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C O N T E N T S

C O N T E N T S *(continued)*

P R O C E E D I N G S

DR. TUCKSON: Good morning to everybody. Good morning, good morning. Welcome to day two. It is amazing. I don't know how, but all of the new folk, even after the painful, tough work of yesterday, have returned.

[Laughter.]

DR. TUCKSON: It is amazing. We are starting on time, of course, which is terrific as well.

If you can see behind me, there are people who are gathering there because they thought that they were going to have a few more minutes to prepare, but it turns out that we are going to wind up being flexible.

We have three of our superstar members who are ending their term. They are gathered over there in the corner commiserating at all the pain, misery, and suffering we have put them through. They are going to get awarded. But to award people in government, you have to have people of rank, and sometimes people of rank have other things that delay them.

So until the rank gets here, we are going to, in the interest of time, march forward. We have our guests here, so what we are going to do is we are going to say to our guests thank you. We are going to say to our guests that you are flexible and we are flexible.

So what will happen, Judy and the rest of you, is that we are going to start. Then rank is going to come. Then we will stop and we are going to honor our departing three. Then we will resume again, because we are all intelligent enough to have more than one idea in our heads at the same time.

That is the way it is going to work. No matter what happens, though, we have to finish the ceremony by 9:30. So I can absolutely tell you there is a hard stop on the ceremony at 9:30 because folk have other things they have to do.

With that, we are going to turn this over to Barbara. I want you all to remember we went through the chart yesterday of what was important, our strategic plan and where we were with it. One of them was education of health professionals. So your job today is to listen and query these experts around ultimately making a decision about whether or not we need to go forward with something in this area and, if we are, what might that be that you might want to do. Or you might decide that the world is in good hands by others and you can turn your attention and intelligence to other weighty matters that are on our agenda.

So you don't always have to decide to do something just because we present it in front of you. With that, you are going to think through and deliberate.

Barbara is going to take us through it, so Barbara Burns McGrath.

DR. McGRATH: Good morning. For the next couple of hours we are going to be talking about genetics education and training of professionals. This is actually the most fun part of the Committee deliberations, I think, where we get to sit back and really learn from the people who are the top in their field in these things and have put together their ideas in a smaller packet of information for us. So I think this morning is going to be very interesting.

It is an area we are revisiting in a roundtable format just as we did a few years ago, and I think the reason is that it is one of those topics that requires or calls for periodic

review or a second look. This is one of those.

I think it is really an important or very appropriate agenda for this Committee. If we think about the pipeline that extends from genetic discovery to utilization, sitting right in the middle are the various health professionals that we are going to be talking about today. All of us here who do research or craft policy know that our great ideas can just pile up and get stuck right there if these people don't implement them appropriately.

So to use a word that has gotten more familiar in our vocabulary in the last year or so, these people that we are going to be talking about today, they are the deciders. They are the ones that move all of our discoveries forward.

As a social mandate, we want these professionals to be knowledgeable about genetic principles so they can, for instance, decide which genetic test is the appropriate one to order and then be able to interpret the results correctly.

We want them to understand complex ethical issues and then make decisions about difficult problems like whether or not to disclose results to asymptomatic children.

We also want them to attend to larger societal issues, like thinking about what is the best way to ensure equitable access to appropriate genetic services to under- or uninsured people in our country, and then to think through is this the best way to eliminate health disparities in our country.

So regulations are written and clinical protocols are created to offer guidance. That was a lot of what we did yesterday. But these folks are the ones that first have to learn about all these suggestions that we have for them, agree that they have relevance in their own professional worlds, and then make their own decisions about how to implement them.

So let me quickly review how we got to today's session and put it into some sort of context. A Reed just mentioned, the need to explore issues around education and training have been part of the strategic plan since its inception. I'm going to talk about how it fits into the Oversight Report in just a minute.

But first, back in 2003, there was a very similar roundtable held that resulted in the 2004 resolution that was presented to the Secretary of Health. Some of you here were involved in that effort. The entire resolution is in Tab 4, but briefly, there were nine resolutions, and I'm going to go over those. They are summarized here a little bit.

The first one is to incorporate in HHS programs and policies into the philosophy that genetic information is an integral part of all health care, then to facilitate this integration by collaborating with state, federal, and private organizations. This one also called for sharing of case studies and practice models specifically.

Another one was to promote and support initiatives that integrate genetics and genomics in education and training of health professionals. This resolution also referred to promoting family history information and supported point-of-care models. It also linked the Health Information Technology Initiative to it.

Another one was to encourage and support programs that promote diversity and cultural competency of the healthcare work force. This included issues associated with disability rights.

Work with relevant organizations to incorporate knowledge of genetics and genomics in accreditation licensure and certification processes. This one was suggesting using the accreditation process as a driver for educational programs.

Support federal programs for faculty training and clinical application-based education models, particularly those that include ELSI issues. This was an emphasis on ELSI issues in terms of educating the work force.

Promoting consumer education and supporting K-12 education programs. The K-12 issue here was intended to establish a pipeline for a future diverse work force.

As you can see, these are rather broad and general. After that committee submitted the resolutions, the SACGHS moved on to other priority areas related to its charter, and you heard a lot about those yesterday.

So in the intervening years after 2004, between 2004 and now, things have changed. It is time for the Committee to take another look. To that end, we have asked key people in the area to come and give their perspectives today. We are doing that now because, as you read the oversight report, it is clear that health professionals have a role to play here, and that is highlighted particularly in Chapter 6 of that report. We hope that this session may contribute to that final report.

Because of this link to the Oversight Report, this session was convened over a very short period of time and some of the folks here had a very short turnaround, in weeks, not months -- for some people it was like one week -- to prepare for this, so we appreciate the effort that they all gave to being here today.

These are the members of the steering committee. Many of them will be presenting today. I would especially like to thank Cathy Fomous of the Committee staff, who actively participated in the decisions and coordinated all the efforts. Cathy, it has really been a pleasure working with you on this.

These are the disciplines that are represented. Some people wore more than one hat. You will see that we do not have consumer representation here. Boundaries were placed to limit today's sessions to issues around professional development. The issue of scientific or genetic literacy is of course very timely and important and probably deserves its own forum.

Similarly, we are not including a review of federal efforts during this session. We are addressing education and training very broadly, including specialists and non-specialists, and consider the continuum from basic education, advanced specialization, to continuing education of those in the work force.

After reviewing the 2004 session and a discussion with the steering committee, these are the topics that were identified to guide today's discussion. The first one we are asking everyone to talk about is addressing the issue of professional education and training from their perspective. The second one is the whole issue of diverse work force. What is happening in their area, any progress made, any new plans for improving that.

What are the emerging issues in the field, and in particular, gene environment interactions that are very complex. We are interested about what efforts are being made to educate people about that, and emphasize its role in clinical care or public health.

Who are the emerging stakeholders that we should be thinking about in the future when we are thinking about education and training. Then, genetic family history is an area that is heavily marketed as a very important tool in individual and population-based care. Practitioners are being urged to integrate it into their practices, and we are just interested to know how is it going. There has been a lot of activity around it, but we haven't heard a lot of results yet.

Then, as I mentioned earlier, we are asking everyone to consider the 2004 resolution as well as the 2007 Oversight Report.

The purpose of this session is to provide an opportunity, as Reed said, to listen and have some conversation with the experts who are gathered here. It is a great opportunity to do that.

The format is that each person will have 15 minutes to present their views, followed by five minutes of questions that can be directed at that particular speaker. We are going to be holding to that time very carefully. There was going to be a break at 10:30. There will be a break someplace in here with the new change, but at some point we will get a break.

Then, after all six presentations are finished, we will then turn to the invited discussants for their comments. Then we will have a little more than a half or so for general discussion.

The goal here today is to decide on next steps. This is something that we are going to decide as a group at the very end, and for that end we would like you to consider two questions while you listen to all of this. The first one, does the topic of genetic education and training continue to be an area of concern consistent with the SACGHS's charter. Second, if so, what elements are most important for us to address considering the scope of our charter.

So, thank you. I'm looking forward to your participation in this. We will start with our first speaker, Mr. Joe McInerney. I will let each speaker introduce themselves and describe what group they are with.

Thank you, Joe.

MR. McINERNEY: Good morning. Thank you for the opportunity to address this group for the second time. I'm very grateful for that. I'm additionally grateful for your willingness to revisit this entire issue of education for health professionals.

I am the director of the National Coalition for Health Professional Education, which is known as NCHPEG, happily enough, so I don't have to repeat the full name too often. We work on genetics education for health professionals full-time. This is what we are devoted to: educating a broad range of health professionals. I have been doing this now for about eight years.

What I would like to do today is share with you some of my perspectives. I don't think I will cover all of those issues that were part of our assignment in 15 minutes, but I would like to give you some sense of the challenges and opportunities that I have been encountering and NCHPEG has been encountering and those many people who work with us have been encountering over the last seven or eight years as we have worked to integrate genetics into education and practice.

I would like to review some data about genetics education and genetics knowledge among health professionals, and then I would like to review with you just one of NCHPEG's many programs very briefly to give you some sense of how we are trying to address what we see as some of the particular barriers and opportunities that arise for us as a genetics community.

Now, I'm not sure you can read this from the back of the room. Can you? I will read it. It is really too much text for one slide, but I will read it to you anyway. Actually, I think I lost some of it.

It begins with a rhetorical question about what practicing physicians should know: "How much genetics knowledge should primary physicians have?" I hasten to point out that we can really extend this beyond physicians.

We will pick it up here now. Should they be able to diagnose, treat, and counsel about genetic diseases. Will it suffice for them to check the literature. I'm skipping ahead. Optimal knowledge must lie between these extremes because a primary physician should have enough knowledge to recognize a problem as genetic and should have enough familiarity with genetic principles to be able to use the literature wisely or to consult with a geneticist intelligently.

Now, that is, I think, quite a reasonable statement. It is a nice summary of some of the questions and problems we face right now. One of the difficulties for us is it was also an apt statement of the problem back in 1979 when this statement appeared in a book edited by Ian Porter and Ernie Hook on service and education in medical genetics. So we are still struggling with some of the same kinds of issues.

This slide is simply to illustrate some of the major challenges to genetics education that we have encountered over the last seven or eight years as we have tried to bring genetics education into curricula for various health professionals. None of these is a startling new piece of information for any of you, but some of them are, I think, more problematic than others because they are, in a very large sense, systemic issues.

It is very important for us to understand that when you encounter any educational system you are encountering a complex ecosystem. As the great ecologist Garrett Harden once said, in a complex ecosystem it is impossible to do just one thing because no matter what you do reverberates throughout the rest of the system.

So, especially for those of you who teach in schools of medicine or in schools that prepare other health professionals, none of these things, as I say, is startlingly new for you.

But I will tell you that one of the issues that has come up over and over again is this issue here: a disconnect between basic sciences and clinical experiences during training. I'm going to talk a little bit more in a minute about the way genetics content is distributed, particularly in medical education, undergraduate medical education that is, before graduate training.

But one of the things we keep hearing here is that if students do encounter the basic science of genetics in the first one or two years of medical school for example, when they get to their clinical rotations they very rarely encounter people who are teaching in those rotations who understand genetics sufficiently well enough to elaborate and bring forward the genetics principles that they have learned in their first two years.

That, I say again, is not simply the case with medical education. We hear this for preparation of all health professionals. There is this separation between the basic science and clinical practice, and it reverberates into practice, as you will see in just a moment.

I'm not going to talk about all of these issues. I will say here that there are some courses -- and you will see in a minute when I show you some data -- where the instructors or the institutions claim that they are integrating genetics across the curriculum. For example, they say, "We don't have a genetics course but it is in biochemistry." I always recall the statement that Bruce Korf made some time ago, maybe even to this group, that often the genetics is so well integrated as to be invisible. So that is an issue.

These challenges then reverberate from education into practice. We will hear more about workplace issues I'm sure from some of our colleagues, but we do have a dearth of genetics professionals. There, of course, is a lack of knowledge about genetics among primary care providers. We still encounter misconceptions, some of them quite startling, for example about the deterministic nature of genetics and that if it is genetic we can't do anything about it anyway so why should we really bother learning about it. We hear that on occasion, happily not too often.

But there are still misconceptions that genetics is associated primarily with rare single-gene disorders and chromosomal anomalies that are circumscribed by two disciplines, primarily pediatrics and obstetrics. We have to work very hard, I think, to counter that perception.

You will hear more later, I'm sure, about inadequate family histories, and lack of referral guidelines is simply, in some sense, a catch-all phrase to say that we do not have enough clinical guidelines related to genetics in general to raise the level of importance of genetics for primary care providers and others who are providing services that we would like to see integrate genetics.

Now, the response we hear most often, and I hear this over and over again when I go out to talk to health professionals about genetics, and particularly if we talk about what is happening at the cutting edge of genetics, is: "That's great stuff, but I want to know what I have to do now." Those of you who are in practice know better than I that time is a significant issue. If we can't give providers something to do that is quite concrete and that is likely to improve patient outcome, it is very difficult for them to think about integrating it into practice.

If we say to them, five years from now we are going to be able to do X, Y, and Z, they say, "Great. Come see me in five years."

Now, this is a bit of a tongue-in-cheek comment from Charlie Epstein when he gave his presidential address at the American College of Medical Genetics, but I think it is important for us to pay attention to this because we as a genetics community believe that genetic information has value in and of itself. But the transmission of that information takes time and somebody has to pay for that somewhere along the way. So this is another issue that keeps coming up again and again.

Genetics is not a discipline that does a lot of stuff. It doesn't order, at this point at least, a lot of tests or a lot of procedures. Still, the information has great value, but how does it get reimbursed.

Now, Ms. Aspinall, who is at the table with us and a colleague from Harvard Medical School, published this paper recently in the Harvard Business Review. The assertion here is that physician behavior is one of four particular barriers that stand in the way of the realization of the promise of personalized medicine. One finds in the paper this assertion: "Most medical schools have yet to fully incorporate genetics and genomics into their curricula."

Let's take a look and see what we know about that and whether in fact we can demonstrate that with some data. In fact, I think this paper is included in your packet. There is a paper that appeared in Academic Medicine back in May by Virginia Thurston and her colleagues at the University of Indiana School of Medicine. They sent a survey to this group, as you see, of 149 U.S. and Canadian medical course directors, and they had a pretty good response rate as of June 2005. I will just review very quickly with you some of these data.

Seventy-seven percent, as you can see, reported that they teach medical genetics in the first year. Only 47 percent incorporate it in the third or fourth year. This harkens back to my previous comment about the separation of genetics from the basic science years and the clinical years, and it is going to come up again, for example right here.

Now, this seems to be a reasonable amount of hours of instruction devoted to genetics. It depends on how one defines it. Eighty-six percent, and we will talk a little bit more about this in a minute, say they cover primarily general concepts, but only 11 percent say they address practical applications of genetics.

Of course, to me, this illustrates the problem all over again that when the students are getting into their clinical years there aren't people who can elaborate the genetics perspectives that they have been introduced to during their first two years, or in whatever the training course is for the health profession in question.

Forty-six percent report a stand-alone course, and 54 percent say they integrate medical genetics into another course. I just told you a little bit about my perspectives on that.

Now, these are some additional data of the most commonly taught topics. This is a very interesting paper, by the way. There are, I think, a lot more analyses that Dr. Thurston and her colleagues can do. There are some data in there about whether these courses are being taught by individuals who are board-certified in genetics or not. What I think the authors are about to do is take a look at some comparisons with respect to the course content to see if there are significant differences in the course content based on the certification of the individuals.

But you see the topics that are covered. I thought this was pretty interesting, that 91.3 percent address multifactorial inheritance, given our increasing concern about common complex disease.

I should point out that the data about the lack of understanding of genetics and the lack of preparation of professionals in genetics, those data come not only from analyses within the healthcare community itself by other health professionals but also from the public at large.

Now, admittedly, this is a selected group. This is an analysis that we conducted in conjunction with our colleagues at Genetic Alliance. We had almost 6,000 responses to this survey of consumers' perceptions of the genetics knowledge of their providers. I will refer you to this paper. In general, the news was not very good. The consumers did not evaluate their providers very well with respect to their understanding of genetics.

Here are the central questions and challenges. There are embedded in here a number of opportunities, of course, but increasingly we struggle with these issues. Which content is appropriate and for whom.

There is a great deal of difference between "accurate" and "complete." Those of you who have developed educational materials know that we struggle with this issue all the time. We are not going to turn, and we don't hope to turn, all other health professionals into geneticists, so they will never have a complete understanding of genetics the way many of the people around this table have it.

But, what is the slice we have to take through that content, is it accurate, how do we ensure that it is accurate, how do we ensure that it is clinically relevant, and which clinical behaviors and attitudes do we want to change and can we.

Presumably, our educational programs are intended not only to increase knowledge but to change behavior. We want people to do something differently. What are those

things we want them to do and how do we measure them.

I'm skipping ahead a little bit to how do we define and measure success, but equally as important is how do we get these materials to people and how do we get them used so that we don't end up with what I like to call "state-of-the-shelf" materials. There is lots of good stuff out there, but many times it sits unused. So, how do we improve implementation and use, and then how do we define and measure success. If we are trying to change clinical behavior, how do we measure that. If we are trying to assert that education will improve patient outcome, how do we measure that. Those are very complicated and costly tasks.

I will skip ahead fairly quickly here, but just to say that we have recently produced a third edition of our Core Competencies in Genetics, which we first presented to this group a very long time ago. Based on feedback from the community, we have pared those down considerably based on surveys and based on what people tell us is really important in teaching and practice. I have some handouts of that that I will be happy to send around.

Here is another program I wanted to tell you about that is one of our attempts to try to provide access to genetics content in a clinically relevant way. I should say, by the way, that we at NCHPEG don't believe we are providing the answer, the only solution. After eight years of doing this, I don't even know what all the questions are, much less the answers.

So this is one potential solution, and we call it Gene Facts. It derived from our observation. As I have said in other settings, this is a little bit more rigorously dichotomous than the reality displays.

But there are these open-source genetics databases that include these characteristics, and subscription databases that include these characteristics. Often the subscription databases are highly clinically relevant. The material is presented in ways that the providers can relate to, but it is often not very sound.

The genetics data, on the other hand, are generally very sound, but the providers can't access them; that is, conceptually or in terms of their practice. They can access them physically but not conceptually and practically.

So we are proposing a middle ground, a point-of-care decision support system where the material is written by primary care providers and geneticists working initially with content abstracted from gene reviews or created de novo from content that doesn't reside on gene reviews. It is our hope that we will be providing information that is clinically relevant and genetically sound.

I should point out that Dr. Khoury's group has provided us with some seed money to get this started and we have made a lot of progress on this. I won't share the template with you now, but I will be happy to send it to any of you who are interested.

Just to let you know, we do have a list of criteria for selection of the first 50 entries that we will put up on this system. I'm not going to go over those with you, but I do want you to know that we have thought about that very carefully.

This is something I shared with you the last time I spoke here three or four years ago. I think it is even more germane now. I think we make a mistake by talking to our non-genetics colleagues about genetic disease all the time because for them, in my view, that simply walls genetics off in ways that we don't want it walled off.

What I'm saying is, and I know it is almost heretical to say it to this group, I really would like us to stop talking about genetic disease as if there is genetic disease and non-genetic

disease. I don't know what the easy locution is yet, but I would like it to express something such as the following: it is not whether the disease is genetic or not genetic, the question is what role is genetic variation playing in the onset and expression of disease now in this particular person. Sometimes it will be quite salient, sometimes the genetic contribution will not be quite so salient.

But I would like us to start thinking a little differently. If we want our colleagues in the other health professions to think genetically and think differently about genetics, I think we have to provide some guidance for them in doing so and not continue to convey the notion that there is this what I consider to be a false dichotomy of genetic and non-genetic disease.

I thank you for your patience, and I will stop there.

[Applause.]

DR. McGRATH: Thank you. I think your perspective of being on the roundtable before in 2003 was really valuable. Thanks for starting us off with that data.

We have a minute or two for some questions.

DR. KHOURY: Thank you very much for this wonderful work that NCHPEG is doing. I would like to pick up the theme of stop using the words "genetic disorder" and "genetic disease." [I will] ask you to guide us a little bit about NCHPEG's activities over the last few years in this context particularly because I heard you a couple of times say genetic information has intrinsic value for us as geneticists. We have to provide guidance.

People sitting on the other side in the context outside genetic diseases for which genetic information has intrinsic value, they don't know whether it has intrinsic value, let's say for pharmacogenomics or for the treatment of Disease XYZ. They don't know whether it has intrinsic value or not.

The question that I want to ask NCHPEG [is], has NCHPEG picked up the concept of evidence-based medicine and working with the primary care providers and the evidence-based communities to see which type of genetic information -- outside of genetic diseases, which we all know we need to diagnose, treat, counsel, et cetera -- needs to actually be moved into mainstream?

The average practitioners are still asking all of us "What should I do now?" So in the absence of evidence-based guidelines to guide them to do this, they may not think that genetic information has intrinsic value. Help us go through this.

DR. JOHNSON: Thanks, Muin. That is a very interesting question. I will have to say we probably haven't addressed the concept of evidence-based medicine as carefully as we should have in most of our programs, or at least in some of our programs.

We do work with our colleagues in the provider community to try to identify those cases and those instances in the clinical setting that are most important and germane to them, and they actually try to help us to frame the discussion about what kinds of evidence will resonate with their providers.

In a more concrete sense, we have, in conjunction with Genetic Alliance, been working on, actually, a CDC-funded project called Access to Credible Genetics information. I don't have the acronym quite right.

But the issue here was for us to try to apply the principles of evidence-based medicine to the selection of genetics information that providers and patients can use to make informed decisions to improve health care. We found that there was not an easy one-to-one correspondence between the types of evidence that AHRQ requires, for example, and the types

of information we want to present to providers and to individuals so they can make decisions.

So we have actually developed a separate toolkit and metric to help individuals, providers and patients alike, judge the completeness and the accuracy of the information that is available to them both in an educational setting and from the literature.

I don't know if that helps answer your question or not.

DR. McGRATH: Thank you.

DR. TUCKSON: We are going to pause for just a moment. Thank you all for your indulgence. We are extremely pleased that three of our members who have rotated off the Committee took the time and the energy to change their schedules to be with us today. One of them has to go off to a meeting that we promised to get her out for, and others will be here for various parts of the day.

We are extremely excited to honor three of our most outstanding members in the history of this Committee. You talk about some hard-working three folk. It really is painful for me because I came in with them and, golly, I just feel so sad that they are not going to be around.

Anyway, let me start with Cindy Berry. Cindy is an attorney at Powell Goldstein, where she is chair of the government relations practice, and a member of the firm's health care practice. Previously, she was general counsel and managing director at Wexler & Walker, Public Policy Associates.

Prior to joining Wexler & Walker, she served as Washington counsel to the AMA and practiced law with Trabue, Sturdivant & DeWitt in Nashville, Tennessee. She also served as legislative assistant to then Representative John Kyle.

Ms. Berry practiced law with a bunch of people before moving to Washington.

[Laughter.]

DR. TUCKSON: See, every firm she has been with has like 18 different people so it just will mess it up completely if you try.

Anyway, her issues are specializing in healthcare law, medical malpractice defense, and commercial litigation.

In addition, she served as vice chair of the Virginia Birth-Related Neurological Injury Compensation Program and was a member of the Secretary's Advisory Committee on Genetic Testing. She received a law degree from Vanderbilt University School of Law and is admitted to practice before the United States Supreme Court.

Cindy, could you join me, please?

The remarks from the Secretary on your certificate are that "You were appointed to SACGHS for your expertise in law and public policy. As a member of our predecessor committee, the Secretary's Advisory Committee on Genetic Testing, you provided important continuity between the two groups. During your tenure with us, you spearheaded the development of the Coverage and Reimbursement Report and made significant contributions to the Committee's extensive work on genetic discrimination, gene patents, and licensing and oversight.

"We thank you for your service, and we are pleased that this certificate of appreciation is being offered to you on behalf of Secretary Leavitt for your public service and commitment to addressing issues raised by the development and the use of genetic technologies."

I will say personally, Cindy, you talk about hard-working but also reliable, trustworthy counsel. She is a safety blanket intellectually, and I want to tell you thank you very

much.

[Presentation of certificate.]

[Applause.]

DR. TUCKSON: Cindy, thank you so much.

I'm really happy now to ask Chira to start to come forward. Chira Chen is representative of the San Francisco Advocacy Corps, a volunteer group that shares the patient's perspective with breast cancer researchers at the University of California. She is a survivor of lymphoma and breast cancer. Her father and mother died from cancer.

MS. CHEN: No, no, not mother.

DR. TUCKSON: I'm always the one who pays for this.

[Laughter.]

DR. TUCKSON: She is a member of the Planning Committee for National Survivors Day in the San Francisco Bay area and does extraordinary volunteer work with cancer patients and their families.

She has served as a reviewer for breast cancer research grant applications for the Department of Defense, providing the most important patient perspective. Professionally, Ms. Chen is a staff research associate at UCSF Comprehensive Cancer Center.

The remarks on her certificate: "Chira Chen was appointed to SACGHS in 2005 for her expertise in consumer advocacy and to bring consumer perspectives to the Committee's deliberations. During her tenure, she provided important insights and contributed significantly to several taskforce efforts, including the Large Population Studies Initiative and Gene Patents and Licensing.

"Ms. Chen, on behalf of Secretary Leavitt and in recognition of your public service and commitment to addressing issues raised by the development and use of genetic technologies, I am pleased on his behalf to present this certificate of appreciation."

I will say personally, again, that it has always been just a joy to have you around the table and knowing that when that patient and consumer perspective needed to be there your voice of conscience was always important. But also, so was your smile and your attitude. You always enlivened and brightened all of our deliberations.

So, thank you for your example and your role-modeling.

[Presentation of certificate.]

[Applause.]

DR. TUCKSON: Finally, for our friend Hunt, who we very rarely call Huntington. They have given me nothing of the official wording. I wanted to read what the Secretary's was going to have.

I'm going to say what I'm going to say, but I have to read what the certificate says, and they have to hand me the certificate.

DR. BILLINGS: Just cry, Reed.

[Laughter.]

DR. TUCKSON: "Dr. Hunt Willard," it says on your certificate, sir. "You were appointed in 2003 for your expertise in human genetics and genomics. Over the past four years," and I know they went like that, "you provided important leadership as chair of our Large Population Studies Taskforce and you guided the development of the Committee's comprehensive report on the policy issues associated with these types of studies.

"You also served on the Pharmacogenomics Taskforce and contributed insights and knowledge on the many other issues addressed by our Committee.

"Dr. Willard, on behalf of Secretary Leavitt, we are pleased to give you this certificate, with our gratitude, for your public service and commitment to addressing issues raised by the development and use of genetic technologies."

I will say that Hunt has been very often a right arm me, trying very hard to keep me in line and to keep making sure that we were informed by the absolute best scientific thinking on these issues. You have been a rock solid rock star, Huntington. Thank you so much.

[Presentation of certificate.]

[Applause.]

DR. TUCKSON: Just to let our three leaving people know, the five new people coming in got a real treat yesterday of how hard this Committee is and how hard we work. I do hope that you can sense that the bonds that are formed by doing this kind of work are pretty intense. So it is just great that we stopped the whole train for a minute to really say thank you to the three of you.

You are welcome to stay and be a part of the deliberations as long as you want. I do hope you recognize that no good deed goes unpunished and that there will be the very long arm of the Committee reaching out and grabbing you for some taskforce or another, i.e., you cannot get away from us.

Once again, everyone, please.

[Applause.]

DR. TUCKSON: To our guests, thank you for your indulgence in allowing us to change the schedule. I appreciate your flexibility.

DR. McGRATH: That was nice. Getting back to it, the next speaker is Norman Kahn, who is with the American Academy of Family Physicians. Thank you.

DR. KAHN: I appreciate very much being here. I'm going to try to represent the position of medical education in this. The reason that I'm sitting here is that I have two video clips. They don't transfer on a thumb drive, so we are going to do our best with the video clips.

There have been a lot of revolutions in health care: antibiotics, aseptic techniques, surgical anesthesia, imaging. If you are part of these, you recognize the kind of revolution that takes place when these come into practice. Immunization, sewage disposal, water purity. The real question is whether or not genomics is going to turn out to be at the same level of revolutionary in the practice of medicine.

Francis Collins' great quote: "Virtually all diseases, except maybe trauma, have a genetic component." I think we all recognize that. As Joe McInerney has pointed out, I don't think that that concept has been integrated well into medical education yet.

I'm going to tell you about two projects, and I'm going to hint at a third one, and illustrate them with a couple of video clips. At the end, I'm going to do a little needs assessment for you to help guide your next steps.

The first project I'm going to talk about was called Genetics in Primary Care. This projected started in 1998. Remember the genome was only sequenced in 2003, so this was really a prescient project. We very much appreciate the agencies that you see on the screen for their foresight in being able to recognize that it was going to be necessary to begin to educate medical faculty about genetics so that they could incorporate genetics into medical education.

The goal, again, was to get it into undergraduate and graduate primary care education.

Now, the next two slides are probably the two most important slides of this presentation. The most important concept that we learn from the GPC project was the concept of primary care through a genetics lens and then genetics through a primary care lens.

When we brought the primary care community -- family medicine, pediatrics, internal medicine, to some extent OB/GYN -- [together] with the genetics community -- medical genetics, genetics counselors, Genetic Alliance, et cetera -- we discovered that there were two different languages, two different cultures, two different perspectives. Each of them was ignorant of the other's perspective. Recognizing the two perspectives was absolutely critical. Otherwise we would not be able to communicate with one another.

The concept of primary care through a genetic lens: as a primary care physician, I need to expand my differential diagnosis to include genetic conditions. I need to use an appropriate family history to identify genetic conditions. I may or may not be able to do a three-generation pedigree, but I certainly need to do an appropriate family history. I need to recognize the importance of non-directive counseling, which is the hallmark of genetic counseling, which is not the hallmark of primary care counseling, as we will see in just a moment. I particularly need to recognize the ethical, legal, and social issues raised by genetic diagnosis.

Now, the genetics community recognized that they needed to see genetics through a primary care lens. If the genetics community is going to contribute to genetics in primary care, then the genetics community learned that they needed to evaluate the utility of genetic information in terms of health outcomes.

The genetics community's basic premise is that all information is valid and valuable. What the primary care community contributed was that it needs to be evaluated. The utility needs to be evaluated in terms of health outcomes.

They need respect for patient preference. The patient is not ready for all information, doesn't prefer to make all decisions themselves.

Protect patients from media hype, and use the potential of longitudinal care. We learned that the genetics community's usual encounter is an hour in a consultation with a patient, whereas the primary care community is seeing people for 15 minutes on a weekly basis, on a monthly basis. People get ready for new information. Those two concepts became very important as we played out the relationships.

There were 19 teams. I think you all have a handout, so I'm not going to go through each of these teams. We put them up there in case you know people at these particular groups. You can talk with them about their experiences.

This was the GPC curriculum. There were seven topics that were recognized in primary care as having a genetic component. Now, what is interesting is, with the possible exception of hemochromatosis, in the primary care community, as Joe pointed out, we wouldn't consider these genetic disorders. Breast cancer is not considered a genetic disorder, nor is colorectal cancer.

Thinking genetically allows us to recognize the family history component, distinguish among the different types of colorectal cancer for example, see which are genetic and which are not. But the concept of patient acceptance is enhanced by not referring to these as genetic disorders.

There were several complementary tools. There is a curriculum. I'm going to

give you the website for that curriculum. You can still get to it. We talked about evidence-based medicine even back then, between 1998 and 2003, and particularly cultural competency.

Here are the websites that you can go to. One of them is a federal website at the Maternal and Child Health Bureau. The other is the Genes R Us website, and you have these handouts.

After 2003 then, having focused on undergraduate and graduate medical education, we decided to focus on practicing clinicians and produced a program called Annual Clinical Focus, which means we spent the whole year of 2005 focusing on genomics and educating primary care clinicians at the practice level.

These are the supporters. These are the folks who paid for this particular endeavor, which we are extremely appreciative of. Again, additional supporters. One of the things that is interesting here is that there aren't very many pharmaceutical companies in the support here. Pharmaceutical companies love to support continuing education for physicians, but they weren't ready to support education in genetics. Very interesting.

These were the cooperating partners. You can see we did this with the nurse practitioners, pediatricians, physician assistants, medical geneticists, the internists, as well as family physicians, a variety of genetics groups, the Heart Association, the March of Dimes.

Here were the topics. You can see there is a lot of overlap in the topics. Family history was added in there. Bipolar disorder. I'm going to show you some usage data that I find particularly interesting.

First, I'm going to stat with a video clip, if I can get the sound to work. We did test the video and I know the video works.

Just to give you a sense, there were eight programs with those eight particular topics. I say past tense because continuing education has a life span. It has an accreditation span of two years. Unfortunately, there are only two of them that remain on the Web. You can get to them if you get there before the end of November, when they expire. There are two remaining, and they are just at the homepage of the American Academy of Family Physicians, AAFP.org. Right there on the homepage you will find "Annual Clinical Focus" underlined, and you can get to these.

But let's see if we can get the video clip to work.

[Video presentation.]

DR. KAHN: I'm going to go on just in the interest of time. I think you get a feel for what we are trying to do with these programs. These are each 30-minute programs. We are trying to model. This in particular is a family physician, but we have a nurse practitioner on one, we have a physician assistant on one, we have a pediatrician on one. We are trying to model the interaction with the patient so that the practitioner gets a sense of what they can do now, today, in practice, not waiting for some new breakthrough.

We had hoped that the Web-based delivery would work. This is the usage data. When we talked to our partners, we had no way of knowing how many people would participate in this. If we got 5,000 visitors or users, we would be thrilled. As of today, we are over 30,000 unique visitors to this program. So we have far exceeded our expectations. My response to you is that this is a very good vehicle for educating clinicians in practice.

I'm just going to let you know that the residency training programs are also core curriculum for incorporating genomics into residency education. It is a particular reprint which I

can get for you if you particularly need it.

Here is my needs assessment for you, as I conclude. Even though we did a five-year faculty development project and even though we spent an entire year with the entire primary care community, educating primary care clinicians, it isn't over. We still see that primary care clinicians need to see primary care through a genetics lens.

Primary care clinicians need to incorporate family history as a standard of practice in each patient's health record. Primary care clinicians need to be sensitive to the ethical, legal, and social issues in helping patients and families approach genetic testing.

Remember there are 2,000 certified medical geneticists and 2,000 genetic counselors in the United States. There are communities that don't have these professionals. A quarter of the U.S. population lives in communities of 10,000 or less. The only clinicians in those communities are family physicians, osteopathic physicians, a few general surgeons, and some nurse practitioners and physician assistants.

Primary care clinicians need to know the evidence-based implications for genetic testing. A full disclosure: I serve on the Advisory Committee for Heritable Disorders and Genetic Diseases in Newborns and Children, which is dealing with implications of newborn screening and particularly the evidence-based implications for genetic testing in that realm.

Primary care clinicians need to be comfortable in interpreting and following up on genetic tests with patients and families. It is not going to be sufficient to say, "Oh, look. Your screening test revealed something. I will get you an appointment with a genetic counselor." That is going to be fine in a large, tertiary care center. It is not going to be fine in most community settings.

Primary care clinicians need to be comfortable managing chronic genetic-based illness using multidisciplinary teams and community resources.

Primary care clinicians need genetic decision support integrated into their electronic health records. You might be interested to know that the integration of electronic health records in the primary care community has really taken off, again much more quickly than some of us would have expected. In the family medicine community, the percentage of offices that are on electronic health records is close to 40 percent, with another 12 percent in the process of purchasing electronic health records. So the need for embedded decision support is right now.

I don't know how many of you have heard Dr. Collins' musical description of the current state of genetics education. I have Dr. Collins' permission to share it with you. In Francis' words, he says, "Oh, go ahead. My life is an open book anyway."

[Laughter.]

DR. KAHN: The Dr. Henley that he refers to is his medical school classmate Doug Henley, who is the CEO of the American Academy of Family Physicians. This was at the annual meeting of the society.

[Video presentation.]

[Applause.]

DR. KAHN: Just an interesting interlude.

DR. GUTTMACHER: Can I just give a little context for this? My major job as deputy director of the National Genome Research Institute is to try to keep Francis from doing this.

[Laughter.]

DR. GUTTMACHER: But I do have help in high places. As some of you know, a couple of weeks ago Francis was awarded the Presidential Medal of Freedom. Two or three weeks before that, he got called by the White House and told that he was going to be bestowed this medal and given directions about where to show up, how many guests he could bring, and that kind of thing. At the very end, the staffer said, "Oh, yes, Dr. Collins. There is one more item that I was specifically asked to tell you about." Francis said, "Yes, what's that?" He said, "No music."

[Laughter.]

DR. KAHN: As speakers are wont to do, Dr. Collins was gracious enough to receive individuals who come down to the stage and talk with him. My job was to guard his guitar because he has a very nice Martin guitar. Hordes of people came down to talk with him, and two people came straight for the guitar. They didn't care about Dr. Collins. They recognized the value of that guitar.

DR. McGRATH: Thank you very much. No one would have any questions after that. You have one?

DR. TUCKSON: Just one quick one.

DR. McGRATH: Go ahead.

DR. TUCKSON: We can get it to later, Norm. I was curious that you didn't mention the boards. I'm just wondering whether or not there is an effort to coordinate the stuff that you guys are doing with the board itself.

DR. KAHN: Not much. I would add that to the needs assessment, Dr. Tuckson. I think that the recognition of the role of genomic medicine in the certifying boards is in need of enhancement.

DR. TUCKSON: We will come back to that in the discussion. Thank you.

DR. WILLIAMS: Reed, just to add to that, there has been a recent assessment that was presented at the American Society of Human Genetics meeting, led by Darrell Waggonner at the University of Chicago, that has looked at least at the national board -- not specialty boards, the National Board of Medical Examiners -- looking at genetic content in Parts 1, 2, and 3. There is decreasing content over the course of those three examinations. But there is some formal work that is going to be published on that in the very near future.

DR. TUCKSON: Thank you.

DR. McGRATH: Dr. Kahn, thanks. I appreciate your perspective on primary care. I especially like your double vision of the lenses. Briefly.

MS. CARR: Yes. I just wanted to say that laughter and humor is one of the best ways to get to the general population. I would suggest that this clip be placed on You Tube.

[Laughter.]

DR. McGRATH: Beth, I'm sorry you have to follow that. That is quite a challenge.

Beth Pestka is at the International Society of Nurses and Genetics.

That clip is probably on You Tube already. It was probably there within moments.

DR. TUCKSON: I'm told that you can Google it, actually.

MS. PESTKA: This is really a wonderful opportunity for me. I am brand new to this type of an experience, so it was very exciting. My family thought I was really

getting important in the world to come to Washington, D.C., and speak to a group such as this.

My name is Elizabeth Pestka. I usually go by "Beth." I was invited to speak to you today on behalf of nursing. There are 2.9 million nurses in this country, so it is the largest group of healthcare providers. I am from Mayo Clinic in Rochester, Minnesota, and we were identified as having an exemplary model of genomics education for our nurses.

Sometimes people will say, "Are all your nurses educated?" It is like, no, I don't promise that. "Are all your nurses competent in genetics and genomics?" No, I won't promise that, either. But we are making some really good headway. So I'm going to share with you an overview of nursing and then, more specifically, what we are doing in relationship to our program.

Prior to 2004, there were many excellent initiatives in the profession of nursing. If you had the opportunity to look at any of the articles that are in the package, you can reference some of those initiatives. But they weren't real condensed and real planned type of activities. There were some scattered things that were making some good inroads.

What happened in 2005 is that two individuals from the International Society of Nurses and Genetics and also who work for the government here in Washington, headed up a group. Their goal was to get more of a concentrated and more of a cohesive plan for how we could integrate genetics and genomics into nursing. They started work in 2004 contacting nursing organizations.

With such a large body of nurses -- there are about 80 recognized nursing organizations in this country -- they actually were able to garner support from 48 of those organizations. That is pretty amazing. That is a very, very high percentage, especially when you think nursing is mostly women and women don't always tend to agree and see eye-to-eye on everything. I think that is really an amazing accomplishment.

In 2005 then, there was a meeting to endorse the essential nursing competencies. The two individuals who deserve an enormous amount of credit for working on this are Dr. Jean Jenkins and Ms. Cathy Calzone. They have done phenomenal work on organizing and bringing consensus to this whole program.

In 2005, they gathered individuals from these organizations who were interested in endorsing genetics and genomics competencies, and did come to consensus. Those competencies are listed in your packets.

What happened then is the support of these organizations, and in 2006, last fall, there was another meeting to identify an integration plan. So this is for implementing these competencies into education as well as into practice. Again, just an extremely excellent oversight and overview of how we are going to be doing this.

In the implementation plan, there are three focus areas. First of all, the nursing academic focus. If we don't prepare nurses of the future, we really won't have anything to work with. Secondly, practicing nurses, and thirdly, regulatory and quality control focus. So there are any number of items identified under each of these categories.

Dr. Ann Cashion is going to speak a little bit more to nursing academic focus a little while later, and so what I'm going to primarily talk about is practicing nurses, because I am a nurse in practice and I work with many thousands of nurses in practice at Mayo Clinic.

What is the plan or what is the theory that this whole implementation plan is based on. It is Everett Rogers' Diffusion of Innovation Theory. I had the opportunity to speak at

length with Dr. Jean Jenkins in September. She came to Mayo Clinic to speak at one of our conferences. She said normally this process takes about 17 years, but she said she is far enough along in her career she doesn't want it to take 17 years. So the plan is set to take place in five years. That is the goal for this plan.

In the Diffusion of Innovation Theory, there is the knowledge stage. So we need to educate nurses on what is genomics. I love it; at Mayo we sometimes have some of our nurses go out to other conferences and they come back and say, "Can you believe it? Nurses at these other places don't even know what genomics is." It is like, think, a few years ago did you know what genomics was?

We do use the term "genomics" quite liberally because we want it to be inclusive. We don't want it to be those pure genetic disorders, so we use the term "genomics" quite liberally.

Then there is the persuasion stage. Why is it important relevant to me. We have certainly learned that it has to be very specific. What is convincing to a nurse in orthopedics isn't convincing to a nurse in hematology, or isn't convincing to a nurse in psychiatry. So it has to be very specific. We have to have information and examples for each subcategory.

Then the decision-making stage. Is genomics worth the effort. Is it really something I want to put my effort into. People are busy. As has already been mentioned, time is of the essence and there are so many competing priorities. Is this worth the effort.

Then the implementation stage. How do I include genomics in my practice. Primarily, what does it look like. Again, this has been alluded to, and I would like to speak to that a little bit more.

Then, finally, the confirmation stage. Am I competent in utilizing genomics in my practice. Again, what does that look like.

At Mayo Clinic, in 2001 our leadership said genomics is the future of health care, no doubt about it. Absolutely no doubt. Our president and CEO, Dr. Denis Cortese, said the future of health care lies in translating genomics advances into practice. Patients will go to the providers that are best informed and best equipped to provide genomic services.

So this has been a prevailing them, and we certainly have been moving in this direction, to the point that we are establishing a center for individualized medicine based on genomics information. We aren't there yet, but we are moving in that direction.

One of the things that was really pivotal for our program at Mayo Clinic was receiving the Magnet Prize. This has been described as the Nobel Prize in nursing. We received it for our nursing genomics program. This was very, very exciting. But again, it brought tremendous national and international attention to genetics and genomics education. People who hadn't considered it before were thinking, "My gracious. This got the Magnet Prize. This must be important stuff."

Actually, even on an international basis. Recently, I was invited to Singapore. They really are seeking Magnet status, and they said, my gosh, this is Mayo Clinic and they got the Magnet Prize for this program. We better consider doing this. So they were really motivated. They were like, if we invite you back next year for two weeks could you get all of our nurses competent in genetics and genomics? I'm like, I don't think I can do that, but I'll come back anyway. It was a great visit.

But it is very important because this has generated a lot of interest and a lot of

focus on genetics and genomics. It was recognized for a very grass-roots effort.

What I would like to say is our organization says genomics is important, the profession says genomics is important, and most importantly, our patients say genomics is important. I have a very brief video clip. This is one of our patients. It is an unscripted, unprompted recording. He did consent to this. But I just want to point out how really important [this is.] Patients come with high hopes for genetics and genomics, and we need to realize those hopes. We need to realize what they are looking for.

This one ties in especially well for this group because he is speaking to pharmacogenomics, which we are actively using.

[Video presentation.]

MS. PESTKA: That is a pretty powerful story. James certainly has high hopes for genetics and genomics. We certainly aren't there yet, but he did come specifically for the pharmacogenomic testing because he has been on so many different medications. He has suffered and struggled so much that he is hoping that this will be something that will be able to help him out as well as his family members.

We have done a series of these video clips related to other diagnoses, and these really sell the education. It grabs that affective component. It is not just the intellectual, it is the affective. These are real patients, these are real issues, and they are looking to us for answers. They are looking to us for care. So it is just a really powerful series.

Just a little overview of what we have done to enable us to receive the Magnet Prize. We have a very diverse program. We started it back in 2001, when the rest of the Mayo Clinic said, okay, we have to educate everybody. Again, it has been incremental and it has been step-wise, but we are moving forward.

We did presentations to the leadership groups, and articles. We started a nursing staff development curriculum, which is a four-hour class that is available to nurses. We have had posters at every single nurses' poster fair for the last years, starting in 2002, just to keep it there and keep it visible. A lot of specialty education methodologies, so we have had a lot of things going on.

We do have a wonderful intranet specific to nursing at Mayo Clinic, and it is being utilized more and more all the time. I haven't been measuring the hits but probably should be because I know the use is increasing. I'm getting more and more correspondence.

In our nursing conferences, we include genetics and genomics in almost every single one. We sponsor about a dozen conferences every year, and is becoming more and more prevalent in our nursing specialty curricula. So when nurses are beyond their general orientation, if they are going into hematology, oncology, of course they get some education on oncogenesis. If they are in psychiatry, they get the pharmacogenomics stuff. If they are in other specialties, there is some genetics and genomics integrated into all of the specialty curricula.

The centerpiece of our whole program, the most exciting part, is our nursing genomics interest group. So we invited all nurses. We have over 6,000 nurses at Mayo Clinic Rochester. Anybody with an interest could join this group. We started out with about 30 people. Now we are up to over 100 individuals, and they are doing marvelous things. Very grass roots, but they are doing marvelous things in teaching their peers.

We started with two things, and we have added a third. You have to learn yourself, you have to teach your peers, and then in this last year we added you have to start role-

modeling the competencies. So we keep it pretty simple, we keep it pretty real, and they don't have to be up and doing big presentations. They can do a bulletin board on their unit. They can do a binder of articles. They can do anything, but they have to do something. That is the expectation and the accountability piece.

Then, there was a list of competencies, but still that was too vague, too general for our nurses. So we said we will help you out here. We will show you what it looks like in the nursing process. So we took those competencies and we put them into the nursing process. We said, part of your assessment should be the family history, pedigree, environmental factors, physical findings, and patients' knowledge and questions. Use that in planning.

Then the interventions would be doing patient education, discussing family risks, discussing preventative measures, testing, treatment, and pharmacogenomics. Nurses are great patient advocates, so discuss some of those ethical, legal, social, cultural issues as well as support services, and then the genetics referral if indicated. Then evaluate the services provided.

So we put it into a format that nurses were used to looking at and they could sooner get their arms around it and say, okay, this is what it looks like. We said we will make it even easier for you. You don't have to do all of these right away. If you do two of the assessment items and two of the intervention items right away, we are happy. That is a good start. So that is what we are working on with our nurses.

Actually, this model is accepted for publication in the American Journal of Nursing. It should be coming out soon.

What are some of the barriers and recommendations. Again, these have been cited already. I'm sure they will be cited by our other speakers. Time for education. Time is valuable, time is limited, time is a challenge. We are saying integrate it into the existing programs. Don't create new programs. There isn't time or resources.

Time to implement and practice. Again, huge challenge. Everybody is busy and racing. There has been more expected with less. But just keep it very simple, relevant, and realistic. Don't expect that everybody is going to do everything because that is not realistic.

Time to evaluate competency. Again, very simple, relevant, and realistic.

Lack of education resources. One of the things that happens is we get invited quite frequently to consult or present at other organizations, and they will say, "You have these marvelous resources," especially our series of recordings. "Can we have those? Can we use those?" The unfortunate part is they were done with private funds. So it is like, I want to share them really badly, but I really have not been at liberty to do that. Occasionally I do. I'm not supposed to. I really want to be able to share the things that we have developed.

What we are developing now is a series of nurses demonstrating exactly how they are using the competencies. It is so incredibly valuable, so I really want to be able to share those. I was invited to speak at a national staff development conference in the summer, and it is like, oh, I would love to be able to share all of those things that we are developing, but I don't have permission to do that.

What we really need is federal funds or non-private funds to develop more relevant resources for all specialty practices. Again, those series of segments that are very relevant to different specialties.

Then we definitely need a centralized location for resources for all nurses to keep that up to date and to identify those gaps and develop things that need to fit into those gaps.

Again, it needs to be very specific. General information just doesn't cut it. It has to be very specific to nurses and very specific to specialties for nurses to really buy it and really embrace it and engage in it.

We definitely believe that genetics and genomics is the future, not only for our organization but for all the different professions in health care. It has just been an exciting opportunity to be working with genetics and genomics. I have had other assignments concurrently, but I always say genetics and genomics is my passion. That is where I really am excited and really see the future going. I really enjoy moving that initiative forward.

Any questions? Yes, sir.

DR. LICINO: I think that the work you are doing is very interesting and very valuable. I really commend you for approaching genetics in such a direct way with patients. I think it really needs to be done.

I was a little concerned about the video with the bipolar patient. I'm a psychiatrist myself. I think that there is a huge mismatch between the patient's expectations and what can be delivered in this lifetime. With psychiatric disorders specifically, let's say for schizophrenia or for bipolar, many people believe the problem has to do with neurodevelopment. Your neurons migrated in a way that they shouldn't have migrated.

There is no genetic intervention they are going to do in the future that is going to make your neurons migrate back the way they should have been so that your brain is going to be rewired based on genetics.

So this man has these very unrealistic expectations. Is it part of your educational effort to address those? Some people don't understand genetics and you have to say, "Here is your gene. You have a contribution to that." But other people are like way over. It is almost like a delusion that he is having that he is going to have three injections and then he is going to be fine.

How do you address that kind of unrealistic expectation side of the equation?

MS. PESTKA: Thank you. I totally agree his expectations are unrealistic. That is part of the education, to frame it in realistic terms. Even the pharmacogenomics doesn't give all the answers. People arrive and they think maybe this will tell exactly which medication and exactly what dose, and it is certainly not sophisticated to that point.

So your point is extremely well taken and we do provide education to inform him. It is hope for him, so it is something. He is excited. But he is unrealistic, so you are right. The education does need to occur.

Yes.

MS. ASPINALL: This is a very impressive program. Have you put together a measurement tool to look at the progress and to monitor how much of the information is being internalized by nurses who have been on the job, by new nurses coming in?

MS. PESTKA: We did one study, and it was a psychiatric nursing conference. My specialty is psychiatry. We did a pre-conference survey, end-of-the-day survey, and a three-month follow-up survey. The nurses did have significant learning. They did retain it and they did apply it. That was the only study that we have done per se.

What we are focusing on now, what we think the real bottom line is, is the competency end of it. Are they going to be able to use it.

MS. ASPINALL: Are you measuring that going forward?

MS. PESTKA: We did one pilot study this last year related to hematology and oncology nurses and whether they were actually applying the competencies. Our results were interesting but not profound, and part of it was in our methods. So what we are working to do is to replicate that study in other settings and say, if we provide adequate education, can we measure that nurses are doing this.

The measurement would be per self-report, that they could report a patient situation where they actually used the competencies and made a difference. Thank you.

DR. TELFAIR: Thank you for the presentation. It is very interesting. Actually, the last question is always one that is very critically important because your model is Diffusion of Innovations. As you know, one of the elements of Diffusion of Innovation of course is looking at it from both the short term and long term. So the question has to do with not only is it early and late adopters of the information itself but then that.

I have two questions. One is, on that note, how are you looking at the immediate education as it relates to the professional relationships you have with other professions in terms of disseminating the information, using the information, that sort of thing. Other professions being not only physicians and physician assistants but people like social workers, psychologists, lay health professionals, and, particularly since you were talking about genetics at the community level, the persons who do things like single-gene counseling and that sort of thing.

Secondly, I am concerned about the same question that came up earlier with the physician. How do you deal with the differential amount of information being provided in the different settings, even at the different levels at which nursing is? The different levels are two-year, four-year, that sort of deal.

That may be a question to come back to later if you want to. Actually, I would ask the same question of the two provider presentations earlier as well. Are we having a forum on that? Okay. So I would like to save that question and then ask the other physicians later. I just wondered if you can think about it, and the other two persons can think about it, and I will come back to it.

MS. PESTKA: Thank you. What we are looking for in our setting is champions. That is what this whole program, the National Nursing Program, is looking for: champions in practice, champions in academics, and then really focusing and spotlighting those champions.

In our own local setting we are looking for champions, and we do have one champion multidisciplinary group. We have numerous ones, but we have one where we did a video segment of different disciplines and exactly what their role is. They actually did a model, and they have six or eight different individuals caring for these high-risk prenatal individuals and families.

They defined what is the nurse doing, what is the genetics counselor doing, what is the physician doing, what is the social worker doing. So it was looking at it from a multidisciplinary perspective because, definitely, we do work in teams and we want to make sure that we are complementing instead of competing or overlapping.

So, excellent question. Thank you for asking that.

DR. McGRATH: Beth, thank you for sharing that really interesting initiative at Mayo. Thank you.

[Applause.]

DR. McGRATH: Our next speaker is Melissa Fries with the American Academy

of Medical Genetics.

DR. FRIES: Good morning. I'm Melissa Fries, and I'm going to speak to you from two roles, one as a practicing medical geneticist, and two, from my role as the chair of the Education Committee for the American College of Medical Genetics. In this, I hope to bring out a little bit of the discussion of some of the ongoing issues relative to the education of medical genetics, what practice actually is like for someone in medical genetics, and what are the roles of our professional societies to assist us in this.

The education right now for a medical geneticist is a residency, formerly a fellowship, prior to 1992. At the moment it is a residency program for which there are 48 ACGME programs in medical genetics. It is a two-year program. Some institutions have three-year programs with other requirements. There is a prerequisite of two prior years of some initial residency training such as pediatrics or OB/GYN or internal medicine.

There are also five-year combined pediatrics and genetics programs and internal medicine and genetics programs. There are several fellowship programs, such as that for maternal/fetal medicine and genetics, which is a four-year fellowship, and a molecular-genetic pathology program, which is a one-year program. All of these residencies are ones that can be entered into after you leave medical school.

The medical genetics residencies [have] 196 positions. I went through the whole listing and counted them. Forty-seven percent of them are filled. That is a staggeringly low fill rate. For anyone who works in ACGME, you recognize that in most places, like family practice, 93 percent of those positions are filled. We are looking at some of our programs where there are four positions, of which there is one fellow or one resident. The fear of all of the programs is that maybe this year we won't have any applicants.

Clearly, this is a huge issue. The positions are there, the programs are in place, but they are not being picked up by medical students.

This has been the subject of a considerable amount of research. The Banbury Summit report in 2005 -- actually, the summit took place in 2004 -- included representatives of many of these major genetic professional organizations both from the United States and Canada. Canada is a key player in much of this and actually has many of their own medical genetics residencies.

The fact that the programs have not filled means that there is a declining number of people that are going to be available to meet the oncoming role. Many people in genetics look like me. This is not artificial hair. So we need new people to come into our program in order to actually take our places. The whole job in medicine is to train your replacement. That is not going to happen if we don't increase this.

The Banbury Summit recommendations at that time worked very hard to try and reach some consensus on increasing recruitment. They wanted to position medical genetics as ideal for students who were seeking an academic career. Clearly, genetics has to go hand in hand with ongoing research and ongoing practice development, so if you have someone who is really interested in that, that is the person that maybe should consider medical genetics.

Their goal also was to seek NIH funding for centers of excellence and to enhance the visibility of medical genetics by working directly with resident and medical student advisory groups.

There was also recognition of the need to strengthen some of the core training

issues as well as partner with other medical specialties and work with some of these joint specialty fellowships. That is where the LFM Genetics Fellowship actually came from.

These have not been idly dismissed, and the continuing process recognizes through Banbury II, which was just recently held -- the report is in press -- that we have to redefine some of our training. We actually plan a Banbury III.

I think one of the things that you want to bring up with the recognition for this is that medical geneticists have a unique role in caring for someone who has a genetic condition. They may actually be the ideal person to be "the medical home" for that person. As you move from your diagnosis as an infant into your role as a teenager, into your role as an adult, you may find that the medical geneticist is one of the key people to actually be able to do that. That requires a change in the training. If you are trained largely to think of things pediatrically, you are not going to be able to actually follow through then into their role as an adult.

Speaking in my own practice, I come from a circumstance where I spent 26 years in the Air Force, where I was an OB/GYN and a practicing geneticist. Most of my role there was in prenatal diagnosis and genetic consultation.

I then moved to a practice in inner city D.C. at a largely academic but very busy inner city hospital. I'm the only geneticist. I'm called the director of genetics and fetal medicine. I am in charge of myself.

[Laughter.]

DR. FRIES: Which is really helpful. But this gives you a sense that even for those places that have medical geneticists, they are rare birds.

What I find is that my practice is guided by these three Rs. The first one is recognition by other professionals. You would think there are so few of us that maybe the hospital, by just mere fact of hiring me, would make some effort to market me. It hasn't happened yet. So marketing and advertising what this person does is, I think, a key function and one of the things that could be done very well.

I was working on an initiative in my own institution for this intranet curbside consultation. If you have that in your own intranet system, just as Beth was commenting, you can then click on that and then, one of these hours when I'm not doing other stuff, I will try to get back to you and tell you if it is one of those that we could work on.

The lack of recognition then leads to my second R, which is the referral process. Many providers, even in fields where you know that there are genetic issues, don't feel a need to refer. They often feel that they can handle those genetic services just as well and it is probably not going to be a beneficial role for the patient. So the referrals are struggles.

And then the final R. I know we have heard a lot about reimbursement, but I have to tell you, the issue of reimbursement in my own institution has created a two-tiered system of genetics because most of the patients are Medicaid. Medicaid patients cannot get a genetic test paid for that is out of state. So if I want to get one of my Medicaid patients tested say for BRCA1 or 2, she either has to pay for it herself or she has to go through Myriad's need program, or hopefully will be able to pay for it through a grant.

But what happens is that then you get an insurance quandary so that you are tiered for that, and patients may not even be referred because the issue of the reimbursement is such a problematic one.

One of the other issues that you find in practicing in a diversity of medical

settings is that there is this ongoing pattern for use of family history. Family history we want to incorporate into all medical fields, but even for experienced genetic providers who do medical family history-taking or genetic family history-taking every day, across demographics this is very difficult. The socioeconomic issues and the cultural issues are enormous.

Immigrant populations may have minimal information as well as problems with literacy and language. We are not just talking about Spanish. I spend a third of my time speaking Spanish to my patients. I have gotten a lot better. But at the same time, my patients may not read Spanish. How are they going to deal with that. How do we deal with those literacy issues.

They may lack information on their parentage. Things that we think, okay, mother, father, you are going to know this, that is not always the case.

The medical issues in the family may not either be discussed, because they may be taboos, or they may be in some ways certainly unknown. So I think that one of our key areas in this is to focus on the development of tools and education across these demographics of language, culture, and literacy. This has to be a key point for integrating this truly into practice.

I cribbed this from Dr. Charles Epstein's article from 2005 about medical genetics, but this basically shows the pedigree of the institutions that are here to help us as professional organizations. You can see that the parent organization, the American Society of Human Genetics, has been around for over 50 years and has given birth to quite a very few children. Actually, they were born quite late in life, although they seem to be still fairly robust.

In 1991, we had the American College of Medical Genetics. In 1980, we had the American Board of Medical Genetics. American Board married late and now is part of the American Board of Medical Specialties, and we have given rise to our sole child right here, the RRC for Genetics in the American Group ACGME. So this is our group of professional medical organizations that are associated with the practice of genetics.

They have all different roles. I would say that one of the key overwhelming roles of all of them is the recognition of the importance of education. I think perhaps in no other field does education play such a huge role. Any genetics interview, any genetics time, is education. It is education for the patient, but in many ways you are educating whoever is around you: your nurse, your genetic counselor, your high school student who is watching over your shoulder to model behaviors.

Geneticists educate as part of their life's blood. So all of our professional organizations recognize this. The American College of Medical Genetics has as its goal the education, resources, and voice for the medical genetics profession, to make genetics services available to and improve the health of the public in general.

The American Society of Human Genetics is a very, very broad organization, but the Information and Education Committee's goal is to identify and promote educational opportunities to increase the understanding of human genetics in North America. They have several specific focuses for that.

Our American Board of Medical Genetics is our certifying organization. This is how to keep us current and how to maintain our certification both for ourselves and for our training programs.

ACMG has been a powerhouse in working these educational initiatives. We discussed the importance of making genetics part of board examinations. The American College

of Medical Genetics has sent several taskforces, at least four times in the past 12 years, in conjunction with the American Society of Human Genetics and the Professors of Human Genetics, to review the questions on USMLE Part 1, 2, and 3.

In looking at them, we found that there are definitely improvements, and this is from Darrell Waggoner's presentation which you just commented on. My [apologies] to Darrell Waggonner because I misspelled things here. I apologize.

But there is definitely an improvement in the incorporation of these basic science questions. There is an increase in the part 2 and Part 3, but the irony is that very often when they give a clinical scenario, family history is not part of it. The patient is presented. A 57-year-old man presents with chest pain and a cough. You don't know that he has a family history of hypercholesterolemia and that he has a family history of diabetes. Any of those other family histories are just not given. So clearly, still, efforts need to go on.

There is definitely some improvement. We hope that this will be expanded with use of virtual patients and clinical scenarios.

ACMGE is also working on the exposures of general clinical genetics with video teleconferences. I would invite you to go to this website, Neurofibromatosis, UnderstandingNF1.org, where there is Bruce Korf interviewing someone who has neurofibromatosis. The intent is to develop about 10 of these video telecasts so that people can have an idea of what geneticists actually do and provide models for those who want to actually look at what this role would be. What is your job going to be like.

ACMGE is also involved in looking at the residency curriculums. We talked about this. This is a collaborative effort to promote the idea of our medical geneticists as the medical home for lifetime care for some of those people with congenital anomalies and genetic conditions.

Another key point we have also addressed is the idea of expanding point-of-care reference systems. ACMGE has developed things called ACT sheets. The ACT sheets are in response to the expanded newborn screening programs where there are at least 29 different things tested for, of which they may come back with positive findings leading many people in the field to both weep, tear their hair, and panic. The ACT sheets are very accessible and very knowledgeable.

One of the interesting and very important issues right now is to incorporate these directly into our electronic medical record systems so that there is an automatic pop-up for them. These protocols are going to be similar models for other activities, such as those on cystic fibrosis, Fragile X, hemoglobinopathies, and then could also be involved with how we work this patient up. What would be ways that could be guided for development of studies on mental retardation or developmental delay, and how do you work towards this. This is actions of the American College.

American Society of Human Genetics has focused on a different aspect, not so much the medical but on the overall understanding of genetics in general. Charlie Epstein, in his presidential address, emphasized that one of the key things that the public has is a fear of genetics, a fear that genetics is going to somehow make a superhuman person, someone will be basically made and we will no longer be able to have our wonderful diversity, that there will be priorities of what is good and what is bad. This is a chronic fear of the public.

The American Society of Human Genetics has worked on this in their expansion

of programs K through 12, and actually K through 16. There are developments of programs. There is a program called GenEdNet.org, a little bit hard to say but a very worthwhile program. There is a database of genetic standards for education at the K through 12 level across all states. So if you want to know how to teach genetics to a kid in kindergarten, you go to GenEdNet.org and you can find out for your state what they will do. It is a wonderful program for all of that.

There are numerous other initiatives that have been involved: DNA Day, essay contests. There is a program called Genetics Education and Outreach, and there is a grant right now that the American Society of Human Genetics has of pairing a geneticist with an educator for training and education.

Clearly, working through the schools is the way to incorporate basic genetic knowledge because your kid is going to be the one taking that piece of paper back to the family and saying, "I want to know what grandma had and what grandpa had." The child is going to be the mover in that particular field.

ASHG also runs a wonderful undergraduate workshop with every meeting that they have, where they are going to be incorporating students and undergraduate educators as well as high school educators from the community in which their meetings are held. There is a key emphasis on education as part of your role as a geneticist.

Finally, the American Board of Medical Genetics is very active in our maintenance of certification, which all of us must meet as physicians. One of the points I would like to emphasize is in our Part 4, where we want to improve our practice models. We will write genetics modules for that that can be translated to other specialties for their utilization in that particular area of training.

I would like to conclude with some of my own thoughts about recommendations for this. There is clearly an improving trend in some points of medical genetics, but it is not enough. We need research on why people make their choices for residencies. A lot of it, I believe, is related to the fact that they don't know anything about what medical genetics does or is.

I would also like to suggest that maybe there is some role for a sponsorship program. If we recognize that medical genetics is a key profession that needs providers, maybe there is a role for a sponsorship program much like we sponsor those who serve in inner cities or rural communities after their training.

I think we all have to recognize that if we are at an academic center our practice patterns are going to reflect some of our initial specialty training. Judith will address a little bit more of that. But I think that we need to recognize within our institutions some of that. We also need to work with this issue of reimbursement.

Finally, I think that all of our professional societies work for education, but it is a work in progress. Education is not enough, as we have spoken before. You have to put it in practice, and you have to develop a competency to reflect that you actually can use that.

Thank you. I welcome any questions.

[Applause.]

DR. McGRATH: Thank you, Melissa. We have five minutes for questions until break. I know we are going to get back to some of the questions that you raised at the half hour,

so we will start with Marc.

DR. KIRCHNER: Thank you for a very nice presentation. I'm sorry to hear that so many of the training programs are open. I guess the obvious question that that prompts is what are the job opportunities? You are the only geneticist at a fairly large center. Obviously there is not room for more or people have not hired one. I think that would drive, of course, the educational needs. Could you expand a little bit on that in terms of where are the job opportunities and is that something that can improve?

DR. FRIES: I believe that Judith is going to speak on workforce issues, and so it may be something that she would be able to defer that. I will let you comment in just a second, Judith.

I think the key point is that there are usually spaces available where people have the creativity to recognize the need. In genetics, we also have to market ourselves. It is not like they are limited, it is simply something that many people have not even recognized that there is a need for that.

Judith, do you want to speak to that?

MS. BENKENDORF: I don't have the statistics on unfilled positions, but I'm sure that is something we can get. I do want to comment about the residency slots. The 196 slots that are approved are not all funded slots. We do have, I think, 75 to 80 funded slots, and we can't even fill those. But what happens in hospitals is they say, "Fine. You want a genetics slot. We only can fund X residency slots, so we will take that funding away from pediatrics or surgery or another department," and that usually doesn't go over too well.

DR. FRIES: Yes.

DR. WILLIAMS: I think the other point that is missing here, and again I refer to your fifth slide, which is the colleges' positioning of medical genetics as ideal for students seeking an academic career.

Speaking as someone who is not an academic and has enjoyed my private genetics practice, there are a number of us out there. I think we may be the only specialty that has positioned ourselves as an academic career. I think most other specialties say you can do academics but there are plenty of opportunities in the private sector as well.

This was a point that I made to the organizers of Banbury I in the sense that there was no representation from the private sector there. I think that was addressed to some degree in Banbury II, but I think we really have to engage with the private sector because there is a lot more money, there are a lot more jobs, and there is a lot more need, quite honestly, in those settings. Then, as Peter pointed out, if those jobs become available, that will to some degree drive interest.

But as we have talked about in the context of healthcare systems and payers and that, once we can really consistently create the recognition that this is really needed out there for the patients that people are taking care of, then I think that will happen. Groups like Northern California Kaiser that have really gotten this message and taken it forward for 30 years are very successful examples of that model.

DR. FRIES: I would agree with that. I would also comment that part of that was a brochure that we developed through the American College on Medical Genetics as a residency program emphasizing, with your comments, that it was an expanding role in the private sector.

Yes, sir.

DR. KHOURY: I would like to come back to your three Rs, Recognition, Referral, and Reimbursement, in light of the unfilled residency slots and in the light of the fact that there is really no marketing of genetics. Here we are in the 21st century and people are selling a whole genome on the street right now as we speak.

There is a major disconnect between the basic science and the marketing of that for the world of practice. This is, to me, a lack of translation in a major way. I wanted to get your thoughts on the idea that maybe we are only selling a very small part of what could be sold.

I'm a member of the College and all these societies, and we are selling genetic services. We are selling information to help families with genetic conditions. I'm not saying we should get away from that because there are a large fraction of individuals and families in the U.S. and everywhere that need those kinds of services. Individually conditions are rare. That is why for average practitioners the a priori probability of finding any particular syndrome or disease is fairly small.

But no one has positioned the medical genetics community so far and the various aspects of it to be the information translators for what genetic information means to the average person or the average encounter of patients with their physicians, whether it is drugs or not.

I want to come back to the concept of evidence-based medicine because that is what the average practitioners need, guidelines and criteria. As long as we in the genetics community keep selling the genetic services model, which applies to a fraction of genetic information, maybe we are missing a larger market out there, the market that allows us to interpret what decoded genetics is trying to do, and the genome profiles and all these combinations of genetic risk factors and pharmacogenomics.

I wanted to get your feel of what that means in a 21st century practice of genomic medicine versus the practice of medical genetics, which is what we all got trained in.

DR. FRIES: Clearly, our practice is going to be an evolution. For many of us, the practice initially started as a spinoff, perhaps of a subspecialty of what we did before: pediatrics, OB/GYN, internal medicine. But clearly, if we are going to make this a 21st century model, you have to work beyond that and you have to expand it.

I think part of that has to go with some of the issues on how insurers are going to support some of the evidence-based medicine as well. Insurers clearly are going to be looking and saying this is a justifiable point to do.

For example, I have one insurer that will not reimburse me as a complex consultation for my genetics consultation. They will only reimburse me for a moderate consultation. It depends. What do you call moderate, what do you call complex. If it takes me 90 minutes to get this information out, that is pretty complex. So some of it has to be driven.

As we said, it is the ecology of our entire changing time. You can't just change one thing, you have to change the whole process. But some of it is you have to make genetics sexy. Genetics is sexy. We deal with sex all the time, so you have to make it that way.

DR. TUCKSON: Please, my god.

[Laughter.]

DR. TUCKSON: Some of us aren't ready for this.

We are going to do this. We are going to do three quickies, real quick, and then we are going to take the break. So, no fooling around. One, two, three.

DR. WILLIAMS: This is just a follow-up to what Muin said. I think what we

need to do is think systematically and look at what healthcare systems need and be the knowledge resource. Some of it is going to be direct hands-on patient care, but some of it is going to be being the intelligent filter of all the information that is coming forward. The economic argument that can be made to systems is that if you don't do this right, you are going to be spending money and valuable resources on things that really don't add value.

So we need to be thinking from a more systematic perspective, and that is something that payers want, too. But we have not been willing as a society in general to engage with those types of things. We have tended to remain in that economic model.

DR. TUCKSON: Andrea.

DR. FERREIRA-GONZALEZ: I was very pleased to see some of the improvements for the USMLE step one questions on the board specific for the genetics. My concern comes, and maybe we can discuss this later at the roundtable, for some movement that is going on through the USMLE to reorganize the curriculum for medical schools to place more emphasis on the clinical sciences and decrease some of the basic science. What is the impact going to be with all this movement down the road.

I don't know if you want to address this now or maybe discuss this later.

DR. TUCKSON: Why don't we tee it up for the discussion. Put it in your notes to make sure we come back to it. Daniel.

MAJ. WATTENDORF: The question I have is regarding this high-complexity visit with a clinical geneticist. In fact, I'm keying in on what Dr. Kahn said and something else that Dr. Khoury said. I think as personalized medicine moves forward, the clinical geneticist arena will expand beyond the high-complexity rare disease visit and really needs to hit the reimbursement for a predictive evaluation and predictive genetic testing, both testing which we don't get reimbursement for easily with CMS reimbursement right now.

The paradigm needs to shift from high complexity visits to within the 15-minute visit where a predictive evaluation of an individual's risk stratification is part of the clinical practice. I don't see the clinical genetics community really moving into that arena.

DR. TUCKSON: Great. We will get that on the discussion as well. That is an important observation. Lastly, Hunt.

DR. WILLARD: Thank you, Reed. There is something symmetrical about this. I will make the same comment I made in 2003, the first time I spoke, and this is going to be my last word.

[Laughter.]

DR. WILLARD: Muin wants evidence-based medicine, but I think we ought to have some evidence-based education. The evidence says, and it has been saying this for 10 years, that medical genetics is not a growth industry as a medical specialty. The growth industry is in genetics and genomics and genomic medicine. There is great excitement. The consumers are there. All the other medical specialties recognize, to greater or lesser degrees, that they need to figure out how to get genetics and genomics into them.

But the specialty of medical genetics, where we are half-empty on the residency end, clearly that is not a growth industry. There are two competing messages. One is, and I think even in your own words you allude to this, there is the need to take care of the traditional business of medical genetics because, clearly, that is a need. There are genetic disorders, there are genetic syndromes, and those kids who become adults need care and there is a medical home

for those.

But for all the other disorders that we don't want to call genetic disorders, to Joe's point, the medical community and consumers don't believe that those are in the purview of medical genetics. Those are in the purview of all the rest of medicine.

So evidence-based education would, I think, tell us to steer our educational efforts elsewhere and then separate that from the somewhat smaller but equally important task of refilling the boat of medical geneticists. We will need a relatively small number, but we need them to deal with "genetic syndromes" going forward.

DR. TUCKSON: Hunt is nothing if not consistent. Thank you for that.

We will stop. At 10:50 Angela will begin. So you have to hustle because Angela will start at 10:50.

[Break.]

DR. TUCKSON: We are in the midst of this discussion. I think we have some very provocative issues out here to grapple with, so these next presentations are going to be key.

I want you all to make sure you are locking down on, again, what are the determinant issues in your mind that would cause you to lean one way or another around creating another subcommittee of the Committee and all the work that it entails. If you were going to do that, what would be the charge to that committee. So keep thinking as you are asking these wonderful questions.

So, are you going to tell us about Angela?

DR. McGRATH: Thank you. This is Angela Trepanier from the American Board of Genetic Counseling. Thanks for coming.

MS. TREPANIER: Hi. Actually, I'm from the National Society of Genetic Counselors. Sorry. There is an American Board of Genetic Counseling, but I'm the president-elect of the National Society of Genetic Counselors. I'm also a program director of genetic counseling graduate programs.

I'm here today to talk to you about genetic counselors' roles in promoting the integration of genetic services into health care. I have framed my comments on the basis of a review of Chapter 6 of the SACGHS Genetic Testing Oversight Report, which was Chapter 5 when I read it the first time, so I was a little confused. But same chapter, different number. Then, also, based on the information that was provided from the 2004 educational resolution.

In brainstorming with colleagues, what we identified were three issues that are really critical to integrating genetics into primary care. The first issue, I think, is that we all need to be on the same page with regard to definitions of commonly used terms like "family history" and "genetic counseling." What do those mean in the different contexts of different healthcare providers.

Second, I just think we all need to be cognizant, which I have already heard, that genomics and genetics and health care is an expanding, moving target. What you are thinking about today in terms of integrating genetics into health care could be completely different than what you are talking about in three years. So I thought it was really timely to revisit the SACGHS 2004 resolution because a lot has changed in the last three years.

Then, finally, I think that there is not a "one size fits all" solution for any health care profession. When we are thinking about solutions for integrating genetic services into care,

we have to think about practice factors that are going to make the solutions that work different for different providers.

That is how I'm going to frame my comments, and then I'm going to talk to you about what we are doing as genetic counselors to try to address some of these issues.

First of all, I wanted to describe what is the scope of genetic counseling services. This is a comprehensive definition which is based on the NSGC's recently developed scope of practice, the American Board of Genetic Counselors' practice-based competencies, which guide training programs, and then NSGC's definition of "genetic counseling."

I'm not going to read through this list, but this is what we consider as genetic counselors as comprehensive genetic counseling. I'm not advocating that this is everything that you have to do for every patient who has some sort of genetic indication. Obviously, that is not the case. As somebody alluded to earlier, this is not in the best interest of all patients for all indications.

But when we are talking about genetic counseling and incorporating it into health care, I think we have to be very clear about what we are asking people to do. If it is this comprehensive list, then perhaps it is time to refer to a genetics professional. But if it is only pieces of this, then perhaps this could be very easily incorporated reasonably into primary care.

I also want to focus on the family history component of genetic counseling services. When you talk about family history, even when you are talking about the three- to four-generation pedigree, that means different things to different people.

In genetic counseling, what that means to us -- and I'm going to read to you from our practice competencies -- is that we can elicit an appropriate and inclusive family history that we can construct the pedigree using the appropriate symbols, that we can structure questions for individual cases and probable diagnosis. That means asking targeted questions based on a deep understanding of the genetics and the natural history and the features of the conditions that we are trying to rule in or rule out.

That we use interview skills to facilitate recall of symptoms that might not be things that people automatically tell you up front, and that we also pursue pertinent history with regard to how the family has been coping with the condition in question.

That is what we are talking about when we are talking about comprehensive family history. Once again, I don't think this is what we always mean when we are talking about incorporating family history into primary care. In some cases, yes, and in those cases, maybe then you need to think about referring to a genetics professional. But in other cases, no. So we need to clearly define what we are asking people to do.

The next point was really what is the role of healthcare professionals without specialty training in genetics. The role is a moving target, as I mentioned. In the Oversight Report, Hayflick's definition was used, and that definition, which is listed on the right side of the screen, I think is still probably appropriate for many genetic scenarios, many of these single-gene conditions. It may be applicable to other scenarios in the future, too; I think that remains to be seen.

But in 2007, obviously there is a need for roles that extend beyond just identification and referral. By sheer virtue of how much genetic information is available and how much can potentially be used, clearly there are not enough genetics healthcare professionals to address all those potential uses. So we have to figure out a way to incorporate genetics into

primary care service just because there is a need to do so.

The gap that has been identified in the Oversight Report and that I have heard from other people, too, is when do you refer to genetics and when do you actually manage in primary care. One of the gaps is just evidence-based medicine, the data that supports when an issue needs to be handled by somebody specifically trained in genetics and when it doesn't.

The third set of factors that I think are really critical in terms of incorporating genetics into primary care are what I call kind of practice factors. Some of those factors have to do with just the disease itself. I think you can develop some practice guidelines based on what you know about the complexities of the disease genetics, the complexities of testing, the complexities of the management, the potential psychosocial impact, the complexity of decision-making involved in dealing with the disease risk or the condition risk, and then the quality of available data, the degree of ambiguity.

There are also other factors that are important in trying to answer this question, and one of them is provider factors. It is whether or not there are competencies that have been developed for providers giving them guidance in terms of what they should be able to do and potentially not be able to do.

It is adherence to those guidelines and those competencies. As you begin to incorporate genetics into primary care and into curricula for medical students, there are going to be some people who have been out in the field who have not had the benefit of getting that information. So there are going to be those differences in the provider's ability to provide genetic services.

Then there is also practice setting and time constraints. No matter how good your guidelines might be and how clear it might be what primary care physicians should do, there are going to be some practice settings that don't lend themselves to incorporating some of this information into the practice. Then there is also the interest of the provider.

The third factor is really the availability of genetic services in the community and the community's willingness to utilize these services -- in some communities there may be a hesitancy to seek genetic services -- the community's access to information about genetic risk, and insurance reimbursement.

All of those factors also need to be taken into consideration when you are figuring out the best model for integrating genetics into primary care.

I wanted to give you an example. This is a case of cystic fibrosis genetic testing, so carrier screening. There really are in this case two indications for CF carrier screening. There is just population screening. In the Oversight Report, it gave some history about when population screening was recommended for everybody who was pregnant or planning a pregnancy. Then there is screening for cystic fibrosis carrier status related to family history.

The purpose of the slide is to identify where the roles overlap when this is provided in primary care in terms of population screening and when there is some difference in what the roles might be when it is because of a family history.

In both cases, you want to identify and introduce the risk. You want to contract. What does the patient want to know, what information do they want to seek. In the population screening, you need to take a family history. You want to make sure there is nobody with cystic fibrosis in the family history. But it is probably more limited.

On the family history side, you want to take that family history, which is more

comprehensive, and you are going to want to know what the family's experience was with cystic fibrosis.

You are going to do limited risk assessment on the population screening side. It is going to be more detailed because you have a family history. You are going to do limited education on the population screening side, more extensive potentially on the family history side.

Both are going to require informed consent. More likely, psychosocial counseling will be required if somebody has a family history, and then you are both going to do follow-up.

Where they differ in terms of genetic testing, even though it is the same genetic test potentially that is going to be involved, because of the different types of mutations that can occur when somebody has cystic fibrosis, you are going to need some genetics expertise to tease out what is the best test of the different types of CF carrier tests that are available for this family.

What is required for primary care providers to effectively provide genetic counseling for cystic fibrosis when it is population carrier screening is really to know to whom to offer the test and how to take the family history, basic information about the symptoms, patients' baseline risk, how to accurately interpret test results, how to refer for genetic services when needed, and then the importance of complying with laboratories' requests for patient information, which plays into interpretation of the results.

Then you also need laboratories that provide interpretable test results that people can read and use and that have professionals available to answer those questions, and then accessible educational resources. Genetic counselors can help with all of these factors.

Because we believe that genetic counselors are really integral in not only providing genetic services but also in advocating about genetic services, we have developed this kind of two-pronged approach to integrating genetics into primary care.

The first prong is really training more genetic counselors and making sure that genetic counselors are adequately prepared for changes in genetic medicine. The second approach is to plan educational programs, conduct presentations for practicing professionals, and everything related to educating healthcare providers and the public about how to integrate services into their practice.

Then, just a little bit of data. This shows the increase in the work force of genetic counselors, and this is people entering into genetic counseling graduate programs by year. There is a little dip, but that is related to the fact that this data is contributed voluntarily through the American Genetic Counseling Program Directors Association.

Basically, the bottom line is that there has been a slow increase in the number of trainees entering in programs, up to 205 in 2007, and the number of programs has increased from 18 to 31 since the inception of the American Board of Genetic Counseling in 1993. So there has been progress in that respect.

The number of certified genetic counselors has increased from 495 to 2,437 since 1993. Since the 2004 educational resolution, almost 1,100 have been certified.

ABGC, which is the certification board, had its first cycle of recertification in 2006, and 316 people recertified and an additional 122 voluntarily recertified. Now the certification exam is on a two-year cycle, so we are definitely making improvements in getting genetic counselors certified.

We have been working towards promoting cultural diversity in the profession.

There was a program directors retreat that was held with midwest program directors to identify some of the factors involved in why there is not more diversity in the genetic counseling profession. What that resulted in is an improved understanding of what the barriers were and some action points. One of the action points was to develop this brochure that you see on the right-hand side.

Other things in terms of improving our education and training. The American Board of Genetic Counseling is conducting a practice analysis in 2008, and that will help them validate the certification exam and make it an even stronger exam.

State licensure. We have had seven states that have passed licensure bills total. We have five that have introduced bills, and 13 have begun the process. So we are working towards more professional recognition and protecting the public.

Finally, the NSGC is pursuing federal recognition of genetic counselors by drafting legislation that, if passed, would amend the Social Security Act so that CMS recognizes us as healthcare professionals.

I would like to also talk briefly about how we are educating other people. This is data from our Professional Status Survey that show what genetic counselors are doing to educate others. I won't read through all these numbers, but the bottom line is that a majority of genetic counselors are involved in educating all other types of healthcare trainees and professionals.

The types of activities they do are very diverse, and they include speaking to lay and community groups, organizing conferences, coordinating or serving on advisory boards, developing genetics curricula, serving on committees, and developing brochures, pamphlets and videos.

Here are some more efforts that you have in your handouts. I just wanted to highlight that some are Web-based, some are in-person educational comprehensive programs. Many are guest lectures. Some are educational material. So, multiple different models.

Then, NSGC in particular has done a number of efforts to train non-genetics healthcare professionals. We have developed a speakers bureau. We have a whole issue of the Journal of Genetic Counseling that is devoted to genetics education. We have representation on key groups that are looking at how to incorporate genetics into health care, and a whole list of other activities.

So the bottom line, I think, is that genetic counselors have the training, the expertise, the motivation, the expertise, and the track record to be key providers of genetics education. For that reason, I think that it is important that not only do we continue our efforts to train other individuals to incorporate genetics into their practice, meeting them where they need to be met. I think that was an important point that Dr. Khan made.

When you are working with primary care professionals, you have to see what they want and what their perspective is and not come to them from your perspective. If you train more genetic counselors, then not only are you increasing the genetics work force but you are potentially also increasing the number of non-genetics professionals who will get training in genetics.

With that in mind, my recommendation for the SACGHS resolution as an additional step is that not only do we need to support training of other healthcare professionals in genetics but we also need to promote and support initiatives to increase the genetics professional work force, its diversity, and cultural competence.

This can be achieved by supporting the development of genetic counseling programs, providing scholarships to support matriculating students who are from underrepresented minority applicants, and supporting initiatives to increase the number of M.D. geneticists, laboratory geneticists, and genetic nurses.

I also have some comments with regard to the Oversight Report that NSGC will submit formally in writing. Because of time constraints, I'm not going to go into those right now. But thank you for your attention.

[Applause.]

DR. McGRATH: We have a minute for questions.

DR. TUCKSON: We are going to get to the discussion, but I need to make sure. First of all, thank you. Terrific as always. You guys are always great.

I have a letter, also, that I think I got from the same group, signed by Katherine Whitcomb [ph.] This issue keeps coming back. Just for the new members, we have been around and around in circles around who it is that ought to certify who is competent to practice genetic counseling and get reimbursed. We have had a number of dueling presentations from well-meaning organizations about whether or not you have to have a master's level training or can you be a nurse who has been in the field for a bunch of years.

You may not be in a position to answer this, and so I want to give you time while you are there maybe to call somebody or something.

[Laughter.]

DR. TUCKSON: The issue is this. I just wondered; we have asked a bunch of times if all of the players could get together and decide on one board of competency certification. So all the competing groups would get together and just work it out without having the strong arm of the law having to come in and smash people's heads together.

This letter doesn't speak to it, either. I'm not sure I know whether anything is going on in that regard.

MS. TREPANIER: I think where we are is, the efforts that we are pursuing to get reimbursement, it is because we are not a recognize healthcare profession. We don't want recognition because we don't want anybody else to be able to provide genetic services. We just want to get paid for it when we provide it.

So as far as from my perspective, there is no issue. Bring them on. The more professionals that have the competencies through their professional organizations through these activities, the better. We just want to get paid for what we do.

DR. TUCKSON: I appreciate that. We have to get to the next one. Just for the discussion I want to make sure everybody understands.

I understand exactly why you would say that. Anybody that has to pay for health care services is basically saying at the end of the day there has to be some kind of clear-cut rule because everybody says that they are qualified to come forward and get paid. You have to have some criteria that says there is a cutoff, who is eligible and who is not.

What we keep asking for at this table is for all of those players who say everybody should get in, one accrediting body that takes care of this whole thing soup to nuts, instead of 15 different accrediting bodies who have dueling accreditation criteria. That is unadministrable.

You answered my question. I just wanted to find out where it was. We can put the rest of that in the discussion.

DR. McGRATH: I'm going to exert my power at the bully pulpit here and ask everyone to save those thoughts for the discussion because I know some of the speakers coming after are going to address that as well. That will be a big topic for the half hour at the end. Thank you.

I would like to introduce Toby Citrin, the next speaker, who is with the Center for Public Health and Community Genomics in Michigan. Thank you for coming.

MR. CITRIN: Thank you. Good morning. I would like to comment very briefly on the significance of using the paradigm of genomics in the public health setting, talk a little about the extent of current and anticipated demand for knowledge in genomics, both as seen from the perspective of the schools and from the perspective of public health practice, to identify some of the barriers that stand in the way, as well as identifying some of the facilitators that are moving the field forward, and then to summarize some of the progress that has been made since your last roundtable, and end with some recommendations.

Your earlier resolution in 2004 makes a point of the distinction between genetics and genomics. This is a very important distinction for public health. The CDC's website of its National Office of Public Health Genomics also defines genomics in a very useful way.

For public health, it is extremely important to be using the genomics paradigm because it fits in quite well with the ecological model of causation of health and disease, which has increasingly been utilized both in the teaching of public health and the practice of public health. Both of the landmark reports on public health by the Institute of Medicine in 2003 make strong recommendations to utilize this ecological model, multiple factors working from the inside out, from the outside in, over the lifetime, and it is very easy to incorporate the genomics framework within the ecological model.

Consistent with that, we are seeing movement both in teaching and in practice from viewing genetics as a separate, almost autonomous field of study and practice into genomics as being worthy of incorporating in all of the fields of public health, both as taught and as practiced.

But let's look at the reality of what is being asked for today both in practice and in academe. When you look at practice, the extent to which genetics or genomics are playing roles in the practice of public health is still quite small. We have traditional newborn screening programs and the expansion of genetic testing within those programs. We have the early evidence of almost experimental utilization of family health history in prevention programs for chronic disease, adding these histories as additional risk factors that are useful in developing prevention programs for chronic disease.

Then we have early signs of the acceptance by some health departments of a role in health education to try to get the public to understand what we mean by genomics and how it relates to their health.

Looking into the future of course, we see an ever growing need for knowledge of genomics in public health practice. A number of commentators speak of the revolution that will take place not just in medicine but in public health which will individualize public health. It almost sounds like an oxymoron. But the increasing knowledge of relative risk on an individual

basis is inevitably going to shift the way that public health designs and implements programs.

What will bring us to that point is the process of translation of research into methods and interventions that are seen as useful in improving population health. Dr. Khoury has been the lead author of a very important article that appeared just a month ago in which he expounds on a four-step process which brings us from gene discovery to health application, from the applications to evidence-based guidelines, from guidelines to practice, and from practice to health impact. It is a paradigm that does not simply apply to clinically provided genetic services. It applies equally to public health interventions.

We are now moving to the academic side. In 2003, in the report on the teaching of public health called "Who Will Keep the Public Healthy?" that had just come out prior to your last roundtable, genomics was identified as one of eight content areas that needed to be taught to everyone going through a school of public health in addition to or as incorporated in the five traditional areas that are the basis for public health education.

So we are seeing evidence that departments of epidemiology have a number of courses in genetic epidemiology. Departments of biostatistics are teaching statistical genetics. Increasing teaching in genetics and its interrelationship to environmental harms and hazards are seen in the teaching of environmental health. Less evident but extremely important is the advent of incorporating genomics in the teaching of public health policy and the teaching of the ethical, legal, and social implications of genomics within departments of health management and policy.

Finally, some of the good news is the significant increase that has been occurring in the incorporation of genomics in the teaching of health behavior and health education. Not so much the influence of genes on behavior but rather the implications of genomics for the way human beings behave in healthier or less healthy manners.

So let's move to the barriers and the facilitators. When one looks at academe, we have the common resistance to any significant changes in curriculum. It has been a constant through a number of the presentations this morning. Insufficient time that people feel already to convey the information that is seen as necessary for public health professionals, and the sense that we are adding yet another overlay on what is already not taught deeply enough.

Certainly, a lack of expertise in most of the faculty to incorporate genomics in their teaching, and to a continuing extent, hopefully lessening over time, non-recognition of the significance of genomics in public health.

[There are] still vestiges of antagonism to the teaching of genomics by those who feel that there is some sort of zero sum gain going on here and the more one talks about genomics and its causation or participation in disease and in health disparities, the less attention one will pay to social and environmental factors.

If you want a good piece of evidence on this antagonism, just take a look at the article that is in your packets by Claudia Chaufan relating, in that case, to the proposed Large Population Study. But one can read in that article this same notion of the less-than-worthwhileness of spending that much time on genomics when we are trying to address serious issues of health disparities.

Let's move to the practice of public health. Same barriers and resistance to change, and more so in the public health setting these days because of ever-tightening budgets, new requirements with respect to preparedness for bioterrorism and communicable diseases.

A good example of how serious this particular barrier is was the talk given by the

outgoing president of the American Public Health Association, Deborah Klein Walker, at the annual meeting of APHA earlier this month, in which she really said that one cannot expect public health departments to take on the new fields of informatics, of genomics, new approaches to disabilities, when public health departments are being starved of resources just to do the most fundamental core functions for which they were formed.

We have continuing evidence of a rather narrow focus on genetics in most health departments, happily not all, where genetics is seen a subset of maternal and child health in the organizational structure of health departments. And, the lack of tools, the lack of evidence-based, off-the-shelf tools coming from genetics research that are seen as useful to public health professionals in addressing major health issues of populations.

Turning to the brighter side, what are the facilitators. Certainly chief among them is the National Office of Public Health Genomics at CDC, which has been the primary place where a continuing array of information of significance to public health and public health practice occurs in workshops and various trainings that emerge from that office. Among its strategies have been the funding of two centers for genomics and public health, one at University of Washington, one that I direct at our university, Michigan, both of which are committed to expanding the knowledge, training, and utilization of genetic tools and information by public health practice.

CDC has also been funding four states -- Michigan, Minnesota, Oregon, and Utah -- to develop comprehensive genetics and genomic programs in order to establish models of how genetics needs to be dealt with comprehensively in a state-level public health department.

A very bright light in the future, just two or three weeks old, the official formation of what is called the Genomics Forum at the American Public Health Association, a group of over 200 people now who are identifying themselves as public health professionals, community people, and academics who are very interested in working together in order to further genomics in the public health framework addressing the goals of public health.

Other facilitators and potential facilitators are the efforts underway to standardize competencies in public health teaching and practice. Several years ago, the CDC launched and funded an effort which ended up with a set of genomic competencies for the public health work force. The website is in your materials.

The Education Committee of the Association of Schools of Public Health has developed a set of competencies for the master's of public health degree. No sanctions or requirements here, but simply advisory to the schools.

Unfortunately, in the enumeration of cross-cutting competencies in this Association of Schools of Public Health effort, the caption is "Public Health Biology" and not genomics. Within public health biology, one of 10 competencies is the competency to explain how genetics and genomics affect disease processes in public health policy and practice. But at least there is a formal adoption by a group representing the schools of the need for genomics education in the schools.

Also of potential significance, and there is controversy on just how significant, but there is a launch coming in August of 2008 of a new examination for a certificate in public health which would apply to people who already have a master's degree but want to have a form of credentialing that is based on a standardized set of competencies in public health.

The examination will incorporate the competencies for the MPH degree that has

come out of the Association of Schools of Public Health, and therefore there will be a genomic component of that certification.

Looking at the number of courses and centers and programs on genomics and genetics in the schools of public health in comparison to your last roundtable, we had a couple of graduate students do a Web search in anticipation of this roundtable, as they did last time.

In 2004, or your meeting in 2003 leading to the 2004 resolution, there were 10 schools of public health that had any kind of genetics programs. Most of them were in the research area. There were only 12 schools of public health that had courses that were identifiably genetics in their topic. Of these, a very small minority of schools had courses in the ethical, legal, and social implications area.

Significant progress since then. All but six of 38 schools of public health that were subject to this Web search have now genetics courses identified as such. Ten have centers focusing on genetics, and seven have actual curriculum tracks that highlight genetics. Our students counted a total of 193 courses identified as genetics or genomics courses in the schools of public health.

A small portion of those, 21, are in the areas of health management, law, ethics, and policy, the bulk of them continuing to be in the departments of epidemiology, biostatistics and, to a growing extent, as I mentioned, health behavior and health education.

Looking at the training of the current work force as distinguished from the future work force, there are hundreds of sources of training materials on genetics and genomics that are available online. To my knowledge, there has not been a comprehensive collation and compilation of these courses. There are several hundred. There are several places where one can go to see an array of these. One of them is the CDC website.

There is an effort underway by two people at the Genetic Alliance, with which our center is about to connect, in order to list and codify the online trainings and provide some sense of level of competency that they address. The Network of Public Health Training Centers has developed a searchable website identifying four courses given by public health training centers that are identified as genomics courses.

Still very little progress in teaching or practice in diversifying the public health work force that incorporates genetics in practice. It is certainly an issue that runs entirely through this field. When one looks in at schools of public health, and it is certainly true of our school, the growing diversity of the student population in schools of public health is not represented in courses or programs in public health.

We have between a quarter and a third of our students who are from underrepresented minorities. In the course I teach, for instance, which is a required course and an elective, out of 29 students I have only one student of color in the entire class. That is very typical and rather sad.

It is quite clear that if public health genomics is going to achieve its potential it is going to have to be more representative of the population that public health sees as most essential to reach with public health interventions.

Finally, recommendations. In addition to trying to find a way out of this problem of achieving diversity, the schools and the health departments need to implement the recommendations on genomics that are in both of the Institute of Medicine reports.

We need to develop a way to gather systematically the data on the extent to which

genomics are included in the teaching of public health, not just labeled as such in course titles but incorporated in a whole variety of public health courses.

We need to achieve a sharing of models of genomic teaching as a way to address the lack of competency in many faculty members to develop their own total courses or to develop their own case studies to incorporate in courses that ought to have a genetic component.

Consideration needs to be given to having genomics identified clearly as a standard for accreditation of schools of public health by the Committee on Education in Public Health, which is the accrediting body.

Public health education not only needs to increase the focus on genetics but also on the ethical, legal, and social implications of genetics. Some of us would like to see the fiscal ability of CDC to broaden the network of genomics in public health centers to a more regional basis in order to serve the needs and the potential of health departments throughout the country.

Thank you.

[Applause.]

DR. McGRATH: Thank you very much. That was a great overview of the public health efforts.

With apologies to everyone, we are going to hold questions again so we can get to the last speakers. I promise we are going to save more than a half hour for general discussions. I apologize for limiting them right now. We are running a little bit late.

The next four discussants who were asked to come were involved in the steering committee. They are taking on the difficult task of limiting their talks to five minutes. They have great perspectives. We will see if that can work.

The first person is David Wilkinson, who is representing the laboratory perspective. He is from the Department of Pathology down in Virginia. Thank you.

DR. WILKINSON: We are going to do it the old-fashioned way, without slides.

First of all, thank you for giving me this opportunity to briefly comment on the state of genetics education amongst clinical laboratory personnel. I would like to start with the most important part of that work force. The core of the people that work in our clinical labs are medical technologists, also referred to as clinical laboratory scientists.

These folks are trained at a minimum at the baccalaureate level. The programs in medical technology or clinical laboratory sciences are accredited by the National Accrediting Agency for Clinical Laboratory Sciences. Their standards do specifically require training in genetics, molecular biology, and molecular diagnostics. They do not specify the exact amount of time or the exact details of the topics. They generally do not have a requirement for any particular or specific course in genetics.

These folks are very well grounded in the basics of genetics. However, we find in our own very sophisticated laboratory, and I'm sure this is true in most places, that these folks, as they come out with their baccalaureate degree in medical technology or clinical lab sciences, are not ready to perform the sophisticated level of testing that we have in a high-complexity laboratory. This would be whether it is in cytogenetics or in molecular diagnostics or other fields related to genetics and genomics. They do require significant on-the-job training.

They are great people to work with, and we can get them up to speed fairly quickly, but they are certainly not ready to go to the bench right out of school.

I would also like to make a few comments about medical student education. Some reference has already been made to the fact that genetics is taught primarily in the first and second year of medical school. In fact, about 77 percent of all genetics course work is done in the first year of medical school. That reference was made earlier to the Thurston paper in Academic Medicine, which gives you some good statistics.

There is actually minimal education in genetics in the third and fourth year of medical school. Hopefully, it is increasingly incorporated into the clinical years, but there is not a lot of specific attention to that.

Now, reference has already been made to the United States Medical Licensing Exam, which currently is administered in three steps. The first step deals with the basic sciences. The first two years of traditional medical school are the basic sciences, including genetics. The first step of the exam, which is the uniform approach to licensing in the United States, covers the basic sciences and is given towards the end of the second year of medical school.

Step two, clinical sciences, is given in the fourth year of medical school and focuses mainly on the clinical education that they receive.

There is a move afoot by the USMLE to compress step one and step two into one exam in the fourth year of medical school. Now, depending on who you listen to, the reason is to increase the requirement for students to retain basic science information into their fourth year of medical school as opposed to learn it the first two years and forget it during the next two years.

However, at least at this point, and this probably gets back to the comments made about resistance to change, most basic science departments, of which pathology is one, are concerned that this may have the effect of deemphasizing the emphasis on the basic sciences.

I have seen editorials in big papers, including The Wall Street Journal, asking, "Why do doctors need to know all this basic science? We are clinicians," which I think is baloney because the basic sciences of course are the basis for clinical science.

So I'm concerned about that move. I think this needs to be studied in great detail before a change is made in that paradigm because, at least right now, they know they have to bone up and be ready to deliver the goods on that step one exam.

Let's move on to graduate medical education. Now, the content of genetics in graduate medical education, the residency training programs, varies quite a bit depending on which residency we are talking about. Let me comment specifically on pathology.

The governing bodies for the content of residency training programs are the residency review committees, the RRCs, which operate under the auspices of the Accrediting Committee for Graduate Medical Education. Our RRC, the pathology RRC, does specifically require training in cytogenetics, molecular biology, and molecular diagnostics. Again, it does not specify the exact amount of time or the exact content, but it has these broad subject areas.

There are about 154 or so residency training programs in the United States. The experience that pathology residents get will vary quite a bit. Of course, pathologists are the physicians who specialize in the diagnosis and management of human disease, and we basically run the clinical labs in America. So we do have people coming out, I think, with somewhat variable experiences.

Now, there is also a subspecialty fellowship in molecular genetic pathology. This

fellowship was already referenced by Dr. Fries. It is jointly administered by the American Board of Pathology and the American Board of Medical Genetics.

This is a one-year fellowship devoted entirely to genetics, including exposure to genetics counseling and molecular diagnostics. You can come through that either having your primary certification in genetics or in pathology.

There are relatively few of these accredited programs. Actually, the accreditation requirements are quite stringent. We are fortunate to have one of those at VCU.

The final step in the continuum of medical education is your continuing education, after you have gone through all of your fellowship training and you are out in practice. The College of American Pathologists is very concerned about education and training and aspects of molecular biology and genetics. Within the College, we have a cluster of committees that are focused on pathology and genetics.

Two of these committees are jointly staffed by members of the College of American Pathologists and members of the American College of Medical Genetics, so these are joint ventures. One of these deals was biochemical and molecular genetics. It is called the Biochemical and Molecular Genetics Resource Committee. The other is the Cytogenetics Resource Committee. These work together bringing these two medical specialties together to oversee both the development of new products as well as ongoing education.

The other areas are histocompatibility, which increasingly is now done with the DNA basis of human identity testing. Microbiology, of course. A lot of microbiology is now based on nucleic acid testing. Another committee called the Molecular Oncology Committee deals particularly with the genomics of cancer.

Another aspect of what these committees do is to manage the development and ongoing changes in the College of American Pathologists and Accreditation Checklist. This is what you use to get accredited, and this is one of the routes by which you can get your CLIA accreditation. You can get a certificate of accreditation through the College of American Pathologists Inspection and Accreditation Program.

The checklists are the things that labs have to follow to make sure that they are being compliant with CLIA. Now, the CLIA regulations have governance over all clinical labs and all clinical testing, including genetic testing. So the CLIA regulations form a very strong foundation for ensuring the quality of clinical laboratory testing in all areas, including genetics.

These committees also manage, create, and evaluate proficiency testing programs that deal with the areas within genetic testing, which is a very important aspect of the CLIA program, which mandates proficiency testing.

Finally, these committees generate educational programs which they provide to practitioners in the area.

[Applause.]

DR. McGRATH: Thank you. I appreciate the challenge of telling so much information with so little time, so I appreciate all of you doing this.

Next is Michael Rackover, who is representing physicians assistants. Thank you.

MR. RACKOVER: Thank you so much for being able to present today. Help is on the way. You have heard that before.

I represent the four physician assistant organizations: the American Academy of

Physician Assistants; the Accreditation Review Commission on the Education for the Physician Assistant, which is the accreditation body; the NCCPA, the National Commission of Certification of Physician Assistants, certification and testing; and the Physician Assistant Education Association, which are the educators.

The last year has been a phenomenal year, which we can talk about our organizational model of success and how to integrate genetics into clinical practice. We met at the NIH with the Human Genome Research Institute. All the organizations came to this meeting. It was a top-down model. It had to be the executive directors, it had to be the presidents of the organizations, and the movers and shakers of the individual organizations to help understand the challenge that we have in providing the clinical services in genetics.

I can give you a short breakdown. I'm going to talk about each organization quickly to give you a sense of who we are and what we can do. Currently, there are about 64,000 physician assistants in clinical practice. Our average age is about 43 years. Gender is 38 percent male and 62 percent female. With minorities in practice, 12 percent of the PAs are in minorities. New grads are at 17 percent. In education right now, the classroom is 23 percent minorities.

Our hope over the next 10 years is that by 2010 we will have about 90,000 practitioners. When we look more to the future, we have to be at 115- to 130,000 practitioners by the year 2020.

The American Academy of Physician Assistants basically provides a survey. We know where we are working. We basically are working all over, in any medical specialty that you can see: HMOs, group practices, hospitals. Eighty-four percent of PAs see outpatients. Fifty-two percent of these PAs also see inpatients. One percent of PAs see patients in nursing homes.

As you can see by the various different charts I have up here, our graph is all over. Wherever you are seeing healthcare providers, physician assistants are part of that team.

Now, at our recent annual meeting, we are giving CMEs specifically in providing genetics education. We also were given a grant by HRSA to work with the NCHPEG in genetics in the physician assistance practice. We now are up on this website. This is continuing medical education for graduate physician assistants.

Now, when you talk about the educational model, we currently have about 139 PA programs across the United States. The number of recent graduates. In the year 2006, we were graduating over 4,800 students. We currently have models for competencies. We have written an article about physician assistant clinical competency guidelines for genetics and genomics on the educational front.

We also were able in the last year to do a survey of genetics education and a needs assessment of the majority of physician assistant programs across the country. This survey represented a 75 percent response rate, and we were looking at how to determine how genetics is taught in physician assistant programs. We also wanted to determine what genetics content is covered, and we also needed to assess faculty needs for supporting a genetics curriculum.

This is the conglomerate of the slides. The challenge we have is how do you restructure an existing over-packed curriculum, but we were able to get the information in. We also realized that it is not seen as a priority by our colleagues. That is part of the marketing that

we have been able to do over the last year. You will see shortly when I talk about it, we now have a standard for all physician assistant education that we have to teach molecular and clinical genetics.

Certainly the problem is lack of time. Eighty-one percent of the programs that were surveyed perceive the need to enhance their genetics curriculum. Sixty-two percent of the programs plan to change their approach to teaching genetics in the near future.

Our hope, and my hope, is to position PA educators as leaders in the teaching of genetics in medical education. What I hope to be able to provide with our physician assistant colleagues is how we are going to monitor and report innovations in genetics education, develop a curriculum, resources, and best practices, create faculty development opportunities, develop assessment tools for students and faculty, and develop a database to track genetics activities and outcomes in PA education.

The standards area, and I talked about the Accreditation Review Commission on Education. In September of 2006, we included in B2.02 the instruction on the professional phase of the program must include instruction in the genetic and molecular mechanisms of health and disease. So we are now seeing that medical genetics for patient care is now taught throughout the whole curriculum, not just the didactic phase but in the clinical education phase.

The National Commission on Certification of Physician Assistants is the NCCPA. They were at the table. We are looking at exam content and we are beginning to code new items on the exam with the genetics code when applicable. They are hiring a new exam writer with experience in genomics that will be added in 2008. During this year, they are promoting their board of directors about medical genetics.

To quickly share with you, the AAPA is the profession, which takes care of the whole organization. We have a major commitment to provide medical genetics education to our graduates. The Physician Assistant Education Association is the educators. We are all now working together to make sure that we have a methodology to train the trainers, get more continuing education for educators in genetics, and we have a commitment to make sure that it is seen in the classroom and it is seen in the clinical education component.

The NCCPA, the certification model, and the accreditation areas are all working together, and we consider this to be an organizational model of success.

I thank you for the five minutes of time. Thank you.

[Applause.]

DR. McGRATH: That was perfect. Thank you.

The next speaker is Ann Cashion, who is representing ISONG.

DR. CASHION: Thank you for asking me here today. I want to give credit to Cindy Prows from the Cincinnati Children's Hospital for these slides. A longer version of this presentation was originally presented earlier this year at a consensus panel for nurses in genetic education.

How do nurses obtain genetic training. There are many avenues to obtain the various levels of genetic knowledge. However, there are about 3 million nurses to update and keep updated, and that is one of the areas that Beth Pestka has spoken about earlier.

Right now, in academic programs, we have less than 10 formal M.S.N. and pre- and post-doctoral programs. Two of the key areas that we have, or the significant players that

have helped build our genomic capacity for nurses are the NINR NIH Summer Genetic Institute, which has had 121 graduates, and the Genetic Education Program for Nurses, that has been conducted through the Cincinnati Children's Hospital. Both of these have provided genetically, genomically trained nurses that have gone back to their individual institutions throughout the U.S. and have tried to incorporate genomics into their teaching models there.

We also have continuing education opportunities, and those were conducted through our professional organizations. Primarily, you will see that through the International Society of Nurses and Genetics, the Oncology Nursing Society, and our women's health and pediatric and developmental organizations.

There are many gaps that we have identified that need to be filled for us to continue this growing momentum amongst schools of nursing to incorporate more genetic and genomic content. Basically, we need to look at the levels of genetics and genomics education needed by faculty, and how do we need to make more existing and future opportunities accessible to more faculty.

Currently, the International Society of Nurses and Genetics has a Genetic and Genomics Nursing Scope and Standards of Practice. This particular document, that is published through the American Nurses Association, has actually helped us identify the different levels of genetic nurses that are out there and what competencies and scope of practice they have.

Another resource that has been provided over the last couple of years is the Essential Nursing Competencies and Curricular Guidelines for Genetics and Genomics. This helps us address what we think practicing nurses should know, so this is all practicing nurses. These are really genetic nurses. So we have that dichotomy going on in our own organization in how we think different levels and different skill sets are needed for the multitude of nurses in practice.

We also look at instructional resources, which are existing CEs, and how can we make them adapt instructional needs of faculty. How do we decrease the burden of adding yet more content to a dense curriculum but still allow for academic freedom and creativity. How do we help clinical faculty structure this content in their clinical settings.

Two big issues for us are that once instructional resources are created, how are they maintained and updated, and how will peer review be a part of this process.

What can we do to influence the use of interdisciplinary courses. This is how we see sharing of our resources being applied. What can we do to influence the use of genetic nursing courses shared among universities without increasing the cost to students. The Southern Research Educational Board has a model for this. We are at this point trying to institute a molecular genomics course that has some clinical content in it as well. It would be actually taught online at one of our universities and then students from other universities could register for that course. So we are looking at shared resources as well.

Thank you very much.

[Applause.]

DR. McGRATH: Ann, thank you.

Our final speaker is Judith Benkendorf with the American College of Medical Genetics. She is going to offer a perspective on the workforce issues. Thank you.

MS. BENKENDORF: Great. Good morning, and thank you. It is a pleasure to

be here. Since I am the last speaker and you have been sitting for a long time, I thought I would give you my bottom line first. This is a talk where I'm going to tell you what you already know: the genetics work force is very small, unequally distributed geographically, and not representative of the broad diversity in the U.S. population.

I think this is especially striking in light of the latest announcements in the last couple of weeks from the consumer genomics service organizations, or as they call their new product, consumer genomics services, by 23 MedicoGenetics and Navigenics.

So, what does our genetics work force look like. I will just tell you that these data come from the American Board of Medical Genetics, the American Board of Genetic Counseling, and also from our workforce study. Both of the publications from that study are in your packets.

There are approximately 4,700 individuals who hold certificates in a medical genetics profession. About half of them are genetic counselors, and the other half are M.D.s and Ph.D.s certified by the American Board of Medical Genetics. Remember not all these individuals are still alive, and not all of them live in the United States and work in the United States, so we are probably a little bit smaller than these numbers.

We are not very diverse. The M.D.s and Ph.D.s are a little bit better. Thirteen percent identify with minority populations, the predominant group being Asian.

As Angela mentioned, the genetic counselors are the fastest growing cohort. We are about half of the work force, but still only about 6 percent are men, 9 percent representing with minority communities. Again, here is the African American communities.

If you look at the U.S. population of about 103 million people, that is one genetic counselor for 127,000 U.S. population. Sitting here in the District of Columbia, I'm told there is one lawyer for every 17 of us. So we are slightly outnumbered.

What do the M.D. geneticists look like. We are less than 0.02 of all the physicians in the United States. We estimate there are about 1,100 active clinical genetics physicians in the United States. In a recent survey done through the American Board of Medical Genetics by the College, we learned that these individuals spend only 45 percent of their time seeing genetic patients. So we have to divide that number out. So we have one full-time equivalent for every about 560,000 people, or 1.8 clinical geneticists per million population.

We get a lot of phone calls. "How many geneticists should there be?" The Royal College of Physicians estimates about one FTE for each 250,000 people as the idea. Again, based on the U.S. population as the Census Bureau posted it in July, we need 1,200 full-time equivalent medical geneticists, but we are not quite halfway there.

This is just a graph of certification trends. Because the board cycle is now compressed to two years, I estimated what this would be if it was a three-year cycle. Maybe it was a little bit generous, but it shows you the trends. It is the genetic counselors that are going up the fastest. I think the clinical geneticists have a little bit of increase, and molecular geneticists, but the rest of it is pretty flat and we are no longer offering the Ph.D. exam after this cycle.

So the medical genetics work force situation is critical. I put that in red. We can't emphasize it enough. The services work force is not expected to meet patient care demand within the next few years. There is a serious mismatch between the explosion of knowledge and the work force size. Young physicians are not entering our field. You can hear this cry. I go to

a number of AAMC-related work force meetings, and they are all saying young physicians are not entering our field. This is not going to help by the fact that there is an emerging national physician shortage coming down the pike.

Many states, at least 17 according to the Work Force Study, have been identified as having an inadequate number of geneticists to meet the demand right now, and the metabolic geneticists are in the most critical need.

The issues of how geneticists work is also a factor. Melissa Fries mentioned this, and I will mention it again from the Banbury II conference, and we are going to address it also in Banbury III. Geneticists have to take care of patients across the life span, from womb to tomb. No more of this saying "I only take patients with this age and this disease." As Bruce Korf says, you are not a neurology unit that says if you have epilepsy go to the medical center across the street. We can no longer afford to do that.

A picture is worth a thousand words. A researcher from Harvard just called me last week and wanted the zip code of all of the active clinical geneticists in the ACMG database. This is only 509 people, but the lighter the state, the fewer the geneticists. You see some states with gray. Idaho, West Virginia, Wyoming, and Alaska have no M.D. geneticists. Obviously, the darker blue have a large population and also more geneticists.

There is a bit of misnomer. Maryland always comes up very high, but remember our clinical genetics friends at the NIH are not doing clinical practice, so that is a bit skewed.

So, what is the status of our metabolic geneticists. On one hand, we are expanding newborn screening and the other Secretary's Advisory Committee is even entertaining the idea of putting more diseases in the panel. Ten thousand new affected individuals who are going to need lifelong chronic disease management and treatment are coming into the system every year. We counted them yesterday; of the 258 people who hold biochemical genetics certificates, exactly 200 of them also have an M.D. degree.

This is the group that is least able to expand services, and this is based on 2003 data from the Work Force Study. Their practices are nearly full. Twenty percent were expected to retire in the next five years, and there is only one more year left in those next five years.

Several states were unable to expand their newborn screening panels to meet the uniform requirement due to a shortage of metabolic physicians. So this is serious.

The approaches to remedying this problem need to be multi-pronged. One thing that I think has been said, maybe not bluntly enough, is there is no federal funding for training medical geneticists. Any time ACMG is asked to provide technical advice on legislation, we ask them to put in funding. In fact, S.1858, which is the newborn screening legislation, was just reported out of the Senate HELP Committee and it does have money in there for education.

We also have one ACMG-F through our foundation, an industry-sponsored M.D. fellowship position for a clinical geneticist. Finally, our board has approved, together with ACMG, the creation of our first clinical geneticist subspecialization. This is the medical biochemical geneticist. So there is now going to be subspecialization in clinical genetics, and I think we are going to be seeing more of that.

As Melissa said, the Banbury conference on the evolving role of the geneticist first talked about the fact that we need to recruit and how can we improve training and recruitment. At the end of that conference, they realized that when different people around the room, the stakeholders, said medical geneticists and the practice of medical genetics, they

weren't all talking about the same thing.

So we brought people together again with broad representation back to Banbury Center a year and a half ago to define the domain of medical genetics practice. How can you write curricula if you don't know what you are training people to do. Can we agree on some principles that underpin this practice.

This document is about to be published in Genetics and Medicine. It will underpin Banbury III. Mike wants me to make sure I tell people that it is not going to be at Banbury, but we still call it Banbury III anyway. This is where we are going to actually develop curriculum and medical genetics training for genetics.

I'm not going to go through all of these principles, but they do come from a pre-published document. I will just highlight a couple of those as follows.

The first one is obviously that we are dedicated to improving the health of individuals, families, and communities, and that we see patients across the life span and for conditions involving all organ systems. We also, obviously, have a public health interest, and we need to be nimble. We need to respond to the rapid pace of discovery with new educational and training and practice paradigms.

A little bit about how we practice. Obviously, we are, and remain, a team-based sport. We are interested in the translation of new technologies into health care, monitoring outcomes and also patient management, and this includes becoming medical homes and coordinating care as appropriate. This goes on and on about the need to expand the training, and many of these concepts have already been said today.

I think I will end with the last slide to say, how are we going to get to where we need to be from here. I'm from the American College of Medical Genetics, and we are here to help.

Muin, in response to your comment, we now have a tagline, which is "Translating Genes into Health." We are working on a branding campaign. We do have a public relations and media advisor working with us, and I think you are going to be seeing the roll-out of really trying to position us more in the eyes of the public as the individuals who actually translate genes and genetic information into health and into health care.

Hunt Willard, in his presidential address in 2001, when he was the ASHG president, talked about opening the tent. We have to open the medical genetics training tent to expand the work force. We have to expand our number of joint training programs, such as pediatrics, medical genetics, internal medicine medical genetics, pathology medical genetics, neurology medical genetics, and the list goes on, as well as some of these subspecialty certificates.

Again, realigning the training efforts to involve common disease, which is often very, very complex. It is not simple genetics and does involve the teaching of gene-environment interactions and health care throughout the life span.

We have to be cognizant of how genetics services will be distributed based on complexity. There is a role for genetics in primary care. There is a role for genetics in specialty care. There is also a role for the medical geneticist.

We also, I think, need to keep our eyes on the ball of the consumer genetics and personalized medicine movement. I'm pleased to say that several of these companies have approached the American College of Medical Genetics looking for ways to use us as resources.

So to position the profession, we want to be able to provide adequate clinical support to the range of service settings, and there are not enough of us to go around. We are already the educators of medical students, but we are supposed to be the educators of everyone else. We are going to see all these patients.

We are going to reach out to rural areas with our new technologies. We are doing a lot to push telemedicine through our National Coordinating Center and the Regional Genetics and Newborn Screening Collaborative.

We are looking at new training modalities. We are integrating our point of care and decision support tools into the electronic medical record. So some of us need to be writing these tools and developing them and promoting them and testing and evaluating them to make sure that they are working.

Am I getting tired going through this list. But we need to, obviously, anticipate future needs and get ahead of the eight-ball and, as I said, develop tools and new practice paradigms so that the work force can grow. It is going to take a multi-pronged approach and a multidisciplinary one.

I think I will stop there. Thank you.

[Applause.]

DR. McGRATH: Thank you, Judith. That was a mind-full.

I'm really happy to open up the table now to use this time to address questions to our guests, who traveled far and wide over short notice to come here and talk with us.

DR. TUCKSON: I just want to give one question as you do this, just for you all to think about. I was trying to follow the discussion as best I could. I just want you to be thinking. This is not necessarily right and you probably have your own constructs or your own notes that are better than mine.

But I'm asking myself over and over again, is there a problem. Is there a problem here that deals with genetic exceptionalism or with just medicine? So, is there something about genetic exceptionalism here, and is there a problem.

Is there a problem with availability of expertise by disciplines, by diversity? Is there a problem because of the integration of expertise into daily clinical practice, whether that is the individual doc or professional level or the infrastructures of coordination across disciplines. Is there a problem.

Is there a problem because of compromised patient care. Do we have any evidence to find out whether this is a big enough issue because somebody is not getting good care as a result of this not being as optimal.

The second big set of questions for me is, is there something, then, that the Secretary can do. Is it through connectivity to others of our reports, like the Oversight discussion we are going to have, or through the Coverage and Reimbursement Report, which has a whole section on this whole idea of who should get reimbursed for what and that whole big thing which I opened up around that genetic counseling deal.

Is there something the Secretary can do around CMS in terms of payment. We will not pay for thus or so unless you have proven that you have kept up with your level of education. In other words, it may not be that you need to beg and plead somebody to go to a CME course or whether they have gotten their boards. If you don't have your board certification

and you haven't kept up with the board certification for family medicine that includes a rational genetics one-on-one, you don't get any bucks. You could be that draconian. I'm not saying this is what we should do.

The last area is, is there something that the profession should be doing themselves and that you use our bully pulpit to urge the profession to do, whether it is, again, the specialty societies, the boards, and what not.

Anyway, I'm not sure it was helpful, but I just wanted you to have something in your mind as you try to think through this now. You are supposed to be asking questions that get you to a conclusion. So don't ask questions just because you are interested. You only get to ask questions that will take you to, in your own mind, a yes or no about whether you want a subcommittee and what you are going to charge that subcommittee to do if you do it.

MR. MILLER: I would just add two questions focusing on framing it. What is the role of SACGHS in this discussion. Also, I'm always interested in what are the metrics that we look to in terms of education. You talk about a dearth of folks, but what are the metrics that we should be looking at.

I have two points, I guess. One is, a number of you mentioned diversity issues, and I acknowledge and agree that that is an important piece, but none of you mentioned it. I would encourage you, when thinking about diversity, to also think about people with disabilities in the profession. In fact, people with disabilities are a medically underserved and underrepresented population when it comes to the health care professions and particularly with respect to genetics.

And, to think of people with disabilities as simply patients as opposed to a community that brings something to the table in terms of understanding the experience of living with a disability or having a disability is something that is very unique and equally valued with respect to diversity issues with respect to race and gender.

My question, though, is I want to piggyback on something else that Reed said in terms of this question that he asked right before the break in terms of who is qualified to make judgments about this. We have all these different groups that are thinking about genetics in different ways. I'm wondering whether there is a role for the SACGHS in bringing groups together and talking about what are qualified genetic professionals. How do we think about genetics in health care and whether the overarching group of this Committee is something that can bring the individual groups together to talk about that issue.

DR. McGRATH: Muin.

DR. KHOURY: First, I would like to thank everyone for this wonderful tour de force this morning. I'm glad to hear Judith's talk about the reorientation of ACMG to translating genes to health. That was really music to my ears. I can go home and be happy about that.

But as we make this transition, so to speak, from the paradigm of taking care of people with genetic diseases and their families and their communities, et cetera, to how to deal with genetic information in general, whether it is microarrays or gene expressions or taking drugs in the practice of medicine and public health, I would like just to get your thoughts about how you think this should be done given the lack of work force.

The numbers are not going up, if anything. The information is going up. I like your statistics. One lawyer per 17 residents of D.C. seems a bit steep. But if you think about how many geneticists you need per snip --

[Laughter.]

DR. KHOURY: -- or per base pairs, those numbers could be a little bit too much.

What is the role of the new genomic person or genomic specialist, whether it is a genetic counselor or a genomic counselor, nurses in genomics, the medical geneticists? What do they have to do to translate genes to health? What is the role of evidence-based practice and how can the new geneticists embrace the concept of evidence-based medicine and use it in translation?

Without that, there is really no payment for services. Let's face it, that is the current model here. It is not the traditional model of genetic information as valuable per se. We have to sell why is it valuable to improve health, or at least metrics of health.

DR. FRIES: Can I comment a little bit on that? I am an epigenemian as well as a geneticist, so I can look from the different roles of the different fields.

Evidence-based medicine, for example in OB/GYN, is largely based on large studies that have been made of practice patterns, whittling down things that have been done from just simply "We have always done it this way" to actual evidence that this makes a difference.

Part of the difference with that in genetics is that when you are looking at it as a residency-based specialty, since 1992, it has really not got that body of practice information.

So you would have to incorporate some of all specialties' practices and incorporate genetics into those in order to assess that. To ask genetics to screen itself for what its best practice guidelines have been is going to be based on a limited number of experiences on patients. So it is going to need a different sort of developmental pattern.

However, that doesn't mean that it can't be done. I think the way that it has to be done is the way that it is done anywhere in medicine. Look at innovative strategies and then set up large sponsored trials of those innovative strategies as a comprehensive group. I think that that is an area where this group could be very influential both in funding and support of those kinds of strategies.

DR. McGRATH: Mara. I'm sorry. I was going to go Mara, then Joseph, then Julio. Oh, I'm so sorry. I missed that. Of course.

DR. KAHN: There are two questions on the table. There is Dr. Tuckson's question and there is Dr. Khoury's question. This may be the only thing that I have to contribute to this conversation, is the answer to this question.

You asked what is the problem and what can the Secretary do about it. I would give you a simple take-away. I think the problem is integrating genomics into the daily practice of whatever our professions are, the daily practice of health care. It is about taking care of people.

What can the Secretary do about integrating genomics, which is not well integrated, into the daily practice. I would suggest to you that the most important thing the Secretary can do is focus on decision support.

Now, there will be a component of education that is necessary. If decision support is integrated into electronic health records or even into office practices that don't have electronic health records, there are other ways to get decision support through the Internet. They have Internet connections.

The clinicians are going to need to be educated on what is in that decision support, and that is really critical, but that clinical decision support at the point of care is where

people are being taken care of. That is how you really integrate this information into clinical practice.

MS. BENKENDORF: I just wanted to make a comment about decision support. I think our newborn screening ACT sheets and algorithms were mentioned, and I just want to tell you where that is going. The idea was, obviously, to get these into the hands of the primary care providers. So the newborn screening laboratories do send those out with all positive test results. The next round of ACT sheets and algorithms that are going to be developed are going to be for genetic tests commonly ordered by non-genetics physicians, again to be disseminated by the laboratories.

We are doing two things. One is that AHIC identified the ACMG and these ACT sheets as a prototype, and they are going to be integrating them into medical records as a point of care education tool through their genetic and genomic testing initiative. So we are going to be evaluating how that works.

The other thing is that the ACMG has obtained funding to convene a meeting, which will probably be this spring, of all the EMR industry folks to talk about decision support tools.

DR. TUCKSON: As the Committee continues to deliberate on this, recognize that we will be hitting these issues again under the Oversight Committee conversation. So there is a considerable part of the things just discussed in the Oversight Committee, so know that we have more than one chance to go after this.

MR. RACKOVER: There is a quick comment I need to make as an educator. We need to get the GINA bill passed. I'm tired of being able to talk with students but they won't get past that. So everybody at this table, before we leave Washington, should make a phone call. Without that, the students don't have to hear that they are concerned about the ethics behind the genetics bill.

DR. TUCKSON: I just want to make sure that you know. Thank you for that. You are preaching to the choir. We have been fighting this a long time. By the way, you can't make that phone call today. After today you can call.

MR. RACKOVER: But tell your friends in Oklahoma to make the call.

MS. ASPINALL: Thank you. Again, this has been an incredibly impressive morning. I was struck by a couple of things. First, how much is being done across so many different organizations. It is great to see that.

That being said, [there are] two missing elements. One is I think there is one constituency not here, which is industry, and how much industry is doing in individual organizations and as a group to come together. I'm not just talking about funding, which has been a piece that several of you mentioned. By no means is a lot of this industry-funded, but I think that that is a component that is important to recognize.

The second thing I was struck by, Reed, is what you said. This may come up this afternoon. What I see is a window of opportunity with the coming of the electronic health records. Somebody mentioned that 40 percent of physicians have them and 12 percent more are on their way. I believe that there is a window of opportunity because I have heard too many times about great new practice guidelines, new tests, and new information that can't fit into a system.

If anyone has changed a system, you only want to do it once, or at least once in a

lifetime. You don't want to have to them revisit it. So I think there is a window of opportunity now.

That being said in terms of context, my question is, do you work together? Is this the first time that all of you have come together or do you really share your best practices across the organizations and put together metrics so you and each other can see how you are doing against your own goals and against a broad, potentially set by this Committee, industry standard?

MR. McINERNEY: I don't think that we have worked together to the extent that you would hope we have in terms of being aware of one another's metrics and helping one another to assess those.

But I can say at least that we have worked with virtually every organization represented at this table, and that is not just because we have reached out to them, it is because they also have reached out to us and to the other people at the table.

So we do work together. It is a small community in many ways. Those of us who are interested in genetics education are sometimes, perhaps, a little too inbred, and geneticists ought to be aware of the danger of that kind of behavior.

But we welcome input from other groups as well. One of the things we do at NCHPEG, and I'm very happy to hear the affirmation for these kinds of approaches, is we reach out to the extent possible to a lot of groups that are not formally in the genetics community and we ask them what do you need. How is genetics manifesting itself in your practice. This goes back to Dr. Kahn's statement about looking at medicine through a genetic lens, or health care through a genetic lens, but then also looking at genetics through a nursing lens or a dietician's lens or a PA's lens.

So we do talk to one another. We try very hard to make the education relevant for the practitioners. Somebody talked about champions before, and that is very important in the Diffusion of Innovations mechanism. We are aware that there are few champions within some of these professions. Some of these professions are very large, like PAs. There is a handful of champions, and we have to clone them, or at least give them the resources to extend their impact somehow.

MS. ASPINALL: I think that is helpful. You don't have to recreate the wheel by sharing the best practices across the organization and, again, with industry. I think there may be a role for a convening organization to help you do that in an efficient way.

MR. McINERNEY: Thank you. That is why NCHPEG came into existence, to try to decrease the extent to which people recreate the wheels. I will tell you, however, that there is a fair amount of parochialism across all disciplines. Not just in health care, in all disciplines. There is some sense that if we didn't develop it ourselves it is not necessarily going to be right for us.

But we just don't have the resources to, I will use the word "squander," on that kind of approach. There is a certain set of core principles, perhaps core competencies in genetics, that are broadly applicable, not withstanding that they have to be elaborated differently for each profession. That is one of the things we help with as well. So we are on that page. We don't want to recreate wheels.

DR. McGRATH: Julio, do you want to go first?

DR. LICINO: I just had a comment, which is that for this meeting we received all this preparation package. Then I, coincidentally, was all out in The New York Times this past

Saturday about the genetics companies offering the 1 million genotypes directly to consumers.

I work in this field, and I was kind of shocked because I know that people offer genotype directly and you can test for this or for that. But to see the whole 1 million [available] to consumers. I'm doing this for research and the price that they charge me for research is exactly the same that they charge for consumers. I think I'm being overcharged because they are making a profit.

[Laughter.]

DR. LICINO: But anyway, I see as a mismatch. I'm in Miami now, but until last year I was at UCLA and I was part of the Medical Genetics Training Program and I had someone who actually trained with me in the program as part of his research training. The research component to his training was all done with me.

So I'm kind of familiar with that structure. I don't know how to say this, but from what I have seen, it has a background and it has evolved from traditional medical genetics. You have now this kind of collision of this explosion of information, which is really not about the risk for typical genetic diseases. It is like susceptibility alleles, some of which may contribute very little to the disease risk.

How equipped are the people who go through these different training programs to deal with this kind of information and be able to interface with the patients? Because the person gets this test and they are going to go to a professional. They say, "I tested for this. What does it mean? Can you digest this for me?"

Are the professionals equipped to do this? Should they be equipped to do this or not? What is the situation? How are you going to handle this kind of a direct-to-consumer, very aggressive effort with traditional medical genetics?

I could see you taking a lot of different positions and justifying them very strongly. You could say we have nothing to do with this. It is not ours. It is not genetics. Or you could embrace it, or you could be cautious, or anything in between. What is your perspective on this?

MR. McINERNEY: I will jump in quickly. I think it depends on how you define "professional." If you are talking about the genetics professionals, I think they are equipped to handle this. I think the average primary care provider is going to be absolutely clueless when confronted with some of these test results.

But the educator in me sees an opportunity here. In fact, we will be meeting with the 23 NV people soon to talk about, I hope, complementary activities to what Judith Benkendorf was discussing from the college.

But if I were trying to integrate genetics, for example, into medical education or into education of PAs or nurses or dieticians, for example, I would take one of those test results into my class and say, look, someday soon when you are in practice, one of your patients is going to walk in with this. What are you going to do. What do you need to know. How do you expect to respond. How are you going to handle this in your practice.

So I think there is a real opportunity for us here because maybe these companies are pushing us faster than we were willing to push ourselves.

DR. CASHION: If I could respond to that also, I actually have an NIH-funded study that is looking at gene-environment influences of weight gain in renal transplant recipients. We are doing adipose gene chips and we are looking at blood as well as dietary nutrition and

exercise.

So I have that part of me, that mind-set, that is working on this. I also teach undergraduate nursing students genetics. People say, well, what do you teach. What is the content in that. Over the last six years, I have found that the most influential content you can teach them is how to be the lifelong learner. We really focus on websites. We go to websites every day and look at them.

We do the Family Health Initiative. They have to do it on their own families. We didn't have the gene chip information that is now out there by the three companies, and I was actually thinking I need to pay that $1,000 and let me go ahead and get this done on myself. But those are the examples that are meaningful to the undergraduate students.

I also teach advanced practice nurses, and they are the ones who are coming in and wanting the BRCA1s and BRCA2s. So again, it is how to look for the knowledge. Whatever I teach them today is not useful two or three months from now. So I really do not teach content as much as I teach how to learn, how to maintain your skills.

MS. PESTKA: I would like to add to that as well. I think much of the answer to this question is not so much in our hands as it is in our patients' hands. As you pointed out, patients are learning. The video snip that I showed of James, James was pretty exuberant in his hopes for genomics. But almost all of our patients come in and they have hopes and they have expectations. We really need to be prepared to deal with those expectations.

I believe there certainly is a place for our experts, our medical geneticists, our genetics counselors, but obviously there are not enough. So we need to have all healthcare providers educated, and then we need to have our body of experts that we can refer to with the really complex cases.

DR. TUCKSON: We have to close out. Was there one last one, Barbara, you had? Toby, go ahead.

MR. CITRIN: I just wanted to at least tie a few of the comments and a couple of the questions together from the public health perspective. It seems to me that Dr. Kahn's comment on the need for integration is very true of public health. It actually addresses Dr. Khoury's question about the size of the work force.

We don't need a larger faculty in our school of public health to incorporate genomics in what we teach. We just need people who are teaching the subjects they teach to incorporate genomics in those subjects. To some extent we have been successful in moving in that direction.

The same is true of public health departments, people who specialize in chronic disease. People who are doing studies of risk factors, who are administering behavioral risk factor surveys need to incorporate family health history in what they do. So it seems to me that it is not a work force question. It is very much a training and education question.

I think this also relates very much to Dr. Tuckson's question about is there a problem. I think the problem is that if we do not move the educational process forward this way, the public will be seeing genetics as segmented because the private sector is moving things in that direction.

The public's consciousness is that genes are more and more responsible for their ills. The net result of that will be genetic tools, a worsening of disparities as these tools are available to some and not to others, a sense of genetic determinism that will result in more

disparities, more stigmatization, and more sense of a rebirth of eugenics, all those horror stories that we are all very familiar with.

The integration of genetics with all the other factors of health and disease; in the public health sector certainly that is our defense against doing less and having the forces that are moving very fast distort people's views of their health and what causes it.

DR. TUCKSON: Eloquently stated. First of all, we have benefitted from a terrific panel. Not only did we get smart people but people who know how to express smart ideas very cogently. We really appreciate it. Let's give them a round of applause, please. This is terrific, just terrific.

[Applause.]

DR. TUCKSON: Now, here is our dilemma. We have a very power-packed session this afternoon on oversight. A lot of this material that we just heard, as I tried to intimate, is part of some of that. But there is still not going to be, in my way of thinking, even under the most blessed of circumstances enough time to get everything squared away.

I would propose that I think there is something here that is going to require a deeper deliberation by us. I would be surprised, but I'm open for someone to object, that would say that in the broadest of unformed terms that there is work that this Committee will want to pursue in this area.

Unless I hear someone scream out that this is all solved and there are no issues here and everyone return to their homes, I think that we are going to wind up creating a subcommittee to take another look at this.

What I cannot do, nor do I think we are prepared for, is to define the agenda or the scope of that work yet. So as the afternoon unfolds, we will be thinking about that a little bit and have something to present to you that will arise out of the oversight conversation.

Barbara, I know that you may not be able to be with us, which is a perfect way to draft you into continuing on, and we will populate the committee with a few interested people. I know that I'm going to draw Mara into this and a few others. Not Joe because he is disruptive sometimes.

[Laughter.]

DR. TUCKSON: But we will do that.

I want to telegraph quickly, before we go to lunch, one thing that is real clear for the new members. I think it was the right question and a great answer came from the committee: "Do you all talk to each other?"

One thing that I think is frustrating for all of us is when we try to ask who is qualified to do what, particularly when it comes to counseling and who then should be qualified to do counseling and what is the reimbursement that ought to go.

This issue comes up over and over and over again. We have asked over and over and over again for all of the factions to sit down together and figure it out, create an umbrella, lay out the issues, and then bring it back.

I think that that now, as I get more mature in this, is an unreasonable expectation. Therefore, I believe that it is a role that we might legitimately play to try to be a convener of the conversation. I don't need to get one more letter from one more organization that just rehearses exactly what they wrote five years ago. Clearly, we are not getting the message. We need to step up to the plate and convene.

That would be at least one thing that we would do. Secondly, I think it is very important that we understand and bring together very specifically those professional disciplines to tell us how they are using their normal regulatory responsibility for who is qualified to do what and to determine the adequacy of those things. We shouldn't be trying to recreate the infrastructure of American medicine.

So I think that those are at least two low-hanging fruit. I think this third low-hanging fruit is real clear, and that is this decision support. We have been told pretty clearly that that has to be looked at. So the AHIC versus other mechanisms in terms of getting this review of what is going on in the EHR is something that is in the Oversight Report, so we will probably hit it there. But I think that is a third area that we are going to want to lock in.

So I at least see three things as a broad, general set of issues. Who is qualified to do what, is a real big thing, and that has to do with clinical practice and it has to do with counseling. Related to who is qualified to do what is who should get paid to do what. That is a little beyond the narrow confines of education, but it is so related that you almost have to look at them together because that determines a whole lot. Once you start asking the question "Who can get paid?" that starts to answer a whole bunch of questions. Then this idea of electronic records. I see those as being some of the things that are there.

With that overly long summary of putting down just a temporary marker, we are going to send you off to eat. Now, the dilemma is that we are at 20 minutes of, almost, and you are supposed to be back here at 1:10. So the dilemma is really yours, not mine. We will see you back at 1:10 because, whether you are here or not, we are starting at 1:10. Have a nice afternoon.

[Lunch recess taken at 12:38 p.m.]

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DR. TUCKSON: Thank you all very much. As always, we start on time. We have a power-packed afternoon, on top of a very power-packed morning, but we are going to pause for just 10 seconds and introduce a very special new friend of the Committee.

Rick Campanelli is the Secretary's counselor for science and public health. He has been an integral part of the Secretary's leadership team since 2002, when he was appointed director of the Office of Civil Rights, and led the significant effort under former Secretary Thompson to finalize the HIPA privacy rule and spearhead OCR's HIPA enforcement program.

When Mike Leavitt became Secretary, he was asked to serve as the first counselor for human services and then as his counselor for science and public health.

Now, I will tell you that Rick has had a rich and varied legal career even before joining HHS, both in the private sector, representing nonprofit organizations, and in the Justice Department as a litigant fighting against unconstitutional conditions in mental health institutions and race-based segregation in state prison systems. Later, in the State Department, he worked on U.S. initiatives to end apartheid. A terrific career.

We appreciate the interest and support that you have shown in our committee since taking Sheila Walcoff's place as counselor a few months ago. You took the time to meet with our Oversight Taskforce in September and just a week ago were briefed by our superstar, Andrea, by Steve, and Marc about the draft Oversight Report that we released for public comment earlier this month.

With that, I just really want to say, also -- I'm sorry, Greg, but you are going to get it again -- Greg Downing is just terrific. We have enjoyed unprecedented relationships with this administration and this Secretary. Not to say anything bad about the fine ones that came before. Lord knows they were all wonderful as well. But you have been super-wonderful this time around.

So with that, Rick, let me welcome you to the Committee.

MR. CAMPANELLI: Thank you very much, Reed. It is great to be with you all. I see around the table friends from various parts of my life, and I'm grateful to be here. When you talk about that varied legal career, my kids refer to it as my checkered legal career.

[Laughter.]

MR. CAMPANELLI: But it has been a privilege to serve since coming to work for Secretary Thompson and Secretary Leavitt.

Secretary Leavitt extends his greetings to you. He is traveling, as I was with him in the last couple of days, on import safety and food safety just before the Thanksgiving holiday here.

But I just want to say that he is so appreciative, as of course am I, for the very hard work that has continuously gone on with this Committee over the year. With that, in this time of Thanksgiving, the only thing I want to say is I'm really looking forward to just being with you. As Reed mentioned, Andrea has been very kind. We had a good talk about the report of the taskforce, and I'm looking forward to hearing more discussion today.

What I just want to say as we approach Thanksgiving, thanks to a few people who are going off. Two of them aren't here, but Chira Chen is down there. We just got to visit. I just wanted to say thank you for your great work on this Committee. Since 2005, right, Chira? The

Committee has had the benefit of your participation, your perspective, and orientation for consumers in making sure that these matters are things that get right to where they are supposed to get, to the bedside, and your participation in the Large Population Studies and also on gene patent and licensing studies.

So I just want to say thank you to you for your service. I know that I say to everybody in this room at the same time that it is a wonderful thing to participate in these things and a sacrifice. It is a very high calling, and we appreciate it.

I also want to say thanks to those who aren't here with us right now, to Cynthia Berry, who was with us with the original Committee and helped in the transition, and also Dr. Hunt Willard, chair of the Large Population Studies Taskforce and the Pharmacologics Taskforce. Just thanks to them for their excellent service on behalf of the Secretary and the Department. Thank you very much.

DR. TUCKSON: Thank you.

[Applause.]

DR. TUCKSON: We know that your schedule is very busy today, and obviously at some point I know you will have to leave us. But please stay as long as possible and don't be shy. When you have to go, we understand.

We have to say one thing to the troubadour.

[Laughter.]

DR. TUCKSON: You cannot get the Presidential Medal of Freedom, the nation's highest civil award, and not be noticed by your colleagues around this table. Now, I know that you are turning red. I don't have to look.

But everyone around this table understands that that is awarded to those who have made especially meritorious contributions to the country, that you got it for your leadership in the Human Genome Project, which obviously revolutionized genetic research, and for paving the way for applications that will greatly expand our ability to diagnose, predict, and treat disease at the individual level.

You got this thing on November 5th at a major White House ceremony, and you were praised for your relentless pursuit of knowledge and your extraordinary intellect. By the way, I did notice with some interest that the President did liken the Human Genome Project to the Apollo project in scope and in the long-term potential that it will have, and that Americans are proud of this wise and humane American scientist who is behind our national scientific effort and achievements in genetics.

So not only that, but Francis, you are not only smart and brilliant but you are collegial. It is a rare gift for a committee like us to have someone who is not only very, very smart but who knows how to play nice with the other boys and girls, who can actually work with other people, not have a massive ego to the point where you can't sit down and have discourse and dialogue, and come up with shared accomplishments.

I think that you have always prided yourself in the way you have demonstrated here as being a member of the team and a member of sharing. But dear Dr. Troubadour, we really do honor you.

[Applause.]

DR. COLLINS: I have to say thank you so much to my dear colleagues for that. That means a lot. I really value the collegiality on this group as it has wrestled with so many

difficult issues. I gather you were exposed to another facet of me earlier today.

[Laughter.]

DR. COLLINS: I will have to tell you one slightly funny aspect of what happened in the Medal of Freedom run-up. When I got the call from the White House, which was a stunning call, to say that this was going to happen, the person who called, someone on the White House staff who I had not met before, described the event. I was of course by that time lying on the floor. At the end of all this presentation about the circumstances and the logistics and what to wear and where to show up and which kind of security issues to worry about, he said, "Oh yes, there is one more thing I was asked to tell you. There will be no singing."

[Laughter.]

DR. COLLINS: To which I guess I would have to assume my reputation had preceded me. The President wanted to be quite clear this was going to be a dignified occasion.

When I did get the award and the President put the medal around my neck, I was feeling pretty inspired. So I turned to him and said, "Mr. President, I feel like singing."

[Laughter.]

DR. COLLINS: And he gave me a very warning sort of look, and I said, "Just kidding."

DR. TUCKSON: Pushing the envelope again.

That really is fun. I think the spirit of collegiality and so forth is very important.

So here we go into this very complicated session. Now, we have rehearsed this a number of times, so all of the five new members of the Committee understand that this is a part of our DNA. We have inherited this issue of oversight. Andrea is going to take us through this.

I want to, again, ask Andrea to emphasize, and I want you to pay attention, to the process that we are under. We have already put forth a draft, a very early draft document, into the public discourse. So the Committee has put forward a draft into the public discourse.

We are soliciting responses from the public through December 21. We are soliciting responses from the public, comprehensively described, through the 21st. Then, with their input and our own new members' and others' review of this, informed legitimately by the public comments as well as our own -- but I'm being very explicit here -- respecting and listening and reacting and responding to the public input, which is what we asked them to do, we will then engage in a second process to deliberate and to determine what it is in fact that we believe. Do you understand what I'm saying?

So we are here today to listen to more public comment, other examples of other experience, that we will then use in its combined wisdom to redeliberate it as a Committee.

So I don't want the Committee today to get caught up in redebating the report, a report that is already in the public discourse asking for comment. It is not appropriate. What you want to do is you want to listen today, you want to raise issues, you want to ask questions, you want to discuss. Then you are going to have a whole lot of energy and time where you are going to grapple with this thing anew. You are going to grapple with it anew after this meeting, or after the 21st when you get the public comment.

With that, we are going to march through this, and then we will have our conversation.

Andrea.

DR. FERREIRA-GONZALEZ: Thank you, Reed. I think we are waiting a little

bit more for the presentation.

DR. TUCKSON: While we are going to reboot the computer, we are going to remind you also, and I think that we got into this wonderful issue the last end of the session, so let's try to lock it in.

We had this notion around the genetic training issues. Francis, you weren't here, but it is an old conversation. We had a whole bunch of stuff on who is qualified to do what and who gets reimbursed to do what. That is a big part of this Oversight Committee conversation.

So one of the things that we have to finish today, if we can, the one deliberative action that we are going to take today, hopefully, is to try to empower a committee that is going to look at the training and education issue within the context of all of this. That might be one thing that we will get closure on before we leave today, at least trying to empower them with a task. The charge will be probably to go and create its parameters, its context.

The Committee will probably have to invent its own priorities, but I think we laid out three at the end of the meeting.

I think that I am certainly going to look for those who want to be a part of that effort to start thinking about self-assigning themselves. Barbara hasn't escaped yet, so as soon she leaves the room I will appoint her chair.

DR. FERREIRA-GONZALEZ: One thing we can do is, everybody has their handouts in their folder. We can pull the handouts and maybe start, and then they can catch up with the different slides so we can keep moving and leave at a decent time.

[PowerPoint presentation.]

DR. FERREIRA-GONZALEZ: Thank you, Reed, and good afternoon, everyone. As Reed said, we have been working diligently over the summer and fall to consider the questions posed by the Secretary. Part of our work is now in the critical stage of undergoing public review, and we are looking forward to the input we receive to help us ensure that the advice we give ultimately to the Secretary is sound, forward-looking, and in the public interest.

We are going to be receiving public comments until December 21st, and our session today is part of that process. We are seeking public comment.

Today we will begin with an academic analysis of regulatory gaps in the oversight of genetic testing and models to address these gaps. We will have about 20 minutes to discuss the findings of these analyses. We will also hear from various stakeholders regarding the oversight of genetic testing.

Sharon Terry, a dear friend to our Committee, will describe the key points that emerged from a summit meeting held in September by the Genetic Alliance. David Mongillo will comment on behalf of the American Clinical Laboratory Association. Patricia Goldberg will present on behalf of ISONG, and Dr. Patrick Terry will also report from the Coalition for 21st Century Medicine.

Before I begin, I want to take a moment to review the Secretary's charge and main elements of our draft report. The draft report is a comprehensive map of the steps needed for the evidence development and oversight for genetic testing. The charge included eight questions about key measures of validity and quality of genetic testing technologies and processes in place to assure their safety and effectiveness.

The Secretary also asked the Committee to consider government and private

sector solutions to gaps in oversight and advised us to focus on the future so our recommendations will be relevant and forward-looking. The Secretary has asked for recommendations by February 2008.

The report was developed by a taskforce comprised of SACGHS members and experts that we have recruited from the federal agencies and the private sector. As you can see here on the slides, there are 33 members. As was commented yesterday, it takes a village.

The taskforce interpreted oversight in broad terms, and I think this is very important, to demonstrate that formal regulatory mechanisms are not the only components of the system. The report frames oversight in a very inclusive and comprehensive way, to include federal and state governments and regulatory agencies, standard-setting organizations, knowledge-generating organizations, private and public sector health care payers, professional societies, health providers, and patients and consumers.

The report was organized in seven chapters and makes 16 recommendations, a little bit fewer than the PGx, that address a number of gaps in the oversight of genetic tests.

The report was released for public comment on November 5th, and this comment period will end on December 21st. To encourage broad input from a wide range of stakeholders, we used several outreach mechanisms, including the federal registry, our website, and a targeted mailing of about 2,000 individuals and organizations. We have also encouraged a number of organizations to inform their members on the opportunities to comment on the report.

This is a snapshot of the website where the report can be downloaded. I will show you the URL in a moment.

We reached out to many stakeholders, including representative nonprofit organizations, advocacy groups, professional organizations, policy groups, healthcare providers, industry, laboratories, government agencies, and other advisory committees. We want to be very inclusive of trying to get public comments back to us.

After the comment period ends, our work will intensify in order to meet the Secretary's request for recommendations by February. The comments will be analyzed and summarized. A lead for each chapter of the report will be tasked with considering the comments and making revisions. As needed, they will call on taskforce members for their expertise.

The steering group, which is five members of the SACGHS members and the taskforce, will have weekly conference calls in January to assess the progress. On January 23rd, the entire taskforce, with the 33 members, will discuss revisions to the recommendations.

At the end of January, we have planned a conference call for the steering group to brief the full Committee on revisions to the recommendations in preparation for discussions of the February meeting. Following the February meeting, we will make revisions to the recommendations to reflect the Committee's input and submit the final recommendations to the Office of the Secretary by the end of February. The final report will be formally submitted in April.

This is the report URL. It is available for downloading.

DR. TUCKSON: Andrea, you have laid it out very nicely. I just want to make sure that in terms of your guidance to your colleagues around the table, once they have had a chance to see the public input at the end of December and have their own reactions and ideas based on that, and revisions to the report that the Committee has put out there, are they encouraged or will they have an opportunity to submit or participate in the taskforce's

deliberations in early January so that their thoughts can be considered in that process prior to it coming for final deliberation to the full Committee?

DR. FERREIRA-GONZALEZ: That's correct. We are very keen on those two particular events. The full taskforce with the 33 members will have a discussion of what we come up with, and then we will bring the SACGHS members, again through another conference call, to tell them where we are with changes to the recommendations to start engaging them and just putting them in the mind frame for the further deliberations that will occur in our February meeting.

DR. TUCKSON: Just to be clear, and you are being responsive, and I appreciate it, because I have a suspicion that certain members of the Committee will be provoked at one point or another to either modify some thinking that they may have had, or had new insights, or we have new members of the Committee, so that they aren't in a position of only being reactive downstream, I think what you have said is that they have mechanisms by which they can get their comments in as the river is flowing, even as they, of course, reserve their right to comment on it once it is more set.

So it is a fluid process. I just want to absolutely emphasize it is a fluid process and you are encouraged to participate in the fluidity of it even though you are going to have some formal deliberations and react to it once it is more gelled.

DR. FERREIRA-GONZALEZ: Thank you, Reed.

DR. TUCKSON: Francis, you had a quick question?

DR. COLLINS: I really appreciate your clarifying that, because we are doing this in a somewhat different process than we have sometimes followed for these reports, by getting the public comment at this point. It is very important to find out what the public thinks. In some instances, that might imply we were already at the penultimate stage where the Committee had essentially already achieved consensus.

I think in this instance the Committee is still wrestling with some of those issues, and I'm glad to hear you endorse the fact that there is still plenty of opportunity here to reconsider what the final product might look like. We should not consider that we have already passed that point.

DR. TUCKSON: I think that is important. What we would say is that we have put forward some considered ideas and thoughts for the public to respond to. We tried to be respectful of the public by giving them something that was legitimate to consider but not so far along the road as to make their input not substantive and meaningful.

So we have tried to strike a very real balance. We gave out something that was serious for people to think about, which we believe we have done, and now we will undertake the process that we have outlined. So we believe we have struck a pretty good balance in all of that.

I'm glad that we have gotten that issue and clarified. I urge you all to participate to the degree that you would like as this stream flows, even though you will still get a chance to respond reactively once it has completely gelled in late January.

DR. FERREIRA-GONZALEZ: Great. Thank you. Let us now turn our attention to Mr. Stuart Hogarth and Dr. David Melzer, who will provide analyses of the oversight of genetic testing.

Mr. Hogarth is a visiting research fellow at the Institute for Science and Society at

the University of Nottingham. Dr. Melzer is a professor of epidemiology and public health at the Peninsula Medical School in Exeter, England.

We will have 40-minute presentations, followed up with a 20-minute discussion.

DR. MELZER: Thank you very much. It is a great honor to be invited to talk here. It is obviously a bit daunting in such illustrious company.

What we would like to cover very briefly is a little bit about the scientific context, which of course you have heard quite a lot and you have some fantastic leaders in the field, but I think this is going to be very important for the regulatory internationally.

Then I'm going to hand it over to Stuart, who is going to talk about the early commercialization of some of this new generation of common disease markers, some of the policy problems, which are global -- this market is taking off in Europe as well as the U.S., and many of the companies cross international boundaries -- some of the policy problems and some of the policy proposals.

What we are talking about is the result of a policy research project funded by the Wellcome Trust in the U.K., and it focused really on a very simple question: how do we ensure that doctors, patients, and healthcare systems can make informed decisions about the use of the new genetic tests.

So we are interested in the three phases of evidence generation. In talking to stakeholders both in the U.S. and Europe, we heard a lot about problems of incentives and the difficulty of financing clinical studies for the generation of clinical evidence.

The next step of course is the evaluation of the evidence by regulators or the public health and medical community and patient groups. Crucial, also, is the sharing of evidence. This issue of secrecy about the evidence, what exactly is in the genetic tests that people are selling, has been a really pressing problem in Europe.

What the project involved was individual interviews and workshops, focus groups both in Europe and the U.S., contacts with people in Canada and Australia, and I would like to thank the FDA for much advice and attending our workshops here in Washington.

Just to be clear, the Wellcome Trust is an independent research philanthropy with no connections with the drug companies anymore. The project was totally independent, and both of us have no conflicts of interest. We have no patents or anything. Our funding has come partly from the NIH and from U.K. research grants.

We are all familiar with genetic testing in the past, the family-based, often high-penetrance disorders where the clinical significance of a marker is fairly clear. But as you were discussing this morning, we are moving into a much different scenario in which there is a statistical association between claimed markers, in which the marker is rather more common in the cases than the controls. Sometimes the marker is only present in a small proportion of the cases. Sometimes some of the controls have the markers as well. So we are talking about predisposing effects in this project.

Again, you are probably all very aware of the enormous explosion in results, especially from the genome-wide studies. So the Wellcome Trust Case Control Consortium, for example, published 28 independent signals for eight conditions. We have seen some wonderful breakthroughs in age-related macular degeneration. In asthma we have seen really quite a big effect for mutation not from the genome-wide studies but from similar work, and even such

syndromes as restless leg syndrome and type II diabetes.

I work with a group in Exeter that does half of the Wellcome Trust case control analysis for type II diabetes, so I'm going to talk about that and use that as an example.

This is the sort of result that one gets from looking at 2,000 cases and 3,000 controls having genotyped some 500,000 markers across the genome. Along the Y axis is the strength of the statistical association, and along the X-axis is the position. What one ends up with is a massive statistical association, and so far we have only really worked through the ones that really stand out, the really big effect one. I think we are up to about 12 that are now robustly proven.

The important message, I think, for policy-makers and regulators is that there are probably many, many more in the bottom of those fountains that are going to prove to be robustly associated and of course many, many more that were just coincidence.

I was one of the many authors on the FTO gene finding that was reported that came out of the diabetes analysis. It was reported in the media as the obesity gene, the fat gene.

This polymorphism homozygote risk status adds about 3 kilograms of fat mass. The effect is there by age 6. It lasts into old age. There is no sign that is a kind of susceptibility to increasing weight gain, which a lot of people have claimed, or somehow susceptibility to continuing to gain weight. It has been portrayed in this extraordinary way in the media as the obesity gene.

On the lighter side, I guess I should translate the best approach, I think, from our media in the U.K. "'Does your butt look big in these jeans? Absolutely,' say scientists." But of course, these are the messages that the public are getting. At the end of the day, it is a 3 kilogram difference. Very small.

To emphasize that point some more, if you look at type II diabetes snips, the first one to be found, actually a little bit far from linkage studies, was the TCF7L2 polymorphism, which is associated in the homozygote state with about 60 to 70 percent increase, a 35 percent increase if you have one.

Many of the new ones are relatively modest. They are wonderful scientific breakthroughs that will lead to wonderful ideas about new interventions, but in terms of risks to individuals, they are down at the 15 percent, 10 percent increase in risk levels. There is a whole set of them, so we are going to have to regulate whole sets of these genes.

Now, older people are an interesting group to look at because if Cause F diabetes is going to develop, type II diabetes is going to develop, it should have developed by age 65. I looked at just the prevalence of this top genotype, TCF7L2, in people age 65 and over against whether they had diabetes or the intermediate stage, impaired fasting glucose, or no diabetes.

As you see, in the risk group there is a very clear association with this marker. Far more of the TT risk group have got it. But most people with the risk status don't have diabetes or the subclinical prodrome, and many people with the so-called protective allele do have diabetes. So I think this really puts into context that these are wonderful scientific breakthroughs opening up new biochemical pathways, but really, it is very early for clinical use. Of course, people are already marketing this test as a diabetes marker.

We played around, along with a lot of other groups, with combining the allele scores across I think there are 12 markers now that are proven. If you add up the number of risk alleles that people are carrying, you do start getting pretty big odds, so pretty big differences in

risk at the extremes. Many people, however, are in the middle. The top 12 markers seem to explain about 5 percent of the variation in type II diabetes risk.

So with 12 markers, we are only explaining 5 percent, and for most purposes, people would be much better off just having their fasting glucose tested.

Another interesting aspect, which I guess you may have heard already, is that many of the things that have been marketed and that people have been working on have failed to replicate in these big, decisive studies. So for example, there was a paper in the New England Journal showing that of the top 85 markers for myocardial infarction, none of them really showed up. If they are true, there must be an extremely small effect.

But what is also very interesting with the top myocardial infarction marker is the pretty big effect: doubling of risk for early myocardial infarction, so really standing out.

What is really interesting is it sits pretty close to a cancer locus, the P16P15 locus. Mutations in this site are involved in malignant melanoma, and we really have no idea what other effect that snip is having. Although not the same snip, the same locus came up as the second-biggest marker for diabetes. So it suggests that we are just beginning to scrape the surface of the biology. It may also have some effects on cancer.

When people get these tests, we have no idea what the overall predictive value for health outcomes as a whole could be. Again, this test is already on the market.

So, conclusions of that rapid context. A rapidly increasing list of markers, a few with large effect, large enough, some of them, to tempt people to use them in paternity testing or pre-implantation testing. For example, the macular degeneration ones and maybe even the myocardial infarction ones. So there are quite possibly high risk applications but also a lot of small effects. Most of the tests we see and have to regulate are going to be sets of markers.

I haven't mentioned this. Most of these studies are from Caucasians. Very little evidence on minority groups at the moment. The predictive value of these markers may be different. Lots of unknowns on the biology.

I will hand it over now to Stuart, who will talk about the market.

MR. HOGARTH: I think David has made very clear that the science is moving fast, and the science is moving very fast into the clinic. For instance, the U.S. company InterGenetics has launched the OncoVue test in Europe this year. This is a polygenic test which is intended to inform women about their risk of breast cancer by using a whole panel of markers and interpretive algorithm, the kind of thing David was just talking about.

Of course, we have just had the launch of deCODE-me by deCODE genetics, an Icelandic company. This move into susceptibility testing is closely linked to the risk of consumer genetics, companies offering their tests direct to consumers over the Web. In the case of deCODE, they are offering genetic risk assessments for 17 common diseases, including AIDS-related macular degeneration, breast cancer, asthma, colorectal cancer, multiple sclerosis, heart disease, and prostate cancer. Their list of diseases will be continually updated as new discoveries are made.

We have just seen the launch of a very similar service from a company called 23-ME in the States, and of course, we have Navigenics and Smart Genetics very close behind.

Another aspect of this market is that it is really very international. So, deCODE based in Iceland, but the test is available in the U.S. InterGenetics are based in the U.S., but

their test launched in the U.K. The company Genetic Health, the one whose website is shown here, offers a range of susceptibility tests from their base in London, but the tests are provided by an Austrian company called Genosense. This Austrian company offers their tests through intermediaries in a number of countries, including Canada and the United States. So we really are facing what is a very international market.

So, what are the concerns about some of these tests. Well, this is a quote from someone who took one of Genetic Health's susceptibility tests for a range of diseases, including heart disease and breast cancer. As you can see, she understood herself from the test results to have a 140-fold increased risk of cancer, but she was very optimistic that she could deal with this by eating more fruit and vegetables.

The reaction from another piece of media coverage from Genetic Health's tests from the British Society of Human Genetics very recently was that they were very concerned that the tests that were being offered were more or less useless and that they were being promoted with unsubstantiated and overblown claims.

Of course, this really is nothing new. There has been longstanding concern about genetic tests moving into the clinic far too fast, particularly in the area of where tests are for more common diseases. Perhaps BRAC is the most high-profile example to date of a test where the claims at market launch went way beyond the data behind the test.

So this policy concern has resulted in a huge amount of work by a series of high-level committees just like yourselves in the U.S., Canada, Australia, Europe, and elsewhere, looking at the policy issues around oversight. I think one of the key conclusions that has come out of many of those reports is that genetic tests shouldn't enter routine clinical practice unless they have had some kind of independent evaluation.

Linked to that concern about evaluation is a concern about trying to deal with the issue of getting good, comprehensive, accurate information to patients and doctors about tests. So we can think of this in terms of regulation as regulation by information disclosure, which is a concept that is now very popular in the consumer protection field, where it is seen as a way of dealing with the asymmetries of information between creators and consumers.

So there is a real concern, as David touched on earlier, where companies aren't even telling people which snips and which genes they are actually testing. In the case of Genetic Health, there is complete secrecy over the panel of genes. So this issue of an asymmetry of information is really important to the whole oversight debate.

The other thing that has come out of the oversight debate to date is a clear idea of what needs to be evaluated and the categories of information that patients and doctors need to be informed about. This Committee would be very familiar with the framework, so I won't dwell on it.

So we want to enhance oversight to ensure more independent evaluation of tests and better information for doctors and patients. But sadly, at the moment we don't have such a system. What we have is really a regulatory system without real teeth and a whole lot of gaps.

Now, of course, you all would be intimately familiar with the gaps in the regulatory system in the United States, but I just want to speak a little bit now about the situation internationally. It is interesting that although we have all gaps in our regulatory systems, there are actually different internationally.

If we look at the United States, we know that the primary kind of gap in terms of

pre-market review of tests is that historically the FDA has not regulated laboratory-developed tests as medical devices. Of course, now the FDA has identified a small subset of tests, IVDMIAs, which they are going to subject to pre-market review.

In Europe, it has really been rather different. The primary regulatory gap in Europe is that we classify nearly all diagnostic tests as low risk. They are therefore exempt from pre-market review. They are the equivalent of a Class 1 device in the U.S. That includes all genetic tests except tests for PKU. Do not ask me why PKU got singled out, but there you go.

Whereas our treatment of laboratory-developed tests is quite different, we think of laboratory-developed tests as medical devices, although we give some exemptions. For instance, healthcare institutions. If you are a pathology laboratory within the National Health Service in the U.K., you are not subject to our device regulations.

Then, if we look at a couple of other countries, Canada essentially has the same regulatory gap as the U.S. insofar as its authority over laboratory-developed tests is unclear. Australia in fact has been busy revising its medical device regulations. It treats laboratory-developed tests as medical devices, and it treats genetic tests as moderate risk. So most of them are subject to pre-market review.

Of course, in a sense, this is rather depressing. We have been talking for over 10 years about how to enhance oversight. We have been talking about in Europe. We have been talking about it in Canada and Australia, and the U.K., and we still have a whole lot of significant gaps in our regulatory system.

But I guess the important thing is in fact policy is moving, as well as the science is moving. It is being commercialized, and the commercial aspects are moving. Policy is moving as well. Of course, you know what is going on in the States, so again I won't bore you with that.

But in terms of elsewhere, in the United Kingdom we did have a couple of years an advisory code that looked after direct consumer testing, although it fell into abeyance for reasons I won't go into now.

We have a new system for evaluating single-gene tests within the National Health Service, the U.K. Genetic Testing Network. The National Screening Committee has been looking at the regulation of commercial screening services, and in the last year the Human Genetics Commission has renewed its interest in the regulation of direct-to-consumer genetic tests. There will be a new report out from the HGC within the next couple of weeks on that issue.

Within Europe, we have had the creation of EuroGenTest, a kind of network of clinicians and lab people across Europe who work on quality assurance issues and other issues around the quality of genetic testing. Our IVD device regulations are imminently going to be revised, and we have been in discussion with the European Commission and member states about ways of enhancing the regulatory system.

Of course, we have had a drug regulator, EMEA, working on pharmacogenetics, not least in collaborations with the FDA. People in Europe have been participating in international initiatives such as the OECD's guidelines for quality assurance for molecular genetic testing.

The Council of Europe, which is something separate to the European Commission, is working on a protocol on genetic testing which really addresses the issue of

direct-to-consumer testing and recommends that tests be offered with individualized medical supervision and predictive tests for monogenic diseases and susceptibility tests should only be offered with counseling. So we have quite a lot going on in Europe.

In Australia, they decided to completely revise their IVD regulations, in part to deal with the laboratory-developed test issue, in part to deal with the issue of genetic tests. They have also issued guidance in the regulation of nutrigenetic tests, which is an international first.

In Canada, they have issued guidance on pharmacogenetic tests, and then if we look internationally, I have already mentioned the OECD guidelines on quality assurance. The Global Harmonization Taskforce, which is a forum within which device regulators get together to talk about how they can harmonize regulation, has been working on issues around IVD regulation.

The International Committee on Harmonization has been working on pharmacogenetics. Muin Khoury, with colleagues in the public health genetics area internationally, not least with some of our colleagues in Cambridge, has been working on HUGENet, which has been a very important initiative.

Of course, in the U.S., probably the most significant issue around pre-market evaluation of tests and the FDA's role has been issuing of the IVDMIA guidance. I would suggest that the IVDMIA guidance probably has correctly identified the area where FDA intervention was most urgently needed. The guidance has brought clarity to FDA's position in a situation where it had been intervening with individual companies on a piecemeal basis.

But really, it opens up the question of what to do with the rest of the laboratory-developed test sector. Obviously, that is an issue which this Committee has been considering in some detail as it has developed its draft report.

Clearly, the guidance leaves most LDTs outside of FDA regulation. It doesn't cover all monopolistic providers. It doesn't cover homebrew tests, where an unlevel playing field would remain between kits and the homebrew test. For example, the Roche Amplichip, an FDA-approved kit for pharmacogenetic testing, has to compete with non-approved tests. It doesn't deal with other tests that we might consider high-risk, perhaps pharmacogenetic tests, perhaps direct-to-consumer testing.

So we spend a lot of time talking about the whole issue of how the technology and the tests are moving very fast and the ethical, legal, and social consequences. We think of these as areas where rapid change is causing disruptions which need today to be dealt with at a policy level.

But I just want to say something about the way that the IVD industry is moving because I think when we are talking about regulation we need to think about what we are regulating. I think that underlying all this technological change, clinical development, and scientific progress are changes in the business of IVDs that are really very significant. It is really crucial to understand this to thinking about how we deal with oversight issues.

In the traditional model of the IVD sector, companies hold intellectual property platforms and they tend to compete with each other. They develop different versions of testing for the same biomarkers, and they compete with each other over who has the best platform and so forth.

This means it is a very competitive industry with low profit margins compared with the pharmaceutical sector. With low profit margins, little protection of investment, and

little experience or infrastructure for doing large-scale clinical validation, the traditional sector has really not focused on doing large studies to demonstrate the clinical validity or clinical utility of new tests.

A model where we have weak IP and biomarkers has meant that no one party is responsible for developing the clinical data and the clinical validity of a new test. So we have academic studies and professional advocates filling the gap, often promoting tests on the basis of ad hoc experience.

There is really a disincentive for doing large-scale clinical studies because any one manufacturer who made such an investment and brought a test to market would immediately find themselves competing with other companies. Indeed this issue is exploited by some IVD companies who specialize in being fast followers. There is an industry maxim which says it is hard to be first.

Now, things are changing. If you look at many of the companies in the molecular diagnostic space, what we see is that they are disrupting the traditional business model. Companies are developing tests based on protection of the gene or the association with the disease, and the emerging market for gene expression and proteomic tests is based, for instance, often on strong IP rights and biomarkers.

Many of these companies are seeing, for some of their tests, significantly higher levels of reimbursement than traditional diagnostics. So potentially, stronger IP and biomarkers, if it gives the company a monopoly on the test and it reduces competition, gives them an incentive to generate clinical data. What we are seeing is companies developing tests, offering them on a monopolistic basis through their own reference laboratories or licensing them to another company, who offers them on a monopolistic basis, and companies are starting to compete in some areas on the quality of their clinical data.

I think that is very important. When we think about oversight, it is all very well to think about how we can improve the evaluation of tests. But if companies don't have any incentives to generate clinical data, then there isn't really any point in creating better systems for evaluating what simply won't be there.

Now, having said there may be some advantages to this new business model, clearly it poses some challenges, as monopolies often do. Obviously, many people have expressed concern that tests offered on a monopolistic basis are not subject to the traditional kind of peer review in the field where lots of different lab directors can take on a test, try it out for themselves, see its strengths and its weaknesses.

There is also a concern that where companies had significant investment to bring a test quickly to market that there is a danger that it will make overblown claims for its tests too soon. We have seen a number of companies in this kind of field where such concerns have been expressed.

That is not to try and say that all companies that are developing tests in this business model are bad players, but in the absence of an effective oversight mechanism, we don't actually have a way for patients and doctors to distinguish between good players and bad players.

So this is a rather provocative slide entitled "Six Reasons to Require Pre-Market Review for Laboratory-Developed Tests as Medical Devices." I hope some of the kind of issues there are fairly straightforward. Obviously, they can pose the same risks as tests. Laboratory-

developed tests are big business. Leading companies are bigger than many kit manufacturers. Even for the small laboratories, they don't get a CLIA exemption, so why should they receive an FDA exemption.

It is clearly possible to do pre-market evaluation of laboratory-developed tests because we have the example of New York State, where many, many, many of the LDTs in the U.S. are subject to pre-market evaluation through the NY state lab regulations. We have the example of FDA regulating and evaluating laboratory-developed tests.

It is also clearly the international trend, if you look at Europe and Australia, and we do have this issue around the business model for reference lab monopolies, which might in some senses pose a particular kind of risk.

So there are lots of different reasons why we might think about regulating laboratory-developed tests as medical devices. But we still have this concern that maybe we might be overreacting. Do we really want to apply statutory pre-market review to all laboratory-developed tests or is it in fact unduly burdensome. So, does one size really fit all. This issue obviously is particularly pertinent in the area of rare disease tests.

So maybe what we need is a range of alternative oversight options. You can really see the implementation problems we have had with trying to deal with the recommendations from successive committees has come up against the issue of how to balance evaluation, innovation, and access. The lack of clarity, I would suggest, on the respective roles of different gatekeepers; so, what is the role of reimbursers, what is the role of clinical practice guidelines, what is the role of the FDA.

We also have the issue of FDA resources, or the equivalent regulatory agencies in other countries. Of course, we have industry reluctance for any kind of enhanced oversight.

So, can we have our cake and eat it. Is there some way that we can develop some kind of more comprehensive system of evaluation whilst ensuring adequate protection to the public and encouraging innovation.

I want to suggest that there is a number of solutions. These are ideas that have come out of our research with stakeholders right across the spectrum from industry through to FDA, patients' groups, et cetera.

One of the solutions is to focus pre-market review on truth in labeling. Another one is to have a far greater emphasis on post-market controls and clarify the role of different gatekeepers. Our research has shown some quite strong support amongst many stakeholders, although not all, it should be said, for the idea that pre-market review should be focused on truth in labeling.

The issue of the role of other oversight mechanisms and the role of other gatekeepers is linked to the idea of responsive regulation. So the idea that state agencies, whether it is the FDA or its equivalent in other countries, are not the only people who have a role in gatekeeping. We need to think about what the appropriate role of all the gatekeepers are.

Although your draft report doesn't mention the idea of responsive regulation by name, I think it has a very cogent analysis of different kinds of compliance mechanisms, ranging from mandatory to incentive-driven, to voluntary and informal.

Clearly, when we think about the three core functions of regulation, information-gathering, standard-setting, and enforcement and compliance, we can see that there are many ways in which a whole range of bodies, organizations, and gatekeepers can be involved in the

process of oversight.

So the crucial issue that I want to explore now is really around this issue of providing accurate, comprehensive information to doctors and patients and the way in which we can use oversight to improve that. I will focus a bit on pre-market review, as well.

What can IVD device regulations do. We think that they should be primarily focused on pre-market review of analytic and clinical validity. They should set clear evidence standards for market entry. They can also monitor performance in the post-market environment. Most importantly, they can ensure truth in labeling and truth in promotion. These are the core functions that we think can be carried out by IVD device regulations.

But there are many things that they can't do. They can't deal with ethical and social issues such as genetic discrimination. They can't regulate clinical practice issues such as informed consent, and they can't evaluate clinical utility, which our research suggested most stakeholders was best left to health technology assessment and clinical practice guidelines.

If we focus on pre-market review and truth in labeling and truth in promotion, again I come back to this idea of regulation by information disclosure. We are trying to balance the need to protect the public with a desire to encourage freedom of choice. This is a kind of minimal approach which reduces the regulatory burden and passes on the responsibility for risk management to doctors and patients, allowing them to make informed choices about when and how to use a test.

But our research also suggested although there was support for this kind of idea there were concerns that doctors generally will not have time to do detailed surveys of the literature on new tests. In fact, we need to think of ways to simplify the information that we provide to doctors and patients, rather as we sometimes do, for instance, with food labels so there is kind of a clearer and easier to understand guide to the quality of evidence that supports new tests.

A scheme like this might take the form of kind of a simplified schema which could indicate where a test lies on the development spectrum from research to well established clinical use, or it might be based on evidence-based medicine standards as developed by the Cochran Collaboration.

Linked to this issue is the whole question of expanding the definition of a label. If we are going to focus on truth in labeling, then we really need to think about the issue that tests are quite different to drugs. With a diagnostic test, in general it is the laboratory, not the patient and doctor, who see the label. So device regulators need to broaden their concept of a label and ensure all those that are offering tests meet the necessary information available to clinicians and the general public. So test manufacturers and test developers should be obliged to keep their labels online, where they can be accessed by all, with samples of test result sheets which show reference ranges and so forth.

In the provision of information, the issue of labeling is another area where there is a clear difference between test kits and laboratory-developed tests. At the moment we don't have the regulatory equivalent of a label for a laboratory-developed test. That is a clear definition of the information that LDT developers should be providing to the users of their tests. This is an issue that FDA has started to address in the IVDMIA guidance, but it is clearly one where some more work is needed.

Regulators can also facilitate information disclosure by making public their

device reviews and subsequent post-marketing data. Now, in this respect, FDA is far further ahead on this than Europe. In the U.S., OIVD publishes review summaries on its website. Sadly, such data is currently treated as confidential in Europe.

However, even in the U.S., it is possible that more might be done. I think FDA could maybe make a better job of making the information easier to find and presenting it in a more understandable way.

So, this kind of model of a very minimal approach to pre-market controls. We found support for this idea, but it was predicated on the idea that that would be balanced with enhanced post-market controls. When I say "post-market controls," I mean all those other forms of oversight that exist once a test is on the market. The role of reimbursers here is really crucial.

I think in the oversight debate there has been real concern in the past about health technology assessment and lack of HTA of genetic tests. That has led to initiatives such as EGAP. But I think although that has been very true in the rare disease field, what we are starting to see with tests with a broader application is that health technology assessment is operating as an effective form of oversight and effective gatekeeper.

If we take the example of the Roche Amplichip, it was approved by the FDA but it has subsequently been subject to a series of very critical HTA reports both in the United States and Canada and in Europe. So I think we can see a really strong and important role for health technology assessment emerging now as tests with broader applications emerge.

But equally important is the role of clinical governance. Many, many committees, including this one, have pointed out the need for increased use of and better funding for clinical guidelines.

There is also a role for independent sources of information. We have some wonderful examples and lab tests online, the GeneTest website, and clearly this Committee has spent some time thinking about how those mechanisms could be used to enhance oversight and they can clearly play an important role. We will talk a little bit more about that.

I focused on the role of the FDA as looking after pre-market evaluation, but clearly there is still quite a lot of concern expressed by industry and other stakeholders about an enhanced role for FDA and there is some concern expressed by FDA about having to take on far more work. I'm sure any of you who know Steve well will know that it is not unusual to get Emails from him on a Sunday afternoon, and it is not terribly clear that people in OIVD can actually work any harder than they are at the moment.

So, are there other alternatives to simply expecting the FDA to do more. I come back to this idea of the enforcement pyramid and responsive regulation and the role of other gatekeepers.

Now, a few years ago when your predecessor committee was working on the issue of oversight, there was a discussion in one of the meetings in which Steve pointed out that in fact FDA had been thinking quite hard about the ways in which it could develop more flexible mechanisms, including self-certification, the use of smaller data sets, different data sets. They clearly got a whole range of flexible regulatory tools that they might be able to bring to bear on this issue.

So what we have seen recently in the last year or so is a discussion about alternative regulatory mechanisms. One of these ideas is the idea of a data registry. It has been around for a while now. It has support from a range of stakeholders. It would appear to address

the problem of information asymmetries without placing an undue burden on test developers.

I think it is also very much the kind of idea your predecessor committee was moving towards as they kind of wound up their work on oversight. I think they were thinking very much about the issue of how do you provide independent sources of information on tests.

Obviously, your draft report recommends a voluntary approach based on expansion of the role of the GeneTest website, and you recommend the registration of the lab and the list of tests offered and, in the case of tests not reviewed by FDA but by other bodies, a statement that they have passed through this review process.

Now, some stakeholders have suggested a more comprehensive registry which provides detailed information about the tests which labs provide. This is a useful suggestion, but it raises two issues, I would suggest. The first one is really the issue that, to act as a trusted source of credible information, a registry must be able to guarantee the quality of information on the registry and it must be able to deal with complaints.

So, how do we address this issue of having a registry but having a registry that has some real kind of authority. What I want to suggest is that one of the ideas is that we actually look upon the role of the FDA as a meta-regulator. When I say a "meta-regulator," what I mean is that the FDA has some kind of overarching role that in certain instances it can be the guarantor of quality. It can ensure, for instance, if there is a complaint, that it will address that. Whilst you can have something like a data registry that would in fact be maintained and managed by other parties.

We can see that it is possible for regulatory agencies to act in this way as a kind of meta-regulator when we look internationally. So if you look at the European model for IVD device regulation where tests are subject to pre-market review, it is not done by the equivalent of the FDA, the regulatory agency. It is done by a notified body, an independent third party who carry out the review.

Now, the model that the Australians are developing is that the Therapeutic Goods Administration, the equivalent of the FDA, has also adopted third-party review. So the professional pathology bodies will act as reviewers, but TGA has a role in standard-setting, can step in where there are complaints, and will review tests which are in the highest-risk category.

So FDA has the authority to empower third-party review and has experimented with it in the past, although I know Steve is not entirely satisfied with his experience of it.

Clearly, there are other agencies within the United States who could act as third parties. We already have the example of New York State, who are conducting pre-market review of a very, very large proportion of the laboratory-developed tests that are currently available in the United States.

In the rare disease field, we have recently seen the example of the CETT initiative, which has developed a system for evidence-based introduction to rare disease tests.

So I think there are some parallels here with the report's recommendations for private sector or public-private partnerships to review LDTs which aren't reviewed by FDA. I think the crucial difference here in the model that I'm suggesting is that the FDA retains an overarching role as the meta-regulator.

We still lack a comprehensive system of oversight which can provide doctors and patients with creditable, comprehensive, and accurate information on genetic tests. The new wave of consumer genetics companies makes this issue all the more pressing. We need to find

ways to balance innovation and regulation. The idea of regulation by information disclosure, the concept of responsive regulation and its tools, including a more flexible approach to pre-market review and a greater emphasis on post-market controls, are perhaps some of the ways we can enhance patient protection whilst nevertheless encouraging innovation.

I just want to mention briefly that David and I are part of a bigger research team with colleagues in a number of places in Cambridge. Thanks a lot.

[Applause.]

DR. FERREIRA-GONZALEZ: Thank you. Thank you very much. That was a very interesting presentation and very comprehensive.

We are going to open it up for questions, and I guess I will take the prerogative, being the chair of the session, to ask you a question.

When you show some of the current oversight for laboratory-developed testing, you show a role for the FDA but I didn't see anything about the CLIA or the role of CMS, which does have currently oversight over laboratory-developed testing. I was just wondering if you can comment on that particular issue.

MR. HOGARTH: Yes, that's right. There is a gap in my analysis as well as a gap in the regulations. So I think this is really a crucial issue, and it is a crucial issue for us in Europe as well. We really need to think about this question that has been thrown up in the last year of the ways in which there are overlaps between the two systems of regulation.

If you start to have FDA involved in laboratory-developed tests, how do you deal with the issues of quality assurance and analytic validity, where clearly CLIA already has a role. That is a very important issue that has to be addressed because what we absolutely don't want is to subject anyone to unnecessary duplication of regulatory requirements.

I think that equally applies perhaps in some cases, or you could argue it would apply to companies who are already subjecting their tests to pre-market review by NY State. I think there really needs to be careful consideration of how you deal with what could become a serious kind of a matter of duplication of effort by regulators, which is a waste of the taxpayers' money, and effort by companies when they could be better spent developing useful innovations for patients and doctors.

DR. MELZER: Could I just add to that a little bit?

DR. FERREIRA-GONZALEZ: Go ahead.

DR. MELZER: I think, especially in Europe, the emphasis on self-certification and review has been mainly on the analytic level. For most purposes, the analytic validity is really very good. The problem for clinicians is the clinical evidence. I think that is why our analysis is focused mostly on the clinical evidence. So clinicians in Europe are getting tests marked with a CE mark that certifies compliance with European directives but it actually means nothing for clinical evidence. That is also very misleading, to have tests that are given a mark of approval on an analytic basis but their clinical validity might be completely absent or claims might be completely misleading.

DR. FERREIRA-GONZALEZ: Francis.

DR. COLLINS: I want to thank you both for a very clear elucidation of the issues and some thought-provoking recommendations. That was really quite a thoughtful presentation.

With regard to the principle that you are putting forward I think as one of your

main themes, namely the disclosure of the evidence that underlies the clinical validity and utility of tests in a fashion that is possible for both patients and providers to be able to get access to that, that does seem to be a general theme that many people are beginning to embrace quite strongly. I guess it is the idea that sunshine is the best disinfectant if you are trying to make sure that this is done in a credible way.

With regard to the specifics of how to do that, I wonder if you could reflect, based upon what you have seen of various models, about the value of having such a registry be purely voluntary versus having it be mandatory in some form or another. Is there a comment you would like to make about those alternatives?

DR. MELZER: Stuart is averting his eyes.

[Laughter.]

DR. MELZER: Reflecting on the European position, the European position requires test-makers to assemble information supporting their claims in a dossier that can be inspected by the regulatory agencies if concerns are raised for tests at the lowest level of vigilance. So there is some specification of what should be in the dossier.

There is a major problem in Europe in that currently this is regarded as commercially sensitive and not made public. Certainly, all our stakeholders in Europe are saying this is absolutely unacceptable. We have the situation where people are marketing tests and refusing to say which snips they are actually testing, what evidence it is based on, and how the results are generated.

But of course, if you look up many of the companies that are marketing in the U.S., you cannot find on their websites which snips they are testing or what the evidence is based on. So there seems to us and the stakeholders we talked to a way forward which involves, say, the central regulator of this country, the FDA, specifying the headings under which information needs to be structured and made available on the Web. I'm talking about clinical information.

You might think of that, for example, as identifying which groups of patients and what the purpose of the test is, saying exactly which snips are going to be covered, what the scientific basis is, and so on. Then the idea would be that that would be made available on the Web.

There is no sign that that is happening voluntarily. Many companies are being very secretive and there is a tendency to perhaps compete on including as many diseases as possible, starting this new wave of genome-wide pseudoanalysis, and so on.

I think many of our stakeholders felt that we need to get to a position where companies don't compete on clinical evidence, on the number of snips they put in, they compete on other things. It would be as crazy as the airlines competing on safety. They don't compete on safety. They all collaborate on safety. We need to get to a point where the clinical evidence part is available. There is no sign that that is happening naturally in Europe yet.

I think there is another thing that worries clinicians, which is it is all happening now. I noted in your report that you are talking about a five-year window of voluntary registration. The worst abuses are going to happen in the next five years. Coming back in five years will leave clinicians with very little to look at on the Web while this revolution starts.

We have had patients, for example, who have taken very poorly supported tests for prostate cancer who are now considering having their prostates out. So it is pretty serious for clinicians. They are being asked to advise patients, and there is nothing on the Web and no sign

of the success of the voluntary kind of ethos of disclosure.

MR. HOGARTH: One of the issues about going for a voluntary system would be why wouldn't you make it mandatory? What are your concerns. Is it that you think it is suddenly going to impose too much of a burden and that people won't be able to provide this information quickly, and so forth.

If your concern is that, one alternative approach is to make it mandatory but have a risk-based prioritization of who you expect to comply with it first, if that is your concern.

I would suggest that it really is not unreasonable to expect someone who has developed and is offering a clinical test to be able to summarize in an evidence dossier the analytic and clinical validity of the test, its likely clinical utility, the indications for its use, and so forth. If someone cannot provide that information, then what on earth are they doing providing the test.

DR. FERREIRA-GONZALEZ: Mara and Steve.

MS. ASPINALL: Thank you so much for your thoughtful report. I have a question about when you looked at the molecular model and you looked at the IVD model. It seemed that you focus on a small number of companies, relatively new companies, that have spent a huge amount of money on the clinical validation, clinical trials, and the science. Have you done comparable analysis on the dozens of existing molecular tests that exist today that are not part of this relatively new generation of different types of tests, or comparable types of tests with different kinds of business models?

MR. HOGARTH: Yes. I think you are absolutely right. Obviously, Mara, there are lots of companies who are developing and commercializing molecular diagnostics, including your own company, Genzyme, who don't follow the business model that I outlined.

But nevertheless, I do think it is a very, very significant development. I think the oversight debate has for too long neglected having an adequate discussion of what emerging business model means or its significances. So I think it is a really important thing to bring to people's attention.

One of the things that we have been thinking about is the fact that historically we have had two big debates going on, one about oversight, one about regulation of genetic tests, and another one about gene patenting and IP and biomarkers. They have been completely separate, generally. In some senses, on the one hand, you are beating companies over the head saying you must provide more clinical data, and on the other hand, you are saying one of the incentives that might allow you to invest more money in clinical studies we think you should have.

We need to think about how those two debates connect up. I think that is a very important idea for this Committee because, of course, you are actually looking at gene patents at the moment as well as developing your Oversight Report. I don't think there are any easy answers there. Absolutely not. Apart from anything else, this is an emergent business model and we don't even know if it is going to work. But nevertheless, it is something I think we need to think about.

MS. ASPINALL: I would agree. I agree very much on the importance, both short-term and long-term, of this new emergence there. But, have you looked at the existing ones that are not in that model and the impact of regulation on the currently existing tests, many of which don't have any IP covering them? Have you looked at the impact of regulation on

those?

MR. HOGARTH: All the companies that we spoke to when we were doing our research, including your own -- I'm very grateful to Bob Yoher [ph] for participating in one of our workshops and speaking to me at length on a number of occasions for probably far longer than he was really happy to.

A lot of the companies that we spoke to don't follow that business model, so we are very much hearing their views as well as the views of other companies.

DR. FERREIRA-GONZALEZ: That is a very good point. Steve.

DR. TEUTSCH: Thank you. This has been very helpful. You talked about getting back to the issue of where in this process you provide oversight and to what level of intensity. I wonder if you could talk a little bit more about the reimbursement level.

If reimbursement were not to occur, if the information that you just indicated should be there, potentially on a mandatory basis, were necessary for reimbursement, then I wonder if you could reflect on whether a voluntary system would be all right. Then, do we have examples where the payers have actually required that so that we know there would be some level of greater scrutiny at that level?

MR. HOGARTH: Probably the most important thing to point out about the whole role of reimbursement as a gatekeeper is that it doesn't function in the area of direct consumer testing. We don't have a health technology assessment for direct-to-consumer tests.

The degree to which some of the companies that have entered the space on a direct-to-consumer basis are doing so because they don't think they would stand a cat in hell's chance of getting approval through a rigorous HTA process I leave for you to speculate on. But I do think that is one of the issues that we need to think about.

I do think the Amplichip is an excellent example of where FDA said, okay, you have told us what you know and what you don't know about the strengths and weaknesses of this test. On that basis, as long as you are careful in your claims for the test and your labeling, then we will allow you onto the market. Reimbursement then took on the role of having a rigorous assessment of the clinical utility of the test.

I don't know if I'm answering your question.

DR. TEUTSCH: No, you are. That's fine. I think EGAP is trying to tackle at least the direct-to-consumer products as well, at least to see if they can. So there is nothing intrinsic in an HGA process that limits it to reimbursement tests.

DR. MELZER: Could I offer a couple of comments? The situation in Europe is rather different because of their nationalized health services, which have a much easier role in reimbursement. But there is a bit of a catch-22, which is some of these tests really do offer real clinical advantages and there is nobody there in the gap between discovery and getting all the clinical cost benefit data to the point where reimbursers might shell out.

Many of these tests will have some components that are under patent and many of the other markers not under patent. So we are going to have a real lack of incentive to do the cost benefit and cost utility studies. I know you flagged this up in your report as an area for public-private partnership, but it would be pretty awful if the reimbursers were so strict that some of the fruits of this wonderful discovery don't come through to patients.

DR. FERREIRA-GONZALEZ: Marc.

DR. WILLIAMS: I have a couple of comments, one related to what you just

talked about. I think it is an issue that has been raised in other contexts about pay for evaluation. You release something out into the marketplace and let the evaluation take place in the context of a post-market and then make a decision as to whether or not there is utility to support the claims. That has been promoted in some ways.

I think there are a number of us that are concerned about, once something gets out there, trying to reel it back in can be a real challenge when clinical practice actually changes, although given the rate of change of clinical practice perhaps that is not a concern. But I would be interested in a reflection on that.

The second comment relates to some of the international aspects that I think, as one of the people that has been working on this report, we didn't have perhaps as broad a view. Not to say that I want to take on the world here, but I think that the point that you made about redundancy and too much regulation or duplicative regulation does have an impact in the sense that a number of these companies are in fact offering these tests in different markets.

In some of the rare disease tests, we have international aspects where the only laboratory that is doing it may be in Italy or Norway. How do we get samples from here to there. I know that some European laboratories have actually undergone CLIA certification so that they can actually provide these tests within the legal construct of the United States healthcare system.

So it seems like perhaps it should at least be reflected in our report that it seems that there are reasons for international discussion so that we can at least achieve some relatively common goals and possibly even look at how the regulators in different places may work together to ease some of the regulatory burden for companies that want to bring things worldwide. So I would appreciate comments on that.

The question I have is that, obviously in the context of this particular discussion, we have been talking about genetic tests, as is appropriate since that is the name of the report, but one of the things we always struggle with in these discussions is whether genetic tests in and of themselves are exceptional compared to other types of tests.

That wasn't something that you addressed in your presentation, and so I guess I would just be interested in your opinions as to whether there are reasons in regulation to treat these types of tests as exceptional.

DR. MELZER: Yes. Post-market review, test review and marketing and then testing it in situ, that is going to be pretty difficult for these predictive markers. Many of them are predicting outcomes late in life and so on. The actual model of how it would be released and how it would be used is going to be pretty difficult to work out.

There is also an issue of where research ethics come in. It has been suggested that some companies are actually using the samples that the public are sending in for testing as research fodder without any kind of institutional review board and without flagging out to people that that is what their samples are actually being used for.

So it is actually quite a difficult situation. I don't think we have any experience yet for these new complex disorders, and I guess we are going to have to see how systems could help generate evidence. It seems, certainly for prediction, to be more an epidemiological problem rather than a health service problem. But there may be specific niches that people are going to come up with wonderful clinical applications that we can't even imagine. That will have to be worked through.

Its national aspects. I'm sure you are very aware that these are before the

Harmonization Taskforce. It does look as if, under the constraints that the various major markets have, you are grappling with identical problems in moving towards kind of similar solutions.

The problem in Europe, obviously, is that we are a loose confederation of countries with much less central authority, much less of a track record of authority than the FDA has. So the whole system has been built on the central regulator reviewing these secondary ones, being a meta-regulator. It is very much seen as a first stage, so people have been thinking about it as an incremental regulatory regime, starting with some very simple that could work across Europe, and then gradually racking it up.

So within the confines of the "real politick," I'm sure there is an enormous scope for harmonization. The companies we talked to seem to be very thirsty for that. They do not want to have to produce different evidence for different markets. I'm sure that a common evidence requirement could be arrived at fairly easily.

What was the third point? Right, genetic exceptionalism. Our team has endless debates about this, and I'm sure you have had it. How can I summarize it. It is true that many other tests have very similar characteristics, if not all the characteristics, of genetic tests, but genetic tests do stand out for a number of reasons. The first is that the public think they are different. The level of education is very low and the gullibility is very high.

So in terms of "real politick" again, there does seem to be something different about genetic tests certainly in the U.K.. Although the strict regulatory system is harmonized with all other tests, it is just part of the device regulations. There are special committees looking at genetic testing.

I think the second issue is we have had these very recent explosion of results. Now, that is probably happening, or going to happen, in proteomics and so on, and it is going to throw out very much the same issues.

So I would argue as a public health person that genetics should probably be seen as an opportunity, a trigger, to improve test regulation throughout. So it shouldn't be ignored because it is just a test because they are different, but it does offer us a political opportunity to get some of that basic information to doctors and patients so they can make sensible decisions.

It is a really good trigger because it is a bit different. When you get tested, you are revealing something about your family, and in the current environment you are likely to overestimate the importance of that and make decisions that would affect other family members.

Do you want to say anything?

MR. HOGARTH: Just something to follow onto the exceptionalism issue. I think once it comes down to what you are asking the device regulator to do, or the regulator or clinical laboratories, they really have to take their standard criteria for the evaluating the risks of the tests and apply them to the genetic tests and decide within those standard criteria are these high-risk tests, moderate-risk tests, low-risk tests. If people aren't happy with that, then you have to say, well, does the regulator have to redefine its criteria for risk classification.

I actually think that you can deal with this just by using the traditional kind of risk classification schema that the regulators have.

On the issue of international cooperation, I was fascinated. I was speaking to an IVD regulator from Canada recently. They said to me, the problem is we don't actually have time and resources to kind of write our own guidance documents, often. So we just take FDA's and we put a slightly Canadian spin on them."

So maybe Steve should be being paid twice or getting some kind of consultancy, although I'm sure the government would never allow that.

So there are lots of interesting examples of international cooperation that I think are very important: the OECD guidelines on quality assurance for instance, FDA's work with EMEA on voluntary genomic data submissions and looking at very complex genomic data for pharmacogenetic tests and so forth together, and of course, very well established mechanisms like standards development and so forth.

I think it is really important to think about how we can lower the burdens for companies by making more consistent standards internationally.

DR. FERREIRA-GONZALEZ: Thank you very much.

Because of time, we are going to cut the questions at this point. If we do have more time at the end, we can invite back our two presenters. So, hold those questions for the end.

Thank you very much, Mr. Hogarth and Dr. Melzer, for being here today and sharing your insights to inform the Committee.

DR. FERREIRA-GONZALEZ: I would like to invite all the public presenters to the front now so we can start moving a little bit faster.

To facilitate the process of hearing from as many stakeholders as possible, we have scheduled an extended public comment period today to focus specifically on oversight. We can move to the front. The groups that are going to be presenting today are Sharon Terry, David Mongillo, Patricia Goldberg, and Dr. Patrick Terry.

We welcome and appreciate the views of the public. We hope that the public comment process will help us collect information and ideas from a wider sphere of stakeholders and members of the public, and that the input that we will receive will help ensure the soundness of the report and the currency, utility, and feasibility of the recommendations.

We are very pleased today to welcome Sharon Terry, president and CEO of the Genetic Alliance, a coalition of over 600 specific advocacy organizations. The Genetic Alliance held a meeting in September on genetic testing, and Ms. Terry is here today to share the key points that emerged from that meeting.

Thank you, Sharon.

MS. TERRY: Thank you. I will try to truncate. Originally, I think Sarah said 30 minutes, and then a 15-minute discussion, but I'm going to try to cram that together. You have covered a lot of the issues that I would cover as well.

So we convened a meeting in September largely because of what we heard from many, many stakeholders across the board, that we really need a place to come together. We brought together a planning committee. I won't go through them all, but they represented the payer community, various industries from biotech and PhRMA, advocacy organizations, policy thinkshops, et cetera, and certainly the provider community as well.

Our starting principles were that we would put our eyes on the prize. The prize was health. So instead of the endpoint of better diagnostics or increased profits or better ability to treat patients, et cetera, we are going all the way to the ultimate outcome, which was health, and really kept that focus throughout the meeting and used it as the lens and the measure by

which we made decisions.

"Truth-telling" was a phrase that in fact even scared some people when we put it in the title. I think there was some sense that we thought we were pointing fingers and saying that certain people weren't telling the truth, when in fact we were just inviting an opportunity for all of us to say exactly what we meant and thought based on the concept of health as the ultimate outcome.

That led us to looking at transparency around both IP and conflicts of interest. While many people from usually academia and not-for-profits stand up and say "I have no conflicts of interest because I don't work for a company," we really redefined that there and said that all of us have conflicts of interest and that it is important to talk about those from every aspect.

So people identified themselves by their conflicts, whether it was from a university, a not-for-profit or a for-profit, or the government. Putting those on the table to start with allowed us to move past them.

We also redefined IP there. Instead of saying this means simply that I'm a patent holder or that I have some kind of intellectual property that is by law mine, we talked about in a larger sense in terms of what I carry, what silos I like to protect, et cetera, and we tried to move beyond them as well.

We started with an intermediate starting point in terms of content quality. What I mean by that is we did not allow presentations that defined clinical utility and analytic validity and the baseline stuff. We really started at an intermediate point and moved quickly to a very advanced point.

We didn't allow any PowerPoint, which got people away from exactly what I'm doing right now, which is talking to you from PowerPoint, but allowed people to move beyond the canned presentation that they usually gave into a place that perhaps was new and really required that panelists speak to one another. So we used moderators and/or interviewers who would make people actually address one another and the questions before them.

The attendees of the meeting break down this way. I know all these slides are very hard to see from the back of the room. But basically, a large contingent of advocates, government, and biotechnology companies, small ones, were about half the attendees. The other attendees broke down with a large number of laboratories, a fair number of pharmaceutical, healthcare agencies, and academia. There were a couple policy people and a few media people. So we had a good diversity in the attendees, and we really left a lot of time. Over half the meeting was discussion rather than presentation.

I'm going to go through some recurring themes, and I'm going to go through them very fast at a really high level because this audience is very much advanced in terms of the issues. But these were the kinds of things that resonated with everyone as we went through the meeting.

The issue that personalized health care itself is creating tensions in the system, striving toward personalized therapies and interventions, issues around education, ones that you have heard over and over, that we need better vehicles across the board in terms of the public and clinicians, et cetera.

Resource allocation. How are we going to do everything from rare disease testing to international and developing world issues, looking at genetics and genomics as maybe the

great divider or the great convener.

Public-private partnerships were mentioned over and over with very strong support, in order to alleviate some of the pressures on the current system. When we peel back many of the layers, we can go all the way back to the mess that the healthcare system is in and not just diagnostics, for example. So the idea that these solutions are not going to come just from government or just from industry but there has to be some collaborations.

Reimbursement. A lot of focus on reimbursement as the ultimate bar, certainly things that you have mentioned here as well. Do payers understand the value. Is the system right. Is the structure right. Can value-based pricing be sustained throughout the entire system, et cetera.

Biobanks. They are not regulated and they certainly often are silos that are contained by one entity or another and are not shared. They are not in the common, so to speak. How important are they; how should they be maintained; what should we look for going forward.

World health. How can the transfer of genomic technology to the developing world be implemented.

A great deal of discussion on evidence. How much clinical evidence is enough. What are the pressures in the marketplace to bring something to market before the scientific need is established or the validation process is done. Issues that you have also discussed. Can all the tests be held to a single standard in terms of evidence, and then, what can we do about post-market data collection.

IP models. There are certainly other industries, like the music industry, the publishing industry, et cetera, that have faced various challenges to their IP models. What have they done in a flat-earth, long-tailed kind of mentality, and what can we apply to the genomics industry.

Strong support for the passage of GINA across the board, that it was certainly an essential piece in getting service delivery. The CETT model, which again Stuart threw up here a bit, has been successful for rare diseases. Can it be expanded into the common disease and general kind of populations tests.

The role of patients in the advocacy community, essentially often considered the bridge between the scientific community and the public, but not always so careful about the messages they bring forward. The kind of hype that the advocacy community might do. Earmarks and IP in a not-traditional kind of context need to be really critically looked at.

Study design. We need predictable, well-designed studies, and we need to streamline the process through the pipeline.

Regulatory. A discussion of who has the regulatory authority for genetic testing and what should be the role of the various federal agencies. How would they be coordinated so that there was transparency and clarity.

Tensions between the product and the process. Issues around the technology taking great leaps, but behavior, whether it is the behavior of clinicians to implement these technologies or the behavior of patients to uptake whether or not they should eat brussels sprouts, is important.

Again, intellectual property. Where should the pre-competitive bar be. It has been moved back and back and back in some sectors, such as the information that is pouring into

various databases on the federal level from the sequencing of the genome on down, but we haven't seen that mirrored in as many places as perhaps it should be.

An issue about registries. Should they be voluntary or mandatory. What kind of data and who should maintain them.

Medical record aggregation. Would the public support large databases or would there be privacy concerns that might override those benefits.

What should the role of professional organizations be. Should they step up to the plate and do more about bad actors, et cetera.

Costs and values. How to determine the difference between those two.

Risk-based regulation. We should perhaps be looking at tests that pose more risks to society and to patients.

Proficiency testing across the board. People believe the critical role of professional testing was essential to understanding quality control. And, how to increase the proficiency testing without placing undue burden on laboratories.

Direct-to-consumer tests need to distinguish between marketing and testing. How can the public be protected from these fraudulent and exaggerated claims.

Test interpretation. A discussion around clinical utility. Providers of course laying stake to the territory that allows them to do the test interpretation, and then some of the discussions with FDA and others around that.

So you can see there was a real richness and diversity of presenters. We went from the pipeline from research all the way through to delivery of services and looked at all the issues. We came up with a number of conclusions, and I didn't really expect us to because I thought our audience was too diverse. But I think it was helpful that we all did try to keep our eyes on the prize of health and did try to do truth-telling.

The report, by the way, will be about 60 pages long and will probably be out somewhere around January or February by the time we finish editing it, et cetera.

The conclusions were, first, that NIH put more requirements on funding and that there be various standards required along the basic and translational research pipeline so that evidence standards were achieved more effectively. Now, that is a chicken-and-egg because evidence standards are not quite clear, but we would recommend that we would start earlier in the process than later and that the research coming out of the RO1s, et cetera, be able to be evidence that would be useful.

The second one was discourse with and responsiveness from the federal agencies that have jurisdiction over genetic testing. The attendees felt that there wasn't always the kind of responsiveness that would lead to more rapid resolution of some of these issues.

Coordination of jurisdiction and activities of CMS and FDA and other relevant agencies. There didn't seem to be good coordination between those agencies and that is desirable.

Clarity and predictability. The current process is not conducive to a growing or stable marketplace. So, some of the same things that Stuart elucidated in a more deep way.

A risk-based regulatory system is desirable, with a caveat that allowances need to be made for volume. So again, some of the same things that Stuart went over in more detail.

Pretty much unanimous that direct-to-consumer tests need a special kind of oversight. Whether they were actually carved out or whether their risk was considered higher,

there should be some way to address that.

Public-private partnerships as a desirable means for ensuring the pipeline of discovery through from research to tests as effective.

Education at all points.

The need for outcomes data collections and clear evidence bars. That was reiterated over and over and would certainly be something that I think focus should be put on.

That the industry itself should have the means to rid itself of bad actors but that regulation should not be based on bad actors, and the balance again between understanding what the industry in general needs in terms of risk to patients, to health, et cetera, versus the kind of various outliers that we have looked at.

A mandatory registry must be established and managed by either a public-private partnership or by a government agency.

This led to a number of action steps for the people assembled. No one entity took control of any one of these steps. Certainly, Genetic Alliance has stepped up on a number of them.

Advocating for enhanced CLIA was important. Promoting a mandatory registry. Convening a summit -- and there were actually several summit ideas -- on reimbursement issues. Another summit on evidence and outcomes data. There was actually another third-party review summit suggestion, and more recently, a suggestion on a summit on models, the models that are emerging lately around genetic testing regulation.

We need to explore the concepts of risk and how we are going to actually divide the lines between various levels of risk. We need to educate Congress, patients, clinicians, and there are probably many more groups we could put in there as well.

We need to work to pass GINA. We need to further examine global perspectives and bring that perspective in more, and we need to report back to the Secretary of HHS or his representatives. We did meet with Greg Downing on October 31st.

In addition, Genetic Alliance has gone on to work with a number of the various entities in the space, like ACLA, 21st Century Medicine Coalition, et cetera, to work together to understand what does this landscape look like and what would be the best solution for us moving forward.

That's the end of my report. Thank you.

[Applause.]

DR. FERREIRA-GONZALEZ: Thank you very much. We didn't mean for you to rush like that.

MS. TERRY: It's okay.

DR. FERREIRA-GONZALEZ: Any questions or comments? Julio.

DR. LICINO: Hi, Sharon. I have a question about is there a difference of opinion. It is really an umbrella group for like 600 organizations, as you were saying. So in terms of direct-to-consumer testing, which you can find susceptibility alleles but you can also find actual genetic disorders, what is the range of perspectives that you get from this very broad group that you have?

MS. TERRY: I should first say that this report does not reflect the 650 advocacy groups. It more reflects the people who attended the meeting. So it is not an advocacy report, really. It is a general report of the people who attended the meeting.

But to answer your question about what do consumers feel about direct-to-consumer testing, my 650 advocacy groups aren't terribly worried about it because it isn't their issue. They are really very much into single-gene disorder testing, and they don't care how they get it, basically.

More broadly, the consumers we represent in terms of public discourse and dialogue have a range of concerns. Most of them are somewhat concerned that what they are getting is what they think they should be getting and so they want to see oversight.

There is, though, certainly a fairly robust subsector who believe they should get any information they want in whatever form they want, and follow more the Amy Harmon article that just came out in The New York Times, having been tested by 23-ME and think that it is important to have this information even if, right on their website they say, tomorrow this could be completely different results. So we do see a huge range.

As a disclaimer or disclosure, I'm on the board of DNA Direct, for example, which really does more direct through health providers, but all of them want to see more clear oversight, more clear guidelines. I'm not sure that they know what they are asking for because I think as we move into that space we will have a better idea of how do we protect the public from fraudulent results and also how do we allow the industries that might result to move health forward, not so much this sort of more recreational stuff.

DR. FERREIRA-GONZALEZ: Kevin.

DR. FITZGERALD: Just picking up on that, Sharon, I noticed you have a couple of points about education. Maybe involved in that was also public engagement, but I'm just wondering if that came up as a specific thing and what sort of methodologies or approaches were recommended.

MS. TERRY: We decided at the meeting not to spend a lot of time on that because there are some good efforts. NCHPEG, whom you heard from this morning, I think is a good effort in that regard. Genetic Alliance itself has a whole wing devoted to quality information and public engagement.

What we got from the meeting, I think, was more a sense of we often silo audiences and we don't understand that these audiences in fact are overlapping. So the public and clinicians and researchers and test developers, et cetera, probably need some common forums where they can have these kinds of discussions. So the idea of more of these summits where people could come together in a cross-talk kind of way resulted.

For example, this report will be written in lay language with lots of glossaries, et cetera, so that the public can read it and not just isolate it to one sector of the stakeholder community.

DR. FERREIRA-GONZALEZ: Joe?

DR. TELFAIR: Thank you, again, for a good report on this. One of the things that struck me, though there is more than one thing. I know I have to be short, so I will.

It seemed that a lot more questions were generated than answered for this. Is that correct? So in your report, if you are going to direct that to consumers, then I'm assuming that there is going to be some degree of summation that is going to go out?

MS. TERRY: Yes.

DR. TELFAIR: All right. The second point, though, is that there seems to be a great deal of overlap with both your conclusions and your action items. For example, you have

three in a row that actually overlap significantly one another, the registry, the reimbursement issues, and then evidence and outcomes data. If we pick one, it depends on what the context is.

I'm just wondering, in the discussion, if you can say a little bit about what was the thinking of demarcating those into separate summits as opposed to looking at those issues as they relate to one another. I'm thinking more in terms of level of application and level of use and practicality.

MS. TERRY: Those are excellent questions, Joseph. Basically, the reason that they sort of fell out of this in separate buckets were because of the depth of the summit that was proposed. In other words, certainly you couldn't do any one of these things without the rest, and there would be some presentation on all the rest. But there would be a great deal of depth.

For example, on reimbursement issues, to go very, very deep on that issue, bringing in the other issues around evidence. Evidence has got to be part of that, but then a whole other summit that would be dedicated to just looking at how do we, from the beginning of the research pipeline to the end, post-market data, look at evidence. So they certainly do overlap.

Our sense with things like this is that if we keep having very broad discussions we don't seem to go down in the weeds as much as we need to. We also don't seem to capture the right audience. Very often the reimbursement community doesn't come to these kinds of meetings. So I think we have to start marking some out and going in depth, but not ignoring the fact that there is a breadth of stakeholders needed at each of these meetings.

DR. FERREIRA-GONZALEZ: Mara and then Francis.

DR. COLLINS: Thanks, Sharon, for a very nice summary. I was fortunate to attend a chunk of the meeting. It really was a very useful exchange because of the way the format was set up. People were not talking past each other, they were really interacting. I think you got much deeper, therefore, into the sense of what needs to be done than often happens.

I want to ask you again about this mandatory registry, which came up in the previous discussion as well. Again, I think the sense of the Truth-Telling meeting was that this was so important that it ought not to be left to chance.

Do you want to say anything about the general sense of the group, if there was any worth reporting, about exactly who should run that registry or what combination of organizations should run that registry? Because, obviously, that becomes a critical question.

MS. TERRY: So while that wasn't exactly nailed at this meeting, and we had a couple of times we could go through that we had a panel discussion about issues like that. We also had a wonderful debate dinner, sort of in the European style, that in fact Stuart moderated. We needed a guy with a British accent to do that for us. Or, not British, sorry.

The sense we got there was that it probably had to be a federal agency, it probably had to be FDA, although again it wasn't nailed down, so I can't say that. If it was a public-private partnership, it might be a professional society or a coalition of laboratories, et cetera, that could be involved in that.

It didn't seem to the assembled masses there that a completely voluntary registry would result in the kind of data that we needed, and that some of that data is already available, although hard to get, and just needs to be expanded on.

So [it could] be married with something like GeneTest, that has begun a Mendelian disorder registry, and then moved over to FDA with some input from NIH. There

was a lot of discussion around how those partnerships could be put together. But the sense was, if this is completely voluntary, it won't get done, which is what we know about all our kids, right?

[Laughter.]

DR. TUCKSON: By the way, as we get to the next point, I just want to make sure to highlight some of these key issues for the Committee, especially for new folk. This is why we have public comment. We had a discussion in our first draft of this and we said, in our initial outreach, the establishment of a voluntary system of genetic test registration through a public-private partnership. Now we are hearing that some of this feedback is perhaps that there should be a mandatory one.

I want to make sure that you ask the right questions to feel good that you understand why they do that. You may well decide based on this kind of feedback to change your original draft report to reflect this kind of input. So I'm highlighting this as one of the key issues that we are looking for as we go forward.

DR. FERREIRA-GONZALEZ: Sharon, let me further comment on these action items that you have and where you promote a mandatory registry. Was that a sense of most of the members? Were there any dissenting members? What was the sense of the group? I'm sure there were different views. What are the other views from the other groups that do not actually endorse a mandatory registration or a system at all?

MS. TERRY: Right. This was the sense of the meeting, at the end of the meeting. Now, certainly, some people had gone home. They didn't get to give their sense. I read these items out at the end and said, "Does anyone object to these items?" No one there objected.

Now, the report will be published, and people, the same way it is happening here, can comment on the report.

Would I say when the whole 200 were there would everybody have raised their hand for a mandatory registry? No. But the vast majority did think that a mandatory registry was probably the only way a registry truly would work.

The other thing I should say here is because we were truth-telling, it wasn't like "Do you want a mandatory registry?" Probably most people don't want one. They don't want a registry at all. They just want to get a good test and go home and know what they have.

But would a mandatory registry give us the result of leading to better health because of A, B, C, D, and all the way to Z? Most people said yes. But again, not unanimously, and like Francis' question, not nailing who exactly is going to run this thing.

DR. FERREIRA-GONZALEZ: Were there discussions on the value of that kind of registry?

MS. TERRY: I'm sorry. Say that again?

DR. FERREIRA-GONZALEZ: Were there discussions on the value of having a registry?

MS. TERRY: The discussion was on the value, whether or not it should be mandatory or voluntary, and who would run it.

DR. FERREIRA-GONZALEZ: Were there any discussions or any comments about genetic exceptionalism?

MS. TERRY: Yes, there was. The discussion pretty much went like what you

just had before in terms of there are some extraordinary things these tests are doing in the sense of moving into a space we haven't regulated before in a particular way. There are also issues around risk, but there are also issues around the fact that these tests are very much like other tests. So the real intrinsic need here is to look at the value of the test, the risk of the test, et cetera, and not so much just chop something off because it is genetics.

MS. ASPINALL: That was part of my question, which is the issue that you talked about in terms of, on the action items, exploring the concepts of risk, because that is something that in various different ways has been part of many of the proposals and much of the discussion, as well as the issue of relative harm. In the EU, there is different regulation. Is there a different amount of harm. Maybe we will get to that from an international point of view.

Can you talk a little bit more about what the concept was about risk, risk profile, and how even at the most basic level now you would think about apportioning risk and potential harm or potential opportunity based on your report?

MS. TERRY: This was another area, and the reason one of the action items is to explore risk is we didn't have enough time there, nor did we have the right experts I don't think, to actually look at risk and to look at it not just in the field of genetic tests but across medicine, understanding it both from the clinician's side, the patient's side, the test developer's side, or the device developer's side.

I think what we saw there in terms of understanding risk was every opinion, from "There is nothing here, there is no risk, and so why has this been pulled out and treated specially?" in the case of, for example, IVDMIAs, to people saying there is a great deal of risk, including some of the advocates who aren't terribly much proponents of new technologies, et cetera.

So what we felt like is we really need to peel back people's complicated and confounded ideas about technology, that technology doesn't inherently mean more risk; about deliverables in terms of decision-making and in terms of life-altering matrices that you might have to go through; in terms of the complexity of algorithms and formularies that bring us to another point; in terms of what a clinician does or doesn't understand. Again, does a clinician understand what it means that my body mass index is too high versus a really complicated test that is done with multiple genes and an algorithm.

So there was no clarity, in my mind, at this particular meeting around this issue, which is why we pulled it out and said we need to really talk about how we are going to assess risk going forward.

DR. FERREIRA-GONZALEZ: Gurvaneet's turn.

DR. RANDHAWA: Thanks. I was going to ask this of Stuart and David, but I think, since you raised the issue here, we can discuss it now. I want to explore further the concept of registries and mandatory data submission.

I'm not quite clear yet as to if you are thinking the registries are there to ultimately improve health. That means we are thinking of linking clinical data and outcomes data with the lab data. There are several issues entangled here. One is of course the data submission by the lab developers prior to approval, getting that in there. Whether or not there are clinical outcomes dealing with that, we already know that is not the case. That wouldn't solve that problem.

Then, after the tests have undergone approval, they will be used in the real

clinical world, where the clinical conditions are in different databases and not really under the control of lab developers.

Then there is a third issue of where do you draw the line between a genomic test or a biomarker and another diagnostic test.

So I'm going to think this through and see how are we going to get a registry that will be having pre-market data, post-market data, health outcomes data, and be mandatory. Can you shed some light on that?

MS. TERRY: No.

[Laughter.]

MS. TERRY: Gurvaneet, those were also issues that were discussed at fairly long and lengthy discussions that didn't lead to a lot of clarity in my mind, either. There are going to have to be some dividing lines, and there is going to have to be an attempt made to get some clarity around this.

The very simplest things could be just that the molecular tests are in a registry like gene tests and that is blown out or expanded on in terms of what labs are doing those tests, whether or not they are CLIA-approved, and whether there has been proficiency testing, all the way to what probably in terms of health would be much more valuable, and that is an aggregation of all the data that you just mentioned.

I think there are lots of other issues inherent in the problems around that, including the fragmentation of our healthcare system, and ones that we probably can't overcome in terms of dividing strict lines between the various kinds of tests and evidence. So I don't have an answer to that, except to say that I think we should wrestle with the question. I think as we do try to integrate genetics into medicine and also improve our healthcare system, we shouldn't just accept that it is fragmented and broken and so we are going to stay broken here, too, but maybe have this field blaze a pathway into the brave new world.

DR. FERREIRA-GONZALEZ: Thank you, Sharon, for sharing these important points. I'm pleased to see that many of them echo some of the findings of our Committee.

Our next speaker is a friend to our Committee whom we have seen before, Mr. David Mongillo, who is the vice president for policy and medical affairs at the American Clinical Laboratory Association.

DR. TUCKSON: As David comes up, I want to mention Amy Harmon's name came up from The New York Times. I do want you to know two articles ago I did call Amy Harmon up and invited her to come, just to be able to meet someone who was doing such terrific, incredible work. She was very eager to do it, but checking with her editors, they wanted her to finish her series before she entertained such a thought because they wanted to be very careful about separating those things out.

So she is going to eventually, I think, accept our invitation to come and meet her and just hear a little more from her about the stories and the folks that she has met and the impressions that they have left. But it will just be whenever she completes this little series of stories. Anyway, I just wanted to make sure that you knew that we had reached out to her.

MR. MONGILLO: Thank you, Andrea, and thanks to the Committee for allowing us the opportunity for public comment. My comments will be brief, partly because we are still digesting the draft report and partly because you have had a very full agenda with a lot

of complex topics. It is not long before everybody gets a chance to go home here.

We think this is an important report. We believe it is critically important and it is really going to serve as a roadmap -- which is, I think, a term that Dr. Tuckson had used when he identified the importance of this report -- a roadmap, really, for the future of genetic testing oversight, which has so many implications for so many components of 21st century medicine.

We want to comment on three what we think are key areas of the report. The first has to do with the oversight role of the federal agencies. We certainly share the Committee's goal to bring the full promise of genetic and molecular medicine to the healthcare system while incorporating the highest quality diagnostic tests. As such, we agree and can work with the Committee to gain consensus on many of the report recommendations.

However, to ensure continued innovation in laboratory medicine and to provide continued patient access to laboratory services, it is critically imperative that CMS, as the agency responsible for CLIA, the Clinical Laboratory Improvement Amendments, continues as the lead agency responsible for the oversight of laboratory-developed genetic test services.

That is in no way suggesting that there isn't a very critical, clear role and definitive role for FDA in this process. In fact, FDA should have a significant role. They should be involved with CLIA in reviewing clinical validity claims and promotion of claims for certain high-risk laboratory-developed genetic tests.

We heard earlier that there were some recommendations made about some of the issues and concerns that people have identified in this area. We think there can be models, and we are posing models, that would deal with the information disclosure, independent validation, enhanced QA and QC, and enforcement if the claims are not met.

As the report stressed, interagency coordination is key and fundamental to ensure that this oversight is least burdensome and does not place unnecessary or duplicative regulation on clinical laboratories providing these genetic test services.

ACLA supports the report's additional recommendations for HHS to convene a workshop with the relevant agencies, as well as stakeholders, to provide input into the development of a risk-based framework for the regulation of genetic laboratory-developed tests, and encourages and supports the development of new and transparent models for private sector or public-private partnerships.

I have heard ACLA's position characterized as sort of the JUNC, "Just Use Normal CLIA" approach, and I want folks to realize that we really are not saying that. We really are proposing models that really incorporate some really, we think, innovative interagency coordination and some, as I said, opportunities for full disclosure, full transparency, third-party reimbursement, enhanced quality assurance and quality control, and enforcement.

The second area that we want to mention briefly is the implementation and timing of the report recommendations on interagency coordination. To allow for a well reasoned and orderly regulatory process, ACLA urges the Committee to include one critically important stipulation in the draft report, namely that the report recommendations should be implemented and understood before the FDA's IVDMIA guidance is finalized or its ASR guidance is enforced.

The benefit and information to be derived from these well thought out recommendations will inform and therefore should precede further guidance and regulatory action.

Finally, the section on effective communication and decision support is particularly noteworthy. We face a critical dilemma for healthcare delivery in the 21st century. Genetic and molecular medicine will revolutionize the ability to capitalize on preventive medicine and target therapeutics but will also become increasingly complex in nature and more available through electronic communication directly to the consumer.

Clinical labs play a critical role in healthcare delivery by allowing for the rapid and timely utilization of health information by providers. The reach of laboratories into physician offices and hospitals by means of health information technology is unparalleled.

ACLA pledges its support in working with the Committee and other interested entities to ensure that clinical decision support systems effectively communicate the appropriate information to providers and consumers in a timely manner and with the necessary level of information to make informed decisions about effective health care.

We are reviewing the full draft report. We plan to provide written comments by December 21st. We thank you for the opportunity to comment and look forward to working with you and the agencies to finalize this process.

DR. FERREIRA-GONZALEZ: Thank you, David. Any questions for David, or comments?

[No response.]

DR. FERREIRA-GONZALEZ: Thank you.

[Applause.]

DR. FERREIRA-GONZALEZ: Our next speaker is Ms. Patricia Goldberg, who is here today representing the International Society of Nurses in Genetics.

MS. GOLDBERG: Good afternoon. I'm Patricia Goldberg, speaking for ISONG today, the International Society of Nurses in Genetics. Our membership spans six continents and represents nurse clinicians, nurse educators, and nurse researchers. ISONG is a specialty nursing organization dedicated to caring for people's genetic health through excellence in the provision of genetic healthcare services by fostering the professional and personal growth of nurses in human genetics.

My brief remarks are part of a longer statement that will be submitted in December responding to the Committee's draft report on the U.S. System of Oversight for Genetic Testing, A Response to the Charge of the Secretary of HHS.

ISONG is enthusiastic when there are advances in genetic testing that our membership can use to improve health care for our patients and the public in general. We consider the work of the SACGHS as a serious public health endeavor and the draft on the genetic testing as a valuable contribution that represents a work in progress.

We take seriously our commitment to our patients and the public's right to the highest quality genetic health care. Patients expect us to ensure that the information they receive is accurate, valid, reliable, and truly useful as they struggle to make informed decisions. We hold this public trust as the highest measure of our success as nurses in genetics.

It is this responsibility that drives our concern regarding the lack of evidence and clinical validity and clinical utility of the genetic tests that are being advanced as useful for common disorders such as diabetes and hypertension.

We suggest that more attention be given by SACGHS to the interpretation and

application of the genetic and genomic results obtained by direct-to-consumer tests for these common disorders. Even though the draft report on the U.S. system of oversight of genetic testing notes that counseling will be given, we believe that there is still too little data on the accuracy, reliability, and true usefulness of the results.

Our responsibility to our patients and the public to provide the highest level of counseling warrants our concern in this area.

Also, as nurses, we are very concerned about the genetic testing being marketed to the consumers outside of the patient-health provider relationship. For example, over the Internet. We urge the Committee to recommend oversight of false marketing of testing aimed to identify made-to-order weight loss plans based on genetic makeup.

Another potentially dangerous area of inappropriate marketing relates to testing to identify the individual's rate of metabolism of drugs so the individual can inform his or her healthcare provider what drugs and dosages to prescribe for them.

We also find the offer of testing for ethnic background to be especially misleading and potentially divisive for families and communities. This medium of exchange requires unique and greater protection for consumers.

ISONG is committed to working towards ensuring that nurses and other clinicians are well prepared to serve responsibly their patients and the public's need for genetic information. ISONG is committed to ensuring that all individuals have appropriate access to genetics and genomic health care, and part of that responsibility includes access to accurate, valid, reliable, and useful information.

Thank you. Any questions?

[Applause.]

DR. FERREIRA-GONZALEZ: Reed, do you want to make a comment?

DR. TUCKSON: I just want to, again, remind folks that was very helpful because, again, in our current iteration of the draft we expressed concern about certain types of health-related genetic tests that are marketed directly to consumers and appear to fall outside of the scope of CLIA. Some nutrigenomic tests, e.g., for caffeine metabolism, and tests to determine the gender of a fetus are examples of health-related genetics tests that are skirting the boundaries of CLIA's authority. There is insufficient oversight of laboratories offering such tests and their potential impact on the public health is increasing concern.

So I think this direction, again, just reminds you that what Patricia has done is to speak specifically to one of the issues that we have highlighted in the report.

DR. FERREIRA-GONZALEZ: Kevin?

DR. FITZGERALD: Yes, I too would like to thank you for the letter and the comments. I would like to dig a little deeper into the specificity, if you are prepared, or to ask ISONG, if they would, to perhaps respond at a later date, if that makes it easier.

But the two areas of specificity which I think we would find particularly helpful would be what sort of oversight regulation did you have in mind specifically, and secondly, even at the end, the very last words you used, you talk about "useful information." Who gets to decide what is useful information? What if the public decides that finding out their genealogy they consider to be useful information. How do we engage in that process of determining what "useful" is and who gets to have input into that?

MS. GOLDBERG: I know they are developing their opinion on that. That is part

of what they will be talking about in December.

DR. FITZGERALD: Thank you.

DR. TELFAIR: I just want to piggyback. That was actually part of a question that I had as well, the specificity, particularly in relationship to the issue of marketing, to whom, and then who makes that decision as to what information actually goes.

One of the concerns is not only just the test and the type of test but also assumptions about the population itself in terms of receptiveness. Then, also, I know that behind all this is this question of duplicity to the public itself and duplicity to these groups. I'm just wondering, given who you all are, could you also cover that?

I respect a lot of what you all do because several of the groups I work with are disease-specific international nursing groups. I know that that is a real concern that they have as well. So I was wondering, piggy-backing on what Kevin suggested, could you also, or will you also be able to address that issue as well?

MS. GOLDBERG: You also want us to address -- I'm not sure what it was.

DR. TELFAIR: There is a question, and I'm actually trying to just be diplomatic about it, because --

[Laughter.]

DR. TELFAIR: What I'm saying in another way is that certain groups are targeted for certain types of drugs in terms of what they believe about that and also in terms of who they are. I was just wondering if in your deliberations related to duplicity, dealing with the issue of duplicity, which seems to be underlying some of the marketing, will you be addressing duplicity as an issue and suggestions on how that could be part of the regulatory process and could be addressed as it relates to specific subpopulations and other groups.

MS. GOLDBERG: I'm not sure, but I will have to ask that of one of our representatives of ISONG that is sitting back in the back, if it is okay. Or she was.

DR. TELFAIR: Knowing who you all are, I have been told that is a legitimate question to ask.

MS. GOLDBERG: Right. We will have to address that with the leadership in time for the December meeting, when they will be submitting another report.

DR. FERREIRA-GONZALEZ: No more questions? Thank you so much again.

Our last speaker, but not the least, is Dr. Patrick Terry, who is here today representing the Coalition for 21st Century Medicine.

DR. TERRY: Thank you. Hello, everyone. Thanks for this opportunity. The Coalition of 21st Century Medicine also wants to thank the overall Committee and the ad hoc working group for putting this report together. I agree with David Mongillo's assessment of the importance and the timeliness of this particular report.

My name is Patrick Terry. I'm one of the cofounders of Genomic Health. Genomic Health's product, OncoType DX, would squarely be in the target area of IVDMIA, so full disclosure on that activity. We are a California-based diagnostic company.

But I'm also one of the founding members of the Coalition for 21st Century Medicine, which is a group that is self-organized around this issue of oversight and regulation and includes industry groups, venture capitalists, academic groups, as well as disease-specific patient organizations, with the concern of balancing oversight and regulation with access and

innovation.

Specifically, I wanted to share with you, as a direct result of the Genetic Alliance Summit, the Coalition has tackled a lot of what was identified as the challenges and the opportunities here for what was, I think, very well described by Stuart's presentation. We fully embrace a lot of Stuart's concepts and proposed solutions. We are crafting a private sector regulatory initiative to present to HHS and to FDA in the near future.

Just quickly to go through it, the proposed framework is based on the following concepts: the importance of advanced diagnostics and their continued development, the importance of reimbursement, the rationale for a revised regulatory framework, which I will speak a little bit more about, and the detailed regulatory and subregulatory approaches that HHS and FDA and CMS can apply and consider moving forward.

The focus of the framework is to identify IVDMIAs as well as multiplexed ASRs that do indeed need enhanced oversight and to clearly define a risk-based approach that includes mandatory pre- and post-market requirements which include a mandatory registry.

The goal of the proposed regulatory framework is to offer specific and detailed regulatory approaches for IVDMIAs and multiplexed ASRs that provide a clear and defined role for CLIA and FDA in a joint framework and that also provide a predictable pathway and a set of expectations for test developers, industry, and the investment community.

The regulatory framework attempts to balance and to achieve a balance between the following principles: innovation, timeliness, transparency, truthfulness, and risk-based regulation.

Finally, in conclusion, the 21st Century Medicine Coalition will formally submit this proposal of a private sector regulatory initiative and will outline alternative model approaches as well as a phased implementation strategy to HHS and to FDA in the near future. The Coalition is more than willing to share that document with the ad hoc working group and to review and discuss further this private sector initiative as you move forward with the finalization of your report.

Again, thank you for your attention.

[Applause.]

DR. TUCKSON: Let me just make sure I got that. That is great. That is terrific. You have a response to this. It sounds like what you are saying is that the private sector is saying it is trying to step up to the plate and diminish the need for more regulation and oversight in this area, that you are responding to the needs.

DR. TERRY: Right.

DR. TUCKSON: So you are going to send that report in.

DR. TERRY: We will submit written comments to the draft report that you are crafting as well.

DR. TUCKSON: All right. That is what I wanted to key [on], that we would get that in time to consider those things that you are doing.

DR. TERRY: Yes.

DR. TUCKSON: Thank you. That was key.

DR. FERREIRA-GONZALEZ: That was the question that I had, are you actually going to respond to the Committee. But as you come up with some final draft or any version of a document that you can share with our steering committee, we will welcome that as part of this

open process that we are trying to establish.

Kevin.

DR. FITZGERALD: Thanks again, Patrick, for that and the work that you are going to be doing. That is great. To follow up on what we have been asking the other panelists, can you give more details about the process you are going to use to ascertain your risk-based platform, your approach, how you are going to identify and delineate risk?

DR. TERRY: There is a variety of model solutions that are being proposed, and there are pluses and minuses to a variety of solutions. I think the context in which we want to present these solutions is not that they are canned fixes but ultimately that these are fodder for further dialogue and debate with the regulators and with HHS.

Part of the solution would be an expert third party review. Another suggestion is return to the post-1976 medical device regulation. During that implementation, there were risk classification panels. [We could] return specifically to a formal mechanism such as that.

So there is a variety of past activities that the agency has implemented in the past to deal with risk and also to add this issue of having important clinical relevance on the table when risk is being assessed for a particular test. So in the absence of the reality of clinical care, you can have a risk-based assessment. But with the reality of benchmarking against trial and error or the standard of care for a particular disease state, the risk-benefit calculation could dramatically change.

So part of the third party mechanism would allow the agency to convene experts between particular disease categories or technical expertise around a test or a technology to participate in a risk-based classification.

DR. FITZGERALD: Great. Thanks very much.

DR. FERREIRA-GONZALEZ: Thank you very much. Any other member of the audience that would like to do public comments at this time?

[No response.]

DR. FERREIRA-GONZALEZ: Before closing the session, I would also like to extend an invitation to other members of the SACGHS Committee that want to be part of the steering committee. You are welcome. Again, we are currently a village, so the more the merrier.

[Laughter.]

DR. FERREIRA-GONZALEZ: So I formally invite you. Just make sure you let Sarah know if you want to be part of the steering committee so you get added to the different rigorous teleconferences that we are going to be having once a week.

MS. CARR: One a week in January. There are five weeks in January.

DR. FERREIRA-GONZALEZ: Oh, I just realized that. Okay.

[Laughter.]

DR. FERREIRA-GONZALEZ: Anyway, I want to thank everybody for very thoughtful, good presentations. We look forward to all your written comments.

[Applause.]

DR. FERREIRA-GONZALEZ: Reed.

DR. TUCKSON: You all know that Andrea has worked her tail off on this thing, so applause to Andrea.

[Applause.]

DR. TUCKSON: Just outstanding.

I can't believe this. Who is the moderator that would have a meeting end early? That is terrific. Joe, you could go on, then. Do you want to make another comment?

[Laughter.]

DR. TUCKSON: I shouldn't have told you to limit yourself.

Let me wrap up a couple of things and then make sure before we close out if there are any other comments from the Committee.

First, let me remind you, although I don't think you need to be, that we developed formal recommendations on the Pharmacogenetics Report and approved the report content. Kevin, terrific job, as usual.

We decided to form this educational taskforce and we have laid out a couple of issues for its charge, trying to ask ourselves the question who is qualified to do what, how do you regulate or oversee who is qualified to provide genetic services, who should get reimbursed, and this idea of looking further into the decision support tools that are available and how they might be advanced and integrated together.

Let me ask who would like to join. The notion is that that taskforce needs to be structured a little bit more. Let me get a sense, before I take any further step. Is it the sense of the Committee in asking this informally such that the new members can weigh in just as those who are rotating off who are still here, Chira, can vote.

Are you the only new member left? Everybody else has abandoned the field here.

Is there a sense that there is an interest to pursue this or am I misreading the Committee's saying you really don't have a lot of excitement about it? Those who are interested, just [indicate].

[Show of hands.]

DR. TUCKSON: There is that sense. Who would like to join Barbara in the effort?

[Show of hands.]

DR. TUCKSON: So Joe, Mara, Sylvia.

MS. CARR: And Marc.

DR. TUCKSON: So it is Andrea.

DR. FERREIRA-GONZALEZ: Okay.

[Laughter.]

DR. TUCKSON: Marc.

MS. CARR: Marc sort of volunteered within some limits.

DR. TUCKSON: Marc, and then which Paul?

MS. CARR: Paul Wise.

DR. TUCKSON: Paul Wise, who is not here. So we have Barbara, Joseph, Mara, Marc, and Paul Wise.

MS. CARR: And Sylvia, right?

DR. TUCKSON: And Sylvia, that's right. Sylvia joined.

So what I think we will do is we will ask the group to consider what it might want to view as its charge and meet informally and then bring it back to the next meeting. That is how we handle that.

Now, for the next meeting, let me let you know. The next meeting is very, very focused on the oversight. This is the oversight meeting. So if you don't like oversight, have the flu or something, don't show through. But it is all about oversight.

However, just to have a little fun, we have invited 23-ME and Navigenics to the meeting, and maybe Amy Harmon will be able to join us at that one. I don't know whether her editors will let her, but maybe she can come to that one as well. So that is really the deal.

Let me ask, are there any points of view that the Committee members would like to express?

PARTICIPANT: Dates for the meeting, just to be specific?

DR. TUCKSON: February 12th and 13th. Now, I'm under strict instructions from Sarah to say to you as you leave that you are demanded to have a happy Thanksgiving and that we urge you to do that. And to the new members, welcome aboard. To the old ones that are leaving, thank you.

Andrea.

MS. CARR: Just to remind the full Committee that you will have a conference call put on your calendars for January 30th, and all the taskforce for the 23rd. They have already got that on their calendars.

DR. TUCKSON: Thank you all very much. Happy Thanksgiving. Great meeting. We got a lot done.

[Whereupon, at 3:48 p.m., the meeting was adjourned.]

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