

U.S. DEPARTMENT OF HEALTH AND HUMAN SERVICES

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

Fourth Meeting

Tuesday,
June 15, 2004

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

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C O N T E N T S

Call to Order

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

Public Health Approach to Genetics

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Director, Office of Genomics and
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Discussion

Discussion of Draft Vision Report

Emily S. Winn-Deen, Ph.D.
Facilitator
Chair, Vision Task Force

Public Comment

Joseph D. McInerney, M.S.
National Coalition for Health Professional
Education in Genetics

Michael P. Murphy, M.Sc.
Gentris Corporation

Kelly Ormond
National Society of Genetic Counselors

Gail Javitt, J.D., M.P.H.
Genetics and Public Policy Center

Report on DTC Marketing Workshop

Alan Guttmacher, M.D.

Discussion

Discussion of Draft DTC Resolution

C. Christopher Hook, M.D.
Facilitator

Discussion of Draft Education Resolution

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

Discussion of Draft Coverage and
Reimbursement Resolution

Cynthia E. Berry, J.D.
Facilitator

Report on NHGRI Working Group on
Large Population Studies

C. Christopher Hook, M.D.

Alan Guttmacher, M.D.

Agenda for Next Meeting

Closing Remarks

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

P R O C E E D I N G S

(8:06 a.m.)

DR. McCABE: Good morning, everyone. I hope everyone had a restful night and is ready to go this morning. Because we will be finishing up at 3 o'clock this afternoon, we have a very tight schedule, and we'll be following that today.

We're going to start the meeting this morning with a presentation by Dr. Muin Khoury on the CDC's public health approach to genomics. Muin has a booklet that -- are you going to pass that around, Muin?

DR. KHOURY: Yes.

DR. McCABE: -- that is really quite intriguing about public health genomics. The committee expressed interest at a previous meeting, and CDC's efforts to address questions about the public health significance of genes and gene variations, as well as on the development and use of evidence-based approaches to establish this knowledge for entire populations, a topic we touched on briefly yesterday.

Muin will describe the CDC's efforts to integrate genomics into public health, and we're looking forward very much to your presentation.

DR. KHOURY: Good morning, everyone. Can you hear me?

There is a book that's going around. Actually, I only have two copies of the book, but each member of the committee will have their own book that's now being mailed as we speak, because we just had them fresh out of the printer last week.

Thank you, Ed, for the introduction and for the opportunity to address the committee this morning. What I'd like to do in the next few minutes is talk about the public health approach to genomics, and I have really three themes. I'd like to set the stage first with a brief discussion of the changing landscape of genetics, because that really affects the way public health does business. The way I captured this here is the concept of the continuum from genetic disease to genetic information. Then I'll move on to describe the public health role in general as an honest broker convening function that's science based, and then I'll describe briefly some of the roadmap activities that CDC and other partner organizations have begun to develop, including a few initiatives. I mentioned some of them yesterday. I'll probably run out of time, but we can discuss those more in detail later on.

Briefly, what do we mean by this continuum business? This is obviously the group not to talk to about this because we've been talking about this paradigm shift. But it seems to me that every time we talk about issues related to genetics, we always fall back on what we know, which is the concept of genetic disease. I'm not trying to minimize that because genetic diseases or single-gene disorders are individually rare, but collectively they account for about 5 to 10 percent of human disease and illness, and they are inherited through the germ line. You have usually a few mutations or many mutations, but in a few genes, and very high penetrance of high lifetime disease risk.

Then all of the environment as we know it, from diet to behavior to chemical to infectious agent, may or may not be there for many of these diseases. We don't know about all of the environment very much. The concept of the delivery of services revolves around the genetic services model, counseling/testing, et cetera. What we are finding ourselves in this new era is that we are moving towards the concept of genetic information that affects all diseases, or the 90 to 95 percent of diseases that we normally don't think of them as genetic, where you have variation in many, many genes and normal variation or variation that puts you at higher or lower risk. Some of that variation is inherited, but you can also detect variation in somatic cells. We talked about this briefly yesterday.

For each one of these genes or their variants, there is a low disease risk, and that's where the role of the environment comes in in a big way. There is really complex gene/environment interaction. The way we're going to increasingly be faced with the situation of integrating this

information in general practice, we'll go back and forth on this but it's an important paradigm shift. It's a continuum, really. It starts with the pure genetic disease to the pure environmental disease, but there is a break somewhere in the middle where the genetic services model will not apply.

Just as an example, for every disease, and I chose the most common human ailment, which is coronary heart disease, you have many single-gene disorders associated with these common diseases. In this case, familial hypercholesterolemia, which is 1 in 500 condition, it's an autosomal dominant condition of the LDL receptor, deficiency is a cause of premature coronary heart disease, but it only accounts for less than 1 percent of heart disease in the population. All of these rare diseases combined probably will account for less than 5 percent of heart disease in the population.

Now, that doesn't mean that these are unimportant diseases. They are very important to the individuals and the families that are affected by them, and as a point of fact there is a public health approach to all of these diseases. I talked briefly yesterday about the fact that familial hypercholesterolemia, which is a treatable condition, you can prevent premature heart disease, the medical system currently today misses about half, if not more, of these cases in the general population because high cholesterol level is so rampant that this entity is missed altogether.

But most of heart disease is due to this. This is a quote from Bob Hegele a few years ago, and for those of you who don't know this gentleman, this is Jim Fixx, and the other person is well known. But Bob said in 1992 that some vegetarians with acceptable cholesterol levels suffer myocardial infarction in their 30s. Other individuals seem to live forever despite personal stress, smoking, obesity, and poor adherence to a Heart Association-approved diet. Really what we're talking about here is a complex puzzle of gene/environment interaction.

By last count, there are probably about 270 risk factors, non-genetic risk factors for heart disease, and each one of these interacts with each one of the genes in our system, and it's very difficult right now to find a pathway for use of that information for the prevention of the 95 percent of heart disease in the general population which is not thought of as genetic.

So occasionally we see articles like this. People are working hard on the prediction of MI using polymorphisms and candidate genes, and this is one example of many that we are seeing, and this happens to be a case/control study in Japan where people looked at a large number of cases of myocardial infarction and looked at 71 candidate genes with 112 polymorphisms in these genes. To cut a long story short, they found a few associations with small odds ratios, and there was an accompanying editorial that said, "Findings should be used to initiate further research, and recommendations for primary prevention cannot be based on these findings."

This is the state of affairs we find ourselves in right now in the use of genetic information in the prevention or management of most human diseases.

So let's come back to public health. Why do we need public health in the first place? What is public health? There has been over the years several pronouncements by the Institute of Medicine. In 1988 they had a meeting that led to a pronouncement called "The Future of Public Health," and then last year they revisited the future of the public's health in another report. The 1988 report did not mention genetics. The one from last year did mention genetics.

But briefly, public health is what we do as a society to assure the conditions for population health. So in public health we focus on three things. We focus on the population, the community as outpatient, not the one-on-one interaction with a patient in a clinic but the community or the population as our unit. We focus on prevention, and we would like for it to be primary prevention; i.e., the prevention of the disease before it happens. If primary prevention is not possible, then we move on to secondary prevention, like early detection, and then tertiary prevention.

Public health is very much science based. The tools of science and public health are

complementary to those in the biomedical sciences, but nevertheless they are tools, and I'll mention some of them later on.

Now, in terms of public health functions, they were laid down in the 1988 report. The three major functions are assessment, policy development, and assurance. These are very important functions, and there are lots of misconceptions about the role of public health when it comes to genetics, because people sometimes think of it as mandated population screening programs or delivery of genetic services, but these two functions are only a small fraction of what public health can do for the delivery of health care and prevention. I'll mention examples of those.

But it's the report from last year that really set a different stage for us as we talk about the role of public health. It talks about the public health system, and it really talks about the partners that work together to assure the conditions for population health. Typically, we tend to think about public health as the government public health infrastructure, which is on the left-hand side. That's us, the federal government and the state and local public health. But those units alone cannot assure the conditions for population health. The public health system as defined by the IOM is all of the partners coming together, including academic, the health care delivery system, employers and businesses, the media and communities.

As a matter of fact, when you think about this, this committee is an example of the public health system in action because you represent the various stakeholders and the groups coming together hopefully to make some policy recommendations and pronouncements that assures genetic information can be used for population health, to improve the health of the public in general. Toby Citrin mentioned yesterday this report, which is another IOM report that was published a couple of years ago about the training of the public health professionals. When you think about it, and the IOM made this estimate, there are probably about half a million professionals in the U.S. that are considered in one way or another as public health professionals.

Public health professionals are those that have a population focus in mind. In other words, they are not engaged in the delivery of health care one-on-one with patients and families but community-based activities. Many of them are actually not trained in public health, but nevertheless they are public health professionals. As Toby Citrin mentioned yesterday, the IOM made a pronouncement a couple of years ago that the public health professionals of the 21st Century will have to deal with critical areas in training, including genomics, and you can read some of the other important areas as well.

Now, what can public health do for genomics as the gene sequence and the gene discovery gets out from the bench to the bedside? What public health brings to the table is an approach to translate all of this new science into activities that improve everybody's health. The way I've captured it here is that I think about public health as contributing in three major areas, or three boxes if you will, three major gaps we're trying to fill.

The first one is probably the most important one at this point, figuring out what does it mean to have genetic variation. What's the role in genomic information in population health? I mean, we have 30,000 to 35,000 genes and thousands of variants, and many, many proteins and protein variants that are going to be discovered, and we are just skimming the surface right now of what that information means to the burden of disease and disability in different community and how this genetic variation interacts with the environment, and the environment, if I didn't say it so far, has been the major point of intervention for public health so far in our quest. So that's an important role to consider, and that's a population research agenda.

The second role is to figure out really, truly, the value added of genetic information in both treatment but primarily in prevention, because right now we have a one-size-fits-all public health approach to the major common chronic diseases that involves behavior modification, diet, exercise,

smoking cessation, et cetera, and we have to figure out scientifically, based on the best available science, why should we change that approach in favor of a personalized prevention medicine approach.

Then the issue of implementation is really crucial because you can discover all the genes and figure out what they do, you can figure out that they are good to be used in a genetic test, but the implementation can be messy in a health care system that's really not prepared for genetics. I'd like to cite to you what Claude Lenfant said last year. Claude Lenfant was the outgoing director of NHLBI and had a nice piece called "Lost in Translation." That's not the title of the movie, by the way. He basically was citing a number of areas in the heart, lung and blood area where basic research has not been translated into practice, and he used one of many examples, the issue of aspirin, that less than a third of patients that need aspirin for the prevention of coronary artery disease are actually using aspirin. At the end of the article he had this rather cynical remark, saying "Let's be realistic. If we didn't do it with aspirin, how can we expect to do it with DNA?"

Now, I don't prescribe necessarily to a pessimistic view of the world but more of an optimist in this department.

Now, CDC and many partners have begun thinking about these issues and drawing a roadmap. It's a bit tortuous right now, having landmarks as we move forward from one box to another, a population health research box, building an evidence base for prevention, and then moving genomics into practice. These things are not necessarily sequential because many genes are on different parts of this continuum. Certainly for rare genetic diseases and newborn screening, we are already in practice. But for many of the common chronic diseases, we are somewhere at the beginning of this map, where genes are coming out of the test tube, if you will, and going down the translation highway.

Since I don't have that much time, I just want to give you a brief overview of the kinds of initiatives that CDC and others are developing, and then we can have some more discussion. In the department of genomics and population health research arena, we have three major initiatives going on: the Human Genome Epidemiology Network, the NHANES projects, and genomics and acute public health investigations. The Human Genome Epidemiology Network is an international collaboration that has been sort of watching over the science of gene discovery and gene disease associations. We have many collaborators from around the world that use epidemiology as the basic science of public health, and those people are engaged in methods development, training, and knowledge base development, and we're also working with NIH and others on the pooling and synthesis of the many cohort studies that are going on around the world. You'll probably hear a bit more from Francis Collins about the U.S.-proposed study later on.

As of May 1st, we've had a number of products that are online. We don't have time to go through this, but a knowledge base and a searchable database is what I would like to show you here briefly, and you can all go online and figure it out. This is sort of a running database that changes from week to week that you can search by either gene -- we use the HuGE nomenclature; disease -- we use ICD codes; or interacting factors, like smoking and drugs, et cetera, that summarizes the status of the epidemiologic knowledge on gene/disease association, gene/environment interaction, gene/gene interaction. I am told that this is a good adjunct for many researchers who are trying to figure out how to get genes out of the test tube into population-based work.

To summarize, this is sort of the literature over the last three years. This has been going up, obviously. I mean, every day there are more papers in this regard. We capture about 50 to 100 articles every week. These are your top 10 genes: ApoE, ACE, MTHFR, and HLA. We don't have time to go through them, but these are the most epidemiologically studied genes in the literature.

The second initiative is the NHANES DNA bank. This is very important because we don't know the prevalence of the major variants of public health significance in the U.S. or around the

world. NHANES is a national survey that CDC does on a regular basis. In the NHANES III cycle back from 1988, DNA was immortalized in about 3,000 nationally representative samples from the U.S., and we're currently, in collaboration with NCI, looking at the prevalence of the top 57 genes of public health significance. I'll leave it at that for now.

The initiative we started last year is figuring out how human genetics and genetic variation can explain outbreak investigations. We're currently in the midst of evaluating which outbreak investigations, which is the bread and butter of many public health activities, both in environmental health and infectious disease outbreaks, figuring out why some people get sick but not others when exposed to the same virus, bacteria, or environmental agent.

The second area along this continuum of building the evidence base, we have two major initiatives, the genetic testing evaluation and family history. For those of you who have been around from SACGT, you may recognize this wheel. SACGT recommended that genetic tests needed to be evaluated along the continuum from analytic validity to the ethical, legal and social implications. For the last three years, we have been engaged in fleshing this out a little bit more through the collaboration with the Foundation for Blood Research using five genetic tests as examples.

We have essentially developed a methodology for how you can begin to evaluate genetic tests as they move from research to practice. At the end of this year we're going to have a methodology meeting where we compare this methodology with other methodologic technology assessments that exist, both in this country and around the world, hopefully coming up with a consensus way of evaluating genetic tests. We're using this information in this next initiative, which will be a collaborative initiative both within the government and with the private sector. We call it EGAPP, or Evaluation of Genomic Applications in Practice and Prevention.

I don't have too much time to go through the specifics, but we are going to be experimenting with a non-federal multidisciplinary independent working group that will begin to, using the tools of methodology assessment, evaluate genetic tests, with a priority for the ones that will be used for prevention and population health, evaluate them one at a time using a stakeholder group for input, and then commissioning reviews through evidence-based centers of the kinds that AHRQ supports, and then coming up with summary statements and recommendations.

The good thing about this project is that it not only involves pronouncements, but there will be some funding for pilot data collection projects to fill some of the gaps that the group will identify. I'll be happy to talk more about this.

Family history is a big one because, as we talked yesterday, it's sort of the initial genomic test, if you will, that we all have, and we don't necessarily have to have a lab test for it, and we know that family history is underutilized in preventive medicine, and it is a risk factor for most common chronic diseases of public health significance. It's frequent. If you look at the major five or six common chronic diseases, half the population has at least a first-degree relative with either cancer, heart disease, or diabetes. It is a risk factor for almost all these diseases. Depending on the number of relatives and the age of onset, those relative risks change. But it's the most consistent risk factor for all common chronic diseases, and yet very few people actually need a genetic work-up as a result of family history.

So the initiative that was started two years ago is now fully under way. We are using the simple classification scheme that Maren Scheuner, Dr. Scheuner from Cedar-Sinai at the time, and now at UCLA, proposed a few years ago to classify people into a qualitative risk classification scheme for any disease depending on their family history: an average risk, a moderately increased risk, and a high risk. We are currently developing a family health tool for five or six common chronic diseases, three cancers, heart disease, diabetes, and using a complex algorithm to classify people into these three groups. We will be conducting a controlled clinical trial to evaluate the clinical utility of this tool in

order to change people's behavior.

The good thing about this tool is it actually bridges the gap between genetics and public health, because in public health we live in this average scheme. We treat everyone in the population as an average person for any given disease, and we give everyone the same recommendation for disease prevention. Geneticists, on the other hand, are always looking for people and families with single-gene disorders, but these are only a few in the general population. Most of us, if not all of us, are not average. We fall in this moderate risk group for most diseases.

Today, we don't know the genetic basis for the moderate risk group. Ten years from now there may be enough genetic discoveries to find out that there could be a genomic profile test that will dissect this moderate group. But the good thing about family history is that it's more than genetics because it involves shared behavior, shared culture, shared diet, and a family-centered prevention approach.

Finally, since I'm running out of time, I just wanted to mention three initiatives in building the capacity and practice, building a public health capacity, developing approaches for population-based monitoring and outcomes research, and I'll mention all the efforts that CDC is doing in ensuring the lab quality of genetic testing and practice, the CLIA efforts that you all know about, et cetera.

But in terms of building the public health capacity, we have three activities, briefly. Back in 2001, as NCHPEG was developing its genomic competencies for health care professionals, CDC and many partners developed genomic competencies for the public health workforce, and there is quite a bit of overlap between the two. You've heard from Toby Citrin yesterday about the development of Centers for Genomics in Public Health. There were three that were funded over the last three years, and hopefully there will be many more, both in schools of public health and medicine across the country.

Last year, CDC began actually funding state chronic disease capacity grants to supplement what HRSA and others are doing on the maternal and child health side with respect to genetics and public health.

Last but not least in terms of outcomes and monitoring, you'll hear more about the direct-to-consumer campaign. But last year, as you know, Medical Genetics had this campaign in two test cities, Atlanta and Denver, and the public health response to this was to do a survey or a series of surveys with health departments in the two exposed cities to the campaign, Atlanta and Denver. We had two control cities, Raleigh and Seattle, in which we had surveys of women that were targeted by the campaign, about 400 in each city, and the health care providers, about 250 in each city. I don't have time to present the results of this since I am running out of time, but here they are, and we can discuss them later on.

So in closing, I'd like to kind of reiterate this long and winding road beyond the bench to the bedside concept. Really, as we all engage in what I call activities in improving the public's health, we have to realize that what we do on the population level really influences to a major extent what is done at the bedside level, and population-level information on either the epidemiology of genes or the evidence base for why we should use a genetic test and how we actually use it in practice and ensure the quality of delivery of the services is really impacting in a major way on the practice of medicine.

So I'd like to close here, and if you have any comments, I'll be glad to take them.

Thanks.

DR. McCABE: Thank you very much, Muin.

Any questions or comments for Dr. Khoury?

(No response.)

DR. McCABE: I'd seen a published report from the Myriad experience, or maybe it

was just some preliminary data that suggested that there had not been much increase. While people were aware of the ads, it had not really changed practices. From looking at your data, it looks like it did change practice. Is that the case, Muin?

DR. KHOURY: Well, I mean, we have some limitations from these data because we don't have actual utilization rates. We have what physicians told us in terms of the interest and their own practices, and it does look like there was a bit of an increase. We are also working with Myriad to analyze their own utilization data for the country and related to the denominators, which is the whole U.S. Census. So I don't have the final word on this, but in the next few months we should be able to actually map it out.

DR. McCABE: Did they continue their ad campaign?

DR. KHOURY: Not to my knowledge. They're pondering whether or not to go national right now.

DR. McCABE: Other questions or comments for Dr. Khoury?

Yes, Agnes.

MS. MASNY: In your systematic review of genetic tests that you showed the wheel, and I don't know if that's in your new book, your report, if you would think that any of the materials from there could be utilized as guidelines for us in the work that we'll be doing to try to both categorize and give guidance for the coverage and reimbursement?

DR. KHOURY: Absolutely. I mean, any of the stuff I mentioned this morning, which was a high-level discussion, there is plenty of material and back-up. So you guys tell me what you need and I'll be happy to give it to you.

DR. McCABE: Thank you very much, Muin, for that very interesting presentation.

It leads nicely to the next issue on our agenda. In March, we decided to prepare a summary of our systematic prioritization process in lieu of a vision report. Though the charter describes a vision and role for this committee, we thought that a summary of our process and the issue briefs themselves would be of use to the Secretary.

I'd like to again thank Emily Winn-Deen for her leadership throughout the prioritization process and her assistance in developing the draft vision report, and I'll now turn to Emily to facilitate the discussion of the report.

DR. WINN-DEEN: Thanks, Ed.

We have to be clear in acknowledging who really did the work on this report, which was the staff for this committee, specifically Fay Shamanski led that effort, and I want to acknowledge her efforts and the fact that she was a very apt ghost writer for the committee.

What I'd like to do this morning is to just sort of go through what I believe is pretty much just a recap of our discussion at the March meeting, make sure that everybody has had a chance to look at this summary, agrees with it. If we have any discussion items, we should put them on the table now because I know that one of our goals is to put this whole set of issue briefs, as well as the summary, up as a public document and one of our work products.

So I guess the first thing I'd like to ask if just if everybody has had a chance to read it and agrees that the written summary is a correct representation of our thought process and our end conclusions from the discussion. I know I had a chance to do a little bit of proofreading, but I don't know if there are any other comments that people would like to make at this time, so I want to do that first.

Hunt?

DR. WILLARD: A point I've made before, as well. I think it's important, especially for written documents that come from this committee, that even though our name is the Advisory Committee for Genetics, Health and Society, that we get the word "genomics" in there as well, at least

periodically in executive summary sections, because there will be some of the audience who will think that what we're saying is relevant to genetics, i.e. the last 20 years, but not necessarily genomics in the next 20 years. I know what we all mean, but I think we should be careful in choosing language that conveys that to our audiences.

DR. WINN-DEEN: So would you like to see everywhere that it says "genetics" changed to "genetics and genomics"?

DR. WILLARD: I wouldn't do it everywhere because that will get tedious to the extreme. But I think even in that very first sentence, for example, it would broaden the sense of that sentence and not hurt it a bit to say "advances in genetics and genomics promise to improve human health," et cetera. Just picking a few spots throughout, especially in summary sections, I think could have effect and be more inclusive.

DR. McCABE: We can even change it in the title to genetics and genomics so it's right up front, "Toward a Vision of the Integration of Genetics and Genomics in Health and Society," if that's acceptable to the committee.

DR. FELIX-AARON: I have a question.

DR. WINN-DEEN: Yes.

DR. FELIX-AARON: Why would we keep both? Why not just genomics? What do we lose by dropping genetics?

DR. WINN-DEEN: I'll give you my answer to the difference between genetics and genomics.

DR. FELIX-AARON: Sure.

DR. WINN-DEEN: Genetics looks just at germline DNA, whereas genomics can also encompass expression analysis, and they're quite different things. So one is what's the basic program, and the second is how is that program expressed at different points in different disease states.

DR. FELIX-AARON: Right. But for the purposes of this group -- I know those are the technical differences, but for the purposes of this group and the work that you do, I mean, I'm asking the question to the group, what would we lose by instead of having genetics/genomics, focusing on just genomics?

DR. McCABE: Yes, I also think that the practice, the clinical practice of this discipline is genetics, as opposed to genomics, which I think of more in the analytical side. But the medical practice is the practice of genetics. So taking it a little bit further, I think it's a subtle difference, but I think that if we want to have credibility within the genetics/genomics communities, we need to try and use the two terms. If we went just with genomics, I think it would be leaving behind the medical practice and some of the issues about germline inheritance.

DR. WINN-DEEN: Okay. So then I think the next thing that we wanted to do was to go through whether the report actually accurately represented our whole process for going through a large set of possible topics and the voting process and triaging and prioritization process that we went through with the main committee, with the ex officios, and in the course of our discussion at the March meeting. So I just want to ask if there's any discussion or if anyone feels that we've failed to capture that in an accurate representation.

(No response.)

DR. WINN-DEEN: Wow. I'm really excited, because after we had all that discussion on whereases yesterday, I was afraid that this might take a really long time.

So I'll take the silence as everybody's ascension that this is an accurate representation of the process and the conclusions of that process.

I guess the third thing I wanted to do was to, if we feel that the overall summary

report, "Toward a Vision of the Integration of Genetics and Genomics in Health and Society," is a good representation of our thought process, I just want to give people a chance if there's any comments on any of the issue briefs, which I think we had a few comments on at the last meeting, and I think most of those comments have been incorporated. But if anyone has any further thoughts or corrections, comments, whatever, on any of the issue briefs, I'd like to open those up as well for any recommendations.

Ed?

DR. McCABE: Before moving on to that, we were just having a little bit of a sidebar here, and one of the things, because of this discussion about genetics versus genomics, that we might try and do is a fairly brief glossary of some of the key terms that we could work on, if that's acceptable to everyone.

DR. WINN-DEEN: Yes. Clearly, I think that would be a useful -- I don't know if it needs to be in the body of the Towards the Vision statement, but just as part of the appendix would be a glossary of terms.

DR. McCABE: Right, and it's not going to be an extensive 30-page glossary, but picking up the key terms like we just discussed I think would be important for people to understand what the real issues are here.

DR. WINN-DEEN: Yes. I actually think that there's a lot of those things that the previous committee already worked on, good definitions, and so we can just pull those together.

DR. McCABE: Yes, that's where I was really thinking we had those glossaries probably largely in hand and can extract from them.

DR. WINN-DEEN: Any dissenting thoughts on that?

(No response.)

DR. WINN-DEEN: Okay. So let's ask staff if they would add that to the appendix as well.

Can we turn to the issue briefs?

DR. FEETHAM: Not to do revisionist history, but in the bullet on the first page of the summary, my recall from the discussions on the third bullet that we talked about, again to be consistent with the name of this committee, it's the ethical/legal/health, which to me is broader than medical. Also there was, as I recall, part of the discussion on the large population studies was the economic. I just bring that up because I think that's what I recall from our discussions, which is broader than what that third bullet is.

DR. WINN-DEEN: Right. So economic, you're talking about sort of who would fund the large population studies?

DR. FEETHAM: Well, I thought that was part of the implications. It's effect on health, but also there's the cost benefit, which was part of the discussion as I recall it, and that was my thinking on it.

DR. McCABE: Just so we have it for the record, investigations of the ethical, legal, health, economic and social implications, is that the way it would read? Or maybe we could put health first, Sarah is suggesting, which I think is a good idea.

DR. WINN-DEEN: Thank you. It's good to have a few comments. I'm underwhelmed by the response right here.

I think that these issue briefs are also quite important for us to have officially blessed as part of this committee, because they are going to in some ways frame the issues, but also direct the way that we think about some of the ones that we have put on a prioritization path. So I'd like to take comments on that.

Deb, if you have some?

DR. LEONARD: In just looking at the bullet points, also "enhancement of oversight of genetic technologies and services." This makes it sound as if there's a need for enhancement, as opposed to assessing the oversight. I mean, it's put in almost a negative light, and we did hear from CLIA and FDA and CAP. There is a lot of oversight of genetic testing services now. So could we change enhancement to assessing the oversight? Because that's really the process that we did. We looked at what was being done and not necessarily enhancing it but evaluating.

DR. WINN-DEEN: So would you say assessment of the need for enhancement, or should we just say assessment?

DR. LEONARD: I think what we did was to assess the oversight. It's just that enhancement makes it sound like there's a deficiency in the oversight of genetic tests, which I don't think currently exists for most areas.

DR. TURNER: Except that isn't this where we got into the testing that you can order online by sending in mouth swabs, and we had that discussion about all the different ways that maybe we wouldn't bless as official testing but that the community at large sees as an opportunity for testing?

DR. LEONARD: But that's the final bullet, assessing the pros and cons of direct-to-consumer marketing of genetic tests. I think that's been separated out as a separate bullet. It may be a fine point, but just looking at how all these others are stated, this is stated in such a way that it sounds as if there needs to be more oversight, rather than we are assessing whether the current oversight is adequate or not.

DR. WINN-DEEN: Right. I think that's fine.

Fay, can you make that change? Okay.

Going once, going twice -- we've got another red light on over there.

DR. SEMERJIAN: I have not been involved in these discussions. This is my first time here. I noticed that in Dr. Murray's presentation, he added an item into his presentation that was not in the printed version about the quality assurance of genetic testing, et cetera, which sort of relates to the same issue.

Perhaps the sensitivity here is with regard to the word "oversight," no? Because I think that there is room for improvement in terms of the quality of measurements, the traceability of measurements to national standards, et cetera, because I think this is a very different testing issue with regard to genetics versus the run of the mill clinical measurements where we have our cholesterol tested many, many times, whereas genetic testing perhaps will be done once in some cases, and you rely on that information for many decisions, that you need a different level of quality assurance, reliability of those measurements.

I'm not sure that we are at that point. I thought this was perhaps part of it, but I thought maybe the issue was do we really want to say oversight, or do you want to say enhancement of quality assurance measurements or quality assurance efforts or something like that? But I think there is room for improvement in that regard in terms of quality assurance.

DR. WINN-DEEN: Ed?

DR. McCABE: What if we state it as "assessment of quality assurance and oversight"? Would that be a way of getting that point in there? Because I think that was part of the discussion in point of fact.

Sarah is commenting that does this get to clinical validity, or is quality assurance more analytical validity?

DR. WINN-DEEN: Well, I think quality assurance is more getting at whether the answer you gave, given the analyte you tested, was the correct answer, which is different than whether that answer has any medical utility. I think they're two separate things, and I think the point that was just

made by NIST is that because things are potentially once in a lifetime tests where you don't have a chance through repeated testing to catch an error, do we have some higher obligation to provide QA/QC kind of mechanisms to assure that the test result is actually a correct result.

DR. McCABE: I'm just trying to figure out how to word it to assist staff. What if we don't make quality assurance, if we leave it at quality, assessment of the quality and oversight of --

DR. WINN-DEEN: I think it might be useful to actually look at the issue brief that goes with that bullet point, which is in the appendix "Oversight of Genetic Technologies Issue Brief." I think there's a lot of discussion about who has oversight responsibilities, what the current status is, but there's not really a separate discussion in that brief on QA and standardization of methods as much as just in the current medical system what groups are responsible for trying to provide the oversight that would be relevant for genetic tests.

Cindy?

MS. BERRY: What if we just said assessment of oversight, blah blah blah, and refinement where appropriate, so we aren't really making a judgment about whether we're definitely going to improve something or that there's something in need of improvement, but we're recognizing the fact that we're always going to have to refine things given changing circumstances. I don't know if that does the trick.

DR. WINN-DEEN: I'll turn to the people who brought up the question. Ed, if you want to comment.

DR. McCABE: Well, I was just looking, and if one reads the issue brief, the issue brief pretty clearly says need for enhanced oversight of genetics tests and leads logically to that point also, issues about protecting the public and access to new and cutting-edge technologies. But I think the way it was stated, perhaps the way you originally said it, assessment of the need for enhancement of oversight, since that clearly is in the issue brief, and by stating it that way it doesn't presume a conclusion.

DR. WINN-DEEN: Okay.

Hunt, you had some comments?

DR. WILLARD: I was just going to point out that in an executive summary like this, all of the other terms are very neutral. They don't tip our hand one way or another, we just list opportunities. So in that sense, Debra's point is absolutely right on target. I don't think we have to sort of lengthen the bullet point to cover all contingencies. We're simply saying we were evaluating it and read further if you want to know what we decided. In that sense, I'd tip more towards neutral terms as much as we can.

DR. WINN-DEEN: So assessment of the need for oversight, or need for enhancement?

DR. WILLARD: I don't think we want to say assessing the need for oversight. That would suggest we actually consider the possibility no one needs to have oversight.

DR. WINN-DEEN: So assessment of the need for enhancement of oversight of genetic technologies and services?

DR. WILLARD: I'm not a big fan of the word "enhancement." I think I'm with Debra on that one.

DR. WINN-DEEN: Okay.

DR. LEONARD: Actually, if you look at the brief, while the next to the last paragraph says "While there seems to be a consensus about the need for enhanced oversight of genetic tests," the beginning of the next paragraph states the question "has a balance between protecting the public and access to new and cutting-edge technologies already been achieved? Do current regulatory

mechanisms strike an adequate balance between access, safety, competition and independence of medical practice?" So those are the questions that we're looking at. I mean, that statement at the first sentence of the previous paragraph is pretty strong, actually, and I never really caught it before this point, because that's saying that there is a need for enhanced oversight, whereas the questions that are being asked at the end are really more balanced.

So I would just be happy with changing it to "assessment of oversight of genetic technologies and services," or "assessment of quality and oversight of genetic technologies and services," because it is, as Hunt points out, more neutral and consistent with the other bullet points as something we're going to look at, without a pre-conclusion about what needs to be done.

DR. WINN-DEEN: Okay. So I'm going to ask -- oops. We've got Judy Yost.

MS. YOST: Yes, I just have a comment, and maybe it's a very subtle point, because I think we had this discussion once before about the terminology when referring to this. My concern again, and maybe it's not legitimate but I think I need to bring it forward, is that that assessment, just leaving it as is, I realize your need for neutrality in an executive summary. However, when you're saying that, it sounds like an active ongoing monitoring of what the oversight is, like you're actually doing -- you know what I mean? -- taking an active part in that oversight, and I don't think that's really what's intended as the role of this committee, frankly.

DR. WINN-DEEN: I think what we decided as a committee was that we would try and just keep this on the radar screen so that we would have periodic reports from the different bodies like CLIA about where things are.

MS. YOST: And that's fine, absolutely. But at that level.

DR. WINN-DEEN: Barbara?

MS. HARRISON: I was thinking maybe a compromise would be to say "assessing the current state of oversight" or "assessing the state" or "the status," so you imply that it's just a one-time look at the topic.

DR. McCABE: And since we were planning to monitor occasionally, as we concluded, I think assessing the status rather than current status would be the appropriate thing.

DR. WINN-DEEN: I'm going to ask Fay to read back what she thinks that bullet point says now so that we can see if we have consensus.

DR. SHAMANSKI: "Assessment of the status of quality and oversight of genetic technologies and services."

DR. WINN-DEEN: Okay. Is everybody happy with that as the bullet?

(No response.)

DR. WINN-DEEN: I'll take silence as a yes. Quick, turn off all the mikes. Joan?

DR. REEDE: In another area, looking at the issue brief on genetics education and training, and given our long conversations yesterday, it really doesn't address issues of diversity in the workforce or cultural competency, and I think those words need to be incorporated within the issue brief.

DR. SHAMANSKI: Do you mean within the issue brief or within the report itself?

DR. REEDE: I think at least within the issue brief, so that when it asks about adequacy of the genetics workforce, there's no mention of diversity there, there's no mention of cultural competency.

DR. WINN-DEEN: So, Joan, do you want to suggest a place where that might go? Should it go somewhere in the first paragraph?

DR. REEDE: It could, if you look at the next to last paragraph, it says there are questions about the adequacy of the genetics workforce, and then it speaks about specialists and

generalists, and I think there to talk about adequacy and talk about adequacy in terms of representation and diversity of groups would be a logical place to put that.

DR. WINN-DEEN: Any dissenting votes on adding those two points to the workforce issue brief?

(No response.)

DR. WINN-DEEN: I think those are really good points and they clearly were brought up as important parts of the training and education of our workforce that's going to be dealing with genetic issues in the future.

Are there other comments on issue briefs that we should take up? Fay?

DR. SHAMANSKI: I just wanted to point out that we did get some public comments on the issue briefs, and I think the committee needs to talk about whether we're going to send those out for public comments. I just want to remind you that when we wrote them, it was just to present a balanced view of the issues, not to present the committee's position on the issues but rather just to give all the background information on which to base your decisions. So the question is whether you want to send those out for public comments and whether we want to change them further, or do we want to keep them in the current state that we had determined previously?

DR. McCABE: In the table folder is the response from AHIP. So America's Health Insurance Plans basically says "We believe concerns" -- I mean, there are a number of things here, but I'll summarize. "We believe concerns about possible genetic discrimination by health insurance plans are largely unfounded." It continues on. Sarah is stating that they point out factual disagreement, and probably we should restate how we spoke of ERISA. It says, "We would also note that the discussion on page 3 of the genetic exceptionalism issue brief incorrectly states the impact of ERISA on state laws dealing with genetic privacy and genetic discrimination and health insurance and employment. ERISA does not apply to insurers or to health information privacy employment. Rather, ERISA is a federal statute that governs pension and welfare benefit plans, including health and disability, income benefit plans." So we can make that factual change.

DR. WINN-DEEN: Right. I think what we should do with the public comments -- this is my personal opinion, but we can get a committee consensus -- is to take the places where we've gotten comments like this that are related to factual things and just do a fact check, because obviously you drew your information from some source and they're drawing from some source, so let's try to fact check it and not just blanketly take the public comment as the correct information but do our due diligence on that, and then I think that would be highly appropriate to make any factual corrections.

I guess from the point of view of the issue briefs, I agree that the point of them was to try and just give a balanced view of what the issue is and what sort of the things are under discussion or that might require further discussion and to try to make sure we've captured those things as well, without drawing a committee conclusion in any way. I think these were intended to frame the issues so that we could then go through each issue and, as we did yesterday with the education, now we're going to make a specific recommendation in our resolution about what we believe should be done, and I think that's completely different than just to frame the issue brief, which is what these were intended to be.

DR. McCABE: I think clearly from discussions that have been held at every one of our meetings, "the concerns about possible genetic discrimination by health insurance plans are largely unfounded," I think this and the previous committee has disagreed with this, the public disagrees, and we in fact will try to bring the public to the next meeting to discuss that where there has been discrimination.

DR. WINN-DEEN: Well, I think it's completely legitimate to say that there is a debate on whether or not it is or isn't that certain stakeholders feel there's no need for legislation because there really haven't been any major abuses, and then there's other lines of evidence that say despite the

fact that maybe there haven't been very many highly publicized cases of abuse, there still is when you do public opinion surveys a feeling among the public that there's a fear. So how do you resolve that? That in itself is a dilemma. How do you resolve the issue of overcoming public fear when there's not actually too much documentation that that fear has a rational basis? But we still have to deal with it. It's still a barrier to the implementation of this and to the practice of medicine.

DR. WINN-DEEN: Other comments on any of the issue briefs? Hunt?

DR. WILLARD: It might be, since this is intended to become a public document, that at the beginning of the appendix where it simply says "Issue Briefs on 12 Priority Issues," that we add a brief paragraph explaining what we mean by issue briefs to essentially argue what you just said, that they're not designed to give an answer, they're designed to lay out the issues for the committee, and by extension for the public, so that people don't read these and believe that we're somehow either trying to make a recommendation or refusing to try to make a recommendation.

DR. WINN-DEEN: Right. That's something staff could do, just write a little summary paragraph with that.

DR. LEONARD: That is kind of explained at the top of the briefs, though, in that statement that's on every page. So something similar to that put at the beginning in bigger letters so you notice it more.

DR. WINN-DEEN: Yes. We just want to make sure that we capture that these are intended to frame the issues rather than reflect any statement of what this committee has arrived at as a conclusion on that particular issue.

DR. McCABE: I would point out, though, that if we finalize it here, then it does become the official view of the Secretary's Advisory Committee on Genetics.

DR. WINN-DEEN: Right, but it's a view basically that this is an issue and here are the questions that remain to be answered, rather than here is our recommendation for what to do about it, which I think are quite different things, and I don't think, for most of these issues, we're quite ready to put our stake in the ground and say this is it, we know exactly what we want to do, go for it. We're framing what we want to do for the next couple of years of this committee's life.

DR. WILLARD: It's particularly acute when you go to the vision statement issue brief. I mean, most people would go there thinking, ah, this is where I'm going to see the vision. In fact, what you get is a lot of questions that say should we have a vision, should we ask someone else to have a vision, and it actually doesn't declare the vision. So I think it's important that people know what these are, and in particular what they're not.

DR. WINN-DEEN: Other discussion from any of the ex officios?

Muin is reaching.

DR. KHOURY: I'm not sure if what I'm going to say might sound a bit too harsh. I missed the last meeting, so I'm not sure how you got to where you are right now. But I think this committee should be more bold, or bolder, and I agree with Hunt here. I was reading the document here, and it meanders along. I think you need to be bold and establish a vision for how this stuff is going to happen. I mean, people know about the issues related to genetics. I was hoping my comments this morning might elicit some reaction, but I guess we all ate too much protein last night.

So just to encourage you to be bold. I think the country needs help in using genetics to improve health and help society. If this is the place to come, just be bold.

DR. McCABE: I would just point out that, as Hunt stated, these really are briefs that we then prioritized in terms of how we would move forward. So they were not the answer; they were the beginning. I would argue that it's not that we don't have a vision, that we voted not to have a vision, but we voted that perhaps that's not where we should spend our time in crafting a vision, but rather to get

down to some specific approaches.

DR. WINN-DEEN: My recollection on the ranking of these various things was -- since I put the vision statement on the table, I sort of paid attention to how it fell out in the ranking, and it didn't get ranked very highly by most of the people who voted on the issue. I think people were more concerned with us spending our time on the actual issues than spending however many hours writing the vision statement, which we could have potentially gone to the "Dilbert" vision statement builder website and picked out the right keywords and had one created for us.

So I think our vision is going to come from the issues that we've chosen to prioritize and trying to take specific action on those issues. So being bold in a specific way rather than in a more general way.

DR. LEONARD: Is "vision" the right word? I just wonder if "vision" -- I mean, maybe this is more our roadmap rather than our vision, because we're going to be creating the vision of what we want to do and actions to take and things as we work through these different issues. This is not really -- you're right, one of the issues was to create a vision statement. What we ended up doing was creating a roadmap.

DR. WINN-DEEN: Yes. So I would be okay with a shift in the summary brief to roadmap instead of vision.

Hunt, let's hear from you.

DR. WILLARD: Two points. One is the unfortunate confluence of terms. So we end up with an issue brief on a vision statement, conclude that we aren't going to spend our time on that, and then write a document that's entitled "Towards a Vision," suggesting that despite our vote we're going ahead and doing that. So some post hoc changing of terms may be of some value.

The other specific suggestion I would have, just again to remind readers and to avoid potential misinterpretation of what these issue briefs are, is perhaps put as the last entry for each of the issue briefs committee outcome or something like that, that reminds the reader how this was then prioritized. This issue became one of the top priorities for 2004, or this issue was determined to be an overarching issue. Then under the vision one it could say what we just all said, that rather than having the committee deliberate on the need for a guiding vision statement, the committee instead decided to do something else. It just would clarify and allow readers six months later to actually understand what thought process we went through.

DR. WINN-DEEN: It might actually be worthwhile, Fay, even ordering the issue briefs instead of alphabetically at this point. I mean alphabetically was completely appropriate when we hadn't ranked them, but maybe at this point it would help also even just in the Table of Contents and in the order that they are presented to the reader on the website or whatever mechanism to put them in the priority order that we have established now so it's very clear to someone scanning it what the priority is. Then our minutes, as we go through our subsequent task force meetings, will have I think a real clear way of capturing, okay, we said these were the top priorities, they were presented at this meeting, the outcome was a specific recommendation. Coverage and reimbursement we obviously have decided is going to take several meetings before we're ready to put out a committee statement.

But others, we hopefully at the end of this meeting will have two committee resolutions. I think that really will also help clarify to people who are following our progress that the priority issues are being dealt with and that there are outcomes.

Hunt?

DR. WILLARD: In terms of the summary statement, there actually are five categories -- we took these 12 potential priorities and put them into five different categories. So I have two suggestions. In the summary statement, I'd actually divide them up, be a little more telegraphic and

divide them into the five sections, use bullet points, help the reader understand. That's what an executive summary is supposed to be for. Then I would agree with Emily to actually then organize the Table of Contents into those five categories. Within each one you can alphabetize. But then it's very clear where one is in terms of those five.

DR. McCABE: So just to help staff, we're on the outcomes section, and I'm looking for it, under Tab 2.

MS. CARR: I was just raising a question about the five categories. We started with four, I think, and then --

DR. WILLARD: Then we have to rewrite the executive summary because that paragraph divides into five. So you've got two issues that are the highest priority, then you have two other issues that are -- sorry, three other issues that are "undertaken" for exploration. Then we've got two others that are short-term action, two others that are monitoring, and three others that are overarching.

DR. McCABE: So rather than the way we had set up the prioritization initially, we could take that penultimate paragraph of the summary and just make it a little easier to discern what the real outcome of that discussion was.

DR. WINN-DEEN: I think just some bullet points and pulling things together so that it's all internally consistent. So the way we categorize things, they're listed that way in the Table of Contents, they're listed that way in the body of the summary, and then they're subgrouped that way in the appendix as well.

Ed?

DR. McCABE: Just to go back to the point about vision being one of our lower priorities, and yet the title of the document, what if we made it rather than "Toward a Vision," which is how we got there without being a vision statement, and I think that was the subtlety, but if we made it "Mapping the Integration of Genetics in Health and Society" or "A Roadmap for the Integration," I was thinking with the mapping being a little bit of the genomic allusion there.

DR. WINN-DEEN: So the question is can we steal Muin's CDC little road logo in helix form.

DR. KHOURY: Public domain. Go ahead.

DR. McCABE: So "Mapping the Integration of Genetics" --

DR. WINN-DEEN: And genomics.

DR. McCABE: And genomics.

DR. WINN-DEEN: I actually like the integration. I think "A Roadmap for the Integration of Genetics and Genomics in Health and Society" or something along those lines would be --

DR. McCABE: Okay. We just want to get it specifically.

DR. WINN-DEEN: Yes. I'd just like to keep the word "integration" there because I think that's really the key thing. There are a lot of activities, and they need to all be somehow coming together in confluence so that this can actually happen.

DR. SHAMANSKI: Could you just review for me what you decided on for the title?
Sorry.

DR. McCABE: I think it is "A Roadmap for the Integration of Genetics and Genomics in Health and Society." Is that correct?

DR. WINN-DEEN: That was my recommendation. Is everybody okay with that?

MR. MARGUS: You're going to call it roadmap? I mean, it's not the roadmap. It's developing a roadmap where the prioritization of the issues now have to be reviewed in order to come up with a roadmap. But it's not actually the roadmap, is it?

DR. WINN-DEEN: Comments?

MR. DANNENFELSER: I recommend going back to mapping. I think it covers that concern.

DR. LEONARD: Brad, why has a roadmap not okay? Because we've set out where we're going to go, and then we're going to go down the road to do it. So in that sense, this is our roadmap of what we're going to be working on.

MR. MARGUS: I see, a roadmap for us. I get it. I thought you meant a roadmap as in the way --

DR. LEONARD: Well, I see it as a roadmap for us, for the committee.

MR. MARGUS: Okay. Yes, I cave completely. Use it.

(Laughter.)

DR. WINN-DEEN: So do we need to say "SACGHS Roadmap"?

DR. McCABE: The subtitle is "A Report of the SACGHS," so I think that's clear.

DR. WINN-DEEN: All right. Now, don't feel compelled to fill the time just because there's time.

DR. McCABE: We have plenty of things to do today.

DR. WINN-DEEN: Yes. I want to make sure we've captured everybody's comments, but we don't have to sit here and wordsmith minutiae just to fill the time until 1 o'clock.

Deb?

DR. LEONARD: Just one quick point. "Health care" is sometimes hyphenated, sometimes split. Could that be consistent throughout?

DR. WINN-DEEN: Agnes?

MS. MASNY: Just a question. Did we finalize the question we had earlier about having this go to the public for comment?

MR. MARGUS: I'll echo what I think we already heard, and that is we spent the last year only trying to go through the issues to figure out which issues we now want to focus on. If someone wanted to make public comment, it wouldn't be about any stance to take on those issues but about whether those issues are important for us to then pursue. While I guess they could still make a comment on this not on particular issues but about whether those issues are important or not, they have had the last year to do that, and we've heard from a lot of public comment. So it doesn't appear to me that we need to do that now.

DR. McCABE: And I was going to ask that at the conclusion of this session we then make a decision -- I was going to propose that we make a decision to finalize the document with the changes recommended and that be the finalized document. It will then go public. Obviously, the public can make comment then. But it's more, I think, as Brad has suggested, it will be comment to help guide us in our future deliberations on these issues. But in terms of what this document sets forth to say, we've had it out there, we have voted upon it. People may disagree with our prioritization, but I think it would be important to move this forward to the Secretary.

DR. WINN-DEEN: Right. So I would agree with that. I think that we certainly are interested, always interested in comments from the public on any of these issues. As I mentioned before, I think the important public comments to incorporate here are the factual comments so that we're not misstating facts. But at this point, unless there's some other aspect to an issue that we haven't considered, I think that would be a legitimate thing to add to an issue brief, if there's one more question that should be on that list of questions to consider. But beyond that, I think we should try to finalize these today and then seek the public comment as we get to each issue and really want to delve into it and get all the public input on that issue prior to making a specific recommendation by the committee.

Ed?

DR. McCABE: Part of my concern is if we consider the calendar, it's important to move this forward with some dispatch.

MR. DANNENFELSER: At the risk of wordsmithing, just a small point on page 5 of the roadmap. The top paragraph is a reference that says "CLIA-certified laboratories." I think that's the first reference to CLIA that I think we could use the full name there.

DR. WINN-DEEN: Right. I think part of the appendix of definitions might be also just a listing of all the key acronyms and what they actually mean.

DR. McCABE: That could now make it 30 pages long, but we'll do our best.

Sarah also had a suggestion in the spirit of truth in advertising from the title, I guess it would be. So using the title that we had decided upon, but then "The Study Priorities of the Secretary" rather than "A Report of The Study Priorities of the Secretary's Advisory Committee on Genetics in Health and Society." So it makes it a little clearer in the subtitle that there was a priority-setting process. Is that okay with everyone?

DR. WINN-DEEN: I'm going to try it one more time. Can we get -- Alan?

DR. GUTTMACHER: Again, at the risk of wordsmithing, I think it might be an important concept. I've got a question on page 1 of the executive summary, the second bullet. "Public discussion of the nature of genetic information (conceptualized in the term genetic exceptionalism)." Is that to suggest that genetic information is equivalent to genetic exceptionalism? What is actually conceptualized? I find that unclear and/or misleading.

DR. SHAMANSKI: It was the nature that we're referring to.

DR. GUTTMACHER: The nature of genetic information is --

DR. SHAMANSKI: Is it unique in some ways from other types of information?

DR. GUTTMACHER: Then I think we should be clear, if that's what we mean.

"Public discussion of whether or not genetic information is unique" or something like that, because there's certainly much to be discussed about the nature of genetic information beyond the question of its uniqueness. If I'm the only one disturbed by this, then just leave me disturbed.

(Laughter.)

DR. SHAMANSKI: That's fine.

DR. WINN-DEEN: Should we just add the word "unique" in front of "nature"?

DR. McCABE: Why don't we state it "Public discussion of whether genetic information is unique medical information"? That's the real nature of this discussion, as I recall it.

DR. WILLARD: Say "unique personal information." It's not limited simply to medical information.

DR. McCABE: "Unique personal information."

MS. CARR: Also, I think the only reason we were -- the genetic exceptionalism term is not exactly -- I mean, it's sort of esoteric. I think it's what the community knows and uses. So we were trying to not use that term --

DR. GUTTMACHER: Oh, sure. Using both terms is fine, but I just wanted it clear.

Thanks.

DR. McCABE: So the way we have it now is "Public discussion of whether genetic information is unique personal information"?

DR. WINN-DEEN: Deb?

DR. LEONARD: Can we leave "medical" in there? Because it's the medical treatment of the genetic information. I mean it's personal information as well, but can we say "medical and personal information"? Because it's the issues surrounding how you treat that medically and whether it goes in the medical record and things like that.

DR. GUTTMACHER: Isn't it the question of whether genetic information is different from other medical information?

DR. SHAMANSKI: Well, it goes beyond that, because our decision in the end was to look at genetic exceptionalism about each issue as we go. So in terms of education, it's whether we're going to treat the education in genetics differently than we're going to treat others. So the idea of genetic exceptionalism goes I think maybe beyond the information, so maybe we need to work with that wording a bit more.

DR. McCABE: Why don't we include that in the prose as an overarching, that genetic exceptionalism is an overarching --

DR. SHAMANSKI: It is in the list.

DR. McCABE: Okay.

DR. WINN-DEEN: Going once, going twice --

DR. FEETHAM: I appreciate your discussion on the medical. But again, I go along with it's the individual's information. Medical to me is more narrow. It's really health information. It's moving towards electronic health records. I mean, I just think it's conceptually much broader than medical.

DR. WINN-DEEN: So if we said "personal health information," would that capture it?

I think what I'd like to do, unless Ed has other comments --

DR. McCABE: Well, Sarah points out that we need to be accurate also, and genetic information is in fact unique personal information. We are unique genetically.

DR. WINN-DEEN: Personal and health?

DR. LEONARD: Could we say "Public discussion of whether genetic information is different from other personal health information"? Because it's really the difference between. Is there a difference, or should it be treated just like all other personal health information?

DR. McCABE: Any objections to that?

(No response.)

DR. WINN-DEEN: All right. So at this point, I think what I'd like to do is ask staff to make the changes that we've requested, to perhaps send them out to the task force or to Ed and myself.

DR. McCABE: I would suggest a fairly small group to work on this. So why not you and me and Emily, if that's acceptable to the committee, that we will then finalize this document and send it on with a cover letter explaining its nature to the Secretary.

DR. WINN-DEEN: Do we need to get the Secretary's approval before we would post these, or what would we do in terms of public access?

MS. CARR: Well, the process will be that we will send it forward to the Secretary, and once it's received, we can post it on our website. We want to make sure the Secretary has it before we make it public as a final document.

DR. WINN-DEEN: That's probably a good order.

DR. McCABE: Just to point out, that's a term of art, "received by the Secretary." It's not like when we receive a letter. It's when it's been formally received. So it may take a process of weeks to be received by the Secretary, just to make it clear. But that's why I would like to move forward, because again looking at the calendar, I think it would be good to get it to the Secretary as soon as possible.

DR. WINN-DEEN: Ed, do you want to go ahead and take our break now, or do you want to go ahead and start the next discussion area?

DR. McCABE: Do I have a sense of the committee? Is there anyone that disagrees

with this process, then, as outlined to the committee?

(No response.)

DR. McCABE: If I do not hear any disagreement, then we will move forward with that process, make the changes with some dispatch, and send the report on to the Secretary.

Thank you very much. This has been a huge amount of work but I think a very important priority-setting exercise that we went through, and I think it's a good summary of the thinking of this committee and will be useful in a lot of different venues.

Actually, we had the break scheduled -- I think that's a good idea. Why don't we take a break now, Emily, and then we'll have the public comment after the break.

Debra?

DR. LEONARD: Can I ask a question? The education resolution, was that revised and will we see that this afternoon during the working session at lunch? Just for information.

DR. McCABE: Why don't we take the break now? Yes, I think we will have time, but let's keep this to a 15-minute break.

MS. CARR: Yes, it's been revised, and I think the plan was to bring it back to the committee to show the revisions at some point during the three-hour open session, I think, and then there may also be some additional discussion of the coverage and reimbursement report. Is that correct?

DR. McCABE: Yes.

Let's take a 15-minute break, and we will resume at about 10:50.

(Recess.)

DR. McCABE: I have four speakers for public comment. We set aside time each day for this. It's very important to the committee, as has been expressed this morning, and we welcome and appreciate the views that are expressed in this public comment period.

I would ask all of the commentators to hold your comments to five minutes. If you can do it in less, it leaves more time for questioning.

The four I have in order, so you'll be aware of your order, are Joe McInerney from NCHPEG; Michael Murphy, president and CEO of Gentriss Corporation; Kelly Ormond from the National Society of Genetic Counselors; and Gail Javitt, policy analyst, Genetics and Public Policy Center.

If there is anyone else who wishes to make public comment that is not on that list, please sign up at the desk outside.

We'll start off with Joe McInerney from NCHPEG.

MR. McINERNEY: Thank you very much. I'll be brief.

DR. McCABE: Joe, why don't you come up -- why don't each of the commentators come up to the table and take one of these mikes at the table, please.

MR. McINERNEY: Thank you very much. I'll be brief. I was listening to the discussion about genetics and genomics, and I certainly don't want to reopen that whole issue for the committee, but I think rather than simply relegating that distinction to the glossary, I would urge you to address that right up front in your document. I know from experience in working with a broad range of health professionals in the last few years that there's a great deal of confusion about what genetics is and what genomics is, and I think this committee should clarify that for people who are coming to this document without any background in genetics.

One of my concerns is that there is an assumption that genomics is somehow going to obviate genetics, or that from this point forward it's going to be only genomics and what we all know as sort of classical or traditional genetics, the study of inherited biological variation and its clinical application in terms of medical genetics, will somehow be left aside. So I would just like to make certain

that that doesn't happen, that that perception or conception does not come through in this document and that the committee takes some time to define the terms up front in the context of the work that's going to follow.

Thanks.

DR. McCABE: Thank you, Joe. Why don't you stay there a minute, Joe. I would agree with you that genetics is the study of inherited traits, that genomics is the study of genomes, and I think that's how we intend to use the terms and will make that clearer in the document.

Okay. Any other questions or comments for Joe?

(No response.)

DR. McCABE: Thank you.

Next we have Michael Murphy, president and CEO of Gentris Corporation.

MR. MURPHY: Thank you. As a means of self-discipline to make sure I cover everything I want to in my five minutes, I'm going to read a pre-written statement.

Good morning. My name is Michael Murphy. I'm president and CEO of Gentris Corporation. I'm also serving on the Pharmacogenomics Advisory Group for the American Association for Clinical Chemistry. This is a group that we've put together from our government liaison to advise FDA and CMS.

Gentris is a clinical pharmacogenomics company. We perform testing for pharmaceutical companies during drug development, and we're also commercializing in vitro diagnostic products for physician-referred testing. It's a pleasure and honor to speak before the committee during the public comments session. I'm speaking in favor of the resolution direct-to-consumer marketing of genetic tests.

Pharmacogenomics is the study of an individual's genetic traits and the relationship it has to variable drug response. The field has made tremendous progress in the last 20 years. Specifically, we've been able to identify the dozen or so genes responsible for drug metabolism and clearance in humans. In addition, we understand the liver enzymes encoded by these genes are involved in the biotransformation of more than 80 percent of all commonly prescribed drugs. We also know that in general, 5 to 7 percent of all patients are so-called poor metabolizers. These patients are at risk for serious adverse drug reactions because they tend to accumulate drugs to toxic levels in their bloodstream.

Adverse drug reactions are now the fourth leading cause of death in the United States, with more than 100,000 lives lost each year. Most experts appreciate that many of the deaths might be prevented once pharmacogenomic testing is utilized prior to drug treatment.

We and others have developed clinical pharmacogenomic tests which can be used prospectively before drug treatment. In fact, most of our pharmaceutical sponsors do just that during clinical trials to make sure they develop safer and more effective drugs. Now we have developed clinical diagnostic products so we can do the same thing for all patients, not just those in clinical trials.

It's clear we're on the verge of early adoption of clinical pharmacogenomic testing into medical practice and health care. Direct-to-consumer marketing of genetic tests has the potential to slow or harm much of the progress we've made in clinical pharmacogenomics. So why shouldn't we let patients have access to these important tests directly, and why shouldn't we market to them?

Pharmacogenomic tests require a translation from a person's genotype to what is called the predicted phenotype. That is, we have to tell the physician if the patient fits the profile for one of four possible metabolizer types, including poor, intermediate, extensive, or ultra-rapid. Even if the laboratories supply this information to patients, there's still a need for a learned intermediary to help in further translating metabolism type to a drug-prescribing recommendation. It's critical to have a medical professional, such as a physician's assistant, nurse, or a doctor, use this information to guide drug

treatment. It is conceivable that patients could use this genetic test result to change their own drug treatment regimen simply based on something they read on a website from groups that market direct to patients. Obviously, this has the potential to harm instead of help.

It's understandable that patients will seek genetic information that might be used for drug treatment. For example, in the May issue of Reader's Digest, a widely read lay publication, they featured an article entitled "Genetic Breakthroughs: Making Medicine Safe." In 2002, the FDA approved Stritera or atamoxitene for attention deficit hyperactivity disorder, ADHD, in children. The FDA took the unprecedented decision to label this drug with warnings about increased adverse events in poor metabolizers of a gene called 2D6. They also included in the label a statement "testing is available," recommending that physicians consider genotyping. So it's not hard to imagine that parents might seek out 2D6 testing for their child about to start this drug treatment.

As most of you know, some companies are marketing to consumers claiming to help patients by testing for genes related to nutrition, so-called nutrigenomics, and even genes related to lifestyle. Most of these tests have not been validated or substantiated in peer reviewed literature or case-control clinical trials. When clinical pharmacogenomic tests like the drug metabolism test we've described are packaged with these unsubstantiated tests, there is the danger that clinical utility might be overlooked or their credibility diminished in the eyes of medical practitioners.

Just recently, on May 11, 2004, the Wall Street Journal ran a short article in the personal health section about several of the companies that offer direct-to-consumer marketing of genetic tests. In the article, drug metabolism tests are lumped together with tests for nutrition and toxins, and the author describes the validity of all the tests as "the science behind many such tests is shaky." Obviously, it's disappointing that on the one hand we've made such great progress towards testing patients for these critical pharmacogenomic traits, and at the same time have them possibly perceived by the general public as unreliable or unimportant.

Gentris and other companies have worked diligently to bring these new tests to market. These tests have the potential to decrease serious adverse events and allow for a more rational practice of medicine. Most in our industry believe that these tests, like other diagnostic tests, are best conducted in CLIA-certified laboratories. We urge the committee to recommend the necessary legal and regulatory changes needed to ensure that progress is not lost so that we can continue progress towards offering these potentially life-saving tests in the best medical setting possible.

Thank you.

DR. McCABE: Thank you very much.

If you have that typed up, if you could provide it to staff along with the copy that you had from the Reader's Digest, any of that material we would appreciate for our record.

Any questions or comments for Mr. Murphy?

Yes, Emily?

DR. WINN-DEEN: So I was wondering what your recommendation is for the CLIA-certified labs who are also marketing directly to consumers. I mean, just because you're a CLIA-certified lab doesn't basically keep you from unscrupulous marketing practices. It just means that you're performing the test correctly, not that the test has any real utility. So is your AACC committee working on some recommendations in that regard?

MR. MURPHY: No, that committee is actually just working on issues about reimbursement, CPT codes, et cetera. We understand that some CLIA labs might offer tests that have little or no medical relevance, and I'm sure they're validating those tests under CLIA guidelines, and I'm sure they're using physicians to order and report those results. I think what's important is that the marketing be honest and fair to the consumer and that they really know, the consumer buying this test

through a physician. We know that consumers will ask physicians to do these tests. We get calls every day. We are a CLIA-registered lab, and we get calls every day from patients who want 2D6 testing. So we have to refer them back to their physician, following CLIA.

The benefit the patient will get for that test needs to be honestly and fairly described to the consumer, and it is not being done so. More importantly, obvious and well-established medical utility tests, like some of the ones that are coming out now for predicting adverse drug reactions, are being mixed with these others. So I think that's the real issue.

DR. McCABE: Other questions or comments?

(No response.)

DR. McCABE: If not, thank you very much.

Can we have Kelly Ormond come to the table, and then also Gail Javitt, if you could come to one of the other microphones at the table.

Kelly Ormond is from the National Society of Genetic Counselors.

You're the incoming president. Is that correct?

MS. ORMOND: Yes, that's correct. Thank you.

Good morning. I am Kelly Ormond, president-elect of the National Society of Genetic Counselors. As you're aware, NSGC represents over 2,000 member genetic counselors practicing in a variety of medical specialties and including academia, research and biotechnology companies. NSGC is the leading voice, authority, and advocate for the genetic counseling profession.

NSGC thanks SACGHS for taking our prior testimonies and support materials into account when developing the draft resolutions and reports. NSGC feels, with one exception that we will discuss today, that the vision report and included issue briefs accurately reflect our understanding of the issues. We encourage SACGHS to continue to address these issues as proposed and discussed today.

We would like to address three areas: the draft resolution on direct-to-consumer advertising; the draft report on coverage and reimbursement of genetic services; and our concern regarding the draft resolution on genetic education and training of health care providers.

First, with regard to direct-to-consumer or DTC marketing, the NSGC code of ethics states that genetic counselors will strive to enable clients to make informed decisions by providing necessary facts regarding genetic testing. As discussed in the issue brief and SACGHS' draft resolution on direct-to-consumer marketing, many consumers view DTC marketing as providing them with additional information and options regarding their genetic health care, but we must be cautious about DTC efforts that provide misleading or inaccurate information.

NSGC supports an individual's right to full disclosure of all appropriate medical information regarding genetic testing, and that genetic counseling services by a board certified or board eligible genetics professional should be an essential component of any genetic testing program that is marketed directly to consumers.

Second, NSGC agrees with SACGHS' statements in the draft coverage and reimbursement report that genetic counselor billing is limited by the current lack of CPT codes for genetic counseling and by the lack of inclusion of genetic counselors as non-physician Medicare providers. While we recognize the challenges in doing so, NSGC encourages SACGHS and the Secretary's office to consider ways to address these two issues. We also ask that SACGHS promote the development of federal funding to support evidence-based studies of both genetic technologies and clinical genetic services.

As was discussed yesterday, this data can be used in discussions with purchasers of benefit packages such as employers to support the inclusion of genetic services and testing as a reimbursable option within health plans. NSGC has prioritized issues of billing and reimbursement as

one of our three primary foci in our recent strategic plan, and are also working on addressing these issues.

Finally and most importantly, NSGC would like to address the draft resolution and issue briefs on genetic education and training of health care providers. First, we applaud SACGHS' efforts to actively consider the issues that impact the genetics workforce in health care and to recognize the educational efforts which are already occurring. Our greatest concern, which was not the focus of yesterday's roundtable discussion, is that this draft resolution does not address the need for additional training of genetic specialists. NSGC strongly believes that the provision of quality genetic medicine requires the involvement of health care providers of all specialties.

Members of NSGC and other professional genetics organizations have been instrumental in developing and implementing educational initiatives for other health care providers, and we expect that they will remain the driving force towards a broader genetics competence in medicine.

While NSGC does not wish to promote the concept that only genetics professionals can address these issues in health care, it is clear that any future delivery models for genetic services will require the input of individuals with specialty training in genetics and genomics. The NCHPEG competencies state that each health care professional should, at a minimum, be able to, number one, appreciate limitations of his or her genetics expertise; number two, understand the social and psychological implications of genetics services; and number three, know how and when to make a referral to a genetics professional.

These competencies make it clear that non-genetics health care professionals should not be expected to provide comprehensive clinical genetic care but rather to work in conjunction with genetic specialists. When one adds to this the fact that most health care providers are not comfortable with genetic information, particularly in the areas of ordering and interpreting genetic tests, and that fewer health care providers see the immediate clinical relevance of genetic testing and related technologies, it becomes clear that if consumers of genetic services are to obtain high-quality health care, we must ensure that specialists are available to support the primary caregivers and referring specialists.

To echo the statements made yesterday by the American Board of Genetic Counseling, the recommendations to ensure that genetics education and training of all health care professionals is adequate will only be successful if there is an adequate genetics workforce to implement these recommendations. It is also clear that the current number of certified genetics providers needs to be expanded.

Additionally, if we are to address the issues in health disparities raised in Healthy People 2010, SACGHS must also consider the limited cultural and ethnic diversity in genetic professionals, and that most of these genetic specialists currently work at academic medical centers, often limited in their ability to provide outreach to underserved regions or populations. Furthermore, there continue to be multiple impediments to increasing the training pipeline for both medical geneticists and genetic counselors. An infusion of federal funding would increase the number of quality genetic training programs in a short time frame.

Genetics professionals, with their experience across various areas of medical specialization and ability to translate complicated genetic information into non-medical terms, are the ideal professionals to help bridge these training gaps. As NSGC testified at prior SACGHS meetings, to meet the increasing needs of genetic medicine, a two-pronged approach is necessary. First, we must increase the number and diversity of practicing genetic specialists trained in the United States. Second, as SACGHS has recommended in the draft resolution, we must increase the knowledge of health care professionals such that they can perform basic components of genetic medicine and develop knowledge of general genetic concepts and referral resources.

To reach the goals of an educated health care provider population, we must actively work to reduce the barriers to training genetic specialists at the same time that we are working to increase the genetics competence of non-specialists.

In conclusion, NSGC urges this committee to actively address the education and training needs for both specialists and non-specialist genetics training to ensure a competent genetics workforce in the future. NSGC is willing to work with SACGHS to develop an issue brief and draft resolution reflecting this approach.

Thank you, and I will provide a written copy of these comments to the committee for your reference.

DR. McCABE: Thank you very much.

Any questions for Ms. Ormond? Comments? Yes, Debra?

DR. LEONARD: So if there is a recommendation to increase the number of people trained as genetic counselors, are there training programs in existence that could expand to accommodate that extra training?

MS. ORMOND: I believe that this issue was covered when Robin Bennett came and presented to the committee several months ago. There are programs which are willing to consider expansion if there's funding to support that, as well as a number of programs which are in development and trying to establish the funding to get those programs underway, and I believe there are also similar issues facing medical geneticist training.

DR. McCABE: Muin, and then Agnes.

DR. KHOURY: I'd like to applaud the efforts of NSGC. Over time, you have been a good voice in this discussion.

MS. ORMOND: Thank you.

DR. KHOURY: In terms of this evolving nature of genetics and genomics in the 21st century, I'm wondering whether NSGC has discussed or considered the training needs of its own workforce in the sense that as we walk through this continuum from a genetic disease focus, where we are focusing on people with conditions and their families and trying to translate information that could be useful for their psychosocial support and decisionmaking to information that is going to be used in the daily practice of medicine, there is that tension. On the one hand I do appreciate and think there is a big role for the practitioner geneticist community, but in the final analysis the number of conditions for which this kind of practice will be needed will probably be no more than 10 percent of human disease.

So how is NSGC going to or has begun to address this range of genomic information, from somatic cell to polymorphisms, and is there a role for something that we might call genomic counseling, and where does genomic counseling end and health education start, and the practice of medicine? So there is that tension between having more specialists versus integrating the genomics knowledge into the practice of daily medicine. Your thoughts on this will be appreciated.

MS. ORMOND: Sure, my pleasure. I think that genetic counselors have always been a very flexible group in finding ways to take the skills that we are trained in and applying them to the various clinical situations. I think that a perfect example of that is our integration into the cancer genetics setting over the past decade or so. Genetic counselors are certainly aware of this issue that you're raising and it considering it actively.

Within our most recent strategic plan we have raised the idea of addressing scope of practice and have set up committees that include medical geneticists as well as many of our members practicing in different clinical areas to look at how we may become integrated into these various areas of genomic medicine, also looking at genetic service delivery, as we do recognize that many of our more traditional approaches to genetic counseling may not be as applicable to the new mode of genomic

medicine. So we are actively considering those issues and trying to incorporate health care professionals with different views into those committees.

DR. McCABE: Agnes?

MS. MASNY: My question was very similar to Muin's, and I was going to ask if the profession and the curriculum development for people who are coming into the field has begun to look at innovative ways to actually have different tracks, maybe as a genetic educator, maybe as a specialist who would be someone who would then train the trainers so there could be more people in the health profession in general who then could have access to this information.

Lastly, historically I know that the genetic counseling profession did have options for people with a public health background, nursing background, to be able to sit for the genetic counseling exam, and then there were specific requirements, of course, just to have the genetic counseling background. Would there be any opportunities to have a separate kind of track where we could make use of other health professionals that already exist to actually expand the amount of genetic counselors that are out there via different mechanisms of either certification, maybe not necessarily genetic counselor, but genetic counselor associate or something, but that would recognize other health professionals who then would have specific training in genetics and then could sit for the board.

MS. ORMOND: I think some of those questions would need to be redirected to the American Board of Genetic Counseling, who does take care of all of the professional certification. I know that they did change their certification processes. I believe 1999 was the last year that individuals who did not graduate from an accredited program could sit for the ABGC board exam. But certainly the training programs are cognizant of the changing needs, and we're always trying to readdress our curriculum to be training for five to 10 years down the road, incorporating many of these new specialty areas, and certainly educating our new students in areas like billing and reimbursement, health education, preventive services.

I am not aware of any programs that have specific tracks established, nor does the certification exam currently have tracks, but I know that these issues have been discussed and I'm sure will continue to be raised. Does that answer your question?

DR. McCABE: One last brief question and brief comment from Hunt, please.

DR. WILLARD: Your call for increased specialty training in genetics and genomics is clear. What isn't clear to me, though, is whether it's your recommendation that that be done only in the specialty of medical genetics, capital M capital G, or whether you can get specialty training in genetics and genomics in all kinds of specialties.

MS. ORMOND: I think both need to happen. I think that there is historically a difference in the approach to management and assessment in medical genetics as compared to some of the other specialties, and I think we can all benefit from having a little bit of both.

DR. McCABE: Thank you very much.

MS. ORMOND: Thank you.

DR. McCABE: Our next presentation or commentor is Gail Javitt, policy analyst for the Genetics and Public Policy Center at Johns Hopkins University.

MS. JAVITT: I actually have some PowerPoint, so if I could approach the podium, that would be helpful.

DR. McCABE: While those are going up, I'd also point out that there is material in your table folder, the comments on the draft resolution on DTC marketing genetic tests.

MS. JAVITT: Good morning. My name is Gail Javitt, and I am a policy analyst with the Genetics and Public Policy Center at Johns Hopkins University. Thank you for the opportunity to present these comments this morning on behalf of the Center and its director, Dr. Kathy Hudson. We

specifically would like to address the draft resolution concerning direct-to-consumer, or DTC, marketing of genetic tests.

The analysis of DTC marketing of genetic testing must clearly distinguish between advertising of genetic tests on the one hand and commercial availability of these tests on the other. Each of these activities is subject to distinct systems of regulatory oversight and is amenable to different possible policy solutions.

With respect to advertising, the draft resolution rightly identifies the FTC as potentially playing a key role in preventing companies from making misleading claims about genetic tests. But while FTC has a broad statutory mandate to protect consumers, this mandate is circumscribed by two factors. First, FTC may prohibit only advertising that is false or misleading. While establishing the falsity of some genetic test ads out there today would likely be neither difficult nor controversial, as to others, but ambiguity and disagreement can be expected.

Concerns about the impact of DTC ads on consumers that are unrelated to their truth or falsity would not likely provide a basis for FTC intervention. Indeed, the government is significantly constrained by the First Amendment in regulating truthful commercial speech.

Second, FTC must choose its enforcement actions carefully based on the nature and magnitude of the harm caused by the advertising in question. Evidence of this nature does not currently exist with respect to DTC genetic testing. We therefore recommend that the committee consider ways to foster data gathering concerning the harms and any benefits of DTC advertising to consumers. This data could then be provided to FTC and used as a basis for that agency's involvement.

With respect to commercial distribution, the draft resolution recommends that genetic tests should not be sold directly to consumers without the informed guidance of an appropriately trained health care professional. Some will view this position as unduly restricting patient choice. Others may feel such guidance should be required only for certainly types of tests, such as those that predict serious disease. Some may question whether health care professionals are adequately prepared to provide guidance and interpretation of these tests.

These are all important issues for the committee to consider, but these comments are intended to address whether as a practical matter there is a means of effectively implementing the committee's recommendation. Currently, no federal or state entity regulates when or under what circumstances genetic testing services may be commercially offered to consumers or health care providers. It is therefore unclear what entity would now have the authority to implement the recommendation.

The draft resolution recommends that FDA enhance oversight of genetic tests while acknowledging that agency's limited oversight for most genetic testing. FDA regulates genetic test kits that are sold as free-standing products and not genetic testing services provided in-house by clinical laboratories. FDA has therefore had limited opportunity to review only a few DNA-based genetic tests, even though there are genetic tests for over 700 genetic diseases.

This is not the first committee to identify FDA as an appropriate body to provide more substantial oversight, and we do not disagree that FDA involvement could be both beneficial and consistent with that agency's broad public health mission. We question FDA's willingness, however, to step into this arena without a clear mandate to do so, particularly in the absence of more concrete evidence of consumer harm.

The draft resolution fails to mention another key player in genetic test oversight. The Center for Medicare and Medicaid Services administers the Clinical Laboratory Improvement Amendments, or CLIA. Laboratories that provide commercial genetic testing services are covered by the statute. Despite recommendations from advisory groups, CMS has not yet issued proficiency testing

standards for most genetic tests. In enacting CLIA, the Congress recognized the crucial public health role played by clinical laboratories. More rigorous oversight of genetic testing laboratories under CLIA could enhance public health protection.

The federal government has not invested in any entity the ability to serve as a gatekeeper, meaning to decide when and whether genetic tests possess sufficient validity or utility to be used in the clinical setting. This is in contrast to the situation for many other clinical tools used by health care providers to diagnose and treat patients. Some would argue that increased government involvement is neither necessary nor desirable. Others believe that, given the increasing importance that genetic testing is assuming in health care, this gap in oversight could threaten public health.

This committee could play an important role in identifying the benefits and drawbacks of a more rigorous system of oversight.

The draft resolution rightly identifies several areas of potential concern related to DTC genetic testing. At the same time, much remains unknown about this enterprise. Is this a trend that will continue to grow? What is the impact of such testing today, and what can we predict about its future impact on consumers? Sound policy formulation in the months and years ahead on this issue will be greatly facilitated by sound empirical evidence. Thus, it is important that this committee identify the entities best equipped to gather such data and foster a mechanism for gathering these data and studying these issues.

In summary, we recommend that attention be given not only to the dubious claims made for some genetic tests but for preventing genetic tests of dubious value from getting on the market in the first place. To that end, we offer the following suggestions. First, the committee should foster data collection concerning consumer impact of DTC genetic testing, including whether and to what extent consumers are obtaining genetic testing through these means, whether such tests are causing harms or providing benefits, and the nature and magnitude of such harms or benefits.

Second, the committee should consider how CLIA could be harnessed to provide greater oversight of labs providing genetic testing services.

Third, the committee should identify the current barriers to greater FDA involvement and consider a means to overcome these barriers.

Finally, the committee should consider the merits and drawbacks of a federal oversight entity that would set standards that genetic tests must meet before they are made commercially available.

Thank you very much.

DR. McCABE: Thank you very much.

Any questions or comments? Yes, Chris?

DR. HOOK: Just a couple of observations and open questions that you raise. You mentioned the term the importance for consumer freedom or consumer access to information, yet in the vast majority of other medical tests that are available, consumers do not have direct access to those. The reason why genetic testing is being marketed in this way is that it can be done by a buccal swab rather than a blood draw or some other type of invasive means of gathering the information.

So conceptually we do restrict access to the majority of other types of medical information gathering processes without direct access by the consumer. So why are we now saying that we need to make an exception in the opposite direction with genetic information and allow them to have access to that when it's much more complex? That's a conceptual question I want to address to you.

My second one is that, again, I agree with you completely that we need to be collecting data, we need to be compiling a database of examples of potential abuse, trying to find how the public is interacting with this, and I think that's very important. But there still seems to be an inference,

or at least that's how I'm taking your comments, that there needs to be blood on the pavement before we have a warrant to intervene, and I'm not sure I agree with that. I think if we can see that harms are going to be done, as for instance Mr. Murphy was pointing out earlier, that we have a significant amount of ambiguity on proven utility even of the cytochrome phenotype systems and various drugs, why do we have to wait to have people be harmed before we do our appropriate job of recognizing the potential for harm and intervening to prevent that?

MS. JAVITT: Let me start with your second question, because I wanted to just make sure that I didn't create a false impression. The distinction that I'm trying to draw is between information provided consumers and products or tests, actual concrete services. With respect to the services, I think that foreseeable harms could indeed be a basis for intervening before there is, as you said, concrete harms.

With respect to providing information to consumers in the commercial context, there are legal constraints that will come into play, and in crafting any oversight system, those need to be considered. The Supreme Court in the past several years has provided a much higher burden on the government to show that the information itself will cause harm, and part of what they've asked for is facts, facts on the ground. That is the distinction that I'm trying to draw.

Was there somebody else who wanted to respond, or was that another question? I thought I saw a hand.

DR. McCABE: Muin has a question or comment.

MS. JAVITT: Oh, okay.

DR. KHOURY: Actually, we didn't have much time to go into the public health response to the Myriad campaign this morning. We might have a chance to discuss it a bit later. But when you make some recommendations about role of different agencies and you put data collection as sort of hanging in there with no jurisdiction for that in any locality, it also begs the question of who is going to do this. The Myriad campaign taught us a few lessons.

The first thing was where the campaigns were running in the populations in Denver and Atlanta, Georgia, the health departments were beginning to get questions from the general public, from women who were concerned, and that led to the mounting of the public health assessment of what really happened. I think as direct-to-consumer in genetics, or in any other thing -- I mean, without the genetic exceptionalism, has the potential for both hurting and helping people -- somebody somewhere needs to keep their finger on the pulse. That's a function that should be well-defined and is truly a public health function that involves going out and collecting data in real communities involving epidemiologic tools and surveys, et cetera.

As this committee begins its discussion, I think we need to fine-tune that function a little bit more, because policy depends on data. If we don't have data, whether we want more regulation or less regulation or more oversight and different kinds of things, the data collection is so key to putting your finger on the pulse so that the right policy decisions can be made. To me, that data collection is inherently and essentially a public health surveillance function.

DR. McCABE: I think that was more of a comment than a question. Do you have any response?

MS. JAVITT: I just didn't want to forget the first question that you had raised. My understanding in terms of providing testing to consumers directly is that it's a state by state decision about to whom labs may receive samples from and report back to, and that's a state decision rather than a federal one. So there isn't necessarily a distinction between genetic testing in that context and other laboratory tests.

DR. McCABE: Matt, I'll let the FTC have the final question here.

MR. DAYNARD: Well, it's really just a couple of points to clarify the Commission's legal authority for the group. I think all ads would be subject to our jurisdiction. If they're on the Internet, for example, they're certainly interstate, or even if they're local in a local paper. If the lab obtains any part of the test from out of state, it's affecting commerce. So that's not a difficult issue.

I agree with you 100 percent that proving whether it's false or lacks substantiation in the form of comparable reliable scientific evidence is another issue altogether, and it may be difficult in many of those cases to make that burden. But we have the jurisdiction.

The second point is the Commission in terms of deception only requires -- Commission law only requires that an ad be likely to mislead consumers in terms of their purchase or use decisions. We don't have to show blood on the floor necessarily. The unfairness jurisdiction might be a different story.

The third point is that there is simply no per se First Amendment protection for deceptive commercial speech. That doesn't mean we don't have to use reasonable means to the end of regulating it. But the Commission doesn't have the problem the FDA has had in a number of areas because it looks at ads before the fact. So it's a much higher First Amendment burden on the FDA, but we don't typically have that problem if we choose our targets wisely.

DR. McCABE: Any comment?

MS. JAVITT: No, thank you.

DR. McCABE: Thank you very much. We appreciate all the commentators for your input.

I'd also remind the committee that there are written comments that we received that are in your table folders.

This is a nice lead-in to our next topic, which is direct-to-consumer marketing. This was ranked as the fourth issue of our top priority issues requiring in-depth study at our last meeting. However, the committee felt that the topic warranted an immediate response to encourage the Federal Trade Commission's efforts in this area. During the next hour we will consider and finalize a draft DTC resolution that was prepared by the DTC task force. The resolution can be found at Table 6 of your briefing book.

I'd like to thank Chris Hook for chairing this task force, as well as Brad Margus, Agnes Masny, Steve Goodman, Matt Daynard, and Tim Leshan for your service on the task force.

Before we begin discussion, I'd like to remind you that NHGRI organized a workshop on March 23rd to consider DTC marketing of genetic technologies and services. So at this time I'd like to ask Alan Guttmacher from NHGRI to update the committee on the outcome of your workshop, Alan.

DR. GUTTMACHER: Sure. Thank you. I'm happy to do that. I should note that Dr. McCabe correctly identified this as a workshop to consider DTC advertising. This was not, for instance, an NIH consensus panel meeting to come up with specific advice or that kind of thing. I think for many reasons it was not that, including the feeling that there had not been enough time perhaps for the field to really look at this to be able to come up with those final detailed kinds of recommendations.

However, clearly, we like the SACGHS, identified this as an area of some potential concern, so we gathered about 50 people together, and they came from several different kinds of backgrounds. There were genetics and other health professionals, there were individuals that represented health consumer organizations, there were individuals from federal agencies, both regulatory and non-regulatory federal agencies, and there were also individuals who came from various industry organizations, or actually industry both organizations and individual companies.

Basically, the morning was spent in looking at the data. We thought that it might make sense to base policy considerations on data, so we spent the morning looking at the data, what do

we know currently, and you will see there's a workshop summary for you that's right after the draft resolution behind Tab 6. That you can see is a nice summary. The first four and a half pages go over the morning session, the data presented, et cetera. Then the rest of it describes the afternoon discussions, and you will see the afternoon was really informed by the morning data, a discussion of both the question of is there reason for concern here, and if so, what are the reasons for concern. Then also a look at how one might move forward.

You will see on the last few pages that there are three major areas where there was some kind of general -- I'm not sure I'd use the term consensus, but general agreement among the group. The first was that it would make sense to facilitate the development of a stakeholder consensus document or white paper outlining best practices for DTC advertising in the realm of genetic tests, and perhaps in genetic services as well. There was some discussion during the day about the distinction, as you just heard, between testing and services.

The second was to coordinate and facilitate the development of a formal petition to the FTC outlining the concerns with current DTC advertising practices for genetic tests. The third was research, including specific collaborations with the private sector, and the idea of developing a research agenda able to inform future advertising practices and any policy development.

So I would just call the summary to your attention. Take a look at it. We know that some of the people involved in the group that came up with the draft resolution were involved in this meeting, and I hope somewhat informed by it.

I'll be happy to answer any questions about the meeting itself if you have any.

DR. McCABE: So any questions for Alan regarding this conference?

Yes, Emily?

DR. WINN-DEEN: So not so much regarding the meeting but just what does NIH view as its next step? Are you going to stop with having convened this sort of state of the state kind of conference, or do you have specific plans to go on and make some consensus conference or kind of recommendation?

DR. GUTTMACHER: I think at this point we probably are waiting to see partly what this committee does; and then again, since our focus particularly is on research issues, I think we would be interested in developing and are planning on developing a more precise research agenda in terms of what are the research questions that need to be answered to enable good science and policy.

DR. McCABE: Yes, Hunt?

DR. WILLARD: Alan, were there any representatives there from industry?

DR. GUTTMACHER: Yes, there were. We invited a number. Some chose not to attend, and some luckily chose to attend, including, you'll see, there are presentations from Myriad.

DR. McCABE: So Myriad attended?

DR. GUTTMACHER: They did, and gave a presentation, in fact.

DR. McCABE: Yes, Kay?

DR. FELIX-AARON: Following up on the earlier question, were there representatives from the provider organizations, particularly not necessarily hospitals but physicians, because I would imagine that provider and clinician perspectives would be important here.

DR. GUTTMACHER: There were.

DR. FELIX-AARON: There were. And who were they?

DR. GUTTMACHER: I don't remember the names. We can get you the list of the people that attended, if you'd like.

DR. FELIX-AARON: All right. That would be very helpful.

DR. GUTTMACHER: Come see me afterwards.

DR. McCABE: Could you provide that to the committee as well, please?

DR. GUTTMACHER: Sure, we'd be happy to. I don't have any reason to think it's not a public document.

DR. McCABE: Thank you.

Other comments or questions?

(No response.)

DR. McCABE: If not, let's move forward.

Thank you very much, Alan.

I'd like to call your attention to the written public comments that are in your table folders in response to the resolution on DTC, and note the public comments we heard this morning.

I'll now turn to Chris Hook to lead the discussion on the DTC resolution.

Chris?

DR. HOOK: Thank you, Ed.

I'd like to begin my comments by thanking the members of the task force, and as Emily had acknowledged, our tremendous debt to Fay Shamanski and the staff for putting this document together on top of the significant amount of time and work that they had done on the other documents that we've been discussing. Thank you very much for that.

In fact, the process was relatively easy for the task force because of that. The first draft had been circulated for comment. Most of the changes were of a clarifying nature. There was some discussion that I should highlight again to make sure that the concerns were addressed. Brad had raised a good point, and that was that as we bring these concerns forward, with the intent of this document really to encourage the FTC and the FDA and other agencies to begin to put these issues on their radar screen, we did not want to completely close the door on the possibility that there may come a time in which some of genetic testing could very easily be done in a marketer or provider direct-to-consumer relationship.

We hadn't necessarily acknowledged that we were at that stage, but we didn't want to close the door so that that was not a possibility. So as we get to that point in the document, we'll bring that up and make sure that others are satisfied with the language that keeps the door open to some extent in that regard.

In terms of reviewing this document, I would ask if the Chair would agree, because of its brevity, that we just read through it in its entirety so that, rather than starting to wordsmith paragraph by paragraph, we're all again reminded that perhaps a concern that someone has at a given point may have indeed been covered later on in the document.

Is that all right, Ed?

DR. McCABE: Sure, Chris. You missed the discussion yesterday. The document may show brevity. I hope our discussion can be informative but as brief as we can accommodate. Thank you.

DR. HOOK: Indeed.

With that, then, I will just quickly run through the document, and then we can begin the wordsmithing thereafter.

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

"Whereas scientists are daily discovering new genes that play a role in disease and health and developing genetic tests that help diagnose and predict disease, at the present time the majority of the more than 1,000 genetic tests available or in development focus on rare diseases or single-gene disorders. For many human genes, definitive links to a particular disease or health outcome have

not been validated. We are only beginning to understand the basis of the complex links between genes, the environment, and common diseases or behaviors;

"Whereas recent marketing practices which can be directed at both advertising and selling genetic tests and services to the general public have included promotions in print media, television, and increasingly the Internet;

"Whereas there may be valid and appropriate genetic tests directly marketed to consumers, we are nonetheless greatly troubled that some entities are misrepresenting genetic information in order to recommend unsubstantiated health and dietary changes to consumers which in some instances may divert individuals from appropriate treatment options;

"Whereas examples of websites that market questionable genetic tests include those offering genetic profiling to assess risk for diseases such as diabetes or heart disease, genetic testing to predict risks of behavior such as addiction or impulsivity, and nutraceutical products tailored to an individual's genetic profile;

"Whereas SACGHS recognizes that many consumers value access to information about new health care technologies and products made possible through direct-to-consumer advertisements;

"Whereas the Food and Drug Administration, in its role in implementing and enforcing the Federal Food, Drug and Cosmetics Act of 1938, regulates devices to assure that they are not misbranded as a result of manufacturer advertisements and promotional labeling;

"Whereas the FDA currently does not regulate the marketing of genetic testing devices;

"Whereas the Federal Trade Commission Act grants the FTC broad jurisdiction over unfair or deceptive acts or practices and false advertisements for drugs, devices, and services, including genetic testing services;

"Whereas SACGHS plans in-depth study of direct-to-consumer marketing in the future, the committee wishes to express concern at this time about marketing of genetic tests.

"As such and in light of the potential health consequences to individuals, SACGHS believes that genetic tests should not be sold directly to consumers without the informed guidance of an appropriately trained health care professional, at least at this time; and that in order to protect the public interest, we urge the Secretary of Health and Human Services to take the following steps to ensure that the marketing of genetic tests is appropriately overseen:

"Direct the FDA to monitor the marketing of genetic tests under its statutory authority and to continue to explore ways the FDA can enhance the oversight of genetic tests offered as services;

"Work closely with the FTC to act against those companies or providers engaged in misleading marketing of genetic tests;

"Engage other colleagues at the federal and state levels, and health professionals and test developers in the private sector, to promote the appropriate use of validated genetic tests and prevent their inappropriate marketing so that the full promise and benefits of these genetic technologies will be realized for the public good; and

"Encourage the public to discuss the implications of a genetic test with a health professional before seeking a genetic test."

DR. McCABE: Okay, so that's a quick read through. I take it that since Chris was not interrupted, it's accepted in toto?

(Laughter.)

DR. HOOK: So moved.

(Laughter.)

DR. HOOK: Actually, Deb was quite busy over here with her pen.

DR. McCABE: I'm sure that people were just being polite, Chris, and now we'll begin to move through it. Let's try and do this from top to bottom in some sort of organized fashion.

So if we could take "Whereas the Secretary's Advisory Committee on Genetics, Health and Society was established," I think that's straight out of our charter, so I don't think there should be a whole lot of discussion about the first whereas.

MR. MARGUS: I actually have a question about the title.

(Laughter.)

MR. MARGUS: Not to change the title, but a question I think the committee has to consider about the whole resolution, and that is that what we are really bothered by about marketing, because marketing is a very broad term, including advertising and/or selling and delivery, and while the marketing is what we have seen so far mostly, and that troubles us, under different scenarios, maybe not sleazy sensational websites, but if a major pharmaceutical company tomorrow had an ad like the Claritin ads, where they advertise, they have all the little fine print, and then they send you to a physician or a person who knows what they're doing, we don't seem to have a problem with that consumer marketing as long as it's being delivered through the right thing.

So if someone were advertising a genetic test in the same way but it was delivered with genetic counselors and everything, would we still have a problem with it? Are we having a problem with all direct marketing, or is it primarily the concept of selling or delivering the genetic test that troubles us? If that's what it is, then maybe we should be a little clearer and not just say broadly, globally marketing, but we should say it's the delivery part.

We don't have to change the title, but somewhere later on -- for example, after all the whereases, the first thing we say is tests should not be sold directly to consumers. So when we get active about it, we're focusing on the delivery, not that we mind the advertising so much. That kind of became less clear to me after the task force finished.

DR. McCABE: I'll let you respond, Chris, but I think it's important to recognize the point that Gail Javitt made in the public comment session, and that is that there is direct-to-consumer marketing and there's direct-to-consumer access to genetic tests, and those we always have to separate, recognizing of course that direct-to-consumer access is not really going to be terribly profitable unless there is recognition that this access is available, which is the direct-to-consumer marketing. So you can have direct-to-consumer marketing with or without direct access, but part of the concern with genetic testing is the access.

Looking at this, I thought that it was pretty straightforward and it did not confuse marketing and access the way I know some other documents have in the past. But I'll let you respond, then, Chris.

DR. HOOK: I would just echo what Ed just stated in that I think because we did focus on the access, that that was in our specific recommendations, that was highlighting what we thought to be the most important point of our concerns or the focus of our concerns. We could change marketing to break it down to say advertising and sales of genetic tests to acknowledge that we recognize the distinction between the two. But I think as you look at the whole document, it's the sales which comes through. So I don't know that we need to change that.

MR. MARGUS: I just wanted to emphasize that I believe the day is going to come, hopefully soon, when there really are legitimate tests that we may want to communicate to the public are now available, not that we want them to be delivered directly, and when those communications become more common, I don't think we're as much against them as we are against them direct access.

DR. WINN-DEEN: I think the other thing that we have to be very clear about are

tests with an established legitimate medical utility and consequence, so either lifestyle modification or treatment or something that you do with that information, versus the thing that you bring up I think in paragraph 4, the tests that are being put out there with unsubstantiated claims or with specific recommendations for, say, nutraceutical intervention, which are not well established as medically useful and might prevent someone from seeking an appropriately validated medical treatment.

That, to me, is the thing that concerns me as well. So I think we've got sort of multiple scenarios, and I think we almost have to work through what is our stance on validated tests like BRCA1 marketed to the consumer. We have a very good case study with Myriad and that test to discuss. Then we've got the unvalidated thing marketed to the consumer. Do we even think that marketing message should be allowed to go out? And then the third is do we recommend or not recommend direct access to the results without the involvement of a health care professional?

DR. HOOK: Could I just reply to that? Then Cynthia, and then Ed.

I agree that ultimately we will need a firm and clear distinction between various scenarios, as you proposed, but I think we're ahead of where we are trying to be with this statement. In other words, we're trying to bring this to a level of awareness to the leadership of the FTC and to other government agencies. We have not gone through the full process that ultimately this committee will undergo where we will look at those various scenarios.

So my question to you, Emily, is the language of this particular document sufficiently obtuse or opaque as to be making firm statements already that we shouldn't be? In other words, at this point in the discussion, are we overstepping what we should be saying at this point?

DR. WINN-DEEN: I guess my concern is are we ready to have a resolution? Because a resolution to me is sort of a call to action, that you have something specific that you would like to make specific recommendations for action by the Secretary. If we're just informing -- I'm not sure we're totally informed as a committee on some of these things and ready to make a resolution. I guess that's my concern here, not that anything that's in this is an incorrect statement, but do we need to, instead of working on this as a resolution, work on it as what are the specific things we're concerned about, where do we need to get information so we could for scenario 1 say this is our recommendation, scenario 2 this is our recommendation. I just don't know if that's a more productive way for the committee to operate than working through a resolution that I'm not sure we have the information at hand to actually make specific recommendations to the Secretary.

DR. HOOK: Cynthia?

MS. BERRY: Well, Brad brought up a good point which I hadn't thought about until he just said it. But if direct access is what we're concerned about, then it does seem to me -- even though I laughed when he was talking about the title, we probably should change the title maybe and have it be direct-to-consumer access instead of talking about marketing, and we could have in the whereas section an acknowledgement that the reason right now we are taking this stance against direct access to these tests and services is because there is misleading marketing going on out there. There are tests that just simply require interpretation by a competent health care provider, genetic counselor, and people need that in order to reap the benefit of these technologies -- and and and, we can sort of go on and lay it out, and then say therefore right now, we think that there should not be direct access, and we can come up with future recommendations.

Maybe there are going to be circumstances when we get additional information, maybe there are services or tests where direct access might be okay once we satisfy ourselves that it is okay. I agree with Emily that for some of these things, we probably don't have all of the information before us yet, but we probably can make at least this preliminary statement in the form of this resolution.

Conversely, if folks felt that we wanted to make a statement about marketing, because

that is one of the conclusions at the end here -- it talks about working with the FTC to act against those engaged in misleading marketing -- then we could change the title to really include both marketing and access if we wanted to make that conclusion that we want to take a stance against direct access, but in the meantime there's this concern that we do have about misleading marketing that we want to work with the FTC on. I have no problem with that either, but I think we should probably clarify with a view towards going down the path that Emily has talked about, collecting additional information, because we probably need to make more specific recommendations once we get that information.

DR. HOOK: Ed?

DR. McCABE: I just want to remind the committee of why we undertook the process of drafting this resolution, and that was because of concerns about direct-to-consumer marketing. So access is an issue. I think this is really focused on the marketing. I think the one area where it could be confusing and we may wish to delete it so as not to open up other doors but to focus it on marketing is under the bullet one of the resolution, and that is the second clause there, "and to continue to explore ways the FDA can enhance the oversight of genetic tests offered as services."

Really among the resolutions, that's the only one dealing with services. The rest are all dealing with marketing, and part of the concern was the information we had received that indicated that there really was misleading marketing going on. Our interest in moving forward quickly before we did the in-depth study to give FTC primarily, but to some extent the FDA as well, the opportunity, based on this resolution, to within their agencies look into direct-to-consumer marketing, and particularly the misleading marketing.

MR. DAYNARD: My question for the committee would be do you believe that one of the aspects of misleading marketing is that it may fail to tell consumers that the intervention of the medical profession is necessary? Because if that's the case, then it makes a little bit more sense to me to include them both in here.

We had a situation once where there was a very low calorie diet being offered to consumers, and very low calorie diets require the intervention of a medical professional because it can be dangerous to have an 8,000-calorie diet. You lose weight very rapidly and it can be obviously very dangerous to your health. But the advertiser wasn't saying that. It was just offering the very low calorie diet. But part of the marketing problem was that it didn't tell consumers you'd better consult a professional, you could be in deep trouble.

So if the committee thinks that part of the misleading marketing is that omission of material information, then maybe you ought to say so in here.

The other -- well, I guess that's enough for the moment.

DR. McCABE: I was just going to comment that I think that's sort of in there under the final bullet of the resolution, "encourage the public to discuss the implications of a genetic test with a health professional before seeking a genetic test."

MR. DAYNARD: Well, then I guess I'm just saying that it really isn't a distinction -- it is a distinction without a difference, because the access is part of the marketing issue. As long as the resolution focuses on the marketing, then I think you've got it right.

DR. McCABE: It does talk about that marketing practices, under the third paragraph, the third whereas, marketing practices can be directed at both advertising and selling genetic tests and services to the public. But again, then it reverts back to the issue about marketing. It says "have included promotions in print media, television, and increasingly the Internet." So I think that access is an issue, but this is focused on marketing, with the exception of that one clause in the first bullet of the resolution, which we could strike to keep it very focused.

DR. SHAMANSKI: I just wanted to comment on that third whereas statement. In

defining marketing practices, we were trying to make it clear that marketing practices include advertising and selling, which includes access. So if it's not clear that access is included within marketing, we can clarify that. But I just wanted to point that out.

DR. HOOK: Matt, I'm sorry. You were tuning in there to her reply.

MR. DAYNARD: Well, because the FTC has no general distinction between marketing and selling. I mean, if you can sell the service without advertising, more power to you. But it's not likely. So anything that's said -- we're talking about a test kit versus coming in for a test, right?

DR. WINN-DEEN: No.

MR. DAYNARD: Tell me what the difference is between marketing and a service here.

DR. WINN-DEEN: Okay. A kit is regulated by the FDA, and it has very specific claims. Most genetic tests are marketed as services where the laboratory develops the reagents themselves, and then they can market that test for whatever purpose. I think that's the issue, that most of the marketing issues are not around kitted reagents, products that the FDA has reviewed. They're around home-brew lab-developed tests that the lab is in some way taking on a responsibility to create a clinical utility, which may or may not be real.

MR. DAYNARD: Okay. I just wanted to point out that as far as the FTC is concerned, there's no distinction. If you advertise a test kit or a service, it's all in the same --

MR. MARGUS: But there has to be a difference between if you advertise and then people can buy it directly from you, or if you advertise and then people have to go through a channel like a physician to get it, or a genetic testing center or something like that, where you're going to get counseling. From the advertising point of view I know it doesn't matter, but the question we had are what are we most concerned about.

Just to be clear, if we want to cover it all, then I think we should just leave it as it is. That one sentence Ed had said to change is a good idea. But in business school, marketing includes the delivery. It includes not just the promotion and the advertising, but also the delivery, the access. My only question at the very beginning was if you don't really have a problem, if you really think about it is our biggest problem the access part, not the advertising, in which case we should make it more clear. But if we want to cover it all, and since the biggest point Chris made is that this is just the resolution to get it on the radar right away, we're telling them that we're going to be deliberating much more, why don't we just say marketing and we'll get back to them on what parts we really care about later.

DR. HOOK: And just to reinforce Brad's comment, I think that there are elements of delivery, as well as advertising, that can be issues of concern for us. So the broader language isn't necessarily inappropriate for our larger set of concerns. Again, I'm hoping people are not looking at this as the final statement of our conclusions about all the different permutations. That work has to be done. But if we're going to partner with the FTC and others, they need to be encouraged to spend the time and the labor to get some of the data we need to make our final recommendations, and that's what the purpose of this was.

Debra, and then Ed.

DR. LEONARD: Well, Gail Javitt pointed out that there is state to state variation in whether you can market medical services directly to consumers. So I think that has to be taken into account as we decide what blanket kind of statements we're going to make, and maybe the marketing of genetic states has to be consistent with state regulations, unless we want to override those.

What? Oh, sorry. I thought there was a comment back there.

The other concern that I have is that once the whereases are done, we're very strong about saying that genetic tests should not be sold, and we're specifying the selling part, directly to

consumers, but I don't see anything in the bullets that is a mechanism for achieving that. So especially the last bullet, which is "encourage the public to discuss the implications of genetic tests with a health professional," and it's not clear who a health professional is, because many doctors don't even know how to interpret genetic test results, "encouraging" is different than "should not be sold directly to consumers." I think that we wimp out in the bullets, basically.

I don't know why we can't just make the statement that's the beginning of the paragraph if we're going to do further investigation. Maybe, following up on Muin's comment, ask the CDC to start collecting information in the public health interest of what marketing is going out to consumers and if there's harm, et cetera. But basically have two bullets, one is that it shouldn't be sold directly to consumers, and ask the CDC to collect more information to inform out future discussions.

DR. McCABE: Well, I was going to, I guess, wimp out completely, then, since we hadn't addressed that in the bullets, and remove that and take the first sentence of the lead-in paragraph to the resolutions and say, "As such and in light of the potential health consequences to individuals, and in order to protect the public interest, we urge the Secretary", and take out that about the selling, because I think that brings us into state issues about differences state to state, and what we were really trying to do was get something quickly out to the Secretary to make some recommendations to those agencies that fall within his purview, which here was directed at the FDA.

I think if you wanted to include some sort of monitoring of direct-to-consumer marketing of genetic tests, that would be good in a bullet, and that could be covered here. But I think it's important that we keep this tight and focused so that we can get it out quickly.

DR. HOOK: Emily, and then Hunt, and then I'm going to suggest a procedural approach thereafter.

DR. WINN-DEEN: Just listening to the discussion, it sounds like what we're really recommending is not that we direct FDA to monitor but that we basically declare a moratorium on direct marketing and access to genetic tests until such time as we can come up with some very specific recommendations for the appropriate level of -- I'll call it oversight, but I don't want to confuse that with the sort of standard lab practice oversight.

But we need to have some very concrete recommendations for the different scenarios. So for the scenario of an established medical utility with a reputable provider, how do you know when a test should be allowed to go directly to a consumer or when it needs to involve a health professional? We need to discuss that in depth and come up with a recommendation. Are there any scenarios where a consumer should be able to get direct access, and what are those?

Then the next level, which is to me the most concerning, are the tests which are being falsely advertised. So they're making claims and/or recommendations which are not substantiated. We need to have a mechanism for policing those things. What is the right mechanism? Is that FTC? Do we just send all that stuff over to Matt and hope that he has some time to deal with it? How do we get to those things? I've seen some of them that I personally find extremely concerning, primarily because they're recommending alternative therapies when there are good established, FDA-approved therapies available which are not mentioned.

So I think I'm a little concerned about direct to the FDA to monitor the marketing of genetic tests as our first -- it sounds, first of all, very directive over an agency which has waffled on whether it even really feels it has the authority to regulate the delivery of information from clinical laboratories. So I'm not sure this is the right time to try and tell it that it must take that authority in hand. But we need to have some clear statement here about for the time being, this should not be done, period, and what are the circumstances under which we would recommend moving away from this should not be done, period, to where is it appropriate.

DR. WILLARD: I guess I'm coming down on a similar side. If I was the Secretary, I'd look at these four bullets, and it's not at all clear what I'm supposed to do, and none of them has any teeth, so it does come off as a very wimpy sort of approach. The closest one to a true action item, other than hopefully give Matt a bigger budget, because we heard at the very first meeting that he can't possibly do all this stuff, so telling the Secretary to tell Matt to do all this stuff isn't going to be very effective.

The closest one is the last bullet, although the language "encourage the public" is not very meaningful. I mean, are we asking for an advisory bulletin? Are we asking for a moratorium? And if we're not ready to say what we want because, after all, we need to study this, and we've already said we want to study it in depth, then perhaps we're at the point that Emily made 10 minutes ago, which is that we don't really need a resolution because we're not resolving anything at this point. We're not resolute in anything until we've done the in-depth study.

DR. HOOK: At this point, I see us potentially going through a couple of approaches. I think that, one, I'd like to clarify, because there's a whole variety of proposals about what this statement is supposed to be accomplishing still circulating among the discussion, and I think we have to come to closure on what it is that we're going to achieve today and what it is we're deferring to a larger amount of effort and intervention in the future.

I would submit that we are, again, attempting to assist Matt and his colleagues and others in getting this to be an issue of consideration by the government. I think this is an opportunity to ask our colleagues in public health to begin collecting the data, as was previously mentioned. I think that we are suggesting that, at the present state of the art and practice, we can change the language to be more firm, but that genetic testing should be done in the context of a relationship with appropriately trained health care providers who can help to know whether it's worth doing the testing or how to interpret the results of that testing.

In terms of going down the road of taking each possible permutation, obviously we're not there yet. But I don't believe that we have to have that information in order to go forward with this discussion.

So I would propose, with the Chair's comment, that we at least quickly review the paragraphs of the whereases, the background information, the fact set that's bringing this question forward. Are there any modifications we need to make to those statements? And hopefully we can do that briefly, and then spend the remainder of our time wordsmithing at least the bullet points at this time as to what we think is appropriate.

Yes?

DR. McCABE: This is Elizabeth Mansfield from FDA.

DR. MANSFIELD: FDA. I'd just like to make the comment that I think that FDA has probably very limited jurisdiction, if any, to monitor the marketing of genetic tests over which they have no oversight. I don't believe that we can do that unless somebody decides that we will have oversight over these tests.

DR. HOOK: Forgive my ignorance, but don't you have oversight monitoring over pregnancy tests and things of that nature that are directly marketed?

DR. MANSFIELD: We have oversight over test kits but not over laboratory-developed genetic tests, which is the majority of genetic tests.

DR. McCABE: But it says "under statutory authority," which would mean at this time kits.

DR. MANSFIELD: We do monitor the marketing of kits, and so far that hasn't been a serious issue.

DR. HOOK: So when a direct-to-consumer marketer of genetic tests sends a packet

out to collect a buccal swab and to return that information, are you monitoring those and giving them approval to do that?

DR. MANSFIELD: Not unless the kit has been cleared or approved by the FDA. Most of those direct-to-consumer are lab-developed tests and are outside our regulatory authority at the moment.

DR. HOOK: But then you would have regulatory authority to step in to make sure that the collection mechanism is appropriate. Is that not correct?

DR. MANSFIELD: No, not that I'm aware of.

DR. HOOK: It's a marketed kit for the collection of --

DR. MANSFIELD: Right, but we don't regulate laboratory-developed tests, so we don't have any jurisdiction over how they're marketed.

MS. HARRISON: Just a point of clarification. Would it be that the collection itself was monitored but not the test that's done on it? Is that the --

DR. MANSFIELD: Collection devices are regulated as collection devices, how they perform; for example, blood tubes and so on. But the actual collection of the sample by whoever is not.

DR. McCABE: I guess before getting into the details on this and spending a bit of time on it, I would just ask our ex officios whether they -- the purpose of this was to assist the ex officios in the interim while we developed a more complete report. So I would ask the ex officios whether they see any value in developing this resolution. If it is not going to be of any value, I think we need to then consider next steps.

Is that okay, Chris?

DR. HOOK: Matt?

MR. DAYNARD: Well, first, I can't speak for the FDA, of course, but I deal with them every day. As you know very well, their charge is to protect the public health and safety. If the committee's concern is the public health and safety in the direct-to-consumer marketing or delivery of these tests, I might suggest you want to make a stronger recommendation now or later to the Secretary to try and get implemented a change to the FD&C Act. I mean, if you don't have authority over these things, it's going to be very difficult for the FTC under any circumstances to do the kind of job you'll want to get done without the help of the FDA, because we can't do our job anyway without the help of the FDA in many other areas.

So that's important. All throughout this I've been wondering, and folks at the FTC have been saying, well, this is really FDA's job. We're talking public health and safety here, aren't we? Why should we be concerned about marketing? I have a decent response for them, but it's not a complete response because they're likely to feel just that way, that if the committee's concern is public health and safety, then, darn it, the FD&C Act should cover them.

But yes, I do feel that a resolution is important because the FTC at some point should be involved, and maybe in the near future this resolution will help get it on the FTC's radar screen. So I am in favor of it.

DR. McCABE: Taking Matt's comment, would it be appropriate, if we decide to move forward with this and just keeping track of these things as they come up, to change the first bullet under the resolution, "Direct the FDA to monitor the public health and safety impact of the marketing of genetic tests under its statutory authority"? Is that something that would be more acceptable, Elizabeth?

DR. MANSFIELD: I don't know that we have any statutory authority to do that now, but if it were to change the Act, if your intent is to get the Secretary to change the Act, then possibly yes.

DR. McCABE: Our concern is for the public health and safety. That's why we decided to move forward with this resolution and we felt that it could not wait for the in-depth study.

DR. MANSFIELD: Yes, I understand that. However, at the moment, lab-developed genetic tests are considered to be practice of medicine or a service, and we don't regulate the practice of medicine or services. So the Act either would have to be changed or someone's mind within the FDA would have to be changed in order to give us oversight.

DR. McCABE: Well, is a better way to go, then, to say "Direct the CDC to monitor the public health and safety impact of marketing of genetics tests"? I mean, if this isn't going to be helpful to FDA, and if we're concerned about the public's health, then perhaps we move it to look at another agency.

DR. HOOK: Please, Paul. Go ahead, Paul, and then we'll go down the list here.

MR. MILLER: This is not particularly my area, but in listening to the conversation, two things sort of strike me. One is that what you might think about doing, if the FDA is an HHS agency, and the FDA is not sort of embracing regulatory authority in this area, is that within the Secretary's purview to interpret the statute in such a way that the FDA does have regulatory authority? Is there enough leeway within the statute? Then, in fact, that would be an action resolution, to say that we recommend to the Secretary that the FDA be used in this way because there is a health and safety issue, there's a gap, and we think that the Secretary should interpret the enabling statute in that way.

I'm not sure sort of the authority of that, but that may be one path to go down or one roadmap to go down, so to speak.

The second thing is that if in fact there is no leeway in the FDA's enabling statute, then it would strike me that that is sort of the problem. Then that would, in a sense, be a recommendation for the Secretary to sort of engage in a legislative agenda to change the statutory authority of the FDA to get them engaged in that such that the FDA would clearly have jurisdiction over these issues, which I think is the sense of this committee, and would enable FTC and FDA to work -- so that the issue doesn't fall through the cracks.

DR. HOOK: In an immediate reply to that, I think that a way to communicate that is in the second portion of the first bullet, to explore the potential need and ways for the FDA to enhance the oversight. We're trying to expand their jurisdiction in that.

MR. DANNENFELSER: If it sounds like there's a general consensus that they should do this if they can do this, I would suggest not changing it to direct the Secretary to make a certain interpretation. That may not seem appropriate. But it certainly would be appropriate for him to ask the general counsel to explore what authority the FDA may already have, and if it's limited to then seek further legislative authority if that's necessary.

DR. HOOK: Joan, then Debra.

DR. MANSFIELD: In fact, it is a question of interpretation, what is a medical device, and our general counsel has pretty much come down, to my knowledge, that in-house developed tests are not medical devices. They're services already. I believe the Secretary could probably affect that interpretation if he chose to, but currently that is the interpretation of general counsel.

DR. HANS: Is that an opinion that's been put out in the public that you could provide to the committee?

DR. MANSFIELD: Actually, I don't know.

DR. McCABE: We have been told in previous meetings that it was still under deliberation but that no opinion had been given. So if that has been an opinion that has been given, it would be helpful to this committee to have that provided to us.

DR. MANSFIELD: Well, perhaps I'm overstepping my bounds in saying that, but to my knowledge that is the interpretation that general counsel has made, and I will find out if that's available to the public.

MR. MILLER: But that's an important issue that really needs to be fleshed out as a starting point for this discussion and for the committee to understand where it needs to go.

DR. HOOK: Joan?

DR. REEDE: I think there are two points. One, I think fleshing this out as we go through our conversation here, this need to reflect the fact that these issues are getting fleshed out, that there are assumptions about what is marketing versus advertising versus direct selling, there are issues about what's the purview of this organization versus that organization, and I think the conclusions of this discussion are not reflected in here. It just leaves the next body to have the same set of questions being asked.

The second part is that I think a lot of what is driving some of this is the need to monitor or to know what's going on in terms of the testing, not the test kits per se, which the FDA is monitoring, but the testing. When I look at the whereas that leads to this, there's nothing that really reflects the fact that we don't have a current mechanism for monitoring what's going on with regards to these tests and the public health, and I think we need that background to lead to a bullet about CDC or anybody else doing that monitoring.

DR. HOOK: Debra? Kay?

DR. FELIX-AARON: In terms of listening to the conversation, I recognize the tension that was stated earlier between whether this committee is ready for a resolution or not. I would like to propose that the committee suggest that if not a resolution but a communication to the Secretary would be helpful. I certainly appreciate the reservation of the committee in thinking that whether the level of discussion is at resolution, but I think there is also value in communicating to the Secretary that those issues are coming up, that the committee is deliberating those issues and would prepare the Secretary for a future resolution if the committee did arrive at that resolution.

DR. HOOK: Deb, then Brad, then Emily.

DR. LEONARD: I am very concerned as a laboratorian who does laboratory-developed tests. We prefer that term instead of home brew, but we can't seem to get it out there. Laboratory-developed tests are regulated under CLIA. So I think we need to be very clear about what we're talking about. It's the direct-to-consumer marketing that's the issue on the table here and not general oversight of all laboratory-developed tests that may be done under CLIA, and that's not clearly differentiated when you make statements about ways the FDA can enhance the oversight of genetic tests offered as services, because my laboratory and many CLIA-certified laboratories offer genetic tests as services because they aren't done through FDA-cleared test kits, but they aren't marketed directly to consumers. They are used in ways that would not be questioned by this committee. They're ordered by health professionals on behalf of consumers to make a diagnosis, et cetera.

So I think any communications have to be very clear that we're not moving into this area of oversight of laboratory-developed tests, which is regulated under CLIA.

MR. MARGUS: Mr. Chairman, I need to interrupt for just a second. I have to actually run to the airport, rush to the airport. Inasmuch as this is my last committee meeting -- I'm rotating off -- I wanted to interrupt and just say to everyone that I've appreciated being on the committee and I've been honored to be on the committee, and I've appreciated everyone's tolerance of my naivete over the last year on certain points. Many of you probably have never seen a professionally trained -- what do we call them? -- appropriately trained health care professional on genetics, but I think all of you should.

I'm now going to leave the committee and go read my horoscope and take action without any advice from a professionally trained advisor and maybe buy a beer without a professionally trained advisor, and maybe drive a motorcycle without a helmet and buy some prescription drugs on the

Internet without any guidance from a professionally trained advisor.

But anyway, I have appreciated it. I'm sorry to interrupt, but I did very much appreciate this year. Thanks.

DR. McCABE: Brad, we very much appreciate your service on the committee as well.

MS. CARR: Before you go, Brad, I just wanted to say that we hope you come back to the October meeting. Both you and Kim, your appointments were expired officially in January, but you've been extended, and until we have a replacement for you, we hope you'll both come because, for one thing, you've got to get your certificate.

DR. McCABE: And just to point out, you've been chaired by also an expired Chair, because likewise my term as Chair was up. So just as I'm continuing to chair as you head off to the airport, we hope you'll come back.

MR. MARGUS: So I guess I'll hold off on all that decadent behavior after all.

(Laughter.)

MR. MARGUS: Thank you.

DR. McCABE: Thank you, and have a safe trip.

DR. HOOK: Emily?

DR. WINN-DEEN: It seems to me that what we need is we need to be very clear that what we maybe are asking for the Secretary to do is to clarify for all who or which agency has the authority to regulate false and misleading advertising and delivery of services to the public. We're not concerned about things delivered through the right health care channels, through CLIA-certified labs. This is my concern, that there are CLIA-certified labs that are also advertising things for which the clinical utility has not been established, for which many of us in this room might say they are making false and misleading claims, and I think we need to be really clear about who is the policeman for that, and I don't think anybody here knows.

We've had a discussion is it FDA if it's a health and safety issue? Should it be FDA? FDA, as a result of the SACGT recommendations, had a pretty clear re-look through their general counsel at what they believe is their statutory authority, and I think Steve Gutman has repeatedly said to this committee that the current belief within the FDA is as Elizabeth represented it today, that they don't believe they have the authority to regulate lab-delivered test results.

So who does have the ability to regulate that, and who should we turn to when there's an issue? I think that's one thing we have to ask the Secretary to clarify, and maybe the way they clarify it is by looking with their general counsel through all the different groups and find out if this is a loophole or if there is some group that just really hasn't been given this as a charge.

So I think that's one thing I'd like to get on the table. The other is that I think that in the paragraph that precedes the bullet points, that we really should pull out the bullet point which reads "SACGHS believes that genetic tests should not be sold directly to consumers without the informed guidance of an appropriately trained health care professional, at least at this time." We should make it very clear that that is our key recommendation, and the rest of the things we're looking for some clarification, guidance, and data gathering. Who is the right group to do the data gathering? Is it CDC? Who is the right group to look out for the health and safety of the general public? Is it the FDA? We just need that very clear so that that can be communicated.

DR. McCABE: Judy has one comment.

I think you're volunteering to take this on, Judy? Is that right?

MS. YOST: Actually, my comment is different. I'm just agreeing with Emily and agreeing with Kay in that I think that this recommendation, or whatever you want to call it, this

resolution, is too conclusive for what we know. So I think that you are, to me -- and this is my personal, not CMS, opinion -- that I think you're interfering with individuals' and the public's freedom of choice here without having enough information to determine that. So I think that the most we can do is what Kay had suggested, send some kind of preliminary statement to the Secretary about our concerns and that what we suggest could be done, including explore whose responsibility this is sort of thing.

But I think that that is way far too definitive for where we are. I don't think we have enough information to stop the public's access to information on the Internet, which is something that's kind of broad. I think the point is --

DR. WINN-DEEN: No, I wasn't suggesting we stop their access to information. It was the delivery of health care results to them directly.

DR. McCABE: What I'm going to do, because our lunch is --

MS. YOST: But there are laws that do require that the public does have access to health-related information, like HIPAA. I mean, there's nothing wrong with encouraging that -- whatever term you want to use, caregiver, health care professional, whatever. You can certainly encourage that that happen, that there be an interface between that public and a health care professional. But I don't believe that you can just recommend stoppage completely of this kind of service, because there is, as you suggest, a hierarchy there. There are perfectly legitimate tests, and we don't have enough information to say whether or not they should include the intervention of a health care professional directly or not.

So since we don't have that hierarchy, I think to do a blanket statement is way too, to me, very strong at this time. That doesn't mean that maybe if we have further data or information that indicates so, that we shouldn't proceed. But I think this is way too preliminary at this point.

DR. HOOK: A quick reply from Sherrie, Joan has a comment, and then I'm going to turn it back over to the Chair.

DR. HANS: Just quickly, reflecting on this discussion and not the previous meeting but the one before where you were discussing with the FTC these issues, it seems that the grave concern and the concern for immediate action that brought you into this discussion really was the false and misleading advertising, and there are many other issues that have sort of come out here that I think the committee intends to pursue much further over the next year or so.

So perhaps what you simply want to do at this time is to just focus on the false and misleading part and encourage the Secretary to work with the FTC or the Secretary to direct the appropriate HHS agencies to work with FTC on that piece of it, and let him know that you'll be coming back with various other issues, because that's also where you actually have an oversight mechanism right now. In access you have no mechanism to begin to address. You can say that folks shouldn't be directly accessing these tests without an intervening health professional, but there is no regulatory/statutory mechanism that you have at this time to get at that, and that doesn't really provide the Secretary at this time with any idea of how to proceed since there's no regulatory hook, if you will.

So my suggestion is that you just focus on the very narrow concern that really raised this issue as something you wanted to deal with immediately at this time.

DR. REEDE: My comments are along the same line. As I've listened to more and more of the discussion, I'm at a point where there are more questions here than there are answers. I think for us to try to draw conclusions with these questions out there and a lack of clarity among all of us I think is premature. So I think, again, being able to follow up on this, the issues are around the sort of false advertising, et cetera, being able to speak directly to that.

I think there is a place to say we need to collect more data, more information about the extend of this or how it might be impacting the public. But to go beyond that, I really feel uncomfortable because there are too many issues that have been opened that I don't understand the

ramifications of.

DR. McCABE: Well, thank you, Chris.

The sense I have of the committee is that what we -- and I'm going to make a proposal and then see if this is acceptable. Rather than taking this as a resolution, take it as a letter to the Secretary informing the Secretary that we have had this information presented to us, that we are concerned about false and misleading advertising, that that is the issue, that our concern is that it's not in the public's benefit and perhaps to the detriment of the public's health, that we will be gathering more information, that we would like to have the Secretary identify ways that agencies under his jurisdiction can begin to work to identify what the impact of this is, and suggest that the Secretary also needs to have agencies work with the FTC to deal with the issue of false and misleading advertising.

Is that acceptable? Is there anyone who disagrees with that approach?

(No response.)

DR. McCABE: So we'll use some of the background from the resolution, but of the resolved parts we will only deal with the single bullet concerning false and misleading advertising.

Yes, Chris?

DR. HOOK: Well, to the extent I think a number of very valuable suggestions and observations have been brought up today, that we could bullet as clarification information gathering recommendations and a whole variety of actions that will be necessary for our subsequent deliberations. But we ought to at least put forward the request for that now in very clear terms based upon just the areas that need to be resolved from our own discussion this morning.

DR. McCABE: I think what I would ask staff to do is take the discussion and help to inform us, but we'll leave that in the letter to the fact that we're planning to do an in-depth study and include those as aspects of the in-depth study. We'll inform the Secretary that we're planning to do the in-depth study. Is that okay? Is there anyone who disagrees or has any comments on that approach before we break for a very brief time to gather our lunch?

(No response.)

DR. McCABE: If not, then I will consider that the silence is empowering us to write that letter to the Secretary, and I will send it out before the next meeting.

Thank you very much, Chris and the task force members, for helping to move us forward on this very important area.

So, 10 minutes. Please gather your lunch, come back in. We're having a working lunch, and we still have a bit of work to do with yesterday's resolution and additional steps to where we go from here.

(Whereupon, at 11:50 a.m., the meeting was recessed for lunch, to reconvene at 12:00 noon.)

AFTERNOON SESSION

(12:11 p.m.)

DR. McCABE: We're going to go ahead and get started. I apologize to everyone for rushing everybody's lunch, and especially I apologize to Joan.

The first topic is going to be the resolution on genetics education and training of health professionals. It's been revised since yesterday, and it's sort of been undergoing continuing revision, and I'll let Joan take the lead.

Again, I'm sorry, Joan.

DR. REEDE: No problem. While we're waiting for it to go on the board, I wanted to make a few comments first. Thank you to the staff for helping us get these revisions in, and to members of the task force, who met this morning before our meeting started to go over some of those revisions.

Secondly, I wanted to make a statement in general about some of my philosophy on wordsmithing and what we're trying to accomplish. One of the things I think that can sometimes be difficult with these kinds of documents is when you try to be all-encompassing and to do everything with one document. So after a while, you actually start to lose what was the purpose of the document. So I think we have tried to capture the conversation from yesterday, the edits from yesterday, and at the same time tried to make this simple and direct, with an understanding that we can come back and this does not have to be the final time that this committee speaks on education and training, and that if there are areas that the committee would like us to explore further or go into more depth in the future, that can be done, as opposed to trying to do everything now.

Now, I'm going to do what I did yesterday, and I had not known that I was going to have to develop skills of looking behind me and in front of me and twisting and speaking at the same time when I came on this committee, so let's see if I can figure out how to do this.

I'm going to go down them, read them as they are. If there are comments, I'm going to try to bring them up as we go forward. If they are small changes, if you could hold on to them. I'm looking more for general concepts that we're off on.

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

"Whereas advances in genomics will lead to a more precise understanding of disease processes and will provide better guidance on the application of therapeutic and preventive strategies that will make significant improvements in health status and outcomes;

"Whereas insufficient education and training in genetics and genomics has led and may continue to lead to inaccurate or delayed disease diagnoses, misguided disease management family planning, increased health disparities, and excessive costs;

"Whereas appropriate and adequate training and education in genomics is crucial for all health care and public health professionals to assure appropriate, effective, and efficient integration of genomic concepts and genetic technologies and services throughout the entire health system;

"Whereas appropriate education in genomics is crucial for the general public to take advantage of the benefits of genetics and genomic advances;

"Whereas education of health care and public health professionals and the public is necessary to assure equitable access to genetic and genomic technologies;

"Whereas education of health care and public health professionals and the public is a necessary component of the application of evidence-based medicine related to genetics and genomics;

"Whereas through a survey of federal agencies on their role and activities in genetics and genomics education, training, and health workforce analysis, it was found that federal efforts are focused on translation and appropriate integration of new genetics and genomics technologies into health care and public health;

"Whereas a solicitation of information from educational and professional organizations identified the following urgent needs in genetics and genomics education and training:

"Inventoried, catalogued, widely relevant clinical and public health applications stemming from advances in genomics;

"Educational models that use such applications to clarify how genetics and genomics,

through the use of family history tools, information technologies, and Web-based practice tools, among others, should be integrated into practice;

"Incorporation of genetic and genomic competencies into national accreditation and re-accreditation standards;

"A broadening of the focus of genetics education and training to incorporate both genetics and genomics;

"Assuring the diversity of the health care and public health workforce and the cultural competence of its members;

"Increasing the presence of faculty appropriately trained in genetics and genomics;

"Training programs that address the interface of an interaction between genomics, their ethical, legal and social implications, and public policy.

"As such and in light of the importance of ensuring that the benefits of the genetics/genomics revolution are accessible to all Americans, SACGHS urges the Secretary to take the following steps to ensure that genetics education training of all health care and public health professionals is adequate:

"Promote and actively incorporate into departmental policies and programs the philosophy that genetic information, which includes family history information, should not be treated as exceptional but rather as part of the spectrum of health information and viewed as an integral part of the practice of all health professionals;

"Incorporate genetics and genomics, including family history tools and point-of-care educational support, into relevant initiatives of the Department of Health and Human Services, including the Secretary's Health Information Technology Initiative, and engage in the dissemination of this knowledge to health care and public health professionals;

"Promote and support initiatives that address the integration of genomics into the education and training of all health professionals.

"In order to facilitate the integration of genomics into health care and public health now, direct HHS agencies to work collaboratively with the state, federal, and private organizations, such as NCHPEG, to support the development, cataloguing, and dissemination of case studies and practice models that demonstrate the current relevance and applicability of genomics to health care and public health;

"Provide adequate program and technical support to federal programs that provide for faculty training in the implementation of clinical application-based genomics education models, particularly models using clinically relevant examples and that incorporate the ethical, legal, and social implications of genetics and genomics;

"Promote communication among all health professionals to enhance the accessibility and widespread dissemination of genomics educational models and applications, and raise awareness among all health professionals, faculty, and professional educational organizations of these resources;

"Work with ASTHO and other relevant organizations to address issues associated with incorporating knowledge of human genetics and genomics into accreditation, licensure, and certification;

"Continue to encourage support and facilitate programs that address the need for workforce diversity and cultural competency of health professionals, including sensitivity to the disability community;

"Provide adequate support for efforts that will incorporate a genetics/genomics focus into pipeline programs supported by HHS;

"Promote culturally appropriate and sensitive public education that provides the

knowledge and skills that consumers require to participate effectively with health professionals in decisions that increasingly are informed by genetic perspectives."

Comments? Questions?

Emily?

DR. WINN-DEEN: So just a minor point. I think NCHPEG and ASTHO should be -- you should say what they are, because in alphabet soup-land, not everybody knows.

DR. REEDE: Okay.

DR. WILLARD: On that same point, I guess I would question why NCHPEG was being singled out. I mean, I recognize that that's something they are doing, but it's not like they're the only organization that has been charged with doing it through some official channels, as opposed to ASTHO, which actually is charged with dealing with some of those issues.

DR. REEDE: I think the conversation yesterday and the general consensus was that NCHPEG should be listed specifically as an example of an organization that is doing this. So that was a general consensus of yesterday's discussion.

DR. LEONARD: In looking at basically our recommendations, many of them say sort of in vague terms what should be done, but I can imagine Secretary Thompson, having presented recommendations to him at one point from laboratory or pathology organizations about genetics and genomics and molecular diagnostic testing, his question was always, with every recommendation, how do you propose that I do that? There are many things on this list that we're not making a specific recommendation. It's relatively vague and philosophical rather than an implementable recommendation.

So, Ed, maybe you've done this a lot more than the rest of us and you could comment on the need to be directly implementable, as opposed to philosophical.

DR. McCABE: I think that there's a role for both, that sometimes if we have mechanisms that we wish to use to recommend for implementation, we should give them, but sometimes it's redirecting issues philosophically as well. So as direct as we can be, we should be.

DR. REEDE: Paul?

MR. MILLER: The paragraph that's up on the screen, to promote and actively incorporate genetic information, should not be treated as exceptional but rather part of the general spectrum of health information and so on, that might flag. That might have an impact if that's the sense of the committee. That might have an impact on the nondiscrimination legislation in the sense that one of the arguments against genetic nondiscrimination legislation is that genetic information is not exceptional, it's just regular old health information. So why should we treat for discrimination purposes the use in privacy and so on of genetic information any differently from anything else?

That is a very strong undercurrent in that debate around the nondiscrimination language, and there are a couple of sort of terms of art or buzzwords in there that I would be concerned if all of a sudden somebody grabs onto that language and says, well, here's the Secretary's Advisory Committee saying that really genetic information should not be treated as exceptional, and therefore undermines this committee's other sentiment around nondiscrimination legislation.

DR. REEDE: Right. I think that part of our discussion in the past had been to look at exceptional based on whatever topic we were looking at in terms of how it would be incorporated. So one suggestion might be that it not be treated as exceptional with regard to education, because I think we're speaking specifically that with regard to education it should not be treated as exceptional but rather as integral. With that change, would that address the issues that you're raising?

MR. MILLER: Yes, I think that would make it much more clear.

DR. GUTTMACHER: I wonder, though, whether -- Joan? Alan, over here. I wonder, though, is "exceptional" really the right word for this context? I think perhaps it isn't. It's not

exceptional versus integral I think in this context. It's the idea that we want it not as freestanding, not as just genetics someplace but that it needs to be integrated.

So I wonder whether we could just get rid of the not treated as exceptional and just say should be treated as part of the spectrum of health information and viewed as an integral part of the practice of all health professionals, something like that.

DR. FEETHAM: That was going to be my comment also. I agree with that.

MR. MILLER: The word "exceptional" is really a buzzword.

DR. FEETHAM: It's integral, that you really want it to be part of the practice.

DR. FELIX-AARON: Yes, I support that.

DR. REEDE: So it sounds like a consensus that "exceptional" gets taken out and what we're really trying to reflect is that it should be integral and not as a stand-alone, separate, optional piece.

Other comments, suggestions, questions, changes?

DR. WILLARD: Just in general, whenever we finally get done with this, there's a lot of inconsistencies, grammatical inconsistencies. The HHS is referred to about four different ways and no obvious rationale for doing it. That's just a staff issue at the very end of the day.

DR. REEDE: If we can leave it to staff to make sure that we're grammatically correct and consistent, and our acronyms are defined, et cetera.

DR. FELIX-AARON: A question, Joan. I don't have the document in front of me, but as you were reading it, I thought that there were many recommendations that we had, and I was just wondering how many recommendations do we have, and do we want to be parsimonious? Again, trying to balance that with the need to have the things that we think are valuable represented in the recommendations.

DR. REEDE: There ended up being 10 recommendations in the end, and that partly came from yesterday's where there were some recommendations that were split from one into two.

DR. FELIX-AARON: I mean, I don't have a clear recommendation, just to share the sense with the group.

DR. REEDE: I think one of our concerns was making sure that it didn't become just a long laundry list that people would not pay attention to but rather ignore. I don't think we ventured into that laundry list territory, but I do think it's something to pay attention to.

DR. LEONARD: Has anybody looked at these and prioritized them so that the ones with greatest significance or impact would be at the top? I don't know that there's necessarily a rule that the ones at the top are paid more attention to, but they might be.

DR. REEDE: We have not tried to prioritize them, but the first step that we took this morning was actually just trying to put them together. So if there were two that related to culture, they actually flowed one behind the other. But we did not try to prioritize them.

I think the consensus from yesterday was that we should start out with the general one that refers to incorporation of these concepts across the various agencies, and that would be the strongest, and then we went from there. Is it the wish of the committee that we try to prioritize these?

MR. MILLER: If I can just make a statement, I'm sort of getting dizzy watching the document jump back and forth. I think I'm on the whirlybird at Disneyland.

What would be helpful for me, since nobody has the printed document in front of them, although I hope nobody has it because then I would feel very lonely not having it, to go through paragraph by paragraph and really tick it off, because it's really hard after the initial read-through and having it jump around to really sort of sign off on the document or understand the document.

DR. REEDE: I agree. I think we can definitely do that. I think we did a part of this

yesterday. My only caution is that we don't extend this into a three-hour discussion of wordsmithing for each piece and not get through it. But what I'm going to ask is the same thing I did yesterday. Sarah, if you could help from there, because this is very awkward for me to try to read around this.

MS. CARR: Do you want me to read it, or do we want to take a pause and get a hard copy and maybe go into the coverage and reimbursement and come back with the hard copy in front of you? We can do that.

DR. REEDE: The hard copy would be easier.

MS. CARR: Do you want to do that, then? Okay.

DR. McCABE: Cindy, are you ready to lead the discussion on coverage and reimbursement?

MS. BERRY: I guess. It's sort of hard to figure out where we left off yesterday. I think we do need to focus, kind of really home in on our recommendations.

MS. SARATA: I have an outline developed for you to work off of.

MS. BERRY: Okay, we're going to get help. I was going to suggest putting the topics up on the screen, but I don't want to interfere with the work that's going on right now. I think it will take us a few minutes.

DR. McCABE: While we're waiting for this to come up, we had a few changes, but the primary changes were in the recommendations and trying to organize the recommendations because they were too numerous. Is that correct? So we got them down to either four or five, depending on whether we considered the broad areas, which were two under the broad areas, and whether we considered that one or two.

The other thing I would remind everyone is that we decided yesterday we would probably not finalize this at this meeting, but we would come up with a second draft of this from the meeting.

MS. BERRY: I think we're ready to go. Staff once again has come through, as they always do. They have put together an outline that attempts to reflect some of the discussion that we had yesterday, because we were struggling with the myriad of topics that we could address in recommendation form. In an attempt to organize that, staff have come up with this outline.

I actually would put something before coverage, and we did talk yesterday about defining what we're talking about when we discuss the term "genetic technologies," whether it's genetic tests, genetic services. What is it precisely we are trying to get covered and reimbursed properly? We need to clarify that a little bit better than we did initially in the first draft of the report.

To the extent that folks have any specific input on that, we should nail that down, I think, because that's really a threshold question. We're not saying cover everything and reimburse everyone no matter who is doing it, no matter what they're doing. We are trying to be focused here. So that will be an important up-front discussion in the report that will be fleshed out a little bit more than it currently is.

I don't know if anybody has any comments on that particular point.

Debra?

DR. LEONARD: But that would be part of the body of the document and not part of the recommendations.

MS. BERRY: That's right. The recommendations, though, will relate to that, because we'll be clear all along that what we're recommending goes back to our initial definition of those technologies.

Hunt?

DR. WILLARD: Again, I'm thinking of the consumer who is reading any of this. It's

the juxtaposition of the terms "genetic services" and "genetic technologies" which -- and I believe we mean different things. As defined in the document, "genetic services" applies only to those who are board certified medical geneticists, counselors, or primary care physicians. It therefore excludes any other specialists who might do lower-case g genetics or genomics work, and that's fine. We can define that any way the committee wishes.

But then the parallel term, "genetics technologies," on the other hand, takes on a much more all-encompassing flavor to it and covers everything under the sun, not what would be traditionally or classically considered "a genetic test," circa 10 years ago, but everything that might come forward for the next 10 or 20 years based on the Human Genome Project. So I'm a little concerned that those two terms invite one to treat them as parallels when, in fact, they're very, very different.

MS. BERRY: Does anyone have any other thoughts about that? Because that is really an important up-front matter that we have to tackle before we can really be precise enough to be useful in our recommendations.

Debra?

DR. LEONARD: I'm not even sure we should be using the word "technologies." Maybe "test" is a better word to use, because I see technology as methods rather than a clinical service that's being provided.

DR. WILLARD: It comes from the charter, however, which uses the term "technologies."

MS. BERRY: What about in the beginning, though? We can define our scope how we wish in the report, where we can say in this case or for purposes of this report, we are referring to just this one aspect of genetic technologies, genetic tests. We can narrow it for purposes of the report without interfering with our charter and our other goals and duties.

DR. LEONARD: I mean, as "genetic technologies" is defined, it's correct. They're technologies, but those technologies are used for tests or testing for clinical purposes, and it's really the tests, using a variety of genetic technologies, that may change over time that we're concerned about. The laboratory tests, and then the other medical services surrounding patient counseling and treatment and everything also is what I would put into services.

So maybe what we need to define are genetic technologies, genetic tests, and genetic services.

MS. BERRY: Anyone else have any thoughts?

Martha?

DR. TURNER: Yes. Just a question we were asking over here is that the written comments that we got from people that are in our folders, I wondered if those had been received in time to incorporate them or to consider the suggestions in these documents for that draft.

MS. SARATA: No, they hadn't.

MS. BERRY: There's mouthing going on.

DR. TURNER: The other question is do we need to do that in this group now, or will that happen later?

MS. GOODWIN: Most of the public comments that we received were received after the briefing books but before this meeting, so most of the public comments are in your table folders and they've been reviewed to the extent that the committee members have been able to review them during this meeting.

MS. BERRY: But we will need to consider them and, to the extent possible, incorporate them in the next iteration of this report, of the draft. But they haven't been incorporated in this particular version that people have looked at.

Well, any other suggestions on the threshold issue of how we want to define what we are proposing to cover and reimburse, or should we just have a go at it with another draft and reflect some of the comments that we've received from the public, and then send a second draft out? I don't mean to cut off debate either, because people may have additional thoughts as you go back to your offices, and you should feel free to email some of those suggestions because I do think we need to nail that down very well. I don't want to gloss over that. It's pretty critical to what we're doing.

Kay?

DR. FELIX-AARON: Just a clarifying question. Are we going to go through the different -- number 1, number 2?

MS. BERRY: I just put a 1a before the coverage that's not reflected in the outline.

DR. FELIX-AARON: Okay. Thanks.

MS. BERRY: Emily?

DR. WINN-DEEN: I just wanted to agree with Debra, that I think in the context that most people in our part of the world, in the diagnostic side of the world use it, we should refer to this as coverage of genetic tests and not of genetic technologies, because the technology is just a means to the end, but what you're developing evidence for is that the genetic test has a clinical utility in medicine, and what you want reimbursement for is when a physician orders a genetic test, that that is reimbursed. It's sort of technology independent, with the exception that the CPT codes code reimbursement by the actual steps that are performed; as they exist today, code for the specific test steps that are required to be performed.

MS. GOODWIN: Are you suggesting, then, changing the title of the report to "Coverage and Reimbursement of Genetic Tests and Services"?

DR. WINN-DEEN: So where you say "genetic technologies and services," I think it should say "genetic testing and services."

MS. GOODWIN: But you're still including the services part of it?

DR. WINN-DEEN: Yes, absolutely.

MS. BERRY: Any other comments on this?

Debra?

DR. LEONARD: But that services is to be distinguished from laboratory-developed tests, which in our previous discussion were referred to as services. That's not the services that we're talking about here but more like medical genetics or genetic counseling, treatment follow-ups, interpretation of the test results, those types of services.

MS. BERRY: We should clarify that in some sort of definition section to really nail that down.

Okay, moving to the first coverage section, we had a lot of discussion yesterday about the evidence base for -- really the lack of an evidence base hampering coverage decisions and determinations. So there was a discussion about doing a technology assessment or some sort of study as to really what is out there. What evidence do we have that supports coverage of certain tests or services?

We did not go into too many specifics. We talked about AHRQ, we talked about other entities. Some have proposed the National Academy of Sciences/Institute of Medicine as a possibility. There may be others.

Do folks on the committee have a preference or an idea for who should actually conduct this? Do we want to recommend a specific entity to the Secretary in our report for recommendations, or do we want to leave that vague? If we want to have a specific recommendation, what is the preference?

Ed?

DR. McCABE: Recognizing that I am a member of the Institute of Medicine, however I would press for this being a study commissioned by the Institute of Medicine. I think that's an independent group and it would carry a lot of weight with the Secretary, I would hope, and I think it would, by making it independent of the agencies of HHS, that hopefully it would be recognized as credible by the Secretary. So I would recommend that we recommend commissioning of a study by the IOM.

MS. BERRY: I have a question for staff. Is the second bullet a second component of the same study, or is there any reason why we couldn't have -- for example, if we decided to go the IOM route, that they could not do both functions, do sort of a review of the evidence, identify gaps in the evidence, and perhaps come up with some recommendations? Do we want to have that all in the same type of report, or should it be bifurcated?

Emily?

DR. WINN-DEEN: I think we need to be clear on this, because I think you've got two different things mixed up here. One is we need to develop a guideline for establishing clinical utility. How do we know when we're there? And then it seems to me what you're talking about here is, if NHGRI or somebody is going to put out an RFA to address gaps, it's going to be for a specific disease area, a specific test where we need more evidence. So we need the generic framework. What steps should one take to collect evidence to demonstrate clinical utility? That's step one, generic guideline. And then the next one is for each new thing that comes along, how do we assure that the right evidence is gathered?

So the NCCLS document, which I think you're referring to, the guidance document that's under preparation on establishing clinical utility, belongs as part of a framework of documents that would be there as guidances to the community on how to establish clinical utility, and then the other things are specific test-by-test evidence, gathering evidence test by test. So I think it's a little bit mixed up the way it's divided here.

MS. BERRY: I think it is, too. Maybe to help staff with this, my recollection of the discussion was that there was this overarching need for a review of existing evidence, and then also to help guide future efforts by helping, whether it's manufacturers or providers, come up with or gather the correct information so that they can make their case about coverage. So this whole evidence issue is kind of an overarching theme.

The second part that staff has outlined for us I think deals more with there are going to be gaps in our current evidence base, but that doesn't mean that certain tests should not be covered or certain services should not be covered right now. What are the criteria that should be applied, whether it's a private insurer or a federal health program, in determining whether something should be covered? That's the clinical utility, clinical validity. I think a lot of that was done by SACGT. If it was done, it predated me.

DR. WINN-DEEN: SACGT went through a whole scenario of trying to figure out how to classify tests into low risk and high risk and how to make a framework that FDA might be able to use, but I'm not sure it really addressed this kind of -- it was like which things need the most regulatory oversight framework, rather than how to develop evidence of clinical utility framework.

MS. BERRY: Muin told us about some of the efforts underway at CDC. I think the point here is that for coming up with criteria, either with evidence or in the absence of sufficient evidence, we don't need to reinvent the wheel, because there are organizations out there that are looking at this, and we need to just inform ourselves. As I remember, Muin, you had the wheel and you had some pretty good information about the work that you're already doing in that.

DR. KHOURY: I think this went pretty fast this morning. What SACGT did was to

develop a framework for the evaluation of genetic tests as they move from research to practice using the acronym ACE, analytic validity, clinical validity, clinical utility, and the ELSI issues. Yes, the intent was to promote the oversight and perhaps push the FDA in the direction of incorporating this kind of acronym in the way they evaluate tests.

But the SACGT also recommended sort of a three-pronged approach. One is an FDA process, two is a CLIA process, and a third is more of a non-regulatory public/private sort of data collection process that we have been trying to work with for a long time. What we did with ACE was to flesh this out a bit more. So we took the four kind of broad evaluation of tests and developed this into a full-blown methodology. So for each acronym, there are many questions that go under that, under the analytic validity, under the clinical validity, and overall there's probably somewhere between 40 and 50 specific questions you'd like to ask of any specific test by intended use.

That work is now kind of winding down. We applied it, with the help of the Foundation for Blood Research, to five genetic tests. BRCA1 was one of them, Factor V Leiden, hemochromatosis, cystic fibrosis, and colorectal cancer testing. What we're doing right now is sort of taking stock of that experimental effort and trying to move into the next phase, working with the other agencies, AHRQ, CMS, NIH, to try to develop this next phase of a framework that uses the evidence-based methodologies that had been developed by the ACE group, but there are so many other technology assessment groups out there, including Blue Cross/Blue Shield has their own, AHRQ has a methodology, the U.K. has technology assessment, Canada has one.

We're going to be convening a group by the end of probably this calendar year hopefully to come up with a consensus way of evaluating genetic tests, again not as an exceptionalism concept, but there are many nuances there that merit maybe a special look for genetics, and then try to move with the implementation of this EGAPP proposal that I mentioned this morning.

So I think these efforts are going to be hopefully pushing us along. I think what the Secretary needs to hear from this group is sort of a need for different kinds of activities to be implemented and coordinated by the various agencies that are under his jurisdiction, because no single agency alone will be able to move this. We're not looking at the oversight regulation concept but the concept of how we can develop an evidence base, working together with academia, the professional organizations, and then put that in play in the real world so that when a new test comes along, you can evaluate it and you can hopefully guide the integration of that test into practice.

MS. BERRY: Kay?

DR. FELIX-AARON: Thank you, Muin, but I think it doesn't address sort of the gap. I mean, I recognize the gap between what Muin describes and where we are in the sense that what I heard yesterday was the need to evaluate the state of the evidence, the current state of the evidence. I think what you described is a wonderful way of evaluating new tests coming into the process. But where we are currently, I think that what we talked about yesterday was a need to develop or to produce a state of the evidence as it relates to genetic tests and services. So that's where I saw the work of the committee was yesterday.

I see up here you've described a way to develop, that what we're proposing is to develop a process for assessing what evidence base is sufficient. I see there's some interface with what Muin is saying, but there's still a gap that currently needs to be addressed so that CMS and other payers do what they need to do in terms of covering services. So that's one point.

The other point I'd like to make in terms of which organization would be best suited to do that, whether it would be an IOM study or, say, another agency like AHRQ that does this type of work. I think there are tradeoffs and there are advantages. IOM clearly has a lot of visibility, and it would definitely raise the issue. They have the credibility. But I think, though, the downside of that or

the other challenges with that, with IOM, are issues around funding and who would fund that report, and the advantages for having an organization like AHRQ do this work is that it does have credibility within HHS, as well as outside the federal agencies. There's already a mechanism and co-funding for that kind of work.

So I think we have two good options, and I think this committee would have to decide which option meets its need today.

MS. BERRY: Does anyone have a comment on that in terms of which organization? Am I correct in assuming that we're all in agreement that we need to get to the first part, which is to do an assessment of the current state of the evidence that exists today? The question is who should do that.

DR. WINN-DEEN: No, I think that's completely dependent on each and every test. For each test, you have to say what is the evidence for that test. There's no way to do an across-the-board what is the evidence for genetic testing kind of a study. I think that would be just a complete waste. What we want is how do we know when something new that comes along is ready to be integrated into medicine, and what's the continuum, what are the steps you have to take to prove that it's ready.

MS. BERRY: I heard yesterday, though, that we need both, that you can do an assessment based on the current science that's out there, what's available and what diseases exist that could benefit by these things, but then our work isn't done. You have to then do exactly what you're talking about, which is to help provide guidance for the future as new technologies come out. But I am not the scientist here, so I defer to others.

But what I heard from yesterday was that there was some need for an assessment of what exists currently, but that may not be correct.

Debra?

DR. LEONARD: Well, I'd like to just reemphasize what Emily said, which is that we're talking about two different things. One is setting up general guidelines that a committee could use to assess whether something has moved from research to clinical utility, and then beyond, once there's clinical utility, making some recommendation that there should be coverage for that service. Those are kind of general guidelines that I think are potentially being developed by NCCLS. I don't know whether other groups are developing those guidelines.

Those are generic and would not move -- that discussion would not move any specific test to clinical utility. Then those guidelines would be applied by the top bullet group that you're talking about there on a test by test, disease by disease basis, looking at the evidence that's out there, and if the evidence is there, then saying this has clinical utility and should be covered. If there isn't, then identifying the gaps that need to be filled, and then that's the third one.

So I agree with what Emily said. This is exactly what Emily said before. There's a three-step process there, and one is identifying the general guidelines that could be used for the task force, then setting up the task force that would do test by test, disease by disease, and then identify that either there is clinical utility out there in the literature or there are gaps, and then those gaps would be put out as an RFA to through research address the gaps in the knowledge so then you could move a test to clinical utility.

MS. BERRY: Are you envisioning a task force that exists in perpetuity so that they are constantly making these assessment?

DR. LEONARD: Yes.

MS. BERRY: Because that's different from --

DR. LEONARD: Or commissioning task forces or one task force that would have a different membership depending upon what test was being addressed, because it's not necessarily professional opinion that you're looking for but just those people who are knowledgeable enough to look

at the evidence that's out there and say it's good enough or not. So it would be probably some constant people who can generally do scientific assessments, but then also bringing in other experts who could provide additional information that relates specifically to that test.

But it wouldn't be a task force that exists for six months and then goes away, because this is going to be an ongoing process for every new test, service, whatever, that comes along.

MS. BERRY: Now you're talking about a federally -- I don't want to use the word chartered, but for lack of a better term, a federally mandated task force that exists? Because how in this case, going back to our genetic exceptionalism, how does this differ, or why would genetic technologies require this kind of federal structure when other services and technologies don't? I mean, each individual insurer can have its own assessment task force, and they don't answer to a federal task force.

DR. LEONARD: I don't know that it necessarily has to be federal. In fact, there's a policy option that's recommended in the document that says that CAP or other professional organizations could provide clinical utility guidelines that would basically drive coverage decisions. So it may not have to be a federal organization. If an organization, a professional organization would step forth to do this, they may need financial support or other resources or something to be able to accomplish that.

I don't mean to monopolize this discussion.

MS. BERRY: No, this is useful, because I have a completely different recollection of what I thought I was hearing yesterday from what you're just articulating. So it's important for, please, everyone to speak up, because this gets to the heart of what we're going to be recommending.

DR. WINN-DEEN: So, Cynthia, maybe the place that would be useful to commission a state of the state kind of thing is in terms of things, what are tests that are actually covered by both Medicare in a routine way, as well as if we can get information on private insurance in a routine way. What are the ones where there's inconsistencies? You know, some do, some don't. Why is there a difference? And then there will be a whole bunch that nobody covers, primarily because there's not enough evidence yet.

But looking at the ones that have established sort of unified coverage, what criteria did they meet? How did they get to that point of having unified coverage? What is the points of disagreement for the ones that have spotty coverage in terms of a gap analysis? So why do some groups cover, some groups don't? That might inform us in terms of trying to understand what the gaps are and how to create a framework that is very clear on what all the pieces of information that one needs to have in order to get a coverage decision.

Then we also would need the third part of that, which is a commitment that once a test reaches that point and you have those points of evidence, that we don't have another endless debate about it, that it's sort of accepted that that is the criteria and there's buy-in for that.

So I think that's actually quite a lengthy thing to try and undertake and get consensus on, but it would be extremely useful. The thing that's different about genetic tests is just that there are so many of them coming along. There really aren't that many new serum markers for heart attack risk or whatever. There's one every couple of years, whereas in genetics we've just got a steady stream of things coming along, not that we're exceptional because it's genetics, but just because of the sheer volume of things that are going to be coming through the pipeline, in my opinion.

DR. LEONARD: I think it has to be some body that is accepted by all coverage entities such that you don't have an iterative process that just goes on and on and on with each. So Reed was saying yesterday that USPSTF is something that insurers buy into what they say, that it does influence greatly the coverage decisions that are made by insurers. Is that correct? I mean, did I hear that yesterday?

DR. McCABE: Yes, you did.

DR. LEONARD: But then I also heard that that process is also very slow and takes forever. So can we get an equivalent body that moves more quickly, or is the process just that slow?

DR. McCABE: I think that body, it's not only slow, it's also incredibly rigorous. If we held medical practice to those standards, we would find that we were doing very little. So I think part of the discussion yesterday was that we need to develop some process, but perhaps we have to have a process that's really workable and will bring more of these tests and services into practice.

DR. LEONARD: But without that end buy-in, and I don't know how you get that up front, but without that final buy-in, the whole process may not be that useful to invest in if it's not really going to influence many, not necessarily all but many of the different insurers that are out there.

MS. BERRY: Kay, and then Muin.

DR. FELIX-AARON: The U.S. Preventive Services Task Force approach would be a test by test, a service by service strategy, and not addressing what Emily said earlier about what is the state, because I think we came to this question as what was the problem. The problem was that those types of services aren't being covered, and that's how we defined the problem. So we said they're not being covered because there's no evidence.

So the question is how would this group want to proceed? Is this group going to proceed by looking at every service that is available now and looking at what the evidence is, or pretty much trying to address what Emily says? There are lots of services coming on board at this point in time, sort of circumscribing the most important services or the most promising tests and seeing what is the evidence for the benefit of those tests for the public, because I think it would be a higher level analysis than a test by test analysis, where you look at a specific test, but looking at the body of this information.

I mean, does it warrant departmental action at this point? I mean, is the body of evidence enough to say that there needs to be some statement about what purchasers should be covering? Because this is a new area, it's a reality, and they should be focusing on genetic testing and services.

MS. BERRY: But can you make an assessment like that of a potential technology that's coming to the fore but hasn't come yet? There's no evidence. You don't really have anything to assess at that point if it hasn't really come out yet. So we're sort of in this limbo land. There are evidentiary gaps, I think, with regard to existing technologies, but then we recognize that as new technologies come out and are developed, we need to provide some guidance. I think this is what I heard others say, we need to provide some useful guidance so that we don't have to go through this over and over again, going back into the literature. Instead, there will be guidance on the front end so that those who provides these services or tests or technologies will have that information, and that will inform coverage decisions more instantly than currently happens.

DR. FELIX-AARON: I hear what you're saying, and I think the group will have to make that decision. What I've heard is that there's a compelling argument to guide the process going forward, but I think that doesn't take away the responsibility for purchasers and other groups to assess the evidence, because clinical trials -- people will have specific questions that they're asking for a particular study, and there will still need to be somebody looking at issues of benefit, looking at effectiveness, and making those types of comparisons.

So I think the guidance is clearly important as we move forward, but I don't think it will remove the need for the work on the back end saying should we cover this or not, the decision-making processes that purchasers have to make.

MS. BERRY: Muin, did you have comments on this?

DR. KHOURY: There are lots of issues that are being discussed, and I think sometimes we mix apples and oranges and pears. I think while there may be a need for a general assessment of the status of the state, where we are with genetic testing, I think we've heard enough over

the last few years that we need a rigorous methodology to begin to look at the validity and utility of genetic tests by intended use, test by test. So not to negate what you just said, Kaytura, but to sort of move it along the test by test methodology.

The process which I described this morning, which I probably did not describe in any reasonable way with that fancy diagram in there, will take us a long way to try to begin to bridge that gap between where we are today and where we need to be in the future. The experiment we did with the ACE project, especially interacting with the U.S. Preventive Services Task Force, I'll take you through that for a minute.

We've established the ACE framework with very detailed questions, and then we funded one of the AHRQ evidence-based centers, the Oregon one, to look at the BRCA1 testing. They are working through the methodology. The U.S. Preventive Services Task Force uses a very rigorous methodology that primarily focuses on clinical utility. It doesn't deal a lot with the ethical issues, or even the analytic validity of the test. What we were told, and we have an ongoing discussion right now with the evidence-based centers and with AHRQ, is that for most of these new genetic tests, the return will be insufficient evidence from that AHRQ, very detailed, rigorous look.

So after this initial phase of trying to put all the technology assessment pieces together, because different organizations have different methods of evaluating tests by intended use, the plan is to put together a working group. We didn't want to call it a task force because we did not want to create another U.S. Preventive Services Task Force but we want to work with the existing one, to create an independent working group that's really not CDC owned or NIH owned or AHRQ owned.

But basically, they will begin to look, test by test, they will decide for themselves, guided by the horizon scan and the stakeholders, first arriving at a consensus for test methodology review, and then review test by test the whole spectrum, from analytic validity to the ELSI, make some pronouncement of what we know and what we don't know through using evidence-based centers, probably using the AHRQ evidence-based center reviews, putting those on websites to try to influence interim policy, because many of them would return insufficient knowledge, lots of gaps, and then working with NIH and others to fund the various research that needs to fill that gap.

I'm trying to follow Debra's comments. All you said here is sort of what this process will move forward to. Again, not one institution, one organization will be able to do this alone. It has to be sort of a joint public/private partnership. Forgive me for keep singing that same tune which, Ed, you probably are tired of me over the last 5 to 10 years, but I view this as an essential way of moving forward, supplementing all these various processes that already exist within HHS and the FDA and CLIA, et cetera.

So I think what you have begun to articulate there is essentially that vision that the diagram I presented this morning tries to move us in that direction, and maybe what we need to do is spend some more concentrated time, maybe the next time or the time after, to flesh this out in a way that engages all the stakeholders, because this is where the rubber meets the road. This is probably the most important thing that will drive the true translation of genetic technologies into practice. I mean, education is important, but without the evidence, there is nothing to integrate. So I do feel passionately and strongly about that, and we will continue to work with our sister agencies on this.

MS. BERRY: Muin, do you see any value in having IOM or somebody else do this first component, or is that not necessary given how far along your task force is moving?

DR. KHOURY: I think the IOM has a wonderful utility. As a matter of fact, after this meeting today, I'm going to the IOM tomorrow. The Disease Prevention and Health Promotion Board is having a meeting to talk about genetics and public health, a special sub-group to evaluate where we are in that process, and they will have another meeting in September. I know NIH at one point talked

to them about a review of the cohort studies and the concept of a cohort study. I don't think that dialogue has yielded some result.

I think if we want to approach the IOM to develop an IOM report, which is a full-blown picture, it has to be well thought out, and it has to be kind of a broad mandate, because the IOM pronouncements take time, they are not cheap to implement, but they have a lot of weight. So if you want to go to the IOM, I would encourage you to think about it, and maybe the feds can sort of talk among themselves in terms of if there is a unifying agenda along that translation pathway that would necessitate a full IOM review. I think that would be a great thing, but it will take time and very deliberate discussion before we go to the IOM.

MS. BERRY: Debra, and then Emily.

DR. LEONARD: So it sounds like you are already fairly far down the road in the planning part of this work group that would do the test by test evaluation using the ACE process, is my understanding.

DR. KHOURY: ACE-plus, which means merging the best tools of the trade, which would be ACE plus the other technologies that are used by the U.S. Preventive Services Task Force, the Cochrane Collaboration, the Canada Technology Assessment. There are lots of groups out there that do this stuff.

DR. LEONARD: So that first part, when we had turned that two-step process into a three-step process, that first step is basically done, or at least in a workable enough form that you could start doing test by test evaluations once you can reach a consensus among all the participants as to what the categories of evaluations are going to be.

So how can we assist this work group in happening and moving forward? Do you need funding? Do you need a commission? How could we move that process along or make a recommendation to the Secretary that would assist in facilitating this process?

DR. KHOURY: I think SACGHS could be a wonderful voice with the Secretary to kind of stress the fact that this process needs to happen, and it has to be a collaborative process across all the agencies that are under the HHS Secretary, and that involves the private sector as well. So promoting the concept of a public/private partnership.

At this point, you can make that assessment and then follow and be engaged in the review of how far along will this process really go forward to fill the gaps that need to be filled. So I think by being engaged, by communicating with the Secretary about the importance of this process, about the collaborative nature of it, I think that would be sufficient at this point.

MS. BERRY: Emily?

DR. WINN-DEEN: So I was going to ask if you'd be willing to give us either a written briefing by forwarding on the materials that you already have between now and our next meeting, or if you don't feel they're quite ready for that, to give us a briefing at the next meeting that's really much more in-depth, walk us through the questionnaire, what are all the lines of evidence, what are the questions, what are the things, and sort of where are you, who are the stakeholders that you're working with, what are their concerns. Maybe we should hear from them independently.

I really think we've identified that this is a critical activity that needs to happen, but we don't need to reinvent it if it's already happening. Well, we either need to bless what you're doing and throw our support behind it or make whatever suggestions that this team might have for how it might be adapted or improved, rather than trying to go through creating a whole new mechanism. I'm not in charge of the agenda for the next meeting, but if we're going to have coverage and reimbursement, I would like to put that as a potential agenda item.

DR. LEONARD: Also to explore whether or not, after you go through this process

for each test, would the insurers then buy into that as evidence to use in their coverage decisions.

DR. KHOURY: That we don't know. It's part of the experiment in the next two to three years. So I think depending on when the next meeting is -- the next meeting is in October, we'll be ready to give you something in writing and maybe have more of a discussion about that.

Now remember, this process is not necessarily to drive coverage but to summarize what is known and what is not known, and then by having the right people at the table, then further discussion could lead to this leveraging of the coverage issues. I mean, the way we started this -- I'm glad you kind of separated 1 and 2 here -- is developing the evidence base. This is sort of what that process is geared to, sort of summarize succinctly what we know and what we don't know, and where the gaps are, so that further research can be done to fill those gaps. Then in the interim, the information that is available can be used for some interim policy or guideline development. If not, then we go back to the drawing board and wait for the research to fill the gaps, and then go at it in another cycle.

So in the meantime, the problem is that people are asking for coverage in the absence of sufficient evidence, and that's something we're going to increasingly face in the world of genetics and genomics. Silence is not really an option. By being completely silent, it's basically not stepping up to the plate and saying is this a good thing or not a good thing. I mean, somebody has to step up to the plate and summarize the status of information so that both consumers and health care providers are armed with the right evidence at any given point in time.

MS. BERRY: A question for the group. We could take several different approaches. One approach would be to, in our report, in the recommendations section, we would state, of course, earlier in the body of the report the nature of the problem, and then in the recommendations section note this working group effort that's going on and talk about the need for supporting that effort and referring to it in some way. Or do we hold our report until that effort is further along, and then the report would simply endorse whatever the approach is that's taken there? I don't know if you have an opinion one way or the other on that.

Ed?

DR. McCABE: I would suggest that we not hold our report, but that we move forward, that we document whatever the state of the art is at the time that we finalize this, and I would hope we could try and finalize it at the next meeting, but we simply document where we are at that point in time.

MS. BERRY: So the recommendation could be something fairly general about the need to develop a well thought out methodology or process for evaluating the evidence and looking at all the factors that insurers or federal health programs need to look at in order to assess whether a particular technology or service is covered, and then refer to this effort as a potential model that we'll be monitoring, without coming to a firm conclusion as to what those precise criteria should be, because it sounds like that effort is under way.

Sherrie?

DR. HANS: One of the concerns that I had in the discussion yesterday and continues in the discussion today is -- and I don't know if this is the intent of the committee or not -- that you're setting up a higher standard for coverage and reimbursement for genetic tests and services than for other medical interventions and treatments. I'm not sure that that's what you want to do.

The other concern that I have is that you've taken a lot of public testimony on this already, but has the question been asked of payers what would be the most useful for them as they go about making their decisions, what is the information that they're looking for? Certainly from the VA's perspective, which is admittedly in an odd category because we're payers, providers, and a public health agency all in one, there are sort of three levels.

One is what are the things that we should tell our docs they cannot order, we will not pay for, we're not going to have folks using those technologies, and that's sort of the basic minimum of do we think this has any validity and does it have any minimal utility at all. Then there's a whole range in there where it's up to the clinical decision-making and perspective of the providers about what is appropriate for the patient that's sitting in front of them, and that's medical decision-making, medical practice that we sort of leave to our physicians.

Then there's a very high level when we use USPSTF or we say is there compelling, overwhelming evidence to say that this is something that, sort of as a public health agency with our population, that we want to ensure that we're pushing forward, that we're making sure that everybody gets this test, that all of our docs are doing this, and that's the next level. So there's the no, there's the medical decision-making, and then there's where do we really want to put our emphasis and where is the real high level of evidence that we're really going to push around.

From just the VA's perspective, helping us understand where to draw those lines through the kind of guidelines and guidance that you've been talking about would be helpful, but I say that in the context that I would hate to see this committee set up a higher standard for this technology for coverage and reimbursement than for other medical interventions.

MS. BERRY: Emily?

DR. WINN-DEEN: I just want to quote my friend, Sam Broder: "Don't let the perfect be the enemy of the good." We need to understand at what point we have enough evidence to put it into practice, to reimburse for a new test, and that's I think what Sherrie was talking about, from no to at the physician's discretion. If the physician feels it's medically necessary, they order it, and it should be reimbursed. So we need that -- for the coverage and reimbursement purpose, that's sort of the threshold that we need to define.

Now, the next level up in genetics I would say is population screening. At what point does everybody need to have this test because it's so important? For that I completely agree, there's another level of evidence that's required to get to that point, and that's maybe to some extent the kinds of tests that CDC has been focused on, ones that are at least candidates for a population screening approach.

So I think that part of what developing the evidence base is is we have to have very clear cutoffs on what pieces of evidence have to be there in the general consensus of insurance providers for a test to be covered, and that gives everybody sort of the same bar to aim for, no matter whether it's test A, B, C or D. You know what you have to develop, you know what to expect. If people want to order it before you've reached that threshold, they know that they're not likely to get coverage.

But on the other hand, once you get to that threshold, then it shouldn't be uneven. It shouldn't matter if you're employed by the government and covered by government insurance or employed in the private sector and covered by private insurance. You should have that covered. I think those are the kind of inequities that we're trying to get past and trying to identify how to deal with that. Then the part we haven't gotten to yet is how do we get the right level of reimbursement associated with that. But if you don't say a test is worth covering, it doesn't matter.

MS. BERRY: Kay, and then if someone can volunteer to wrap up the two, because I do think there are two parts here, and we need to really home in on what will be our two recommendations under this coverage section. I think we're getting there, but I'm not positive yet.

Kay, you had a comment?

DR. FELIX-AARON: I hear what Sherrie says in terms of the description, the different bars, and I agree in terms of making those types of distinctions. But what we also hear is purchasers and people who are making decisions, they're making payment decisions or making clinical decisions, asking for guidance as to what should be covered or what services should be offered. So I

think that the bar is high, but I think that it's not only for genetic services. It's for newer services as medical practice has moved more and more to a recognition, that anecdotal experience, that sort of clinical practice needs to be supplemented by rigorous evaluations of what is the best course of action, whether it be in the area of payment or whether it be in the area of what types of services should be provided to patients. We also get that response, the need for more guidance.

MS. BERRY: Anyone volunteer to summarize, then? Come on, we can do it. Just two recommendations.

All right, evidence base. Do we need a study? Do we need an assessment? The state of the state.

Debra?

DR. LEONARD: I don't think so.

MS. BERRY: No.

DR. LEONARD: I think that the first bullet should be -- I don't know whether you want to say specifically the CDC's work group that they're organizing, but develop a mechanism for assessing when the evidence base is sufficient for coverage or for establishing clinical utility or for moving it up from don't do this test to medical decision-making use of the test, and this would be done test by test.

The outcome of that, which are the sub-bullets -- you have also identified gaps, but the outcomes of that would be either that process establishes the clinical utility or it doesn't establish the clinical utility, evidence is there, and we'll identify the gaps in the evidence base and various applications, which will identify areas for research that need to be done. Then the second bullet would be that if that second step happens, that there's a mechanism by which to get an RFA out there to have that type of research done that will then take it back up through this iterative process and hopefully end up with that the clinical utility is established by the process.

Does that make sense to you guys who are writing this down?

MS. BERRY: Hunt?

DR. WILLARD: I'm confused on the potential for an RFA here. So for a particular genetic test -- I mean, this is an ongoing process that changes between Monday and Tuesday as the potential for a test is developed and different cohorts are evaluated with different odds ratios, or whatever. So I can't imagine a situation in which one would have gone through a sufficient cycle of responding to an RFA and going out there to find out that, oh, while we were waiting to do that, we got the answer a year and a half ago.

I realize there are some questions that are more complex than I just spelled out, and the utility for population screening for CF alleles is a prime example, or hemochromatosis or what have you, but to me it wouldn't be a general RFA. It would almost have to wait for a failed process where there isn't sufficient knowledge coming from the regular pipeline that we're all engaged in as new tests come out of our institutions or other institutions. It's only when there fails to be a consensus reached, perhaps because there's different populations, perhaps because there's different technologies, whatever, that one would finally get to the point of saying, gee, we need a much more concerted effort to try to see whether the answer is thumbs up or thumbs down.

MS. BERRY: Ed?

DR. McCABE: That's where I would see it. I wouldn't see it as a blanket RFA for all genetic tests. I would see it as a targeted RFA when tests with potentially high value, gaps were identified, it's not clear that they're proceeding down the road toward implementation, and hemochromatosis is an obvious example, where you need large population studies to carry that out and determine the penetrance of the various alleles and that sort of thing.

So I see this as not a blanket RFA for all genetic tests but certainly a prioritization, identification of those that could impact heavily on the public's health.

MS. BERRY: Muin?

DR. KHOURY: Actually, I was going to use the example of hemochromatosis as the poster child of this process. Hemochromatosis happened at the time -- I mean, the gene was discovered in 1996. There was a rush to pronouncement that we need population screening. We and NHGRI put together sort of a working group, an expert panel that essentially looked at the evidence. I mean, we didn't do it in this AHRQ type method but just convened the expert panel and looked at things, and then decided that there was not enough evidence, and the research gap was what is the penetrance of the hemochromatosis with respect to various health outcomes.

A feedback group went back to NHGRI and I think NHLBI on this, and they funded this gigantic cohort of 100,000 people to begin to look at the natural history of the hemochromatosis gene. Now, what we need to do here -- I mean, the reason why this is important is because we will have many such applications. As time moves forward, we may be hit with two or three similar claims every week. It hasn't happened, so this is a good time for us to plan for it, and I guess always the value added of any process would be why not leave it to the existing mechanisms.

I think this group and other groups have decided that genetics may put a pressure on the system because just the magnitude of the quantity of genetic tests that may be hitting us in the next decade or two may overwhelm the system and the ability of evidence-based groups like AHRQ, U.S. Preventive Services Task Force to cope with it.

So I think the hemochromatosis model is a good example, and you multiply that two or three or four or ten times and you begin to develop that kind of genetic process that examines the evidence using specific quantifiable methodologies, identifies the gaps for that particular test, goes back to the research and the feedback loop while more knowledge is accumulated, while at the same time communicating in a transparent way what we know and what we don't know so that the right coverage decisions are made, the public is more informed, and the health care providers are more informed so there is a feedback loop to everyone.

DR. McCABE: I just wanted to let everyone know what I asked Cindy before, and that is that we try and wrap this up by quarter of the hour so that we have an hour left to deal with what we had left about two and a half hours for in the schedule. I know that will be pushing it, but if we can try and give recommendations to staff to help with the redrafting of this.

MS. BERRY: So we're abandoning the notion of a state of the state study. Do we have consensus there? We're not doing that.

Hunt?

DR. WILLARD: I would agree with that point, but following on Muin, two comments for staff. One is, when in doubt, keep writing, in the case of a particular test or in the case of a particular association between an allele and a clinical outcome to be sure we're not talking about general, one-size-fits-all for all genetic tests but that it relates to a specific one.

Then in the report, I think the case story of hemochromatosis is a wonderful one to put actually into the report itself, because it does demonstrate if there was anything that looked like it should have been a slam dunk, that was probably it. Of course, it turns out to be very, very different from that. So it does illustrate exactly how this all may play out for 100 other tests.

DR. McCABE: It turned out to be a dunk rather than a slam.

DR. GUTTMACHER: Can I just emphasize what Hunt said at the end of that, and that is what it means for 100 other tests, because I think as we move forward we can't believe there's going to be any kind of a mechanism that can actually vet every single genetic test that comes down the

line. So part of the interest in hemochromatosis I think was using that as a paradigm for a certain kind of genetic testing. I think we need to look at those particularly. We need to think of ways of developing paradigms that we can then use, because as Hunt also brought up, any genetic test, the use of it will change over the course of a couple of months in different populations, all kinds of issues.

So there's no body that one could have, particularly in the current American medical system, that would be able to sort of vet each test, and we shouldn't ask for that. Instead, we should look at ways that we vet processes and ways of thinking about things so that they be used wisely.

MS. BERRY: Suzanne, Amanda, do you all have enough information based on the discussion about the working group and their efforts, and maybe sort of the nuggets of a possible recommendation for inclusion in our report regarding the criteria?

MS. SARATA: Just one quick question for Debra. Could you clarify, when you say develop a mechanism for assessing when the evidence base is sufficient, did we decide who was going to be responsible for doing that? Was it a federal agency, interagency --

DR. LEONARD: (Inaudible.)

MS. SARATA: But you said not to refer directly to the --

DR. LEONARD: Well, that's up to the group whether you want to do that. But we haven't heard about that in detail. We heard about it at this meeting, so it's a matter of -- I would refer to that process as something that the Secretary could enhance, facilitate, support, once we've heard about it in more detail and know that that would be a mechanism. But it sounds, at least from what we've heard, that it would be a mechanism to do what we're asking to be done. So I don't know what everybody else thinks about mentioning the CDC. If they present at the next meeting, and this report is going to be finalized at the next meeting, then we could put it in temporarily, and if we disagree with that once we've heard the CDC's presentation on the work group, take it out or change it.

MS. BERRY: Muin, do you envision the model or the methodology that you all are going through in the working group as something that once it's finalized, however long that takes, it could serve as a model for private insurers, or is this something that only some sort of federal task force, group, entity could undertake? Is it translatable into the private sector?

DR. KHOURY: Yes, potentially. I think the best way to characterize this process, as my friend Elliott Hillback from Genzyme always said, it is an iterative process. We've iterated for the last few years. We've reached a point that we are closing in on the methodologies for the review. I mean, that's step one, because whatever group you basically form has to be armed with a set of methodologies so that if you form another group, they can come up with the same conclusions because of the idiosyncracies of the system.

The second is now the experiment over the next two to three years is to test the feasibility of this approach. Alan mentioned, and other groups, that there are existing other processes, and what we need to do is test whether a process like this might work in the current set-up of our health care delivery system, given that there is Medicare, Medicaid, private sector, et cetera. So by constructing very carefully a process that brings all the partners to the table, and evaluating it, because part of the experiment is an evaluation component, within three years we'll know whether this is a model to implement and sustain, or not to implement or sustain.

We're in the beginning process of Phase II. Phase I was the development of the methodologies, and we're finishing with that. Phase II is the development of a model process to see whether it will work, and then package it in a way that fits with the existing processes that we have under the medical system right now. So within three years we'll have an answer, but within a year we'll know whether at least -- I mean, you'll be hearing more of the attributes of how that works, and this group can really weigh in in a big way to steer it one way or another as the experiment unfolds, I think.

MS. BERRY: So Amanda, what I'm hearing, then, is that I don't think we necessarily want at this point to recommend that there be some sort of federal entity or structure for evaluating all genetic tests as they come along, but rather that we are taking a good, hard look at this approach that's underway and we'll be evaluating it as it progresses, with a view towards determining if it's a model that can be used across all federal health programs and in the private sector and elsewhere.

Debra?

DR. LEONARD: So moving on to number 2 at the bottom, I don't think it's so much develop criteria for coverage, because that's kind of what you're doing in 1.

MS. BERRY: Right.

DR. LEONARD: So really what we want to do in 2 is facilitate the use of the evidence base as criteria for coverage. So it's basically using what's in 1 or facilitating the use of that by CMS, which we do. The Secretary of Health and Human Services does have influence over CMS. But then also to explore whether other insurers would use this evidence base in their coverage decisions, and I don't know a mechanism for doing that. It can directly be done with CMS by some mechanism that could be developed, but what you do for other insurers -- but we've also heard that what CMS does influences what other insurers do. So that may be a way to have an influence in and of itself.

MS. BERRY: Also in that number 2, I'd actually move the last bullet that's on the last page of the outline that you have, where it's overarching barriers, the preventive nature of the genetic tests or technology. That actually could be moved up into the coverage section because we heard a lot of discussion yesterday about one of the barriers to coverage in Medicare is the fact that there is a statutory exclusion with regard to screening tests and services. So there was discussion about legislation that's been introduced or is about to be introduced in Congress that would allow Medicare to cover certain preventive technologies.

So I think maybe, unless folks disagree, that could be a part of our coverage recommendations. We could talk about the screening exclusion, that perhaps that should be changed. That would require, of course, a legislative change. It's not something the Secretary could do unilaterally, and we of course can't lobby Congress to do it, but it could be something that we reference in the report that the Secretary could focus on and, as mentioned here, make reference in the administration's submission to Congress, budgetary submission.

Ed?

DR. McCABE: I would suggest that perhaps we could deal with that last page by that recommendation, and then we had already, I thought, whether now 2 and 3, provider education and training and health disparities, that we intended merely to make those as paragraphs to elucidate the problem in the body of it. I thought that was where the discussion was yesterday. So we had removed them from recommendations per se and made them just something we would reference as they related to coverage and reimbursement and not in the grand scheme of the education and training or health disparities.

MS. BERRY: That's my understanding.

DR. McCABE: So that leaves us with reimbursement to cover in the next three minutes.

MS. BERRY: Debra?

DR. LEONARD: Can I take one of those minutes? What is not on here is the CPT modifier system will reduce denials but there's still the issue that's not listed anywhere on here, which is the inadequacy of the level of reimbursement for the cost of doing these technologies. So there are royalty fees, but just the reimbursement level for the CPT codes that do exist is not adequate for the cost of doing the tests, in general.

MS. GOODWIN: Can I prompt you a little further and ask you what do you recommend as a mechanism for making changes?

DR. LEONARD: Like I said yesterday, the whole reimbursement for CPT codes issue is very complex, and when you raise the reimbursement for one CPT code, they kind of want to reduce the amount paid for other CPT codes. I don't know how you hit a balance, but if we are going to move toward genomic medicine, where this genetic technology-based testing is going to be used more and more and more, it's not a viable system as it currently exists because the payment is inadequate to cover the cost. That's an issue that has to be addressed if we're going to move forward.

I don't know within the current reimbursement for CPT code, establishing that reimbursement level, how you do that, because I've heard that it may end up opening up the entire laboratory fee schedule for review, and I'm not sure that pathologists -- I mean, I may be dead as soon as I walk out of this room if that is the conclusion of this discussion. So it's a very complex issue, and I don't know how you change that.

So one way, maybe when we're doing the discussion next time, is to have someone come and inform us of how you do changes in reimbursement.

DR. McCABE: That would be someone from the AMA and the group that's involved in that process. We could identify the appropriate individuals.

MS. GOODWIN: You're talking, though, about changes to the actual coding development process?

DR. LEONARD: No, the codes exist, although there's discussion of bringing online new codes. But that process happens. It's the codes, the 14 codes that we currently have where, when you put those together in combinations, the amount paid by Medicare for those services does not cover the cost of doing that test.

MS. GOODWIN: So perhaps AMA is not the right group, then, to bring here?

DR. LEONARD: Well, it's AMA that establishes the reimbursement level, but those reimbursement levels were established back in 1980-something when these codes were developed and there wasn't a good idea of how much it costs to do this testing. So that's the reimbursement level that still exists for those codes.

MS. GOODWIN: My understanding is, though, that AMA doesn't set the reimbursement level. They simply set up the framework for the codes, but the AMA is not involved in actually determining the payment rate associated with each CPT code.

DR. LEONARD: Well, Mark Synovec from CAP deals with this all the time, and I know he understands the process, but I have yet to absorb that into my brain. You're right, the AMA is the one that establishes the new codes, but then there's a complex process of collecting information about the time and the cost and the professional components and technical components of everything to establish reimbursement.

We've gone over our three minutes.

MS. BERRY: Hunt, did you have something?

DR. WILLARD: I was only going to ask the question, and I feel your pain, Debra, but to what extent is any of this specific to a genetic test as opposed to the introduction of all kinds of new tests, including putting in a variety of medical devices? I mean, there must be all kinds of groups that say we're not being reimbursed adequately for what we've put into this. So I would caution against -- unless we can identify that there are specific issues related to genetic testing, I would caution against sort of railing against the CPT reimbursement system because I don't think that's going to get us very far and we'll undercut some of our credibility, unless we can specifically target it where there are inadequacies to how they're dealing with genetic tests as opposed to four other classes of tests.

DR. LEONARD: I just know that I do genetic testing and all kinds of molecular testing in my laboratory, and I'm very often faced with the issue that we want to bill another institution, because if we bill insurers, we don't get paid, and yet the other institution says they won't send us because they're going to eat the cost. But they're perfectly happy to send it to my lab and have my institution eat the cost of doing this testing. So there is something inadequate in the system for molecular tests because they are not broadly available everywhere, like CBCs and other laboratory tests. If there's inadequate reimbursement, there is inadequate reimbursement, it's not evenly distributed across the health care system.

So if we're going to be moving more and more in that direction of doing more and more genetic tests, the system has to be fixed. It may be that there are problems in radiology and there are problems in other specialty areas. I don't know those, but I am acutely aware in my laboratory that there are discussions that go on about people not wanting to be billed for the test because they know that they aren't going to get paid, and yet they need the test because it's standard of care.

MS. BERRY: Ed?

DR. McCABE: Can I ask for some other big questions? And then we'll have to give it back to staff and the committee to work on in the interval between now and the next meeting.

MS. BERRY: Emily?

DR. WINN-DEEN: I just wanted to see if we could get a couple of words into the reimbursement system relating to health economic value of tests rather than cost reimbursement or under-cost reimbursement. I think if we're trying to frame what are the key issues, I think those are the key issues. Not only are we not recognizing the value that these tests have to the overall management of the patient, that is where in some cases they save a whole lot of money in hospital stays or other places, but we also need to recognize that many of them are under-reimbursed, and we should just frame those as issues.

MS. BERRY: Judy, I don't know if you can help us with this, or someone else who might know. If somebody wanted to influence the level of reimbursement for a particular CPT code, how do you do that? I mean, is there a recommendation that we can come up with, is there something that's missing that currently does not exist that results in this inadequate level of reimbursement and that we could recommend something to the Secretary to provide that missing piece that would help increase rates? What is the problem there? I'm not familiar enough with how the reimbursement is actually set for individual codes.

Ed?

DR. McCABE: As I've been taking notes about possibilities for the next meeting, I have down that we should educate ourselves about the process of establishing CPT code reimbursement.

DR. WILLARD: Didn't we do that?

DR. McCABE: No.

DR. WILLARD: We had somebody come talk to us about that. He was adding up the cost of a Southern blot and a PCR and everything.

DR. McCABE: Yes, but this has to do with how you influence the system, because that's the way people do it now in order to get increased reimbursement, but it's not how you influence the system to change the system.

Can I ask that if there aren't any other big issues, I want to be sure that we have time to deal with the education and training. I think this is a huge undertaking that we've begun here. I appreciate very much Cindy and her group's efforts on this behalf. But I think we're close to completing the resolution on genetic education and training of health professionals. I want to be sure we have time to look that over one more time before we go on to planning the next meeting and other issues.

Joan?

DR. REEDE: I think everyone has a copy, a hard copy of what was previously read to you. I don't want to go back through reading it again.

Have you had an opportunity to look at it?

DR. McCABE: Why don't we give everybody three minutes to read through it. It's not a terribly long document, so you can read it through probably faster than if we read it aloud for you.

Is there anyone who doesn't have this hard copy before you?

(No response.)

DR. REEDE: I'm assuming we've had our three minutes time to review. If we could take them a page at a time, are there any comments or thoughts in terms of the first page of whereas?

DR. TURNER: Number 6.

DR. McCABE: I'm sorry, we lost your mike. If you could put it on again.

DR. TURNER: I'm sorry. Number 6, to assure equitable access, I'm not sure that we're not overstating it to say that education of health care and public health care professionals is necessary to assure equitable access. It's a piece of it.

DR. REEDE: How would you like to change that? "Is a necessary component to assure"?

DR. TURNER: Yes. It's a piece of it.

DR. REEDE: Add the word "component."

DR. TURNER: Yes, I think that would work fine.

DR. REEDE: Other comments?

(No response.)

DR. REEDE: If we could move to page 2, comments or thoughts?

DR. WILLARD: Joan?

DR. REEDE: Yes?

DR. WILLARD: In the last of the bullets, oddly enough, the word "genomics" is singular and not plural. So it should be "genomics, its ethical, legal and social implications."

DR. REEDE: Thank you.

Others? Debra?

DR. LEONARD: In reading through the bullets, I think on the second page, not the bullets at the top but the little paragraphs at the bottom, "Promote and support initiatives that address the integration of genomics into education and training of all health professionals," that's really the same or a component of the next bullet. So you could say, "In order to promote, support, and facilitate the integration of genomics into health care and public health now, direct HHS." So those two things I think could be combined into one.

DR. REEDE: So what I'm hearing is that removing the third recommendation and incorporating it into number 4. "In order to promote, support, and facilitate the integration of genomics," et cetera.

DR. KHOURY: Since we are one Department, I guess we don't need HHS to direct us. What I would recommend is that you would say that for HHS to develop a plan to support the development cataloguing dissemination of blah blah blah case studies. So basically we leave it up to our Secretary to decide how he's going to work with the agencies, rather than just direct us to work in a fragmented way but develop a plan for how the Department will do this as one entity.

DR. REEDE: Fine.

DR. HANS: I'm not sure, Debra, that those two actually are the same. I mean, one is the --

DR. LEONARD: Right, because I realized that the last one goes on to talk about developing case studies.

DR. HANS: Right, cataloguing and disseminating case studies. The other is a more broad, general, integrating into.

DR. LEONARD: Right. I stand corrected.

DR. REEDE: So given that we said two different things, are we keeping them separate? Is it the committee's wish to keep them separate?

DR. LEONARD: Yes, keep them separate.

DR. REEDE: Fine.

MS. CARR: I'm sorry to go back to the bullets, but in the first one and the last one, we're only using genomics, and I'm wondering if we need to add genetics and genomics in both those places, and then it becomes "their," not "its," I guess.

DR. REEDE: I think in most of the document we've used genetics and genomics, and I think to be consistent we could use the same language here.

DR. FEETHAM: That's also true of being consistent with using health care and public health professionals wherever that belongs.

On the second page in the larger paragraph or divider, using the term "genetics/genomics revolution," I think we're beyond that language, something I've learned from Alan and others quite a while ago, that maybe it's the benefits of genetics/genomics knowledge, not having to say revolution.

DR. REEDE: Fine.

DR. GUTTMACHER: I'd like to volunteer the most picayune comment, and that is down at the bottom, the listing of organizations, it's actually the Coalition for Health Professionals rather than of Health Professional Education in Genetics. Just to show you I was paying attention.

DR. REEDE: I think the other part is there are some others that responded either in oral or written testimony that are not here, so we'll be correcting that and listing them alphabetically and all of those types of things.

If there are no other comments on page 2, could we turn to page 3?

DR. WILLARD: The two that begin at the top, provide and promote, those two seem very, very similar to me, and there must be a way to combine those and save 45 percent of the words, because they seem very repetitive to me. One is training for the implementation of models, and then the next one is promote communication to enhance dissemination of those models. There's not a lot of difference there to me.

DR. REEDE: Do you want to suggest language?

DR. WILLARD: I have infinite respect for the staff to be able to merge those two somehow.

DR. LEONARD: Or you could simply say "for faculty training and the implementation and dissemination of clinical application-based genomic," because basically you just want to get them out there, which is the point of the second one.

DR. REEDE: And I think also the first one, a major part of that was the faculty training, because that was a recurrent theme. So we'll leave it to staff to wordsmith that.

DR. WILLARD: Also, the one that refers to pipeline programs, first of all, it isn't clear to me at least. Maybe the Secretary knows what that is. But second of all, that didn't seem too different than the third paragraph on the previous page, "promote and support initiatives that address the integration of genomics into the education and training of all health professionals." Isn't that the sense of this one, to support training programs?

DR. REEDE: I think pipeline programs does not refer necessarily to health professional training but actually to the programs that may be K-12 or college or other programs that would bring people into the health professions and provide this background. So the pipeline programs refers to a different level in the academic pipeline.

DR. WILLARD: That's very helpful, but then I would suggest we spell it out and not call them pipeline programs, or put parentheses, K-12 programs or something like that.

DR. McCABE: Why don't we call it inter-educational K through 12 and undergraduate pipeline programs?

DR. REEDE: Fine.

Debra?

DR. LEONARD: So I hate to add something, but it was brought to our attention several times that many of the competency recommendations are that generalists be able to identify when they have a genetics issue or one that would need specific genetic counseling, and that the genetic counselors are gearing up to deal with more complex disease traits, et cetera, and that there are a paucity of genetic counselors that will likely be insufficient for the growing need. Do we want to make a recommendation, as we've been requested to, about specific genetics training programs in addition to the general programs, which is what all these different recommendations really deal with?

I know Muin has some concern as to whether or not we will need genetic counselors, but I cannot believe that interpretation of the genetics or genomics of complex disease traits is going to be any simpler, really, than the genetics of single-gene diseases, and I think we still will need specialists, and we may need expanded specialists who can deal with these, especially the whole time aspect of doing genetic counseling with family members that most general practitioners don't have the time to do or the expertise.

So that's something that has been brought to this committee by different groups, and I don't think we've addressed it in this resolution.

DR. REEDE: I think part of our deliberations, this didn't come up as something to include yesterday, but we also determined not to include anything for specific disciplines. So one of the things that came back was that we need more nurses in order to be able to deal with these issues, and we thought that if we started to deal with specific disciplines as in nursing, as in genetic counselors and others, that they may take the form of another kind of study or recommendation, as opposed to this, which is really looking across the full spectrum of the health professionals. So most of this and most of the comments that we got back really pointed to commonalities across the full spectrum of health professionals that needed to be addressed, and so that's what we tried to incorporate here, with an understanding that we may need to come back and look at specific disciplines in terms of issues.

MS. ZELLMER: The only comment that I was going to make, Debra, is my experience has been that I know probably 100 families with rare genetic disorders, and I would say a very small percentage of them have actually got genetic counseling. I think until we resolve the issue of families getting better information from their more general practitioners and getting referred to genetic counselors, I think we need to resolve that issue before we tackle the need for more genetic specialists.

DR. LEONARD: It's just that in anticipating the response, you train the generalist, and the competencies are not listed that they have to know how to do the interpretation of the tests. The competencies generally say you recognize when you need a specialist and you refer. So we're going to train all the generalists to refer, but there's not going to be anybody to refer them to. Or is there? Hunt, you may have a better sense of the numbers, but I'm hearing from the National Society of Genetic Counselors and the American Board of Genetic Counseling.

DR. WILLARD: Part of what you said I don't disagree with. What I think is still a

contentious issue is whether the only people who "have sufficient knowledge of genetic testing" will be those who are genetic counselors or certified medical geneticists. I do not think you get buy-in to that conclusion perhaps even around this table, much less if you went very far outside this room.

DR. LEONARD: Well, true. I'm neither of those, and I do genetics. So there are other organizations and groups that do this. So maybe it's not genetic counselors or medical geneticists, but we have not at all addressed -- I mean, this is all general, and if that's what this document is supposed to be, that's fine. But I would like to remind the committee that I don't know if there will be enough specialists as we move forward.

DR. WINN-DEEN: So perhaps what we could do is add one bullet point that just addresses the need to, without saying specifically how large this number needs to be, but in support of this expansion of genetics and genomic medicine, that we also need to train an appropriate number of referral specialists.

DR. REEDE: Sarah?

MS. CARR: Well, I just wanted to ask Suzanne Feetham if you could recall for us what the scope of the workforce analysis is that HRSA is doing now and that we heard about in October, and whether or not it might be prudent to wait for the findings of that analysis before making recommendations about specific specialties. Will that analysis make a determination about whether genetic counselors are going to be in under supply, or are now?

DR. FEETHAM: There were two studies. A study that was completed a couple of years ago was on genetic counselors, and in that study it did acknowledge that there was a small number, 1,800 at that point in time, and that that was insufficient to meet the needs. But there was also attention, as I recall, in the recommendations that where this was going into primary care, that quadrupling the number was still not the issue. It was what we've been talking about.

The current study is looking at genetics in primary care and with a specialist, and I don't expect that it will be more of a description of a practice than giving numbers. I don't expect numbers to be coming out of that, but I would expect that, again, what we've been hearing is that we will need and always need genetic specialists, and we have a high need for this knowledge in primary care. It will give us more description of the practice by both genetic specialists and primary care in this current environment.

DR. REEDE: Ed?

DR. McCABE: I was going to comment that --

DR. FEETHAM: Did you have another --

MS. CARR: My recollection of the first study was that it didn't draw any conclusions about the adequacy of the supply, and looking to the second study to provide more specific recommendations in that area. But if it's not going to do that, then it's important for the committee to know that, I think.

DR. FEETHAM: Well, I don't think it's the type of study that's going to come out and give you a ratio of specialists and primary care. I mean, it's more a description of the practice base and hopefully giving a baseline for the future.

DR. McCABE: I think we're a long way from Debra's concern that we're going to have so many generalists educated in genetics that we are going to overwhelm the specialists in genetics. So I would either leave it general, which is the tone that we have it here. If we were to insert something, I would say something general like evaluate the adequacy of the specialized genetics workforce. But I'm not sure we need another workforce study. We've had those, it's inadequate, but it's a different issue than inserting genetics education across the board, which is really what I think this is about.

DR. HOOK: And along that line, it's not an either/or. It's not generalist versus

medical geneticist. I mean, a lot of the counseling that's going to take place is coming from other subspecialists -- neurologists, hematologists, gastroenterologists -- who you indicate at earlier points should have as part of their training a very thorough understanding of the genetic anomalies within their organ system subgroup, and that's also a referral base that doesn't have to go to a specific medical geneticist but that you have covered in the principles that you've previously articulated.

DR. REEDE: Alan?

DR. GUTTMACHER: No matter where one comes down on the question of specialists versus generalists, I think in the end it actually is more pertinent to the resolution on reimbursement. We've treated that reimbursement largely by genetic testing. I think the real driver of the number of genetics professionals is not the training programs. People are in training programs for a couple of years. You're then out practicing for a lifetime. It's who gets reimbursed to what degree for doing what that will actually drive the need for genetic counselors and medical geneticists, primary care people doing genetics, et cetera. That's much more the driver, I think.

I think that no matter where we come out on this, if we're really going to influence that, I think it's probably more important to influence on the reimbursement end rather than from the educational funding end.

DR. REEDE: Any other comments or suggestions with regard to the recommendation?

(No response.)

DR. McCABE: So could I take it, then, that we will leave the specialized genetics training out of this, that we'll focus more on improving genetics education and training across the board, and that that will be another issue for a later day? Okay.

Thank you very much, Joan.

At the last meeting we talked about large population studies and pharmacogenomics. These were identified as needing in-depth study. I want to now talk about a meeting. There was a March meeting. At our March meeting, Francis Collins invited us to appoint a liaison to the NHGRI working group organized to consider large population studies. Chris Hook was appointed as our liaison to the NHGRI working group, and I'd like to thank Chris for taking on this responsibility and ask Chris to update our committee with respect to the working group's activities, and then have Alan Guttmacher following Chris update us on efforts by NIH on this issue.

Chris?

DR. HOOK: Thank you, Ed. I'll be brief given the number of things you still have to cover.

The working group has had two face to face meetings and one phone conference to date, and I want to thank my colleagues in the AGES group for setting up a phone line for me because it's been very difficult to travel with some home front issues. But the discussions have been lively. It has been an education in population genetics and the incredible logistical concerns that are covered in this. Obviously, it's still a work very much in progress.

Covering the broad front of issues, such as the ability to utilize existing population cohort studies in a new collaborative or consolidated sort of a fashion versus starting a new project on its own with all of the issues of trying to secure funding and so on for a project of that nature. There are very complex issues in terms of understanding the power, understanding recruitment, single versus rolling informed consent, dealing with a population base which essentially could be followed for 50 to 100 years over the lifetimes of individuals, from infancy to their demise, and how do you account for the changes in the technology that will take place during that period of time, and a variety of other significant issues.

There is at least one more face to face meeting planned in August, I believe it is, and some more phone conversations before a report will be rendered by the group.

A couple of points in regard to the handouts that you've received. There is Dr. Collins' article in Nature from May 27, which lays out at least some of the issues that the group is considering in trying to develop this large cohort study, and a modification to the request for information for public comment has been extended to June 30th. So they are still receiving statements from individuals concerning that. They have already received a significant volume to date, but obviously with the number of questions that are on the table, there is still interest in getting further input.

I think at this point I'll turn it over to Alan for further comment.

DR. GUTTMACHER: Thanks, Chris, for your involvement in this, which we have really appreciated.

I'm not sure there's a whole lot to add to Chris' very good summary. The information he mentioned to you is in I think the table folders that were provided this morning. Again, I would just, as Chris did, call your attention to the request for information, which is open for another couple of weeks. It has 14 specific points. It requests folks in the scientific community and the public who have any expertise or points they'd like to make about these 14 questions specifically or other questions they think would be of use. We invite comments. There have been scores of comments received already, and they've actually been very helpful to the working group to see the breadth of this.

A lot of this does get to the question of how one works with existing cohorts because there's certainly a role for those, but also to think what are the ways that a new cohort might add to our understanding of all this.

I don't think there's really a whole lot else to add. We hope that by the end of the summer/early fall, this working group will have achieved its purpose of trying to really figure out the science and to a small degree the logistics of what such a study might look like, and then it will be up to higher-ups at the NIH and other agencies of DHHS and the administration to figure out whether it makes sense to go forward with such a study or not.

DR. McCABE: Any questions for Chris and Alan about this process and the large cohort study?

(No response.)

DR. McCABE: Please take a look at those 14 points, and if you wish to have input to them, please respond to them.

Okay, thank you.

The next thing I'd like to talk about is the agenda for the next meeting, and then we'll talk about other issues beyond that. Yesterday we talked about including on the agenda at the next meeting two processes or two groups of individuals, and I have them down as genetic discrimination, bringing real people with real problems related to genetic discrimination. That was the terminology that was used, and we specified some specific types of genetic discrimination, and we talked about speaking with the Genetic Alliance and other groups to help us identify those individuals; also talking to Paul about if he could help us identify individuals that have come through the EEOC.

The other group that we had talked about like that were coverage and reimbursement, again identifying individuals who -- I have it down as the impact on the health care for individuals refused coverage and/or not reimbursed for genetic testing, genetic services. So this would also require some identification of those individuals, but we had talked about both of those groups yesterday.

Then the options that have come up in our deliberations today are an update -- again, these are options, and we can't fit them all into the agenda, so we'll have to make some selections -- an update on the CDC working group effort for ACE+, the process of establishing CPT code reimbursement,

both of which will relate to the draft document that we're hoping to finalize at the next meeting; update of the National Academy of Sciences group on patent and licensure, and then large population studies, if that had been finalized.

So what is everybody's wishes? I think if we do the genetic discrimination coverage and reimbursement, we bring in people who have been impacted by those issues, that's probably pretty close to two half days right there. So we're left with another half day, or we can fill in parts of those half days. They may not be full half days on those, but they're going to be significant investments in time for those.

The first two topics under the options relate to the coverage and reimbursement, CDC working group effort, ACE+, process of establishing CPT code reimbursement. One could argue that the patent and licensure through the National Academy of Sciences is relevant to that. Large population studies will be completed by mid-October? If it's up in the air, my guess is it probably won't. So maybe we could postpone that to the following meeting and just put a placeholder in the following meeting for the large population studies.

So now we're down to three.

MS. CARR: I think we also have to build in time for a review of the revised version of the coverage and reimbursement report. Is it the committee's hope that we will revise the draft and go out for public comments with the revised draft, or do we feel we need to sort of work on it some more and then look at it again in October and then go out for public comment?

MS. BERRY: The latter.

MS. CARR: The latter. Okay.

DR. McCABE: Any disagreement on that? The head of the task force said the latter.

MS. BERRY: The task force of one.

MS. CARR: And maybe that should be broadened out.

MS. BERRY: I think that would be a good idea, because then we can get everyone's input, and that might make our review more efficient so that we could have a broader group, everybody look at the revised version once staff has had a chance to put that together. So I don't know who would want to. Are you going to force people to join?

DR. McCABE: Do we have volunteers? If not, we will force people to join.

Put Debra's name down.

DR. LEONARD: I want to ask a question. Since the committee has gone over this document, is it something that the whole document, once its revised, could go back out to the entire committee and ask for comments back? So that the discussion time -- I mean, basically doing the discussion by email, electronic communications. I don't know if that works for everyone or not.

DR. McCABE: I think Cindy was hoping for some additional help in reviewing the next draft to make some changes there before it went out to the committee. I think given the amount of work that staff has, this is unlikely to happen within the next two to four weeks, in which case it's going to be coming back fairly close to the time of the next meeting.

Do I have any volunteers in addition to Debra who wish to?

DR. LEONARD: I've already been put on one task force, and because I might be --

DR. McCABE: I'm just teasing you, Debra.

Anyone?

DR. WINN-DEEN: I'm willing to volunteer, and specifically I think we need a few people who can help if staff, as they try and translate all our discussion here into things, has some questions they need to just bounce back and forth, we need a little referral group to just help them with that.

DR. LEONARD: Well, I did already volunteer to help with one part of it, which I will do.

DR. McCABE: Okay.

Anyone among the ex officios to help with that?

MS. CARR: Maybe Muin, since we're going to try to incorporate your -- and I think we should volunteer Reed Tuckson in his absence.

(Laughter.)

DR. McCABE: In keeping with the tradition of all committees. Reed had a lot of input, too. So it would be good to make sure he's satisfied with how his edits are incorporated. That's how we'll justify doing this to him.

So that gives us three topics that are clear, genetic discrimination, coverage and reimbursement with individuals, as well as the review of the revised document. CDC working group effort, ACE+ -- how long will it take you to do that, Muin?

DR. KHOURY: It depends when the committee wants to hear. October will be at the beginning of this next phase. If you want an abbreviated version, maybe we can take a half hour or less. If you want something more of a discussion, I think it will be an hour-plus. But I wouldn't think you'd need more than half an hour.

DR. McCABE: Okay. If you can do it in a half hour, that would be great. So we'll allot 30 minutes to that.

The process of establishing CPT code reimbursement. Are people interested in that for the next meeting for our education?

DR. REEDE: I think if we're going to be talking about reimbursement, that it would be nice to understand that better.

DR. McCABE: Okay. So we'll try and identify someone who can speak to that process.

DR. LEONARD: I would suggest Mark Synovec because he does that, and that's practically his professional life's work.

DR. McCABE: Can you provide his contact information to Sarah and her staff, please?

DR. WILLARD: Can we clarify? He's a pathologist who is trying to fight that system, or is he on the inside of that system trying to help?

DR. LEONARD: He is a pathologist, but he works with the AMA CPT Editorial Board, he works with the reimbursement side. He has gone through this process many times and understands how it works. At least we could contact him. If he's not the one to speak, he could say who in different organizations, government agencies or whatever, are the appropriate people to inform us.

DR. McCABE: And likewise, I know there's somebody -- that would be from the pathology side, and then the American College of Medical Genetics has someone from the medical genetics side, and I'm not sure who that is.

DR. WILLARD: The reason I raised the point now, and I would bring it up also in the context of ABMG, is that to me it actually would be useful to have someone on the other side of the desk who can explain why it isn't just as simple as saying this is what it actually costs, and gee, I'd like to be reimbursed for 100 percent of my expenses, because I suspect people on the other side of the desk will just shake their head and go where are you coming from? This doesn't happen. So it has to be someone on the other side who sits there and does the thumbs up and thumbs down on these things.

DR. LEONARD: And does it across the board, not just laboratory tests but genetic services, genetic counseling, all of the aspects that are related to genetics.

DR. McCABE: Okay. Then we have two topics. One is in preparation for February. As people are leaving, we can volunteer them now for these other work groups.

(Laughter.)

DR. McCABE: If we're going to talk about pharmacogenomics, then we need to put together a group to address the topic of pharmacogenomics. Anyone who wishes to volunteer for this? So Emily with that group. Anyone else? Anyone who is left who would like to?

DR. WILLARD: It's not clear to me what you're asking. You're not looking for speakers.

DR. McCABE: This is to plan for --

DR. WILLARD: Plan that session.

DR. McCABE: This is to plan a session on pharmacogenomics.

DR. WILLARD: I'm happy to do that.

DR. McCABE: So we're talking about Emily, Hunt, Debra, Chris. That's the group. And Suzanne. Anybody else among the ex officios who I may have missed? Kay.

So we're left with the one topic we haven't decided.

Oh, Sarah is telling me we need a group for large population studies. Who would like to do that? Chris, I think you're stuck with that since you are our liaison. Any interest in that?

DR. WILLARD: Interest, yes, but do I want to be on both of them?

DR. McCABE: Can we call upon you, Alan, to do that, and Muin?

DR. LEONARD: For pharmacogenomics, should there be someone from FDA on that work group? That would be useful to inform us.

DR. WINN-DEEN: There could be. I just didn't want to volunteer Steve.

DR. LEONARD: You can always volunteer Steve. He's not here.

(Laughter.)

DR. WINN-DEEN: There's a lot of FDA guidance documents that are going to be coming out with updates. Maybe you want to say something about what the timing of those are so that we can plan an update to this committee as well.

DR. LEONARD: I think just putting somebody from the FDA on this work group to help plan the pharmacogenomics sessions for February would do that. I don't think we have to discuss it now necessarily.

DR. WINN-DEEN: There's more than just the pharmacogenetics guidance document that's going to come out. Microarray and companion diagnostics are also on the list, right?

DR. MANSFIELD: Did you want to know that now?

DR. WINN-DEEN: Would you be prepared to do something in October, or should we just plan for February?

DR. McCABE: I don't think we have room in October, unfortunately.

DR. MANSFIELD: The voluntary genomics submission is supposed to be finalized this year. It's sort of creeping. I don't know that anything else will be finalized this year. The combination therapeutic diagnostic workshop is going to be in July, so maybe a draft will be out this year.

DR. McCABE: Okay, good. So you just volunteered Steve, I take it. Okay. Let the minutes reflect that.

Then Sarah and I just conferred and we think we could do something very brief on the National Academy of Sciences' process on patent and licensure. We could fit that in in October also. Anybody who doesn't want that in there?

(No response.)

DR. McCABE: I think it would help with the coverage and reimbursement, actually, because it is part of that story. So something very brief, probably another 20 to 30-minute process.

Are there other topics that people would like to bring to the floor? Other topics that we should be thinking about with even longer-range planning?

(No response.)

DR. McCABE: If not, I think that wraps us up. Everyone travel safely who is traveling, either short or long distances from here, and we'll see you in October. Thank you.

(Whereupon, at 2:39 p.m., the meeting was adjourned.)