DEPARTMENT OF HEALTH AND HUMAN SERVICES

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C O N T E N T S

P R O C E E D I N G S

[8:33 a.m.]

DR. TEUTSCH: Good morning, everyone. Welcome to the second day of the Secretary's Advisory Committee meeting. We have a special event this morning. Rick Campanelli, who is the Secretary's counselor for science and public health and has been extraordinarily supportive of our Committee since taking over that post, is here this morning and wants to say a few words to Reed.

I'm not going to make it a lengthy introduction since we have had the pleasure of having Rick here on several occasions. So let me turn the podium over to Rick. Thanks.

Presentation of Certificate of Appreciation to Dr. Tuckson

Richard Campanelli, J.D.

DR. CAMPANELLI: Good morning. I first want to start by saying thank you, Steve. Thank you for serving as chairman now, and welcome. We are grateful for your service on this and your willingness to serve. With all your background in the government and in the private sector, you bring a wealth of experience and you are

slipping into big shoes. But we know and have great confidence that you are going to do a great job, and we are very grateful to you.

We are grateful to all of you. The Secretary is grateful to all of you for your willingness to serve and all that you have done.

I'm grateful that I'm here and that I'm dressed. We had no power this morning in my house.

[Laughter.]

DR. CAMPANELLI: The ethics folks spoke up there. I just want to confess to you that I [couldn't] see. It was pitch dark when I left home, and pitch dark at home. I didn't see until I was on the subway that I was wearing a different suit than I thought I was, but the tie basically goes with the suit.

I am very pleased to be here this morning to have the chance to speak with all of you and, on behalf of the Secretary, to recognize Reed's important work.

I don't need to tell you how rapidly things are changing in this world of genetics and molecular health. New genetic associations are being announced every week. New products are being offered not only to medical

professionals but also to the general public.

About a month ago I saw a product that offered a dating service relying on genetic matching. Greg Downing just recently got married, and I'm pretty sure he did not use that service.

[Laughter.]

DR. CAMPANELLI: I'm not sure, if you are genetically matched, if that is a good or a bad thing. I'm not sure which one that is.

But things are moving so quickly, and your Committee is really at the crux of the interface of both the pioneering work that is going on there and the need to really be wise and prudent about how we go forward. Unlike the pioneers of the days of the wagon trains, these wagon trains are moving very swiftly, so we so much appreciate your work.

Particularly, the Secretary is grateful for and understands and recognizes the work of this Committee. Since coming here he has looked to the promise of better health and better health care through genomic advances and genomic and molecular medicine, and also the challenges that we face.

As we look at these things, we are looking, and you are looking, in many of the things that Reed has helped to guide us through and guide all of you through, at what is the interface of the role of government and what is the right way for us all to look at both what is happening in the private sector and how can we all work together well so we can effectively advance the ball and do it prudently in this developing area.

Along that way you all have been so effective, but particularly you have been effective because, Reed, you have been so effective in your position. I know you are probably getting a little tired of hearing all these good things. Maybe not, maybe not.

[Laughter.]

DR. CAMPANELLI: But it is a joy to have the privilege of saying to you again this morning, as I know you have heard from some of your colleagues, and you will continue to hear, the gratitude that we all feel to you for your leadership.

Leadership is a hard thing to define. There are thousands, maybe tens of thousands, of books on leadership out there. I always think the thing that

people look at when they look at these books, or when I look at when I look at them, is I just look at who is the author. Who is writing the story, and do they have a story where their personal experience is one that I have seen and they are known to be effective as a leader.

Reed, if you write that book on leadership, I know that everybody in here and many of us in the Department are going to read that book. You convey a few things to us that are effective as leadership that are particularly unique.

You have the big picture, but you keep your feet on the ground. You have the background to understand the challenges in health care today, but you have had your feet firmly planted in a whole bunch of different grounds.

I refer to my own career sometimes as a checkered past, but you have a distinguished and checkered past. You have been the commissioner of public health here in the District. You have been a university president involved in the health and science area. You have been someone who is responsible for professional standards in the nation's largest physicians

organization. You know reimbursement and quality issues from the standpoint of a payer.

On top of all that, you are someone who understands the implications of this new and challenging area of genomic and molecular medicine.

You are also a great leader because you understand about when to put your foot on the accelerator and when to tap the brakes, lightly or otherwise. In a group like this, the size of this group, with the complexity of the group and its subgroups, all of us recognize the need to be able to hit the accelerator when it is time to do it. You all have done a great job of doing that, and Reed, you have led in that way. Also, tapping the brakes when it is right time to make sure we have the information we need so we can make the recommendations that are so important.

All of that is so important to the Secretary, whom you are advising, to the Department, and to the public, because certainly the message of this Committee, importantly, is heard by the public, and is going to be more so as the public is really clamoring for leadership in this arena.

Under your leadership you have developed important recommendations and background pieces on coverage and reimbursement of genetic tests, genetic discrimination directed to consumer marketing of genetic tests, genetics education for health professionals, and more, including of course your new report on pharmacogenomics. We are looking forward to the report on genetic oversight that is now in the works. Although you won't still be here for it, your stamp is certainly on it.

I was talking to a few people before you came in this morning about the fact that this is the last meeting you will be -- probably not the last meeting you will be at but the last meeting where you have been serving as chair. In Washington it is true that not everyone is the nicest person you will meet, but when you meet a person who is personally kind and who brings good faith and good humor to a task like you have, it is rare to see that combination of characteristics with somebody as a leader.

We are all blessed by that, and we wish you godspeed in all your next endeavors. I would just like

to conclude by reading this letter from the Secretary. When we can work it out in your schedule, we want to make sure that you and he get together. We are working to set that up.

"Dear Dr. Tuckson, thank you for serving as chair of the Secretary's Advisory Committee on Genetics, Health, and Society. The success of the Department of Health and Human Services has been due in large measure to the willingness of people like you to participate on these advisory committees, and on this one in particular. I appreciate the many hours you have spent in preparation for meetings and other activities, sacrificing your private interests to advise on the planning and operation of our programs. In recognition of your contributions to the Secretary's Advisory Committee on Genetics, Health, and Society, I am pleased to forward the enclosed certificate to you."

Thank you very much, Reed.

[Applause.]

DR. TUCKSON: I will be very brief. We have an awful lot of work to do.

Let me just say that this is extraordinary. The thing that I know more than anything I know is that as chairperson my role is really teeny. The main thing that you can do if you are going to be in any way successful at this is just let the smart people do their thing and get out of the way. I think to any extent that you like anything that I did, it was only because I got the heck out of the way and let you do what you are supposed to do. That is really what it is all about.

One other quick word I would say is that I really enjoyed working on behalf of Secretary Leavitt and, before him, Secretary Thompson. But, I really did enjoy my relationship with Secretary Leavitt, and I will say there is something very important in what he says.

Public citizenship during these times is extraordinary. To be able to get citizens to be willing to put in the amount of effort that you have put in in service to the country is something that I think is overlooked. I don't think there are a lot of people who understand or value not only how much work but the

results and the product of that work.

When we started off our meeting yesterday, it took a long time to read all of the things that you have achieved as a Committee. That was a wonderful legacy that you have achieved, and I know that you will have an even better one in the days to come. We are very, very, as Americans, proud of your service to the country, and I hope that you feel good about what you have achieved.

The last thing is, you know that we don't do anything without the staff, and we never can thank the staff enough, each and every one of them. But Sarah Carr is absolutely the best at what she does of anyone I have ever met in my life. She is just first-rate, and the team that supports her is first-rate, all the way through to every logistical detail. Abbe and all the people that do what you do, you are fantastic as well.

With that, Steve, you are the absolutely right choice to get us where we need to go, and I need to get the heck out of the way and let the meeting go on.

[Applause.]

Opening Remarks

Steven M. Teutsch, M.D., M.P.H.

DR. TEUTSCH: It is a great privilege for me to follow Reed as chair of this Committee. I can't echo enough Rick's words about what it is like to follow Reed. Under his leadership this Committee has made great strides in addressing many of the important issues that we face in the field of genetics.

It is a daunting task to follow someone with Reed's vision, his leadership, his generosity of spirit, and extraordinary clarity of focus, and still a man who has a great sense of humor.

I have a terrible memory for names, so that may be one place where, Reed, I can emulate you.

[Laughter.]

DR. TEUTSCH: I first heard you speak, and frankly, I don't remember whether it was 15 or 25 years ago, when you gave a great speech at a Healthy People introduction to the country. I said, I need to follow that man. I guess now I get my chance.

[Laughter.]

DR. TEUTSCH: It has been a singular treat for

me to work with you here on SACGHS. I want you to know that you actually are not being liberated. Sarah gave me your phone number, and I will be calling.

We all know that the field of genomics is at a transformational crossroads. The Human Genome Project has opened vast new vistas. We have new technologies that have yielded important advances in basic science and have provided us great new tools for breakthroughs and innovations, for risk assessment, diagnosis, new therapies, and prevention.

When I was in medical school, which was the last time I studied genetics, frankly, genetics was a basic science in the study of rare conditions. At that time, that was a pretty specialized subject. We are clearly at a juncture where it is no longer just about tragic, uncommon diseases but about common, chronic conditions: diabetes, heart disease, cancer, arthritis. It is about complex genetics and interactions with the environment.

I think our task in large measure is to help create an environment that stimulates research and innovation, helps us understand the value of new

technologies, that facilitates the appropriate use of those technologies, and helps the healthcare system function effectively and efficiently. And perhaps most importantly, to improve the health of the American people.

At the same time, we must assure access to those technologies and fairness in their distribution. We need to make sure that there are appropriate protections for individuals and their families and that an educated population can understand the real opportunities and not be daunted by the high and unrealistic hopes.

I am reminded of what T.S. Eliot wrote probably close to a century ago. Where is the wisdom that we have lost to knowledge, and where is the knowledge that we have lost to information? We will have lots of information and lots of data, and our task is to help people make good choices.

We are facing brave new paradigms and challenges. The $1,000 genome will put incredible volumes of information in the hands of clinicians and patients. How can we make sure that that information is

tapped and targeted and used well. How do we avoid harm from information overload or misinformation. How do we build effective information and clinical support systems. Most importantly, how do we protect the public from harms.

Vaccines aside, virtually no healthcare technology really saves money. Indeed new technologies are one of the major drivers of increasing cost in our healthcare system. As the cost of health care rises inexorably and we face budget constraints, how do we make intelligent choices about the opportunities before us.

I also recognize, of course, that the field of genetics has many, many stakeholders, more than I realized when I joined this Committee two years ago. We need to listen carefully to all of them. They have important things to tell us.

Our real purpose, though, is not in meeting the needs of individual stakeholders but in improving the health of Americans. I'm constantly reminded that remembering that simple fact keeps me grounded, focused, and motivated.

While we are ultimately accountable to the

American people, our primary audience is the Secretary of Health and Human Services. Our job is to provide him with wise guidance and counsel so that he can manage and lead the HHS agencies, seek appropriate resources, and recommend legislation.

As most of you know, my field is public health and health policy, not really genetics. This Committee is blessed with an incredibly talented group of people with deep knowledge and experience in many aspects of genetic health and health care, healthcare policy, and personal experience with genetic conditions. It is that richness which gives me tremendous optimism that we can build on the legacy of SACGHS to make even greater contributions.

Our liaisons bring not only their personal knowledge and experience but a direct line into the workings of their organizations, the opportunities and the challenge that each of those organizations face. We need to be responsive to their needs as well as those of the Secretary. I look forward to working with each of you and engaging you fully into the work of the Committee.

Finally, as you all know, we have an extraordinarily talented and devoted staff, who I trust will not only keep me on the straight and narrow but will continue to maintain the Committee's productivity.

We have lots of work before us. We will spend the end of the day brainstorming about needs and priorities. In the end, though, all is for naught if our reports aren't used. Our job is not done when our reports are complete. We all know important work that we have been involved with that only serves to keep bookshelves well anchored. We will need to follow through on our recommendations to assure that they are implemented, to continue to work with the Secretary and the agencies to make sure that the recommendations are translated into reality.

With that, let's turn to the real major work of the day and to complete the work on the Oversight Committee. I think we made good progress yesterday, and we will continue to do that. As we did yesterday, though, we will do that informed by the public input, and I believe we still have several individuals whom I would like to call up to do that.

The first one is Pam Dixon. Pam, are you here, from the World Privacy Forum?

I'm going to ask each of you, as I did yesterday, to please hold your comments to five minutes, since we have a very large agenda.

Is Kimberly Layton here? If you don't mind just coming up so that we can have a quick transition from one of you to the other.

DR. BILLINGS: Steve, I wanted to make one comment about the public comments from yesterday. There was some discussion after the 23 and Me presentation about their laboratory oversight and their views about laboratory oversight.

I just wanted to make it clear that from my knowledge the tests that are run by 23 and Me are significantly run from a CLIA-certified laboratory, in fact from a laboratory that is regularly FDA inspected. So for whatever their position is on oversight, they are using a reviewed lab.

DR. TEUTSCH: I think we will welcome them here when we get a chance to, hopefully for the July meeting.

While we get the slides up for Pam, Kimberly

Layton, are you here? Not here yet? Robert DiTullio, are you here? Great. Kathy, are you here? Everybody has slides. Let's just wait a second until we know which slides we have up.

Kathy provided us some comments as a taskforce member yesterday and has some additional comments, I think, that she wanted to talk about from the perspective of the Policy Institute. So Kathy, thank you.

PUBLIC COMMENTS

Comments from Kathy Hudson, Ph.D.

Genetics and Public Policy Center

[PowerPoint presentation.]

DR. HUDSON: Good morning. My name is still Kathy Hudson and I'm still from the Genetics and Public Policy Center. The public believes and expects that genetic tests that they take to make important health-related decisions are analytically and clinically valid. As the taskforce report has clearly documented, they cannot have that confidence today.

Your recommendations need to make sure that there is adequate evidence and that that evidence is transparent to the public. As Marc Williams suggested,

we need to lift that curtain.

Yesterday's discussion, as you may have surmised by the murmuring in the audience, was troubling in several respects. There was a constant refrain that increased oversight will stifle innovation. In the absence of evidence that such stifling has or will occur, today manufacturers of IVD kits are subject to FDA regulation and if they were being stifled we would have expected to hear about it in the public comments.

To the contrary, the comments of AdvaMed, a trade association for device manufacturers, and Roche argue that more and not less oversight is needed.

There was no discussion about the deleterious impact yesterday of the status quo on innovation. IVD manufacturers face significant disincentives to produce validated test kits. The problem, of course, is that for any test kit a manufacturer can present evidence to FDA and go to market and the very next day Joe's Genetic Tests R Us can offer the very same test or make identical claims without having the oversight from FDA.

The absence of discussion of this yesterday may reflect that while there are significant numbers of LDT

providers on the Committee there are no IVD manufacturers on the Committee.

The Committee will not fulfill its mandate unless it makes recommendations that substantially level the playing field for businesses that can and are innovating in this space at a time doing the work necessary to get FDA approval.

Yesterday Steve made recommendations of what FDA does do and can do, and there were suggestions that perhaps we should wait and push a pause button on FDA oversight until various committees have met and registries are formed and we have achieved world peace. I would really make a very strong suggestion that you not handcuff FDA.

There was also considerable discussion yesterday about direct-to-consumer genetic testing. I want to make five points about direct-to-consumer testing. First, the map that was provided by Lewin was a map describing current oversight. I pointed out yesterday that the Lewin Group inaccurately showed that there is a non-CLIA regulatory pathway for genetic tests.

With the exception of those tests where it is

unclear whether or not those tests provide a health assessment, and that is a distinct minority, selling an LDT without CLIA certification is against the law. I would argue we do not want to include a pathway on our regulatory map that includes breaking the law.

Second, on a related point, the vast majority of DTC tests are subject to CLIA and they make explicit or implicit claims of health assessments. We have recently done a review of the direct-to-consumer tests, and that has been passed around. The majority of those claim that they are providing those tests from CLIA-certified labs. Of course that is difficult to verify because there is no publicly available list of CLIA-certified labs. We called Judy Yost to verify those claims.

Our review in this slide is already outdated, and it is about a week old. It shows that there are 30 companies offering health or health-related tests direct to consumer. So I would suggest that we don't want to make DTC companies the scapegoat here. There is a much bigger problem with all laboratory-developed tests, and it would be misleading and inaccurate to point the finger

solely at DTC providers. The failures in oversight apply across the board.

Third, there were a number of inaccuracies in statements about the regulatory status of direct-to-consumer tests. The definition of clinical lab is one that examines samples derived from the human body to provide information about the diagnosis and treatment of disease or for the assessment of health of human beings. This definition and all the CLIA regs cover labs whether they are being sold direct to consumers or through a provider.

Concerns were also raised about skirting oversight by claiming that genotype provides research information. Paul, you referred to 23 and Me's comments. There is an exemption in CLIA for research but only if those research results are not provided back to the research subject. So even if someone was saying that they were conducting research, they would have to perform those tests in a CLIA-certified lab if they are providing the results back, as 23 and Me is, and they are operating in a CLIA-certified lab.

Finally, yesterday the FTC representative said

that interagency collaboration on DTC is working. I'm not sure what that means. Since the issuance of a consumer alert 18 months ago, which was prompted, I think, largely by a GAO investigation and a Senate hearing, we haven't heard anything more about FTC's efforts on direct-to-consumer testing. What progress have we actually made.

Matt told the Committee that there have been no enforcement actions, this despite numerous consumer complaints to the agency, a class action lawsuit, and numerous clearly faults or misleading statements on DTC websites. Perhaps the Secretary could ask for or recommend that the Secretary check in on the progress of this collaboration and FTC's evaluation of these faults and misleading claims.

In closing, I ask that at the end of your deliberation you read carefully over your recommendations, and Reed asked that the Committee do this yesterday, to make the recommendations as specific as possible. In a year if we read these recommendations, will we be able to tell if there has been measurable progress or are they so mushy that we can't really

discern whether or not there has been progress. Thank you.

DR. TEUTSCH: Thank you, Kathy. Appreciate that. Any comments for Kathy?

DR. BILLINGS: I have one. Kathy, on this list, does that mean that each of these entities is making health-related claims around all these SNPs that have been associated with disease?

DR. HUDSON: Along the top are what they are offering tests for. So there is obesity. I would argue that is a health assessment. There are some that get a little on the borderline, but most of those are explicit health-related, disease-related claims. We haven't included ancestry or sort of recreational, "who were you related to" kinds of stuff.

DR. BILLINGS: Thank you.

DR. HUDSON: Muin?

DR. KHOURY: Kathy, do you have an answer to what the person from 23 and Me said yesterday when I asked her about the difference between health-related claims and otherwise? Because they have a service to try to inform and educate the public and they view this as

not giving advice on health-related issues. I don't want to single them out, but I think most of these on the list would probably do the same.

DR. HUDSON: So 23 and Me offers several services and one of those is clearly health-related, giving you information about your risk relative to the general population based on genome-wide association studies for a set of clearly health-related conditions: diabetes, et cetera. There are other parts of their service that I would argue are not health assessments but are providing genetic information.

We are in the process now of doing a careful evaluation, in fact using some of the work that you have led, Muin, from EGAPP. We are comparing what evidence EGAPP has found for various tests to the claims that are being made by the DTC providers and finding significant variance.

DR. TEUTSCH: Great. Thank you. Appreciate those thoughts, Kathy. Let's move on, then, to Robert DiTullio from AdvaMed. It looks like we are good to go with some slides.

Comments by Robert DiTullio

AdvaMed

[PowerPoint presentation.]

MR. DiTULLIO: Good morning, ladies and gentlemen. My name is Robert DiTullio, and I'm with Sequinom, a molecular diagnostics and research company in San Diego, California. I'm also co-chair of the AdvaMed's Diagnostics Taskforce. As such, I'm here to present AdvaMed's least burdensome proposal for the regulation of all diagnostic tests.

AdvaMed is the world's largest association representing manufacturers of medical devices, diagnostic products, and medical information systems. AdvaMed member companies produce the medical devices, diagnostic products, and health information systems that are transforming health care through earlier disease detection, less invasive procedures, and more effective treatments.

As some background, in 1997 FDAMA had a requirement for the least burdensome approach to regulation. More recently, MDUFMA had qualitative goals for the consideration of exempting some of the lower-risk

tests. Very recently, the SACGHS Committee drafted a report and in that report they highlighted the need for improvement in the current regulatory scheme.

We at AdvaMed, through our taskforce and our membership, have been working on this proposal for more than a year. Our number one, main underpinning of all of this first and foremost has been that patient safety is the key.

To address safety and effectiveness, we know that there are more than 1,000 genetic disorders where tests are developed in labs and these are not subject to FDA or CMS/CLIA evaluations of safety and effectiveness prior to use on patients. We advocate timely access to all safe and effective diagnostics regardless of where they are manufactured or used using a risk-based approach. We promote the application, as FDAMA required, of the least burdensome approach in doing so.

As the SACGHS report indicated, we need to modernize the regulatory scheme, and this proposal advocates doing so with the least burdensome approach, doing so by realigning the intensity of regulatory oversight with patient risk benefit ratio in mind, and

allowing FDA to focus their limited resources on only the highest risks. We promote the FDA oversight of safety and effectiveness of all diagnostic tests regardless of site.

We are presenting this proposal and the underpinnings are seven key principles. The first principle is that all clinical labs should be subject to CLIA requirements and quality standards. We believe FDA should oversee safety and effectiveness of all diagnostic tests no matter where they are made because they have the same risk benefit profile for patients.

We promote FDA oversight of tests, and that oversight should focus primarily on the risk of harm associated with how the test result is used to treat patients, not only on new technology or the novelty of the analyte.

To further the third principle, we believe that low-risk tests and well standardized tests should be exempt from FDA pre-market review or only subject to labeling review of the performance claims. This would allow the FDA's resources to be used toward the higher risk tests, and these should be cleared or approved using

a risk-based approach that aligns data submission requirements and the intensity of the review with the risks.

We also promote the fact that patient access to specialized test categories should not be disadvantaged.

FDA and CMS should harmonize regulatory requirements for diagnostic tests and leverage each other's standards and resources for oversight of lab-developed tests. The new oversight system should be implemented through notice and comment rulemaking and guidance as appropriate.

Our seventh principle is that CMS must recognize that all new diagnostics must receive timely and adequate reimbursement.

DR. TEUTSCH: That is helpful. Do we have any comments or questions?

MR. DiTULLIO: There is still some more.

DR. TEUTSCH: I know. We are at five minutes, so if you can finish up in just a few seconds.

MR. DiTULLIO: Yes. Actually, I was not finished, but --

DR. TEUTSCH: Do you have another point or two

you would like to make?

MR. DiTULLIO: Actually, what I would like to do is to propose some questions for consideration for the final report of SACGHS. One such question is, will formal, risk-based, independent review of critical elements, such as intended use, analytical and clinical data, limitations, et cetera, take place before the test is commercialized and available to patients? Will it be assured that claims are commensurate with data provided?

Another compelling question we believe, are more limited post-market reporting requirements such as NDRs and recalls alone adequate to assure patient safety? I thank you for the opportunity to comment.

DR. TEUTSCH: Thank you.

DR. BILLINGS: Under your model of FDA oversight of all tests, what role does CLIA play?

MR. DiTULLIO: CLIA plays a role of making sure that all the laboratories follow the existing CLIA regulations with regard to their quality standards and also, as I said in one of the principles, there should be some meeting of the minds between the FDA about a future version of how pre-market regulation might be had.

DR. TEUTSCH: Muin, did you have a comment?

DR. KHOURY: The concept of safety and effectiveness according to AdvaMed, can you go over that? Because I'm struggling with two ideas here: clinical validity of a diagnostic test, meaning sensitivity, specificity, et cetera, and then clinical utility. Are you suggesting the FDA regulate clinical utility as well or just to go after clinical validity? Some of the discussion here yesterday was focused on clinical validity.

MR. DiTULLIO: The FDA process should remain as it has been all along. That is what AdvaMed is proposing. What we are proposing is that they focus only on the higher risk products and do so in a risk-based approach. We are not advocating a change to how FDA currently does their review.

DR. TEUTSCH: Jim and then Reed.

DR. EVANS: Can you just give me a quick example of a low-risk test and an example of a high-risk test?

MR. DiTULLIO: BUN or urea is a low-risk test, and there is no reason for there to be any review of

that. A high-risk test could be a viral load HIV.

DR. EVANS: And the criteria for determining low risk and high risk is?

MR. DiTULLIO: In one of the slides that I wasn't able to show was that, through the qualitative goals that were given by MDUFMA, we agreed that we were going to make a presentation of an exemption proposal for the low-risk devices and present that to the agency by the middle of this year. We are going to do so with a tier-triage approach where we took into account risk and the novelty of the analyte, the novelty of the technology in a matrix fashion. We are also planning on presenting a flow chart that will help the FDA implement this.

DR. EVANS: I don't mean to belabor this, but risk for? I just haven't --

MR. DiTULLIO: Risk for how the tests are used on patients.

DR. EVANS: So for example, a test that is wrong and the impact that might have on the patient.

MR. DiTULLIO: That's right.

DR. TEUTSCH: Reed.

DR. TUCKSON: Jim got mine. Thanks.

DR. TEUTSCH: Thank you so much.

MR. DiTULLIO: Thank you very much.

DR. TEUTSCH: We also very much appreciate the extensive comments you provided as part of the earlier process. Thank you very much. Appreciate it.

MR. DiTULLIO: Thank you. So, welcome to Pam Dixon from the World Privacy Forum.

Comments by Pam Dixon

World Privacy Forum

[PowerPoint presentation.]

MS. DIXON: Good morning. Thank you for waiting. I'm Pam Dixon, executive director of the World Privacy Forum. We are based in San Diego. We are a nonprofit public interest research group. We focus on in-depth analysis of privacy issues and also more longitudinal research of these same kinds of issues. One of our focus areas is on healthcare privacy issues.

Our take is a little bit different than pretty much everything else that I have heard so far in this meeting. We are really interested in the aspects of privacy that we felt were slightly underrepresented in the otherwise very, very thoughtful and deliberative

report.

Our concern is will marketing interests and misused science crowd out legitimate genetic testing and privacy. What we are looking at is really occurring outside the clinical sphere. We believe that you guys are doing an excellent job of looking at the issues within the clinical sphere, but we think that there are other issues and mischief potentials outside that sphere. That is just what I want to walk you through a little bit today.

One of the things that we really looked at in this area is something that is already occurring in the healthcare sector, which is privacy activities related to consumer-consented healthcare data.

For example, right now if you go to something called DirectMeg.com, which is a big direct marketing magazine for marketing companies, and you go to something called the List Finder, you just search the List Finder's 60,000 marketing lists. I typed in "diabetes." The reason I typed this in is because this is a mature market. As you can see, there are 406 lists containing "diabetes."

Now, when you look at these lists, some of them are for magazines and what not, but most of them are generated from actual consumer healthcare data.

This is something called a data card. A data card basically tells you what is being sold about the consumer. In this particular case, you have 2,186,700 consumers who are known and identifiable to this list, and there are 400 data points about the consumer.

You can e-mail them, you can find out all sorts of things about them. It is 53 percent female, 47 percent male. The source of the data was often e-mail. But anyhow, you can select whether they are type I or type II diabetes. You can look at the average household income, which is $48,000 per year.

Then, if you look over here, these are selects. Selects are something that you can choose to purchase along with the base list. You can purchase the age of the person, the age of their children, their education level, their ethnicity, their gender, and again you already saw the income, the prescriptions and what over-the-counter medications they take, and all sorts of other marketing activities and purchasing activities that the

consumer has engaged in.

So that is just one of the diabetes lists. I typed in "genetic," and we are early on this, very, very early, but I found a list. If it weren't sad, it would be humorous. These are 54,000 primarily men who expressed interest in Ferrari Hair Centers.

[Laughter.]

MS. DIXON: I apologize if I'm offending anyone in here. Anyhow, I don't know about you, but this sentence is very interesting to me. "The Ferrari concept of genetic hair restoration," blah, blah, blah. You get the idea. Anyhow, these people who opted into this list in some way can be sold, trussed up, and delivered to the marketing company.

The problem here is, this is goofy, but in the future we expect this to look much more like the diabetes list, where you have a person's name, home address, number of their kids, maybe even names of their kids, education level, income, and everything else you might want to know.

This is actually just a random list I pulled. This actually is of mental and behavioral disorders of,

again, individually identifiable consumers. At the very top you will see, "Ventee, an Experian company, has the industry's largest and most comprehensive consumer database of self-reported online data compiled from three reliable sources, including online surveys, direct response e-mail marketing, and consumers visiting Ventee websites."

So that is just the point. This is not clinical data that is leaking. This is consumer-reported data. Our concern is that as this area of direct-to-consumer advertising and genetic testing and also consumer-initiated genetic testing matures and also the price drops, I think we are looking at a situation where this kind of thing can really get worse and start to impact consumers.

I think the outcome that we all want to avoid is a wild west data rodeo where consumers have initiated genetic testing through some kind of Web portal or online site and the genetic test can be a fake genetic test, or it can be a real one, but the point is that the data is collected and then used for marketing purposes.

Now, in the genomic world, you have a consumer

whose information impacts them, their employability, insurability, and other potential harm, but also,their family, their progeny. So this is a persistent privacy issue.

We submitted comments last December. I will leave those to you. You can look them up online. We made three recommendations in those comments. First, we asked that privacy be expressly included in the draft report as an issue to be looked at, including privacy outside the clinical setting.

The second recommendation was to task a group. We worked with this and we were thinking who could it be. We thought maybe NCBHS. But anyhow, to figure out how that might look to address the specific privacy issues that come up in this context because they are complex, and not to be flip here in showing you genetic Ferrari whatever, but it is a difficult task. When you mix the complexity of genomic work and then also privacy work, it gets quite difficult.

But a recommendation we wanted to add is, the Federal Trade Commission right now is working on and asking for input on what to do about advertising to

consumers online and whether or not medical information or healthcare information should be included in that tracking kinds of advertising or not.

We are asking the Committee to think about working with the FTC to urge them to say that no, genetic data and requests for genetic tests on websites and this sort of thing should be off the table in terms of advertising, being able to use this data for marketing purposes, or any purposes other than a person's health care. Thank you very much.

DR. TEUTSCH: Thank you very much, Pam. These are obviously important issues. Marc.

DR. WILLIAMS: I also wanted to say thank you for that. I must admit, as I have reviewed the comments I thought that they were eye-opening. I think you will find that we incorporated some of the suggestions that you had made in our recommendations.

The specific question I wanted to ask you is, it doesn't appear from your presentation that you have actually identified any of the direct-to-consumer genetic testing companies that are actively asking for permission to collect this information, but I'm just curious.

Looking at this list that Kathy Hudson presented, how thoroughly have you looked at the landscape, and are you aware of anywhere they are asking consumers basically to check and say would you be willing for us to share your information.

MS. DIXON: We are aware of some companies that are already doing that. We are debating on how we want to approach that issue, whether we want to do a substantive longitudinal research study or if there is some other mechanism.

So our first step in addressing this issue is to address it broadly without going after any particular company and see what this Committee came up. We are hoping for a deliberative process that is thoughtful and hoping for the best.

That being said, we are very concerned about some of the information our research in this area has turned up. One of the great issues is that a privacy policy really is a very thin scrap of contractual material to separate a consumer from harm. The privacy policies, some of them are quite dense. I think it would be difficult for a consumer to read and have a really

clear understanding of what is happening.

So that is one concern. But then, of course, the second concern is right now a lot of the actors in the field are primarily good actors. We are thinking that down the line there will be a proliferation of bad actors who make the current landscape look like Disneyland.

DR. WILLIAMS: I would just follow up on that and say that in our discussion of those recommendations yesterday -- I don't think you were here for that.

MS. DIXON: No. I was trying to get here.

DR. WILLIAMS: I can tell you that when I presented, inadequately, but attempted to present the information that you had provided for us that there was quite a bit of interest from FTC about this because it seemed to be something that they were not specifically aware of. So I think that there would be a receptive ear if you have some data that suggests there really is some untoward activity.

MS. DIXON: Thank you very much.

DR. TEUTSCH: Joseph.

DR. TELFAIR: Thank you, again, for the

presentation. I think Marc's last statement almost covered what I was going to ask. As you are considering your study, one of the concerns I know that has come up is the building of the evidence that there is harm, not just potential for harm but actual harm itself. I was wondering in your considerations of your design of your study, are you going to begin to put into the study a means by which you can detect that through either firsthand cases, secondhand cases, or whatever?

MS. DIXON: Absolutely. We will have a peer-reviewed methodology before we ever begin. So if you are volunteering, that would be fabulous. But yes, thank you.

DR. TEUTSCH: Thank you very much, Pam. These are particularly important issues and we appreciate your demonstrating them so vividly and also for your comments earlier.

MS. DIXON: Thank you very much.

DR. TEUTSCH: Is Kimberly Layton here? With the weather she is probably stuck somewhere because she comes from Virginia. We will give her another chance if she is able to get here later.

Let's move on to the main task of the day. As all of you know, what we have to do is get to our set of final recommendations and get to approval today. So I will turn the gavel, as it were, back to Andrea and to Reed.

Reed somehow ended up way down there. Can you see us down there, Reed?

DR. TUCKSON: Hello. When you are gone, you are gone.

DR. TEUTSCH: You get the corner spot.

So we are going to start off where we left yesterday in the discussions on the recommendations for Chapter 4.

SESSION ON OVERSIGHT OF GENETIC TESTING

Development of Final Recommendations

DR. FERREIRA-GONZALEZ: After you all left, we stayed here and continued to muddle through. We have come up with an alternative version for Recommendation 4. I'm going to give you some time to read this recommendation. Actually, I could read it. Let me go ahead and read it.

"The Committee is concerned by the gap in

oversight related to clinical validity. The Committee believes that it is imperative for this gap to be closed as expeditiously as possible. To this end, the Committee makes the following recommendations:

"All high-risk LDTs should be reviewed by the FDA in a manner that takes advantage of its current experience in evaluating laboratory-developed tests. In order to accomplish this recommendation, the Committee recommends convening a multi-stakeholder public and private sector group to determine the criteria for risk stratification and a process for systematically applying these criteria.

"The multi-stakeholder group should also explicitly address and seek to eliminate duplicative oversight procedures.

"For all other tests, this multi-stakeholder group is also charged with the development of a review process that meets the needs of protection of the public. This group should also consider existing regulatory models and data sources, e.g. New York State, and responsibility for overseeing this review process should be defined by this group.

"To expedite and facilitate the review process, the Committee recommends the establishment of a registry as noted in Recommendation 3."

Yes, sir.

DR. FITZGERALD: I know we are trying to get these recommendations to evolve in a more precise way. My concern is that we have in Part A, a term that we then say we don't know what it means and we need to define it. So "all high-risk LDTs," and then later on we say we are not quite sure what high-risk is and we don't define it in the report.

Now I'm worried just about the logic of saying these should be reviewed but we don't know what they are. In the end, when we do get the stratification, we may think that all moderate and high risk need to be reviewed.

I know we went from "complexity." We said yesterday "high complexity," which was not clear, either. I guess we are still struggling to figure out exactly how we want to categorize this, but I don't want to use a term that we then have no standards for. Not in the recommendation.

DR. TUCKSON: What do you suggest?

DR. FERREIRA-GONZALEZ: So move the bullet as Part A and where they convene to determine what is high risk.

DR. FITZGERALD: Exactly.

DR. FERREIRA-GONZALEZ: And then what high risk is then has to go through the FDA review.

DR. FITZGERALD: Right. So what we are saying is, regardless of what they come up with. We don't know what high risk is.

DR. FERREIRA-GONZALEZ: Well, there is some definition today of what high risk is. We might not fully agree to that definition of high risk. Steve?

DR. GUTMAN: Implicit here is the fact, as I said yesterday, we have been classifying products according to risk for 32 years. The original classification system was recommended by the FDA but it actually all went to advisory panels. FDA actually isn't responsible for final risk determination on anything except the fundamentally new products that went around in the late '70s or early '80s. We tended to follow the precedent set in the late '70s or early '80s.

So I guess I have either a concern, or maybe not a concern but certainly an issue to put on the table, which is, is there a proposal that there be one set of risks for commercial IVDs made by Abbott and Roche and BD and Beckman Dickinson and then we will have a different but special risk system for LDTs? If that is the case, then this would be a very effective way of accomplishing a two-tiered risk system, one that now exists and has been used for 30 years and one that you would put prospectively because of the unique status of LDTs.

That wouldn't create parity and it might create confusion and chaos for everyone to make LDTs, but it would be an option.

DR. BILLINGS: Can I ask a point of clarification to that, Steve? Are you saying that a small lab in the Midwest who develops markets or produces a test for inflammatory disease is aware of the FDA risk system and knows that you might classify that as a type I or Class 1 or Class 2 risk?

DR. GUTMAN: No, I doubt that a small lab would be aware of the risk system.

DR. BILLINGS: Right. So despite --

DR. GUTMAN: That is a communication issue.

DR. BILLINGS: Well, it is a functional issue of how FDA has decided to use its enforcement.

DR. FERREIRA-GONZALEZ: So let's go back to what we currently have on the table. In Bullet C, we said for all other tests this multi-stakeholder group is also charged with the development and review process that meets the needs and protections of the public. Maybe also, to get to some of these points, here we might be creating a parallel system. I think it is okay the way we have defined it here.

The notion that maybe we can have C be the first one, where this stakeholders group develops or evaluates to develop a review process that is appropriate for this type of testing that meets the protection of the public, which is underlined. Unless they look at models of data sources that maybe we can also evaluate new models. I think Mara brought up some of the issues yesterday that maybe they need to be considered for this.

We can say that maybe, in the meantime, as this process moves forward, the high risks, as they are classified today, have to go through FDA review.

DR. GUTMAN: We certainly would prefer the ability to move forward on a risk base and go after the high risk. The proposal which is still emerging is one that would take, I think, existing risk products that are currently certainly not high risk but perhaps moderate or some of the higher low risk. I don't know that it would actually change their risk but it would change our review practice, a very major emphasis that I don't think was clear. I don't think there was time on the slides we have seen yet on the proposal, so I don't know if it has been submitted yet to this group or not.

But a critical issue wasn't changing the risk, it was assuming that the risk was okay. It was suggesting that perhaps for certain risk products we would change our approach to how to handle them. I would actually argue that BUN might not be such a low risk product. You are not going to do a mastectomy or a prostatectomy based on a BUN, but you are going to be making decisions.

But the controls that they were suggesting, because it is a well established analyte, was that it may be -- I'm suggesting this -- exempt from pre-market

review and it be captured by the quality system regs.

So from my perspective, God forbid, I do think we could refine our risk system. I hate this recommendation because it strikes me as exactly whoever said, "Gee, if you recommend you go to a committee to decide how to recommend, you are not going to get anything done for three years." So I actually hate Recommendation A. I think FDA ought to be allowed to use its work and consult and expand.

We are not even sure the registry is legal in terms of what FDA statutory authority currently exists, but if the registry was done, frankly, at Westinghouse or at GE or at the McDonald's, the point I was trying to make yesterday was that it has to be credible. If it doesn't have pre-market controls, then it has to have strong post-market controls.

DR. FERREIRA-GONZALEZ: It seems to me that you are currently undergoing a process of gathering information about other models of the process of review. What you just mentioned goes in line with Recommendation C here, at least, where we have convened multi-stakeholders to develop a review process that meets the

needs of protection of the public.

DR. GUTMAN: I don't object.

DR. FERREIRA-GONZALEZ: They need to look ta the current models that exist there and maybe look at new models there. That could be a recommendation. But there is still a concern in the public about the high risk. Muin, Mara, Jim.

DR. KHOURY: I'm getting a bit confused here. I know we did this as a result of the discussion yesterday, but if I look at this cold, which I tend to do in the morning, it just doesn't make sense to me. What are we saying here? Why can't we be more direct? We recommend that FDA does something. Or, we recommend HHS does something.

This looks like it is all written in a torturous way. The Committee makes the following recommendations. It seems to me it ought to be the other way around. All LDTs should be reviewed by the FDA. The FDA should do something. What is it we are recommending the FDA to do? Is the FDA convening the stakeholder group?

DR. FERREIRA-GONZALEZ: No, we can say HHS. We

can say HHS should take the lead. We can add that the HHS convene the stakeholders.

DR. KHOURY: Going back to the framework here, I'm a bit confused. Is this preamble only concerned with the clinical validity of tests? This is all a registry of clinical validity or a registry of safety?

DR. FERREIRA-GONZALEZ: No, this is not the registry.

DR. TUCKSON: The registry is another recommendation.

DR. FERREIRA-GONZALEZ: This is just clinical validity and the review of those tests. When we go to the registry, again we are going to have to figure out what we put in there.

DR. KHOURY: But why can't we be a bit more explicit about what we are asking the FDA to do? We are saying do this. The multi-stakeholder group should do something. So we have one group here, a stakeholder group. You have the FDA, you have all the agencies. It is not clear what we are asking.

DR. TUCKSON: If you start out then with the firm recommendation, the recommendation is that all high-

risk LDTs should be reviewed by the FDA, and they ought to do it being informed by what they currently do, whatever the adequacy or inadequacy. Boom, done.

Now, the only point that the next point was is to say we aren't in a position, unless Kevin is, to say what is the definition of high risk. Let me just ask, are we in a position to say what the definition of high risk is? Do we have criteria for that?

DR. TEUTSCH: No, not right now.

DR. TUCKSON: So if we don't, all we are simply saying is then there needs to be some process urgently undertaken to define what is high risk. Now, do we want to be more directive than that? How much more directive can we be about high risk?

DR. KHOURY: Remember, Reed, in SACGT we spent almost a year under Riley Burke's able taskforce trying to define high risk and low risk. I think at that time we didn't succeed very well.

DR. TUCKSON: So, what is your guidance?

DR. KHOURY: Let the FDA do its thing.

DR. TUCKSON: Let the FDA go forward. Let them figure it out without having to create a committee.

DR. FERREIRA-GONZALEZ: Steve, what is your definition of high risk?

DR. GUTMAN: High risk is like life-threatening. It is actually in the reg. I don't have the reg with me. But high risk is making cancer decisions, mastectomies or not mastectomies, intervening in cardiac care. I would argue that there are many things that people would construe as high risk that we would call moderate risk. Glucose, which I think is actually a very high risk that you are going to dose insulin, we call that moderate risk.

So our risk classification systems tends to actually err on the side on the side of gentleness.

DR. TUCKSON: Let's be real clear, then. The first decision here is do we leave it up to the FDA alone to determine what they consider to be high risk and they then go and review. All you should be saying now is, either you want the FDA to be given the authority to do it or you feel like there needs to be a multi-stakeholder committee to advise on the definition of high risk. Which is it?

DR. WILLIAMS: I have two specific comments

related directly to that issue, which is right on. The first is that we are not being responsive to the public comment that we have received on this if we say just let's continue with the way things are currently working with risk stratification.

We have had numerous public comments that have said there are issues with trying to apply risk stratification as it is currently done to LDTs. Therefore, we think it is appropriate, and the reason we incorporated this recommendation, that we need to bring the stakeholders together, which I think could be done in a very short period of time, to say what tweaks do we need to make to this system to actually make it work.

I understand Muin's concern about the SACGT spending a year on this, but that was Reed's point. This isn't the group to do the risk stratification.

DR. TUCKSON: Now, is there anyone who agrees with Muin's point that you do not want to go to the extra step of having a convening group to talk about it, just turn it to the FDA and let them do their thing? Is there anybody that wants to support Muin's point?

DR. FERREIRA-GONZALEZ: I don't see the problem

of convening a group. If FDA had deliberations with the group, they might come up to the final decision that yes, that is the way to do it, and then we are okay.

DR. TUCKSON: I'm looking for supporters of Muin only. We already know the other point is on the table. Sylvia.

MS. AU: I think we are discussing why don't we have them go ahead with what FDA uses for risk stratification while the group is seeing if that is the correct one or we have to add on so that then the high risk, once defined by the FDA, must really be high risk and they should be doing something.

DR. TUCKSON: So a simultaneous strategy.

MS. AU: Yes.

DR. TUCKSON: So we have two options on the table. I only want you to speak to the options on the table. Or make a different option. You can add a new option.

DR. FERREIRA-GONZALEZ: I have Mara first, who has waited for a while, Kevin, and then Jim.

MS. ASPINALL: I think the two options are, or maybe there are three: convene the group; don't convene

the group, just have the FDA do it; and do it simultaneously. Let the FDA do what it is doing now and convene the group.

I would speak against the simultaneous approach. One is I think it creates confusion in the market. For LDTs, which are being potentially newly regulated, you would say between February and May we are going with this definition and the group is coming together and may change the definition. I think that is very hard for laboratories. First of all, it gives you a disincentive to get involved soon because the rules might change. So you will say, I will just wait it out.

And I think that human nature being the way it is, if we convene a committee and they are already doing it one way, I think it gives the committee less incentive to get it done quickly. So I like Proposition A, which Marc spoke about, which is getting the stakeholders together, getting it done, but I'm going to come back later today to put a timeline on it so it doesn't go a long time.

DR. TUCKSON: Any other comments need to be just that clear. Is there anybody that has another point

of view? Because otherwise we are going to take a poll.

DR. FERREIRA-GONZALEZ: Anybody with a different point of view?

DR. FITZGERALD: So in the meantime, Mara, what happens? Nothing?

MS. ASPINALL: In the meantime, there is draft guidance out now, which I don't know if this is finalized, but that we put this in quickly, the stakeholders get together within a period of -- I don't know if we can manage weeks but certainly not more than a few months, and we get clarity and we have something that sticks, not something that we have another interim period, which I think creates confusion and awkwardness.

I believe that many companies will game the system pro or con in a way that doesn't serve the best interest. We should put in the system and go forward so laboratories get the clarity of where we are going.

DR. FITZGERALD: May I respond to that? Here is my concern. I understand your concern. Looking at it from the flip side, if one looks at the experience of trying to wrestle with this issue up to this point, as Muin pointed out, this is not something that is easily

determined. So you might be able to force a complex group of stakeholders together, tell them at the end of six months you want an answer, and not get a good answer in that period of time.

I'm just saying that is a possibility. If that were to occur, you have put all your eggs in one basket. So my concern is to say why not let FDA do what FDA is doing. I don't know if we want to call it "simultaneously" or "in parallel" or whatever. I would just throw out "in order to accomplish this recommendation," that phrase, from that little bullet point.

So A, FDA reviews high-risk LDTs according to your current standards. B, the Committee recommends convening a multi-stakeholder public group. Then the other B just folds into that because that is the same multi-stakeholder group.

DR. FERREIRA-GONZALEZ: This is critical. It might hamper access to certain testing because laboratories don't know what is coming down the pike.

DR. FITZGERALD: Right.

DR. FERREIRA-GONZALEZ: So I concur with Mara

that we have to have, maybe, the stakeholders convene in a very short time and then come up with a decision before they go through this continued high risk. If I have a high risk and I know that there is another group that is going to be coming up with a different definition of high risk, I'm just not going to put it through.

DR. TUCKSON: One last comment and then we are going to poll the sense of the Committee to get a consensus here.

DR. BILLINGS: Just to Kevin, if nothing happens, we still have that very complex framework that the Lewin Group presented yesterday that is in existence. FDA has been acting to do its regulatory thing with that framework in place. So if the Committee takes nine months or six months to figure out what a high risk thing is, it is not like there is nothing left. So I don't think we have to fear a great deal of the absence of any oversight.

DR. TUCKSON: Now, let's just start to see if we can't push to a quick consensus here because we really do have to move this. You have a couple of options on the table, and it boils down to how many of you think

that we ought to define the FDA as moving forward in this field now and not worry about the convening of other body, that the FDA should do this, they have the authority to move forward, and that is what happens? How many of you are of that posture?

[Show of hands.]

DR. TUCKSON: Did I see four? Oh, yes, the other side. Ex officios. Everybody is on this. This is not a formal vote. This is a consensus. So, four. We have four altogether.

Now, how many of you believe that the FDA ought to move forward, because they already are. You can't stop them. They exist, as Paul's point was made. They are doing their jobs, getting paid by the citizens. While that is happening, urgent convening of a group that will recommend as we have here? So the convening of a group occurs and that will then ultimately inform the process. How many of you are in favor of that strategy?

[Show of hands.]

DR. TUCKSON: That is the overwhelming majority. All right. I think you have a pretty clear sense, so I think that you have declared where you really

are. Yes, Joe.

DR. TELFAIR: My question is in terms of the turnaround on this. There is more than one type of methodology for convening a multi-stakeholder group in terms of convening. There is more than one strategy. There is a rapid strategy or there is the more traditional strategy. Since I don't know what is it you all were discussing, I'm just wondering is that something that could also be a recommendation as a strategy that actually speeds the process a little bit more?

DR. FERREIRA-GONZALEZ: We have Jim and then Mara.

DR. EVANS: Two things. I think we get sidetracked by this whole issue of finding high complexity or high risk. Correct me if I'm wrong; it seems to me that the reason you even bring up high risk is a real-world constraint and a necessity to triage FDA's limited resources and approach this huge universe of LDTs.

I think that what we can say is that FDA needs to tackle this, that the bottom line is that all LDTs must be safe and must be sufficiently regulated, and that

we can tell them there are gaps here and this needs to be done posthaste.

As far as triaging which ones they feel are most in urgent need of oversight, that is something the FDA has been doing for a long time, I imagine, figuring out what they need to tackle first. There is nothing wrong with us saying that this has to be done quickly and maybe a very blatant statement that all LDTs need to be addressed by FDA.

DR. TUCKSON: Jim, I think you are speaking to the sense of what we have as a consensus. You are adding specific stuff around urgency. Mara had a point about urgency. I think we all recognize that. So the writing can capture the urgency words in here. Everybody is talking about not playing around.

You might want to have the subcommittee, as you capture the final language, get to Muin's point a bit by just stressing in the body of the narrative that the point is that the FDA is doing what it is doing, move forward. FDA should not be screwing around waiting, sitting on its hands for nine months until some committee [meets.] I think the sense of the Committee is pretty

clear on those points. Muin didn't get everything he wanted, but he got a lot of it.

So the consensus is there. Let's move to the only other issue that you didn't describe. There was a throw-out from Steve which I think people need to deal with quickly, and that was this issue of these parallel programs for LDTs versus IVD/IVDMIAs.

I don't know if I understood it. Anybody else understand whether or not we have created a single system or two different systems? Steve, do you want to explain that real quick, what we just did just now in terms of the consensus?

DR. GUTMAN: I'm not sure I can. I'm not sure I'm following because I'm hearing different things.

The current risk system, whatever else you can say about FDA's risk system, whether you swear by it or swear at it, is highly public. Even our interest in IVDMIAs isn't exactly a state secret. So everything we are interested in and the direction we are going, the current classification, our initial foray into future regulations, it is just all open.

DR. TUCKSON: So Steve, the consensus is that

you will keep doing what you are doing. Now, having said that, did we say anything else that creates a parallel universe, or is it one?

DR. GUTMAN: That is not what I thought the vote actually came out. That is what we would like, but in all honesty, I wasn't sure that that was the direction that I was getting.

DR. TUCKSON: Mara.

MS. ASPINALL: I think Reed's comment and Steve's comment may be sort of crossing. I think there were two issues. One is, I thought the last vote said, which I agree, have a multi-stakeholder group in an urgent manner, and deals with Joe's issue. I would say it is too prescriptive to say the details of how they do it, but rather give them the timeline.

To Kevin's issue, they may not come up with the perfect example, but work expands to the time allotted. It may not be perfect in six months, but it may not be perfect in three months, so let's give them a short period of time and get it done to emphasize the urgency.

But I think that was the sense of the Committee on that issue. Your question, Reed, to me is a different

one, which is, is there a different system for IVDs than there is for LDTs. I don't think we have discussed that, so I don't think there is a consensus or a proposal. Well, maybe there is a proposal, but I don't think we have discussed that formally.

DR. FERREIRA-GONZALEZ: I think we did in the text of the report. We made a distinction between the product and the LDTs that we consider services. So already in the report we are bringing out that there are differences between these two.

MS. ASPINALL: I would like to suggest that we continue to recommend that there are differences. In reality, there are important differences both in what is done by the laboratory, the number of laboratories doing it on any one piece of data or analyte, as well as the volume that is typical of the average LDT versus the average IVD.

That was one of the very important things we saw in the public comments. It is just, quite frankly, infeasible for many LDTs to go through the exact same process an IVD might go through because of the relative numbers of putting together the data and the relative

resources. We heard a lot of that from the university laboratories as well. It is not feasible to go forward and therefore it becomes an access issue.

DR. FERREIRA-GONZALEZ: Marc, do you have a comment?

DR. WILLIAMS: We are obviously all hearing different things. I heard Jim very clearly state that his recommendation was that basically to avoid parallelism that all LDTs and all IVDMIAs undergo a triage process by FDA using their currently available standards, so that we basically eliminate this avoidance of FDA oversight.

We would also then look at this multi-stakeholder group to address some of these other issues so that if we can refine the process to address this, that that would be the way to move forward to try and facilitate that.

Again, I think you don't say to FDA "You don't do this." I think the point is that we really want to be directive but if we are really concerned about the end user, the public, then you have to say we can't allow tests to come out where we have no data about anything

relating to clinical application of the test. That is just not acceptable.

DR. TUCKSON: Let's get our philosophies straight, then. We have been struggling with this dichotomy forever. The poor small folk, we can't overwhelm them with a whole bunch of regulations. Yet everybody, we have also said, has to be regulated. At some level, you can't have anybody slipping through the cracks, so we are closing the crack.

Now the notion is, should the process coequal you regardless of whether you are an LDT or an IVDMIA folk. We have two different points of view on the table. One is saying yes, there should be a difference, and one is saying that there shouldn't.

Other than the people that have spoken, who is strongly in favor of one side of that equation or the other? Where are you on this issue? This is a big issue. Do you go easy or do you recognize a differential in these two populations?

MS. ASPINALL: Not necessarily easy.

DR. TUCKSON: Differential. One process or two. Kevin looks confused.

MS. ASPINALL: I just said not necessarily easy, just different.

DR. TUCKSON: So, where are you at? Confused, not sure? See it as a difference without a distinction?

DR. FERREIRA-GONZALEZ: We are asking in C for another group of multi-stakeholders to be charged with the development of a review process that meets the needs of protection of the public. Can that group wrestle with this notion?

DR. TUCKSON: It is a punt. So you want to punt. Muin doesn't want anybody punting anymore. He is tired of punters. Steve, would you again, just very quickly, say again what are your concerns about having parallel systems from an administrative point of view?

DR. GUTMAN: The concern about the parallel system is what Mara said. You need to be careful and like what you wish for, and I don't wish to chill technology and I don't wish to review 1,200 genetic tests, many of which may be very exotic and rare.

So that is a negative side. AdvaMed got a little bit truncated there, but I think they have a point. A lab test is a lab test. If I'm a patient,

whether my lab test is graded by the Mayo Clinic, by Steve Gutman Lab of Rockville or by Genzyme, I want the damn thing to work and I want somebody to be responsible for it working.

So as a patient, whether it is FDA or CMS or HHS or AHRQ or CDC, the damn thing should [work.] Somebody has to stick up their hand and say "I'm taking responsibility that this has been quality controlled not by the sponsor."

DR. TUCKSON: Paul's hand is up, and I want to get there, but that sure resonated as clarity. Let me just make sure I know where you ended.

I think everybody is on board with you -- I'm watching the heads nod -- when you say the doggone thing should work, there should be an independent party that says it should work, and so forth and so on. Your first part of getting to that, do you say that you are prepared to accept from your experience two different mechanisms to get there?

DR. GUTMAN: I think if it is a rare test, whether it is made by Abbott or whether it is offered by the Mayo Clinic, it is a rare test and we have to be

careful about what we wish for.

DR. TUCKSON: So, two different.

MS. ASPINALL: No, he's saying the same.

DR. TUCKSON: It's one? So one process.

DR. GUTMAN: Yes. Maybe it needs to be tampered with, it has to be fooled around with, but yes.

DR. TUCKSON: Clear. I stand corrected. I'm trying to hear. One. Paul.

DR. FERREIRA-GONZALEZ: You talked about a system, but you just said that you can't actually review all of them.

DR. GUTMAN: No, so I do think that the Department is faced with a challenge.

DR. FERREIRA-GONZALEZ: Exactly.

DR. TUCKSON: Everybody, Paul has his hand up.

DR. MILLER: I had better say something important and relevant.

[Laughter.]

DR. MILLER: It strikes me that in a sense the group is coalescing around some ideas but that there are different perspectives about that same idea. The way I see it is that there is a policy issue about what the

process should be. Quite frankly, I'm hearing that there is quite a good deal of agreement about what the policy is. You have just stated that we have heard the heads nod.

Where the breakdown lies is in a sense how do we get to actually effectuating that policy. Does the reality work. I would submit that if we agree on the policy, and this is a policy body, that we frame it in a way back to HHS to say here is where the group is and this is what we think is the right thing to do. It is not incumbent upon this Committee to work out where the resources are or whatever. It is for this Committee to say here is what the right thing to do is, here is what our panel of experts says, and let's move forward and make that work.

DR. TUCKSON: So we have Julio and then James. With that, try to get us specifically on where you are on this specific thing.

DR. LICINO: My comment was a little broader than that. Just studying up some of the things that were discussed yesterday -- I wasn't here but I went through the materials as to what is being discussed now -- people

were saying that this has to do even with freedom of information, just telling people who they are. It is just freedom of information and giving your genetic blueprint.

But if you are telling people that they have a risk of myocardial infarction, they are going to change their lifestyle. They can change their life dramatically one way. If it tells them that they have a risk of Alzheimer's, they may completely shift their whole entire lives in a different way.

So I think in a sense these tests are even more important and more in need of regulation than BUN and 057 or 120 or 130. We split the hair over the precise accuracy --

DR. TUCKSON: That is good, Julio. That is terrific. Jim, what I need you on is one or two systems. That is all I need to hear now.

DR. EVANS: You aren't going to get that from me.

[Laughter.]

DR. EVANS: I will be clear and tell you that I think we need to quit parsing. We are parsing on

parallel and differentiating LDTs versus IVDMIAs. We are differentiating about high complexity, et cetera. That is not what we should be doing. We should be saying just what Steve said, that every test that a patient gets should be reliable. Number one. Number two, all that parsing is just code for the fact that real-world constraints make it difficult to implement, which is what Paul is saying.

So what we need to do is say all tests need to be reliable and that the FDA, it seems to me, has a long track record of looking at that. We should say to the Secretary this is a difficult issue because of the constraints and you have to figure out how to get this implemented, and it might mean advocating for more resources to do it. Because you have a flood of new tests that need to be evaluated.

DR. TUCKSON: Jim, let me just ask you. You are very clear. The only question I have for you is, do you feel like if that is what we did -- some people would put the phrase "if that is all that we did" -- that we would have then less than ambitious and thereby slowing the process down by not being more direct?

DR. EVANS: No, on the contrary, I think it is extraordinarily ambitious to say to the Secretary you need to be sure that every test has undergone scrutiny and it may mean figuring out how to get more resources to do that. I don't think it is more ambitious to make more committees.

DR. TUCKSON: So we all know where we are, we have already gotten one set of consensus there. I opened up this door because I wanted to make sure that people were not going to read this report, as we already can tell, as through a filter of whether or not we are saying there should be two different tracks, two different systems for LDTs and IVDMIAs.

Jim, I think you are sort of saying rise above that. I think Paul is saying rise above that debate and simply say, look, it is not important. Just you figure that out. What we are saying with great clarity is this is what the goal has to be.

DR. EVANS: I think there should be a bullet that says this is going to be a bigger and bigger problem. It is already a problem. It is going to be a bigger problem and you need to figure out how the

resources are going to be devoted to this.

DR. TUCKSON: There may be an emerging consensus of that is what you mean to say. If there is somebody who feels strongly that this is not what you want to be associated with, you need to quickly and succinctly tell us that. I see Kevin's hand was up.

DR. FITZGERALD: No, I'm just going to make it happen.

DR. TUCKSON: You are a doer.

DR. MILLER: Rather than just stating the policy and giving it to the Secretary, give the Secretary at least some guidance into how to come out the other end in solving that.

DR. FERREIRA-GONZALEZ: That is what we have here.

DR. TUCKSON: Therefore, let's take the look now at what we have.

DR. FERREIRA-GONZALEZ: Mara and Kevin.

MS. ASPINALL: I would agree with Paul and what Steve and I think Jim said. Making the fundamental statement that all tests, not IVDMIAs but LDTs and IVDs, have to be safe and effective and the FDA is involved, is

a huge step.

DR. TUCKSON: Hold it right there, Mara. Hold it right there. I just want to make sure we get exactly what you are saying. Are you saying in A all --

MS. ASPINALL: No, this isn't even an A. This is really above A. I think it is what everybody was saying.

DR. TUCKSON: Do we want to use the words "all tests, LDTs, and IVDMIAs"? I just wanted to make sure I knew where you were headed.

MS. ASPINALL: With that, in answer to your comment, I think that is a huge step. Many of the stakeholders have said that CLIA alone is enough to have done that. So I want to recognize that this is a statement in a way that is not a small one and is a significant policy. Not all the public comment folks would agree with that because many say CLIA alone today is allowing these tests to be safe and effective.

But what I want to acknowledge, and Andrea mentioned it from my comment yesterday, and my concern and therefore support of two systems is not so much that we need two different systems but we need to acknowledge

that tests are different from devices and that service tests are different from product tests.

I very much agree with that statement that all tests, regardless of what they are, because a patient doesn't know the difference on who is doing it or whether it is on a central lab or a decentralized lab. They don't need to know. That doesn't help them to know.

But what I'm concerned about is taking the square peg and putting it only a few holes that we have available. What I want to make sure, which is consistent with what I said yesterday, even if it means a bigger regulatory environment, is that we have something that is appropriate to both types of tests as they exist now.

DR. TUCKSON: Andrea has given a list of responders. As you respond to that list, you need to focus in on these two pages, A, B, C, and D, and what changes, if any, would you make, so we can bring this to closure.

DR. FERREIRA-GONZALEZ: Kevin, what do you have?

DR. FITZGERALD: Following up on this, we take "high risk" out of A. So we are going to say "all LDTs."

Now, Steve, what do we say should be reviewed, evaluated, addressed? I don't want to get into a technological --

DR. GUTMAN: All tests.

DR. FITZGERALD: So all tests should be what, addressed?

DR. GUTMAN: Yes.

DR. FITZGERALD: "Addressed" is good? Okay. "Addressed by FDA in a manner." "Tests," yes. I'm sorry. "Should be addressed by FDA in a manner that takes advantage of its current experience in evaluating laboratory tests." "This should address." Don't say "review" it or "evaluate" it because that might be too technical.

"This step by HHS entails commitment of significant resources."

DR. FERREIRA-GONZALEZ: Where are you now?

DR. FITZGERALD: Right at the end. Just put that in. New sentence.

DR. FERREIRA-GONZALEZ: Here, above the bullet?

DR. FITZGERALD: I'm sorry. Yes. "This step by HHS will require commitment of significant resources

in order to avoid potential harms," and then we could say "(e.g. patient and public health and stifling technological innovation.)" One of the potential harms is stifling technological innovation. So in other words, this has to be done right. Otherwise you get both bad effects.

DR. FERREIRA-GONZALEZ: Isn't it also stifling innovation but access to the testing?

DR. FITZGERALD: I'm sorry? You can add to the list if you want.

DR. FERREIRA-GONZALEZ: So the public harm will be also access to the test.

DR. FITZGERALD: Yes, access. Sure.

DR. EVANS: I was just going to say, one of the things that I have learned from Reed over the last couple of years is that it is easy to demand that more money be spent. In a way, this is demanding that more money be spent. I think that this is actually a case where it makes sense to demand that.

This may sound like wordsmithing, but because it is a big deal I do think it should be B. We don't want to come across that we are cavalier, that we can

solve all these problems by throwing money at it. Just give them some money. When we do that, I think it should be deliberative and careful and it should not be a throw-away. I think it should be a point.

DR. BILLINGS: Kevin, can you clarify for me how your change picks up what Mara had said just before you about the bifurcation of service and product?

DR. FITZGERALD: My understanding currently is that is one of the areas that we are having difficulty parsing, and the idea of what we are talking about here for LDTs versus IVDs, IVDMIAs, all that sort of thing. By putting in just "tests," I thought we could be broad enough. That was the idea, to get above that particular distinction and say in place we have the possibility of addressing this situation via the FDA.

Now, that doesn't mean that the way things are currently is going to be sufficient do to that, but the template is there. That means you have to have the resources committed in order to step this up.

DR. TUCKSON: So, do we have consensus on this change, other than maybe moving this step to a sub-bullet?

MS. ASPINALL: I think we can deal with that later.

DR. TUCKSON: I think the other modification earlier, from the other discussion, was that we would eliminate "In order to accomplish this recommendation" and we would simply put "The Committee recommends."

DR. FERREIRA-GONZALEZ: Yes, "The Committee recommends."

DR. TUCKSON: So, do we have it? Yes, Mara.

MS. ASPINALL: I don't think Jim meant the same thing, but I think B is absolutely essential, that we address explicitly. I would get rid of "seek to" and I would say eliminate "duplicative oversight procedures." Can we go back? Just the word "seek to" and talk about being specific. None of the labs can exist over duplicative oversight for which you have two things to do that are against each other. You just can't do it.

DR. BILLINGS: There are actually three things. You will have FDA, CLIA, and state.

MS. ASPINALL: Which is even worse. But I would agree. So I would just say it is absolutely essential to get rid of the word "seek to."

DR. TUCKSON: You said should eliminate?

MS. ASPINALL: Eliminate "duplicative," however many there are.

DR. FERREIRA-GONZALEZ: "The multi-stakeholder groups should also explicitly address and eliminate."

MS. ASPINALL: That is good. "Address and eliminate."

DR. TUCKSON: Good. Anybody have any problem with that? I think that is straightforward. Moving quickly. Yes?

DR. KHOURY: "The Committee recommends convening a multi-stakeholder public and private sector group." Is that by HHS, by the Committee, or by FDA?

DR. TUCKSON: HHS. "Recommends that HHS convene a." Good pick-up. Last comment?

MR. DANNENFELSER: Are they going to have the authority to eliminate by themselves or they can only recommend that these duplicative procedures be eliminated?

DR. TUCKSON: To the extent that it is within the purview of HHS. They can't eliminate states.

MR. DANNENFELSER: It says "the multi-

stakeholder group." That is why I'm just wondering. It is kind of the broader outside people and so on.

DR. TUCKSON: Again, the Committee is convened by HHS, so anything within HHS can be eliminated. It can't tell New York what to do, but it can certainly be informed by and try.

DR. TEUTSCH: Yes, the group itself can't actually eliminate it.

DR. FERREIRA-GONZALEZ: They will have to recommend how to.

DR. TUCKSON: Oh, I see what you are saying.

MS. ASPINALL: I think that is a detail. I would rather have the strength of the language that says "eliminate." It is really just New York and Washington, and there are a lot of relationships between there. I would rather have it as "eliminate."

DR. TUCKSON: Good. Let's proceed, please. Let's go to No. C. Anything there? Oh, go back to B.

MS. ASPINALL: There is no C now.

DR. TUCKSON: This, as I remember it, refers to the --

MS. ASPINALL: We don't have that anymore.

DR. TUCKSON: It goes away.

MS. ASPINALL: No, no, no. Just the phrase "for all other tests."

DR. TUCKSON: What is the difference there?

MS. ASPINALL: Yes. I don't think we need this.

DR. TUCKSON: Yes, it is over.

DR. FERREIRA-GONZALEZ: But I think we can also look at "consider existing regulatory models of data sources" also. We can consider new models, too.

DR. TUCKSON: But that is the committee.

DR. FERREIRA-GONZALEZ: No, no, no. Taking a portion of this and putting it back into A.

DR. TUCKSON: What portion would you put back?

DR. FERREIRA-GONZALEZ: I would like to put back that the group should also consider existing regulatory models, data sources, and new models of oversight.

DR. FITZGERALD: Wait, wait, wait. That bullet in A, that sub-bullet, I thought we were going to combine with B. It is all one thing that the multi-stakeholder group does. Otherwise we have two things on the multi-

stakeholder group.

All you have to do, Cathy, is move the B in front of the bullet. That's right.

DR. TUCKSON: So the highlighted part before "responsibility." Right there.

MS. ASPINALL: But add the phrase "and new models." "Existing and new regulatory models."

DR. TUCKSON: Would you cut that and paste it into the earlier one?

MS. ASPINALL: And then put it where Kevin said.

DR. FOMOUS: You want it at the end of here; is that right? No?

MS. ASPINALL: Yes. What Kevin is suggesting is also put the B in front of the bullet point. So these would be the three things the Committee would do.

DR. FERREIRA-GONZALEZ: But we need to add "the current existing regulatory models and data sources and consider also new models."

DR. TUCKSON: So, "existing and new."

DR. FOMOUS: The sentence that is in B about explicitly addressing duplicative oversight procedures,

where do you want that within this bullet paragraph? Do you want this sentence to come at the end of this?

DR. TUCKSON: I thought it was the third bullet.

DR. FERREIRA-GONZALEZ: We will have three bullets, I guess.

DR. TUCKSON: Basically, if you take Jim, you will have four bullets. Three bullets with Jim.

DR. FITZGERALD: Basically, Cathy, B is talking about the multi-stakeholder group. There are three things that the multi-stakeholder group is going to do within B, which actually is going to turn to C once Jim's sentence becomes B.

DR. EVANS: Well put.

DR. FITZGERALD: Thank you.

DR. TUCKSON: This is just now graphing here.

DR. FITZGERALD: We can figure that out later if you want.

DR. TUCKSON: I think we have the sense of it. Again, you have Jim's B about the money and then you have three bullets under C.

DR. FITZGERALD: And we will take care of that

later.

DR. TUCKSON: So, you all got it? Move to the next issue. We didn't do D. You are eliminating C. C goes. Now you are left with D, which is an appropriate transition to start to talk about the registry, which is where we are headed.

DR. FERREIRA-GONZALEZ: Do we also want to talk about or say something that any system that comes up from the deliberation, there has to be some period that allows the laboratory industry and even the IVD manufacturers to step up to the part?

DR. FITZGERALD: No one is going to just immediately expect --

DR. FERREIRA-GONZALEZ: But we have to tell them that we might want to wait for two years before anything would change?

PARTICIPANTS: No.

DR. TUCKSON: No. Now Andrea is going to take us through a discussion on the registry because the last statement on this one is this process is going to be informed by this as yet undefined registry. So we need to go do the registry.

DR. FERREIRA-GONZALEZ: Recommendation No. 3. Recommendation 3 supports a mandatory system of genetic test registration that uses CLIA registration data as the foundation. This recommendation was significantly revised from the draft recommendation, which called for a voluntary system of registration through a public-private partnership.

During the discussions of this recommendation with the taskforce, we had agreement that registration should be mandatory, but the taskforce was split on where such a registry should be housed, either at CMS or FDA.

The public comments also did not offer a clear indication of which agency should house the registry. A few comments articulated a preference for CMS or FDA, but most remained silent on this issue or suggested registration with a government regulatory body or publicly supported website.

Based on the split decision regarding a home for the registry, SACGHS staff explored the issue with our ex officios from CMS and FDA. I would like to pause to thank Judy Yost, Steve Gutman, and Liz Manfield for their patience with our questions and making concerted

efforts to seek answers within their respective agencies.

These discussions led to unanswered questions about the legal authority to gather and publicly display certain data elements. Because of this rather significant development, the steering group modified this recommendation, and this is something that you have here now.

Let me read this recommendation as you only recently received it and may not have time to review it.

Recommendation 3, which is the newest version, states that "There are considerable information gaps about the number and identity of laboratories performing genetic tests and the specific genetic tests being performed. To gain a better understanding of the genetic tests being offered as laboratory-developed tests and to enhance the transparency in this field, SACGHS reviewed proposals for a voluntary or mandatory test registry and considered the benefits and burdens of each type of system.

"The Committee decided that a mandatory, publicly available, Web-based registry that is well staffed to maintain an accurate and database will offer

the best approach to address the information gaps.

"Since genetic tests are not unique from other laboratory tests for oversight purposes, the registry should include all LDTs.

"The Committee also discussed whether such a database should reside at CDC, CMS, or FDA. Based on the exploratory work, SACGHS concludes that the concept of a mandatory registry offers promise but recognizes that there are unresolved issues, including practical and legal questions, that require further analysis before a final decision can be made about how and where to implement the registry.

"So with that preamble, in light of these unresolved issues, SACGHS recommends the following course of action:

"CDC, in collaboration with CMS and FDA, should convene a stakeholder meeting by September 2008 to determine the data elements to be included in the test registry. CDC should cast a wide net for a broad stakeholder representation, including representatives from the private sector who can represent a role for the public-private partnership in developing a registry.

"CDC, through this stakeholder effort, should assess the level of effort as well as the burden on the laboratory and the impact on the other key stakeholders such as patients, physicians, and payers, necessary to obtain each data element, including linking to reliable sources of existing information.

"HHS should perform the requisite legal analysis to determine what data elements, as determined by the CDC stakeholder group, can be required by CDC, CMS, and/or FDA. For example, if clinical validity is a required data element, the legal analysis should determine whether CDC, CMS, or FDA currently have the statutory authority to require reporting of this information for all LDTs.

"If these agencies do not currently have the necessary statutory authority, the legal analysis should identify specific statutory provisions that may be needed in order to effect the system of enhanced reporting requirements and a statutory authority should be sought.

"HHS should appoint and fund a lead agency to develop and maintain the mandatory registry for LDTs. The lead agency should work collaboratively with its

sister agencies to create a comprehensive registry and minimize duplicative collection of registry information. The lead agency should have the qualified personnel who are experienced in developing and updating large databases in a timely and accurate manner.

"While awaiting completion of the above processes, HHS should use short-term voluntary approaches such as incentivizing laboratories to register with Gene Test and encouraging laboratories to make their test menu and clinical validity data for these tests publicly available on laboratory websites."

DR. BILLINGS: Andrea, we just decided, at least in the discussion we had before, that FDA is going to review all tests. So they are going to have information, presumably, on all tests. So aside from Bullet C, which basically says that the lead agency ought to be FDA and that they ought to work with other agencies to make sure that they are reviewing all tests, why do we need this registry? Except to make sure that the public has better access to test information.

DR. FERREIRA-GONZALEZ: I don't know that we are recommending that FDA will review all tests. We are

recommending FDA look at everything but not necessarily actually physically reviewing low-risk and so forth. They might be using different mechanisms for this.

DR. BILLINGS: Well, if they have information on all the tests, why can't --

DR. FERREIRA-GONZALEZ: They might not get all the information for all the tests. Mara, Muin, and Kevin.

MS. ASPINALL: I think, Paul, to your question, we recommended that the FDA in overview has responsibility for all tests but that doesn't necessarily mean pre-market review and it doesn't mean that there is an easy-to-find directory. One of the public speakers mentioned it. You can't get a list of all the CLIA labs today.

I think what we want as we talk about public health and transparency is the ability to get a comprehensive and complete list of tests that are available and what the specifications are. As we heard for Gene Tests from several people, as effective as it is, it is not comprehensive. I think that that, to me, is dangerous because people look at it as if it is

comprehensive and may make certain conclusions based on it.

So I think this works very well with the other recommendation, and we should have the transparency for the public but also physicians to be able to keep up with the rapidly changing and increasing number of tests to say what is there, who does it, what are the basics about these tests. We could debate what is on the registry, but I think this is a very important point that we have a window of opportunity to recommend. It would be a wonderful statement a year from now that we have the ability to look up all tests that exist in the U.S. that are available to physicians and patients.

DR. FERREIRA-GONZALEZ: I think what we have recommended is that the FDA look at all the tests but not that they are physically going to go and review all the tests. That is what the specific recommendation say.

Now, in the meantime, as this process evolves, this registry can provide important information to the consumers. As this registry evolves even faster, it can be developed, but then the FDA can look at what is already in there and necessary to be reviewed or not from

there. Muin.

DR. KHOURY: Can we go back to just the beginning of this? Recommendation A. For those of you that have been at this table for many, many years, my friend Elliott Hillback from Genzyme used to serve on the SACGHT. I have this running joke, Muin and Elliott always saying we need to inform the public and the providers what we know and what we don't know about the genetic tests at any given point in time. It has to be authoritative and updated. We don't have that right now.

I think we have pockets of this through Gene Test, through the SEP process, through EGAPP, and it is not only about clinical validity. It is about the whole package, the whole package from analytic validity to proficiency testing, to clinical validity, to clinical utility. Whatever data are available out there so that providers and the public can make the right decisions about the use of these tests, and also for these things to be reimbursed.

So I view the registry as a vital concept to go through this morass here of trying to pool all the information together.

Now, Recommendation 4 was a breakthrough recommendation because you just asked the FDA to do something, which I think is profound, and you just created a stakeholder group to help with that process. Now, how many stakeholder groups do we want to create between now and September to create the registry?

I would maintain to you that you keep this as a high level recommendation and you task HHS to create the registry but not discuss the data elements. We know what they are: analytic validity, clinical validity, clinical utility. Let's not go through this granularity of saying we need to do A and B and C. Basically, you are asking HHS to implement the concept, and you can give them some guidance.

DR. FERREIRA-GONZALEZ: I still think it is very important to engage the stakeholders. You say we know what the data elements that we need are, but how much data and how much in-depth? That is very important. It goes to the key of how successful it is. So you still need to engage the stakeholders to really see the burden of all this.

DR. KHOURY: I think there is a vital

distinction between what you are saying and what I'm saying. The recommendation is to create the registry. In the process of the creation of the registry, things have to happen, including evaluating authorities, legal authority of collecting data, engaging stakeholders. But, just basically, create the registry. Give it to HHS to implement. The groups, CMS, CDC, NIH, will get together.

DR. FERREIRA-GONZALEZ: What you are saying is add to this saying HHS should create a registry.

DR. KHOURY: Exactly.

DR. FERREIRA-GONZALEZ: Then we can leave CDC in collaboration because that is the process.

DR. KHOURY: Right now, what you are giving us is sort of A, B, and C, which seem to be a bit confusing. HHS should appoint and fund a lead agency. That is No. C. You are putting the cart before the horse. If the lead agency is NIH, then NIH should do the convening function of the data elements. Why should we give CDC something to do before HHS acts on the lead agency?

DR. FERREIRA-GONZALEZ: But if you don't know the data elements, how would you do the legal analysis?

DR. KHOURY: The lead agency, in collaboration with all the others, will figure out the data elements. But you are giving recommendations to HHS to do something.

DR. FERREIRA-GONZALEZ: I think we are okay with saying you have to create a registry.

DR. TEUTSCH: What I'm hearing Muin say is we are creating a registry which contains up-to-date information available to the public on analytic validity, clinical validity, clinical utility, and availability of the tests. That is what I'm hearing. That is what we are asking them to do. Now we just talk about how to get there. But that is the recommendation.

One point of clarification. Since we put "tests" in No. 4, are we just putting "tests" here or are we putting LDTs? "Tests," right? "Tests."

MS. ASPINALL: I would suggest, again, this should be "tests." I just can't say enough that this is a historic moment. It is the only part of the healthcare system we don't have the ability to get everything that exists. So I like what Muin added, and I think this is a key area that addresses lots of the stakeholders and lots

of the public comments of stakeholders to say we want transparency inside the medical community and outside the medical community. This does it.

DR. KHOURY: One more thing. Just like you did with the FDA, you have to put resources behind this. Right now, the creation of the registry involves --

DR. FERREIRA-GONZALEZ: That is what we said in the recommendation. We will fund it, Web-based, accessible. We have some language.

So, the idea is to add a new A.

DR. KHOURY: Switch C to A. "HHS will appoint the lead agency," and then that would be who is doing the convening. If it is NIH, fine. If it is CDC, fine. Or we can keep it going at the HHS level for a while. But don't ask CDC to do something in a vacuum. I think we all want to work together because this is complicated. It is not going to be done by just one agency.

DR. TUCKSON: Andrea, let me just make one comment. You keep driving the train here. Sarah, if staff could tally from everything we have done so far in terms of recommendations to this point and each time we make a new one. We need to put up a slide at break that

shows how many committees we have commissioned and how many of them have money associated with it.

So that, when we come back at the end of our discussion and start to really fine-tune before we close this whole thing off today, we will know whether or not we have committed too many committees and too much money.

Sarah, if you could just [do that.] Please continue the discussion, Andrea, but I just want to make sure we have that list.

DR. FERREIRA-GONZALEZ: Paul Wise.

DR. WISE: I hesitate a little bit to bring this up at this point, but I'm new to the Committee and maybe you could help me a little bit with this process.

I'm increasingly uncomfortable that this is the proper forum to hash out these kinds of issues. I feel that in some ways the time pressure to get this out is forcing a lack of serious consideration of things that may be extremely important in the end.

I could use some guidance. If this is the way it always works and every report goes through this kind of detailed conversation among the Committee at not quite the last moment but somewhat near the last moment, then

fine. But if this isn't typical, then is there a larger question about whether this should go back to the committee for more detailed conversations, talking more with some of the people who represent the different agencies in the government.

DR. TUCKSON: Paul, thank you. It is a great point. I will just give you a little quick history. First of all, inevitably you will find in every report that we do an enormous tension and pressure around time and deadlines and that sort of thing. However, you often have the opportunity to postpone certain decisions if you are feeling an unreadiness.

In this case it is a little different because we have a request from the Secretary to deliver a product by a date. Therefore, we are under a little more unusual pressure in this case. That is why you are feeling it.

Be that as it may, you can imagine that, like most things in nature, it abhors a vacuum. You will fill up every second of every day and you will machinate and gnash teeth on every issue for every report. But this one is a little different because of the timeline.

DR. MILLER: We would love to have you on some

of these subcommittees if you think that we are not having too many meetings.

[Laughter.]

DR. FERREIRA-GONZALEZ: The deadline for the recommendations is February 29, but the document is the end of April.

DR. EVANS: Sweet. The one thing I would second with Paul is that I do feel like we are doing a little bit more in the way of substantive discussion here than we usually do at this stage. I would just make the plea that we make sure that all of us do our jobs when we get the report. If we see things, don't just rubber-stamp it.

DR. FERREIRA-GONZALEZ: You have until February 20th on the report.

DR. TUCKSON: Not on the recommendations. The recommendations we have to have by the end of the day.

DR. FERREIRA-GONZALEZ: Now, the rest of the Committee realized that the steering committee that worked on this document plus the taskforce worked over the summer and the holidays, so I'm not feeling bad asking you to read this. It is all upon all of us to

actually make sure that there is consistency in the language, that inaccuracies are brought up to Cathy, who is the designated keeper of this document, to make sure we all support a product that we are proud of.

Going back to Recommendation 3 on the registry, I'm feeling a bit uncomfortable on keeping the elements right now without really having an evaluation of what it means to say "clinical validity." Do you want a number for clinical validity? We need to be very careful what we are asking here.

DR. WILLIAMS: I'm not hearing Muin say that there isn't a role for convening a group to discuss what elements are needed. What I'm hearing Muin say is that that recommendation which is currently C needs to be A because HHS will make a determination of what the lead agency is and they will task the lead agency then to do the other things we are talking about. That will in fact include evaluation of the elements for what is necessary and then what is practical.

DR. FERREIRA-GONZALEZ: I just heard some elements given out here. That is why I'm bringing it up. I think what we are talking about is Recommendation C

becoming A now, and then in A, we can change "CDC" to "The lead agency, in collaboration with the sister agencies."

DR. WILLIAMS: They will decide what they want to do.

DR. FERREIRA-GONZALEZ: There are data elements that are already being collected. Can we avoid duplicating. That is where I'm trying to go.

So we have now A, "Appoint and fund a lead agency." No. B becomes "The lead agency." I do want the collaboration. Instead of "CDC," "The lead agency."

DR. TEUTSCH: I think we know what the general content is, which were the things that we just talked about, analytic validity, clinical validity. How you measure them and the detailed metrics are not for us to work out and for the agency. We know what the general content of the registry is and probably need to say what that is. Not the detailed elements.

DR. FERREIRA-GONZALEZ: No, I think we still need to be convening the stakeholders to really see the ramification of what that data is, are we going to get that data, what will it take from the laboratories to

obtain that data.

So what we have now as A is "HHS should appoint and fund a lead agency to develop and maintain the mandatory registry." B is "The lead agency, in collaboration with sister agencies, convene a group of stakeholders by September this year to determine the data elements to be included in the test registry."

Second slide. Let's go back.

DR. WILLIAMS: All the "CDC" in that has to change to "lead agency."

To speak to Steve's point, I think we can maybe solve this by saying in that first sentence, "To determine the data elements that address analytic validity, clinical validity, clinical utility," and what is the last? "Accessibility to be included."

DR. FERREIRA-GONZALEZ: Do we really want to tell --

DR. WILLIAMS: Yes, because the report basically says here are the gaps and here is why we need a registry. So yes, we need to specifically articulate that.

DR. FERREIRA-GONZALEZ: I would still argue to

have the stakeholders get together and go over --

DR. WILLIAMS: No, we don't want the stakeholders saying we don't need to collect clinical utility. We are saying we have proven that we do need to collect it.

DR. FERREIRA-GONZALEZ: You are going to have stakeholders, the payers and other ones, there that might say that. So, I don't think so.

DR. KHOURY: I disagree wholly with you, Andrea. I think we need to lay out a little bit more.

DR. TUCKSON: Let's just deal with this, then. I think that there is, again, a clear dichotomy here. Let's just make sure we know where the Committee is. How many of you on this, just so we get a sense of you, think we ought to be very directive in saying that there should be a registry that includes the elements that we have described? How many of you think we need to be absolutely explicit and we say that there should be that, not leave that decision up to a multi-stakeholder group?

Let's start with the registry. The registry itself. There should be a registry and you don't leave that up to "Mother, May I?" with a committee.

[Show of hands.]

DR. TUCKSON: Who feels like there ought to be you leave up the determination of whether there should be a registry to a multi-stakeholder committee?

PARTICIPANTS: No, no, no.

DR. TUCKSON: I'm just asking to get it clear. I just want it piece by piece. We have it clearer than that.

Piece 2. Define the elements now.

DR. WILLIAMS: Basically, what I said was we don't need to drill down on the specifics, but the elements that need to be included have to address analytic validity, clinical validity, clinical utility, and accessibility of data.

DR. TUCKSON: Great. Those of you who feel that those elements should be in the registry, let us know by a show of hands.

[Show of hands.]

DR. TUCKSON: Those of you who feel like you ought not dictate but leave it to some committee, raise their hands.

[Show of hands.]

DR. TUCKSON: So we have a clear sense of the Committee. You have resolved this issue. Move forward.

DR. BILLINGS: Can I ask a question, Reed? Let's say we prescribe the set of elements for the registry and a test doesn't have adequate clinical utility. What does it matter?

DR. TEUTSCH: It is just blank.

DR. BILLINGS: It is blank. Fine.

DR. TUCKSON: Muin?

DR. KHOURY: As part of the EGAPP process, we have been spending a lot of time discussing data elements and breaking down analytic validity, clinical validity, clinical utility, and all of these things into a series of questions. I think by last count we probably had 45 or 50 questions. We don't have to hash them here today, but the broad elements are what can be put out there.

The fact that the test doesn't have clinical utility is not necessarily bad. It could be left blank. But the customer and the providers and the payers need to know.

DR. TUCKSON: So we have that issue resolved. Move on, Andrea.

DR. FERREIRA-GONZALEZ: Yes. So that is B. Now we have A, "Appoint a lead agency." Now we have C, which is in the meantime. Any comments or edits to this C?

[No response.]

DR. FERREIRA-GONZALEZ: Can we move forward to the next recommendation?

DR. TEUTSCH: We are already a half hour beyond our break time. Why don't we take a 15-minute break. First of all, is everybody in agreement with these recommendations? Okay. So, why don't we take a 15-minute break. Plan to come back at five minutes after the hour.

DR. FERREIRA-GONZALEZ: What do we have; Nos. 5 and 6?

DR. TEUTSCH: We have Nos. 5 and 6, and then of course we have to go back and review the overarching, and we need to go back to revisit some of the ones we did not finalize.

[Break.]

DR. TEUTSCH: Folks, let's reconvene. Cathy, did you have a response to a query from Reed? I'm not

even sure which query it is.

DR. FOMOUS: Yes, I do. Reed asked how many of our recommendations are calling for public-private partnerships or stakeholder groups as part of the recommendation. So in direct answer to Reed's question, there are three. There are our revised Recommendations 3 and 4 in Chapter 4 that call for these multi-stakeholder groups. They theoretically could be the same group of people. Then Recommendation 1 in Chapter 5, asking for a multi-stakeholder group to look at clinical utility issues.

Now, in addition, there are a couple of recommendations that suggest that relevant agencies might want to engage a group of stakeholders just for additional information. That kind of connoted less permanence.

DR. TUCKSON: Thank you. Let's keep track of that as the conversation goes forward. My suggestion is that when we get to the end, before we finalize all of our vote, we take one more look at exactly how much bucks we spent and how many committees we convened and see whether that affects any decisions.

DR. WILLIAMS: May I ask a question related to that? Is there a threshold over which we say that this is not important and therefore we shouldn't make the recommendation?

DR. TUCKSON: I don't know. I have no idea. What it may do, also, is say certain things may need to be combined. I don't know. I have no idea what it is going to look like.

DR. FERREIRA-GONZALEZ: Why don't we keep plowing through and then we will get to that point.

DR. TEUTSCH: Yes, I'm looking forward to that moment. So, Andrea and Reed, walk us through the last of these.

DR. FERREIRA-GONZALEZ: Recommendation 3 for the registry, we are going to add the wording of "least burdensome."

So Recommendation 5 requests enforcement of existing regulations. We revised Part A to include that laboratories without CLIA certificates cannot be reimbursed by Medicare and Medicaid but these restrictions have no consequences for laboratories that perform direct-to-consumer testing.

We did not make any changes to Part B of the recommendation.

Do you have any questions about this recommendation?

[No response.]

DR. FERREIRA-GONZALEZ: That is refreshing. Do you have any edits?

DR. EVANS: Yes, I'm sorry.

DR. FERREIRA-GONZALEZ: Jim.

[Laughter.]

DR. EVANS: I'm just wondering if we want to make it a little bit stronger. It seems awfully tepid, saying they should explore mechanisms, develop new authorities and resources. Say they should find ways to close that loophole.

DR. FERREIRA-GONZALEZ: So, what language would you recommend? If you are going to speak up, you have to come up with some.

DR. EVANS: How about just forget "explore mechanisms and seek." Just "should develop the authority" or "implement its authority in order to effectively enforce."

DR. FERREIRA-GONZALEZ: But they might not have the authority, so they need to seek.

DR. EVANS: "Should develop the authority" or "attain the authority" or just something a little stronger.

DR. FERREIRA-GONZALEZ: Judy is not here, so.

DR. EVANS: "Should develop the authority." How about "develop the authority"?

DR. TEUTSCH: Jeff, do you happen to know what the authority is?

DR. ROCHE: No, I don't.

DR. TEUTSCH: How about just saying "should strengthen its enforcement efforts against laboratories."

DR. FERREIRA-GONZALEZ: Whatever they have to do, seek authority or not, then they will do it.

DR. EVANS: There you go.

DR. FERREIRA-GONZALEZ: Repeat that again, Steve?

DR. TEUTSCH: "HHS should strengthen its enforcement efforts against laboratories." You just delete everything from "explore" down to "to."

DR. FERREIRA-GONZALEZ: There we go. Very

good. Can we move to the next recommendation?

DR. TUCKSON: I just want to make sure. Until they step up, does this leave a hole?

DR. FERREIRA-GONZALEZ: Yes.

DR. TEUTSCH: Yes.

DR. TUCKSON: I just want to be even more explicit. We need to make sure that everybody who reads this understands that you have to close the hole. This is like saying they should step up, we hope that. I think you have to say the explicit intent here is to close the hole.

DR. EVANS: "Should close this gap."

DR. TUCKSON: And that the only way you can do that, we are saying here, is --

DR. FERREIRA-GONZALEZ: But, isn't the first sentence saying that?

DR. TUCKSON: It says "Further efforts are needed to prevent laboratories." I guess that says close the hole.

DR. FERREIRA-GONZALEZ: Actually, it should be CMS.

DR. TUCKSON: The question is, you can't impose

them on uncertified laboratories. You can't impose it on an uncertified lab. Remind me what the uncertified lab is, again?

DR. FERREIRA-GONZALEZ: A laboratory that doesn't have a CLIA certificate. Today the problem is that when they go to inspect the laboratory or they have come upon a laboratory and they don't have a CLIA certificate, they cannot close down the laboratory.

DR. TUCKSON: That is kind of crazy. It doesn't make sense to the average person. In other words, I inspect you, you don't qualify, I can't inspect you. That can't be it. That is what it sounds like. Because you are so bad, I can't do anything to you.

DR. TEUTSCH: They have to be referred to the inspector general for enforcement, is the problem.

DR. FERREIRA-GONZALEZ: Yes. Then the inspector general at that point intervenes. So we are asking them to have something direct. If they come across a laboratory and they don't have a CLIA certificate and they are doing clinical laboratory testing, that they can do something to that laboratory right there.

DR. TUCKSON: That is what this is saying, that they can now stop you.

DR. FERREIRA-GONZALEZ: Without having to go through another.

DR. TUCKSON: Enforcement action. So "CMS should strengthen its enforcement action against" -- no. "CMS should have enforcement action against."

DR. TEUTSCH: "Should obtain and strengthen," is that the issue?

DR. KHOURY: A quick question. Are we conflating DTC with no CLIA certification?

DR. FERREIRA-GONZALEZ: No, no, no. We are talking about laboratories --

DR. KHOURY: The section in green. If you read it again, "Labs without CLIA certification cannot be reimbursed, but this restriction has no consequences." Can you explain that again?

DR. FOMOUS: The first part of this sentence would have consequence for labs that are actually performing clinical testing that is related to health care or tying into results that patients need. There would be a consequence for that. But for a lot of these

DTC companies that we are concerned about, they don't care about reimbursement because the consumer is paying them directly.

DR. KHOURY: If a DTC company has CLIA certification, what will happen?

DR. FERREIRA-GONZALEZ: If a DTC laboratory has CLIA certification? Nothing. They have CLIA certification. They are okay. If it is a laboratory, DTC or not, that doesn't have a CLIA certificate --

DR. KHOURY: Some of them do.

DR. FERREIRA-GONZALEZ: Even if you do have a CLIA certificate, then you are not covered in here.

DR. WILLIAMS: We shouldn't confuse the two issues. We have addressed in other recommendations the fact that some of the tests that are being done in CLIA-certified labs don't have validity and utility. So we are trying to address that in a different area.

This is specifically addressing those laboratories that do not have CLIA certification that are performing tests that we consider to be health-related and should fall under CLIA. The only enforcement ability that CMS currently has is to not reimburse. We are

seeking enforcement ability beyond that, which is to say you must cease and desist.

DR. FERREIRA-GONZALEZ: Steve.

DR. GUTMAN: I'm sorry that Judy is not here, but I'm sure that is wrong. It is illegal in this country to offer a lab result for a medical purpose on any person in a non-CLIA-certified lab. I don't know what her tools are.

DR. FERREIRA-GONZALEZ: We understand that it is illegal.

DR. GUTMAN: Now, they may not be enforcing them as enthusiastically as you would like, but --

DR. FERREIRA-GONZALEZ: But it is not only enforcing, it is the way that she currently has the tools to enforce are going through a different mechanism. There is nothing very direct right there to do something about that.

DR. GUTMAN: I would argue it is very direct. I don't want to pick any names, but if you look at the companies that were described at the Smith hearing, many of them were non-CLIA-conformant before. They are all CLIA-conformant now. I think she has more tools than you

understand.

DR. FERREIRA-GONZALEZ: She was okay with this recommendation.

DR. GUTMAN: I'm sorry she is not [here.]

DR. TEUTSCH: Judy, are you on the phone, by any chance?

[No response.]

DR. GUTMAN: I think we should call her and ask her about this. Unless she is wanting encouragement to use those tools more liberally, I'm certain she has them.

DR. FERREIRA-GONZALEZ: She has seen these recommendations and we haven't heard anything specific from her.

DR. GUTMAN: She is the CLIA expert, not me, so I will shut up.

MS. CARR: She has been on the phone on and off this morning, and I think she is en route back to her office. I think she will be on shortly. I just Emailed her to see. We will try to get her. But she did review this recommendation. She actually provided us input that helped us clarify the role the inspector general has.

DR. FERREIRA-GONZALEZ: Yes, that is how we

learned about the inspector general, that route, from her.

DR. GUTMAN: Again, she is the expert, I'm not. But I do know she has tools. So her motive must have been perhaps giving her more --

DR. FERREIRA-GONZALEZ: It is more direct.

DR. FOMOUS: So, did we want to change the wording here?

DR. FERREIRA-GONZALEZ: Yes. Reed, what do you want to change here? Is Reed there?

DR. TUCKSON: I don't like this "further," but anyway, that is wordsmithing. First of all, what I would like you to consider is taking "further efforts are needed" and saying "to prevent laboratories from performing genetic tests without appropriate CLIA certification," here is what you should do. I think that is what it needs to be. You just go straight to the heart.

So, "To prevent laboratories from performing genetic tests without appropriate CLIA certification, the Committee makes the following recommendations." I guess that is repeating, maybe, the preamble. But anyway,

without getting into that.

So then you need to say that "The CLIA program has an array of enforcement actions available but those actions cannot be imposed."

DR. TEUTSCH: Judy, are you on the phone yet?

MS. YOST: Hello. I just got to my office.

DR. TEUTSCH: Oh, Judy, good. You are very timely. We are looking at Recommendation 5 in Chapter 4. Steve raised the issue regarding your comment on the need for CMS to secure additional enforcement activities against labs which are not CLIA-certified. We believe you reviewed this recommendation, but are there issues here that you need to raise about whether you already have those authorities?

DR. TUCKSON: Judy, this is Reed. I'm the one that is struggling with it, and Steve has introduced it well. What we are specifically trying to get at here is that the impression that we are left with from the original recommendation is that you do not have the authority to specifically regulate non-CLIA-certified labs and, as a result, the only power you have is to go to the inspector general to have that person's office

fill the gap.

We are trying to understand that because it seems to us that it doesn't make any sense, that what we ought to basically say is you should have the authority to regulate the labs, whether they are CLIA-certified or not.

MS. YOST: The legal answer is yes, that is essentially what the regulations provide for. They provide that if we have to take an action against a laboratory that does not have a certificate, in that case it would have to be referred to the OIG. However, we have put into place mechanisms for those circumstances when we come across them and have dealt with them rather successfully.

That doesn't say that we probably maybe need some more, but we have been able to deal with them fairly successfully by sending them a rather unfriendly letter that says that they must cease and desist their testing because they are operating outside of federal law. In most cases, that is rather effective. We do not allow them to initiate testing again unless they apply and their application is approved to begin testing again.

That has worked.

So it is not a total black-and-white answer.

DR. TUCKSON: That is helpful. That is very helpful. In terms of moving this forward, if we were to just take out the ambiguity of all of this and the jerry-rigging of it and scaring people, what if we were to simply make logical sense and recommend that CMS ought to have the authority within its body unambiguously to perform this function. Is that something that has to be done through statutory change?

MS. YOST: I would assume you could do it through regulation.

DR. TUCKSON: Through regulation?

MS. YOST: We can throw it in the PT regulation.

DR. TUCKSON: I know you have the awkward position of not being able to actually write it, but we are asking you specifically so that we don't have confusion. We would recommend that the Secretary take steps to seek -- who writes the regulation?

MS. YOST: CMS and CDC write the regulations. For enforcement, CMS would do it.

DR. TUCKSON: So, can you write this sentence for me? If we said that the Committee recommends that the Secretary of Health cause the following agencies to write the regulations that will permit CMS to regulate non-CLIA-certified labs.

DR. TEUTSCH: "Should secure the regulatory authority."

DR. TUCKSON: From where? "Secure the regulatory authority from"? Congress?

MS. CARR: It doesn't need statutory.

DR. FERREIRA-GONZALEZ: It is not statutory.

DR. TUCKSON: So, where do they get it?

MS. CARR: Themselves.

DR. WILLIAMS: They write a Federal Register announcement and get comments.

DR. TUCKSON: So they get it from themselves?

DR. WILLIAMS: Right. So they should assume regulatory authority.

DR. TUCKSON: So there is the language. Judy, would you be okay with that?

MS. YOST: Yes, that's fine. That is why I didn't say anything else with what was there, because I

had a feeling that that would happen anyway. But I did want to explain that we do have some and it has worked. In fact, for the illustrious folks we had on the phone yesterday, the laboratory that they are currently using just underwent that process successfully.

DR. TEUTSCH: It should say that "CMS should assume the regulatory authority to allow it to take enforcement actions against laboratories that perform."

Any other comments on this?

[No response.]

DR. FERREIRA-GONZALEZ: So we have the language. Do you have it, Cathy?

[Pause.]

DR. TEUTSCH: "Regulatory authority to allow it to take enforcement actions against laboratories."

DR. FERREIRA-GONZALEZ: So we have to put "to allow it to take." No, don't take the "enforcement."

[Pause.]

DR. TEUTSCH: It already has authority.

DR. FERREIRA-GONZALEZ: They already have authority.

DR. TEUTSCH: What are you asking it to do is

be able to enforce those actions directly itself as opposed to referring it to a third party.

DR. FERREIRA-GONZALEZ: Hold on, hold on. "Should exercise its regulatory authority to allow."

[Pause.]

MS. YOST: This is Judy again. I wanted to mention, too, because I don't know where you are in the process of this because I kept getting disconnected from the telephone. I don't know if you had any further discussion about the DTC labs, which is kind of related here. I know some of the concerns about those labs --

DR. FERREIRA-GONZALEZ: It is coming up next, Judy.

DR. TEUTSCH: It is the next item.

MS. YOST: Oh, okay. I will be quiet.

DR. TEUTSCH: Anything else here?

DR. FERREIRA-GONZALEZ: Yes. Anything else that we want to add to Recommendation No. 5?

[No response.]

DR. FERREIRA-GONZALEZ: Let's move to Recommendation 6. Now, remember we had some issues in Chapter 6, Recommendation 5 on how we were going to tie

it up with this one. So let's get back to Recommendation 6.

Recommendation 6 calls for the expanding CLIA regulations for CMS's statutory authority through CLIA to encompass certain direct-to-consumer tests that appear to fall outside CLIA's scope. We revised this recommendation to include FDA's authority and regulatory process.

Do you have any questions about this recommendation? Judy, you made some comment about this. Is there anything you want to add to this recommendation?

MS. YOST: I actually was just going to talk about the DTC labs because I know there is a lot of concern about the ones who are advertising over the Internet and whether or not they are CLIA-certified.

I just wanted to let the folks know that we have taken it upon ourselves and we are collaborating with both CDC and FDA that when a laboratory like this is identified that we will follow up to investigate what, if any, type of testing the laboratory is performing. For those that are within the current scope of CLIA, that

they do obtain a CLIA certificate.

Those efforts have been rather successful, to the point where we currently have 64 laboratories identified that we have reviewed and not only do we just do an initial investigation, we do continuous follow-up until we are satisfied that they are not only enrolled but in compliance.

So I just thought you should know that we haven't been sitting around.

DR. TEUTSCH: That is very helpful, Judy. This is really talking about expanding the scope.

MS. YOST: Right. I realize that. But I did want to throw that in there because I think that people think we are just ignoring that, and we are not. We are very well aware of it and we are going to try and set up something more formal to accommodate that.

With regard to scope, we have had discussions with our attorneys and clearly, right now at least, in order to come under the purview of CLIA an entity would have to meet the current statutory definition of a laboratory. So that is what our limitations are currently.

DR. FERREIRA-GONZALEZ: Any comments? Marc.

DR. WILLIAMS: The only point I would make is that it seems now, as we look at this, that Recommendation 5-B would fit better as part of this recommendation because this deals with issues of claims which will be directly impacting the determination of health-related. So I just think moving 5-B and incorporating it No. 6 would make it clear.

DR. FERREIRA-GONZALEZ: Yes, I think it is a good idea.

DR. TEUTSCH: The one before isn't limited just to these DTC, whereas Recommendation 6 is.

DR. WILLIAMS: All right. I withdraw my [suggestion.]

DR. FERREIRA-GONZALEZ: We don't have any comments. Any edits to this one?

[No response.]

DR. TEUTSCH: We have been through them all once now, correct?

DR. FERREIRA-GONZALEZ: Yes, yes. Well, we still have one more. Sorry to tell you. We have an overarching recommendation.

DR. TEUTSCH: Oh, correct.

DR. FERREIRA-GONZALEZ: So we have an overarching recommendation that outlines steps to enhance interagency coordination for oversight activities. Do we have any comments? Yes.

DR. WISE: I'm concerned that this is pretty weak. It doesn't speak to the concerns that Reed has been raising straight through all these conversations.

Right now it reads sort of as an insider's report, which is totally understandable given the technical complexities and what has to be dealt with. But genetic testing is also an issue of great public concern and there has to be a framing, I think.

We may want to take advantage of the overarching recommendations to help frame the public presence of this report in language to say genetic testing is expanding greatly, however oversight of genetic testing currently is inadequate. Therefore, the issues we have identified looking at the gaps, there are jurisdictional problems that would relate to coordination, there are authority issues, there is quality control, and then there is dissemination of

appropriate innovation to the people who need it. All the gaps fit into those categories.

So, is there a way to use either the overarching recommendations or something up front that would help frame the public presence of this report in a way that translates the technical conversations and the technical language that is in there into something that makes this more accessible and controls the public presence more than the way it is written now.

DR. FERREIRA-GONZALEZ: So, what would you recommend?

DR. WISE: If people are happy with the idea that in fact the report needs to do this, to try to accomplish this, then my suggestion would be to not elevate what we need just to get along better and then elevate the other themes that have come through, one including better coordination, but the issue of filling major gaps in regulatory control.

There are sentences in here, but to elevate, to use this mechanism to elevate the critical positions, the legitimacy, the justification for this report in a way that is accessible. If this isn't the best way, then I'm

happy to try to do it a different way.

DR. TEUTSCH: Paul, you can see it in the executive summary. Between the header, "Recommendations," there is some text that is sort of the preamble to all of them. What I'm hearing you say is you are not thinking just to this, you are speaking to all this set of recommendations.

I wonder if we can't craft some language that will sit in that space. These are the issues we have found and the recommendations below speak to them.

DR. TUCKSON: Steve, one thing I would speak to is I do like where Paul is going and your comment. The word that is missing in all of this is the word "accountability."

To me, I think that that is the overarching recommendation. It is saying that you take all the things Paul said and, at the end of the day, the recommendation is that the Secretary must use all of his or her power in terms of the agencies reporting to them and is accountable for protecting the public in this regard. Then the recommendations that come down the pike start to get more specific about those things, but the

word "accountability" is where I see the overarching.

DR. FERREIRA-GONZALEZ: That is a different issue. the issue that we have here, too, is that there might be different activities happening out of the Secretary's office that might be duplicative or somewhat of an overlap. That is what we are trying to get here, that the Secretary needed to have a better coordination of the different activities that are currently happening under his office.

DR. WISE: The issue is, should that be the overarching recommendation, the one that is elevated above all others. There has not been a report about the federal government that has not included this issue, ever. My concern is, given the importance of this issue, that we need a framing recommendation that truly gets to the heart of why this is so important.

I agree; I think the accountability may be the overarching recommendation and that the preamble and the executive summary or that text that is there now can be reworked to be more focused, more clear, and to state that action steps are going to be required to optimize the benefits of this new technology but also to prevent

the harm that also is potential outcomes.

To take the gap analysis that is very technical, and that is required to be technical, and to reframe it as the three or four big ticket arenas of action that are going to be required. It doesn't have to be the overarching recommendation. I like Reed's. But to have something right up front that frames this report and that sets the foundation for the interpretation of the technical language that ensues.

DR. FERREIRA-GONZALEZ: Muin.

DR. KHOURY: I want to second what Paul was saying here. I think that you can beef up the preamble and all the background, but given how the Committee has worked so hard to identify this monster here plus all the gaps, et cetera, the recommendation to the Secretary to say "Take steps to enhance interagency coordination" is rather weak.

I have been in other committees where, at the end, an interagency working group was created, for example. Even that was weak because it got disintegrated over time.

But you have to tell something to the Secretary

that is a little bit more substantive than just taking steps to enhance interagency coordination. That is what they are doing all the time. What kind of steps do you want them to take.

One approach is to create a working group that would oversee the implementation of the recommendations or do all this in part of the Personalized Healthcare Initiative, which genetics fits nicely under.

It will obviously be left to the next administration to implement, but you need to send a stronger signal than just "take steps to enhance interagency coordination." Just different words.

DR. FERREIRA-GONZALEZ: Joseph and Marc.

DR. TELFAIR: This is just a question related to this. As a public health person, one thing that we look at a lot is who are our target populations and who we deal with. I think "the interagency coordination for the purpose of reporting to," and then whoever we are deciding who the target population needs to be accountable to, be it providers, be it the public, or whatever, as a strong statement coming out of an overarching recommendation.

If you look at everything else that has been said over all these recommendations, you have parceled out who these target populations are and who should be accountable or moving towards.

I would just say it should start off with an outcome statement right off the bat that is pretty strongly stated and includes what it is you are trying to do. The Committee itself can look at what they are recommending, but it should be for the purpose or for the expectation that whoever, the public, the providers, the other persons who are constituents, will be able to enact or be able to be involved with this group.

Something in that neck of the woods seems to make sense. I think you start off with a very strong outcome statement to move forward with that.

DR. FERREIRA-GONZALEZ: Marc.

DR. WILLIAMS: Personally, I think if we could capture what Reed said and substitute that as an overarching recommendation for what we have here. As I read through this again, most of it is represented in the other sub-recommendations.

DR. FERREIRA-GONZALEZ: This was also speaking

to some issues that we started identifying, that within the Office of the Secretary there are different agencies or groups that are working on similar issues at the same time.

DR. WILLIAMS: I recognize that, but I think we reflect that in all of our recommendations because we have the same alphabet soup that is appearing in all of them. I think, to be very clear, the issue is protection of the public and gaps must be closed. That should be the overarching recommendation, and the rest of it is going to fall out.

DR. FERREIRA-GONZALEZ: Jim.

DR. EVANS: I agree exactly with what Paul and Marc have said. I think that this is all about the fact that gaps exist, harm could result from those gaps, the gaps have to be closed. The rest of the recommendations all pertain to ways that we recommend to close those gaps.

But I think an overt statement at the start along those lines would strengthen the report immensely.

DR. FERREIRA-GONZALEZ: Kevin and then Reed.

DR. FITZGERALD: If we just take what we have

at the beginning there, which I think is still a nice little setup, there is this complex oversight system, many dedicated people. Hold on, wait a minute. Then say, "Nonetheless, the Committee also found significant gaps in the system that could and do lead to harm. Therefore, the Committee recommends," or we can put it that way. "The Committee states that the Secretary of Health and Human Services should take accountability for addressing these issues. We have, in the following, put forward some specific recommendations," but ultimately it all ends up in the Secretary's lap. Period.

DR. FERREIRA-GONZALEZ: Julio?

DR. LICINO: What is the actual harm that has occurred as a result of the current system? If there is, we should document it. If there isn't, we should say "potential." But I'm not aware myself of actual harm to anybody due to the current system. I may be missing something.

DR. TUCKSON: Folks may want to start thinking in their mind whether the word is "potential" or "actual" or do we know enough to know. Because you don't know what you don't know. I think the stuff that we have been

hearing around the direct-to-consumer stuff where you just have no idea what pop-ups are going where; you just don't know. It may be that we have some documentation that we want to bring forward or we just want to say "potential."

Be that as it may, I think I liked everything that Kevin said to advance the ball. The only modification, and I think it is not even really much difference, it is very much informed by Muin showing the map. It sounded like I was hearing Kevin say the Secretary is accountable for getting these recommendations done.

I think what I'm saying is, look, the Committee has done its very best by providing a set of recommendations. It is almost to Paul Wise's point. We have done the best we can under the time constraints that we had. And I don't want to diminish our recommendations by doing that.

We have given a variety of recommendations which we think move it forward. At the end of the day, this is complicated. The Secretary ultimately is accountable for making this complex puzzle make sense to

protect the public. At the end of the day, that is where this falls. People should not be hoping, praying, trusting that we have it all. At the end of the day, Mr. Secretary, you have a bunch of people who are very smart and very dedicated. You have to make this happen.

DR. WILLIAMS: The point I would just make about the harms is that the text of the report clearly identifies those harms for which there is literature, support, and clearly identifies those harms that are potential or plausible but for which there is no documentation. I don't think we need to revisit that.

DR. FERREIRA-GONZALEZ: No. So we need to start working on the language.

DR. TUCKSON: One last question I have before Andrea brings us to whatever our summary is. I think we heard that we didn't add any more committees and any more money, so I think we just need to take one more look at that summary again.

But also, did we resolve Mara's point? I'm not sure whether we answered what is in and what is out as the definition of a genetic test. Did we resolve that?

DR. FERREIRA-GONZALEZ: Don't go there.

DR. TUCKSON: Don't go there? I can leave it alone? It's too late? We are all right? All right. There is a strong consensus to not raise that. I just wanted to make sure we weren't forgetting something.

So, give us the numbers again. How many committees did we create?

DR. FERREIRA-GONZALEZ: Have we finished with the overarching recommendations? I don't see him writing.

DR. TUCKSON: So, what was the amount of money and the committees again?

DR. TEUTSCH: Three committees.

DR. TUCKSON: Three committees. What were those again?

DR. TEUTSCH: Registry, utility, and --

DR. FOMOUS: FDA review and registry, which could be one and the same, and then one for utility.

DR. TUCKSON: That is why I wanted you to slow down. You said they could be one and the same. Let's go back and understand that.

DR. FOMOUS: Registry and FDA review.

DR. TUCKSON: So registry and FDA could be one

and the same. Now, this may be beyond your neuronal capacity at this point, since we have dangling participles here, Marc. But, is anybody prepared to think about can we put those two together and is that going to be harmful?

I really, obviously, am trying to get us to where you don't have three, if you can get two, because it just gets to be a god-awful nightmare trying to administer this stuff.

PARTICIPANT: To me that seems to be micro managing. We don't need to specifically articulate that they have to be separate committees, and I think we could leave that to the Secretary to decide what is the best way to do that.

DR. TUCKSON: I'm suggesting you take what Marc said and capture it in the letter of transmittal from the Committee. What I don't want to have happen is a loyal staffer to the Secretary walks in and says, "Secretary, I want to brief you on the Committee's report. They are asking you to create a massive new government infrastructure, three committees with 50 people on them. Public-private people from all over the world have to be

convened twice. The costs for travel are going to be, blah, blah, blah. Somebody has to staff it," and the Secretary and these people are out of their minds.

So if you can get away with limiting this to two, that would be terrific. But if you are saying that is micro managing, then, Marc, maybe the sense of the Committee is, Mr. Secretary, we think that you have discretion on how you administer this and we are sensitive to the cost effectiveness of what we are proposing.

DR. FERREIRA-GONZALEZ: Look at the map and the gaps. It might not be feasible to have a single group looking at all this.

But we still have to go back to Chapter 6, Recommendation 5.

DR. TEUTSCH: Are we done with this?

DR. FERREIRA-GONZALEZ: No, they are wordsmithing.

DR. WILLIAMS: Actually, Andrea, for Chapter 6, Recommendation 5, I believe that as we have rewritten Chapter 4, 5-B and 6, that what was left in Recommendation 5 in Chapter 6 is now completely

redundant. I think it is captured in Chapter 4, Recommendations 5-B and 6. So I think we should just take it out.

Now, that is not my decision to make, but that is the conclusion that I come to.

DR. FERREIRA-GONZALEZ: Why don't we take it out as we go through it again.

DR. WILLIAMS: That is what I'm saying. Let's not visit it now.

[Pause.]

DR. WILLIAMS: So, start with "The Committee."

DR. FOMOUS: We are not going to use any of this?

DR. WILLIAMS: Yes, we will be, but it is moving around too much. Ready? "The Committee found significant gaps in the U.S. system of oversight of genetic testing that could and do lead to harms." Julio, we can talk about this, but we do have data that say there are harms.

"The Committee formulated a number of recommendations that, if implemented and sufficiently supported, could close these gaps. The Secretary of HHS

must take responsibility for closing these gaps and fostering the public health."

That's it. They wanted something simple, direct, and overarching.

"Public's health"? That would be all right.

[Pause.]

DR. FERREIRA-GONZALEZ: I'm still confused about this. That is why we are giving him the report. He must do.

DR. MILLER: I would just change that second --

DR. FERREIRA-GONZALEZ: Remember we are an advisory group.

DR. WILLIAMS: And we are advising him.

DR. FERREIRA-GONZALEZ: Yes, all these recommendations.

DR. MILLER: I would just say "The Secretary of HHS is responsible for closing these gaps." Because anybody can say "I'm taking responsibility for everything." Secretary Leavitt, close these gaps.

[Laughter.]

DR. TEUTSCH: How about just saying "should close these gaps"?

DR. MILLER: No. He is responsible for closing the gaps.

DR. LICINO: Can't we just say "closing these gaps" --

DR. FERREIRA-GONZALEZ: Julio, turn over your speaker. We are taping.

DR. LICINO: Can't we just say that closing these gaps is in the public health interest"?

PARTICIPANTS: No.

DR. FOMOUS: Can we just alter the end of it a little bit to say --

DR. MILLER: Please.

[Laughter.]

DR. FOMOUS: Please, because I think it will make it even stronger -- "is responsible for closing these gaps to foster" or "to optimize the public's health"?

DR. FITZGERALD: I guess the idea there was one can talk about closing the gaps but there is also the possibility of going beyond just closing gaps. One can, once the gaps are closed, still continue to work to foster the good that can come from these technologies.

So if you limit it to just closing the gaps, then the work is done. But I think some of what we have been talking about is the fact that there is more to do than just closing gaps.

DR. FROHBOESE: That gets at the main point, that there are gaps, here is what we recommend, and not only is it just about the gaps but the big picture is the public's health. That is the big picture.

DR. FERREIRA-GONZALEZ: This is an overarching recommendation, but we still are going to have another recommendation as part of this one to talk about the coordination?

DR. TEUTSCH: No.

DR. LICINO: Just one thing. I'm a little troubled by this. He convenes an advisory committee who says "You have to do this." I would say that it is our advice to the Secretary of HHS that these gaps be closed to foster the public's health," because that is what we are supposed to do, give advice. We are not supposed to tell him, "It is your job to do this." It is very strange. I feel kind of strange about this.

DR. MILLER: I would say that he has convened

this panel of experts to look at a particular issue. I don't think we are saying anything particularly radical here by saying that the HHS Secretary is responsible for closing these gaps or responsible for his agency. We are laying it on his desk to say here is what we think you need to do.

DR. TELFAIR: There are two things. First of all, it is an expectation that it is his responsibility. It is an expectation. If we read through pretty much all the comments, particularly the public comments, there is an expectation listed here.

Second of all, by enforcing an after-the-outcomes effort, which is what we are talking about when you look at the "and fostering the public's health," is adding teeth to a lot of the things that are going to come after it with this set of recommendations. That is pretty clear what we are asking to be done.

DR. FITZGERALD: If we are worried, we could always say, "And if you have any questions about this, see Reed Tuckson."

DR. FERREIRA-GONZALEZ: Martin.

MR. DANNENFELSER: How about a sentence like,

replacing that last sentence, "This action is consistent with the Secretary's responsibility for fostering the public's health"?

DR. MILLER: With all due respect, let's just say what we mean. The Secretary has prompted this Committee to go through extraordinary efforts to get him a document because he wants something before he leaves. That is where we are. We are just saying, you asked us a series of very important questions, we found some very significant problems, we came up with our best efforts to think about it. It is now your responsibility to do something.

I don't think that that is overreaching. I don't think it is impolite. I just think it is appropriate.

DR. FERREIRA-GONZALEZ: We are done.

DR. TEUTSCH: I want to go back to one thing Marc said. I'm okay with removing the one recommendation, but we need to make sure that in Chapter 4 where we talked about the oversight of marketing and other such things that we are explicit that that should include the DTC and tests as well.

DR. WILLIAMS: Yes. That just is report modification.

DR. TEUTSCH: Right. That is the editorial kind of thing that we can take care of.

DR. FERREIRA-GONZALEZ: Yes, we have been through everything. Do we want to go through all the recommendations?

DR. TEUTSCH: Why don't we do this. Why don't we read through the recommendations one time now. Are there other things that are missing that are not here? This is our last chance to add new recommendations.

[No response.]

DR. TEUTSCH: Do you want to go ahead and vote now and walk through it? Sarah, what is your advice on how we proceed?

MS. CARR: If the Committee will consider it, I think you ought to try come to a consensus now on the body of your work here. Final recommendations and that the draft report is in spirit ready to be sent to the Secretary after you have an opportunity by February 20th to provide some additional comments. No additional input on the recommendations.

DR. TEUTSCH: So, do we have them so that we can read through all of the recommendations?

DR. FERREIRA-GONZALEZ: Just a reminder on the voting. Questions to consider in voting for the recommendations. Are these recommendations the optimal way to address the opportunities and challenges identified in the report, and are these the recommendations that the secretary of SACGHS should make to the Secretary. Those two we have to keep in mind.

Now we are going to go in order, actually. Just let's go. Just go through it.

Chapter 6, Recommendation 1.

DR. TEUTSCH: Let's go through it in the right order this time.

[Pause.]

DR. WILLIAMS: Mr. Chair? Dr. Chair? It seems to make sense at this point, since we really have significant organizational things, that we should take a lunch break, reorder the recommendations so that we can go through them in order.

DR. FERREIRA-GONZALEZ: What time is Reed leaving? Reed is going to be leaving at one o'clock.

MS. CARR: Do you feel like you can't just go through them?

DR. WILLIAMS: I'm just looking at recommendations that are right now all over the place. It just seems to me we will lose a lot of time trying to find which recommendation is where and then putting the next one up. But I think we have to go through them in order.

DR. TEUTSCH: Cathy, where are you? How easy is it going to be to walk through these from what you have there?

DR. FOMOUS: If we do them backwards like we reviewed them initially, we can do them very rapidly. If we go Nos. 6 through 4.

DR. TEUTSCH: No, we need to do them in the right order.

DR. FERREIRA-GONZALEZ: We need to do them in order.

DR. FOMOUS: It will be fine. I mean, there will be a slight pause as we go from chapter to chapter, but that is all.

DR. TEUTSCH: Let's try and walk through them

one at a time.

DR. FOMOUS: I would like to recommend, though, that we reserve the overarching recommendation until the end so that we review all the recommendations and make sure that one really captures them.

DR. TEUTSCH: That's fine. You can read it now and we can revisit it at the end if we need to. So, why don't you go ahead.

DR. FERREIRA-GONZALEZ: Chapter 4, Recommendation 1.

DR. TEUTSCH: Are you going to read them to us?

DR. FERREIRA-GONZALEZ: Do you want me to read them? Okay.

"For a number of years CMS has been planning to address gaps in the oversight of laboratories that conduct genetic tests with the addition of a genetic testing specialty under CLIA. Recently, CMS changed directions and is now addressing these gaps with a multifaceted action plan. SACGHS considered CMS rationale and reviewed the agency's action plan. SACGHS carefully considered the recommendations of prior groups as well as the perspective of the stakeholders who

support the specialty.

"In the end, the Committee came to the conclusion that identified gaps can be addressed without the creation of a genetic testing specialty. SACGHS proposes the following recommendations to support and/or augment the CMS action plan.

"Recommendation 1-A. Currently, CLIA requires all non-waived tests to undergo some form of performance assessment, but only 83 specific analytes, none of which are genetic tests per se, are required to undergo the type of assessment called proficiency testing. PT is currently considered to be the most rigorous form of performance assessment.

"In principle, genetic tests and all other non-waived laboratory tests should be required to undergo PT. However, such a goal cannot be achieved immediately. Consequently, the following actions should be taken.

"CMS should require PT for all non-waived laboratory tests for which PT products are available. For tests without PT products, laboratories must use alternative assessment methods, as required under CLIA regulations.

"In order to promote the development of new PT products and facilitate performance assessment efforts, HHS should fund studies of the effectiveness of other types of performance assessment methods to determine whether they are as robust as PT, and support innovations in the way PT is performed, such as through methodology-based processes.

"CMS should consult or contract with experts in the field to train inspectors of genetic testing laboratories. Training by such experts will enhance the inspectors' understanding of the technologies, processes, and procedures utilized by genetic testing laboratories and equip them to assess compliance with CLIA requirements.

"In addition, CMS should identify and evaluate innovative alternative mechanisms to inspect genetic testing laboratories.

"As recommended in the 2006 Government Accountability Office Report on Clinical Laboratory Quality, CMS should use revenues generated by the CLIA program to hire sufficient staff to fulfill CLIA's statutory responsibilities. The program should be

exempted from any hiring constraints imposed by or on the agency.

"Recommendation No. 2. Currently, there are gaps in the extent to which analytical validity and clinical validity data can be generated and evaluated for genetic tests. To address these gaps, SACGHS recommends supporting public resources for genetic testing through the following actions.

"In consultation with relevant agencies, HHS should assure funding for development and characterization of reference methods, materials, and samples; for example, positive and negative controls and samples from different ethnic and geographic populations for assay, analyte, and platform validation, quality control, performance assessment, and standardization.

"HHS should assure funding for the development of a mechanism to establish and support a laboratory-oriented consortium to provide a forum for sharing information regarding method validation, quality control, and performance issues.

"HHS agencies, including NIH and CDC, should continue to work with public and private partners to

support, develop, and enhance public reference databases to enable more effective and efficient collection of mutations and polymorphisms data and expand clinical reference sequence databases, and provide summary data on gene disease associations to inform clinical validity assessments, e.g. RefSeqGene or HuGENet.

"Such initiatives should be structured to encourage robust participation, for example, and may need to consider mechanisms for anonymous reporting and of protections from liability to encourage information sharing among members.

"HHS should provide the necessary support for the development and dissemination by professional organizations of additional standards and guidance for applying genetic tests in clinical practice. CMS should work with professional organizations to develop interpretive guidelines to enhance inspector training and laboratories.

"Recommendation 3. There are considerable information gaps about the number and identity of laboratories performing genetic tests and the specific genetic tests being performed. To gain a better

understanding of the genetic tests being offered as laboratory-developed tests and to enhance the transparency in this field, SACGHS reviewed proposals for a voluntary or mandatory test registry and considered the benefits and burdens of each type of system.

"The community cited that a mandatory, publicly available, Web-based registry that is well staffed to maintain an accurate and current database would offer the best approach to address the information gaps. Since genetic tests are not unique from other laboratory tests for oversight purposes, the registry should include all LDTs. The Committee also discussed whether such a database should reside at CDC, CMS, or FDA.

"Based on exploratory work, SACGHS concludes that a mandatory registry should be established. The Committee recognizes that there are unresolved issues, including practical and legal questions, that require further analysis before a final decision can be made about how and where to implement the registry.

"HHS recommends the following course of action:

"A) HHS should appoint and fund a lead agency to develop and maintain the mandatory registry for LDTs.

The lead agency should work collaboratively with its sister agencies to create a comprehensive registry and minimize duplicative collection of registry information. The lead agency should have qualified personnel who are experienced in developing and updating large databases in a timely and accurate manner.

"The lead agency, in collaboration with its sister agencies, should convene a stakeholders meeting by September 2008 to determine the data elements associated with analytical validity, clinical validity, clinical utility, and accessibility of data that should be included in the registry.

"The lead agency should cast a wide net for a broad stakeholder representation, including representatives from the private sector who can represent a role for public-private partnership in developing the registry.

"The lead agency, through the stakeholders effort, should assess the level of effort as well as the burden on the laboratory and the impact on other key stakeholders such as patients, physicians, and payers, necessary to obtain each data element, including linking

to reliable sources of existing information."

DR. KHOURY: Just, a registry for LDTs or all genetic tests?

DR. TEUTSCH: It should be genetic tests. You will notice copy edits and things like that which we are going to need to try and take care of. I don't think we need to do that as we go through this. So we will talk about that at the end.

DR. GUTMAN: I was going to keep my mouth shut, but you do have an opaque statement about exploring legal authorities. I'm certainly not a lawyer and I hardly can represent the FDA, so I certainly couldn't represent CDC or CMS or HHS in general. But there actually is at least a possibility that in order to do this there would need to be some statutory change.

DR. FERREIRA-GONZALEZ: Yes, we understand that.

DR. GUTMAN: It makes it awkward to have an October '08 meeting if you --

DR. FERREIRA-GONZALEZ: No, the October is for the stakeholders to gather the information on the elements.

"C. While awaiting completion of the other processes, HHS should use short-term voluntary approaches such as incentivizing laboratories to register with Gene Test and encouraging laboratories to make their test menus and clinical validity data for these tests publicly available on laboratory websites.

"Recommendation 4. There has been much debate in the past decade regarding FDA's role in regulating laboratory-developed tests. SACGHS supports FDA regulation of LDT and the" --

MS. ASPINALL: Excuse me. I'm not sure that is correct.

DR. FERREIRA-GONZALEZ: Yes, I was going to say.

[Pause.]

DR. FERREIRA-GONZALEZ: It was the clinical validity. There we go. That is the one.

"The Committee is concerned by the gap in oversight related to clinical validity. The Committee believes that it is imperative for this gap to be closed as expeditiously as possible. To this end, the Committee makes the following recommendations:

"All laboratory tests should be addressed by the FDA in a manner that takes advantage of its current experience in evaluating laboratory tests. This step by HHS will require commitment of significant resources in order to avoid potential harms (patient and public health, staffing, technological innovation.)

"The Committee recommends that HHS convene a multi-stakeholder public and private sector group to determine the criteria for risk stratification and a process for systematically applying these criteria. This group should also consider new and existing regulatory models and data sources, such as New York State. The multi-stakeholder group should also explicitly address and eliminate duplicative oversight procedures.

"To expedite and facilitate the review process, the Committee recommends establishing a registry as noted in Recommendation 3.

"Recommendation 5. SACGHS fact-finding also identified gaps in the enforcement of existing regulations. To prevent laboratories from performing genetic tests without appropriate CLIA certification, the following steps should be taken.

"The CLIA program has an array of enforcement actions available, but those actions cannot be imposed on an uncertified laboratory. Instead, CMS must report the laboratory to the HHS inspector general for action. Laboratories without CLIA certificates cannot be reimbursed by Medicare or Medicaid, but this restriction has no consequence for laboratories that perform direct-to-consumer testing.

"CMS should exercise its regulatory authority to take enforcement actions against laboratories that perform genetic tests for clinical purposes without proper CLIA certification. CMS should step up its efforts to make publicly available a list of laboratories that have been cited by the CLIA for condition level deficiencies."

DR. KHOURY: Andrea?

MS. ASPINALL: I have a question. Is this another one where we take out "genetic"? Because we just talked about all tests. Even if we don't define genetic, if a lab is doing a test.

DR. FERREIRA-GONZALEZ: Yes.

DR. KHOURY: Andrea, do we need the section in

green? Can we just take it out? I always find it confusing. We have to acknowledge it, but it interrupts the flow. Maybe we should move it elsewhere.

DR. TEUTSCH: We can take care of that in editing.

DR. FERREIRA-GONZALEZ: That is in editing.

"Appropriate federal agencies, including CDC, CMS, FDA, and FTC should strengthen monitoring and enforcement efforts against laboratories and companies that make false and misleading claims about genetic tests."

DR. TUCKSON: Including the DTC.

DR. FERREIRA-GONZALEZ: Including the DTC.

DR. TUCKSON: You don't have to type it now.

DR. FERREIRA-GONZALEZ: "Recommendation 6. SACGHS is concerned about certain types of health-related genetic tests that are marketed directly to consumers and appear to fall outside the scope of CLIA. Some nutrigenomic tests, for example a test for caffeine metabolism, and tests to determine the gender of a fetus are examples of health-related genetic tests that are skirting the boundaries of CLIA authority. There is

insufficient oversight of laboratories offering such tests and their potential impact on the public health is an increasing concern.

"SACGHS recommends that CLIA regulations or, if necessary, CLIA statutory authority, along with FDA risk-based regulatory authority and regulatory processes, should be expanded to encompass the full range of health-related genetic tests, including those offered directly to consumers. Relevant agencies such as CMS, CDC, FDA, and FTC, should collaborate in an effort to develop an appropriate definition of health-related genetic tests that FDA and CMS could use as the basis for expanding their scope."

DR. LICINO: Question. When you say they are health-related genetic tests, could people use that as a loophole and say this is just information about your biology, who you are?

DR. FERREIRA-GONZALEZ: This is what we are trying to close. That is exactly the loophole we are trying to close.

DR. LICINO: But then, should you take the "health-related" there and just say "genetic tests"?

Because all of them have some health relevance.

DR. FERREIRA-GONZALEZ: No.

DR. LICINO: It could be like a genealogy. Let's say if you do the $1,000 genome thing comes there and becomes available, then they say, okay, I'm doing it to see where I come from, but the information is sequenced and can be used any way. Could people use that as a loophole and say I'm not doing it for health-related reasons, I'm doing it just to get a genetic history?

DR. WILLIAMS: I think that is why the point of the last thing, which was to develop an appropriate definition. Again, I don't think we can necessarily do that, but we can't leave it to the company to make the definition. That definition has to be defined by the agencies that we want to have regulatory authority.

DR. FERREIRA-GONZALEZ: The exact example that you described could be brought in, too. Martin.

MR. DANNENFELSER: But, a gender-related test may not be health-related. So I don't know that if that language applies. I think the idea of taking "health-related" out makes sense.

DR. MILLER: So, can you just take out "health-

related" and just say certain types of genetic tests that are marketed?

MS. ASPINALL: Isn't it just tests? Again, this definition is moving, technology moves. I think some of those tests are being used in health-directed ways. I think the second sentence makes sense.

DR. FERREIRA-GONZALEZ: "Genetic tests."

MS. ASPINALL: Just certain types of tests.

DR. FERREIRA-GONZALEZ: Certain types of tests that are marketed directly to consumers.

MS. ASPINALL: It is consistent with what we said in the rest.

DR. FERREIRA-GONZALEZ: But then, "should collaborate in developing an appropriate definition of health-related." We leave that there.

DR. FROHBOESE: It is repeated again at the bottom, as well. The last line. "Health-related genetic tests."

DR. FERREIRA-GONZALEZ: Are we okay with this? We can work through that. That is a detail.

Next one. Chapter 5. Recommendation 1. "Information on clinical utility is critical for managing

patients, developing professional guidelines, and making information on clinical utility of genetics tests. There is inadequate data on which to base utility assessments and only a few studies have been done of the clinical utility of specific genetic tests.

"More fundamentally, insufficient analysis has been done on the standards of evidence upon which the clinical utility of genetic tests should be evaluated, and evidence-based methods applicable to genetic testing have been developed.

"Further, policy analysis is also needed to define the process by which clinical utility assessments will be applied. To fill these needs, SACGHS recommends the following:

"HHS should create and fund a sustainable public-private entity of stakeholders to assess the clinical utility of genetic tests. An example is building on CDC's Evaluation of Genomic Applications in Practice and Prevention, EGAPP, Initiative.

"This entity would identify major evidentiary needs; establish evidentiary standards and level of certainty required for different situations such as

coverage, reimbursement, quality improvement, and clinical management; establish priorities for research and development; augment existing methods for assessing clinical utility as well as analytical and clinical validity, such as those used by EGAPP and the U.S. Preventive Services Taskforce, with relevant modeling tools; identify sources of data and mechanisms for making them usable for research, including the use of data from the electronic medical records; recommend additional studies to assess clinical effectiveness; achieve consensus on minimal evidence criteria to facilitate the conduct of focused, quick turnaround time of systematic review; increase the number of systematic evidence reviews and make recommendations based on their results; facilitate the development and dissemination of evidence-based clinical practice guidelines and clinical decision support tools for genetic/genomic tests; establish priorities for implementation in routine clinical practice; and publish the results of these assessments or make them available to the public via designated HHS or other publicly supported, like Gene Test, websites.

"To fill gaps in our knowledge of analytical

validity, clinical validity and clinical utility, utilization, economic value, and population health impact of genetic tests, a federal or public-private initiative should develop and fund a research agenda to fill those gaps, including the initial development and thorough evaluation of genetic tests and the development of evidence-based clinical practice guidelines for the use of those tests, and disseminate these findings to the public via designated HHS or other publicly supported websites, such as Gene Test.

"Recommendation 2. Healthcare payers are increasingly requiring evidence of clinical utility before they will pay for genetic tests. Therefore, coverage and reimbursement decisions play a critical role in stimulating innovation and facilitating access to genetic testing. In February 2006, SACGHS issued a report that made recommendations for developing evidence of clinical utility and addressing other barriers to the coverage and reimbursement of genetic tests and services in the public and private sectors. SACGHS offers the following recommendation concerning the development of clinical utility evidence.

"As the issues identified in the Coverage and Reimbursement of Genetic Tests and Services Report ar still current, SACGHS urges HHS to act on the report's recommendations.

"In addition, public and private healthcare payers, in collaboration with relevant groups such as test developers and clinical laboratorians, should develop mechanisms such as development of phased reimbursement to facilitate the collection of clinical utility evidence for high-priority tests and applications. Implementation of innovative approaches should be accompanied by careful evaluation to assess whether they enhance or hinder innovation, understanding effectiveness, and appropriate utilization.

"Recommendation 3. The value of genetic tests to patients is realized only when they are used appropriately. In addition, quality improvement processes are needed to assure that genetic tests are delivered consistently to appropriate patients. Furthermore, an ongoing process is needed to identify opportunities for improving the use of genetic testing, including the collection of post-market outcome data.

SACGHS therefore makes the following recommendations.

"HHS should conduct public health surveillance to assess surrogate and health outcomes practice measures, including the proper utilization and the public health impact on genetic testing. Information should be linked to quality improvement practices that affect patient outcomes and the provision of health services.

"Data on specific genetic testing results will be required to permit understanding of the significance of genetic variance and new detection methods to improve the utility of testing.

"Recommendation 4. The clinical utility and value of genetic testing is inextricably linked to methods to improve care processes and decision support. Interoperable electronic health records will play a central role in the translation of guidelines into care practices through their decision support and educational functions. They will serve as a critical resource for assessing clinical utility and quality of care. SACGHS therefore makes the following recommendations.

"HHS should ensure the coordination and implementation of efforts, including the deliberation of

SACGHS and AHIC, or its successor, and other workgroups addressing personalized health care, population health and clinical care connections, and confidentiality, privacy, and security to advance the appropriate use of patient-level data for research and for enhancing the quality of decision-making.

"Chapter 6, Recommendation 1. There are documented deficiencies in genetic knowledge in all relevant stakeholder groups. In addition to the creation of the SACGHS Education Taskforce, SACGHS recommends the following strategies to address these deficiencies.

"HHS should work with all relevant governmental agencies and interested private parties to identify and address deficiencies in knowledge about appropriate genetic and genomic test applications in practice and education of key groups, such as healthcare practitioners, public health workers, public and private payers, and consumers. This educational effort should take into account the differences in language, culture, ethnicity, and perspectives on disability, as well as issues of medical literacy, access to electronic information sources such as the Internet, and

deficiencies in public infrastructure such as libraries that can affect the use and understanding of genetic information.

"Based upon increased research regarding analytical validity, clinical validity, and clinical utility, sufficient resources should be provided for the translation of this knowledge into evidence-based clinical practice guidelines that enhance the quality of clinical care and public health outcomes. See also Recommendation 3, Chapter 5.

"Although FDA has asserted its authority over clinical decision support systems, the extent to which the agency intends to regulate such systems is not clear. Given that clinical decision support systems will be necessary to communicate information appropriately in the pre- and post-analytic period and because these systems contain elements that involve the practice of medicine, clarification of the nature and scope of FDA oversight of such support systems is critical.

"SACGHS recommends that FDA should engage with other relevant federal agencies, advisory committees to the Secretary of HHS such as AHIC and the Newborn Genetic

Testing, and stakeholders to gather perspectives on the appropriate regulatory framework for clinical decision support systems in light of the change in healthcare delivery and healthcare data collection systems.

"As part of this process, FDA should prepare a guidance document articulating the basis of its authority to regulate clinical decision support systems, as well as rationale and approach to such regulation explaining in particular which features of the system constitute a device.

"The need for genetic expertise to support best genetic testing practices has been identified as an essential element for the provision and interpretation of appropriate genetic tests. Access to genetic expertise could be addressed in part by solving problems in the reimbursement of genetic tests and services. SACGHS recommends that HHS act on the recommendations of the 2006 SACGHS Coverage and Reimbursement of Genetic Tests and Services Report.

"There are extensive gaps in knowledge about genetic tests and their impact on patient care. Prioritizing activities under the authority of HHS would

help to close these gaps and enhance the quality of patient care. SACGHS recommends that HHS allocate resources to AHRQ, CDC, HRSA, and NIH to design and support programmatic and research efforts in order to encourage development and assist in the evaluation and dissemination of tools, particularly computerized tools, for clinical decision support in the ordering, interpretation, and application of genetic tests; and address current inadequacies in clinical information needed for test interpretation.

"These efforts will require engaging providers and payers as well as providing incentives and protections in order to ensure participation in design and dissemination of tools, implementation of clinical decision support, and contribution of necessary data."

DR. FOMOUS: The next one I think you want to decide if you want in or out.

DR. TEUTSCH: The next one we deleted, right?

DR. WILLIAMS: My sense was it should be deleted, but in the presentation of all the recommendations we wanted to assure that the Committee agreed with what I think, which is we have covered this

already back in Chapter 4.

DR. FERREIRA-GONZALEZ: What about the privacy issues?

DR. WILLIAMS: We talked a lot about the need for this. We now have a recommendation in Chapter 4 that deals with the marketing of DTC tests, but it is general, as part of oversight of all of those marketing programs, including them, and doesn't specifically speak to this issue.

I think the preamble, which is the first paragraph, which is setting the stage, does present some unique aspects that probably need to be folded in, but the recommendation per se is covered. Basically, I think we need to work the text from the preamble into the relevant [section.]

DR. TEUTSCH: If people are all right with that, we will do that as part of the editing process and drop this recommendation because it is dealt with elsewhere.

Andrea, thank you very much. That is a real tour de force just to read.

MS. ASPINALL: We have one comment.

DR. TEUTSCH: We have one more? Oh, we have the overarching one. Did you read it or not?

DR. FERREIRA-GONZALEZ: Hold on, hold on.

DR. FROHBOESE: But, does the preamble adequately cover the issue of privacy?

DR. WILLIAMS: It covers it enough from the perspective of the recommendations. The text of the report has additional information on that. The setup for the recommendations is essentially just to give a little bit of a taste of what is actually in the report that is discussed in more detail.

MS. ASPINALL: I'm sorry. Can I go back to Chapter 6, Recommendation 3? I thought we were waiting until the end of the chapters. The issue that we have talked about several times and I think is critical -- so tell me if I'm missing something -- is the need for appropriate reimbursement for these tests.

DR. TEUTSCH: That is in this report.

MS. ASPINALL: It is in this report, which I understand, and I think it is great that we refer to it. I just read the preamble here as the need for genetic expertise focused on genetic counseling and other

services but not the tests themselves.

DR. TEUTSCH: That is correct. This is the chapter on decision support and education.

MS. ASPINALL: That is where I was waiting until the end, but maybe it is appropriate here. I think it was the sense of the Committee to say it more globally, that in light of the importance of the tests and, quite frankly, additional requirements that we are going to have as a result of it, that we need to ensure that reimbursement is reviewed. That is not the FDA, and it is perfect for HHS, which has the CMS component.

DR. TEUTSCH: That is another recommendation, either No. 4 or No. 5.

DR. FERREIRA-GONZALEZ: Chapter 5. Do you want to go back to Chapter 5?

DR. TEUTSCH: We have referenced this report twice, once relating to tests, once relating to expertise.

MS. ASPINALL: Which one is it?

DR. FERREIRA-GONZALEZ: Chapter 5.

MS. ASPINALL: I didn't see it.

DR. FOMOUS: No. 5-2.

MS. ASPINALL: So that was why I wrote down 5-2. Can you just go back to the first piece of it? This, to me, was not about the overall reimbursement but rather to facilitate the collection of clinical utility evidence which was referenced in the other report, as opposed to more broadly.

DR. FERREIRA-GONZALEZ: Do you want to have a specific statement to also look at the reimbursement?

MS. ASPINALL: Yes.

DR. FERREIRA-GONZALEZ: Because it is covered in the report.

MS. ASPINALL: Right. I do. I am happy to have it anyplace, and maybe it is in the overarching place at the beginning, that this report is only complete in addition to the 2006 Coverage and Reimbursement Report. That would be fine. Nos. 5-2 and 6-3 are very specific and important, but it is fine if it is in the overarching. I was waiting until we saw these because they are very specific on individual issues.

DR. TEUTSCH: The whole report, Mara, on this topic was not a subject of great focus here except insofar as it dealt with oversight. We tried to allude

to it at least on several occasions plus in the text. So we understand that that is an issue and that is, obviously, why we are continuing to address it.

MS. ASPINALL: I guess given that a lot of people will look at the recommendations, the two recommendations at 5-2 and 6-3 that mention it, mention it in reference to a specific aspect, clinical utility or genetic counseling or genetic expertise. What I was saying at a higher level is this report needs to be connected with the Reimbursement and Coverage Report.

DR. FERREIRA-GONZALEZ: Where would you recommend that we put that? You want a third mention to this report.

DR. TEUTSCH: The other way to go is in the text. I think it is important that we do make these links because this relates to much of our work.

Could we ask you to take a look and make sure that it is clear in the text of the report that we are addressing the reimbursement issue, that it is taken up there, it is an important issue in making sure that there is access?

MS. ASPINALL: I think it is in the text. I'm

saying it needs to rise to the overarching piece of the recommendations because I think it gets lost in 500 pages.

DR. TEUTSCH: Marc.

DR. WILLIAMS: I would make two suggestions. One is that we are obviously still engaged with the Secretary on the Coverage and Reimbursement Report, and many of the issues that are still outstanding there are being addressed. I personally don't think we need to keep beating this about the head and neck. I think it is relevant where it is in the recommendations. It is adequately addressed in the text.

I would suggest we just do a thumbs up/thumbs down in terms of how people feel about this. Is this something that is important to represent as an overarching? I would argue not. But if people feel strongly that way, then we can work on where to put it in.

DR. FERREIRA-GONZALEZ: Any language that we can add to any of these two recommendations?

DR. WILLIAMS: Not really, not really.

DR. TEUTSCH: Let me just get a sense of the

group. How many people think we need either a recommendation or to modify something to add additional reference to the Reimbursement Report about the need for testing?

[Show of hands.]

DR. TEUTSCH: How many feel that we do not need to do that?

[Show of hands.]

DR. TEUTSCH: I take it the sense is we all recognize it is an important issue, and if we can strengthen it somewhere else we will have other ways to deal with that.

That is a real tour de force, Andrea. Let's see if we can get to a vote on the recommendations.

DR. FERREIRA-GONZALEZ: Are we doing the overarching?

DR. TEUTSCH: Do you want to go back to that?

DR. FERREIRA-GONZALEZ: Yes, the overarching.

DR. TEUTSCH: Read it.

DR. TUCKSON: Are you going to vote individually, Steve, or just can you try to vote en masse?

DR. FERREIRA-GONZALEZ: The overarching recommendation.

DR. TEUTSCH: We will. No, we just skipped over that and Andrea asked that we come back to it.

DR. FERREIRA-GONZALEZ: "The Committee found significant gaps in the U.S. system of oversight of genetic testing that could and do lead to harms. The Committee formulated a number of recommendations that, if implemented and sufficiently supported, could close these gaps. The Secretary of HHS is responsible for closing these gaps and fostering the public's health."

DR. FITZGERALD: I understand that there was some concern about the forcefulness of that final statement, but one can look at it in a different way that presents the same logic. Take that last sentence and imagine it in this form: "If these gaps are to be closed and the public's health fostered, then the Secretary of HHS must take responsibility for this process."

I think the fear is that this is coming across as some kind of determination by this Committee that the Secretary should take up some extra job or responsibility or something.

MS. ASPINALL: No.

DR. FITZGERALD: I think what we are trying to say is no, this is just part of the reality. If this is going to be done, the Secretary of HHS is the one to do it.

DR. TEUTSCH: Right. The Secretary, of course, has asked us to do it because, presumably, he believes that he has the responsibility for doing it.

DR. FERREIRA-GONZALEZ: Robinsue.

DR. FROHBOESE: I think, in keeping with that, aren't we missing something here? I'm wondering whether "responsibility" is the right concept or the fact that we need to tie the fact that the Committee formulated a number of recommendations. Don't we want to say that the Secretary can and should close these gaps by following these recommendations?

I think by trying to couch this in terms of responsibility we are missing that linkage. The Committee found gaps, formulated recommendations, and then the last thought is that the Secretary can and should close these gaps by following the recommendations.

DR. LICINO: Robin and Reed, both of you were

not here. We had a discussion about this. The original language was "The Secretary of HHS must close these gaps," and then we discussed here how do we tell the Secretary what to do without sounding too presumptuous. That is the challenge, I think.

DR. TUCKSON: Because you are his advisory committee. He asked us to tell him how to do it. Whatever the discussion was, I don't feel a certain shyness here. The only word that I'm concerned about is the word that we are saying these recommendations "could." I thought we felt pretty good these recommendations "will." We didn't come at this to say that there are big holes left.

So I think we should say that these recommendations would close these gaps, and I do like the way Kevin phrased it. I thought it was very nice.

DR. FITZGERALD: I don't think we can say "will close these gaps" because if they are not implemented they won't do it.

DR. FERREIRA-GONZALEZ: How about the language?

DR. TUCKSON: I think we want to just be real careful that you don't make it sound like you are

undercutting your own recommendations by saying, hey, these are some nice ideas.

DR. FERREIRA-GONZALEZ: How about the last sentence?

DR. FITZGERALD: My understanding is the reason we put in that word was because Reed was pushing for accountability.

DR. FROHBOESE: But, can we get at it by saying "The Secretary "can and should close these gaps by"?

MS. ASPINALL: I like the wording. Because, otherwise, it is not connected to the other paragraph.

DR. FERREIRA-GONZALEZ: Can you repeat the wording?

DR. FROHBOESE: I'm recommending that we say "The Secretary of HHS can and should close these gaps by implementing the Committee's."

DR. EVANS: Or we can just put that last sentence as the second sentence and say, "The Secretary of HHS can and should close these gaps by implementing the recommendations that are contained."

DR. FERREIRA-GONZALEZ: "The Secretary can and should close these gaps to foster the public health."

DR. FITZGERALD: One thing I don't think we want to lose again is what Phyllis also pointed out. It is not just a matter of closing gaps. That doesn't solve all the issues. There are more. So we don't want just to worry about closing gaps. We want to actually move beyond that once that happens.

MS. ASPINALL: So, Kevin, with that, do we want to start with "The Committee found significant gaps and necessary improvements" or "potential improvements in the U.S. system"? You are right -- well, not just you, Kevin, but everybody -- it is not just about gaps that we are filling. It is about improvements.

DR. FERREIRA-GONZALEZ: Joe?

DR. TELFAIR: I'm very clear that you need to plainly state what it is that you want to do. To me, changing that, with all due respect, is not plain and is not straightforward. It is just saying this is your responsibility, this is what you do, and that is it. That is just laying it on the table. Also, it reinforces the fact that there are things that are beyond the main outcome, which is gaps. There are things beyond that that need to be done, and it is perpetual. It doesn't

just stop at that point.

DR. FOMOUS: I think you need some sort of language in there that sounds like a recommendation because I think the prior sentence that we had in there was like this is all nifty and dandy, you are responsible. Where is the recommendation for action?

DR. TUCKSON: Maybe one way to do it is instead of saying "The Secretary of HHS can and should," "We recommend that the Secretary of HHS close these gaps." "We recommend that he close the gaps."

DR. FERREIRA-GONZALEZ: Yes, that's it. That's it.

MS. ASPINALL: What about gaps and improvements?

DR. FERREIRA-GONZALEZ: Hold on. Let's finish with this and then we will go back.

DR. TELFAIR: But there is an implied endpoint there if you just say "close the gaps." We already know that this is a perpetuating situation. Even though this Committee at this point in time has identified these, there are anticipatorily going to be other means that are going to come up. It was made pretty clear through both

the public comment and in things we have heard that this is only basically the tip of the iceberg right now and there is more to come.

DR. TUCKSON: So maybe one slight way you could do it [is], "The Committee recommends that the Secretary of HHS close these gaps by implementing the recommendations and continuing to advance," "continuing to monitor and respond appropriately"? No, that's crappy. Never mind.

MS. ASPINALL: How about just "implement the recommendations"? We are giving recommendations. We are saying, listen, do it.

DR. EVANS: If you want to say something about the public's health, just say it at the end of the middle sentence. "Would close these gaps and foster the public's health. Period. The Committee recommends the Secretary of HHS close these gaps by implementing the recommendations."

PARTICIPANTS: No.

DR. EVANS: "Implement the recommendations." Period.

PARTICIPANTS: Period.

DR. FERREIRA-GONZALEZ: Mara, you had a comment?

MS. ASPINALL: I had a different comment from Sylvia's comment, I think. Somebody said it. The issue about "The Committee found significant gaps and potential improvements possible in the U.S. system." "Opportunities for improvement in the U.S. system." It is not as if it is all about gaps, it is also about improvements.

DR. TEUTSCH: Can we get a sense if folks are all right with this recommendation as it is?

MS. ASPINALL: Can we say "recommendations," period.

DR. FERREIRA-GONZALEZ: So, how about in the second sentence, "would close the gaps."

PARTICIPANTS: No.

MS. CARR: "Enhance the public health" after gaps? Second sentence.

PARTICIPANT: "Would close these gaps and enhance the public's health."

PARTICIPANTS: Yes.

DR. TEUTSCH: You can put that up there.

MS. ASPINALL: But I'm still going to come back with, are we really okay with saying the report is only about gaps?

DR. WILLIAMS: We are talking about enhancing the public's health. I think we addressed it.

MS. ASPINALL: How about, we are enhancing it through just --

DR. FERREIRA-GONZALEZ: "Enhance the system." We need to enhance the system.

MS. ASPINALL: We are just closing gaps, is what this sounds like. I think we are doing more than that.

MS. CARR: Enhance the oversight system.

DR. WILLIAMS: So you could say, "The Committee recommends that the Secretary of HHS enhance the oversight system to close these gaps."

DR. TEUTSCH: It should say "implement the recommendations." "Implement the recommendations and assume responsibility for improving the oversight of genetic tests." That is the ongoing part.

MS. ASPINALL: I think it just easier in the first sentence to say "close gaps and opportunities for

improvement."

DR. LICINO: "To enhance" or "and enhance"? Would "close these gaps in order to enhance the public's health," right?

DR. FERREIRA-GONZALEZ: No, no. Paul. We need to finalize this.

DR. TEUTSCH: We need to get to closure. Paul?

DR. LICINO: You need "public health" twice. I like public health and all its advocates, but you need it twice.

DR. TEUTSCH: Paul.

DR. MILLER: The train may have left the station by now, but the title of this is "Overarching Recommendation," and the overarching recommendation is to implement the recommendations.

[Laughter.]

DR. MILLER: That just seems to be a little circular to me. What I thought originally, as I was thinking about an overarching recommendation, is in a sense going back to what, I forget whether it was Steve or Reed, said it. The loyal staffer comes in and says, "SACGHS has come up with this report." The Secretary

says, "Well, that's great. What's the bottom line?" The bottom line is, in a sense, what is the real push here.

I like that first thing saying, hey, there are significant gaps and there is a way to solve these gaps. Again, the train may have left the station, but ultimately, what is the core thing, if you can capture it in a thought or a thing, in terms of the overarching point? Because of changes in technology and the greater use of genetic tests, more resources and more oversight and stakeholder input are needed to come up with something.

DR. EVANS: Closing the gaps, isn't that the overarching recommendation? Didn't we decide that, look, what has to be done? There are gaps. They have to be closed. Isn't that our overarching recommendation?

DR. MILLER: I'm just saying that this is an opportunity to say something more concrete, and I wonder if we are taking that opportunity. That is my point.

DR. TEUTSCH: I know we need to bring this to closure. I wonder if you could, with some forbearance, allow the steering committee to work on this and get it finalized.

DR. FERREIRA-GONZALEZ: We keep going around and around.

DR. TEUTSCH: I don't think we can wordsmith this by committee at this moment.

DR. FOMOUS: Instead of calling this an overarching recommendation, could we call it an overarching scene? Then there wouldn't be this sense that you have to be recommending an action to capture all of the recommendations.

DR. MILLER: I think you do need it. I think the report is stronger by having an overarching recommendation that follows from, we found significant gaps and here is something concrete that you need to do.

DR. TUCKSON: I think the suggestion from the chairman is that we have a sense of what is needed. The committee is going to have to work this one a little bit. It is not a technical recommendation. It is a matter of style here. I think Paul's comments are well taken here. I think the Committee understands it.

I would pile in that as you think about it offline that I realize that we have lost the key, which I think is the overarching issue, accountability. Alphabet

soup. Americans are concerned. There is somebody in charge. Deal with it.

What I would suggest from a process point of view is that we have, I think, gotten the recommendations done. The Committee can grapple with this and send it back to us for wordsmithing or approval on the final.

I think we need to take a vote and get this done. It is almost one o'clock. That is our drop-dead date. We have four minutes before the end of the hour, and we have a critical quorum issue to deal with. So unless there is a whole big issue that somebody else has, we should vote.

DR. TEUTSCH: We are going to vote on the recommendations in each of the chapters. We are going to have the steering committee work on this overarching one. What I would like to do is to proceed to a vote as we have talked about. So, other than editorial changes and some wordsmithing on this, that is what we will be voting on.

Before we actually take our vote, we need to make sure that we have a record that two of our members, Paul Billings and Paul Miller, still have some pending

paperwork, so that they are not going to be able to vote because of a delay in that process. Apologies to both of you.

For the remainder, we do need to take a vote. Do I have a motion in favor of accepting these recommendations?

DR. TUCKSON: I move.

[So moved.]

DR. WILLIAMS: Second.

[Motion seconded.]

DR. TEUTSCH: One from Reed, and a second from Marc. All in favor?

[There was a chorus of "ayes."]

DR. FOMOUS: Show of hands, please? And keep them up.

[Show of hands.]

DR. TEUTSCH: All opposed?

[No response.]

DR. TEUTSCH: Any abstentions?

[Motion carried.]

DR. LICINO: Can I make a motion?

[Laughter.]

DR. TEUTSCH: Go ahead, Julio.

DR. LICINO: I would like to make a motion for us to approve this as is without further editing.

DR. FERREIRA-GONZALEZ: No, the steering committee has an idea.

PARTICIPANT: It may wind up like that.

DR. TEUTSCH: It may. I get a sense that there is a significant amount to do there.

DR. TUCKSON: Mr. Chair, can I make a motion? I would like to make a motion that we give a resounding --

DR. TEUTSCH: This is what I was --

DR. TUCKSON: Oh, if that is what you were going to do, Mr. Chairman, it has more power coming from you.

DR. TEUTSCH: No. I think this has been an extraordinary effort on the part of Andrea and all of the steering committee, the taskforce members, the staff.

DR. FERREIRA-GONZALEZ: The staff.

DR. TEUTSCH: An enormous amount, and all of you.

DR. TUCKSON: And the public.

DR. TEUTSCH: And the public.

DR. TUCKSON: And the public, and the public, and the public.

MS. CARR: Implicit in what you just voted on is that you have also approved the spirit of the report going forward. Everybody understands that. You need to get edits by February 20th to Cathy.

PARTICIPANT: Yes. Can you just go through this process of edits by February 20th and then the completion by the 29th?

DR. TEUTSCH: The recommendations by the 29th will be finalized and sent to the Secretary. We will copy-edit the document and then the final version will go in April.

MS. CARR: By April. But we will copy-edit the recommendations and you will see what the steering group came up with on the overarching. You will see all of that again. But, no more edits to the recommendations.

MS. ASPINALL: Except as we have talked about them going through it because there were still some clarifications in putting them together.

MS. CARR: We won't be receiving more edits or

seeking more edits from you guys on the recommendations.

DR. FERREIRA-GONZALEZ: We are just going to be cleaning up the language to assure that we have the "genetics" replaced and so forth.

DR. TUCKSON: I didn't hear the applause.

[Applause.]

DR. TEUTSCH: Thanks, everyone. That has been an enormous amount of work. We were going to take a 45-minute break for lunch. That will put us back here -- hopefully they are still serving -- at quarter to two.

[Lunch recess taken at 1:00 p.m.]

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A F T E R N O O N S E S S I O N

[Reconvened 1:48 p.m.]

DR. TEUTSCH: We are recharged after our morning's exertions. Now we get to turn to, hopefully, some fun stuff. We have two items for this afternoon. One is to review the charge for the Genetics Education and Training Taskforce, and then we get to blue-sky what our future might look like.

First, I need to remind everyone that this is the taskforce that we got underway in November. Reed had given them some preliminary direction and charge, and the taskforce has been busy trying to sharpen up what they are going to do.

We owe a special debt of gratitude to Barbara Burns McGrath for a couple of things. One is, as you may or may not know, she had to do a lot of this work from the other wide of the world, where she was, and then she has had a lot of things to deal with in the last week. So we are particularly grateful that she actually was able to find a few hours to come and be here not only for this morning's session but to be part of this discussion.

What we have asked Barbara to do is to go ahead

and present the draft charge for the Genetics Education and Training Taskforce and then to lead us in a discussion. What we need to do, of course, is get to agreement. We will vote on the charge at the end.

You will find the materials in Tab 4 of your briefing books. We really appreciate, Barbara, all your extraordinary efforts to be here and to lead us through this discussion.

SESSION ON GENETICS EDUCATION TASKFORCE

Draft Charge for the Education Taskforce

Barbara Burns McGrath, R.N., Ph.D.

[PowerPoint presentation.]

DR. McGRATH: Thanks, Steve. It makes me nervous to start another taskforce when we are just putting one to bed. As everyone keeps saying during breaks, let's use this one as a learning curve. So, we can learn from the Oversight Committee, for better or worse.

DR. TEUTSCH: You mean we get to do it again?

[Laughter.]

DR. McGRATH: I will give a little bit of an overview of what we are up to for about 20 minutes. Then

we have, for about another half hour, some time to talk about what is going on with the Committee.

The title of it is an Education and Training Taskforce, and it is made up of several of us on the Committee here and several ex officio members. Our staff representative is Cathy Fomous, who has been very much involved in this.

The purpose for today is simply to give a bit of an update to all of you on what we have been doing over the last couple months. One thing we did was to create a draft charge. We will go over that. We would like some feedback on it because the goal for today is to reach consensus on a final version of that charge.

In terms of background about this committee, the history of the education and training interests with SACGHS goes back to its very beginning, when it was one of the priority areas. Following that, there was a meeting around 2003 that resulted in a resolution that was written in 2004. We have used that as our starting point. Then we met last year in November for the first time and had a session on that. The resolution that was written in 2004 was a great starting point.

The whole thing is in your tab area as well. If you read it, you might come to the same conclusion that we did. It was fairly broad. Nothing to argue about in there. It was all logical recommendations that anyone might want to make. So it gives us a little bit of guidance, but we are hoping to move it forward.

Last fall, we did have what I thought was a pretty interesting session where we first identified some really interesting people in the community who know this area well. Ten folks were here and talked about education and training from each discipline's perspective. The areas that they covered were professional as well as education, diversity in the work place, family history, and emerging issues, as well as who the emerging stakeholders might be.

At the end of that meeting in November, there was a discussion. A vote was taken by the committee, first saying that the topic continues to be of interest and is consistent with our charter, and second, that these issues can best be addressed through a taskforce. So the taskforce was formed.

Between November and now, that task group has

been meeting on Email and phone conferences, and our one order of activity was to develop a charge so we know what direction we should be heading. That is what we will be looking at.

I thought about whether reading all of this aloud would be a good idea, or have a little silent reading, but I'm afraid if we do silent reading it will get kind of dozy in here. So I will read the charge and then we can talk about it. I ask for comments to hold to the very end so you get the flavor of the whole draft. It is not that long. Then we will open it up to discussions to help us fine-tune it a little bit more.

The first thing is the need for the charge. "Advances in genetics and genomics are leading to a better understanding of disease processes and improved application of genetic testing to guide health decisions. With increased integration of genetics into other medical disciplines, however, health professionals with or without training or expertise in genetics are challenged to keep pace with this dynamic and rapidly evolving field.

"Education will have to address the growing

importance of genetics in common disease, which likely will require more knowledge and understanding about risk assessment and communication.

"In addition, the accelerated growth of direct-to-consumer genetic services highlights the need for informed decision-making." That is the need.

"To realize the benefits of genetic technologies and protect against potential harms, the education of the healthcare professionals, the public health work force, and the general public is critical. For these reasons, the Secretary's Advisory Committee on Genetics, Health, and Society has formed a taskforce to build on the findings of the Committee's 2004 Resolution on Genetic Education and Training of Health Professionals."

The overall aim. "The taskforce is charged with developing a plan to identify the education and training needs of health professionals, lay health educators, and the general public in order to optimize the benefits of genetic and genomic services for all Americans. This plan will also outline the steps required to meet these needs and evaluate the efficacy of

educational and training efforts. The plan includes, but is not limited to, the following activities."

So there are basically eight goals that we have created in this process.

"No. 1, Assembling evidence to determine which recommendations from the 2004 Education Resolution were implemented and which ones require additional efforts;

"No. 2, Identifying the education and training needs specific to genetics and genomics for health professionals involved in providing care for individuals and for those involved in the development of guidelines, policies, and strategies for incorporating genetics and genomics into clinical care;

"No. 3, Identifying the education and training needs of lay health educators who are non-credentialed individuals from the local area trained to promote health and provide general health care services for a specific condition or program;

"No. 4, Identifying the education needs specific to genetics and genomics for medical directors, administrators, and policymakers in the public and private sectors to inform policy development,

legislation, coverage and reimbursement decisions, and other issues that directly or indirectly impact the provision of genetic services;

"No. 5, Identifying the education needs of patients and consumers to assist them in informed decision-making about the use of genetic services and enhance their understanding and utilization of results and how these results impact decisions about prevention or treatment;

"No. 6, Identifying effective educational tools that can be incorporated into electronic health records, personal health records, and clinical decision support systems that would enhance the appropriate integration of genetic and genomic technologies throughout the healthcare system without adversely impacting privacy, access, and work flow. In addition, identify gaps where such tools do not currently exist and develop recommendations on how to address these gaps;

"No. 7, Assessing the use of evaluative research methods to determine the efficacy of genetics and genomics education and training; and

"No. 8, Promoting active involvement by health

professional governing bodies that influence education and training (for example, residency review, National Board of Medical Examiners, and so forth) to be more proactive in their requirements for genetics in curricula, clinical training, and licensing and certification and continuing ed requirements."

So those are the goals that we created. The idea is that after we fine-tune these, perhaps make them more specific, that will give us some guidance about what other members to add to this taskforce. So we will be adding ad hoc members based on the goals that we end up deciding on today, and they could come from any of these sorts of organizations or other ones that we identify today.

So the next steps are to decide on the initial activities that we think are important for this taskforce to do; form workgroups, because there will be too many. We need to divide them up. Select ad hoc members as needed; and, the report on our progress at the July 2008 meeting.

I'm interested in hearing your thoughts on the scope of this taskforce. I think there is a danger of it

getting too large so that we take on things that are perhaps handled in other groups or have been handled in other ways. So I would like to get some feedback on the scope.

My other plea is that I hope we end up with goals that are measurable so that at the end of the period of time the next task group that comes on education in five years' time will be able to look at our work and really have a clear sense of whether they were achieved or not, rather than just having them be pleasant suggestions.

Those are my two pleas. I'm asking for help to help us decide as a group where our scope should be, where we should draw the boundaries, and help us to make these more measurable.

That is all I needed to say now. I think we can open it up to suggestions and feedback.

Discussion and Finalization of Taskforce Charge

DR. BILLINGS: One simple suggestion is that, in looking at the older recommendations, you have a specific one that says you are going to look at the old report and which ones need more work and which ones have

been implemented. Do you also want to include some evaluation of their impact or the effect of them? You talk about evaluations later on. Maybe you want to start by evaluating the ones that were actually called upon that were provided before.

DR. McGRATH: And evaluate their impact specifically.

DR. BILLINGS: Right. To recommend education can be a kind of banal recommendation unless education leads to some better outcome.

The other thing I was going to suggest was, you are dealing with, potentially, physicians, non-physician health providers, patients, and consumers. They seem to be potentially targets of this educational assessment activity.

DR. McGRATH: And policy-makers.

DR. BILLINGS: And policy-makers, right. That is a pretty broad swathe.

DR. McGRATH: Yes, it is.

DR. BILLINGS: I wonder whether narrowing that somewhat might be to the best interest of being effective.

DR. McGRATH: Let's list that again. We have healthcare providers or practitioners, patients, consumers --

DR. BILLINGS: I see them as different. I see each one of those as individual groups.

DR. McGRATH: Yes, just as a list. That, that, lay health educators, policy-makers, and then we have some language about credentialing bodies, which could be five or six. So, what do we think about that?

DR. FOMOUS: Can I just say something to Paul's first point? I will call on the taskforce members to either tell me I'm remembering this completely incorrectly or fill in the details. It seems like we did have a discussion at one point about evaluating the impact of the 2004 recommendations. I think there was concern that this might actually bog us down because it would be difficult to make that evaluation because the recommendations were rather broad, it would be hard to determine whether things had been carried out or what impact they had.

I know this was a point of discussion. Maybe others can fill in where we want to go with that.

DR. WILLIAMS: I'm wondering if maybe a compromise to that would be, since our report was advisory to the Secretary, would it be possible to engage with the Secretary's staff to say this report came out in 2004. Did it lead to any tangible activities from the Secretary or secretariat agencies that you could point us towards. Is that a fair question to ask back to staff?

MS. AU: I thought that was what we had decided on one of our calls, that was a question that we were going to ask. Because we weren't sure whether it was a report or some other thing that caused the education to happen.

DR. TELFAIR: I would agree. That was considered one of the first steps that we were going to take, actually. So, do the background work to start with.

DR. McGRATH: That is great. That is the first goal, to request that office to give us a report and from there we take the next step. Great.

DR. GEOLOT: I was not on the conference call, but I agree with Paul. It just seems like the charge is so broad when you include lay health educators as well as

patients. Is the intent of this report to look at the capacity of health professionals and others in the genetics field to provide the information that is needed by patients? I'm trying to figure out how lay health educators and patients are part of a charge in terms of looking at educational and training needs.

DR. McGRATH: Joe?

DR. TELFAIR: Just as a point of clarification, as I think you maybe implied through your question, lay health educators do come from the consumer population. The point there is that there are a number of organizations and groups that use lay health educators as their primary educators to the public and to consumers.

So one of the concerns always is that they themselves are as up-to-date as possible and receive the best training as possible such that when they engage in that work that is there. That is the thrust of that because of the nature of that.

That is at least my understanding. The rest of the committee can correct me, but that is one of the main reasons why they are in there. The whole idea here is to begin to look at the means by which the public and others

receive, digest, utilize, and assess information for the public's good.

DR. EVANS: I wasn't able to be at the fall meeting. I'm certainly sensitive to the idea that, gosh, you are taking on a lot by naming these different stakeholders. I guess what I would argue, though, is that it is very hard to separate provider education and public education. They go so hand in glove. Anybody who takes care of patients will tell you that a well educated patient population is extremely important to getting done what you need to get done. I think that genetics is new enough and a fast enough moving target so that it presents big challenges for both.

Although it would be nice to parcel it out and say, okay, we will just focus on the providers or we will focus on the population, ultimately I'm not sure if that makes a lot of sense. I kind of feel like they have to be attacked together.

DR. FITZGERALD: To build on that a little bit, taking a cue from our discussion earlier today of the role that HHS can play as a coordinator or an agency that has the resources to be a focal agency in this area,

there have already been a wide variety of efforts addressing education of a particular group with genetic information

I am aware of [several] with religious groups, for instance, because a lot of people end up having to deal with people in their tradition with their genetic questions. So there has been quite an effort to educate clergy along these lines. Obviously, the genetic counselors have been deeply involved for a long time with genetic education.

So one of the ways you could look at this would be, in a sense, to help coordinate that in a way that it has not been done before. You have a variety of individual efforts out there, some of which have been relatively successful but narrow. Try to see how one then can maybe come up with something that isn't necessarily a "one size fits all," but at least you are aware of what everybody else is doing and what seems to work in their particular venue.

MS. ASPINALL: I very much agree with what Kevin said, and I thought that at our last meeting what was so impressive in our brief conversation was that 12

people came with public comment, or actually formally as part of the agenda at the last meeting. At least I felt, one after another, wow, that looks like a great program; wow, that looks like a great program; oh, they are implementing it here.

One of the questions I asked is, do you guys ever talk to each other? The answer was no, or "We know each other but we don't really do it."

I have to think about how it would change the draft, the goals of the draft charge, but the sense I have is that we are not recreating the wheel, we are working as an organizing body to ensure the best demonstrated practices. Given the amount of time and resources that we have to do it, creating the best document and connecting the folks who are doing it to say these are the best practices, may be in point the highest leverage we could have.

From my bias from that perspective, it would focus on working with the organizations that are already doing it as opposed to focusing, for instance, specifically on a consumer body, which is just a huge task. It goes back to what Paul said earlier. I think

we narrow it by working with the organizations who are already working at it and leveraging their work as opposed to creating new work.

DR. KHOURY: Just to echo some of the stuff I heard, there is a lot of genetics education going on, so much of it, actually, that just to assemble that data might take a couple of years. So I'm all for not reinventing the wheel and doing something a little bit more creative, and I was thinking about this as part of the earlier deliberations between November and now as well as on an ongoing basis with the public health community.

It seems to me there are two things to keep in mind. One, the report is going to the HHS Secretary. So this group is going to ask, like we did in the morning, HHS to do something. So just keep that in mind.

All the agencies in HHS do a huge amount of training or funding for training. CDC does, HRSA does, NIH does, AHRQ. All of us are involved with that. Keep that in mind if you are asking the Secretary to do something that is a little bit different than what we are doing now.

The second thing is the various stakeholders and the need for a more literate public in genetics and genomics, whether the providers or the consumers. A case in point is selling the GWAS platforms on the street right now, the million SNPs or 500,000 SNPs. It is a great educational opportunity. You can focus on that as a way to say what does the public need to know about 1 million genetic variants. Some of them are already rushing to the Internet to order these tests, whether for recreational purposes or health-related purposes. The providers are not necessarily in tune with how to interpret that rapidly emerging knowledge.

One thing I want to say [is] the geneticists themselves, and I am one of them, have been trying to force others to learn our stuff. They keep telling us, we don't want to learn your stuff. It is not relevant in my day-to-day life.

So the geneticists need some education as well as to what other forms of adaptation they need to look at to take their tools and adapt them to a public or provider situation that is not very receptive until the time comes for decision-making or general education

purposes.

So I'm all for creation. I think this group can really sink their teeth in. Before we go on a big data collection exercise that could take us two years collecting what HRSA is doing and what the public health schools are doing, [we need to] think about these draft goals and say, at the end of it, what do we actually want to say to HHS.

If we end up saying to them what we said in 2004, sorry, that is not good enough because it sounds like a U.N. pronouncement. Whereas, whereas, whereas, do more, do more, do more. That is not good enough for me. I need more guidance from this group. Thank you.

DR. RANDHAWA: I just want to raise a different point.

DR. WILLIAMS: I think that Muin is on the right track, although I might frame it a bit differently. I think we need to be cognizant that in some sense our scope is defined by what the Secretary has ability to control, which means we really have to look at the activities that occur under the Secretary's aegis that are involved in education. That may be HRSA, it may be

CDC.

That is still a big thing, but at least it gives us an idea of the targets. In other words, I don't think we should be necessarily looking beyond that to create something new. I think there is plenty to do within what the Secretary has control over.

The second thing relating to Muin's point is that we have in the past, as a genetics community, taken a top-down approach. I think it has been very well exemplified by family history, where we say everyone has to take a three-generation pedigree. No, they don't. In fact, they will tell us very clearly what they really need to do. We have done very little in the way of actually developing a research, if you will, agenda to learn what is really needed so that we can match the needs with our expectations. I think that would be a highly valuable exercise to pursue.

The third point I wanted to make is again related to things that the Secretary does have control over and for which there is a great deal of enthusiasm. In fact, the only reason I agreed to sign on to this taskforce was the huge opportunity that we have right now

to deal with point-of-care, just in time education, to deal with the point that Muin just made.

They don't need to be educated until they need to make a decision, and that is where clinical decision support and point-of-care education can really provide that knowledge just in time to support the decision or provide the information that is needed. The efforts that are being done with a lot of energy and resources from the Secretary now relating to the AHIC, or now AHIC 2, plus what we have already discussed in the context of the Oversight Report, provide a huge opportunity to really leverage that new learning methodology and apply that that really wasn't, I think, envisioned much in 2004.

DR. FROSST: In response to Muin's comments about things that are ongoing and issues [about] direct to consumer, the 23 and Me Model. Here is your genome, here is something you have never thought about before.

Following to the idea that there are things that the Secretary can do, and perhaps this is a framework in which we can be thinking, I thought the taskforce and the Committee should be aware, and possibly are already aware, that NIH has formed a trans-NIH

communication group surrounding complex genetics and diseases and the 23 and Me and Navigenics, and is moving forward with an agenda of their own and a charter and things like that. Alan Guttmacher is the head of that. It would certainly be useful to coordinate those efforts.

DR. RANDHAWA: I have a slightly related but different point. I was looking at the different goals of this group and [specifically] Goal No. 6, which is on the effective educational tools that can be incorporated into electronic health records. I think this is a good goal to have, but what we miss here is, apart from identifying the different populations and groups that we need to educate, to also have a list of the different kinds of educational mechanisms of modalities.

This is just listing one, which is, I'm presuming, more electronic and decision support at the point of care. I think [that] is essential, but there are other kinds, whether it is Web-based, whether it is paper-based, whether it is person-to-person education. There are different kinds of modalities. It might be useful to have not only a list of the different populations but a list of the different modalities and

which ones would work better to educate which populations in which settings. That aspect really hasn't come out here.

DR. McGRATH: That is a great idea. Paul.

DR. MILLER: Two things. I want to associate myself with your comments about the deliverables and suggest maybe two ways of thinking about that. I think that is critical, particularly for an issue that, as others have said, has already been so much on the table and a lot of work has been done.

One is, in addition to going to the Secretary's office and asking them where they are with the '04 recommendations, I think it would be really helpful and useful to go to the stakeholder community and say, hey, there were these recommendations out there. What do you think the value of that was. What do you think was the value added. Get that piece, in a sense, from the community to say this was what we really needed, this wasn't. But, to get some feedback on that.

Then I would say to almost double back and think about, well, what is your end product. What do you want to come out of this, in a sense. Almost begin to

draft, given the amount of information that is already out there about the issue, what is it that you want to ultimately accomplish. Think backwards before you think forward. In that way you can begin to really tie in what your deliverables are with how you establish your goals and the path there from the very get-go. Begin to go backwards and then forwards.

DR. McGRATH: Just to make sure I got your idea, are you suggesting, once we identify the stakeholders, that we go to each one and do a needs assessment with each group?

DR. MILLER: Do it, I would say, in a more informal way. Say we are looking at these again, we had these things, what was it about the last process that you thought was really useful and really delivered something of value to your community or to what you do. What, maybe, sounded good but was a frustration ultimately, not because it wasn't implemented but just [because] there was no "there" there. Have that kind of conversation so, in a sense, you can leave that one paradigm and begin to fashion something new. Does that make sense?

DR. McGRATH: Yes, it does. Before we leave

this -- I don't know if we are leaving it or not -- we have this list now about five or six stakeholder groups. I first raised the question is this too many. I will raise it now. Is this enough. Are we covering the right people.

It starts with healthcare providers or professionals. I think we all know what that is. Then we have patients and consumers, and we have lay health educators; policy administrators, people who are involved in making those sorts of reimbursement decisions; and then governing bodies, for things like credentialing.

Is there any group that we are leaving off? Other than healthcare providers. So far that is how we have been thinking about.

DR. BILLINGS: As others probably know here, there have been specific educational programs, for instance for judges. There might be some lessons to be learned from those programs. For instance, taking on the issue of education for consumers is a huge bite and gets into public school education and a lot of things which we may or may not want to take on.

MS. AU: I think, though, for education of

consumers I would really just focus on the things that have been funded through HHS. One of the problems I have is they have this scatter philosophy where the different agencies fund different types of education all over the place and nothing is ever really sustained after the funding. This scatter education, from my experience, hasn't really helped that much.

I think that one of the things that would probably help the Secretary is the recommendation that there is more coordination because this is not going to get sustained if you are only going to fund small projects for a small amount of time.

DR. KHOURY: To continue this discussion, we are focusing on the "who." I think Gurvaneet mentioned the "how" a little bit. Electronic health records is a mechanism of a "how." I think the "what" is important, too. Phyllis mentioned that there is this NIH Communication Workgroup that is focusing on essentially the 1 million SNP chip, GWAS on the street. I think [we need to go] through the "what," "who," and "why." We are doing the "why."

Then there is this issue of "when" do you do

that kind of training in the life cycle from research to practice. Think about all the other activities that SACGHS has taken on: oversight, reimbursement, pharmacogenomics, large-scale population studies. In all of these things there is always an educational component that we say "See the Education Taskforce." We just said this this morning. That may be under the rubric of "what."

I think this Committee, based on the input of so many stakeholders, can come back at the end and distill those nuggets into actionable things. For example, if there is a need for the Secretary to create a taskforce around educating the public and the providers around 23 and Me, because it is a teachable moment and the public has the right to know, then it could be a focused recommendation at the end.

I'm very sensitive to what you just said about the scattering and non-sustainability of efforts within HHS. I'm one of them, so I can relate to that.

[Laughter.]

DR. KHOURY: You fund something for three or four years and then you either lose your funding or you

give up on the fundees and then you do something else.

This group has the opportunity over a period of a few months to take a look at the field, where we are and where we are moving, and give a coherent recommendation to the Secretary. We all look at these recommendations, even if we don't sometimes coordinate with each other, and we are no fools. We are saying what the action is. It is in GWAS now. It will be in proteomics next year, or pharmacogenomics.

But having, at the end, complete recommendations that can help us direct our funding no matter how small and then maybe push it in a certain direction that we may be missing right now.

Last but not least, one more point. Training and educating everyone is a big thing. Nobody can do it. The educators themselves don't know enough about genetics to educate others, although geneticists and others can benefit from principles of education and training. If there is a train-the-trainer type model somehow integrated into this, we can integrate those ideas.

Just a smattering of my thoughts here. Thank

you.

DR. TELFAIR: It goes back to a little bit earlier in terms of the need to look at the goals and be sort of discrete. I just want to bring to everyone's attention that Goal 7. This is actually a big area. I think we have had a lot of discussion around it. Everything that we have said on Goal 7 is very relevant to what was just said.

Going back to [the comment of] my colleague here about looking from basically what would be a public health end approach, which is looking at your outcomes and working your way backwards, but also looking at what is there and what is used, looking at the populations, [learning] from them.

I would just say that as we are looking at the goals, this is a key one to keep in mind as we begin to make decisions about what we can recommend and what is very doable in looking at the work itself. So I would say that is one of the issues as well as we are thinking about this. It does cover the "who," "what," "when," "where," and "how" sort of thing.

DR. McGRATH: I find that way of thinking, the

"who," "what," and "how," really useful. The "who" is the stakeholders, that I think we have kind of a list on. The "what" I think goes to Gurvaneet's comment on No. 6. If we enlarge that, that will tell us the "what." I'm not sure what the "how" is. Evaluation, to me, is kind of floating out there. I guess it covers everything, but that one is a little broad for me.

But it also seems like maybe we could start thinking about can we get a little more focused. We have a good list of stakeholders. We have been looking at tools. Maybe thinking about who the ad hoc members should be, or maybe talking more about this evaluation issue. That is the one I'm most vague on myself. Evaluate what? Are we assessing evaluation of educational tools, or for each one of the stakeholders are we going to evaluate the pros and cons of their methods?

Joe, do you want to talk about that one?

DR. TELFAIR: Yes. I think originally the recommendation on evaluation was pretty prescriptive. I think the discussion was that we can't be that prescriptive with it. Out of necessity the methodology

itself requires a certain degree of prescription. But since we can't do that, then that is what the compromise goal was, was to look at evaluative methods that would be used.

So the idea is that you do have, for example, different tools and different approaches to providing education to different groups. Taking Ms. Au's statement about the scatter approach, you have these things where things are funded and then they go away but you don't have evidence as to did they even work in the first place or not.

There are a lot of things that you can incorporate: how are they used or not, if it is or not used, how do you know or don't know. Those are key questions that drive a lot of this work. That is why there are so many different groups and subgroups and other efforts looking at this whole question of building the evidence, building some idea of what works best with different groups. There is just not enough of it there. We can make some pretty clear recommendations.

Evaluation will tell you it should be there from the very beginning, but this may be one of those

efforts where you may have to wait and make some early decisions about what exactly it is you want to do. Then you can start talking about what evaluative methods you might want to use.

So I would say it is vague now because there are still some decisions to be made, but I would suggest that it is going to be clearer once some of these other decisions that are being discussed are worked through.

DR. McGRATH: Gurvaneet and then Mara.

DR. RANDHAWA: Just to add to that point, I think apart from the methods of evaluation it would be useful to have a sense of the outcomes you are thinking about. Is it primarily increased knowledge, which may be very short term and then, after the intervention is done, a few months later, it is all back to baseline. Is it decreasing the variety of some psychological measures, which again may be short term or long term. Is it actually an improved decision-making at some level. Or is it actually health outcomes over the long term. What happened with the intervention subsequent to the original education.

This is probably going to vary depending upon

what context, what genetic test, and what scenario is being considered. But that would be a useful framework for everyone to focus at first on.

DR. McGRATH: I agree. Mara.

MS. ASPINALL: Two comments. First, in a context without comment on the specific comments previously, what time frame do we have to get this done?

DR. McGRATH: February 29th. No.

[Laughter.]

MS. ASPINALL: That makes it very easy. We can just sit down and be done.

I just think that it is relevant to the amount of time that we have to be able to even come close to the 10 goals. So I think, as I listen to this conversation, my head goes to, wow, they are all really important things and lots of [people] internal to HHS and broader would say these are great goals. If we have six months or a year or five years would change my estimate of how we would have to do that.

I just want to finish after you are done.

DR. TEUTSCH: I think that this is not on a crash project like we have just been through. This will

be on a more standard timeline for us, which probably means over the next year and a half or slightly more to have a completed report. I was going to ask Barbara to make sure of that.

The next meeting in July we actually get back having had the benefit of this conversation to finalize the charges, get the workgroups, get the ad hoc members that we need -- and if there are any that we need to add to the list we should hear them now -- and then tell us about the plans at the meeting in July. Then they will be able to talk to us about what next steps they are going to be taking to gather the information that we have just been discussing and the timeline for the report.

I think we are probably looking at 2010, or 2009, rather.

MS. ASPINALL: I had a year in the back of my mind. But if I say a year to two years, which amidst other priorities of the Committee is a long time but is not a huge amount of time given the reasonable resources we have, I'm going to go out for maybe some criticism here. I think it is very tough, if not impossible, to achieve all these goals in a substantive way. So I'm

going to suggest we look at those and really narrow them down to a smaller number without a value judgment that the others aren't good but to get to a more specific, achievable list.

DR. TEUTSCH: We would love to hear that. Do you all have some specific suggestions? What can be pruned or focused here? I heard some suggestions regarding those that are going to be germane to HHS that are actionable. I have heard a framing here that is going to be about who this is targeted to, who we are educating, some of the specific mechanisms, and then what we are educating about as ways to frame this. I would be interested in how we scope it down some more and then, of course, who else we need to have involved.

Paul and then Marc.

DR. MILLER: I don't have this information, but go back to say what has been useful and not useful in the past in terms of not doing the same thing but what kinds of deliverables are going to be relevant to the community to achieve certain goals. I think taking a look at that is also going to help winnow down not only what is possible but what is really going to be value added.

DR. WILLIAMS: This is a tool, not a prune. Not being involved in the previous taskforce, I think even within the last three years there have been some very good studies that have been done that address the issue that Gurvaneet talked about. It is not just getting them to learn it but to retain it. I think there has been some very good literature showing what really works in terms of retention. That is the tool that we should use to prune away some things to say this is just not possible.

DR. TEUTSCH: Any final thoughts? Scott.

LT COL McLEAN: I understand the desire not to get too detailed about exactly what you want to prepare, but I think there are a lot of people looking at very detailed aspects of education. I think we can serve them by giving more of a strategic plan for how to approach that and coordinate that.

The other thing that I would like to mention is that the other word in the basic charge is "training," which implies action, behavior as opposed to just knowledge. If we focus on education as just a knowledge status, I think we are going to be missing really what we

want to do, which is to change healthcare behavior.

So how you use that in outcomes, as Gurvaneet mentioned, to see exactly where you take that knowledge and what difference it really makes I think is really important for us.

DR. TEUTSCH: Last comment. Muin.

DR. KHOURY: This is all good, but I want to challenge the group here. I'm not the chair of this, but if everybody thinks very quickly about what is the first thing that comes to your mind -- this is sort of a psychological game here -- [as to] the most important priority in this field of education in genetics and genomics for the provider community and the general public. Take away all the other stakeholders. What first comes to your mind?

Given all the background noise of who is doing what, we can work through, of course with the help of other stakeholders, the "what" and then go see if the "what" is being delivered and what the success is.

I could say, for example, one of the "what"s is knowledge of family history in a decision support environment both for the public and the providers. I

could say your knowledge about interpreting GWAS data.

See, Marc, has a different list. What comes first to your mind?

LT COL McLEAN: From my practice where I do this every day, living longer, living better. People that don't die because they know what to do for their health and who can inform their relatives that they are at risk and prevent disease and prevent death.

DR. KHOURY: If we assemble that, we have a starting point that could focus our energy over the next year rather than just saying educate, train, everything is good.

This group has been working for three or four years now already. Think about your other reports and the priorities for genetics, health, and society: reimbursement, pharmacogenomics, research, all of these. We are not missing the big holes in this area.

DR. BILLINGS: I would just ask this task group to frame, as you look at your task of access, public awareness, and exceptionalism again, is there something specific about genetics education and health related to this which we ought to highlight, as opposed to things

that are true about risk education across the board or health education across the board. Similarly, about access to genetics education, which is specific about this, and the public awareness issues.

DR. McGRATH: That makes sense.

DR. BILLINGS: I would think you would want to use that as a potential frame to think about.

DR. McGRATH: I want to just respond to Muin. If we do that, that means realistically we are eliminating the policy administrator group, the governing bodies, and the judges and clergy, which I don't have a strong opinion about. I could see that group being handled in a different arena. Of course they are important. It doesn't mean we are eliminating them because they are not important, but that is the implication of what you are suggesting.

I would like to hear the rest of the taskforce. Let me know what you think about that. Or by Email. Joe.

DR. TELFAIR: I just have a comment. In the public health education arena itself, there is this whole concept of functional knowledge and what that really is.

What is the information that is really needed for you to be able to do these things, and in what context or contexts is that, which is more an ecological context, which means you engage those around you. The core of that, of course, is going to be the population or the group of people around that.

If you take the idea of what are the priority areas and set the priorities of what it is you can do, you still have to keep in mind, even in that context, what realistically can be recommended outside of this. What if we did that. What if we just focused. Can we come up at the end of the day with a set of recommendations that are doable, functional, and that can really be carried forth given the existing arena that you have.

If you look, there are commonalities across pretty much all these areas of what CDC is doing, NIH is a doing, to a lesser extent to what AHRQ is doing, and those sorts of things. There are commonalities across those things, but there is a context to put it in. So if we consider this, we have to say what is our priority focus. Then we can work our way out.

I would suggest that if we did this that we take it as a step-wise approach to do this, which is what are the two populations you really want to focus initially and, as we spread out ecologically, there are going to be those that we have to deal with. So if our focus is going to be on providers and clients, then we also say, okay, what is next related to them, which is going to be these other folk, the non-health person, the administrators of policy, or whatever.

What that would allow us to do is to take a more systematic, functional approach where these outcomes are related to that, but it still allows us to get where we are going. So that is just another way of thinking about it, but to me it is a little more doable and functional and you can really wrap your hands around that a little bit better than trying to take on the bigger picture.

DR. TEUTSCH: Absolute last comment from Jim and then we are going to move on.

DR. EVANS: I want to say I like the way that Muin phrased it. I think it is worth polling by Email, et cetera, the group. My response [is], I think we need

to take into account what Paul has brought up. Where is genetics and the need for educational aspects. Where does it differ from all the other educational aspects in diabetes.

What I come up with when I think about that is the understanding of probabilistic risk. That is such a paramount issue that, while not absent in other areas, has risen to the fore in genetics. It cuts across the various "who"s of stakeholders.

So we might want to think about identifying certain aspects that are highly germane to genetics and then let the "who"s fall out from there and focusing. Like Mara says, getting all of this done in a year and a half seems unlikely.

DR. TEUTSCH: I think we are going to task all of you to come back with a timeline and a plan for July. What I'm hearing is we are not talking about education broadly, we are talking about specific aspects that relate to the genetic field for these specific groups that we are talking about and that are actionable by HHS, hopefully well informed by what is actually out there that makes a difference. So thank you very much,

Barbara, for that.

I'm going to suggest that we not take a break. We had a late lunch, and I know you are all energized. I would like to move forward to the --

[Laughter.]

SESSION ON PLANNING FOR JULY PRIORITY SETTING

Future SACGHS Priorities: Issues and Planning Process

Steven M. Teutsch, M.D., M.P.H.

[PowerPoint presentation.]

DR. TEUTSCH: I think there is more coffee back there, so even if you are not energized you can reenergize. So, as folks need a break, I understand that you may want to take rolling breaks.

You heard yesterday from Reed about the major projects that have been completed and the many of them that are actually nearing completion. What I want to do now is to devote some time today to a brainstorming session on new priorities that might be appropriate for the Committee to take up.

The point is not to come to any final decisions today but simply to come up with a list of things that we need to consider, and then we will work through them

probably at our next meeting in July. So the goal today is really just to identify some of the issues that we might want to take up.

The priorities that we have been working under and which Reed went over with us in some detail yesterday were established through a systematic process that the SACGHS undertook in 2003 and 2004. In Tab 5 you will find a summary of the process that was used at that time, which in general seems to have worked very well. I want to walk you through my best understanding of what that was, since I wasn't there. Fortunately, I think a few people are still around the table who actually were there and can talk to us, and we will get some of their input on how it worked.

So what was done is initially the Committee members and the ex officios came up with a list of about 19 topics that were suggested during a brainstorming session somewhat analogous to what we are going to be doing this afternoon. A taskforce was formed to narrow down that list and investigate the remaining issues in preparation for a meeting which they had in March 2004.

Now, between the meetings the taskforce, with

input from the Committee members and ex officios, narrowed the list from 19 to 11, which is what you see there. In narrowing that list to 11, the Committee considered a variety of questions. I'm not going to read them all but the criteria, if you will, for winnowing down the list.

Four of them I want to highlight here, and the first one is, does the government have jurisdiction or authority over the issue. So, do we have an audience that we are supposed to be talking to. Second, does the issue that is under consideration raise concerns that only the government can address or would government involvement be duplicative of other efforts. Third, is another body addressing the issue or actually better equipped to address the issue. As we all know, we are only one of many bodies that advise the HHS. Finally, has a policy solution to the issue already been worked out. That is, do we have something to contribute.

Once the list was winnowed down from the 19 to the 11, issue briefs were prepared on each of the 11 issues that were selected. Hopefully, some of you had a chance to look in Tab 5 at those issue briefs.

At the next SACGHS meeting then, members and ex officios deliberated on each of the 11 issues and organized them into three categories. They were issues that required in-depth study, issues that required short-term action or monitoring, and overarching issues that would be considered within the context of all the other issues.

You will see on the far left, under the category of "Short-Term Action," the vision statement, or roadmap as it eventually came to be called, was written to describe the 11 priority issues, the reasons they were selected, and the process for identifying them. It includes the issue briefs that were prepared as background for the priority-setting deliberations, and all of that is in Tab 5.

Genetic discrimination, genetics education and training, patents and access, and oversight were categorized as issues requiring monitoring. The coverage and reimbursement, large population studies, pharmacogenomics, and DTC market were categorized as those requiring in-depth study. Then the overarching issues are access, public awareness, and genetic

exceptionalism.

The next slide shows the priority issues which were first established in 2004, and some of those issues have actually changed categories, the ones that are there in blue. For instance, oversight was initially categorized as requiring monitoring but was subsequently elevated to requiring in-depth studies.

So in brief, that is how the priority-setting process was done in 2003 and 2004. Before we jump into all of this I would like to ask for a few volunteers to actually serve on a committee that is going to help work through this process so that some of you can take very close notes as we go through the subsequent discussion.

The purpose of this group is to continue the brainstorming on the issues suggested today, to develop a plan for the priority-setting process, and to identify what we need to do in terms of materials and background for that July session. The point is, get folks identified so you can be particularly vigilant as the ideas flow this afternoon.

So, could I get some volunteers to serve on that taskforce? Paul, great. Paul 2. Paul the Lesser,

Paul the Greater. Jim and Mara. Ex officios are fine. I got Muin and Gurvaneet and Mara. I think that is a pretty good group.

Anyone volunteer to be chair of this group? I will participate as well.

Now, I think it would be helpful to actually hear, how many of the folks who actually were on that planning process back in 2003 and 2004 are actually in the room? Some of the ex officios actually were there.

DR. FROSST: I was in the audience taking copious notes, but yes, we were involved.

DR. TEUTSCH: Perfect. Muin, were you here for that?

DR. KHOURY: I think I was involved in the group, but --

MS. ASPINALL: Muin has the tenure.

DR. KHOURY: I'm the longest-serving person at this table.

DR. TEUTSCH: Anyway, since no one is willing to volunteer prior experience, I would like to open the discussion to the rest of the Committee just in terms of the process because clearly we could follow roughly the

same priority process that we did before.

Now Muin has a comment.

DR. KHOURY: The reason why some of these things are on the board is sort of a gestalt of what was happening back in 2004. For example, the large population study was a very timely issue that Francis Collins brought to the table because of the desire for NIH to do a large cohort study, which now has been rephrased.

DR. FROSST: For the U.S. to do one.

DR. KHOURY: Yes, for the U.S. A big cohort study around gene and environment. Now, that led to an in-depth analysis and now there is an initiative called GEI, the Gene-Environment Initiative. So there has been some movement. That was brought to that table because of the fed bringing that to the table.

The direct-to-consumer marketing was brought to the table because --

DR. TEUTSCH: Muin, I think what we really want to know is just about process right now. We will walk through some of the actual topics and what is on the table as a holdover. I think the question is, is that a

process that we are comfortable with in general or are there other suggestions about just how we are going to sort through this in general.

The process was the generation of the brainstorming list, forming the committee to winnow that down, developing issue briefs. Then there was actually a voting process in there. The staff developed these, and they are in your folder so you get a sense of what they look like. Then, bringing that back to the entire committee to review and make some decisions about.

Yes, Mara.

MS. ASPINALL: I have a question. It sounded like it was somewhat part of the '04 piece, but shouldn't we ask the Secretary and his staff what priorities they have?

DR. TEUTSCH: Right. I'm going to walk you through at least some of the ones we have gotten from the Secretary. I think Greg is here and he can help us with that.

MS. ASPINALL: What a perfect setup.

DR. TEUTSCH: I think that there are issues like the issue briefs, which are a lot of work. Is that

something that is going to be helpful for our process going forward. That is the kind of thing that would be very helpful to get people's sense of. What were the important parts of the process.

MS. ASPINALL: Right now or for the next meeting?

DR. TEUTSCH: What I'm asking is, as part of a process for the priority-setting would you like to see those done again, realizing the work that is involved. Barbara?

DR. McGRATH: I know when I came on board you gave me the book of those, and that was critically important for me to read just to get a sense of the philosophy of the group, what was on that list and what wasn't on that list. So I think they are great, if we have new ones coming up, to have that similar sort of background.

MS. ASPINALL: I would agree. I think they are great. I actually think they are sufficiently good that I don't know if they could be useful in getting out more broadly to some of the public or stakeholders on some of these issues because I think that they are a great 80/20.

They are not the comprehensive but they really, with a little bit of effort, outline the issues well enough that I would love to look for opportunities to use them more broadly.

DR. TEUTSCH: I think the Vision Report, which actually included those briefs, which was made available publicly, was done probably for a couple reasons. One is to let the public know what we thought we were trying to do, but also then to share that information broadly. So the extent we want to devote the energy to a similar exercise, that is one of the things that could be done.

Maybe it would be helpful, before we actually start brainstorming, to again look at the original priority issues, which you see up there. We have already completed the report on the vision statement, coverage and reimbursement, the large population studies. We are about to complete the report on oversight and pharmacogenomics. We have written letters to the Secretary on genetic discrimination and direct-to-consumer marketing, both of which we continue to monitor. We wrote a resolution on genetics education and training of health professionals, which we just talked about and

just created a new taskforce to revisit the topic, and we are in the middle of our work on the gene patents.

Our newest priority, which we would call here "evaluation," which is what Reed explained yesterday, which really was looking at translational and economic issues, was added in March 2007 but was put on hold at that time because of the need to get on expeditiously with the Oversight Report.

We know that the IOM Roundtable on Translating Genomics-Based Research for Health is considering this issue as well.

We also have the three overarching issues of access, public awareness, and genetic exceptionalism.

So the genetics education and patents will remain on our list. I think one of the things for us to think about, then is are there any others that need to be revisited or require ongoing monitoring, and do any of the overarching issues actually stand on their own as stand-alone issues. Do we need issue briefs updating us on evaluation or on the work being done by other bodies.

So we will think about those as we go through. Let me turn now, partly to answer your question, Mara,

about suggestions for new issues. We have received several suggestions already, and the Office of the Secretary provided the following topics for us to consider. Let me at least put them on the table.

One is on international genomics infrastructure for clinical research. The next is on primary care practice-based approaches for the integration of CME, curricular and medical boards. Third is on clinical research standards for biospecimen collection. The next one is on economic and diagnostic value of multiplexed genomic tests and how are costs integrated into the commercial development plans to determine what factors the developers actually use to assess value, and co-development of molecular targeted agents and diagnostic biomarkers.

I can read them very well, or at least I can read them. Greg, we just ran through the list here of the ones that were contributed by the Secretary. Did you want to add any additional comments on any of those? I think one of the things we want to do today is get some clarity on what all of these things mean.

MR. DOWNING: I think, as opposed to how the

oversight issue came as a specific charge, these were ideas in the journey of looking across the whole spectrum of personalized health care related to the initiative as to where further policy direction and activity the Committee might have some value.

There are aspects of this that I think touch on some of the other ongoing activities here. So we don't want to necessarily influence and move off any of the priority areas you are developing, but I can talk quickly through some of these if you would like.

Some of them reflect on some of the large population-based needs and resources for developing further research infrastructure, capabilities, and knowledge on some of the aspects of genome-wide association studies, for example, the aspects of biospecimens, and the characterization of them and the classification of them, the starting materials. The clinical resources behind the sequencing projects themselves has been something that has been pointed out to us. The Secretary was recently abroad in a number of countries and has brought back to us some specific follow-ups to ongoing international efforts.

It has been a while since I have really gone back through the records of what this Committee has done on international genomics projects, but it seems to me with the scope and the breadth of where a lot of the genome-wide association projects are going that there are multiple opportunities for international partnership. There may be within the agencies themselves enough activity ongoing there that this isn't something that has to be dealt with here.

But in terms of data sharing and the aspects of sharing the specimens and the characterization of the specimens, those are some of the things that we have stumbled across as being some obstacles that perhaps this group may want to weigh in on.

The extension of that to the clinical applications of genetic testing in other countries and that infrastructure and capacity is something. We are often contacted about where can health officials from other countries go to to find out more about how these are being integrated into clinical practice.

So if I had to say of the new things that he has brought to us recently, it has been the international

domain of our context of this, not from necessarily just the research side but also into the translational side of it.

Can you go back? It has been a couple weeks since I saw these. Thank you. I apologize. I haven't followed specifically the one activity that you and Gurvaneet had tag-teamed last fall on as a potential area that relates more to the evidence development processes. A critical need that we see is that there are pieces falling into place about the aspects of being able to foster projects that support the evidence development. Not necessarily analysis of the evidence but where does all the evidence come from to support the clinical applications of these. That is a broad-ranging and complex issue but it is a big hole, as I'm sure you know from your experiences.

Then I think the primary care aspects. Some things that we have discussed offline with some of the staff are that the specialty areas have been dealt with and the educational components to this, but in terms of where we hear, frankly, a large amount of interest is from the primary care communities. They recognize they

don't need to know all of the specifics but like to be able to start to plan to integrate from a preventive health perspective or be able to maintain current knowledge of understanding how to integrate various types of genetic tests into clinical practice and primary care.

I think Marc has shared a great deal of interest and information with us as well, so I think this is somewhat consequent with your last discussion.

We were asked to provide comments about directions and new things that we had seen through the Secretary's Initiative that might bring some interest to this Committee, but I want to be very careful to characterize these as not being of the same nature of the genetic testing oversight issues that we have brought forward.

I would be happy to elaborate.

DR. TEUTSCH: Why don't you stay here. First of all, are there some questions or clarification?

DR. WILLIAMS: I just think that as we were thinking about pruning the tree in our last discussion, Bullet No. 2 here would seem to be responsive to that particular thing and should fall under the aegis of the

Education Taskforce.

DR. TEUTSCH: I think we are going to need to do some consolidation. We will get back to the taskforce group. But, are there clarifications of at least the issues that the Secretary has at least raised? Yes.

DR. FERREIRA-GONZALEZ: Greg, for the clinical research standards for biospecimen collection, NCI already has an initiative. I know it is oncology-oriented, but they already have an initiative to work in this area developing these standards. Do you know anything about that?

MR. DOWNING: Yes, probably more than I should.

[Laughter.]

MR. DOWNING: Nevertheless, we have been off to a number of communities with a number of different approaches to integrating genomic information into primary care or into health care overall.

One aspect of this that keeps rising to us are, great, we are working on all these health IT standards and all these other aspects of it, and recognizing that they are professional organizations as well that are behind this, but in terms of characterizing where does

the genetic information come from in the literature, if you go back and find these studies there is not any way you can identify whether the lineage of these specimens and so forth has been credentialed.

I'm making this up on the fly, but the aspects of being able to say does this information come from a bona fide source of annotation and so forth. I'm not a clinical pathologist, but is this really what it says it is.

I think some of this is addressing areas where recently a lot of attention to characterization of other cell lines and others has come into question in terms of some standards aspects to it, being able to go back in the literature and say, well, I have the same cell line, I can't find the same pattern of polymorphisms that somebody else has reported in Paper XYZ.

I think that this goes in the category of accountability and the ability to trace this back to where are the levels of competence you have that that really represents a patient with that condition and it is reproducible.

I'm not suggesting that this is a new area that

requires a whole lot of attention and focus, but it is something that we have been seeing rising, particularly with a lot of the clinical genomic characterization studies that are ongoing. A lot of standards efforts have gone into the technology and now into the health IT side, but the biospecimen components themselves still wax that.

Some of the attendant issues related to that are, if you are having a federally funded study for that support, some of that specimen goes back into a common repository where, if for some valid reason one needs to go back and reidentify that particular gene sequence or the associated genes with it, that there is a mechanism or a way to go back and validate or verify that.

To some extent, some of the stem cell work parlays over into this a little bit in the ability to say, do we have the identity of this particular specimen or cell accurately portrayed and it is reproducible.

DR. FERREIRA-GONZALEZ: This is a little bit different focus than the NCI activity because the NCI activity is more how do you qualify the quality of the specimen. Here it is the information. Those are two

different issues.

MR. DOWNING: I agree, and they are probably both valid areas to explore. This is, I think, probably in a bit higher domain of being able to provide a credentialing process. If you are going into a large public database and you tell me it validates that that specimen is truly where it comes from and is associated with that particular condition.

So I don't think we are thinking of any huge, elaborate study, but I think there is a fair amount of effort underway and a lot of communities are saying, what are other groups doing around this area that help with standardizing or credentialing the tissues that are being utilized for these very comprehensive studies.

DR. TELFAIR: Thanks. I just have a question. It is two slides up. The other way. Forward. There you go. This is a question I have to get an understanding of the priority in thinking here. I thought that is what we were trying to do.

DR. TEUTSCH: There are a couple of these we haven't gotten to. Let me run through the rest of the things that are on the table, at least that have been put

here, and then open it up to everybody else to put them on. Greg, if you can stay, we can benefit from clarification.

MR. DOWNING: I'm going to probably have to leave at a quarter after for a meeting.

DR. TEUTSCH: Then let me ask, are there other questions directed to Greg in terms of clarifying what the issues are?

DR. KHOURY: A question to Greg. The international arena is very important. There is so much stuff going on with genetic clinical research and population research and biobanks around the world. If this group spends a lot of energy dealing with this, would the recommendations that go back to HHS be useful? I guess they are because HHS has recommended for that to be at the table.

But the question to you, Greg, is in what way does HHS want to learn a lot of these informational efforts and what kind of advice can SACGHS give HHS along those lines?

MR. DOWNING: The aspects of what have been done in terms of broad international basic research

projects overall I think have been well noted in the annals of science now in terms of how communities came together largely through consortium efforts and basic discovery research activities. I think where we have been seeing some of the gaps are, if there are difficulties in acquiring specimens or tissues or whatever, that these resources are recognized as another opportunity to go to.

I don't want to say that we have completely identified a specific target in here, but in the focus of translating this into clinical and medical practice the meaning of this information, looking at different populations in different countries, there is, I think, a lot of interest that comes to the Secretary. His overall interest in this for many years now has given him a little bit of a compass in terms of how you put together communities to do this in more and more overarching ways.

So I don't think that there is a particular challenge or a problem here. It is the aspect of being able to share information, recognizing what the challenges are in other communities that may not have the same breadth and depth of the science infrastructure but

are still very interested in the whole aspect of taking what is basic science now into solving major clinical challenges that may not necessarily be ones that we deal with on a high priority basis here but may have some relevance in other countries.

It may be, from the standpoint of looking at what are priorities in clinical research areas of applying genomics in other communities and countries, that that may be the opportunity for further collaboration in research. Maybe the challenge is introducing these in regulatory frameworks in other countries and how they accommodate this into their healthcare delivery system and approaches like that.

I know you have taken great care in looking at the regulatory side of this, but in terms of the health care practice side of the applications of genomics, genomic tests and so forth, there are still a lot of questions from many other communities about this.

We have had contingents recently from Japan and China who have been very interested overall in the population-based health approaches to health planning around several of the genetic tests and capabilities

around certain types of cancer and cardiovascular disease.

So the emphasis here is really looking at are there ways to share common approaches in clinical research of genomics in broader communities and different contexts than what we would do just here in the U.S.

So I don't think there is any particular agenda here, just recognizing that the way health care is practiced in many parts of the world is different than here. The capabilities of doing clinical research in many of these countries to solve some of their health problems is not as well developed as it may be in this country. There are probably some areas that we can benefit from in terms of looking more broadly.

I don't think we are trying to shape a new research agenda here. It is the aspects of understanding how different cultures are perceiving the integration of genomic technologies into health care.

DR. TEUTSCH: Marc.

DR. WILLIAMS: Adding onto that, you mentioned research specifically, but we have also identified in different venues international issues relating to

availability of rare genetic tests and the CLIA certification implications of that transfer of biospecimens across international borders and issues like that. Is that something that could be considered under that rubric of international issues?

MR. DOWNING: Yes. This was really meant to be a very broad characterization of one area that we thought transcended one particular agency in the Department and was something that we hear a lot about. The Secretary travels a lot, and this is one of the things he has brought back to us to further investigate and develop some approaches.

It could be as simple as identifying issues and leave it at that, but at the same time there may be some ways to use his leadership in some ways. I know from his meetings at the G8 this summer and at the WHO level there have been detailed discussions around the aspects of where genomics is moving in terms of health care. So I think, to this extent, if there are notions or directions about that, that would be helpful to him in operating either in leadership capacity or partnerships or addressing some of the technical issues that are coming

up relevant to advancing research areas. That would be helpful to us.

Robinsue can help me here, but I should mention Bill Steiger's office at Global Health Affairs has been very much supportive of our efforts here in the Personalized Healthcare Initiative overall and frequently asks us to meet with the science authorities from other parts of the world. We have not done a careful analysis overall of what people are doing, but we recognize that this is an area that this body may be able to help out in some ways.

DR. TEUTSCH: Mara.

MS. ASPINALL: Just quickly as I'm cognizant of the time, when you talk about codevelopment of molecular targeted agents and diagnostics with biomarkers, there have also been a number of studies there. Is that in search of, and I put the last two together, personalized health care and how the next generation of drugs can or should be linked to diagnostics?

MR. DOWNING: I just, in the last several months sort of stumbled upon this notion of nomenclature about codevelopment can mean a lot of different things at

various stages of drug discovery and development through various clinical phases of development and into basically companion diagnostics and therapeutics. We have not really been able to find a very good characterization of what that means at each of those stages and phases.

I'm not suggesting this gets into the aspects of developing guidance, but FDA has indicated that they are interested in developing guidance in this area.

But the attendant issues of how a particular test is used and the processes of looking at the test and the drug during its life cycle through development and clinical trials areas, what does that look like and what are the characteristics of that. What are the requirements of regulatory submission. If you intend to use that test along with the drug in the clinical practice overall, how do you design studies and so forth to do that.

This was an area that we put in here because we understand there is interest within FDA overall. Part of our report last year dealt with the areas. There is substantial industry interest in this as well. We haven't had a detailed discussion with FDA leadership

about this yet, but we haven't found a lot of background information about what that process looks like and how companies think about it. There has been relatively little discussion to which we can really point to say here is what the future might look like in that kind of context.

MS. ASPINALL: I'm wondering in the context of almost combining it with the one ahead of it because many of these tests today may be multiplex tests. Part of the issue is looking at the relative value of the test versus the therapeutic and how they combine. It is a new field.

MR. DOWNING: Right. I think we were looking at the steps of this [being] the extension of a biomarker into a laboratory test that ultimately might be used in clinical practice. So that is one of the aspects of why we wanted to really focus on how do you design your R & D components to this, if you will, if your intention is to ultimately apply that marker as a test in clinical practice.

I'm sure they can be combined, but I was trying to distinguish the two approaches.

DR. TEUTSCH: Greg, we know you have other

commitments, so thank you. I don't know if you were here this morning to know that we got approval of the recommendations for the oversight test, so they will be headed your way.

MR. DOWNING: Yes, the lights were flickering down on the fourth floor.

[Laughter.]

MR. DOWNING: I was waiting for the lightning bolt to come. All I can say is we have had some internal discussions about how we would embrace what you have to share with us. We realize what we have asked of you over the last almost year now and have been very impressed with the degree of engagement and the thoughtfulness of the Committee members' comments. We look forward to reviewing those.

DR. TEUTSCH: Thank you. We know they are going to fall on receptive ears and an enormous amount of effort has been devoted. So we will be interested in following up.

MR. DOWNING: Thank you.

DR. TEUTSCH: Thanks so much for your time.

MR. DOWNING: You're welcome.

DR. TEUTSCH: Before we open the floor, just two items, just to be aware there are other things that have been put on the table as issues. At the end of Tab 5 is an article from Nature Reviews Genetics which actually specifically requests that SACGHS provide guidance and set standards for determining what data from whole genome sequencing should be included in electronic health records.

Second, just to remind you that we had anticipated having a meeting in July to deal with the whole issue of personalized genome services, which has gotten a lot of attention. We heard a little bit about it yesterday from the public comments. As you know, several companies -- 23 and Me, Navigenics, Gnome, and others -- have launched direct-to-consumer genomic screening and analyses, which raises a whole variety of policy issues that we may want to address that are within our charter.

Since they touch on so many critical issues, the staff has actually proposed having a half-day session in July that will help us learn more about these companies and their services. So I would like to think

that that will be on our list of things that we may want to address.

I wonder if I could ask Sylvia, would you mind working with staff to help organize that session? That would be very helpful.

MS. AU: Volunteers? No.

[Laughter.]

DR. WILLIAMS: I apologize because I'm going to have to disappear for flight reasons. But if the floor is open for a topic?

DR. TEUTSCH: Right. I know several of you have to leave, so I would love to get the ideas. I know Paul has an early flight. Why don't you get yours on the table and we will try and get them down as best we can.

DR. WILLIAMS: Very quickly, I think that we need to explore the reality of the $1,000 genome and the impact that that will have on what we consider to be a genetic test. It is painful to think about just after going through all the oversight, but the reality is that if the price point comes down for a whole genome then genetic testing becomes an informatics query. It does not become a lab test.

As much as we have already stretched the 1972 regulations on devices, this may well prove to be the camel that broke its back. So I think that would be an interesting thing to do at least a future scan and topic on.

DR. TEUTSCH: Great. Paul, I know you have to [go.]

DR. BILLINGS: I have a couple. One follows directly on from I think what Marc is getting at, which is we have seen already in the last couple of days talking about what is health-related genetics and genetic testing and what is cosmetic, recreational, ancestral, whatever you want to call it. I would think that a paper or an activity from this group at least fleshing out how technology is driving the changes in that and just the whole idea would be a really interesting thing.

A couple of other things occurred to me. One is, it is striking that we see no discussion anymore about gene therapy. When I was first coming up to Washington, there were any number of groups that were meeting on that issue. So it does seem to me that some update, reevaluation, whatever, in that area might be of

some interest.

The other issue that was on my list was, again, the translation from our work on pharmacogenetics and genomics to the whole concept of personalized medicine and what is the yin and yang or the pro and con or whatever of personalized medicine.

DR. TEUTSCH: I think these are great issues to put down. I think there are lots of them that actually go on, whether it's genetically modified organisms, whether it is about stem cell research. All of these things could be within our purview. There are other committees that have obviously worked on them. But I think we should get them all on the table now so that we can begin to sort through them and figure out whether we are the right place to house them. So, think broadly.

Let's throw it open widely. Julio, Jim, Mara.

DR. LICINO: This was said before, but I think maybe we [can] make it very specific. I'm just [suggesting], either at the discussion that we are going to have in July or as part of these recommendations, essentially the interface between the informational tool that people are going to be getting more and more, which

you described, and your medical records. When should those things go to your medical records, who decides if they go to your medical records, can you place them in your medical records yourself.

The way I see it, I think that some of that information may end up in people's medical records not always completely voluntarily. If you are a prisoner, if you are in jail, if you are in the military, your DNA is collected. If you are at school, if you are a missing person. No, seriously, because that is how they find missing people now. You start to query and they can be described.

So this interface between law enforcement, informational things, and your medical records. How is that looked at.

DR. TEUTSCH: The privacy and controls.

DR. LICINO: Where does that happen.

DR. FROHBOESE: Directly following on what you just said, the idea of privacy, privacy of genetic information in an age when more and more of it is out there for more and more uses. I'm not sure if privacy as a big concept fits with what you were just saying or with

other issues that we have come around, but it is an issue that has been around for a long time and is not going to go away, and I think is only going to get more and more intense as more of our genetic information is out there in more sources.

DR. TEUTSCH: Jim.

DR. EVANS: I have been struck in the last three years on the Committee in how often the issues that we address are directly affected by the structure of healthcare delivery and specifically the fragmented structure of healthcare delivery in the country.

The most explicit example of that was in the Large Population Study, in which we stated that one reason that dissuaded us from more firmly embracing that idea was our system of healthcare delivery, to use the word "system" generously, and moreover to achieve the benefits of something like a large population study we needed a less fragmented system.

I think that there are important aspects of genetic medicine that inform us about the type of healthcare delivery and healthcare reform that this nation should be pursuing. I think that is an issue that

is very much on the radar screen of the public right now, healthcare reform.

I laid out in an article in JAMA about a month and a half ago at least some of the details about how genetic medicine influences what we should be seeking as we engage in healthcare reform. Since that is likely to be a national topic, and clearly the executive branch has a huge role in that, I think we should consider addressing what aspects of the emergence of genetic medicine can inform the Secretary in the directions we should go.

DR. TEUTSCH: Very good. Mara first and then Muin.

MS. ASPINALL: A couple things. First, let me add to the structure of healthcare delivery. I think we need to add to that the economic incentives that are related to that system and how that drives care or lack of care both in a public health perspective and an individual --

DR. EVANS: Relating back to genetics.

MS. ASPINALL: Exactly. Relating back to genetics and to personalized medicine, which is a lot of

overlap but not entirely overlap.

On the one that was mentioned before that in terms of privacy and control of genetic information with the medical record, I guess I go a little bit more broadly. I guess I think that is the secondary bullet. The first bullet is what do we need in electronic medical records that is related to genetics. If somebody came down and said, as there was legislation only two years ago, this is the standardized federal health EMR that exists, what would our answer be? What would we want in that EMR in a rather specific way. At some point somebody is going to ask or somebody is going to create it.

Given that it has been talked about here in a number of different areas that the EMR itself very well may be the key organizing mechanism for all of us as a society and as individuals, I think we should give an opinion for what goes in there when you are putting that together. So, what is needed in the EMR in terms of genetic information.

No. 3 is related to Greg's point about drug and diagnostic interaction. Some studies would say 60 to 70

percent of drugs, particularly in oncology, in phase 2 or phase 3 trials today will require some sort of not inheritable but gene-based information in order to personalize the use of that drug. How do we believe that system should be set up in order to ensure that those tests and companion therapeutics are indeed used appropriately and as necessary.

DR. TEUTSCH: Great. Muin.

DR. KHOURY: I'm going to use sort of a genetic type called The Roadmap Less Traveled.

[Laughter.]

DR. KHOURY: We are into finding genes these days, and lots of energy and resources of the government, with GWAS and others, are geared to finding genes. Test developers want to put them into diagnostic tests and move them through the oversight as quickly as possible. We have discussed the whole morning how we do that or how it is currently done.

In spite of all of this, there is this big gaping hole after you develop these tests, which is sort of that first step on the roadmap. I call it T1. You develop that promising application and what to do with it

afterwards. In other words, it is clinical utility and how do you go from there to the development of evidence-based guidelines so that third party payers can pay for it. It is a big gaping hole.

I think Greg mentioned a little bit about that, although it didn't make it on his list. He said, how do we stimulate that kind of research, not the data gathering we are doing as let's say part of EGAPP, where we are looking at the information and saying there is not enough information on clinical utility in the real world to make it worthwhile covering.

That is T2 and beyond and is such a big hole right now. We can oversight to death if we want to. We can develop registries. We can get the FDA to do their thing. But HHS, maybe in collaboration with the private sector, [needs to] begin to invest money in doing the kinds of trials and observational studies that would allow us to evaluate the utility and the validity of a promising application so that we can develop those evidence-based guidelines that we all need so that they can become integrated into health practice.

This is sort of the rubric under which this can

go on, The Roadmap Less Traveled, or it could be The Road to Outcomes Research or some other conglomerate depending on what other ideas come in.

DR. RANDHAWA: I would suggest three topics. The first one is, it may be useful to have a white paper on research priorities for pharmacogenomics. The premise of this is there are hundreds of drugs available, hundreds of genes that may potentially predict hundreds of different outcomes. So I don't think it's feasible to do outcomes research on every possible combination of drug, gene, and outcome. Is there a way or are there criteria we can think of that would help prioritize what are the high value, high target pharmacogenomic applications.

For the other two topics I will take Steve's comment literally about thinking outside the box. One would be genetic modification labeling of food, and the second would be genetics and cloning.

DR. TEUTSCH: Genetics and clothing. Oh, cloning or clothing?

DR. RANDHAWA: Cloning.

DR. TEUTSCH: Here we have genetics and

clothing.

DR. RANDHAWA: I meant cloning.

DR. TEUTSCH: Paul, you had some things I know you wanted to put on the table. Do you want to raise them now and then we will get to Scott?

LT COL McLEAN: I have seen a paper or two recently about genetic screening for populations beyond the newborn period, either mid childhood, adolescence, or adulthood. I think Beth Hutcher wrote a paper about that, which is an interesting topic. I mean, there is an entire advisory committee for just newborns. If you move that into other populations, it would be very interesting and raise a different set of problems.

DR. TEUTSCH: Paul.

DR. WISE: I have two. One that I would put as a subconcern under the issue that Jim raised about health care reform and that is the implication of new genetics for relationships over the life course. What we are seeing is new genetic insights creating new precursor conditions that never existed before.

Basically, any genetic predisposition for an adult-onset disease is automatically or perhaps a

pediatric concern. In other words, The New York Times article about 23 and Me ended with the reporter saying "I'm going to get this for my six-year-old daughter."

There are a lot of issues that are raised. However, the structure of health care in the United States is in no way capable of facilitating appropriate response to those kinds of insights. In other words, Medicare doesn't talk to Medicaid for Kids. Insurance programs that have 30 percent turnover per year have no incentive to look long-term at preventive or precursor management. FDA will have new issues about extending the use of medications into precursor areas that really have not been examined. You begin to look at the whole structure of healthcare delivery and assessment in the United States over the life course and things begin to break down very quickly.

But I like the framing of this as a subset of the broader issue of the implications of the new genetics for health care reform.

The other issue that I would like to get on the table, and I know that it has come up here earlier, is just the implications of the new genetics for minority

health. There are equity issues that are of special status that are captured by the excess uber-concern that the Committee has because of the special history genetics has played in these communities, the special requirements for the provision of genetic services in these communities, but also because public discourse around minority health is heavily influenced increasingly by genetic insights.

It is not sufficient to talk about this merely as a reimbursement issue or an access issue. It has special requirements because many would feel that the justice requirements would insist that any technical capacity to improve health, policy should respond in ways to preferentially address the needs of the neediest communities. It may require a special concern being voiced and examined by this Committee in this regard.

DR. TEUTSCH: Barbara.

DR. McGRATH: Exactly. A follow-up on that, or ditto that. Two little issues with that. One is that in most of our reports we make some statement that the new genetics decreases health disparities. We kind of use it as a phrase, and I'm not exactly sure how we should

address that, but I think we should look at it a little more deeply and not use it as a throw-away.

The other phrase we use a lot is "genetic exceptionalism." We all agree there is something to it. You mentioned probabilistic risk. But by making it an overriding theme, we somehow aren't addressing it. It is very ephemeral. I'm not sure what a taskgroup for us would look at that, but I think we have things like stigma discrimination, probabilistic risk. We have enough features on it that it would be useful, I think, for us to figure out a way to address some of those.

My third little thing is a question. When I came on the Committee, the issue that was most in the press that I saw was stem cell research. I was kind of surprised that that wasn't talked about at all here. This is my question: is it not under HHS? It is less of a topic now, but it certainly is in the press a lot but not much in this room much. That is a question.

DR. LICINO: I think the stem cell discussion is very important. I'm going to skip that, but just going back to --

[Laughter.]

DR. LICINO: I think if we had something like on, let's say, genetics health diversity and health disparities, it would be very nice. I think it may impact on different groups very differently.

DR. TEUTSCH: I wasn't here to tell you exactly why stem cell was or was not on the agenda, but it seems to me that is something that we need to put down here for at least consideration going forward.

MS. CARR: Can you be more specific about that, though? What would you put on that? What is the issue?

DR. McGRATH: What is HHS's policy on promoting stem cell research. That is why I'm not sure if it is under HHS or not.

MS. CARR: The other thing that I would point out [to] this Committee is that the focus of SACGT's work was more in the healthcare arena and public health than in research. Large population studies was certainly a research topic and certainly research topics are part of your charter, but I think because of the many, many opportunities and needs related to the integration of genetics into health care, it seems to me that that is why that has been such a focus.

I will say on that I think there are other mechanisms to receive advice about that very important issue. Not to say you shouldn't [think about] this because that is what this is, throwing things at the list.

DR. TEUTSCH: Denise, did you have something earlier on consent or something related to privacy that you wanted to raise?

DR. GEOLOT: As we talk about privacy and population studies and the use of genetics, you have the whole issue of informed consent. Even though there is another advisory committee that focuses just on informed consent, it might be a good idea to explore meeting with them to see what their views are in terms of informed consent with children and with regard to genetics and broader issues.

DR. TEUTSCH: Sylvia.

MS. AU: I want to add to that minority health issue. For minorities it is not only health issues, there is identity. There is a lot of movement towards genetic testing for ethnic background. So there are a lot of other issues besides health-related issues for

minorities and genetic testing. I would like to make sure that that is included under that topic, too.

DR. TEUTSCH: Mara.

MS. ASPINALL: I apologize. I have to head out. Maybe it is implied in a number of these, and it reminded about stem cells, but to deal with new and evolving technologies. We talk about a number of things in the context of today, but how to integrate new technologies, whether it is proteomics or the genome piece. There are some specific ones, but there is going to be a series of new technologies. Each one can be evaluated in and of itself, or is there an overarching mechanism that says how to recognize a new technology and how to integrate it into a system.

DR. TEUTSCH: Do you have a comment on this issue?

DR. FITZGERALD: Yes, just following up on what Mara said. Again, if you are going to go into that and even some of the other things that have been mentioned, this is the Committee for Genetics, Health, and Society, but as we all know, what is genetics, as we have just seen in our last thing on genetic tests, is being

blurred. Even to go back and review the vision statement and the terrain of this Committee, you are going to say, what if it is an epigenetic issue? Does that fall under SACGHS or is that something else?

That is something I think the Committee is going to have to wrestle with at least a little bit because we are going to keep coming up against this genetic issue.

DR. TEUTSCH: What is it, right?

DR. FITZGERALD: What is it.

DR. TEUTSCH: Yes?

DR. DAYNARD: Just to build on something that Paul the Wise had mentioned, perhaps the idea that the aged are a separate community, a minority community, whose special needs as far as health care and the impact of genetic testing and genetic techniques on that population might be worth considering as a separate population sub-issue.

DR. TEUTSCH: Are there others? I see we are gradually losing our critical mass. I want to run one thing by you. One thing I think I would like to do, and I just want to get your concurrence, is to actually

direct solicit the ex officios. Within their agencies I think there probably are some specific issues that they are facing that we should elicit. I just wanted to see if that was consistent with where you are thinking.

DR. EVANS: Should we ask for public comment about important issues that the Secretary's Committee should take up? It would seem logical.

DR. TEUTSCH: Do we have a process for such?

MS. CARR: We can, sure.

DR. TEUTSCH: Do we have a listserv, at least?

DR. EVANS: Some would say, whoa, we are going to start being logical now?

DR. TEUTSCH: A foolish consistency. Other thoughts? We have a really rich list of topics. Paul the Wise, the table remains open. We have talked about at least through the 22nd, and if we are going to have more of an ongoing process it could be longer than that. So if there are other topics that people want to get on the table, let's do it. It would be good to start with a rich count.

LT COL McLEAN: One of the minor themes that ran throughout was identifying subpopulations within our

society which may not be getting as much of the benefit as possible. Maybe we should look at the population more systematically to find the other subpopulations that are not receiving the benefit of the genetic advances we have.

DR. KHOURY: One more.

DR. TEUTSCH: Muin, don't disappoint us.

DR. KHOURY: I'm not. Over the years I have seen an interesting dialogue and sometimes clash between the two worlds, the world of genetic and genomic medicine and the world of evidence-based medicine. I would like to see whether or not this Committee can weigh in on this dialogue.

DR. TEUTSCH: That really raises some different issues. There are some really interesting issues on evidence-based medicine about the individual versus population issues that come up in many guises. It would be an interesting thing for us to address.

Let me wrap up. I'm sure most of you won't mind leaving a few minutes early.

Next Steps and Concluding Remarks

Steven M. Teutsch, M.D., M.P.H.

DR. TEUTSCH: I think it has been an extremely productive meeting, and I want to thank everybody for all their attentiveness, the hard work. Aside from the bittersweetness of Reed leaving, we have managed to get quite a bit accomplished. We got a letter out to the Secretary regarding reimbursement and CMS. We -- I wouldn't say miraculously because Andrea was a known thing all along -- got our recommendations wound up on the oversight of genetic tests, which is just an enormous accomplishment. That feels awfully good.

We have firmed up the charge and tasks for the Educational Taskforce, and I think we have a good initial discussion on priority setting. Of course, we will have our group formed here to help lead us through that.

It looks like Sarah is gesturing like she has something else to say.

MS. CARR: We have something else to show.

DR. TEUTSCH: What else do we have to show?

While you are showing this, just to let everybody know, the next meeting, which will be July 7th and 8th, will be back here at the Humphrey Building.

Oh, what we have done. We have walked through

these. Obviously, on oversight, we finalized the recommendations and approved the transmittal of the report in spirit. We will get any final comments from you on the text. We talked about the Education Group and priority setting. We approved the charge, with some refinements and in draft.

And, the Priority-Setting Taskgroup we just formed. Paul Wise will be chairing it with Mara, myself, Paul, Jim, Muin, and Gurvaneet. We will be going through a process hopefully so we will have a substantial discussion of that in July. Then we finalize the letter which you saw yesterday to the Secretary.

Next steps. For the Oversight Report, finish the copy-editing and the final transmission of the recommendations to the Secretary by the end of the month and the finalization of the entire report by April 30th.

Final comments on the text are due February 22nd, and the steering committee will see the final wording of the overarching recommendation.

The Education Taskforce will be developing a plan and timeline, and Priority-Setting will help us develop the materials and process for the priority-

setting process for July. We will be working to convene the ex officios, or at least contact them, to solicit their feedback on the topics.

Sylvia and Julio will be helping organize the session for personal genome services for July as well.

So for July, what we have on the table right now is the priority-setting process. Hopefully we will get to finalizing those topics. We have the personalized genome services, and we will try and get the companies in and have that panel. The series of panels that Sylvia is going to help us to figure out as to who is all going to be there, and then the update on the Education and Patents Taskforces.

Sounds like a full load to me. Thanks, everyone. Any other final words?

[No response.]

DR. TEUTSCH: Thank you all.

[Applause.]

DR. TEUTSCH: Safe travels.

[Whereupon, at 3:53 p.m., the meeting was adjourned.]

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CERTIFICATION

This is to certify that the attached proceedings

BEFORE THE: **Secretary's Advisory Committee on Genetics, Health, and Society**

HELD: **February 13, 2008**

were convened as herein appears, and that this is the official transcript thereof for the file of the Department or Commission.

SONIA GONZALEZ, Court Reporter