DEPARTMENT OF HEALTH AND HUMAN SERVICES

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ON
GENETICS, HEALTH, AND SOCIETY
(SACGHS)

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P R O C E E D I N G S

[8:03 a.m.]

Opening Remarks

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

DR. TEUTSCH: Good morning, everyone. I think we will get going. We have, hopefully, an interesting agenda today to plan our work going forward.

Before we launch into that, I first want to recognize, at the end of the table here, Matt Daynard. Matt, yesterday we went through transitions with folks. I understand you will be making one yourself and retiring.

DR. DAYNARD: That's correct.

DR. TEUTSCH: We wanted to acknowledge that fact and express our appreciation for all that you have done in this field with the Committee.

DR. DAYNARD: Thank you very much.

DR. TEUTSCH: Many thanks, and all the best.

DR. DAYNARD: It has been an honor to be part of this Committee for the last seven or eight years, or whenever it started. I appreciate it very

much. I wish you all great luck. The Committee does amazing work.

DR. TEUTSCH: Great. Thanks so much.

[Applause.]

DR. TEUTSCH: Later on, just to let you know, Dan Wattendorf will be sitting in for Scott McLean and Kerry Leibig will be joining us as the new ex officio from EEOC.

Today we are going to return to work that we began back in February to begin to plan out our work for the future and to identify the high-priority issues that we should be taking up.

The background materials are all in Tab 5 of your briefing book. You will find the slides in your table folders.

As a reminder, in February we reviewed the process that the Committee used in 2004 to establish the priority issues that we have been working on ever since and agreed that since we were nearing completion of that study agenda we should begin to look ahead to identify some of the emerging issues and unresolved issues that continue to need our attention.

We did some brainstorming back then and in July made some preliminary decisions about priority topics. You will remember the diagram that Paul Wise regaled us with as he helped lead us through that discussion.

Today our goal is to finalize our future study topics and how we will be addressing them, and then agree on a strategic plan for getting the work done.

I should say that although this may be "final", it is not really final because we hope that we will be able to take the results of that work and have a chance to meet with the incoming Secretary, and presumably his staff, to talk about how that dovetails with their priorities so that we can get some good alignment and begin to work together.

Paul Wise has been leading this. I really appreciate all his work as chair of the Priority-Setting Task Force. It has been more than a little work over the last few months. Paul will lead us through a discussion of the steps and decisions we have already taken. Then members of the task force

will review the issues we identified in July and lay out some policy options, questions, and action steps.

I shouldn't say "policy options." We are not reaching conclusions on those today. But we will be looking at policy questions and action steps in each area that we could take as we move forward.

We will want to get some sense later on in the day of the relative priority. I don't expect we will take votes, but clearly it is a long list of things that are potentially on our plate. We want to get a good sense as to the order in which we might tackle those.

With that, let me turn the floor over to Paul, and we will get going.

Review of Priority-Setting Process and Proposed Priority Issues

Paul Wise, M.D., M.P.H.

[PowerPoint presentation.]

DR. WISE: Thanks very much, Steve. Just to remind people, the Priority-Setting Task Force included members of the Committee and ex officios. You can see here on this list the membership.

We have also starred the people who were designated as cluster leads to help take us through the issues that we have identified as being potentially of highest concern.

I also want to, in addition to thanking the cluster leads, thank the staff, who have worked extremely hard and produced very high-quality work in helping us move through this agenda. That of course includes Sarah, but also Cathy Fomous, Darren Greninger, Kathi Hanna, and Linda Smith, who have really done a remarkable job putting this together and generating the issue briefs that we will discuss in a few moments.

I just want to quickly review the priority-setting process that we have used. Seventy-three potential priority issues were generated through a brainstorming session that we had at our February meeting earlier this year. We subsequently had discussions with ex officio members to make sure that we had a good sense of their concerns and the concerns of greatest interest to their respective agencies or departments.

We also solicited public comments and got a broad range of very helpful suggestions. In addition, we had specific interviews with both the content experts but also what we call vision leaders, to help make sure that we in fact had the best broad advice as to where we should be heading over the next few years.

These issues were then ranked by Committee members individually using a Likert scale. We then examined those priority issues and areas that emerged by just ranking the ones that were felt by Committee members to be the most important and the most relevant to our work, but also looking at affinities between the different issues to try to organize and create a coherent structure for the issues that were ranked high. Then we developed and confirmed that there were in fact a relatively small group of clusters of issues, based on both their content and the affinity within the patterns of voting that the members conducted.

The next steps were then to take those clusters and to develop issue briefs, which were distributed and really, I think, did a very nice job

capturing the central elements of the clusters that were identified as being most important and relevant.

These clusters, just to remind everybody, were seven in number and included coverage and reimbursement for genetic services; ensuring the clinical utility of genetic information; genetics education and training, with attention to workforce diversity; informed consent, privacy, and discrimination issues in genomic data sharing; implications of consumer-initiated use of genomic services; public health applications of genomics research, with attention to health disparities; and genetics and the future of the healthcare system.

Just to identify what our goals are for today's conversation, we will first have presentations of each of the cluster areas by the cluster leads. There was a format that was suggested for the presentation of these issues. The order of cluster presentations in no way reflects anything about how we valued these issues. They are not ranked on the basis of any criteria. It is strictly the way we thought the order would be most helpful for presentation.

We then want to discuss the specific policy questions and propose action steps in each cluster, and then to develop an overarching and flexible action plan.

There is no formal vote-taking today. We are really just looking for several things. One is to make sure that we got the issue right, that the central elements are truly reflected in the issue briefs and the presentations, and also to see if through the discussion we can develop or at least get a sense of a consensus of which of the seven deserve greatest attention, particularly in conversations with the incoming administration.

We will work through the seven clusters. These will be relatively short presentations. If you have questions for clarification or something that you really feel you need to voice at this time, that would be fine to ask the cluster leads. Otherwise, we have a lot of time set aside after the cluster presentations for discussion, for criticism, and additions. We would hope that the heart of the conversation would be after all seven clusters have

been presented.

Comments or suggestions about this plan of action?

[No response.]

DR. WISE: Cluster No. 1.

Discussion of Proposed Priority Issue Areas

Cluster No. 1: Coverage and Reimbursement for Genetic Services

Marc Williams, M.D.

[PowerPoint presentation.]

DR. WILLIAMS: A wise man once said that when anybody says it is not about the money, it is about the money. I think anybody that has listened to any of the discussions that we have had at this Committee, both at this meeting and any other meetings, know that issues of coverage and reimbursement come up frequently.

I first got involved with the old SACGT on reimbursement issues back in 2000, working with Suzanne Goodwin, and have been working on and off with this through a workgroup on the report that is listed there under the first sub-bullet. In some ways, a lot

of what we are going to be reporting here are things that are already in progress and that we just need to continue to do.

Essentially, as we have heard, there have been some unresolved issues from the February 2006 SACGHS report. We are basically continuing to pursue those unresolved issues. We are looking at strategies to hopefully remove some of the obstacles to implementation of some of the recommendations that we put forward.

But we have also been looking at identifying new issues. This, again, ranges from things relating to Medicare coding, billing, payment, and reimbursement policies.

The report that I referenced had nine recommendations, and there have been a number of things that have occurred since that report which we have heard about at previous meetings, so I am not going to walk you through these.

We also heard from Steve yesterday in regards to the last sub-bullet there that we have been told and are expecting a letter from the Secretary's

representatives later this month regarding some of the follow-up issues that we had discussed at our last conference call and also in letters that have been going back and forth this year.

The policy questions that we are really interested in focusing on are approaches to revised payment rates to reflect the true cost of the genetic test. We know that the Medicare fee schedule at the present time has a lot of these tests relatively undervalued compared to their true cost. While there have been some efforts to look at invoking things such as inherent reasonableness to address some of these fee schedule issues, we have not been particularly successful in terms of being able to make any movement there.

This is an issue relating to offering tests and to some degree, as we talked about yesterday, also impacts access to testing.

The billing related to certified genetic counselors has been an ongoing issue, in particular how we can get access to CPT E&M codes to enhance access to genetic counseling services, which we think

will be increasingly important based on the increasing visibility of genetics in clinical practice and the importance of this in terms of informed decision-making prior to embarking on testing.

As we saw some of the costs of the tests yesterday, I think all of us that are at least even mildly affiliated on the payer side would really like to make sure that the people that are going forward with testing are good candidates for testing and actually that is what they want to do. So genetic counselors have an important role to play, at least in the traditional genetic testing field.

There have been new issues that have arisen since the 2006 report. One of them relates to the application of reimbursement audits, specifically medically unlikely edits, to procedure-specific CPT codes where, in the course of processing DNA, certain CPT codes are done in multiples, sometimes many multiples. There was a movement to apply medically unlikely edits so that those multiples would be kicked out and only one CPT code of a given type would be paid for. So we have been working to say this is

probably not actually reflective of the work that is happening in the laboratory.

That at least is on hold at the present time, but we haven't had any resolution to that issue.

We have talked a lot in this Committee about family history. We heard yesterday from Steve about the roll-out of Version 2 of the Surgeon General's Tool. There has been a lot of effort from a number of agencies both within DHHS and also throughout other areas of the government that are providing health care about using this Family History Tool as the de facto standard for family history collection within government-provided health care.

But we still are left with the issue of how do we actually use the family history and then how do we fairly reimburse for that. In particular, the idea is using family history in the definition of a personal history of disease so that we can meet reasonable and necessary standards for Medicare coverage.

We have put forward several possible recommendations, or as Jim would say, a range of

recommendations.

[Laughter.]

DR. WILLIAMS: I'm sorry. I should have had another sip of coffee before I started, perhaps.

But, is there a way that we could use some of the current evidence groups such as EGAPP or USPSTF to define cases in which family history of a disease could be considered to be personal history, which would allow coverage for some interventions.

We are also looking ahead to August 2009 and the NIH-sponsored State of the Science Conference on Family History, which will really probably give us the best assessment of where the current evidence is relating to the science of family history.

Again, how can we look at the current reimbursement system and say, if you are doing something beyond just asking "Do you have a family history of anything?", if you are actually doing analysis of pedigrees and this sort of thing, which is work that is outside the current family history within the evaluation and management codes, then is there a way to reimburse people for doing the extra effort

there.

In conjunction with the Pharmacogenomics report that came out a year or so after the Reimbursement report, the idea is to raise pharmacogenomic testing as a national coverage decision issue that could be looked at by CMS and then how that would fit into the idea about whether pharmacogenomic testing would be considered to be a diagnostic test. In that case there could be consideration of coverage from CMS in situations where the evidence warrants, versus the concern that pharmacogenomic testing would be looked at as a predispositional test, which right now is excluded from coverage under the Medicare statute. So this is an issue that really needs to be resolved.

Then, ultimately, how do reimbursement issues impact access to genetic services and how can we improve that in populations that are currently being underserved.

Possible action steps. We are going to continue to monitor the recommendations from the 2006 Coverage and Reimbursement Report. We have ongoing

discussions with CMS officials relating to that laundry list of items there. Hopefully we will get some additional communication before the end of the year that will let us know what has been done and what needs to be done.

We are engaging with the laboratory community to again look at generating support for the application of this inherent reasonableness authority to the clinical laboratory fee schedule. That might provide some relief for some of the molecular codes, although what we were told in our phone call was that if we open inherent reasonableness, it is open for everything.

So the tradeoff could be, while we get some increase in molecular codes, there may be decreases. So, does the laboratory community as a whole look at this as a benefit to open this or would this be something where there would be more harm than benefit that could potentially accrue.

We want to look at the authorization act, the Patient Providers Act of 2008, looking at clinical preventive services and coverage through CMS, if

USPSTF recommends that and if the MedCAC concurs, to see whether or not there are some aspects of what we are doing, particularly around family history, that could be folded into that.

We are trying to encourage collection of demographic data so that we can have a better sense of access to and utilization of genetic services in underserved populations.

So that is a very brief overview of what we are doing. Most of it is a continuation of things that this Committee has already signed off on. Hopefully, we will be able to pursue those things. I'm happy to answer any questions.

DR. WISE: Questions for clarification?

DR. FERREIRA-GONZALEZ: Do we discuss now if we need to add a point or later in the afternoon?

DR. WISE: If it is a quick addition or a quick point or a clarification question, it would be great to do it now. If it is a more complex suggestion or issue, then we should probably leave it for the full discussion.

DR. FERREIRA-GONZALEZ: There are a number

of laboratory associations in the community and even in industry that are looking at review of the coding for genetic testing as a monitor. We might want to engage these professional organizations on an ongoing basis to maybe come back to us to report on some of the efforts. Maybe we can be informed so that all these different groups, who might not be talking to each other, can actually work in a single group.

DR. WISE: Why don't we move on, then, to Cluster No. 2.

Cluster No. 2: Ensuring the Clinical Utility of Genetic Information

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

[PowerPoint presentation.]

DR. TEUTSCH: Great. Thanks, Paul. I have to disagree a little bit with my colleague, Marc Williams. It is about the value, not just following the money. We are going from money to value.

That is ensuring the clinical utility of genetic information. This is a topic we have already begun to discuss.

Actually, I should say, Paul Billings, I

think I heard you get on the phone; is that right?

[No response.]

DR. TEUTSCH: I think Paul has joined us from the West Coast.

DR. BILLINGS: I'm here, yes.

DR. TEUTSCH: Great. I'm sure you will chime in.

DR. BILLINGS: I doubt it, but go ahead. There seem to be enough chimes in the room, thanks very much.

[Laughter.]

DR. TEUTSCH: Clinical utility is an issue that we have also been tackling over the last few years in different ways. There are multiple challenges in establishing the clinical utility of tests.

One is the general paucity of clinical studies that actually do look at the clinical utility. Even when they exist, we don't have a clear set of accepted evidentiary standards against which to judge the studies, particularly for their different applications, everything from screening and prevention

on to pharmacogenomics and things of that nature.

At the moment, there is no organization that is actually dedicated to performing utility assessments. There are a number of them that are involved to varying degrees.

There are also a set of concerns about how we are going to deal with the avalanche of information that is going to come out of whole-genome sequencing and how to assure that the information there is going to be applied in a way that actually leads to real benefit for patients. Some organization will be needed to perform some utility assessments on that so that we use the information well.

There are a number of groups that have begun this work. EGAPP is probably the one that is most clearly dedicated to it. That is the group that is out of CDC. It has been working to define standards and has actually been performing some assessments, but it is small and only beginning to tackle a modest number of these at the moment.

There are a variety of commercial and non-commercial entities that actually do technology

assessments. You have two of them up here. Hayes is one that provides information largely to the payer community. BlueCross BlueShield Tech actually makes their evaluations public. There are EPCs and other organizations that do some assessment of genetic tests as part of their larger efforts to assess technologies.

There is also the IOM Roundtable on Translating Genomic-Based Research for Health, which is very much concerned with these issues and then how we get effective technologies actually translated into the healthcare system.

The reports on Pharmacogenomics, Coverage and Reimbursement, and Oversight have all touched on this issue to varying degrees. We have recommended that the Secretary create a public-private partnership and a group that would define the types of underlying studies that are needed for assessments and the standards by which they should be judged, as well as to do those evaluations and help with the dissemination of clinical guidelines based on those assessments.

As many of you, I think, are aware, there are bills in Congress that do that, not specifically for genomics but do that generally within health care. There are other proposals, of course, that would like to do that exclusively in the public sector.

The kinds of issues that we think we could address are which groups would be most effective in defining the evidentiary standards and actually doing the reviews. A second would be how the government can better inform those involved in research and development about those evidentiary needs so that the appropriate studies will be done and we will have the information that will allow us to assess specific technologies and how they can be applied for specific conditions.

Although we have recommended in the past that this be a public-private organization that would be responsible for assessing the clinical utility of genetic tests, as I indicated, that is just one of several options that would be available for how this might get done. We could revisit the issue not so much of what it would do but more how it should be

organized and structured, as well as a little bit about the scope.

All of this, of course, ties in more clearly with how to integrate what we know about genetic tests and effective technologies in the healthcare system, and will tie into the topic of the future of the healthcare system, which Mara will be talking about a little later.

These are some of the things that we could do. We could provide a forum for discussion to help define the evidentiary needs and standards for evaluating clinical utility. We could recommend that any governmental organization or group tasked with assessing clinical utility apply different clinical utility assessment methods for different clinical users of genetic tests so that we have that range.

We could develop some brief reports on how the clinical utility assessments, which are usually mostly very scientifically oriented, can also incorporate many of the important contextual issues: cost, cost effectiveness, ethics, legal issues, feasibility, acceptability, et cetera, to better

inform the users, particularly patients, regulators, payers, healthcare providers, and performance measurement specialists, about how they can use that information in their decision-making.

This isn't quite worded right. We can look at how to better inform those involved in research and development about the evidentiary standards so that we can provide some better direction to the research community.

The next one is about government organizations or groups that can establish evidentiary standards for clinical utility assessments, that can create methods for assessing them, and can actually do those utility assessments on a more systematic basis than what we have right now.

So that's it. Jim.

DR. EVANS: One question. Maybe this is better discussed later. But, since a common theme in this is the observation that we don't really have the kinds of studies to assess clinical utility and that that is needed, and getting back to the other theme of the morning that it is about money, is it worthwhile

to look at mechanisms for encouraging or obtaining funding specifically for those purposes. There is a finite pie. Sometimes it is more appealing to fund certain things. These are sometimes seen as boring studies, but in the end they are obviously incredibly important.

DR. TEUTSCH: They are fascinating, Jim.

[Laughter.]

DR. TEUTSCH: No, I think it would be part of helping to provide that. I think some of the legislation that is out there today that is not specifically in the genomics area actually carves out a fairly broad list of things. They actually talk about sponsoring or conducting research, all the way from, for those kinds of things, doing clinical trials on through the economic evaluations.

I think we would want to talk about what the scope should be and how that should be used to inform the research committees. Pardon?

DR. EVANS: These are very expensive kinds of studies, but there is no shortcut for a lot of them.

DR. TEUTSCH: I don't want to get into a long discussion here, but there are different ways. This gets to evidentiary standards. When do you actually need an RCT, right? When is a simple decision model going to be adequate, right?

DR. EVANS: Sometimes it is adequate, yes.

DR. TEUTSCH: For the most part, if you ask people in this field about how to do these things, they are talking about clinical epidemiology mostly. We are not talking about, for instance, how do you understand the biological mechanisms of action and use that to inform the likelihood. How do you know when a biological mechanism is likely to be, in our understanding, informative or misleading. We have plenty of examples of both.

There is a whole range of things here that could be done under this general rubric.

DR. EVANS: Sounds great.

DR. TEUTSCH: Other comments?

[No response.]

DR. WISE: Thank you very much, Steve. We will move on to Cluster No. 3.

Cluster No. 3: Genetics Education and Training

Barbara Burns McGrath, R.N., Ph.D.

[PowerPoint presentation.]

DR. McGRATH: Thank you. We have a suggestion that it is about the money or it is about the value. I'm going to suggest it is about the knowledge.

This topic about the need for basic genetic education and ongoing training resurfaces a lot. It was one of the initial priority-setting topics within the initial SAC. Then it rose again to high on the list of the data gathering we did with this group and the public comment. I have a feeling it is going to continue to resurface every five to 10 years as long as we are talking about these things.

There are reasons for that. We started a task force based on some other priority setting in November of 2007. You have heard a fair amount about our activities, so I will make this fairly brief since the Committee has heard about this a lot. But in terms of addressing the need at the priority-setting, I will review it a little bit.

In our committee, we decided to identify three groups to focus on for our short-term goals, and those are the needs of health professionals with and without expertise in genetics, the needs of public health providers, and the needs of patients and consumers. Each of those groups are represented by task force heads. They are collecting data on those in different ways.

In terms of background, why is this important. The data continues to come in that clinicians and consumers are being increasingly expected to have greater and more sophisticated knowledge, but the education perhaps isn't keeping up with that.

The policy questions. What we would like to do, and what we are planning on doing, is looking at the initiatives and programs that are out there. There are quite a few. We are at this point collecting data on all the ones that are being implemented and being planned and trying to evaluate whether those are adequate. If any of those are particularly good, we will see if we can use those as

models so we don't have to reinvent the wheel.

With that, what is the role of the federal government in all of this. There is no shortage of information about education, but we need to keep our eye on the idea of how can HHS help with education and training. We will then look to see the role of the federal government in this.

An important area is what role can the federal government take in promoting and supporting diversity and cultural competency of the healthcare professionals and the work force. This is a really important area. We would like to use this as one way to deal with the Healthy People 2010 recommendations that we address health disparities in the country by looking at it through the angle of increasing the diversity of the healthcare work force through the angle of genetics education and training. This is perhaps a newer area that we are going to be looking at.

The other one is the whole notion of accreditation, licensure, and certification. That is a role that the government can play. We will look at

how that might be improved or changed or what is happening in that area.

The next couple policy questions are dealing with patients and consumers. Of course, we all know that there is lots more information reaching patients and consumers that isn't necessarily being filtered. We would like to broaden that by looking at various experts who are working in the area of communication and patient education by looking a little deeper at the work of academic researchers as well as clinician educators and lay health educators, as well as what industry is doing to address their interest in educating consumers.

I think we heard a lot about this at our last meeting. We saw a lot of the promotional material that is reaching consumers directly about genetic services. We would like to take a look at that. Some of it is very sophisticated. It is being used not just for marketing but it is educating consumers as well as health professionals. That might be an unintended purpose of it, but we might as well look at that and see if there is something to be

gained from that or if we have something to learn from that.

Along with that, is there a role for FDA to be involved in monitoring that in terms of some of the things that the Oversight Committee brought up in the last report.

We have some action steps. There are some very short-term ones. At the last phone conference it was suggested that we really could sit down very briefly and quickly with FDA to see whether it is under their purview to deal with issues around the medical device promotional materials that we are seeing, as well as talking with industry about establishing voluntary standards for promotional materials in terms of their educational properties.

The more longer-term ones. All three of the groups are very busy working on gathering data, doing interviews, collecting surveys, and collecting existing materials. We will have a report ready for public comments this coming summer, the summer of 2009, with hopes that the final report will be finished in the year 2010.

As I started off by saying, we have identified three groups: health providers, public health practitioners, and patients and consumers. There are other groups that continue to emerge as being involved that have needs for education and training. We have a list there. The next round may be to look at some of those people as the next level, not the first line of contact but the next layer of people who are involved in the decision-making around genetics to see what their education and training needs are.

I think that's it. Thank you.

DR. WISE: Yes, please.

MR. KIRCHNER: Thank you very much for a nice report. I'm just wondering what interaction you envision between the educational responsibilities and recommendations you have made and the work of Task Force No. 2. It seems to me that the two ought to be very much related. It is just as important to notify people about tests that have not been validated and therefore perhaps should not be used, as about tests that have already been proven to have a valid

relationship to risk factors and therefore potential intervention.

DR. McGRATH: Absolutely. I would consider that the whole notion of clinical utility needs to be shared with providers and practitioners as well as consumers and patients. Absolutely. Thank you.

DR. WILLIAMS: If I could just add to that, as Steve mentioned in his report, the EGAPP working group and then the associated stakeholders group of EGAPP, one of their tasks is to try and actually disseminate the information, whether it is positive or negative, relating to that utility. So it does seem to be a natural point of reinforcement.

DR. WISE: Part of the discussion that I hope follows the cluster presentations will be to look for connections across the clusters as much as individual ideas or suggestions within each cluster. I think it is going to be very important for us to look for commonalities and ways to create coherent linkages. In fact, reclustering of the clusters may in fact be the most helpful thing we can do.

Other comments? I just want to thank

Barbara for all your work with the task force. It has been truly impressive and fits very well among the highest priorities that were identified by the Committee.

We will move on to Cluster No. 4, please. Kevin.

Cluster No. 4: Informed Consent, Privacy, and Discrimination Issues that Relate to Genomic Data Sharing

Kevin FitzGerald, S.J., Ph.D., Ph.D.

[PowerPoint presentation.]

DR. FITZGERALD: Money, value, knowledge? This is the 21st century, people. It is about power. Information is power.

So, what we are going to do with that information, how that information is going to be given to people, and how they use that information to make their choices, of course, fits into a concept that we have been working with for quite some time called informed consent, where people are supposed to get all the relevant information they need in order to make an informed decision.

Of course, the problem with personalized medicine is all the information is now relevant and all of it will have to be put together somehow so that we can come up with a comprehensive view of an individual's health status. As we go about pursuing these laudable goals, it is going to raise some really interesting issues that we need to address as far as privacy, confidentiality, and informed consent are concerned.

What we are seeing right now, which is currently challenging, is the pursuit of the large, population-based databases, where a lot of this information would be pulled together. The whole idea would be, obviously, to pull it all together so you can associate various aspects of health and disease in a picture that will of course, as we heard yesterday, create a disease signature.

The question is, what will that signature be, just how individualistic will it be, and how identifiable will it be.

We have already noticed that the evolving research paradigms in this whole idea of personalized

medicine may in fact force us into a new conceptualization of informed consent or perhaps a new conceptualization of some kind of way of moving forward in a world where the information perhaps cannot be held in some kind of anonymous state.

This is going to be true for both research and clinical practice, since I think they will be much more greatly intertwined than they have been in the past. Obviously, this will require perhaps levels of vigilance and attention that we have not had to apply up to this point.

This is not anything new, in one sense. There have been activities ongoing, as you can see there, since the NBAC in 1999. But these are areas that other people are looking at. One of the questions will be how will we work with others, or need we work with others, to address some of these things.

That leads us, then, to some of our policy questions. If there are these new issues raised by this data, how do we begin to address that. Are they truly new. Are they just extensions of what we have

wrestled with before in the '70s and the '80s when the Belmont Report was looking at the effects of research on human subjects. In one sense, we are all going to be the human subjects. We will all be part of the research, since it is going to be pulling all the information together.

How do we take these challenges to the public? How do we engage them, not just tell them but engage them, in the process of trying to understand this and delineate the issues which are of some significance and importance?

How do we cross generational divides? We were talking about education. I don't think the next generation is going to be too worried about this. They can't be. All our students have all their information on Facebook already. What is there to hide?

[Laughter.]

DR. FITZGERALD: So, will there be different levels of concern when we talk about information being made public. If so, what do we do about consent needs for these large-scale population studies. How do we

consent the population. These are some interesting questions.

Then, how can the consent process be improved? What are the strategies? We can use some of the stuff that Barbara was talking about, and that her task force is looking at. How do we educate and engage people? One strategy is, you may be familiar with what are called teach-backs. You teach somebody something, and then they teach it back to you or to a third party. Obviously, when you come to truly understand something is when you can teach it to somebody else.

What is the role of SACGHS in this? What is the role of HHS? Obviously, these are big issues and big questions. They are probably beyond the purview of both SACGHS and HHS, but obviously we may have a key role to play in these.

As I said, we are wrestling with some new concepts and new ways of looking at these things. What are the implications, especially with computer algorithms that are now out there that we discovered in the journals in August? They can pull individual

sequence data out of an aggregated, supposedly anonymized database.

If that is true and we start coming up with these disease signatures, what does it mean when someone publishes a journal article and there is a disease signature article? I can look at that and say there is only one person on the face of the Earth that has this particular signature. All I have to do is link it to that person.

Then, how does the legislation we have now, HIPAA and GINA, affect this process? What about the proposed legislation; what is the pipeline that we need to look at?

These are some possible action steps for ourselves. One that isn't up there that we probably could take into consideration goes back to Marc's emphasis on money. We could start our own personal information website and charge, and make lots of money. But, maybe that doesn't fit.

Sarah, is that allowed? No? Oh, shoot.

[Laughter.]

DR. FITZGERALD: All my best ideas.

One thing we can do is monitor the process, especially for GINA and perhaps other proposed legislation as it comes along, to see how they actually get applied, what the gaps appear to be, and how those gaps may need to be addressed.

Certainly, again, soliciting public input. One of the things I think that could certainly derail this move toward personalized medicine would be if we lost public confidence in the process because the public became suspicious in some way or was worried about the fact that things were being done that were somehow not transparent. How do we do that?

Then, again, we could write a report, which is what we do well. Can we come up with a report that dives into this, which I think would be a rather complex topic? It would involve, again, ideas of risk. What people consider to be the risks of this information being available publicly.

Again, I would imagine you would have quite a diversity of perspectives on that and a diversity, also, in the sense of what the harms and benefits might be. That, too, would be something that I think

would need to be addressed.

Of course, the main thing with all this is how are we going to use that information. Of course, the person following me with Cluster No. 5 is going to solve all those problems, so I'm just going to stop here. No pressure.

DR. WISE: Any questions or comments?

DR. FOX: Just a suggestion. On your first possible action step, I would add VA to the list of organizations you might want to collaborate with.

DR. FITZGERALD: Absolutely.

DR. WISE: Thank you. Other points of clarification?

[No response.]

DR. WISE: Sylvia.

Cluster No. 5: Implications of Consumer-Initiated Use of Genomic Services

Sylvia Mann Au, M.S.

[PowerPoint presentation.]

MS. AU: Cluster No. 5. I agree with Kevin; it is about power. But I think in this cluster it is about empowerment. Now there is pressure, isn't

there?

We are all trying to sell our clusters. Unless you have been hiding in a cave without wireless access, you have been inundated by direct-to-consumer genetic testing articles in the media. This cluster came about because, of course, the number of personal genomic services marketed directly to the public has increased in the past few years, which is definitely an understatement.

This is a new model that doesn't have direct involvement of a personal health care provider. So we are wondering whether a comprehensive consumer protection strategy may be needed in this type of medical testing.

Our concerns include the relative value of the information provided, of course going back to Cluster No. 2, clinical utility; the level of consumer understanding, which again is the education cluster; the provider community's ability to understand and translate information for patients, again the education cluster; and the potential risk of misuse of information by consumers or third parties, going back

to Kevin's cluster. So you can see that this cluster encompasses every other cluster that we have on the list.

Genome service companies offer a vastly different array of services, ranging from risk assessment to recreational testing such as match-making. As of yesterday, in the U.S. you may now test your children for their sports ability. I believe it is $149. That was in a New York Times article this weekend.

Past SACGHS activities include letters that the Committee wrote to the Secretary in 2002, 2004, and 2006, expressing concerns about the advertising claims made by companies offering these direct-to-consumer genetic services. We also had an information-gathering session at the July 2008 SACGHS meeting to explore what was going on in the landscape of genomic services.

There has been a definite explosion of U.S. and international activities related to this area: research studies, educational resources, workshops. So there are a lot of activities going on right now in

this area.

Not surprisingly, because this area is fraught with so many questions, we have the most policy questions of any cluster. For oversight, of course, we wonder whether these genomic tests will be regulated similarly to other complex laboratory tests.

As to our Oversight report that we did in SACGHS, which we will never forget, we wonder if those recommendations will be sufficient to relate to these direct-to-consumer genomic services.

We have concerns, of course, about clinical validity and utility, which again came up in the Oversight report. What are the best formulas for calculating these risks. What are the criteria to determine whether association between a particular genetic marker and a phenotype is strong enough for that marker to be included in the genetic testing and reported out.

Continuing concerns about clinical validity and utility. Should there be standards for formatting the raw data from the whole-genome scans. How will the clinical validity and utility of such tests be

assessed and communicated to consumers. When is it that sufficient data will have been produced to change previously recommended risk calculations.

Issues for consumers and healthcare professionals. Are requirements for public education and informed consent needed before testing. What are the appropriate roles and responsibilities of the healthcare providers, consumers, and public health programs in this non-traditional approach to genetic testing. Do personal genome services actually fill some specific healthcare or public health need. Are providers and consumers adequately prepared for the information provided by these services. What are the benefits and potential drawbacks of direct-to-consumer personal genomic services.

How will the healthcare system and providers be affected by the availability of these personal genome services. What is known about consumer interest in personal genome services and consumer understanding of these services. What are the criteria that should be considered in determining the value of the personal genome service. What are the

criteria for determining whether previously tested individuals should be contacted to inform them of modified risk, or should we let individuals fend for themselves and contact the companies for follow-up.

Of course, for advertising, what are the criteria that the companies need to follow before offering these services and marketing them.

As Kevin said, we have privacy and discrimination concerns. What are the privacy concerns. Probably not much with the next generation on Facebook.

[Laughter.]

MS. AU: What cautions and benefits do consumers consider when sharing their genomic information with others, such as their family members, social networks, clinicians, employers. Does GINA apply to this type of personal genome service, and are these companies actually covered by GINA.

Then, for disparities, could personal genome services actually exacerbate health disparities? Most of these are paid out of pocket and not covered by insurance right now.

Our possible action steps are, of course, to monitor the outcome of all these federal and non-federal workshops, work activities, and educational activities that are going on. Short-term actions would include a development of a checklist that patients could look at when they are trying to determine whether or not they want to participate in these direct-to-consumer genomic services. The Personalized Medicine Coalition has come up with a basic checklist that has started already.

We could also do a brief report on selected key issues so we don't have to delve into every policy question. Or, we can do that lovely in-depth report that we love to do on every single issue that we can think of under the sun, and work on this for the next 10 years.

DR. WISE: Comments and suggestions? Marc.

DR. WILLIAMS: At the risk of adding to the list --

[Laughter.]

DR. WILLIAMS: I'm taking the risk.

MS. AU: That's okay. You're not rolling

off.

DR. WILLIAMS: That's right. Although, after dinner last night, I might.

[Laughter.]

DR. WILLIAMS: You can take that any way you want.

I was struck a couple of meetings ago when I think it was a representative from the World Privacy Forum spoke to this group about the concerns about not only the information from the testing but the fact that some companies may in fact be using information that the consumer is providing at the time of testing to sell to others. I don't see that represented there. Since one of the focuses of this relates to potential consumer harm, I think we should fold that in somehow.

DR. FOMOUS: We had a more general question about that, but we can make sure that we capture that.

DR. WISE: Other comments or questions?

[No response.]

DR. WISE: We will move on to the next cluster. Joseph.

Cluster No. 6: Public Health Applications of Genomics Research

Joseph Telfair, Dr.P.H., M.P.H., M.S.W.

[PowerPoint presentation.]

DR. TELFAIR: Thank you very much. We in public health are very solution-oriented. To us, it is about the work and getting the work done.

I will cut to the chase. First of all, I would like to say it is really an honor to present this information. I do want to thank Dr. Fomous and particularly Dr. Kolor, who is sitting at the end of the table here, who worked with her group at CDC to help us to formulate this.

Public health, as many of you know, is a broad and diverse table. We try to look at the world ecologically to fit between the physical and the social environment in many ways, to the benefit of the general population.

We want to clarify some of our terms. Public health genomics is a multidisciplinary field that really is concerned about effectiveness and responsible translation of genomic-based knowledge and

technology, with a focus on population health.

We focus on policy and actions that are needed to promote health and to prevent and control disease. We also focus on the interplay of genes, the environment, both physical and social, and behaviors. We want to ensure that the benefits of genetics and genomics are realized across many diverse populations and groups. We do this through our main public health priorities, which are assessment, policy development, and assurance. We do this from a knowledge base, an evidence base. Research forms the core.

Like the previous work before us, there is some overlap. We expect that there will be some cross-clustering grouping of our work. But the point here is that public health is very broad and allows us to look at a number of these things.

Now, assessment as we define it is really the systematic collection of analyses and dissemination of information. It focuses on epidemiologic and laboratory research, investigations, and monitoring of community health problems and risk factors.

We also work towards policy development and basically taking what we have learned and promoting that from the translation of advancement in human genetics in terms of prevention and other opportunities. We do this through communication, through education, and through promotion of prevention for both clinical and population settings. Many of you may know this already.

We also then, because we have worked very hard at this, want to assure that this actually happens, moving from research and process to actions and then to being accountable for what gets done.

We do this through bolstering the public's confidence that this information is used appropriately and that the services we do meet agreed-upon goals for effectiveness, accessibility, and quality in research. We look also at how to assess that to make sure things are happening. We do this through a clear evaluation methodology and then quality assurance and quality control.

Then we look to ways we can work together to enforce laws and policy standards and to assure that

we have the ability to get this done through the development and assurance of a competent work force.

In our policy questions, then, given that broad area, we want to really nail down what to do. So we have questions about the characteristics of the diverse systems of health care, how management and delivery influence the provision of genetic tests, and then, subsequently, how clinical or preventive services work.

We also were asking what are the leading opportunities and responsibilities for public health systems to contribute to the development and implementation of the new genomic knowledge and technologies to improve health, to prevent disease, and to address health disparities.

Specifically, we want to drill down to look at a couple things. First of all, there are the opportunities, challenges, and benefits of incorporating genomics into existing and future public health investigations and surveillance systems to advance knowledge.

There are also opportunities and

responsibilities for incorporating evidence-based genomics and knowledge and technologies into public health programs to improve health and prevent disease, the actual application of this work.

We also wanted to look at the public health infrastructure and to partner, we believe. Part of our major focus in public health is collaboration and working within and across healthcare delivery systems, employers, businesses, communities, academia, media, and others, particularly consumers.

We wanted to also know what steps can be taken to address ethical, legal, and social issues in public health genomics research and practice. Drilling down, then, we want to know how does informed consent for DNA testing in public health differ from informed consent for other public health services and in clinical practice. Dr. FitzGerald brought this up. It is a clear issue because our question is, under what circumstances is new consent for archive specimens needed also for public health investigation.

We wanted to know what are the immediate and long-term benefits and risks of population-based

disease registries, as well as how can concerns about potential stigmatization of the population groups result from research on testing programs be addressed.

We wanted to know what policies should be in place to share large amounts of data collected through gene, environment, and disease association studies, and we want to look at emerging concerns as technologies evolve. Will it become possible to test for multiple layers of biological challenges which reveal chinks in the bodily integrity before classical clinical symptoms emerge. Will advances in technologies and knowledge shift current conceptions of injury in toxic tort suits or the preexisting condition exclusion in GINA.

We also wanted to know what tools are needed to understand how genes and environmental factors, physical and social, interact to perturb biological pathways and cause injury or disease. How does the federal investment in genomics encourage translation into population health benefits. Is it cost effective to tailor interventions based on genetic information.

Lastly, what steps must be taken to assure a

competent public health workforce with a sufficient knowledge base and skills to ensure that the appropriate use of genetic information to promote health and prevent disease, as well as to educate the general public to be informed consumers of genomic applications.

Given that, we wanted to know how can the public health agencies prepare the workforce and their constituencies to ensure that information about gene-environment interaction is used appropriately.

So, how to get there. For short-term actions, we believe we can organize sessions such as SACGHS meetings to expose the field of public health to genomics policy questions associated with advances in understanding gene-environment interactions and for in-depth discussion of the potential for genetic and genomic testing to exacerbate and lessen health disparities. Thus, the work with these clusters.

We believe that we can perform a systems review of relevant agencies to assess mechanisms that are already in place or can be in place to disseminate information about the distribution of genotypes in

different populations and to assure effectiveness, accessibility, and quality of services.

We also wanted to look at the potential of using SACGHS as a forum to promote collaboration within and between DHHS agencies for efforts such as preventing the stigmatization of individuals, families, or populations at risk for or with genetic conditions and for implementing an assessment process that will provide guidance for how and when genetic tests can be used to promote health and prevent disease.

You can see our focus is on getting the work done. Again, we suggest brief reports on selected public health topics such as the impact of genetic and genomic testing on health disparities, how characteristics of different healthcare systems influence provision of genetic tests and subsequent clinical provision of preventive services, building a competent public health workforce to ensure appropriate use of genetic information to promote health and prevent disease, or whether it is cost effective to tailor interventions based on genetic

information.

Of course, like everyone else, we really believe that this needs to be looked at very much in-depth, particularly at issues related to public health genomics in the areas of disparities, gene-environment interactions, and population-level testing. We left off, I think, workforce development.

DR. WISE: Comments, suggestions, or questions for Joseph?

[No response.]

DR. TELFAIR: It is clear. What can I say?

DR. WISE: We will have time to come back to these issues and discuss them both more globally but also in greater depth.

We will move on to Cluster No. 7. Mara.

Cluster No. 7: Genetics and the Future of the Health Care System

Mara Aspinall, M.B.A.

[PowerPoint presentation.]

MS. ASPINALL: Thank you, Paul. As Cluster No. 7, I can say it is not about money, value, education, power, empowerment, or responsibility; it

is about all of those. It is not even about today but really about the future. Cluster No. 7 is not looking at any one issue but looking very broadly across the healthcare spectrum to say how do we get prepared for the future that we all talk about. What came up yesterday is, today is useful but how do we ensure the infrastructure for the future.

In Cluster No. 7 there is a focus of two key questions. Personalized health care: is it achievable; what are the costs and what are the benefits. Secondly, what are the infrastructure changes needed to foster or even adopt personalized medicine or personalized health care in the broader perspective. Those are the key pieces.

When we look at the background, I'm not telling anyone anything new here, but the current system is not working. It has high cost and poor outcomes. That doesn't mean there aren't parts of it that are working well, but as we have heard many times, the current trend and system is unsustainable and we need to move beyond that. As we found out in many areas, both health care and more broadly -- the

nursing shortage is a great example -- we need to be prepared for how the future will change.

Let's look at those two key focus areas. The first one is value, cost, and achievability. The assumptions that we made here are that genomic medicine has the ability to benefit public health and that new technologies will come along, some of which will decrease cost but some of which are going to increase cost, depending on how widely or narrowly you think about the cost.

But this is still a technology and a science that, whether you call it in its infancy or adolescence, is not as robust as it will be into the future. So, considerable research will continue to be needed.

Lastly, and really the core of personalized health care, is that the greater understanding of the epigenetics, the genetics of subpopulations, is critical to benefit all people and that we need to understand how to target those interventions, whether they be diagnostics, drugs, devices, or other interventions. That is the key.

The second piece is, what is the infrastructure that we need. This overlaps with several of the other clusters. First, in healthcare delivery, how do we ensure that we have cost effective delivery and what we believe is a likely need for increased genetic counseling. In this cluster, we define genetic counseling quite broadly.

Secondly, we talked about workforce. We are likely to need additional clinical lab workforce, even if they are doing different jobs than they are doing today.

Thirdly, health information technology. Current HHS Secretary Leavitt has made a big deal about the critical piece of integrating health IT into the delivery of care and into personalized medicine.

Fourth, systems to monitor that so we don't roll out new systems of genomic medicine and not have the ability to say is it effective or not.

Lastly, should the government play a role in incentivizing business pursuits -- and we mean by that the broadest definition of the industry, not just for-profit but not-for-profit -- in the pursuit of

diagnostics and genetic therapies.

We have a number of policy questions that come out of this piece. Again, the objective here is to really look forward and say how should the government -- by that, individual agencies through HHS -- help invest resources in genomic medicine.

No. 2, should the government be part of adopting a new healthcare delivery. You will see in a minute there are a number of different models that have come out. Should the government promote them. Should they get involved. If we anticipate that the medical home model might be the one, what is the infrastructure that needs to be set up to ensure that we can handle the onslaught that is likely to occur.

No. 3, financial incentives in the workforce. Are we facing a crisis that many would say today that we do not have enough people in the healthcare workforce to be able to offer these kinds of tests and this kind of science going forward.

No. 4, health information technology. What else can HHS do to promote this. Specifically, we talked about development of electronic health records

and the digital storage of health data. Again, assumed many of the clusters is we are gathering much more information than we have ever had. How do we ensure that we can access that information in the future.

No. 5, quality. It is the core of the system and the core to what has been described in terms of confidence in the system. How can we ensure that indeed the quality is there and that we are able to monitor this for public health benefits. Are there surveillance systems in addition to what we have discussed before that need to be put in where we need to put the ground work in today.

Then, finally, the issue of incentives for genetic diagnostics and targeted therapeutics. This is not just about diagnostics, this is about genomic medicine in its broadest piece.

Lastly, how can the government think about these differences in population that ensure that we are focused not just on subsets that exist today but important variations amongst the populations in the future.

So, the short-term action steps. First, we talked about getting together the chief medical officers from the health plans. That is public and private health plans. They have been a group which we would say has been somewhat underrepresented in our discussions and they are a critical piece to ensuring that the infrastructure going forward is covered. We wanted to put them together, both public and private, to get their view of the future and ensure they are part of the team.

What we then looked at, and not on the slide, is working with the other HHS agencies. Many of the agencies have begun to look at what they believe is the future of health care, and we want to ensure that we are not recreating work that has already been done in other agencies.

In the last few slides, we look at brief reports. I won't go through each of these individually. They basically mirror the policy questions that say how do we look at this.

The reason we talk about brief reports, quite frankly, for a big piece of this is that it is

possible for this to go on into the future such that we won't be looking into the future. We think it was critical to ensure and put a stake in the ground on a couple of key issues, particularly around tools and incentives and the health information technology.

Lastly, there were two areas that we did believe may warrant some in-depth work. In the area of health care delivery, one example is the medical home model, the customized care centers, and genetic counseling to work with some of the other clusters to ensure that we understand what the infrastructure will need to do that going forward.

Lastly, the genetic and epigenetic variations, in which, again, working with other agencies is critical in the true delivery of personalized health care. What we wanted to ensure is, if there was data that was needed for populations that are today not represented, that we begin getting that data today and acknowledging what the gaps are so that when we have the ability to go forward that there are not groups that are left out.

Thank you.

DR. WISE: Questions.

DR. EVANS: I just have one question. It seems to me that on two of the slides, Nos. 64 and 67, there is a fairly exclusive focus on encouraging students to pursue clinical laboratory careers, which confuses me. I think if we are talking about the healthcare system that perhaps an emphasis on both laboratorians and clinicians who focus on genomic aspects of health care. I wouldn't want to confine it to laboratorians.

MS. ASPINALL: I think that is a great addition and we should broaden it. Because of the attention on the crisis literally today there was a focus on that, but I think as we look forward we need to look more broadly. Thank you.

DR. WISE: Yes, please.

DR. DREYFUSS: This is the mirror image of the questions you were asking yesterday. In several different places you talk about incentives to find diagnostics and therapeutics. I would rather have that say "promote the development of diagnostics and therapeutics" because in fact sometimes I think we see

that these incentives get in the way of actually promoting them. So it is not just creating new rights, necessarily. It might be creating the right to use research that somebody else has a right on.

MS. ASPINALL: I think that makes a lot of sense. That was the spirit in which we were intending to do it. We will ensure that it is changed that way.

DR. WISE: Steve.

DR. TEUTSCH: Just one addition to the EHR story, which was mostly about data storage and records. It is really, I think, given the information that is likely to come out and the need to manage it intelligently and the difficulty of keeping it in everybody's heads, the clinical decision support part of that translates all this information into something that is useful, interpretable, and actionable.

MS. ASPINALL: Yes, so it is more specific. I think that makes sense. The report that we got yesterday on personalized health care actually has some wording that might be useful to incorporate into that. So we will change that as well.

DR. WISE: Marc.

DR. WILLIAMS: Just to add on to that, I think that an overarching theme for all of the different clusters is the idea that almost all of them will need robust information technology in a variety of different forms to actually make it happen. I think that is implicit in all the presentations, but we probably need to be more explicit about that and also be very intentional about looking to partner with the other groups that are working on this.

In particular, as the new public-private partnership that is going to be the second iteration of the American Health Information Community gets up and running, we really need to make sure that the engagement we have had with that group to this point continues.

DR. WISE: Other comments or questions specifically on No. 7?

[No response.]

DR. WISE: Thanks, Mara.

Discussion

DR. WISE: We have heard that it is all about money, value, knowledge, power, empowerment,

responsibility, the future, but particularly to help guide our discussion, it is important to always remember that it is always about ego.

DR. WILLIAMS: On that note, it is time to go over these cluster issues in greater detail to, number one, make sure that we are reflecting in each cluster description the action steps that were outlined that were reflecting the insights and wishes of the Committee.

Also, we want to look at linkages. We very clearly picked up linkages that cut across. Now, we could turn all these clusters into one cluster, but that wouldn't be of particular utility for our work as we move into the future. But there may be ways to group these clusters in our thinking and particularly in the way we present them to the public and particularly to the new administration.

Steve suggested that we can walk through each cluster and have an opportunity to talk in greater detail. Maybe, Kathi, you can put up Cluster No. 1.

Also, in your handout in your book, at the

end of Tab 5 is a summary grid of the major points made in each of the cluster presentations, particularly the outline of the action steps. So you could also use that to help ground your thinking and the conversation regarding each of the clusters.

DR. TEUTSCH: What we have to get by the end of the day is a pretty clear sense of how we want to organize this and what our priorities are. Although we didn't say it explicitly, I think we have a number of things that we can do, and we have captured them here.

There are things that we have already made recommendations about that we can just monitor. We have things that we can do over a fairly short term or in a brief report or some more in-depth reports. We need to figure out what that portfolio of work ought to look like.

So as we march through each of these, I think it would be helpful for us to begin to think about, within each cluster, which of these things we think are most important. Clearly, this body of things that we have laid out far exceeds our capacity.

So as we begin to sort through them and think about how they might be rearranged and so forth, we can begin to get a sense as to which of these things are of the greatest importance. It will help us to sort through and consolidate clusters if that is what we need, and begin to set some clearer priorities and organize our work going forward.

DR. WISE: It is fair to say, though, that we still remain flexible about these next steps given that the administration coming in may outline priorities that would require our being responsive.

DR. TEUTSCH: Absolutely.

DR. WISE: So while, as Steve suggested, it would be very helpful to get some clarity about where the Committee feels we should be going, we recognize that we will likely have to be flexible in responding to new concerns and priorities of the new administration.

So, discussion regarding Cluster No. 1. Please, Marc.

DR. WILLIAMS: The good news, if there is any, regarding Cluster No. 1 is that we are not

indicating we need to do any reports, since we just did one. There is a lot of work that is ongoing that is going to continue to happen.

As I was reflecting, though, on this and the discussion from yesterday and today, the thing that probably isn't represented there that does have an impact that we should be intentional about is looking at coding systems.

The current CPT and ICD-9, which is going to be transitioning to ICD-10, how is that impacting. It certainly has an impact on reimbursement, as we talked about yesterday. It has an impact in terms of collecting data, which is going to influence Steve's cluster about utility.

So that may be a cross-cutting issue that should be added to our portfolio for study.

MS. ASPINALL: I would wholeheartedly agree.

DR. FERREIRA-GONZALEZ: I want to continue to echo that, but I think this could be a short-term action that we could engage in already.

Like I said earlier, we know the system is broken. It's not working for us. The professional

organizations are already trying to look at these issues, and we need to engage them actively. Maybe we can have them come together in different groups so they can talk to each other or for us to continue to evaluate that. They have very in-depth expertise because that is what they do every day. That might be a way to go about this.

DR. WISE: I have a question. This issue has come up. It has been the subject of considerable deliberation by the Committee and discussion. Yet not a lot has actually happened in response. It is still an issue, still a problem.

What do you see as the primary obstacles to moving forward with this, and how would that elevate some of these steps to ensure that we are really finally going to be addressing the essential barriers to making headway on this?

DR. WILLIAMS: I'll take a crack at it. Some of the barriers relate to a lack of clarity relating to some of the interpretations of the statute and regs.

In the letters that we have been writing, we

have asked specifically for some clarification. Much as we were talking about yesterday, understanding exactly what can and can't be done will inform strategies going forward about how to address some of these issues. Lacking that clarification does hold this up.

So the hope is that some of the questions that we articulated very clearly about wanting some direction and clarification will be forthcoming in the letter later this month. But if that doesn't happen, I would certainly see as the new administration settles in that we revisit those issues and say we really would like some clarity around these issues.

The second barrier is that different stakeholders hold different pieces of the puzzle. ICD is maintained in one area. CPT is under the control of the AMA. So there are lots of different players in there. As Andrea has pointed out, it is sometimes difficult to get the engagement or to get everybody at the table.

So maybe there is a role for the Secretary to convene, perhaps with SACGHS as the facilitator of

that, everybody around the table and say how can we work these things out. But it is a very arcane system to have to navigate and to try and pull all the different pieces together. That does slow progress.

DR. WISE: Jim.

DR. EVANS: Not that I have a solution for it, but just to point out that you are also, in this context, fighting a long entrenched system of procedure-based reimbursement. Genetic services, at least so far, until we invent the genetoscope, is not a procedure-oriented field.

So it is a very broad issue. There are other constituencies in the same fix: general internists, pediatricians, and psychiatrists. So it is a rather pervasive issue and this somewhat irrational system is an entrenched system. That is an obstacle. I'm not sure exactly how you get around it, but maybe enlisting the kind of support of other large and important specialties for which this is an issue.

DR. WILLIAMS: Yes, that is very well taken. Of course, the problem that we have is not only on the clinical side, where there are the issues that you

have articulated. Genetic counselors, being a relatively new profession, really don't have any official standing in any of the statutes in terms of being articulated, as opposed to nurses and physician's assistants. That also creates a barrier.

But even on the laboratory side, which is a procedurally-based specialty, it is clear that the reimbursement around those procedures is not adequate for the actual cost of the things that are being done. So that is an issue.

There was another point, but it has lost me.

DR. WISE: Steve.

DR. TEUTSCH: I was just going to ask Jeff a question. I know I'm putting you on the spot a bit, Jeff, but you have been wrestling with these issues. We have had some wonderful dialogues with you and colleagues at CMS, and you have been wrestling with these issues. Can you elucidate some of the things that we might do that would help move this forward?

DR. ROCHE: Hi. Good morning. For those of you who may not know me, I'm Jeff Roche. I'm an alternative for Dr. Straube from CMS.

CMS is indeed an interesting institutional road to navigate. I'm, at the moment, about six weeks past a year, so I'm a relative rookie.

One of the more interesting publications that I found that was useful is something that many of you may already know about which describes how CMS addresses payment and reimbursement issues related to new technologies, which is a larger basket with which genetic and genomic testing issues and genetic service issues in general are in competition. This may be very useful.

Again, it isn't a one-pager. If it were available to anyone to read, it might be very useful.

I welcome the opportunity to continue to work with Sarah and others from the NIH staff who have been trying to get us to consider these questions, which, I think as Marc correctly and accurately pointed out, are based on an old system designed for a different purpose, which is confronting a rapidly changing field and which has both promise and, in some ways, areas where the promise has not been fully demonstrated.

I call to mind perhaps a recent editorial in the New England Journal of Medicine just last Thursday where the headline was "Payment Now, Possible Benefits Later." The discussion was actually fairly stringent about CMS's perhaps errors in covering certain imaging procedures and the major effect on Medicare costs that that error may have led to.

I think we want to be sensitive to the needs of any community, especially the changing community in health care, but I think we need to be aware that the consequences of our decisions at CMS, as for almost any other government agency of course, have enormous consequences.

So, as I say, we continue to look forward to engaging with both your staff and other Committee members and trying to resolve some of the specific issues, but I think this cluster does point out very nicely.

DR. TEUTSCH: It gets back to Jim's point. As we look towards health reform, it probably also means financing reform in how we pay for services, whether they are going to be episode or bundled or

some other ways of reimbursement. Whether we handle this under the coverage and reimbursement set of issues or whether we handle it under health reform, it is clearly one of those things that is going to be tightly linked because it so strongly shapes how care gets delivered in this country.

DR. WISE: Did somebody have a comment?

DR. WILLIAMS: The other point I was going to make is about the money. The systems we have heard about are not a target-rich environment.

When we are talking about that we should get a bigger piece of the pie, that inevitably means that somebody else is going to get a smaller piece of the pie. That always is problematic when it comes to those types of discussions. That is just the environment that we have.

This ties in with Steve's cluster. We could make a much better case for ourselves if we moved away from being the faith-based specialty that we have been in genetics, which is you have to believe that what I'm doing is really good. In God we trust all of the data. Paul has heard me say that before.

But the bottom line is we do need to develop evidence around the value of what we do because then we are not just whining about the fact that we are not being paid.

DR. WISE: Andrea.

DR. FERREIRA-GONZALEZ: I think that the issue that Jim brings up is a good one. I think there is a lot of momentum right now not only in the laboratory community and the genetics community but across all medicine to start looking at some of these issues.

I think this issue crosses different clusters. Even if we look at Cluster No. 1 where we are going to monitor this, I would strongly recommend that this becomes another point in the clusters of the future. It has a huge impact on how we actually move. Even though it might be in two different areas, we need to connect them.

DR. WILLIAMS: That is a really good point. Mara can speak to this. She and I were the only two Committee members that were at the Summit on Personalized Health Care.

I haven't read the report thoroughly, but in talking to some of you who have actually been through the report already, it probably wasn't conveyed strongly enough in the report about the feeling of the attendees there that none of this is going to happen without substantial reform to the current system. That was really a key element of that summit. I think that that is a really right-on point. I agree that this needs to be carried forward with what Mara has been doing.

MS. ASPINALL: I was just going to add, people may be familiar with it. About six weeks ago, there was a summit in Utah around personalized health care. The report that we have was a key piece of that, really looking at many of the issues that overlap with Cluster No. 7, which says if personalized health care is going to happen naturally, how do we ensure we have enough resources, beds, systems, information, data, to have that happen.

But during this summit, which was relatively small, we were broken up into four areas. One of the key areas had two major recommendations, and I would

say the number one recommendation coming out of the business area -- and again, "business" doesn't mean for-profit businesses but commerce in the broadest definition -- was reform of the reimbursement system.

So I think this one has to be a priority. There are a number of groups working on this, as Andrea mentioned, in the professional community, and as Marc knows, I was asked to head up a piece of that from the Utah summit on looking at a fundamentally new system and potentially replacing the CPT code system in order to do that.

I think the good news is, with health reform potentially coming, there was a real groundswell of support to say we need to look at this and how do we do it.

What I'm intrigued with in Cluster No. 7 is, if the reimbursement system changes there is no question utilization is going to change. If utilization is going to change, how can we ensure that we are ready for that and people are not therefore denied access for very different reasons because we don't have the resources there to serve. That is how

I see the two working together.

DR. FERREIRA-GONZALEZ: Also, I think we need to work together with the clinical utility piece. As we continue to look at new ways that things are going to be reimbursed, the knowledge is generated to have a very good idea on the clinical utility of the tests over time. So, how do we make sure that as these technologies move forward into practice or not, that we gather more information and make decisions but, at the same time, make sure whatever we communicate or continue to work on within these three different clusters doesn't stifle the movement of personalized medicine.

MS. ASPINALL: That was the second recommendation coming out of Utah, ensuring that the public and professionals have confidence in the products coming out. The clinical utility piece may go a long way toward ensuring that confidence so that people aren't saying, well, does that work or not work. If we don't have confidence in the system, we can't change the reimbursement and then we can't assume adoption.

DR. FERREIRA-GONZALEZ: But to gather that clinical utility information sometimes takes time. We need to see how that process over a gray area of material is now black or white. That is something to keep in the back of our minds.

DR. WISE: Julio.

DR. LICINIO: One of the things, though, that I think is very important here that we are somewhat missing is that the big paradigm shift now is this direct-to-consumer thing. By the time a doctor or a healthcare provider or clinical geneticist or genetic counselor talks to somebody about some genetic finding that may be relevant to health, the person has already paid $400, which is relatively affordable, to 23andMe and has their 500,000 SNPs all there.

They actually have a service that they call, I think, a Genome Browser. If a paper comes out today in the New York Times, they can immediately go and check do I have that SNP or not, even before the scientific community is aware of the finding.

I don't think we are doing it in a deliberate way, but I think we are being very

traditional and paternalistic. I think we are trying to find guidelines or policies to make recommendations to the field. But, is that what the public really wants.

I think that one of the recommendations should be a broad survey or community engagement process. I have done this before. It becomes very sticky: who speaks for whom? Who represents what? You cannot engage the whole country. So, which group do you engage; how representative are they. That is why before the word used was "consultation." That held with some people but not with others, so that has been changed to "engagement."

But I think that some element of community engagement should get an impression from the public. Or, recommend some kind of broad survey as to what the public really wants or needs. We may be preaching one thing, but if the public is willing to pay and get the information anyway in spite of the risks, they will do it.

DR. EVANS: Does that really belong in this cluster or does that fit better in Sylvia's cluster?

DR. LICINIO: Probably in Sylvia's.

DR. EVANS: I think your points are well taken, but I don't think they are necessarily part and parcel of reimbursement by third-party payers because of exactly the issue that many of these people will pay out of pocket for such information.

DR. WILLIAMS: Yes. I think the overarching theme that I'm hearing that really reflects all the comments is that clearly we as a relatively small group are not somehow going to reform the system. Yes, I know we all would like to.

I think that, as I see this, we don't necessarily have a good idea about where things are going to go, whether it is reform of the traditional healthcare system, whether it is a consumer-driven system that has radical alterations in reimbursement, or how evidence is going to play into this.

It seems to me that the role of Cluster No. 1 is to be able to assess where things are going, to hopefully be engaged as best as possible with the movers and shakers that seem to be making a difference in terms of the reform effort, and then being nimble

to say what are the strategies that would work best given whatever system we end up with so that we can basically be ready to provide what is needed at the time that new things are rolling out.

It seems to me less a task of pulling everything together and creating something that is a static document in some ways and is more of an ongoing, nimble process to try and respond to a rapidly changing environment, and just basically trying to develop as best we can the information connections that allow us to really have a good sense of what is happening.

DR. TEUTSCH: To build on what you just said, it seems to me that what I'm hearing here is that this particular cluster is dealing with a whole set of tactical issues that we are really facing right now. They deal with the system as it currently is and how to optimize those issues.

There is a whole set of linked strategic issues about what the future is going to look like which probably belongs more in the health reform future issues.

So it might be a way to keep our eyes on, yes, we have a whole bunch of these acute, short-term issues that we need to address, but we understand that probably over the longer term we need to deal with the broader issues. If we are going to move to a personalized healthcare system that adds real value, what do we need for that system.

DR. WISE: Other comments or suggestions before we move on?

[No response.]

DR. WISE: Why don't we talk formally about Cluster No. 2. Oh, I'm sorry.

MR. KIRCHNER: Peter Kirchner from DOE. I wanted to bring up the relationship between clusters again. That relates in particular to the reimbursement and how you are going to address the importance of reimbursement for any given area to the information coming out of Cluster No. 2 regarding the strength of the associations that have been created.

I'm just wondering whether there is some kind of a plan as to what will be a minimum amount or a necessary amount of evidence before you can address

that.

Some time ago, the internal medicine organizations used to put out data of high correlation versus medium versus low, depending on the amount of evidence that was available for a given treatment or a given association. I don't know if anything like that might be useful in trying to address this difficult issue of when do you cross that threshold so that it is justifiable to spend public money on reimbursement.

DR. WILLIAMS: I think that is an excellent point. That is clearly something that is at the center of the healthcare reform debate in this country. The issue is that the reimbursement system, right now, essentially, is disconnected from the system of evidence and quality. It is basically based on work units and how much you do, and an evaluation system that, as Jim pointed out, tends to favor procedures over non-procedural activities.

I won't use any denigrating characterizations about thinking and not thinking, as some have chosen to do.

I think you are absolutely right. We are

going to have to take the step in this country to move in that direction where reimbursement is really tied to best practice, to quality outcomes, and to evidence-based medicine.

How that is ultimately going to play out, and how that is going to look is not clear, but it is incumbent, I think, on the field, and in particular, under Steve's Cluster No. 2, that we are able to deliver on the evidentiary standards that are going to emerge and then use those to tie into how the reimbursement reform is going to work.

MR. KIRCHNER: The other thing I wanted to mention is that, in addition to reimbursement, should there be consideration in your cluster regarding payment for additional research needed to bring the level of evidence up higher.

Now, CMS has actually been doing that for the last several years. I remember when it announced that it would begin to reimburse certain types of research as long as it met very rigid protocol standards.

That, I thought, was very important and

might be applicable to the kinds of things that you want to do.

DR. WILLIAMS: I think that's a good point. The idea of coverage with evidence development has had some visibility. Jeffrey could certainly comment on this. I think that, as with anything, there have been some good things and some bad things that have come out of it.

The problem, of course, is that we sometimes decry the idea of how long it takes for physicians to adopt something. What we forget is that it takes them just as long to unadopt something.

I think one of the problems that has been looked at with coverage with evidence development is the idea that we allow a procedure and develop the evidence. Then we find out this really doesn't work, so we decide we are just not going to do it. But now everybody is in the practice of doing it and it is hard to extract that out. I think that has been reflected in some of the issues around imaging and, in particular, some of the more expensive treatments in the oncology arena.

Of course, the other thing that gets involved in that that we haven't even talked about, the real elephant in the room, is how a single liability case can dramatically impact the reimbursement landscape.

If we look at the bone marrow transplantation in breast cancer situation, one lawsuit against a payer for tens of millions of dollars essentially overnight changed the reimbursement policy in this country and caused payers to cover a procedure which, as evidence was developed through coverage, we realized really was not worthwhile. There are estimates that it cost this country in the range of $500 billion. I think that is the right number of zeroes associated with that. But it was a heck a lot of money, not to mention the morbidity and mortality associated with a procedure that was essentially in the vast majority of cases futile.

While I think that we are also going to see how this can play out, we should realize that that is also a double-edged sword. But it certainly is

something that we would want to consider as part of our toolkit.

DR. WISE: This discussion of Cluster No. 1 has identified that there are clearly linkages across different clusters and that coverage and reimbursement issues will depend heavily on the success of Cluster No. 2 for sure and that will be an important contributor to Cluster No. 7, healthcare reform.

But as much as Cluster No. 1 puts a demand on Cluster No. 2, Cluster No. 2 also must respond very much to the immediacy of Cluster No. 1, reimbursement and coverage, and be intensely practical and relevant in how it goes about its business in order to truly be engaged into the issues raised by Cluster Nos. 1 and 7.

Why don't we move to Cluster No. 2 and discuss it more formally, even though we just moved into it to an extent. Any specific comments regarding Cluster No. 2, which is ensuring the clinical utility of genetic information? Marc.

DR. WILLIAMS: I think the biggest thing that this group can do is to really highlight the

funding disparity across the translational research arena.

As we heard from Muin when he presented at a previous meeting looking at the T1 through T4 translation, about 97 percent of the current research dollars are residing in the T1 basic science arena, which leaves very little for even establishing what might be considered rudimentary clinical validity, not to mention evidence around utility.

I think that one of the strategies that we should definitely look at and promote as members of SACGHS is some redistribution, if you will, of the research funding to move more monies into the T2 and T3 areas. That will allow development of evidence around clinical validity and clinical utility, which we all recognize is going to be exceedingly important if we are really going to move this down the road.

DR. GUTTMACHER: You are asking for redistribution, not an increase in spending, to allow that?

DR. WILLIAMS: That is what I said.

DR. GUTTMACHER: So you are taking that

money away from other areas of research. I just want to make that clear.

DR. WILLIAMS: Yes.

DR. GUTTMACHER: I think some of us might have difficulty with that.

DR. WISE: Alan, tell us what the difficulties might be.

DR. GUTTMACHER: Well, exactly what research are we taking the money away from?

[Laughter.]

DR. WILLIAMS: You have heard me say this before, but I will put it into a public context. In some ways I think we have a hammer-and-nail problem. The Human Genome Project is, arguably, one of the greatest scientific achievements of the past millennium, if you will. I don't think that is overstating it.

One of the consequences of the funding for the Genome Project was the creation of a tremendous amount of sequencing capacity. In some ways, that has created a huge hammer so that every problem that then appears from a research perspective is to say, can we

solve this with sequencing.

So we go from genome to HAPMAP, to genome-wide association studies, to sequencing organisms and all this stuff. There is a lot of that infrastructure. There tends to be a bit of tunnel vision looking at, can we continue to solve the problems that we are dealing with by using this incredible technology.

My contention would be that while there is still value and knowledge to be gained from doing that, we can't continue to focus the vast majority of the research efforts into that. We have to take some of that and use different techniques to be able to say how we can do something with this information that is actually going to provide direct benefit to health care in this country. That requires a different model than we currently have been having.

That may be a simplistic view of it, but it does seem to me to be part of the problem.

DR. GUTTMACHER: I would suggest that might be a form of genetic exceptionalism. What you are really arguing, I think, is that the federal research

dollars in general should be spent less on basic research and more on applied research, health services research, et cetera.

If that is what you are arguing, you might want to put it in that context rather than saying genomics per se. I think that is a larger discussion. This Committee, of course, does advise the Secretary of Health and Human Services, so that may be within the agenda of the Committee.

But I would think it is a zero-sum gain to some degree. Or one could ask for increased funding. Those are two different options. But I think one needs to be aware that of course good applied research needs to be based upon good basic research, which I know you wouldn't argue with.

I certainly would not argue against the idea that we ought to know more about what we are doing in health and health care, but we shouldn't just be in favor of apple pie without thinking about exactly what else we are not going to be able to cook and whether the apple pie is going to taste good.

DR. WILLIAMS: I certainly wouldn't disagree

with expanding the discussion. I took the G in SACGHS seriously there. But you are absolutely right. I don't think that this is a problem that is unique to genetics and genomics. I think it is a problem that we are struggling with in terms of that balance of basic science research, which is absolutely critical, and then moving that into the translational realm.

I think there has been a relatively strong argument made that there is, and there has been, an imbalance between those two areas that probably does need to be addressed. Yes, it does mean that it is unlikely that there is going to be additional money injected into the system. It is not impossible, but unlikely. That means that there may be some redistribution that will be necessary.

But looking at what our task here is relating to the charge that we have been given by the Secretary, which is to address the needs of the American people relating to these new opportunities to improve health and reduce disparities, I think it is fair to say that we do need some additional evidence of utility to be able to move that agenda forward.

That does require some commitment to research.

DR. GUTTMACHER: I would certainly agree that evidence of utility is necessary and research should be done in that area, but I would also argue, of course, that until we understand the basic science of genetic factors in health and disease there is much less point in applying that to health care.

We need to understand where it is along that pipeline of base translation. One could certainly discuss it, but we really need to understand those factors before we run out and study their use in health. We need to understand them and, as we understand them, incorporate them. That is an area of research that I think would be worthwhile.

DR. WISE: On that note, I have been impressed that the conversation between how much money should go for basic research versus applied research has been overwhelmed by the fact that applied research or comparative effectiveness or clinical utility research is going to be demanded by cost containment and by restructuring healthcare services. It won't have much to do with the conversation we just have

had, and we need to be cautious that we don't allow this to become a tension that could be unhelpful.

But, to what extent should the clinical utility arguments be framed within healthcare reform and cost containment arguments? In other words, it is a shift in the frame of how we think about clinical utility and it also puts a different set of demands on the clinical utility community to provide products that directly and quickly can address issues of comparative effectiveness and cost reduction.

DR. TEUTSCH: On that note, why don't I suggest, since we have come to 10 o'clock, that we go ahead and take a break. At 10:15 we can come back and get public comments. Hopefully, we have folks here who do have some words for us. Then we will return to more on clinical utility, for which I have already seen some hands emerge. We will get back to those.

Go ahead and take a 15-minute break and meet back at 10:15.

[Break.]

Public Comments

DR. TEUTSCH: One of the important things

that this Committee does is to serve as a public forum for deliberations on all of the issues surrounding the human health and societal issues in the area of genetic technologies. So we always appreciate it when we get comments from the public and welcome all the perspectives they have to share with us.

I have a list of two individuals who are scheduled. If there are others, please let me know.

We would like to first hear from Sue Friedman, if you are here, who is the executive director of FORCE, Facing Our Risk of Cancer Empowered. Dr. Friedman, we look forward to your comments. Thanks for being here.

Comments by Sue Friedman, Ph.D.

Facing Our Risk of Cancer Empowered (FORCE)

DR. FRIEDMAN: Thank you for having me. I want to thank the Secretary's Advisory Committee for inviting me to present today. I am founder and director of the national nonprofit organization FORCE, which stands for Facing Our Risk of Cancer Empowered. We deal specifically with hereditary breast and ovarian cancer and families that have been affected by

the disease.

I came here from Florida to your weather, so I'm very motivated to speak before this panel. This is freezing cold for me.

Part of our mission is about advocating for the health and well-being of our community of people and families affected by these hereditary cancers. The goal of my testimony is really to alert the Committee about a growing issue that we are seeing and really trying to document, but it is a problem.

Once a test is out there and once it has been offered, the consumer has assumed that it has clinical utility and that it has been validated. I saw that you are looking at those issues. I think it is really important.

One of the problems with the tests that are already out there at CLIA-approved laboratories is that there really is very little oversight. We know there is a lack of knowledge and information in the healthcare community and on the part of consumers, and that gap is being filled in by the companies that are developing the tests. Certainly, I think, they have a

place at the table, but I don't think they should be the exclusive source of information to not just consumers but the healthcare community.

What we are seeing is not just a direct-to-consumer marketing of genetic tests but also a direct-to-doctor marketing of genetic tests that really wouldn't have been allowed had they been pharmaceutical companies and had to go through the FDA oversight process.

We are seeing this, literally, daily. I often staff our help line and we get calls every day. It is wasted dollars. The wrong tests are being ordered. The wrong individuals within a family are being tested and people are being given wrong information about what the results mean. A lot of this is based on the fact that the company that develops these tests is providing doctors and consumers with all the information that they are getting. There is no one else filling in the gap.

Obviously, this doesn't happen when people are referred to genetics experts. There are standard-of-care guidelines for hereditary breast and ovarian

cancer. I sit on the NCCN panel that developed standard-of-care guidelines for genetic testing. It does say that there needs to be a three-generation pedigree, there should be access to genetic experts, and there are some clear guidelines. These are not being followed.

We are hearing a lot of cases, and I provided some examples. I sent a letter that outlined one woman's experience. She was allowed to fill out her own genetic test. She ordered the wrong test, and this was at her OB/gynecologist's office. Based on her test results, she proceeded with a lumpectomy, believing that her breast cancer was not hereditary, only to find out after her lumpectomy that the wrong test had been ordered. That would not have happened, in my opinion, had she sought out expertise from a genetics expert.

Her letter is very telling because we hear this a lot. People love their OB/gynecologists, so they don't want to indict them. They almost feel bad that their OB/gynecologist didn't know how to guide them through this. In many cases, we can look back

and find out that that OB/gynecologist just had a recent visit from the company that is making the genetic test that they are selling and that the information that they are getting is not complete information about the test, what it means, and how to properly do a risk assessment on someone to determine if they are the appropriate person for genetic testing and if this is the appropriate genetic test.

We are seeing cases where people are being told their tests were normal when they have a mutation. We have seen cases where people have been told while they were driving that they carry a BRCA mutation. We had one 23-year-old who was told by a nurse that her risk for breast cancer was 85 percent. She was 23 and her risk was not 85 percent at that moment. So, inappropriate information is given at inappropriate times.

We have reports of people being ordered full-sequencing testing when a $300 or $350 test would have been more appropriate. Insurance companies are footing the bills on this, or the consumer is footing the bill, or taxpayers are footing the bill. This is

happening a lot.

With respect to where the information is coming from and where consumers are getting the information, I have followed sales representatives at conference calls and listened as they have promoted testing to doctors and nurses and said that they do not need to refer people to genetics experts. I have heard that on more than one occasion. At a professional society meeting I saw a nurse raise a continuing education guideline booklet that was produced by a genetic test lab and say this is all you need to start doing genetic testing in your office. That booklet only spoke about the test that that lab produced.

Unfortunately, we know that the healthcare community in some ways is only getting information from one area.

For our community to improve things, as you are determining where to go from here, I think it is really important that there be at least one government agency that has oversight and jurisdiction over genetic tests even from CLIA-approved labs and has

oversight as to how they are marketed to consumers and to physicians. Currently, at least to my understanding, there really isn't oversight and these companies can say pretty much whatever they want. They can be the single source of information for physicians and consumers.

Consumers need to know and be given access to trained experts in genetics. I know there is an argument that there aren't enough genetics experts. Part of it, though, is it is really hard to argue that a 23-year-old woman without cancer had such an emergency for having genetic testing that she had to have it in her doctor's office as opposed to being referred to one of the many good genetic clinics in the City of Chicago. We are seeing this in big cities, where there isn't a long wait and where there is no immediacy. So I don't know that that argument always holds water.

I think consumers don't know that they are being denied standard of care or even that there are experts in genetics. I think that is part of it. I think people have a right to know that, especially if

they are getting below-standard of care genetic services.

Laboratories need to be held accountable for their marketing materials for consumers and for physicians. I don't think it is enough to just scrutinize what laboratories are saying to the consumer. We also need to be looking at what they are saying to physicians and what they are telling physicians that they can and cannot do.

I certainly am not qualified to say who can and cannot do genetic counseling, but I don't think that the laboratory should be doing it, either. I think they are setting the bar very low.

We need an agency to track adverse events. Currently, because there is no FDA labeling for some of these tests because they are coming from CLIA-approved labs, there really is no off-label use of the test and there really is no way to say what is and is not an adverse effect and to be able to track it.

I do believe that it should not be up to the test developers to govern themselves or determine the appropriate amount of information, nor to designate

the minimal competency for conveying this information.

Thank you for your time and attention.

DR. TEUTSCH: Thank you. Thank you for putting a face to those issues. We did write a report on oversight, but I don't think we addressed this issue very completely. So this is important.

Why don't we take a couple of comments. Jim.

DR. EVANS: I just wanted to add my thanks to you for coming. I had not met Sue before but I have been very familiar, as somebody who takes care of patients, with genetic predisposition to breast and ovarian cancer. I have been very familiar with your website. Your organization is of huge use to patients.

It is very useful to get your impressions that these types of things are occurring. It has been my anecdotal experience, but that is just anecdotal experience. I would just ask, if you can try to keep track in a systematic way of these things, that will be very helpful.

I would also just echo your plea that there

be sources of information for patients. I think it is something we should keep in mind. We should have sources of information for patients that are apart from the commercially driven sources by some of these testing laboratories. We all have conflicts of interest, but patients need access to differently conflicted types of information. Thanks.

DR. FRIEDMAN: If I can just make one comment on that. It is my understanding that the New York State Department of Health, when Myriad did their direct-to-consumer marketing campaign in New York, developed posters that went in the primary care and OB/gynecologists' offices that did tell people about what standard of care was with regard to genetics.

The other thing that I would say is, FORCE is happy to be a source of information. Really, it is an honor that you speak highly of our organization. Funding is a big issue for us on an ongoing basis. I know you were talking about funding before. It is very hard for us to meet the gaps and meet those needs. We do need assistance with that. So, thank you.

DR. TEUTSCH: Great. Thank you very much. That is very helpful.

The second speaker is Amy Miller. Dr. Miller is the public policy director for the Personalized Medicine Coalition. Welcome. We look forward to what you have to say.

Comments by Amy Miller, Ph.D.

Personalized Medicine Coalition

DR. A. MILLER: Thank you. My name is Amy Miller. I am the public policy director of the Personalized Medicine Coalition, an organization that represents all the stakeholder groups within the framework of personalized medicine. I have spoken to this group before on a number of occasions about your work. Today I am speaking about consumer genomics and our work in that arena.

It is unavoidable to recognize that consumer genomics has received more attention than any other one product or sector or aspect of personalized medicine. Part of the PMC's charge is to educate consumers and doctors on personalized medicine.

To that end and based on some federal

conversations that have taken place over 2008, PMC has taken three different tacts to address consumer genomics. One is, we organized the leading consumer genomics companies to come together around standards of operation in their field. The companies that have joined PMC in this effort in particular are 23andME, DECODE, and Navigenics.

The aspects of standards of practice they have agreed on are scientific, and they are going to be presenting that work at a CDC conference on consumer genomics later in this month.

They have agreed on a number of scientific standards. Where they haven't agreed, they have agreed to be transparent. They have put together a brief document on that work and will be sending that out in advance of the CDC conference.

During the CDC conference, their scientific teams will be available to answer questions from the field.

The second is a consumer guide. Part of the PMC's work is educational. We have worked on a consumer guide on Warfarin dosing, for example. We

think that to inform the consumer guide we need to hear from consumers. So we are going to have a roundtable where we are going to have consumers and healthcare providers that work with these particular consumer groups and talk about what the standards are in this field and what consumers want from these products.

As has been mentioned, they are available. They are being used and they are being purchased. So we need to know what consumers find useful and risky about these tests. We are looking to develop a very balanced document that addresses some of the scientific issues and some of the concerns about these products and then what they can be used for.

Those are the three efforts that PMC is doing in consumer genomics. I would be happy to keep this group informed of that work and in any way assist the SACGHS in their work in this area. Thank you.

DR. TEUTSCH: Thank you, Amy. We appreciate the work that you are doing to try and get us to a good set of standards.

Any comments or questions for Amy?

[No response.]

DR. TEUTSCH: Good. Thank you very much.

Are there any others who desire to make public comments?

[No response.]

DR. TEUTSCH: If not, we will return to our primary task today, which is to review the priorities.

We were in the midst of a discussion on utility. I will turn it back over to Paul. I know that there were a couple questions, I think from Jim and Andrea.

Discussion of Proposed Priority Issue Areas

DR. FERREIRA-GONZALEZ: I think we were discussing research dollars for the gathering of this evidence. I want to bring that issue back because I think it is a very, very important issue.

As we continue to move forward in developing the infrastructure for the evidence development, we are going to continue to find out that there are gaps in that knowledge. As there are gaps that knowledge, the research will have to be done, but there has to be funding. Laboratories will not be able to perform

some of these studies or will not have the power to do collective, multi-site studies to really gather the information that will be needed.

So I think a focus of this group has to also be trying to identify or trying to recommend sources of funding for that research.

DR. EVANS: Alan addressed my issue.

DR. TEUTSCH: One of the things I would like to throw into this mix in this discussion is that the wide clinical utility is of particular interest to the future of healthcare issues. We have touched on it. So many issues that we are facing in personalized health care don't fit into the old paradigm of the clinical epidemiology or RCTH. If you go into personalized health care you are talking about smaller groups and more tailoring, and large clinical trials and other kinds of things are going to be very difficult to do.

We have been dealing all along with rare clinical disorders for which, clearly, that kind of work is never going to happen, all the way up to dealing with very common diseases with complicated

genomic profiles. Getting to an understanding of the value in all of this is going to be a whole lot more complicated than many of the things that the traditional clinical EPI community has been doing in the area of clinical utility in terms of the nature of the studies, evidentiary standards, and so forth.

My feeling is, if we are going to get there, not only is it going to be relevant to the reimbursement issues that we talked about but it is going to be very critical to how this will ever fit into health reform in a way that we can be assured is actually going to deliver real value. It is going to be tough.

DR. EVANS: I agree with you. I think it is also important to keep in mind that some of the rules haven't changed, even though it is a new landscape. I think there is tremendous, understandable impatience with the desire to translate and get things to the bedside. That is perfectly understandable.

But I think that one of the reasons the focus is on genetics in this context is because we have had an unprecedented burgeoning of basic science

knowledge in genetics, more than in any other field, arguably, for quite some time. So the gap is simply more apparent in genetics.

I agree with you there are going to have to be novel ways of figuring out when has evidence been sufficiently met, but in the end it is a slow process. Impatience can cause problems with it, too.

DR. WILLIAMS: I think, though, that one of the things that could come out of this is that we could improve efficiency if the people that were in the earlier stages of research were given a view of ultimately what evidence is going to be needed to move things into the clinical arena. That could potentially influence how research questions are asked earlier in the translation pathway. Then, when the movement comes, we are not dealing with what we are frequently encountering when we look at EGAPP or other reports, which is a lot of gaps in evidence and key aspects that just are not available.

It seems like communication across the continuum of research might more efficiently more allow us to have the answers to those questions at the

appropriate time. That really wouldn't add cost to the infrastructure, at least to a significant degree. It would certainly be less costly than going back and redoing the study to answer a key piece of evidence that was not addressed.

DR. FITZGERALD: Actually, I think that is one of our recommendations in the Pharmacogenomics report. You are right along that line.

DR. WISE: Other comments or suggestions specifically related to the clinical utility, Cluster No. 2?

[No response.]

DR. WISE: Great. Could we move on to Joseph's cluster? Scheduling issues require that we move to Cluster No. 6. Joseph?

DR. TELFAIR: Yes. Thank you very much. I know it is a skip-over for a lot of the other work. Just to cut to the chase, we in public health believe very strongly that the Committee in its deliberations should consider what is a balanced view. Much of the discussion so far this morning really has been on the issue of translation. But I think what was just said

by Dr. Williams a second ago is actually where our stance would be. We need to really look at the very beginning at how you pull this information in.

The paradigm is really the idea of assessment. Many times the program is up and running before the assessment actually is even considered or takes place. So you have to go back and relook at things.

I would just bring to your attention to two parts of the short-term actions. The second paragraph is the idea of a systems review. The question really becomes where do the different elements fit together and then where are the commonalities and the differences. But a systems review really means looking at the different areas in which program function and the issues that we are looking at fall together. Then, where are the commonalities and what can be done.

The bottom line is really the last part of the sentence, which is both the differences but the assurance of effectiveness, accessibility, and quality of services. Then the question is, how do you move

from the basic science to this area. One way is to look at what is being done and what are the common areas that are being looked at.

The other part I would really push is that you do it in a systematic way. If you do a systems review or a review of what agencies are actually doing in order to meet this goal, then you move into how can you work together to effect that to meet the other needs that are consistent with what the push for this actual Committee is, which is dealing with these other issues related to application, risk assessment, et cetera.

I would leave it at that for conversation. I think that cuts pretty much to the chase on that.

The other element that I would add would be also the social, ecological, and environmental fit, which is looking at the interaction of genes, environment, and health applications.

DR. WISE: Marc.

DR. WILLIAMS: I would just strongly endorse what Joseph said about the systems review. As we have heard presentations at different meetings -- and the

most recent example was yesterday afternoon -- we hear different groups that come to us and talk about what they are doing and then we realize, wait a second, there are at least three different groups that are looking at biospecimens that could be used for a variety of purposes in terms of standardization of proficiency testing, et cetera. That is an inefficient way to do it because we are essentially doing some degree of duplication of effort.

Inasmuch as we can assess what everybody is doing and look for areas of commonality and use that to build consistency, we can get more bang for our buck. We don't have to be spending money on duplication. I think that is a great idea.

DR. WISE: Other comments or questions?

[No response.]

DR. WISE: Thank you, Joseph. That was great.

DR. TELFAIR: Like I said, when it's clear, it's clear.

[Laughter.]

DR. WISE: We are convinced.

DR. TELFAIR: Thank you.

DR. WISE: We will then move to Cluster No. 3, Barbara's genetics education and training.

DR. McGRATH: Actually, before we discuss it, I just wanted to say that I have been listening this morning and, obviously, yesterday as well. Our first action item was a small one, to talk to FDA about devices and educational standards. After listening to Cluster No. 7, I think it really fits.

I don't want to punt, but it is just a suggestion. It's a thought. Perhaps we could work together on that one; let's put it that way. That was one thought I had after listening to your presentation. Anyway, that is one thought.

The other actions really are what we are doing on the Committee, which is heavy data gathering and, more importantly, synthesizing the data from a lot of different places about existing programs and looking to the future. I think that is a logical way to go. I would welcome any suggestions for other ways.

But what I'm thinking about is the whole

notion of looking to the future more. Part of it is Julio's comment, and I think we are trying to do that, to avoid the old way of looking at it and to think about what are the needs in the future of the new generations of people on Facebook. We are all dealing with information differently, so we need to get out of that old paradigm.

The other one is that what is frustrating to me is that there are lots of specialty agencies who are involved in biomedical education and are in the traditional silos of medicine, genetics, internal medicine, OB/GYN, that way we have been dealing with biomedical care in our healthcare system since it was organized. Yet, on the other hand, we have this dialogue going around with these terms like "systems medicine" that Leroy Hood has been using. That is on the horizon. It is not our landscape yet, but it is on the horizon.

I don't know if that is a direction to go in, but if we really head off in that direction, that really changes the way those silos are set up about education and training. It might be worth thinking a

little bit about looking to the future of that as well, while not ignoring what is happening now because we have to deal with the landscape. That is something I have been listening to the last couple days.

I think those are my only new thoughts on it.

DR. DREYFUSS: I was very struck yesterday by various comments that patents were important for education. I should say shocked. Just as we said yesterday that utility is its own issue and quality is its own issue and we shouldn't be mixing patents with that, it seems to me that we shouldn't be mixing patents with education, either. As the speaker just pointed out, the kind of education that is provided by people who have a very strong interest in the sale of whatever it is they are selling is not going to be really good education.

So I would really endorse doing more on this particular topic so that the pressure isn't on either patentees -- and you do have this in what I take to be this cluster -- or industry groups alone but also includes patient advocates and healthcare providers.

I think on numerous occasions we have found that relying on patentees for educating people about what their patented products are is not a recipe for a good way of utilizing public resources.

DR. WISE: Mara.

MS. ASPINALL: A few comments. First, on the cluster, I think this is one of the most important ones and absolutely essential. I would agree, though, that the first short-term action is not consistent with the rest of the in-depth report or the policy questions, partly because it doesn't fit here. I'm not sure it fits into No. 6, but I think it get into what is a very current issue around the FDA requirements and laboratory tests. There are a number of associations and groups working on that.

There are questions right now about how those tests will be or will not be touched by the FDA. So I would also endorse Barbara's comment that the short-term action in No. 1 about working with the FDA officials does not make sense.

However, as Rochelle said, to broaden No. 2 to understand the regulation today, I would broaden it

to say it is not just to encourage the development of voluntary standards but to understand what the standards are today, how they work, and therefore what recommendations we might have in the future.

I would probably take issue with what Rochelle said. As the speaker said, somebody who owns the patent or has an interest in it may produce a great piece of material. I don't think we can assume it is necessarily a bad piece of material. But in and of itself there may be a perceived or real bias that says it would be better coming from a neutral organization.

But there are high-quality materials coming out of individual companies and out of universities that hold the patents, and there are poor materials coming out of universities and companies holding the patents or not holding the patents or involved in the commerce. What we are talking about here, I think, is the broader scope, which is ensuring a regular process so we don't have to depend on the individual involved in the commerce to ensure that we have the right materials for the purveyors of health care, whether

they be physicians, genetic educators, nurses, or others.

In summary, eliminate No. 1, expand No. 2 under short-term action. I think the in-depth report, though, is very consistent with the policy issues you raised.

DR. WISE: Joseph.

DR. TELFAIR: I would agree with the last statement on the in-depth report and would add that it is important to consider that education is not only multidisciplinary but is also a multidirectional process that has to be comprehensive, particularly in this arena such that you are looking at the general public, specific consumers, and professionals of all types.

The second thing I would say is that because it is that kind of process it is going to be important to assure that there is clear understanding and there a means of both monitoring and evaluating the specific outcomes of the education process itself, given that it is multidirectional. Those are critical pieces. I would just add that to this action step, if possible.

Thank you.

DR. WISE: Marc.

DR. WILLIAMS: I would just request that we make something explicit that is implicit. That relates to discussions both from Mara and from Steve about the way medicine and delivery of medicine are going to change. We are going to be moving toward very complex information sets that are going to have to be combined and that are going to require informatics tools like clinical decision support. Some of that, I think, is going to be offloaded into personal health records and algorithms that run on those that the individual can control.

There has to be education embedded around those types of tools so that if an individual says, wait, I'm getting this message, what is this based on, that they can rapidly find that information within the context of the clinical decision. The idea of "just in time" point of care education within electronic health records and personal health records is going to be critical.

The doubling time of medical knowledge has

changed from when I graduated medical school. Then it was about 30 years, so I only had to relearn everything I had learned once in my career. It is now seven years. That means that somebody graduating from medical school now is going to have to, essentially, relearn everything four times in their practice career, and actually, depending on how their 401(k) looks, maybe six times.

[Laughter.]

DR. WILLIAMS: But clearly, traditional educational approaches, while important, are not going to be sufficient to do this. We really have to make sure that we are responding to that future.

DR. McGRATH: In response to that, I have more of a question to the group. Even if you just limit yourself to the "just in time" primary care providers, it is a number of different silos, if you will, or specialty groups. In other reports -- I'm thinking of Oversight in particular -- we have suggested that when we felt like it was dysfunctional because there was little communication one of our strongest recommendations was coordination across

groups.

I don't know if that might be something that this task force should put as one of its things to consider. Is there a recommendation that there should be better coordination across everyone using the electronic health record, which is many groups. That is a question.

DR. WILLIAMS: Just a couple of responses to that. First of all, I think that there have been efforts to try and create those types of groups. I think the National Coalition for Health Professional Education in Genetics, NCHPEG, is a good example of that. They have really tried to do some cross-disciplinary educational efforts.

I think that there is clearly a movement within the medical informatics community to say if we are going to have guidelines and the guidelines are going to be embedded within electronic health records, then there have to be some standards relating to computability and how the information is obtained. There is actually talk about establishing a national electronic clinical decision support repository, much

like Guidelines.Gov.

If that actually moves forward, that would be an important partnership to link onto to say what are the educational things that you need to have that would associate with these vetted clinical decision support issues. Those are the types of partnerships that I think would be important.

I think we need to also recognize that, assuming that the incoming administration continues what has been a strong push of the current administration, which is to have a fully interoperable electronic health record in this country by 2014, we have a very good window of opportunity where there is going to be a lot of energy and investment to make that happen.

DR. WISE: Steve.

DR. TEUTSCH: I heard a question here about the information that comes from industry and how that gets done. We didn't address that all that completely in the Oversight report, as I recall. This Committee worked with FTC before, as well as with FDA, on labeling and promotional information. I wonder if

that is something that we want to have at least as a short-term thing to talk about in light of the comments we heard. Clearly, there is a lot of promotion that has escaped the FDA labeling system.

DR. DAYNARD: I just have one comment. We heard a speaker and the Committee members talk about promotion that may need additional oversight and reigning in. The problem is as follows. When the FTC challenges advertising, it is for one of two reasons. One is the claim is just blatantly false and is never going to be true, like losing 30 pounds in 30 days or something. Unless you have the right gene, I don't know.

The second would be because they lack substantiation. Substantiation is what the scientific community says the evidentiary standard is, RCTs for example. So when we are looking at a promotion that says you can link a genotype to a healthy living recommendation and that that is going to help a specific genotype person, the question is, what is the evidentiary standard to show that that is true. Is it case control association studies, which is what is

happening now? Is it an RCT that may take a long time and a few million bucks?

And, is the FTC the right agency to say what these evidentiary standards should be. If we challenge a claim and say, you didn't meet the evidentiary standard, they will say, what is that evidentiary standard that you are saying we didn't meet? Gee, I don't know, because the community doesn't know.

This is going to be a continuing issue. I'm not sure, frankly, where the FTC is going to come out on it. But the question still stands to you all: What is the evidentiary standard? It is another linkage that has to be laid out.

DR. WISE: Peter.

MR. KIRCHNER: On your short-terms actions, or perhaps longer-term actions, you didn't include the establishment of some type of a Web-based information area that would list what evidence is accumulated for linking associations. This, of course, could be indexed in two ways, one by genetic findings and specific genetic markers, and the opposite direction

would be by specific disorders.

I think this could be maintained by a group of professional editors that would assess the reliability of published data because you only want to update it with things that are very secure in terms of their contents.

I would think something like that would be extremely valuable both to physicians who, like myself, do not have enough expertise in this area, but also of course to the public. There would be some indication then as to how reliable some information has become or what is missing still.

DR. McGRATH: We have talked in the group about that. There is a number of them out there. Part of the data gathering is to pull them together to look at them because they are like genomic Wikipedias out there. It is like the Wild West. I think that would be a good contribution.

MR. KIRCHNER: It is a major task of how to pull that data together, but I think it would be of great utility to everybody in health care and also to the public.

DR. WISE: Yes, please, Robinsue.

DR. FROHBOESE: Robinsue Frohboese from the Office for Civil Rights. I just have a quick question. I know that in this Committee's 2004 resolution that workforce diversity and cultural competence were critical issues. I think that in the summary of the clusters workforce diversity was in parentheses. It wasn't highlighted in our background materials. I wondered whether in the report that is in progress culture competence and workforce diversity are going to be key issues that are addressed.

DR. McGRATH: Yes. It is a bullet, not a parentheses. It has risen up to the top.

DR. WISE: Important point. Other comments or suggestions?

[No response.]

DR. WISE: Kevin, you are up.

DR. FITZGERALD: I'm just going to punt to Sylvia. Actually, I think this issue is not one that anybody would disagree with. Everybody is certainly interested in protecting privacy and confidentiality, and for continued application of informed consent.

I think the issue is a little different from some of the others, in the fact that it sprung out of the pursuit of personalized health care and the application of the various advances in technology that we want to do, which brought about this scenario that maybe didn't have to happen but is certainly, I think, happening.

The whole idea of the application of these technologies and the pulling together of all this information in some kind of accessible form in large databases with interoperable healthcare records and all that, brings up this issue.

One of the things, I guess, that we really need to wrestle with is what is the role of SACGHS in addressing this. I'm not sure that this is the place where it should happen. I think there are arguments for and against. Maybe that is what we could explore a little bit.

I will just put it in some context here. It is not that, again, this is an issue that hasn't been looked at or even experienced by other places. I think the experience in Iceland over the past several

years would be instructive.

I think in the United States we have represented here at the table two groups that are in the midst, probably, of addressing some of these issues. I know Ellen mentioned the VA being involved. Also, DOD. Daniel is here. The idea is that we have two large groups managing numerous people's healthcare records. How is that going to be integrated and what will the issues be coming out of that.

We have a third issue that is moving very rapidly on the horizon, and that is the newborn screening issue. What are we going to do with that information. If one wants to be logistically efficient, that should go immediately into some kind of national database that we could start now and use for longitudinal study.

Again, I think the issues are with us. How we wrestle with it and what our role is, is the question. I think we would have to take into consideration Department of Justice issues and Commerce issues. There would be other things outside of HHS, like civil rights.

Again, it is something, I think, that could lead to some rich discussion for this group as part of a larger sphere. That, I think, is the question that is really before us. What role do we play. Are we some kind of a not objective but, in a sense, less invested third party that can provide some kind of distance. I don't know. I think that is what we have to wrestle with.

DR. EVANS: As you think about these issues, I would just encourage everybody to read the short article by Patrick Taylor that is in our briefing books about some of the nuances of consent. It touches on privacy issues. It is good to heed some of these things as we go forward.

DR. FITZGERALD: Just one other issue on that, just to give you another sense of how things are moving. If you look in our materials on page 24 under Tab 5, just go back to the SACGT committee. We wrote, "The major distinction between consent to research and consent to treatment is that, in the first, there should be no presumed benefit and, in the second, there is no reason to proceed without a presumption of

benefit."

My sense now is, with personalized health care, the idea is that it is all supposed to provide a benefit. So research, clinical, it doesn't matter. It is all the same thing now. So, how did that indeed change the landscape for us.

DR. WISE: Marc.

DR. WILLIAMS: As you were setting up the issue and were listing some of the stakeholders, it seemed to me that maybe one of the things that could happen in the short term would be to have one of our educational sessions focus on different stakeholders' approaches to privacy. Certainly, the people that you have already listed would be very good, but I'm thinking now of two private groups that have gone into this in a relatively large way.

One would be the Marshfield Personalized Healthcare Coalition and their approach to consenting individuals and reconsenting and recontacting. Then, the Vanderbilt program for residual blood specimens and use for research.

Then, echoing what Jim had said, [we could

talk to] someone like that author or Zach Kohane, who has also written on differences in terms of how we can approach the consenting process.

I think that would be a fascinating session that might really help. The other group, by the way, would probably be representation from the direct-to-consumer folks, who would also have a perspective on consenting and privacy.

I think it would be a very interesting and very rich session that might well provide important information that would set the tone for the report.

DR. FITZGERALD: That is an excellent idea. It was one of the things I think we bounced around. Obviously, we are not going to recommend it without the Committee's support, but that is a great idea, if people want to do that. That could be a first step.

When is the HRSA meeting? February. Do you know the dates? As I said, there are a lot of people looking at this issue.

One of the things we would have to do is check to make sure we are not reinventing the wheel on this.

DR. CAROME: Mike Carome from OHRP. There is a higher-order issue that I think is implied in the cluster discussion here but is not explicit, and that is when does research involving genetic data and associated clinical information rise to the level of being research on human subjects. If it doesn't involve human subjects, then you don't have to get informed consent, at least under the regulations. So some of this would be moot.

A lot of this turns on part of the definition of human subjects, which has to do with obtaining individually identifiable private information. What does "individually identifiable" mean.

There is certainly a great deal of research involving stored specimens, stored DNA, and stored clinical information that is done in a way in which it is coded or all identifiers are deleted and not replaced with a code. Under guidance from our office, we have opined that that doesn't involve human subjects. So the consent discussion is cut short.

I think one of the questions implied by

Policy Question No. 6 is whether with evolving technologies in genetics and information technologies, are things that we considered not identifiable now identifiable and therefore we need to change the paradigm somewhat. So that is something it might be important to explicitly identify in the policy discussion here.

DR. FITZGERALD: Sure. Actually, one of the ways we could look at this is either this move toward personalized health care is going to put you out of work or you are going to become as big as DOD, one way or the other.

DR. WISE: Other comments?

DR. FITZGERALD: Is there a sense among the group that, although HRSA is doing an educational session, we need one for ourselves? Would that be a good short-term first step?

DR. FERREIRA-GONZALEZ: Do we have any representation at that conference? Is anybody from our Committee going?

MS. CARR: Actually, Joseph's term on SACGHS is ending, as is Kevin's. At the next meeting they

will be coming back and we will be saying goodbye to them when our new members are on board. Dr. McGrath, at least we think, is going to be the new liaison to that committee.

MS. AU: I will be at the meeting because I'm PI of one of the regional collaboratives but not for the Committee.

DR. FERREIRA-GONZALEZ: That might be a way we can hear back in our Committee as to what are the findings and then make a decision if we need to continue or gather more information.

DR. FITZGERALD: We could put that under "monitor." Monitor the meeting and then decide based on what we find from that. That is great.

DR. WISE: Other comments?

[No response.]

DR. WISE: Moving on to Cluster No. 5, Sylvia. Thank you, Kevin.

MS. AU: I think the challenge with this cluster is that there is such a broad range of policy questions. Some may be easier to answer, like whether the Oversight report covers direct-to-consumer genomic

testing. It also overlaps with every other cluster. Basically, I can punt to everybody else and we can collapse this cluster into nothing.

I think that, looking at our possible action steps, it basically comes down to where does this Committee want to weigh in on the curve. If you monitor and then you comment, then you weigh in lower on the curve. If we actually are proactively going to do a detailed report, like we often do, then we would probably be closer to the beginning of the curve.

There is a lot of interest on the Committee in this subject. I just don't know where the Committee thinks that it can do the most benefit in this area.

DR. EVANS: I think there is a lot of interest and a lot of expertise on this Committee in this type of thing. My own personal feeling would be that monitoring would probably be too passive of an activity.

MS. AU: Darn, Jim.

DR. EVANS: And I think you should head it up.

DR. WISE: Joseph.

DR. TELFAIR: I agree that monitoring may be light but evaluation of outcomes is not. I would make that argument. There is a difference between the monitoring process and the evaluation process. So the short-term action step may be a long-term action step because this is something that should be ongoing. I would argue for that.

But it should be informed. It should be informed. I would move back to a recommendation that I made related to public health, which is starting with a review of what actually is going on and then developing the assessment based on that, with some clear, agreed-upon outcomes that need to be looked at.

That is important, and that is doable. The recommendations from that would be doable.

MS. AU: So, maybe doing an assessment and seeing if there are key policy issues that we should address and then which things we need to punt to others to include in their clusters.

DR. TELFAIR: Yes, I would agree with that.

DR. TEUTSCH: Can you comment on that issue?

We did spend a lot of time at our last meeting with an assessment of the landscape and whether we need to go back and actually do that or can we just build on that.

DR. TELFAIR: Is that question to me?

DR. TEUTSCH: Either of you.

DR. TELFAIR: A systems review actually takes into account work that has been done and then uses information that is actually missing as well. You would actually begin to look at what is the existing evidence across the groups. If that is adequate based on whatever group is using that, then you would go from there.

But the key here is to develop accessible outcomes that would work, particularly with this challenge, which is moving in that direction. We would have to be able to make sure that the decisions that were made and the evidence that is there is actually very focused and targeted and will allow you to look at the outcomes you have agreed upon. That is what I'm talking about. It is a combination of those things.

MS. AU: Because this is such a moving target, there have definitely been huge updates in the last six months.

DR. EVANS: I would just make a plea to make sure we do this in a timely fashion. We could be reassessing forever. This is a rapidly moving field. I would urge us to move on it. If we are not going to do that, then we should just monitor.

DR. WISE: I have Peter and then Kevin.

MR. KIRCHNER: I see a strong interaction between what you are trying to address here and the educational component, which is so terribly important. I would think that in some ways effective action to counter misinformation that might come from promotion of such tests directly to the public can be addressed with the strong educational approach that has been described by Barbara. I would think that you would want to see some kind of strong interaction.

DR. FITZGERALD: I'm just wondering if this could be pursued as a consequence of the Oversight report in the sense of saying we touched on it in the Oversight report. We certainly mentioned how

important an area it was. Then we could just say, building on that, now we are going to take this little piece of the Oversight report and expand it within that context, over that same framework, so we don't get into all the morass that we might. But using that as a boundary, we would try and expand upon that from there.

DR. McGRATH: That would be one approach. The other one would be to make this a stand-alone topic that SACGHS works on. When the charter was written or when the original missions were written, it wasn't such an urgent or current issue. It has, over the years, really grown. Whenever we talk about it, there is a lot of new information and a lot of emotion. So maybe that is one of those that should get its own separate category.

I don't know what other agency in the government is going to be looking at this, so maybe this is something we should take on as in the spirit of our original mission, even though it wasn't spelled out like that. It does seem to fit with our overall mission.

MS. AU: I think one of the important things will be, if we want to move quickly on this, what are the options. Our detailed reports take a long time, other than the Oversight report. But we don't want to repeat that, unless Andrea wants to chair it again.

I think that that is one of the decisions we have to make.

DR. WISE: Can I just ask why we think this is so important? For somebody coming from outside the field, this seems like a gimmick. It doesn't seem crucial. If we were to move it forward as a high priority, I think we would need to frame it in a way that engages a broader challenge to the healthcare system and to public awareness about the importance, the relevance, and the implications of genetic insights in a way that doesn't make it look petty for a committee to take on in some meaningful way.

I would just take a step back and ask why do we think this is such a crucial thing.

DR. WILLIAMS: My response to that would be to look at analogous movements of consumer-driven care. The two that I would highlight would be

complementary and alternative medicine and nutriceuticals.

I think the complementary and alternative medicine story is a very interesting one because there was obviously a huge interest in this. It ultimately led to the formation of an institute within the NIH specifically devoted to looking at the science and evidence behind complementary and alternative medicine, essentially saying we have empiric observations that there may be something here. Should we not then take a look at this from the perspective of science.

It really addresses the issue that Matthew brought up, which is what is the evidentiary standard to say that this is good or this is not good.

That really hits home for me. In some ways, we are the emperor with no clothes. We are saying, you need to have some evidence, but if we really honestly look at the evidence that we are all developing around the things that we do on a day-to-day basis, it is pretty thin. We don't have a lot to hold up, either. We may not be naked, but we are in a

skimpy negligee, I think, at best.

[Laughter.]

DR. WILLIAMS: But we know who would head it, and I will leave it at that.

At any rate, I think that there really is value there. I think also that the nutriceutical argument is if you look at the amount of consumer spending relating to things that are not necessarily well understood and where there clearly have been examples of very significant harm that have resulted to the public, these are the types of examples that really, to me, say this is something that we do need to try and get a handle on.

I think we do need to come at it from a fair perspective, which is to say there is something there. We know that this is important. We know that this means something to people. We know that maybe this is the lever that we need to get people to change behaviors, which ultimately will make them healthier. How can we pull this together.

I think it is important from that perspective, and I would strongly endorse being

proactive and being relatively formal about engaging on this.

DR. EVANS: I'm just really intrigued by you bringing that up, Paul. I think that many of us who are immersed in genetics think, oh, this must be important. Given the media attention these types of things have gotten, we think, wow, it is the next big thing.

I was recently told by somebody who should know these things that, frankly, there has been very little uptake of this, aside from the splashy articles in the press, et cetera. I was very heartened by that because I think it tells us the public is more savvy than we sometimes give them credit for.

My feeling is, however, that we might be right after all, that this might be something that catches on, and there are real concerns with it. That is why I like the idea of a short-term action that, in a relatively expeditious manner, comes up with a checklist or something useful to people, something that can be promulgated in an efficient manner, that is easy to use, that brings some light to this field.

But in the best of possible worlds, perhaps that wouldn't be needed because people don't really buy into the hype.

DR. FITZGERALD: I also think that this is an area that provides a more extensive perspective beyond its own current scope. That is in the ongoing debate whether or not health care is just another consumer good to be, of course, driven by market forces and consumer desire. Or, is it a societal obligation that is to be delivered by a certified professional community. Those are radically different concepts.

DR. LICINIO: Automobiles now have become a societal obligation.

DR. FITZGERALD: I think this is one of those things that, the way it exists currently, is of such a magnitude it does raise that conceptual issue. On that level, too, I think it is worth delving into.

DR. WISE: Barbara.

DR. McGRATH: One of the reasons I think it is important is that it might be standing in for other things. I think the uptake is low, not that many

people find genetics all that interesting.

But I do think it may be a way that consumers and all of us start learning about health care, and behavior change happens there. So it is a window through which to look at other things.

I think the CAM example is exactly right. That moved the science forward. But I go back even further to the HIV and AIDS activism of the '80s that moved that science forward. One thing that that did was it highlighted who the science wasn't working for: populations and subgroups whose needs were not being met. I think if we were to highlight some of this it might shine a light on the groups that genetic services and the genetic technologies are not particularly helping. It would be another way to look into the whole issue of disparities, which I think are really critical.

DR. TEUTSCH: On the same issue, I think that if you look at these emergent technologies, the best time to deal with them and help shape them is early. Once they are out there and in widespread use, it is really hard to influence them because they have

a life of their own.

So in some sense, just on a timing basis it is clearly topical. It is an opportunity for us to at least have some influence over the development, which is probably timely.

DR. FROHBOESE: I actually had a question for Sylvia. Of course we see the health disparities theme throughout all of the clusters. I just wanted to get a little bit more information about this thought of doing a report on how direct-to-consumer marketing may be impacting health disparities. How would you get at that issue and what is the tie that you see there?

MS. AU: I think that that was just one of the policy questions that we came up with. Of course, the amazing task force that would be formed would come up with the amazing way to collect this data.

I think it is really difficult because the uptake is low already. People have to pay out of pocket for these tests. So it would be difficult to measure any significant health disparity at this time.

But I think it definitely is a point that we

can extrapolate from other instances where society has access to pay-out-of-pocket kinds of medical care. This would be a similar down-the-road kind of thing.

DR. WISE: I think as we have talked about this in the past, particularly in relation to minority health and reducing disparities, one is differential access and differential provision of these services. To the extent that they are beneficial, they would then enter the conversation about disparity creation.

But the other is how widespread consumer engagement with genetic services will alter public discourse about questions of equity in society and public programs, and larger questions of social inequalities as well as health inequalities.

So beyond access, I do think that there is the potential for altering public discourse around a whole variety of social issues because of direct consumer community engagement with genetic services.

Other comments or questions about this? Mara.

MS. ASPINALL: I had a comment on the timing. We are going to have to come back at the end

of this process to prioritize everything. I'm assuming that we will not have the resources to pursue at the same time all seven of the clusters.

In the context of this, given the conversation around the short-term nature, I very much agree with what Steve and Jim said. If we were to attach a value to this, we should do it sooner rather than later. As we heard in the public comments, there are other people focusing on this. The timing is critical.

So I might suggest that we maybe lower-prioritize the in-depth report but higher-prioritize the brief report to be able to make a statement on this issue.

I'm worried about this. We have heard a lot of great things, at least on the first few, and maybe some more, but ultimately the toughest decision we are going to have today is to say which ones do we want to do ahead of the others. Putting this one in a slightly different category to be a brief report of a short-term nature I think may help with the ultimate decision-making.

DR. WISE: That is very helpful. Other comments?

[No response.]

DR. WISE: Thank you, Sylvia.

DR. BILLINGS: I just have one comment that I would like to make. Mara has made an overarching comment, so I thought I might interject at this point.

It might be also useful, either today or as we think about prioritizing, to look at whether there are gaps or areas very prominently which are not covered by our clusters which we do still think are part of the purview of the Committee.

One area might be the treatment of people with genetic disorders. That is, progress either in monogenic or polygenic treatments of the conditions under which people with genetic disorders receive health care in the United States.

That is not specifically addressed. It may come under a couple of the clusters, but that might be an area of obvious interest to this particular Committee.

The other area, in my view, is the

relationship between the work of this Committee and its topics and the research portfolio at the NIH, as well as the interface between the National Center for Human Genome Research and this Committee and what the interplay between topic areas is.

Those would be just two areas that might be gaps. We may decide that we are covering them adequately or don't need to cover them, but those are a couple of areas that strike me as things that it might be people would be interested in.

DR. WISE: Thank you, Paul. Any comments or responses?

DR. FITZGERALD: Paul, this is Kevin. Just for clarification, when you say people with genetic disorders I presume you mean clear mendelian kinds of disorders?

DR. BILLINGS: It is a changing paradigm. We just engaged in a discussion about consumers and their searching for what may not even be validated risk factors. I think we have an obligation as we invest more in genetic technology to ensure that the treatment of people with the older-style genetic

disorders continues to improve. I think that is an ethical obligation as well as a practical one. We ought to state it and make sure that we are collecting evidence of trends in that area; let's put it that way.

DR. FITZGERALD: Thank you.

DR. WISE: Other comments or suggestions?

[No response.]

DR. WISE: I think those two will be important, certainly, in the consideration of our next cluster. We can see how it might or might not relate to the future of healthcare systems.

Mara, do you want to take us through Cluster No. 7? Then we will have time for general discussion and begin a conversation about how we would prioritize these in just a flexible, general way.

MS. ASPINALL: We go back to the words that we talked about before, "preparedness" and "future." The first piece is getting together the folks on the health plans to be able to look at how they see the future. One of the things that we had done previous to this as part of the initiative was talk to

futurists more broadly. This is where this original initiative came about.

That, coupled with the idea that healthcare reform may very well be a hallmark of the next administration, gave us the opportunity to say we really need to be not just looking six to 12 months out but really five years out as to what the infrastructure needs are.

I think a piece of it is the chief medical officers of public and private institutions but also having the ability to scan the various agencies in HHS to understand what planning they have done so we are not recreating the wheel with others within the HHS environment and there look at the potential infrastructure needs in the future.

The objective would be to put together a report to be able to outline that future and the key steps that need to be taken in the short term that will help achieve the future that we see.

DR. WISE: Marc.

DR. WILLIAMS: Just a couple of things. I think from what you said, I'm seeing something

different in what is written. When I see health plans, I assume that that is an insurance. I think that it would be important to be inclusive and to have chief medical officers not only from payers but also from integrated health systems, hospitals, academic medical centers, et cetera. I think they are all going to have an input.

As much as I think we like to think that we docs are in charge of the future of health and how that is going, the reality is that it is really a partnership with administrative leaders. So we really need to have some innovative administrative leaders that are not CMOs but CEOs from those same organizations. They will have the business perspective in terms of where they think things are going.

I think it is a great idea. I just would vote to be more broadly inclusive.

MS. ASPINALL: I think that makes a lot of sense and that we should do it. What we meant by "plans" is the broadest definition but that it is not just the M.D.s and their key strategic thinkers and

those pieces. The emphasis, which may not be clear, is this is public and private as well, and to look to the chief strategy officers, whether they are called that or not, from the agencies, again to ensure that we are not recreating the wheel.

MS. AU: I think that we can build off some of the work that Deb Doyle in the State of Washington got funding for. She did bring together leaders of healthcare plans and third-party payers to discuss what they were doing currently about genetic reimbursement and then what they thought the future was. I believe the work was completed about two or three years ago, so the report should be out there somewhere.

MS. ASPINALL: That's great. I'm not familiar with it. That would be helpful.

I'm glad you brought up the University of Washington. During the break somebody also mentioned to me talking to the key healthcare providers in terms of the medical associations and the groups of hospital systems in and of themselves. This implies just the reimbursement piece, but we really need to be broader

on that. So, does the hospital of the future, in anticipation of genomic medicine, look very different in terms of its in-patient/out-patient mix, in terms of its information systems. So we would also include that.

DR. WISE: Other comments or suggestions?

DR. WILLIAMS: Are we going to go through other parts of this or is this the discussion piece?

MS. ASPINALL: We can keep going through any parts you would like to. What we haven't changed on the slides on the screen but I have changed on my slides is the initial earlier comments about clinical lab careers and broadening that, as we discussed earlier.

DR. WILLIAMS: I would like to second what Paul had put forward. If there was one thing that was really exciting about the American Society of Human Genetics meeting last month, it was just how close it looks that we will actually have some treatments for traditional untreatable diseases based on small molecules and taking advantage of axon skipping and other things of that nature, not to mention some of

the interesting work that is going on with RNAs.

I think we may in fact, in the future window of five to 10 years, have some extremely effective therapies for some of the traditionally untreatable genetic disorders. I agree with Paul; I think it would be a shame if we let that drop off the radar. I think it probably does fit within your cluster.

Whether we need to do anything at a high priority level right now other than just to monitor where things are I don't know, but I think it should be represented. I know there are at least a couple of them that are in phase two, and maybe even one in Duchenne in phase three, clinical trials?

MS. ASPINALL: Yes. But basically, what you are talking about is that with the advent of genomic medicine in a broader scope there are diseases that are today not fully addressed that may become long-term chronic diseases.

The example that I would use here, and I think it absolutely fits, is AIDS. As the AIDS community changed from a disease with a very finite life span to a long-term disease, we needed to change

the infrastructure, whether that meant hospitals, reimbursement, life insurance plans. Quite frankly, that shift happened quickly enough that many of the institutions were not ready to do that.

If you look at probably two or three scenarios about these kinds of inventions occurring and being successful, how do we indeed put together the infrastructure to be ready for that without undue cost.

DR. WISE: Joseph.

DR. TELFAIR: Just a point of clarification on that. When you are looking at the planning, do you anticipate as part of the discussion looking at both workforce development as well as education of the workforce? Once you make decisions about where it is going, then who is going to actually be there to do the work. That is where I'm going. Do you anticipate that as part of the thing?

MS. ASPINALL: We do talk about that, and we talk about it as one of the policy questions. I'm wary of recreating the work of the other clusters. We would take this at a much higher level as opposed to

getting to any specifics of this type of material or this type of education. Rather, we would more broadly talk about the type of healthcare providers.

For instance, one of the futurists really talked about the dramatic change -- if you believe in personalized health care and much more precision -- that there will be fewer physicians providing care and more non-physician care. That would be an example of the high level that we would look towards. If that is the case, how do we set out an education program for non-physicians.

We would not, I would anticipate in this cluster, get into the level of detail that says how would you educate them. That would be handled with other clusters. But we would look at that big piece to say what is the mix and how does it change.

DR. TELFAIR: I guess I brought it up because of the admonishment with which you started off your beginning statements with, which is the question of integration and priority setting.

I recognize and respect the fact that you were trying to stay away from that, but I also

recognize that we have to set some priorities. We have to look at how there is some integration across these clusters. That is what my question is. It seems that actually would be less efficient than looking at where integration might be. It shouldn't mean, to me, working with some of the other clusters. We are going to have to do that anyway.

MS. ASPINALL: Yes, yes. I think that is a good point.

DR. WISE: But it does seem like for this Committee to engage this issue, which of course touches every committee that exists related to HHS and beyond, we are making a special claim of relevance. In other words, healthcare reform cannot realistically move forward without engaging in a very purposeful way the explosion in genetic insight and capability.

The second is that this is an intensely anticipatory project for us to take on. In other words, this is really tilted forward and looking at very big-picture issues to ensure that the healthcare reform conversation is not only about changing CPT codes over the next six months, although that may be

very important. Particularly given the trajectory of genetics and genetic capabilities, healthcare reform must engage these issues in a very meaningful but also in a highly anticipatory way.

Am I hearing that correctly?

MS. ASPINALL: I think that is well said.

DR. WISE: I think, Joseph, you raised the fact that genetics in the service of reforming the healthcare delivery system could in fact embrace clinical utility. It certainly, as I mentioned, relates to reimbursement policy shifts and workforce.

So it clearly will have strong linkages to other clusters. I wouldn't call it a task force yet. It may be that in our priority setting we could suggest that if healthcare reform becomes a framing activity for us that it include other issues that would then not be seen as the highest initial priority but would be included in the anticipatory special claim arguments that the Committee would make in this area.

MS. ASPINALL: I think that that is right. I think the challenge in front of us in doing that is

keeping up with the potential progress in the real world outside of this room and ensuring that we remain relevant in a way that if healthcare reform moves quickly -- and I don't think we all have the answer to that now -- that we will have a seat at that table short-term. One of the priorities may be stating that to ensure that our interest in doing that is clear to the next administration.

DR. WISE: Other comments specifically on Cluster No. 7?

[No response.]

Determination of Priority Issue Areas and Action Plan

DR. WISE: Steve, did you want to make a comment?

DR. TEUTSCH: Yes. As we move into now trying to triage all of this in an orderly fashion, I just want to remind everybody that at the end of the grid that you have in Tab 5 is a list of the things we have already done and the reports we have already issued.

Not all that we have recommended, strangely enough, has actually come to be. We actually are

continuing to monitor the recommendations that we have already made for the reports on genetic discrimination, where indeed there has been substantive progress, but we are in the midst of seeing what comes out of the Oversight of Genetic Testing Report and the pharmacogenomics one as well. Then there will probably remain policy issues related to the large population studies that were made several years ago.

So we will continue to monitor all of that work. We should have that in our minds as we begin to think about what the new projects are that we want to take forward and what the nature of that work is. I just wanted to remind everyone that that is there as well.

DR. WISE: Thank you, Steve. Just to remind everybody, we are not going to be voting on priorities here. This is not a formal listing but rather to get a sense of the group of how we would prioritize these cluster issues, how we may want to relate one to the other, and to provide some guidance on next steps, recognizing that we need to be flexible in how we

approach this. We need to be responsive to the new administration's priorities and their needs from us.

I will just open the conversation for general comments. Please, Marc.

DR. WILLIAMS: This is a daunting task, to say the least. But thinking about it from a process perspective, and reflecting on the conversations that we have had already and the investment that all of us that have taken leadership in one of these areas have made, it seems to me that maybe the way to think about this going forward would be not to necessarily take the seven clusters and try and arrange them in some type of a rank order but to reflect that each of the clusters has certain things to bring to topics that the Committee as a whole may feel heavily invested in.

The concept that I would put forward as a straw man would be to perhaps leave the seven clusters as they are with leadership to keep appraising what is happening in that area but then to focus in on what are the areas where we really think we have some opportunities to leverage.

For example, we are in the middle of the

Education report. We have heard about how several of these clusters are going to be relating. If that is going to be something that we prioritize, can we then have the cluster leads in the other areas say, this reflects directly onto that, therefore this is going to move up as a priority to support that particular effort of the Committee as a whole.

It seems to me that using that sort of a modular approach, particularly in terms of trying to be nimble in an environment that is going to change rapidly and in ways that are likely to be unexpected, might allow us to maintain expertise around these very important areas and yet readjust the focus within each area to support a communal effort.

DR. WISE: Please. Kevin.

DR. FITZGERALD: Joe and I would like to suggest an alternative approach, that you do everything, and do it all with really large reports.

[Laughter.]

DR. FITZGERALD: And start in March.

DR. WILLIAMS: They are representing those going off the Committee.

DR. WISE: Thank you for that helpful suggestion.

[Laughter.]

DR. WISE: Other general comments, particularly about process and how to think about going forward?

DR. EVANS: I was just going to say, one of the good things that could help is to identify specific niches. Here is something quick we can do within one of these. Then the rest of that particular agenda can be put a little bit on the back burner.

MR. KIRCHNER: Actually, to be somewhat concrete and hopefully helpful, I think there are a couple things that came up that we could put in an order. For instance, on the informed consent thing, the HRSA meeting will be happening in March. We could have something set aside where we would decide what sort of educational segment you would want, say, in July. That is one thing we could just put down right away.

Similarly, with Mara's suggestion of pulling people together, once you look at the report from the

University of Washington you could at least frame questions for the next meeting, which then would allow you to go out and look to see who are the people you would need to bring in to answer those questions.

It would be another thing that we could do concretely. Line all these things up for the March meeting and then we can jump off from there. You might have a better sense of where the next administration is pushing, at least, at that time.

MS. ASPINALL: I like that idea, but I wonder if we have time for that and whether we need to move faster to start things before the March meeting, or at least prioritize them.

One option -- and obviously things are still in flux, but we have some data -- is to be able to attempt to meet with the new administration relatively quickly. Maybe seven is not such a large number to propose and say these are our seven priorities. This is a committee reporting to the Secretary, so the Secretary's preferences are pretty important in the context, but this is the process that we went through and these are the seven that we have.

In the January time frame, if that is not too unrealistic, we could present all seven of them, unless there are some that the group today would like to say they would like to take off the list. But assuming that is not the case, present those in the January time frame and say these are our seven, we would like your input, what is most important to you as the new incoming Secretary. We would then have the ability to hit the ground running so that by the time we are at our March meeting we already have their input and can begin to move forward.

DR. WISE: South Dakota is beautiful in January.

I think that is helpful, particularly as it relates to the next thing on our agenda, which is putting together a brief report of our activities and plans with a cover letter that would precisely introduce not only the Committee to the new administration but what we feel are our strategic contributions to the issues of the day.

In going through the different clusters and listening to the conversation and the very helpful

public comments, I was struck that there were not quite principles but what I think of as strategic contributions that could help us frame the seven clusters.

One is, and clearly it is going to be crucial, genetics in the service of reforming the healthcare delivery system. That includes Cluster Nos. 1, 2, and 3, and certainly No. 7 is the overarching one. But the clinical utility, coverage and reimbursement, and ensuring that there is a workforce capable of actually implementing what everybody is hoping for in healthcare reform, are going to be crucial.

Second is the idea that genetics will be crucial to improving public health and population-based prevention. That is clearly Cluster No. 6, but there is also a larger framing construction that would allow us to engage in those issues and which came up very high in the ranking that we had prior.

Third, individual engagement with genetics and protections and the public's growing awareness and engagement with genetics. Direct-to-consumer

marketing is merely one important component of that, but this issue is going to be crucial, and protections for individual engagement are going to be as well.

The last is to ensure that the new genetic technologies will enhance equity in health outcomes. We ensure that we will reduce disparities in health as the health of all is improved. There is no single cluster for that strategic contribution but rather we have decided that that would in fact be a component of all.

In thinking about how we would frame our seven clusters, we could just list these as our priorities. But there are different levels and they have different histories. I'm looking for ways to frame our seven in ways that would definitely be clear as to why we picked these seven.

So I'm just coming up with, say in our cover letter to the new Secretary, to say we need to make sure that genetics is a central part of healthcare reform. Then we have specific priorities that we think are the best ways to do that.

Second, public health and prevention clearly

is going to be engaged by this administration. Genetics actually has a meaningful role.

Third, we need to talk about public engagement, public awareness, and protections.

Lastly, health equity is a crucial component of everything we do. We need to ensure that the genetic insights and capabilities address these issues in a meaningful way.

I will just throw that out again just to concretize the situation but really building on what your suggestions would be.

In the document that follows the cover letter, we have all seven. We would include all seven clusters in greater detail. They all have components of the issue briefs. But this overall framing I think is important as to what we are about and what we feel the new administration needs to address.

Do you have a comment?

MS. ASPINALL: I think it is great. I agree with doing it that way and organizing it. I think you described three major fundamental areas for which the seven would then fall, but having the information on

the seven is a great step forward and just organizes it in a little bit more context.

I would obviously like the ability to edit things given the comments that we had today going forward, but I think getting it out and emphasizing to the new Secretary having his and the staff's view of that before our March meeting, will allow us to hit the ground running quickly.

DR. EVANS: In that editing, the one thing that I think is really important is, as it stands now, there is quite a prominent slant on how healthcare reform can bring genetics into the fore. I think it is really important to go the other way. I think it is really important to emphasize to the Secretary and to the public that the advent of genetics in medicine is going to drive medical care. It is going to affect medical care in that other direction. I think that is very important to articulate.

DR. WISE: Other comments about prioritization?

[No response.]

DR. WISE: Steve, did you want to make a

comment? Are we fulfilling what you hoped to accomplish?

DR. TEUTSCH: I think we are fulfilling it. I think we still have the actual heavy lifting. I actually like your framework about the clinical care system, where it is going, where the population system needs to go, and then how individuals engage with things, as well as disparities.

I think that is a rubric that people can relate to and understand. Then we can get to the pieces below it.

I do think, for our own purposes, we need to think about, given all the things that are on the table and the areas where we might actually make a substantive impact over both the short term and then over the longer term, how would we think about which of these issues we want to tackle in a way that we can manage within the resources we have available to us. I do think we should have that discussion.

I do think this is actually helpful. Your reduction from seven to four is actually pretty helpful in communicating effectively, what are the

components. Maybe one thing we could do is get that up so people can see it. Then we should have a little bit of a discussion about what we think would be the most important short, immediate kinds of things that we can do, as well as a couple of larger reports that we really could undertake over the next few years.

How many do we usually manage at one time; two or three, right? She is thinking one. We manage, usually, a couple to three at a time.

DR. WISE: The Workforce is going to move forward. Is that correct?

DR. TEUTSCH: That is ongoing. The Patent report is still in the midst. That is two. But then, hopefully, we will get to the end of the Patents over the next year and we should be prepared to take on what we think would be --

DR. WILLIAMS: Jim just fell off his chair.

[Laughter.]

DR. TEUTSCH: That means Jim can take on the next one because he will be in such fine shape.

But we should think about what are the next important topics that we actually want to take on and

then which of these we really want to get on with in some more shorter-term agendas.

I would be very interested in hearing Committee members' thoughts as to, given all of the important issues that have been put on the table, which are the ones that are likely to be the ones where we can make the most difference.

DR. WISE: Andrea.

DR. FERREIRA-GONZALEZ: There is an issue that touches all laboratorians, and it has been touching us for about the last five years, or even more and we haven't realized, which is the critical shortage of laboratory personnel. We have this as a brief report in Cluster No. 7.

We are at a very critical time point where our current workforce, working not only in genetic laboratories but the entire laboratory community, is reaching a mean age of about 40 to 45 years old. We don't have a lot of people going into this type of work.

One of the issues that we have is that we don't have enough schools. Schools are being closed

due to lack of funding. But also, there doesn't seem to be enough incentive for young individuals to go into the field. Another problem that we have is that we cannot retain them. They usually go to work in IT, information technology.

This is a critical issue that I think we are currently facing in many different areas in the country. Maybe we could start developing a brief report or a white paper where we can start investigating the issues of where we are and what can be done. That could have a huge impact on the crisis that we currently have.

DR. WISE: Is that part of the Workforce purview at this point? Is that a central element?

DR. McGRATH: We talked about including laboratorians and decided to put them next. But certainly this is not cast in stone. We could move it up and include it as part of the three groups. But there was a decision made not to.

DR. WISE: Other comments?

MS. ASPINALL: Even if it is part of the education group, that is, I think, a little bit

different from what Andrea is talking about, which is the availability of the personnel. We take it up in No. 7 but again on more of a long-term basis, less of a short-term basis in terms of reacting to what many have described as a crisis in the field.

DR. FITZGERALD: But, on that note, Mara, in your long-term view, one could differentiate between the things that are in crisis now, or will soon be, which could derail the long term. So, would we be able to break out of your report those issues? First of all, identify the workforce issues and other issues that have to be addressed in the short term if we are ever going to get to the long term. That may be something that this Committee could do which would be unique.

MS. ASPINALL: We could do that. We actually highlighted that in one of the policy questions for exactly that reason. We got some comments to broaden that. But I think the perceived current crisis is in the laboratorians of all types.

DR. FERREIRA-GONZALEZ: That is why I'm bringing it up a little bit separate but within this

group. If we are going to develop a brief report in the long term, this could take, with all the other reports that we have, two or three years to really come out. I think we have to start investigating this very proactively. Maybe there is something that we can recommend.

DR. TEUTSCH: Barbara, I hope your memory is sharper than mine. When we made the decision for your committee I thought we made the decision to exclude laboratorians explicitly? We can always revisit that. I wonder, given that decision, what has changed. Maybe I should address this to Andrea.

If it is clearly established that there is already a problem, is there something that we could do that is something less than a full report that could begin to help with the solution to that rather than to evaluate it?

DR. FERREIRA-GONZALEZ: One of the questions is do we include it in the Education report with a very specific scope, which is to start looking at specific areas of the crisis: why are there no schools, why are we not attracting or even retaining

people, and what changes can be made. That could be part of the Education report, but I'm not sure how far along that has already gone and how wide the scope of that report is. Is it going to get diluted in everything else that we do.

I think we can do a brief report or maybe a white paper where we can get something up and going to deal with these specific issues.

DR. WISE: Mara is next.

MS. ASPINALL: I still believe that laboratorians may be included in the Education, but I think that is a different issue than what Andrea is talking about. The kinds of things that people have talked about in this field and we could take a stance on short term are seed funding -- I'm familiar with this in the education arena -- for community colleges to take on programs in laboratory medicine. That has been incredibly effective in the education field, where, as a result, in about 10 years the number of programs that were available both at the two-year and then four-year colleges was tremendously enhanced.

The private industry councils and the

workforce development monies, which are increasing in the context of a recession where retraining happens and there are federal dollars not necessarily from HHS but ones more broadly, can be directed to careers. This happened in the nurse community, where there was a tremendous amount of money funneled off specifically to train nurses for the next generation of nursing. This was about 15 years ago. Even after that, we are still dealing with a nurse shortage.

So there are some very short-term pieces that, in my mind, don't require legislative support or new laws fundamentally to do it but to prioritize laboratory medicine in a way that brings what very well may be a larger number of unemployed individuals into the field in a short period of time.

It is not quite that easy because there is a fair amount of education. So you don't pull a lever today and have it work tomorrow. But it is comparable to what happened in the nursing shortage, where both universities and companies got together, priorities were made in these private industry councils and in workforce development money. Those, to me, are the

kinds of initiatives that we might be able to put together in a relatively short time in a white paper.

DR. WISE: Marc.

DR. WILLIAMS: A comment and a question. The comment relates to the reimbursement aspect of that particular issue, which is particularly for Ph.D. laboratorians. There are some reimbursement issues -- you are shaking your head no. There aren't any reimbursement issues?

DR. FERREIRA-GONZALEZ: The laboratories get reimbursed for the CPT codes and the different procedures, but I professionally cannot bill for it.

DR. WILLIAMS: Correct. But when you are talking about retention, and I know this from speaking with other institutions, some of the issues have been, why should we pay for these folks to be there if they can't bill for their services or we have to do work-arounds, or whatever.

I think there are some reimbursement aspects of it that may impact retention to some degree.

DR. FERREIRA-GONZALEZ: I think you are right on the money on that. I'm not talking about the

Ph.D.-level individuals like myself but the medical technologists of the bench. We are talking about a crisis at the bench. So the issue is also that, due to the short reimbursement, we don't have a free market to be able to increase salary support for these individuals. It is not because we don't have people, it is because we don't have money to pay them more. Normally, we lose them to information technology, to be honest with you, at least in our case.

So reimbursement could be tied into decisions, but I like some of these ideas about the white paper and trying to see how we can reeducate some of the individuals that are in the workforce to do this.

DR. WILLIAMS: The question I have relates specifically to the creation of the white paper. I don't recall this specifically, but what, if anything, was addressed within the several Banbury conferences on education and genetics? Was there much time spent around that?

In other words, we shouldn't create a white paper if Banbury has addressed this. If they haven't

adequately addressed it, then I think that would be a worthwhile investment of time.

DR. FERREIRA-GONZALEZ: I think this goes beyond the inherited disorders genetics field into other areas. It is not just the cytogenetic technologies and the molecular biology technologies. It goes to all parts of the laboratory. We have to assume that genetics is percolating to every area of the laboratory. So it is just not that narrow. It has to be a broader scope.

DR. WISE: I was just going to ask about the role of the Committee. I know other groups have tried to address this issue and have put out reports. HRSA has. The Bureau of Health Professions relatively recently put out a large report specifically on this issue. Some professional groups have been working and advocating on this issue.

What would you see would be the role of this Committee given that the clinical lab workforce issue is much beyond genetics? What would you see the role of this Committee as being given the other reports and other work being done more broadly around clinical

workforce?

DR. FERREIRA-GONZALEZ: I think we can look at what other groups have been doing or what has been reported and see if there are areas that we continue to discuss and then contribute to because there are gaps or nothing has been moved forward. I think we can start surveying what has already been done and then move forward from there.

DR. WISE: Barbara.

DR. McGRATH: I was just going to ask that same question. I think there is a difference between genetics education, like basic education and continuing education and training, versus workforce issues, which is getting people into the pipeline. Maybe that is a little bit where the line is.

I don't know agencies as well as the rest of you, but I think HRSA has often picked up that workforce part of the territory. I'm not sure about that.

DR. WISE: Kevin, did you have a comment?

DR. FITZGERALD: This falls under Mara's purview in the future piece, so we are talking near

future. In the near future, if we have the $1,000 genome, what does that do to the demand for clinical laboratorians? We can put it in that context, too. If this is really going to ramp things up, that is something else we need to look at.

DR. FERREIRA-GONZALEZ: Exactly. If you have the $1,000 genome or everybody gets screened for genetic disorders, like carrier screening, we still have to have people to run the tests. It creates an issue, but it goes beyond just the molecular biologist in the genetics laboratory. You have to have individuals in other areas of the laboratories where testing is going to be done.

As we continue the implementation and move these personalized medicines and genomic medicines, it will require more testing in the laboratory. We need to have a workforce and retain that workforce.

DR. EVANS: Just so we don't forget, the ripple effects will be huge. We will need a lot more genetic counselors, for example.

DR. FITZGERALD: And maybe even some clinical geneticists, but I don't know.

DR. WISE: Other comments more broadly? Steve.

DR. TEUTSCH: I want to push folks a little bit here. We have heard a lot of things. I don't have a magic number, but three to five short-term terms, three to five monitoring things that we can afford to pick up, and then a couple or three longer-term projects. I would be really interested in what people think those things should be. I can push back on each of these cluster leaders to talk about what those might be, but I would be very interested in which people think are likely to be the most impactful that we should take up.

DR. FITZGERALD: Just on the monitoring, I think we have some for those. First of al, the informed consent one, because there is a HRSA meeting coming up. OHRP may be looking into this too at some point in time, I'm presuming. So that may be adequately addressed by other people and we don't need to wade into that. That could be a monitoring.

The DTC issue, again, is something that we need to monitor to decide how we want to frame it for

this Committee. Although I know there is some desire here to move quickly on that, I'm not sure we have decided how we want to weigh in on that yet. So that is already there.

Then the short-term needs. We could certainly pick two or three of those, as we have already discussed. I think you are right; genetic counselors have to be in there, too, because that is obviously part of that ripple effect. The long term I guess I will leave to others.

DR. EVANS: In response to Steve's plea for specifics, I would just throw out there that, of the things we have discussed this morning, I think that two short-term items that could be addressed, one very short term, would be something along the lines of what Mara suggested, a letter to Daschle emphasizing the importance of genetics and the changes it will bring about and that it has to be factored into plans for healthcare reform.

I think that a second, relatively short-term item could be something along the lines of a checklist in the DTC arena, as has been suggested.

Then I would personally advocate that, given the incredible importance of reimbursement for the practical functioning of the field, including genetic counselors, that would be a high priority for an in-depth report that should be pursued and initiated quickly.

DR. WILLIAMS: So you think we need a new in-depth report.

DR. EVANS: I saw the puzzlement. What I'm saying is we need to act on those and proceed vigorously with what has already been done.

DR. WILLIAMS: Right. I think that that would be good.

DR. EVANS: I think you should do it again.

DR. WILLIAMS: I know. Right. I was going to throw my BlackBerry at you.

[Laughter.]

DR. WILLIAMS: I'm sure it was almost implied in your sense that that will be something that we will be doing. I don't see that as an item that is even on the table for debate.

DR. TEUTSCH: Let me see if I can try this

out on you. Having listened to this discussion, I'm going to try and run through each of these. You can tell me where I'm miles off base.

Under coverage and reimbursement services, what I heard is that in fact the most important things we have to do are monitor and look to the implementation of the things that have already been out there.

DR. EVANS: Yes. Not just monitor, push.

DR. TEUTSCH: Yes. But we are clearly having an interaction, particularly with CMS, on those issues that we will continue to, yes, more than monitor. But it is in the sense that they are there and our job is to work with the organizations to help move them forward.

We heard that the clinical utility is important but, under the rubric that we talked about, emerged under the future of the healthcare system. I'm not convinced that we need anything there because we have already made some of the salient recommendations as part of the Oversight and Pharmacogenomics reports.

We just heard about genetic education. With that, we probably need to add the laboratorian component in a stronger way. But that is already underway.

DR. FERREIRA-GONZALEZ: No. It is the definition. That is what we were talking about with Barbara.

DR. TEUTSCH: You talked about a white paper and some shorter-term things. I'm sorry. I didn't mean in the context of a larger report.

We talked about informed consent. This is going to be really important in the privacy issues as we get to the $1,000 genome, the EHRs, and all of that, as to how we are going to do that research. It is going to be central to the clinical utility if we are going to be able to use those kinds of resources.

I heard that we are going to at least listen to what comes out of the HRSA conference and then identify whether there is something that needs to be done there. That ties into what Michael was talking about with protection of human subjects.

I also heard strong interest in at least

doing some short-term assessment of the DTC, direct-to-consumer testing. That would be a short-term thing that we would probably want to take up in a way that we could stay on top of that, more than just watching it. Details to be worked out.

Public health applications. There is a lot in there. I haven't quite got my head around exactly what that is going to be. We talked about performing a systems review. That gets you on to things, but even within that there is a lot that can be done. I would be interested in others' thoughts about what can be done there.

Then I'm going to push a little bit on Mara because it seems to me that the big one is about health reform and the key things that can be done. Clearly, we could do an in-depth report and spend several years, but I know you don't want to do that. I don't want to do that, either. I want to pick out a few things within that area that we can get on with. But I suspect that that is going to be an ongoing major effort.

MS. ASPINALL: Are asking for comment now?

DR. TEUTSCH: I'm trying to put out a straw man of what I have taken away from the conversation. That is my interpretation of most of it. So yes, I am looking for some thoughts. Paul.

DR. BILLINGS: Sorry. It is hard for me to raise my hand and get noticed.

I didn't hear whether the regs and recommendations associated with GINA, which is in my view an immediate issue rather than a long-term issue, are part of the discrimination cluster or whether we have spent our wad on that one already.

DR. TEUTSCH: It is currently listed as one of the things that we are going to be monitoring. We have those four items that we have already issued reports on.

DR. BILLINGS: Sure, sure.

DR. TEUTSCH: That was one of the items within that.

DR. BILLINGS: But more than monitoring, given that we are at a crucial period and that the incoming administration will have some impact on the regulations and enforcement of the legislation, do we

want to be a little more aggressive on that issue?

MS. LEIBIG: Hi. I'm Kerry Leibig. I am from the EEOC. I can tell you at least in terms of Title 2 of GINA we are moving along as quickly as we can. Actually, the person who was most involved in drafting the notice of proposed rulemaking for those regs has recently left and accepted a job with Department of Justice. I'm his replacement. You are familiar with Peter Gray. He has been working on this issue a long time.

I'm doing my best to fill in for him. We have high hopes that the NPRM will be coming out soon, at least in terms of Title 2 employment discrimination.

DR. TEUTSCH: Right. So that will be sent out for public comment; is that correct?

MS. LEIBIG: Right, right. Under the Federal Rules of Civil Procedure, they would be published for a 60-day comment period. Then we would take in all the comments and then come out with a final rule after our commission signs off on them. So we are still in the steps of drafting the notice of

public rulemaking, which is our proposal for what the regs will be, which we will then get comments on.

DR. TEUTSCH: So that is one part of the implementation. Yes.

DR. WILLIAMS: Just a clarification. Will all of the titles come out for public rulemaking at the same time or will they be issued independently?

MS. LEIBIG: They will be issued independently. EEOC only has the authority and knowledge to do it on Title 2, which is the employment section.

DR. TEUTSCH: We have the insurance one, too. I think that is due for release we said in November of next year?

MS. CARR: The law becomes effective in November. My understanding is that the HHS, agencies within HHS, Treasury, and Labor are working together on the health insurance provisions. I'm not sure whether that will come out as an interim final rule or as a rulemaking for a proposed comment. I'm not really sure. Alan, I don't know if you know more about any of that. I wish Robinsue were here. I'm

sure she would be able to fill in some details for us.

DR. GUTTMACHER: Yes, that is consistent with what I know. I'm not sure whether they have made any decision. But there may be a decision; I don't know.

MS. CARR: Perhaps at our March meeting we might want to have a fuller report. I bet things will be clearer by then.

DR. TEUTSCH: That would still be timely, wouldn't it?

MS. LEIBIG: I would hope that the NPRM would be published prior to that and we would be working on the comments that we received. Our hope is to publish a final rule by May, but it depends on how things go at the Commission. The new administration is going to have to weigh in.

DR. TEUTSCH: Paul, what did you have in mind beyond trying to coordinate with these agencies?

DR. BILLINGS: If there are key components from the Committee's point of view of the law and either stricter interpretations of potential rules or less strict interpretations of rules. As we have just

heard, the new administration is going to have its say on the construct of these enforcements. We should educate the leaders on that.

DR. WISE: Thank you, Paul. We are heading towards lunch.

DR. TEUTSCH: Yes. Good.

MS. ASPINALL: I have a narrow comment and a broad comment. On the narrow comment, which is in answer to your question about Cluster No. 7, I think I could, given the comments that we heard today from the Committee and the public, have probably three priorities within Cluster No. 7 that I would focus on: workforce, health information technology, and monitoring and evaluating effectiveness.

If you wanted me to at least put a straw man out to prioritize within No. 7, I could do that. But I think the bigger issue to maybe think about over lunch is, to me there look like two very different ways to go. One says we leave all seven priorities, organize them the way Paul described, send them to the new Secretary, and leave it at that. Continue to focus on the two that we have going in the interim and

wait until we get feedback. That is one approach, which I think would be a reasonable approach.

The other approach is that we prioritize amongst the seven and either start working on them or send them to the new Secretary with a prioritized list amongst the seven. My concern is that is hard to do. I would like to know what the new administration would like to do. I don't think seven is so large a number that it is overwhelming or looking scattered.

DR. TEUTSCH: I was not proposing that we don't give them the seven. I was just proposing that we begin to clarify our own thinking about how we would take on all the pieces within that. There are other things. For instance, you had laid out the idea that over the short term we could actually convene a group of chief medical officers and other kinds of people that we could actually bring together in the near term. It is hard to believe that this isn't going to become an important topic. We could then get on with the agenda so that when we meet in March we are not back here again.

MS. ASPINALL: That is exactly what I'm

trying to do so that we don't have to go through this again in March and we have clear marching orders amongst ourselves and with the new administration. I'm with you.

DR. TEUTSCH: That is basically what I was looking for. So, yes, we have these broad issues. Yes, they will probably say they are interested in all of them. But if we have a little bit of clarity where we can begin to take it on, then I think we will have a richer discussion with them at a different level of granularity. Then we can move forward.

Why don't you think about that over lunch. I'm not sure whether the hypoglycemia is worse before lunch or after lunch. It probably depends on your insulin status.

DR. WISE: Before we break, I want to thank everybody for being humane with the leader of this task force and all your really informed, very helpful contributions over the last nine months or so. We really appreciate it, particularly to Sarah and the staff and to the members of the task force. Thank you.

DR. TEUTSCH: Great. Thank you, Paul. This has been great. We will probably revisit this, if there are any other comments after lunch. The rest of the day we are going to be reviewing the draft report, which you had in Tab 6. I think it was also handed out in your folders. This is the note that we are going to have to send to the incoming Secretary. So we would like to get your feedback on that.

My guess, for those of you making travel plans, is that we will wrap up a bit early. I think we have made good progress. But, why don't we take our break for lunch and be back at 1:15.

[Lunch recess taken at 12:32 p.m.]

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AFTERNOON SESSION

[Reconvened at 1:25 p.m.]

Review of Draft Progress Report

Steven Teutsch, M.D., M.P.H. and Paul Wise, M.D., M.P.H.

DR. TEUTSCH: Welcome back to our final session for this SACGHS meeting. Thanks to all of you for returning. I know people will start drifting out.

I first want to express my thanks, of course, to Sarah and her incredible staff, who, meeting after meeting, somehow make those of us who sit up here look good. It is incredibly appreciated. They do a terrific job behind the scenes, and it is much appreciated.

Thanks to Abby and her folks, who help us out with so many of the logistics.

We have just a couple things that we want to wrap up here before we conclude the meeting. What we wanted to do is bring to closure the discussion we had this morning. What you have up here is the framework that Paul had suggested to us as we take this forward to the new administration. This is partly so you can

see the actual words that we intend to use, first related to our energies that will be devoted to improving the healthcare system and how genetics fits in with that.

The second is on genetics and public health and population-based prevention. I have to do a mea culpa here because I gave that rather short shrift in my summary. My public health roots have come back to haunt me. I can't go to L.A. County and join the health department if I don't flesh this out a little bit.

I breezed through some of the agenda. I will work with Katy Kolor and with Joseph to try and identify the one or two items that we can move forward with that are pretty specific that we can have as part of those discussions.

DR. EVANS: That is the hard part because it is a very broad topic. It is hard to focus on.

DR. TEUTSCH: It is. But I'm afraid if we say something like "Assess the systems," even I will fall asleep.

DR. EVANS: I would advocate focusing on

something along the lines of using genomic data to stratify populations for risk in the context of screening, something like that.

DR. TEUTSCH: Right. This actually embraces two things. We have the clinical part of the public health in the first bullet. The second part is really the population-level stuff. Yes, it could include some of the screening, but it probably needs to deal with some of the environment-gene interactions and the risks that accrue to communities, subpopulations, and things like that.

So I will work with folks to do that. Obviously, it is not going to be cast in concrete, but I think that was the distinction we were trying to make. Those of us in public health think that the clinical system is part of that rather than separate, but that is a whole different set of ego problems.

The third one is, we talked about the individual engagement that deals with, certainly, the privacy and protections issues, as well as the direct-to-consumer genetic testing.

Finally, there is that major cross-cutting

issue that we all feel passionate about but it is hard to get your hands around it. It is a discrete kind of thing about the equity issues in this whole area.

Jim?

DR. EVANS: Equity issues?

DR. TEUTSCH: It is disparities, right? Fairness, disparities. Do you prefer "disparities" to "equity"?

DR. EVANS: I don't know. It just caught me by surprise.

MS. ASPINALL: I like "equity."

DR. EVANS: "Equity" is fine.

MS. ASPINALL: "Equity" also includes access as well.

DR. TEUTSCH: Yes. Across multiple dimensions.

MS. ASPINALL: Yes, that is why I'm saying I like "equity." "Disparities," to me, has a different implication. "Equity" is broader.

DR. TEUTSCH: "Equity" is, I think, where we want to go, as opposed to where we are. Is that okay? That was your word; so that is okay with you, right?

DR. WISE: There are people who have built their careers on the difference between disparities, equity, inequality, and injustice.

DR. TEUTSCH: What I would like to do is just bring this to closure. Are there any other issues generally with this framing and what we discussed this morning?

[No response.]

DR. TEUTSCH: Hearing that postprandial slump, we will move to the last major thing. In July we talked about the fact that we would like to engage the new administration in a timely fashion. What you have in Tab 6, and I think also in your folder, is a draft letter, which I assume you have all studied assiduously, to the presumed Secretary, Tom Daschle. We have a little bit to incorporate, which is largely this, that we will incorporate into this letter.

Aside from a general framing of where we have been, some of the priorities that we think we should take up that are based on our prior reports that relate to some things that we thought were ready for quick action that he can build on the activities

of the current administration, we want to talk about the future directions where we want to engage. That is basically the framing in the cover letter.

Then, in the I don't know what we call it. The attachment, the appendix, which says The Integration of Genetic Technologies Into Health Care and Public Health: A Progress Report and Future Directions for SACGHS, begins to flesh out the issues that we just identified in the cover letter.

Many of you have had a chance to look through this. I would welcome any comments.

Paul has played a critical role in drafting all of this. Paul, anything you would like to add to that?

DR. WISE: I think our approach to this was that the cover letter was going to make the broad, general case for both the existence of this Committee and where we think the central issues are going. We have more background and more elaborate discussions, still relatively brief, in the appended document.

We didn't want this to be a laundry list of everything that the Committee has done. We didn't

want it to be a laundry list of all of the things we are considering doing but rather a framing letter that would be accompanied by the more in-depth document that would be perhaps the basis for beginning the conversation between the Committee leadership and the new administration, to begin that co-navigation process that we discussed.

DR. FROHBOESE: I think the letter is very well drafted, will be very helpful to the new Secretary, and will be instrumental in making sure that this Committee's views are before the Secretary at the earliest possible moment.

Now that we have this framing of the strategic contributions, it may be good to incorporate those concepts in this letter.

DR. WISE: That was the intention. It will probably be bullets in that paragraph that covers it very broadly. But I didn't want to presume to insert these kinds of reclustering or condensation until the Committee had an opportunity to really discuss all the clusters and how people saw it put together. So if people are happy with this, we would then put this in

basically as our principles of moving forward, or what we view as our strategic contributions to the efforts of the new administration in this arena. I think our idea was to take this and incorporate it into the cover letter.

DR. TEUTSCH: While you have the floor, Robinsue, we always benefit from your wisdom on how best to facilitate these things. You have seen a transition or two. Any other things that you would suggest that we do so that we can best engage the new administration on these issues? I'm putting you on the spot, of course.

DR. FROHBOESE: No, I'm happy to contribute my views. Right now the Department is going through a transition planning process. So there is an opportunity for each operating division and staff division to meet with members of President-Elect Obama's transition team. We are actively going through this interview process right now and providing material.

I think certainly all of the ex officio members, in going through these interviews with the

presidential-elect transition team certainly can bring forward their involvement with this Committee and the visions that we are setting forth now.

I think once the new Secretary is in place, then working through the Office of the Secretary and getting this letter directly to him identifying who the point person is going to be in the Secretary's office to handle these issues and establishing that contact will be the best way to get the information across.

DR. TEUTSCH: Thank you. Thoughts on the letter? Did we catch them at a weak moment or something, Paul?

DR. WISE: We just did a good job on the letter.

DR. TEUTSCH: We could have done this before lunch. I'm sorry, Sylvia.

MS. AU: Can we just highlight the opportunities for immediate action, Nos. 1, 2, 3, and 4. Can we make them bold or something?

MS. ASPINALL: I'm sorry. Where is that?

MS. AU: On page 6.

DR. TEUTSCH: Oh, you are talking about the attachment.

MS. AU: It just blends into the letter.

DR. TEUTSCH: Sure. I agree. Those are things we do want to highlight. We debated about how to actually format that even in the cover letter. They are there as well, but it is hard to highlight everything in a cover letter. Yes?

MS. ASPINALL: I love Sylvia's suggestion. Maybe the opportunities for immediate action should go in the cover letter, not in the attachment.

DR. TEUTSCH: They are there, right?

DR. WISE: No, it is not.

DR. TEUTSCH: Which version am I looking at?

DR. WISE: I'm the one probably responsible for yanking them out of the cover letter.

MS. ASPINALL: So it is you we should blame.

DR. TEUTSCH: They are there. They just are not formatted this way.

DR. WISE: I'm the one to blame. The reason was because we are making, in the cover letter, an existential case. We are talking about healthcare

reform and we are talking about broad issues of public policy. Then in the next few sentences we are talking about specific, immediate action steps, which is just nowhere going to be on anybody's radar screen until the assistants to the assistants to the assistant secretaries are in place.

My concern was just the mismatch of scale. It was jarring. It really had that cut-and-paste kind of feel. So my suggestion, to respond to Sylvia's suggestion, would be to elevate the immediate action steps within the attached document, make them bold, move it up in the document, but not to put it in the cover letter because of the mismatch of the scale of the immediate action steps with the main purpose of what we would like to get across.

MS. AU: I think that makes sense. Somehow online it looks a little bit different. There is so much information here it is hard for anybody to get through easily, but it is well written. I like the paragraphs. Maybe the opportunities for immediate action become a separate attachment that is independent and then you have the summaries there.

I understand about not putting it in the cover letter as you have now described it, but maybe having two attachments there so it is actually separable and looks different from the rest of it. Even at the beginning or end of what is a pretty substantive, six-page document, it may get lost.

DR. TEUTSCH: That is a good thought. Yes, Charles.

DR. KECKLER: I think that is a good decision. Following up on Robinsue's point, one thing that you might want to consider during the transition is, I think it is very important to figure out as soon as that person is in place or as soon as possible who the point person will be that is coming in as part of the political administration, either the counselor for science or whatever structure is going to be put in place.

You may want to consider sending the document, the cover letter and/or the attached progress report, to that person along with the Secretary and to send it at the time when that person is there. I'm sure the confirmation won't actually

take that long for the Secretary, but that person may actually become identified before the Secretary is confirmed. It may not be a political and Senate-confirmed appointee.

So you may want to time it and get that document to that person along with the Secretary as the target addressee.

DR. TEUTSCH: That is a great point. We have had an extraordinarily constructive relationship with Rick Campanelli and with Greg Downing and all the folks in the current administration. We really look forward to fostering that with the new administration.

Other thoughts, folks? Anything you would like to talk about for two hours? Is there anything else? Sarah, anything else?

MS. CARR: You got your core goals done.

Concluding Remarks

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

DR. TEUTSCH: Let me see if I can't summarize, then. It has been a great couple of days.

As you recall, the Council on Linkages Between Academia and Public Health Practice is looking

at competencies for the public health workforce. Sylvia and Joseph brought this to our attention and have worked to draft a statement about the competency in the area of genetics that we would like to have included in the statement of the workforce competencies.

If you have comments we will take them now. But we have until what, the 15th; is that the date? It is due by the 15th. So we will need them presumably by the end of the week. If you have suggestions on how that is worded, please get them to Cathy.

MS. AU: They only do this every 10 years, so it is probably important for us to get some in there.

DR. TEUTSCH: So please do.

Just to pull things together, I'm constantly amazed at the productivity of this group and how much we actually get done. Just to quickly go over where we have been, yesterday Jim led us through a discussion of the Patents report and a set of not-so-easy options for us to consider. Having reviewed that

and gotten some additional suggestions that I think were all very constructive, we will tidy up that report and it will be sent out for public comment hopefully in the early February time frame. Then we will have 60 days to look at that.

Then it goes back to Jim and colleagues to look at the comments we are likely to get, which we can anticipate as being plentiful. Hopefully we will get a brief feedback on that in June and we can finalize that report by October. That is great. That is a complex and important bit of work. I think as we have heard here with the $1,000 genome and how that is going to interact with the existing patents and what should be patented, there should be lots to discuss as grist for the mill for this Committee. So that was great.

We had the update on the metrics that are being developed and the standards from NIST, from FDA, and from CDC. It is always fascinating to see what goes on down in the trenches of all of these agencies, so it is good to see all that important work that lays the ground work for the credibility of the data that

all of us use.

Then, today, many thanks to Paul and all the folks who have been working so hard on trying to get us ready for the next set of issues that we need to address. I think we had a really constructive discussion and I think it went amazingly smoothly, and that is a credit to everybody's hard work up front to get this framed, get the input, and then to work through that.

Finally, we have the draft that we will be getting in final form for the incoming administration. We are ready to engage with all of them and take it along.

Most importantly, thanks to all of you, ex officio members, guests, Committee members, and particularly staff, for all of the incredible work that you do and for helping this Committee stay productive and relevant.

Thank you, and safe travels. Have wonderful holidays. We will look forward to seeing you at our next scheduled meeting, March 12th and 13th, presumably somewhere nearby. Are you pointing here

because it is here? We will be back here at the Humphrey Building.

Thanks, everyone. Safe travels.

[Whereupon, at 1:48 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 (SACGHS)**

HELD: **December 1-2, 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