON: I agree with you. I mean, I am struggling, because I'm not sure that I understood the question that you posed, but I just wanted to respond to Muin's point. The U.S. Preventive Services Task Force addresses really basic questions. Is there enough evidence to recommend a particular service? You get a number of letters.

The challenge here is that the evidence, and I go back to Steve's point about the evidence base for making those types of evaluations is that chances are that evidence base is really often non-existent, and so the recommendation that comes out is I, insufficient evidence. Not that it is discouraging those services, but that there is not sufficient evidence.

So I think that the U.S. Preventive Services Task Force does a lot of good work, but I haven't heard around the table sort of questions that we have that would be appropriate for that particular body. However, AHRQ also has the evidence-based practice centers, which addresses more policy, sort of like higher level questions. If this committee had a particular question that it wanted to address, our EPC programs could address a particular question.

The EPC program is such that it not only relies on the published literature, but is increasingly looking at the gray literature, literature in reports, and increasingly pooling reports and experiences, sort of practical experiences to draw conclusions, and to provide

guidance.

So provided that this group came up with a clear question and wanted guidance on how it ought to tackle a particular question that had policy relevance as well as clinical relevance, the EPC program and AHRQ could address that.

DR. McCABE: I've asked Cindy's permission to come to the mike at the podium, just to save my neck, trying to crane 180 degrees. I think if you look here, let's look at the recommendations, the possible topics for recommendations.

As I look through the all insurers category, I don't really see any topics there that don't apply to Medicare and Medicaid, and I see some that actually collapse into some of the other topics. So that if we look, for example, for coverage decisions, under that, there are issues like the screening exclusion under coverage decisions. Also, if we go under all insurers, we see evidence-based coverage decision, we see a need for a, rather than a lack of uniformity, we could say a need for uniformity to make it more positive in coverage decisionmaking. Also reimbursement determinations come to some extent, under that.

Basically all of these issues apply to Medicare and Medicaid, as well as to all insurers. I mean, that's by definition, Medicare and Medicaid are all insurers. The issue is is there anything there that we should exclude as being too numerous in terms of the menu to the Secretary? Or should we look at collapsing them into fewer categories as a way of organizing this.

MS. BERRY: Steve?

DR. PHURROUGH: Well, I'll try and give you some concrete recommendations. I think the issues as you were, I think, trying to elaborate, Ed, are not around the kind of payer. The issues are around what is the evidence base, what are the barriers to applying the technology using that evidence base, and then perhaps there are some specific Medicare or alt payer issues.

So I would say that the first priority is developing an evidence base, and it is a recommendation that the Secretary task his appropriate agencies, which I think are AHRQ, NIH, CDC, and perhaps even HRSA, with systems research to commission a technology assessment that defines the current genetic technologies, the evidence base for those, and identifies the gaps and knowledge it needs further trials to determine. I think that's number one.

I think number two is define the Secretary's task CMS, and in fact we've already made our recommendation that the Secretary provide in its recommendations to Congress, whatever that is, that the screening evaluations become a routine part of the Medicare portfolio, and that CMS be allowed to make those decisions based upon the best data that is at hand.

Three are other barriers, such as CPT codes and reimbursement determinations, and we could ask the Secretary to task the appropriate agencies to define, well, I'm running out of concrete ideas. I think rather than saying what should Medicare do, and what should all insurers do, I think there are things across the realm, and I think the first is the evidence base, and I think the other is incorporating screening into the entire insurer population, and not just private.

DR. McCABE: But using evidence base in the grandeur, not U.S. Preventive Services Task Force's slow, methodical, perhaps too slow and too methodical sometimes approach, but look at where utilizing best practices and literature could define an evidence base for this, the way we use it more typically in medical practice.

DR. PHURROUGH: Exactly.
DR. TUCKSON: I've got to sign off. Can I just make one quick comment?
DR. McCABE: Of course.
DR. TUCKSON: Thanks, Ed.
I like what we just heard from AHRQ, I think it sounds good, and I think we ought to fall for those kinds of larger institute studies that AHRQ can do outside of the U.S. Preventive Services Task Force.
The only other comment that I would want to emphasize though, is that there ought to be very clear criteria for when recommendations about going forward for extensive things ought to be done when there is no evidence basis. I can clearly understand there are times when you have to go forward, but I think we've got to be very clear about what may precipitate that recommendation.
Secondly, in my analysis, I did call for a little attention on looking at the actual quality of performances as based upon professional guidelines for the delivery in this area. The guidelines need the big step from the evidence, and then evaluation of what you actually paid for from the taxpayer's point of view, and private purchasers based on those guidelines. That is something that we didn't get into, and I just want to put it on the list for consideration maybe sometime going forward. Thank you very much.
DR. McCABE: Thank you for being with us this afternoon, Reed.
DR. TUCKSON: Thanks.
MS. BERRY: Emily?
DR. WINN-DEEN: I guess I just want to second that comment that maybe the broad categories are one, coverage. What is needed to establish that a test should be covered? This goes back to SACGT's long discussion about clinical utility, when is clinical utility established, and then what are the other things that follow from that?
Then the second thing is once you have determined that something should be covered, how do you assure adequate reimbursement, and that gets into this whole is the CPT code system working, is the reimbursement associated with method codes the right way to do it? That's another whole set of discussion items.
Then I like the sort of final everything else broader issues category of things that if coverage was there and reimbursement was there, what are the other things that we would need to be concerned about in bringing this to the practice of medicine? So what are the remaining barriers to entry? Because I think a lot of people think that they are all coverage and reimbursement, and I think we clearly in our morning session on education and training, identified that if a doc doesn't know how to use something, or a system doesn't know how to provide the right context for it and make people aware that they even should be tested, that it won't be utilized either.
I think if we divide things into those set of bins, it would help our discussion, and help make very focused recommendations. How do we improve coverage? How do we improve reimbursement? What are the things within HHS that can be activated in each of those areas? It just might simplify our job.
DR. McCABE: May I just ask one very specific question? So would people agree that one of the things that the Secretary might do would be to request input by whatever mechanism, specific mechanisms, have been recommended, but request input on how to improve those coverage decisions? Because that is the kind of thing that we can

recommend, that a study be chartered, or whatever.

DR. WINN-DEEN: Right, and I think just one little sidebar, there is an NCCLS group working on setting a guidance document on determining clinical utility. For example, I think it would be worthwhile for us to look at that draft document at the point when NCCLS is ready to share it, to see if that kind of a consensus process, which is what NCCLS goes through, would also serve us, separate from some government processes that might also be in the works to set out what is evidence-based medicine, and how do we know when we've gotten there?

MS. BERRY: Kay?

DR. FELIX-AARON: I wanted to draw the committee's attention to the fact that groups in the private sector, our federal partners, often request that the agency conduct studies. For example, the AMA could come to us and say, I want you to conduct a study on health literacy, and we do those studies.

So I wanted to raise again, it could go through the Secretary, and Sarah, I think you would have to guide us in terms of what types of interactions this committee can have with the agency. But in addition to going through the Secretary, it may be possible for this committee to actually request the agency conduct such a study, and that this committee may be able to go directly to the agency and work with the agency to create, implement, and conduct a study that addresses the specific questions we have laid out this afternoon.

MS. BERRY: Debra?

DR. LEONARD: I guess I have kind of a generic question. We talk about developing an evidence base, but really that is done one disease, one test at a time. I don't see how you develop a process to do that, because those who would need to be at the table would differ, depending upon what disease or what test you're talking about as to whether there is clinical utility for doing that. So can you create a generic process that would work for everything, for making coverage decisions?

DR. McCABE: After the experience with the SACGT, I would argue you probably could not make a generic decision that would cover everything, but that should not prevent us from getting started somewhere. So you can focus on what it is that you can begin to make decisions about, and identify a process for that, at least.

DR. PHURROUGH: Well, there are fairly standard processes that most payers go through in making coverage decisions. They vary somewhat in process, but the concepts are fairly similar. What we would like to see, those of us who are making these decisions, are to begin with, is sort of what is the state of the science now?

The state of the science isn't gathering experts together. The state of the science is let's pool through our normal processes, every piece of literature that has been published, and determine based upon that, what the state of the art is today. And then there will be gaps, and you'll list all the tests that are there, and you'll see where the gaps are. You'll list particular tests that have been proposed, perhaps they may have utility, and there will be complete gaps, because there is no published literature there. But evidence-based medicine by most carriers, by most payers, is an expert opinion. It is what has been published in the literature, based upon clinical trials.

DR. LEONARD: Can I follow up? So then the question is if you go through this process and identify the gaps, it seems that there are tests out there that are being used, but they are not covered, and there may be clinical utility, but it is not in the published

literature. Could then a request be made to NIH to put out an RFA for this series of diseases or tests for there to be funding to develop the evidence base, so that there could be coverage?

DR. PHURROUGH: I think the Secretary would be very hesitant to make a recommendation like that until we did have that evidence base.

DR. LEONARD: No, this is funding to develop, to see whether it should or shouldn't be covered. That requires research, and generally research that is not very well funded at this point.

DR. PHURROUGH: Correct. He would first want to know what is the state of evidence today, and then look forward, yes.

DR. LEONARD: Exactly. So there would be a body that would say okay, we've looked at these various tests and diseases that are out there being used, and for these three, we find plenty of evidence, and we're going to cover them. For these ten, we don't find enough evidence. Could that then be reflected in an NIH-type/NHGRI-type of RFA to ask for studies to address this?

But then you would also have to have guidelines out there about what kind of studies, what kind of scientific evidence you would need, how the studies should be done prospective, whatever, in order to provide appropriate evidence, but those types of studies are not well funded.

DR. PHURROUGH: That's correct.

DR. LEONARD: And so you never get there.

DR. PHURROUGH: A good technology assessment will tell you what trials need to be done, and be fairly specific, these particular tasks need these kinds of trials. And then I would assume this committee would make the recommendation to the Secretary that he task his appropriate agencies to look at those particular trials. It may not be NIH, it might well be AHRQ, and in fact, it could be CMS who may in some instances, provide reimbursement for technologies that aren't proven in the context of trials. We do pay for clinical trials. I'm not sure health systems research would fall into that category, but the Secretary can task his agencies to do whatever.

But I don't think he would do that until we first had what is the state of the art today, and what are the gaps, and then let me go out and fill those gaps. I think we need to study technology assessment first.

DR. McCABE: And I think what Debra is arguing is that there are quite a few gaps, if we have really started to look there. Certainly it fits with what CDC is doing, and what many of the agencies around here are doing, because it is recognized that there are huge gaps here. It clearly fits into the roadmap. You can't do translation if there is nothing to translate from, or to, which are some of these gaps that you would have.

I just wanted to go back over this organization, and be sure, because what I want to leave here this afternoon with is some guidance to staff about how to move from the document we have. We've had a lot of written input and a lot of oral input this afternoon. But the key is the recommendations, and we need to organize those.

So Steve, you had given us sort of an overview of an organization. Emily has, I think, taken that another step, and I want to be sure that everybody is in agreement about these categories, and then I want to talk about the broader issues and be sure that everybody agrees that they even need to be there.

So the coverage issues. The coverage issues, that would come under your

first heading, would it not, Steve? Because part of the coverage issues are that evidence base. How to assure adequate reimbursement? I don't remember what your second issue was, but would that be acceptable as a major category?

DR. PHURROUGH: I'm not real thrilled with a lot of discussion around reimbursement, but then that's because we argue reimbursement very commonly in CMS. But I think there are issues around what prevents coverage, and reimbursement is one of those. It may have a better title than reimbursement that I'm not thinking of at the moment, but reimbursement is an issue around why things aren't covered, and evidence is one of them. CPT codes is another.

DR. McCABE: I think for the practitioner community, one of the things we've heard over and over is that reimbursement is a huge issue, because people aren't doing these things if they're not getting reimbursed for them.

DR. PHURROUGH: We hear that for every single thing that we pay for. No one is yet to say we pay too much for something, we have yet to hear that complaint.

DR. McCABE: The real problem in genetics is, as Debra pointed out before, all genetic testing is covered under one code.

DR. LEONARD: Fourteen codes, to be exact.

DR. McCABE: Well, a very limited number of codes end up having to be creatively bundled and rebundled and everything to get anything to move forward.

What was your second category, Steve?

DR. PHURROUGH: My second category were Medicare-specific issues that all insurers don't have, and that's the screening exclusion issue. That's a specific Medicare barrier that others don't have.

DR. LEONARD: However, other insurers I have heard, speak and say that they don't cover preventive strategies. And so there are other insurers that do that. Not all, but some.

DR. PHURROUGH: By choice, not by law.

DR. LEONARD: Right.

DR. PHURROUGH: That's the difficulty we have, is the specific law that permits that.

DR. McCABE: Well, I need some guidance from the committee, then, so that we can leave here and have some instructions to staff.

DR. LEONARD: Can I comment? I don't care whether CMS likes the reimbursement issue or not. I think that this is a major issue, that the reimbursement is so inadequate, it doesn't even cover the cost. I'm not talking about the charge, I'm talking about the cost of doing the testing, so that it is a barrier to wide availability of the testing competition for pricing, and all sorts of other things.

If we are moving to genetic/genomic medicine in the future, that is going to be based on genetic/genomic testing that is not going to be paid for adequately to cover the costs. So I don't know, I mean, it is a very complex system of how reimbursement levels for CPT codes is set. I don't understand it completely, but it is not simple.

It is also constrained by the new Medicare Act that says that the laboratory fee schedule is frozen until 2009, and there are a lot of constraints on even addressing the reimbursement issues. But I'm not sure that we're having a lot of recommendations or discussions here about how to change the reimbursement for genetic testing and genetic

services. Not only the testing, but the genetic counseling, and other types of services which are also inadequate.

DR. McCABE: So Debra has taken a strong position in support of reimbursement. Is there any member of the committee that would wish us to take reimbursement off of the table?

MS. BERRY: I think it needs to be on the table, but it is sort of a linear thing, because it doesn't become an issue if there is no coverage. I mean, they aren't going to pay for something that's not covered. So the coverage part is the first threshold question, so reimbursement is by itself, it covers all of the insurers, Medicare, Medicaid, private sector, and it is critical. It is something that we are tasked with looking at, because it is a significant barrier to access. So we need to sort of think of them, and I don't know if it is linear, horizontal or vertical.

DR. LEONARD: Well, the problem is that there are CPT codes out there, and they are generic codes, so you can use those codes for any kind of testing, covered or not covered. It is not clear to me how insurance companies know what they are paying for, whether to cover it or not, since the CPT codes are generic.

That is going to be remedied somewhat by the CPT code modifier system that is supposedly coming this fall. But it is not clear until that happens and laboratories start using those modifier codes as to how much that will decrease denials because of coverage decisions. They will now know what they are covering or not covering and can say yes or no, because instead of just saying no because they don't have a clue what the test is, and why it was done.

But that still doesn't address once they decide to cover it, that it is a covered test, that what you get paid is inadequate for what it costs you to do it. So you're right, it is a linear process, but I don't think that we cannot go to the next step of at least figuring out what to do about the inadequate reimbursement, supposing that something is covered, and address those.

You don't really have to address the coverage issues before you look at the reimbursement issues, because it either is or isn't covered. The reimbursement is inadequate, even if it is covered. So the reimbursement is a separate issue, and you don't necessarily have to decide the coverage before you look at the reimbursement concerns.

MS. BERRY: Agnes?

MS. MASNY: The only place we would need to look at coverage then would be just the screening issue, because that is where if Medicare doesn't cover screening, or see screening tests as covered, and I think that was your point earlier about looking to the commission and agency to define, or commission HHS to allow CMS to incorporate screening, so that would be a coverage issue.

DR HANS: It is actually in my written comments, but I would just remind the committee that HHS actually does have a health delivery system that it is also responsible for, and that is through the Indian Health Service. So you actually have within the purview of this committee, a delivery system.

While I don't have any specific recommendations, because I'm not quite sure where this is going, I would just sort of put a mental placeholder in that as you move toward more concrete recommendations, that you then think, does there need to be a footnote for IHS? Just to think through what are the implications for the health delivery system that controls

pretty much all the elements that you're talking about here directly.

I'm not sure that there would be any special circumstances, but I would just want to put that on the table for folks to remember. Of course it is rather self-serving, because if there are things for IHS, there is likely to be also for VA, and we would therefore find that recommendation is more helpful for us.

DR. McCABE: The cultural sensitivity issue is certainly one for IHS and some of the Indian nations, and genetics does not fit into their belief system, and so they really have decided as a nation, not to go down this road.

MS. BERRY: Debra?

DR. LEONARD: One of the areas of coverage that may be able to be more specifically addressed, which Muin had brought up, is when the use of a genetic counselor is appropriate. We talk about moving to more generic services being delivered as part of all health care professional services, but maybe if criteria for coverage of genetic counseling services could be developed, then it would be clear when those could be covered, and when not necessarily covered. I don't know if those already exist, maybe someone can comment.

That would be a separate issue from reimbursement for those services. But I think defining when as we move toward the 90 fevers that are dealt with by generalists, and the ten fevers that are dealt with by specialists, maybe we need to distinguish when you would pay for the special genetic counseling services.

DR. McCABE: I would like to move us on, then to the two topics under broad issues, and discuss inclusion or exclusion of those, and if they are to be included, then the rationale for including them under coverage and reimbursement of genetic technologies and services.

Neither of them are dealt with in the policy implications, and that is why I think it is important that we assist staff with this at this point in time. Health disparities, the way I see that included, is that if there are populations that are not covered, and/or not reimbursed, then that will lead to a health disparity. But could we have some additional help from the committee for staff in terms of the rationale for that one being included in a coverage and reimbursement manual?

MS. BERRY: Just to play devil's advocate, I actually regard both of those issues as being so important as needing to stand on their own. I don't see them in the context of coverage and reimbursement, but I see them as broader than that. They could lend themselves to their own reports, each one. So I'm not clear as to how we would fit them into a coverage and reimbursement report, or if we should.

DR. HANS: Let me ask a question and see if you think that it is an issue. If you set down some criteria for making coverage decisions based on some evidence, what if that evidence does not adequately cover certain ethnic populations? What then coverage recommendations do you make for those populations? And is that a health disparities issue there?

DR. LEONARD: I think what you're getting at is the broader issue of the science disparities by ethnic group that will influence the evidence base that will be use to make coverage decisions, that will then influence the health disparities. So you have to go all the way back to the science.

DR. HANS: Absolutely, that's the train. But if you are making a policy decision, and for Caucasians, the test has been validated, what do you then do as a policy

entity? Do you say you cover it for anybody, but the evidence doesn't exist for the other ethnic populations?

I don't know how you begin to help policymakers figure out how to make those decisions. Or do you wait until you have evidence that covers all ethnic populations before you make a coverage decision?

DR. McCABE: How about provider education and training, then? I think the argument has been made that we do need the evidence base for populations if we are to have recommendations. What about provider education and training?

Again, and I understand Cindy's point that these are huge topics. But if we focus them, I think we can focus the topic and take one little piece of it for coverage and reimbursement, not try and deal with all of health disparities, but just as they apply to this topic. Would that be then more reasonable, Cindy?

MS. BERRY: Then how does provider education relate to coverage, for example? We know all the issues about the inadequacy of the education for our providers and the gaps and public awareness and all of that, but how does that relate to coverage? Emily?

DR. WINN-DEEN: I want to make a comment on health disparities, and then I'll answer your question. I think the other thing that we have to do, besides setting up evidence-based studies that take into account at least the three major branches of the human tree, and make some kind of statement about whether a test is or isn't appropriate for each of those branches, let alone all the twigs and leaves that are off of those branches.

We also have to recognize the economic disparities that exist, and whether there is a group of people who are willing to self-pay, and thus gain access to things earlier, while coverage decisions and evidence-based medicine are churning along, and whether that creates some additional health disparities or access issues, however you want to put it.

I think there is a sort of genetic group disparity where we have to deal with that, and we set up evidence studies. But there is also the economic disparities that people who have the means will pay for almost anything. To what extent do we feel as a policy group we need to address that? And at what point are people spending money on things for which there isn't evidence? Is that just their own buyer beware, people pay for a lot of snake oil, and only a little bit of it works?

So that's one thing. The other, in terms of provider education and training, I think that again, comes down to potentially an issue of disparities, where if you are seeing somebody at a highly acclaimed university medical center, you might get a different level of provider education than if you are seeing someone in a rural outpatient clinic. It just depends on where you live, and the kind of training that the people who you are seeing may have received.

So again, you have the issue of getting equal access, equal treatment availability to everyone, without having equal provider ability to deliver that.

MS. BERRY: My point is that I agree with everything you're saying, and I think of these issues as being at the core of the overall access umbrella that we've been struggling with, and I think they do merit further attention, and maybe their own report. But I'm still not clear on how they directly relate to coverage and reimbursement.

Not that I think that they aren't important, but I want to focus the report on those two issues, since that is what it is supposed to do, and then address these others, which we have to get our arms around in a broader context, because it cuts beyond those two issues of

coverage and reimbursement, I think.

Debra, and then Martin.

DR. LEONARD: Well, the health disparities, I think directly is related to coverage and reimbursements since they are different systems, and they are uninsured and underinsured. If Medicaid is decided state by state, it may be harder to influence that coverage, and so that may be less adequate coverage than something that can be influenced nationally.

So the way coverage and reimbursement is done does create health disparities, and maybe we should look at those issues. The provider education and training, aren't we doing that in the other resolution? I don't really see how that affects coverage and reimbursement, since coverage and reimbursement is decided on an evidence base that is not related to whether physicians know how to use tests appropriately.

So in my mind, that broader issue is really more addressed in the other resolution that we discussed this morning, and not so much a piece of the coverage and reimbursement.

MS. BERRY: Martin?

MR. DANNENFELSER: Yes. It would seem right now that if people have the economic means to pay for things out-of-pocket, they are going to pay for them. So the idea that we wouldn't provide coverage because somebody else might get it, I don't see the issue there. I think that people of lesser means are not going to have the capability now. So if you make it available, make the coverage available, then you are providing them the means, and you are increasing their access.

So I think we can get to the point of trying to micromanage or letting, as was said before, let the perfect become the enemy of the good, if we're going to say that people don't have access now, then we're going to provide them access, we're enhancing, we want to do what we can to make sure that the level of training and services are equitable, or as close to equal as possible. But I think if we wait until everything is perfect, we'll never be able to act.

MS. BERRY: Barbara, did you have a comment?

MS. HARRISON: I basically wanted to echo exactly what Debra said. I can definitely see the link between health disparities and how that needs to be addressed in our report, because what comes out of our recommendations about coverage is going to directly affect the access that people have to genetic services, and I think this point has been brought up before, not just talking about disparities among ethnic groups, but also economically. If we're talking about things like Medicare, Medicaid, and what it is covering, then to me, it is just very plain about how that affects health disparities. So we can have that be a continual theme throughout the report as to why this is so important.

But again, agreeing with previous comments on the provider education piece, I don't necessarily see it being directly affected by our report on coverage and reimbursement. Definitely to access to services, but not in this report.

MS. BERRY: Emily?

DR. WINN-DEEN: Yes, so I guess I think the only place it belongs in the report is at the end where we say just having coverage and reimbursement does not assure that this becomes practice of medicine, that there are still other barriers that we have to deal with.

So that is where I'd just like to see sort of a sentence at the end that says, this is a huge barrier. If we overcome coverage and reimbursement, we are 90 percent of the way there in delivering this promise of genetics to the practice of medicine. But there still are

some other things that have to be in place for that to really be totally efficient. That's the only role that I see in it in this report.

DR. LEONARD: And can't we reference the other resolution?

DR. McCABE: Yes. I think it is always good to cross-reference ourselves, and I think it is also important to, and I think perhaps in that paragraph we could use some of the data that Reed provided us with that shows that even where there are U.S. Preventive Services Task Force guidelines, they are still not fully utilized. He has some excellent data in his comments that he sent to us.

Well, I want to thank everyone for a very productive discussion, and for a very productive day. I want to especially thank Dr. Boughman, Dr. Reede, Ms. Berry, and all of our panelists and public commentors.

We are now going to adjourn for the day. Members who are planning to join us for dinner this evening, please meet in the lobby at 6:40 p.m. so that we can then carpool to dinner.

To remind everyone, we are starting a half hour earlier tomorrow, so we will reconvene at 8:00 tomorrow morning. Thank you, and we're adjourned for the evening.

(Whereupon, at 5:22 p.m., the meeting was recessed, to reconvene at 8:00 a.m. on Tuesday, June 15, 2004.)