Discussion of Coverage and Reimbursement Report
October 18, 2004

DR. TUCKSON: Well, listen, let me get everybody back and thank you. For the record, the ex officios had to buy their own food. I get all these new challenges when I come into the job. It's incredible.

We want to get started on the next session. We're a little behind time. The committee remembers that at the March priority-setting meeting, we identified coverage and reimbursement of genetic technologies and services as a high priority issue requiring in-depth study. At the June meeting, the committee considered a preliminary draft report on coverage and reimbursement which was revised over the last couple of months.

Let me just say I want to really, really thank the staff for these revisions. While the report still will be revised further, there has been a considerable enhancement of an already very good piece. But they have really, really changed the report. I can see a significant improvement as they have listened to the inputs from lots of different stakeholders in this. It now includes revisions that they have worked with that they have received since June. The revised report is in Tab 5 of the briefing book.

Cindi Berry really needs to be thanked for her leadership on this, as well as Debra Leonard, Emily Winn-Deen, Muin Khoury, and Sean Tunis from CMS. I really want to thank Suzanne as well as Amanda for their staff support.

So to lead us through this discussion, I'm really pleased to ask Cindi to take it away.

MS. BERRY: Thanks, Reed.

I want to echo Reed's word of thanks to the staff, because this was an enormous undertaking. Reports of this magnitude don't just appear mysteriously. To my knowledge, they haven't invented a machine that will take down everybody's comments and then synthesize it and make sure everything is grammatically correct and organized and insightful. That is something that the staff did, and we owe them a debt of gratitude because there is no way that any committee can draft a document such as this in the amount of time that it has taken and produce the results that these folks have produced.

So Sarah, Suzanne, Amanda, Fay and others, incredible job.

I'm going to switch these two, the report purpose and goal. I'll start with the goal, really to improve access to genetic technologies, genetic services, genetic tests in particular. We have in there the word "appropriate," and that is key because no one here is suggesting that all genetic tests and services should be paid for and covered no matter what. We're talking about appropriate coverage and appropriate reimbursement, and that is the overarching goal, one of the goals of this committee.

So to that end we said, as you'll recall from previous meetings, how can we do that? How can we accomplish that? What are the barriers to achieving that goal? Two of the largest barriers really have to

do with coverage and reimbursement. In many cases there is inadequate, insufficient, or non-existent coverage of genetic tests and services, and to the extent that some are covered, the reimbursement is not adequate and is a further barrier to access.

So the goal of the report or the purpose of the report is to sort of do a comprehensive analysis of the state of play with regard to coverage and reimbursement, and then offer recommendations to address the discrete barriers to access and the problems that we identify with regard to coverage and reimbursement.

As you'll recall, at the June meeting we did take a look at a first draft of the report, and we had a robust discussion, but we didn't finish the discussion because there are so many issues involved with coverage and reimbursement. We didn't get too far, so the suggestion was made, and we implemented it, to form a coverage and reimbursement task force which would be tasked with further examining the report, the objectives, the barriers, revise the report based on some of the recommendations we heard at the meeting, based on input from members of the public.

We held a task force meeting on September the 8th to further delve into the issues that we identified and recommended some additional deliberations. This is the task force. Then our September 8th meeting consisted of the following participants. We had some outside folks because we had a lot of questions in terms of how operationally genetic tests are actually provided, the reimbursement issues, what are the challenges there. So we had a lot of questions. Based on that input, further revisions were made to the draft report that you already saw.

These were the goals of the meeting, develop some concrete recommendations for the full committee to consider. The task force did not want to and did not attempt to say we're going to just come up with recommendations and everyone is going to bless them. Not at all. What we were doing is developing a list of possible recommendations for the full committee's consideration, and you'll see as we go through the report, and if you already read the report you'll see that there are numerous recommendations under certain sections. Some are additive, some are somewhat mutually exclusive, and in some cases we really didn't have any recommendations.

The purpose of today's session and tomorrow's session is to really get everyone's input on the committee, ex officio members, and members of the public so that we can come up with very thoughtful, concrete, and productive recommendations to addressing some of the barriers that we identify.

A final goal of the September meeting was to plan this session.

I next want to turn to Dr. Linda Bradley, who is going to give us a presentation. It was done really at the request of the committee from the last meeting. We discussed the need for some sort of mechanism for assessing when the evidence base is sufficient for establishing clinical utility and making coverage decisions, as well as a mechanism for addressing gaps when there is no real evidence to support coverage necessarily for genetic technologies. It was brought to our attention that the EGAPP project, Evaluation of Genomic Applications in Practice and Prevention, which is really the next step in the ACCE process Muin had identified for us the last time, really would be a potential model for us to consider.

So at our last meeting we requested a presentation on the EGAPP project so that we can assess whether

it's something that we want to incorporate, either some of its work or all of its work in our coverage and reimbursement report.

So with that, I'd turn to Dr. Linda Bradley for her to brief us on this project.

DR. BRADLEY: Mr. Chairman and members of the committee, we really want to thank you for allowing us this opportunity to provide a brief review of a recently launched model project by the CDC.

It's important to point out that this project does not represent a new concept as much as an evolution of ideas and methods dating back to the 1997 report of the Task Force on Genetic Testing. As most of you know, this report emphasized the need for evidence-based review of new tests during transition from research to practice, and for a coordinated process to collect data in pre- and post-market periods. It also described assessment criteria, analytic validity, clinical validity, and clinical utility in the context of genetic tests.

SACGT, through its subsequent deliberations, affirmed the task force assessment criteria and added emphasis on ethical and social issues as a component of evaluation. SACGT also encouraged collaboration for data collection and education and made other specific recommendations, among them that CDC play a coordinating role in data gathering and analysis.

In 2000, CDC took a step in addressing the need for pre-market review of data by funding a cooperative agreement with the Foundation for Blood Research to evaluate the ACCE model system, which has been referred to here. ACCE, as most of you know, simply reflects the components of evaluation laid out by the task force and SACGT. The ACCE process is based on the premise that you first define the specific disorder or phenotype to be tested for, then the test, and the setting in which the test is to be performed -- for example, diagnosis or predictive testing -- then an analytic framework -- in this case, sets of targeted questions can be used to systematically review evidence on each component -- analytic validity, clinical validity, clinical utility, and related ethical, legal, and social implications.

This process was designed to assess availability, quality and usefulness of data on DNA-based tests for disorders with a genetic component. ACCE differed from some other standard methods, and some characteristics of the ACCE process included a broad focus. ACCE aims to provide a first look at all the available data, not just the published literature, and also reviewed all evaluation components, including analytic validity.

ACCE used an ad hoc approach to grading quality of evidence versus a more structured approach in order to extract maximum information. ACCE reports included review, analysis and integration of data, and identification of gaps in knowledge and the data needed to resolve them. An objective of ACCE was not to suggest policy or make recommendations but to provide complete, accurate, up to date summaries in formats useful to a range of audiences.

Five ACCE reviews have been posted for comment, and you can see the titles here. We've learned a great deal from this process. I think two points that I would raise is that we learned, as I expected, that this information is eagerly received. We've had rapid uptake and application of the evidence developed. For instance, data developed for the CF report was utilized in preparing the 2004 revision of ACMG's

mutation panel recently published in Genetics and Medicine.

We've also learned that in moving beyond the published data, we can uncover new and useful information. For example, in the absence of any other data, new estimates of analytic validity in the CF, HFE, and Factor V reports were derived from external proficiency testing data. Though not an ideal approach, it provided a reassuring snapshot of U.S. performance as not perfect but really pretty good. It also highlighted situations in which understanding of analytic performance could impact how a test is implemented in some settings.

So as we considered the next step from a public health perspective, the question really was what might a non-regulatory process for evaluation of genetic tests look like? Well, certainly evaluation needs to occur at two key points. The first is transition from research and development to clinical practice, ideally before a test enters widespread use. It should include systematic review of evidence on clinical validity, and with the rapid development of new complex technologies with very little performance data, systematic review of analytic validity needs to be part of this process, too. In some cases, certainly a first step.

Assessment of risks and benefits, while focused on outcomes, needs to include more, including consideration of resources for the testing process, counseling and education, the need for pilot trials, and cost-effectiveness analysis. We need to identify more effective approaches for assessing ethical, legal, and social implications of testing, and there needs to be a plan for dissemination of the information developed to all relevant target audiences.

The second key point for evaluation is in the post-market period to assess performance in practice and public health impact, beginning with very basic information that we currently lack on utilization and access. We need to be able to document problems and successes with implementation and fit with health care delivery systems, and we need to be able to update the knowledge base after the test has been in practice.

Evaluation of Genomic Applications in Practice and Prevention, or EGAPP for short, is a three-year model project. The goal is to establish and evaluate a systematic mechanism for pre- and post-market assessment of genetic tests and other genomic applications in the U.S., hopefully one that can be sustained in some form beyond this model project. EGAPP is a public health initiative with a population focus, and like ACCE the objective is a first or early look at new tests and technologies to determine what is known and to identify important gaps in knowledge.

The project plans to utilize information and recommendations developed through this and other advisory processes, as well as the knowledge gained from the ACCE project. Partnerships and collaborations are vital to the success of this project. Examples include existing evidence-based processes -- for example, the Agency for Healthcare Research and Quality, the U.S. Preventive Services Task Force, evidence-based practice centers, and the CDC's Task Force on Community Preventive Services. We began talking with these groups early in the planning process, and ongoing discussions have been helpful as we sought to define the scope of the project. These and other agencies and organizations, many represented here today, will have a continuing role in advising the project.

We're also interested in collaborations with the international health technology assessment community,

and we are developing very productive contacts with groups in Canada, the United Kingdom and the Netherlands who are involved in similar projects.

We're also interested in relationships with other projects and initiatives. There are certainly too many to cover here, but just to mention a few, with regard to quality assurance and improving laboratory practice, CDC's Division of Laboratory Services is currently developing a process to obtain and distribute quality control materials for genetic testing to labs, researchers, and the diagnostic industry. CDC Division of Laboratory Services, in collaboration with Emory University and the National Institutes of Health Office of Rare Disease, held a May meeting on promoting quality laboratory testing for rare diseases. Outcomes included the formation of this laboratory network and planning for a second conference to move into implementation of the recommendations developed. There are also a number of other initiatives related to policy, programs and services, and research.

The process aims to provide a clear linkage between the evidence developed and the recommendations made, minimizing conflicts of interest in the review process, but keeping in mind some very good advice from Al Berg of the University of Washington and the U.S. Preventive Services Task Force, that evidence-based requires that the linkage be transparent, explicit, and publicly accountable, not that it be objective. The project will develop a plan for effective dissemination of information to target audiences.

The question of are genetic tests different or exceptional comes up here again, and whether or not genetic tests are exceptional or different in other ways, methods for assessment of genetic tests have basic similarities to those used for other tests. However, there does seem to be an increased awareness and concern about genetic testing and a public perception that it is different, and I think the compelling testimony you heard this morning about potential harms makes that very clear.
EGAPP is focused on genetic tests and other genomic applications, responding to the demand from health care professionals, policymakers, and the public for a source of reliable and reasonably objective information about appropriate use of genetic tests. However, the knowledge gained about successful evaluation approaches, methodologies and infrastructure should certainly be applicable to assessment of other tests or emerging health care technologies.

Technical and logistic support for the project will be provided by RTI International, a non-profit contract research organization, and this contract was awarded in late August. RTI brings to the project a wide range of scientific expertise, but one resource we feel is very relevant to this process is the RTI University of North Carolina Evidence-Based Practice Center.

So the central element of the project is the working group, independent, non-federal, multidisciplinary, made up of 10 to 12 experts from fields such as health care, genomics, epidemiology, health technology assessment evidence-based review, public health, health economics, and potentially others. Two in-person meetings are planned in year 1, three meetings in each of years 2 and 3.

Proposed roles of the working group -- and this is the concept -- is that the working group would first develop an organizational plan defining protocols for evidence-based review and development of recommendations. The working group will certainly consider input from stakeholders, develop criteria for selecting topics, and then select and prioritize topics for review. When a topic is selected, the working group will request that RTI commission or conduct an evidence-based review. The working group will

ensure appropriate review of reports and develop recommendations based on the evidence. They will consider needs and strategies for post-implementation monitoring and data collection studies and will take part in evaluation of the project.

Stakeholders, very important. The project will identify and engage a wide range of stakeholders. The primary focus for this project is health care providers and consumers. A secondary focus is policymakers and health care payers and purchasers. RTI will conduct needs assessments and set up a process for ongoing dialogue with stakeholders. Basic information that will be sought from stakeholders in the early parts of the process include recommendations on specific topics for immediate consideration by the working group, an also on the content and format of the information needed by the stakeholders and useful from their perspectives. Stakeholders will also be a source of content experts, and their roles will include technical assistance, review of reports, and involvement in development of informational messages for key target audiences.

This just provides sort of a very simplistic overview of how the process might work, beginning with the working group and a large group of stakeholders. The stakeholders provide input on topics and priorities. The working group makes decisions on their criteria, selects a topic, and requests an evidence-based review. I put the little RTI on there just to remind you that as we go through this, RTI underlies this process in terms of making these things happen. The request will go to an evidence practice center where systematic review will be done that identifies the gaps and the data needed to fill them. Then that information comes back to the working group for appropriate review and comment, and then they will prepare recommendations, reports, and disseminate these materials to the target audiences of consumers, providers, policymakers, and purchasers and payers of health care, with an opportunity again for stakeholder input on the development of these targeted informational messages.

Under certain circumstances, the working group may decide to refer a topic out to other groups for further appraisal, for instance the U.S. Preventive Services Task Force, and they will be involved in developing collaborative projects for pilot data collection with stakeholders.

The first year of EGAPP really begins, obviously, with process development, recruitment of the EGAPP working group, two organizational meetings to follow, and the development of the working protocols by the working group. There will be a methodology conference which I'll come back to in a moment, preliminary needs assessment activities, two small pilot data collection studies, and an evaluation that's based mainly in this first year on process.

In years 2 and 3 we'll see continuing support of the working group, commissioning and oversight of evidence-based reviews, four full reviews in these two years, and three what we're calling fast track reviews. There will be dissemination of reports, working group recommendations, and informational messages. There will also be ongoing dialogue with stakeholders who will be involved in the development of these informational messages for target audiences, as I mentioned, and who will provide feedback on the value of the process and the products.

There will be two pilot data collection studies in each of those years, and there is a comprehensive plan to evaluate the success of the process, the quality and usefulness of the products, and the impact or value of the project overall. At the end I think we really want to be able to consider mechanisms for sustaining

whatever we can validate from this process for evaluating genetic tests.

So why talk about methodology when there are standard and sometimes gold standard methodologies that are being used? Well, it's been pointed out, and I think many people have seen this, that the standard process and methodologies -- for instance, the U.S. Preventive Services Task Force -- may not be as effective when we're dealing with conditions that are uncommon to rare, where interventions and clinical outcomes are not well defined, where the evidence base is limited, and that obviously is going to be the case very often here, and where there are problems with study design or quality of data. We have already noted that ethical, legal, and social issues are less amenable to the evidence-based approach. We need to think about that. We need to consider the influence of advocacy, which I'll talk about in a moment.

So the plan that we're working very hard on right now is to have a methodology meeting in July of 2005 to bring together a relatively small group of U.S. and some international experts in evidence-based review, health technology assessment, epidemiology, genomics, and health economics, particularly those having experience in evaluation of genetic tests, for a working meeting that focuses on elements of evaluation, selecting and defining topics, developing an analytic framework for evidence-based review, literature searches, grading quality of evidence, and translating evidence to recommendations.

We will also address questions about the use of unpublished data sets and the gray literature. We'll consider how to deal with proprietary data. We'll seek information and agreement, in some cases we hope, on minimum standards for determining when a test is ready to move into clinical practice, how much information is enough, what is a reasonable threshold for quality of evidence for genetic tests, is the threshold different for different evaluation components such as analytic validity and clinical validity, and how do we optimize the quality of data to be collected in the future. The results will be used to inform the EGAPP working group deliberations and will be published, we hope.

We feel that the timing is right to use what's been learned and to move forward from ACCE. Certainly the situation is not going to become simpler. More tests are certainly coming, testing will move into primary care, and health care providers and the public need a source of reasonably objective advice about appropriate use of tests. In the short term, whatever is learned will be useful and is likely to provide information to address questions posed by this committee related to oversight of genetic technologies, coverage and reimbursement, access, potentially public awareness and understanding.

In the long term, we hope to create an expectation that a certain level of review will occur prior to acceptance into routine practice. We hope to facilitate standardization of data collection formats. We hope to identify specific gaps that may stimulate research, and we hope that what we learn will support the need for postmarket review of testing practices, clinical guidelines, and recommendations based on new information.

Thank you for your attention, and I'd be happy to answer any questions you might have.

DR. TUCKSON: Any questions?

MS. MASNY: My question is, is your process for review the same process as the methodology meetings that you're going to have, or are they two separate types?

DR. BRADLEY: Yes, that's a good question. We know that the standard methodologies that are being used in many evidence-based review practices are going to be problematic with genetic tests because we have a lack of quality evidence in many cases. So what we're hoping to do is to have this methodology conference in order to look at different ways that we might approach some of these issues. What comes out of that methodology conference or meeting in January will certainly inform the working group's deliberations on how they want to proceed, but it will be separate.

DR. TUCKSON: Have the requests gone out, the letters inviting people to the meeting in January? Do we have a sense of who you're inviting?

DR. BRADLEY: We're certainly working on a list, and I think we're about to make final decisions in the next week or so. It's been a tortuous process trying to decide on who are the right people to invite to the meeting, as you can imagine.

DR. LEONARD: This may sound like I'm self-serving, but it's a little disturbing to me to see in the stakeholders list laboratories. Since you're talking about genetic testing, I liken this to investigators who want to study breast cancer or lung cancer and they have no knowledge of what those tumors are, and so they treat them all the same. You really need an intimate knowledge that breast cancer comes in lobular and adenocarcinoma forms and stuff, what you're actually talking about.

So to see the laboratories that are doing this testing as a stakeholder rather than an active participant in this process informing the group as to the details of the testing and what it means to do it one way versus another way and applications is a little disturbing to me.

DR. BRADLEY: Well, in fact, they will be filling just that role. As I told you, we see a number of roles for stakeholders. It's a very broad group in there, and I appreciate what you're saying. But, in fact, they will have a very important role in this process as experts and will be involved in providing technical assistance to the working group, doing review of reports that come out in their preliminary stages so that the working group can be sure that the evidence is accurate and complete and there aren't issues that they don't understand. I think, very importantly, we're hoping to get guidance from this group about what priority topics are and why, and also I think a very, very important role, especially for laboratorians and people in the genomics and genetics communities, is at the other end of the process. Once you have an evidence-based review, the evidence has been collected and it's been presented, helping to develop appropriate informational messages to go out to these different target audiences.

So I think, in fact, that it's a very big role of stakeholders, and I think obviously laboratorians are going to be a big part of that.

DR. TUCKSON: Thank you very much.

Cindi, I think we can move on.
MS. BERRY: What we'll do now is go through the version of the report that you have seen. The intent is to go through section by section, outline the report as it is currently drafted, discuss the barriers that have been identified, then discuss the recommendations that have been suggested. This discussion, which will

continue into tomorrow, will provide input for additional revisions. We will also solicit further input from members of the public. There will be a notice that goes out so that we can get formal written comments from anyone who would like to comment on the draft report. So it is by no means final. In fact, today is by no means a final discussion of the report. This is just the next phase in this process.

So to start out with the structure of the report, it will give you an overview of how it is laid out. Of course, we have a preface, introduction. These are the basic chapters, I suppose, if you will, and you'll see the background section goes into fairly good detail of how health care is structured in the United States, both public programs as well as the private programs, private insurance. It goes through coverage decisions, payment decisions, how they are handled operationally, and the billing process. Then the final section outlines the barriers to access for genetic technologies, coverage and reimbursement, and then potential recommendations under each section.

We probably don't need to discuss the preface too much, and the introduction as well. It gives some general background on what we're talking about with regard to this report, genetic tests and services. What are genetic tests and services? What is it that we're trying to improve coverage of? It talks about, again, how the health care system is structured and what are the constraints under our current system, and it outlines the purpose, the goal, and the objectives of the report.

This section, of course, as I mentioned, outlines precisely what it is we're talking about when we talk about coverage and reimbursement for genetic tests and services. It describes them in a sort of a general sense. It talks about how they are different from other tests and other health care services, and it explains some of the challenges that they pose to the health care system.

We've got the overview of the U.S. health care financing system, Medicare, Medicaid and SCHIP, the public programs, private insurance programs, a discussion of managed care, and there is a bit of a discussion on the uninsured and the underinsured.

Feel free to stop me as I'm going through this. I intend to go through this part of it pretty quickly. You all have seen the report and we're sort of going through how it's structured. But if there's something that you want to comment on and you feel needs to be addressed in this aspect of the report that I'm just glossing over, just raise your hand and we'll stop.

Coverage decisions. This section of the report --

DR. LEONARD: Cindi?

MS. BERRY: Yes?

DR. LEONARD: Can I ask a question about something that still remains in here and is inconsistent with another draft document that we're working on and we'll discuss tomorrow? We define genetic tests where we're doing the overall thing -- it's not our vision statement but whatever that thing is that Emily did. We defined genetic tests or genetics as inheritable and only inheritable, and genomics is defined as the broader inheritable and acquired. Yet on page 19, when it says "What are Genetic Tests?," we persist in including the acquired testing in the definition of a genetic test. While I agree that would go in the

definition of a genomic test, I still strongly disagree that that goes into the definition of a genetic test and, in fact, is inconsistent with the definition of "genetics" that we have in our other document. So we're not even being internally consistent between the two documents.
MS. BERRY: Suzanne, I'm trying to recall where we pulled that definition from.

MS. GOODWIN: The current definition in the report as it stands was SACGT's definition. So if there is modification that this committee would like to make to that definition, you certainly can do so. If there's discussion amongst the committee members about whether they want to do that, I suppose now may be a good time to do so.

MS. BERRY: I couldn't remember where it came from, but it seemed to me that it was from some prior committee work and that it was some sort of a consensus. I am no expert in this field, so I defer to others in terms of modification to that definition. We should be consistent. We shouldn't have different definitions in different documents that are the work product of the full committee.

DR. LEONARD: It does come from SACGT?

MS. GOODWIN: Right, the oversight report, and there was a lot of deliberation around that at the time. But certainly we want for the different reports to be consistent.

DR. LEONARD: Right, and I could refer you to the other report if you want, or whoever. But there's a clear definition of genetics and genomics. So if we're referring to a genetic test, then I would think that's a test to look at genetic diseases, and that genetic definition does not include acquired. That's in genomics.

MS. GOODWIN: If you look on page 19, there's a definition for genetic and genomic technologies, but there is no definition of a genomic test.

DR. LEONARD: Right.

MS. GOODWIN: So you would like changed, then, the definition of genetic test that --

DR. LEONARD: I would, but I don't know what other committee members think about this. Ed and Reed, you were part of the previous SACGT deliberations that included acquired, but it is concerning that looking at the overview document of our deliberations on how to come up with our priority setting, that we do define genetics without including acquired, and that's included in genomics, which I agree, acquired does fall into genomic testing if you want to define that kind of testing.

DR. McCABE: I'm trying to read this through quickly. I see the sidebar on 19. Is that what you're referring to?

DR. LEONARD: Yes. It's the first sentence. It says, "A genetic test analysis performed on DNA/RNA genes and/or chromosomes to detect heritable or acquired genotype mutations, phenotypes or carrier types." It's the "or acquired" that bothers me, and I can refer you to the other document.

DR. McCABE: Okay. So I think if you then look down to what are genetic/genomic technologies, I think that's where it talks about acquired there. So it looks like if we went back to the vision report and brought page 2 of a roadmap for the integration of genetics and genomics into health and society -- is that what you're referring to? Yes. So I think one could go back to those definitions.

DR. TUCKSON: The road map is in Tab 3, and we're talking about page 2.

DR. McCABE: I think it's important for us to be internally consistent, and I don't see any need for us to be consistent with the work of a previous committee, recognizing that some years have elapsed since that. I think the SACGT definition was taken actually from an earlier committee. So there's been quite a bit of time between when that was crafted. So I would suggest that we just bring it into consistency with the road map. That would seem to make sense.

DR. TUCKSON: Is everybody on the same page? So bottom line is we're talking about trying to take the definition in the reimbursement document and substitute that one with the box on page 2 so that we're consistent in terms of how we do this.

MS. CARR: I just wanted to point out that when SACGT was deliberating, perhaps it was an oversight on our part, but the issue about the term "genomics" didn't really -- we didn't have deep deliberations about that the way we did here in the last meeting. So we weren't trying to make that distinction. That's what the definition in the road map report does, tries to clarify what we mean by both those terms and that they're both important. But if we were going to use that as the basis for what we have in the coverage and reimbursement report, it still seems like we need to do a little bit of work to translate that concept, those concepts into what a test is, or what a genetic test is and what a genomic test is. Is that what you're saying, Debra?

It might be helpful to have, maybe not right now, but you or Emily or others and Ed involved in actually helping us do that, because I'm not sure it's a straightforward translation.

DR. McCABE: Right. As I look at it, the definition on page 2 is really a definition of genetics and genomics. It doesn't translate into the testing. I think if you actually look at what are genetic/genomic technologies, that would probably be acceptable to translate that for testing, for genomic testing, and just separate them out. I think it could be done by a small group.

DR. TUCKSON: Great, and I think maybe what we might also do is, one of the challenges and jobs that we have -- and Sarah, I want to make sure here -- is to educate the public. So we ought to take every opportunity to remind people about the fundamental blocking and tackling terms. So I think what we should do perhaps, or consider as a small group, is state the definition first in the vision statement and then say what this means in terms of genetic tests and technologies, so you can see the logical progression of the ideas. But I think we've got to take every chance we can to educate.

DR. LEONARD: I know this sounds like a small point, but in the mindset it's not, because genetics, where it is inheritable, has different ethical, legal, and social implications than acquired mutations. While they're done using the same technologies in the laboratory, the surrounding issues are quite different.

MR. MARGUS: But Debra, for coverage in this case, is the intent to include genomic somatic changes and all that, or is the intent to focus only on germ line inherited stuff?

DR. LEONARD: To include.

MR. MARGUS: To include everything.

DR. LEONARD: Yes. It's just that it's confusing to have a definition of a genetic test where we define genetics elsewhere --

MR. MARGUS: We just need to make it consistent, but (inaudible) include it all.

DR. LEONARD: Right. I definitely want genomics included.

DR. TUCKSON: So we'll have Ed and Debra work on this with you, Suzanne. So Ed and Debra will work on this. Thank you for doing that.

MS. BERRY: The next section of the report focuses on how coverage decisions are made in both the public sector and the private sector. It outlines the Medicare coverage decisionmaking process, national coverage decisions as well as local coverage decisions. It reviews in general how these decisions are made in the private sector, and then reviews the state of play in terms of coverage for genetic tests and services in both the public and private sector and the role of economic evaluations.

Then we move on to payment decisions, the clinical laboratory fee schedule, and the existing payment rates for genetic tests and services, and then of course the billing process, how are codes developed, how are these tests and services coded, what are the billing practices in Medicare as well as in the private sector.

DR. LEONARD: So I will provide those CPT codes, but a question. Do you want them as genetic CPT codes, or there are genetic ones and genomic? The same CPT codes cover both.

DR. McCABE: Well, I would think if we're going to be inclusive, we need to clarify that so that those reading it will understand.

MS. BERRY: Here's where we get to the meat, barriers and recommendations. The report outlines specific barriers to coverage and reimbursement in the Medicare program and other public programs, as well as in the private sector. Then in the blue boxes in the report -- and if you have simply a xerox copy, I suppose it's gray. But anyway, in the boxes is where we outline some potential recommendations for the committee to consider to address each of the barriers or most of the barriers that have been identified.

Now, how we propose going through this is really a systematic analysis of the report in that part of the report called "Barriers and Recommendations." We want to focus on that section, pages 49 to 73 of the report, and go through each one in a systematic fashion, and then discuss whether we should make a recommendation, first of all. If we should, if there is a recommendation outlined in the blue/gray box, whether it's something that the committee would like to put forward, and are there alternative

recommendations that the committee feels we should include in the report.

Of course, our objective here is to reach a consensus. If there is no consensus or we can't come up with a recommendation to address a particular barrier, then it won't go in the report. Our idea, though, is to, at the end of the day or the end of tomorrow, produce something concrete that can be utilized by the Secretary and by others as a helpful guide to improve policy in this area.

DR. LEONARD: Cindi, can I ask a question?

MS. BERRY: Yes, Debra.

DR. LEONARD: Unfortunately, I just took a bite of brownie. The question is, is this going to go, after we make our recommendations, directly to the Secretary, or is this something that goes out for public comment first?

MS. BERRY: Public comment.

DR. LEONARD: Oh, great. Okay. I missed that step.

MS. BERRY: Turning to Medicare, one of the first things that was identified, and we talked about it at our last meeting, and it's in the report here, is the screening exclusion in Medicare. CMS policy is such that tests that are performed in the absence of signs, symptoms and complaints or personal history are not covered unless explicitly authorized by statute. So a predictive test or presymptomatic genetic test is not covered by Medicare because of this policy. Preventive services that have been covered by Medicare have been specifically authorized by Congress, and we don't have that here. So this exclusion is one of the critical barriers to coverage of genetic tests and services in the Medicare program.

DR. WINN-DEEN: I think it's important for us to say major barrier for some genetic tests, because this paragraph the way it's written, it sort of implies that all genetic tests are screening tests and that this is a barrier for everything. This is a barrier for just a subset of genetic tests.

MS. BERRY: That's right.

DR. WINN-DEEN: Carrier screening, presymptomatic screening, those kinds of things. So I think we should just make that clear in the opening sentence, particularly since, for whatever reason, this, which I think is a subset of all genetic tests, is the first thing that appears. I might suggest that we put the order in a little bit different way so we deal with the things that affect all genetic tests first and then some of these subset things later just in terms of organization of this section.

DR. LEONARD: Especially since in the Medicare community it's less likely to be a presymptomatic test.

DR. WINN-DEEN: So from the point of view of presymptomatic screening, and probably even the majority of carrier screening, Medicare is not the carrier that's going to be paying for that. So this becomes one of those minor extra points instead of a major point, and I think we should start with major points.

MS. BERRY: Ed?

DR. McCABE: Just before we leave that, I'd also point out on page 61 is preventive nature of genetic services. It would just seem like as we're reshuffling and reorganizing, this is a nice lead-in to why screening is important and why it's important to eliminate the screening exclusion, and there is no recommendation that goes with that, so it would fit nicely together.

MS. GOODWIN: The way this chapter is organized is the Medicare-specific barriers are listed first, and then the preventive nature of genetic services section is listed under a barrier that's applicable to both public and private insurers. So in combining the two, there would need to be some reorganization of each section. So do you have any suggestions on how best to do that? We tried to keep separate the barriers that were specific to Medicare and the ones that were applicable across the board.

DR. WINN-DEEN: Again, you might want to do it the other way around, the ones that are done across the board first and then do the Medicare-specific things, because Medicare is a subset of all insurance.

MS. BERRY: I think probably the reason it was initially done this way, and it certainly isn't critical that we leave it in this order at all, but I think, if I recall correctly, that the thinking was that Medicare oftentimes is the model that many in the private sector follow. So how Medicare goes, the private sector goes as well, but not always, because I think as we've discovered, in many cases the private insurers are taking more of a lead and are being a little bit more progressive in the area of genetics and genetic tests and services. But I have no objection at all, unless anyone else does, to reordering it so that we do the barriers that are applicable to both public and private sector first and then do the Medicare section after that. I think that was really the only reason that it was done the way it is.

DR. WINN-DEEN: And I think in part it's because of data on what's going on in Medicare is easily and publicly available, and what's actually going on in private insurance is not always as easy to get at, exactly what they're reimbursing, exactly how they're making their decisions, since they're not obligated to be public about that.

MS. BERRY: Right. Does anyone have any objection to changing the order?

(No response.)

MS. BERRY: We'll do it. We'll still go through the report the way it's currently configured, but we'll change the order in the next draft.

The screening exclusion barrier, the recommendations that are outlined, there is sort of an order to this. CMS had informed us that number 4, which is for them to unilaterally change their interpretation of the law to allow them to cover screening services without any legislative action, is very unlikely to occur and is not particularly feasible. I think some of the folks at CMS thought number 1, which would be for Congress to change the law, would be a better approach. Having said that, we're all aware of how difficult it is to get Congress to change the Medicare statute by adding a benefit category for preventive services.

We don't want to give the impression that we think that's just a cake walk and we'll just put that as number 1 and everybody likes that and let's just be done with it, because that has its own challenges as well. These four recommendations are not necessarily mutually exclusive in any way. It's just that the original order was what might be the most effective recommendation, not necessarily the easiest or the most feasible.

I don't know if anyone has any suggestions, additional recommendations they want to put up for consideration before we go through each one.

DR. McCABE: I'm sorry. I don't have any additional ones, but I think, especially given the visibility that family history is going to have, and perhaps it's in here, but we need to make sure we take the information that Alan Guttmacher presented, which represents a coalition among the HHS agencies, and really play that up since that's something that the Secretary is going to be quite aware of. I would just fill that in here with the website and all of that information.

I think number 1 is going to require a culture shift in American medicine, really, and culture is always hard to change, but it's essential that this change. So while it may not happen overnight, perhaps at least we can begin hammering that we need to move from acute intervention to prevention.

DR. LEONARD: How difficult is number 2 to do, though, in the context of this family history tool that's being rolled out? It would be a nice thing to coordinate that family history that we're emphasizing constitutes medical justification for a test being done. So how does 2 happen? Can somebody inform us?

MR. KAY: Hi. This is Terrence Kay. I'm from CMS. Actually for number 2, we have a fairly straightforward process. We have it on our webpage of how anyone, the committee or anyone can ask for a national coverage decision and what kind of information we would look for. So process-wise, I think it's fairly straightforward.

I think the major issue is going to be -- in general, when I've listened to the discussions today, whether the issues are legislative or regulatory, I think for communication purposes -- for example, I heard today that there are over 1,000 genetic tests. So I would think -- I obviously couldn't speak for Congress, but I would think both Congress and the agency would be thinking, gee, what really is going on here? What are we walking into? It almost seems like, if you look at the history of preventive services as they've gotten added to the Medicare benefit, whether it's mammograms or colon cancer screening or whatever, that there was a case made, a specific case made for those services, and in a way that everyone could understand.

For right now for genetic tests, just sort of listening to the conversation and reviewing the material, I think a lot of folks, frankly, would be like me. I'm not a clinician, but it would be sort of hard to understand the implications. Earlier today we were talking about a thousand genetic tests. Would you want to cover all of those? Which ones are really most beneficial for Medicare? It's a long answer to your question.

The process to do number 2 is straightforward, but I think the comment that somebody made here a little while ago that CMS had indicated that it might be unlikely that we would do number 2, all I would say is

that we take the committee very seriously and we're very interested in your recommendations, and we have tried to attend all the meetings. I also would put in a plug, a lot of appreciation for the involvement of the staff for this committee and how cooperative and responsive they've been to comments we've made that have been reflected in the draft report. I think it's been a very productive relationship, and we understand a lot more than when we started. Likewise, you've gained appreciation for that.

We can do number 2. We'll certainly give it serious consideration. But we're very much looking for an evidence basis for decision-making on national coverage decisions, and I think a fair amount of thought would need to be given to how best to craft the argument so that the agency would be persuaded to make a change like that.

DR. LEONARD: So if I'm understanding you correctly, I don't understand how national coverage decisions are made, and I haven't read the website. But you're implying that someone has to bring this to you as a proposal? It's not something CMS does actively themselves. And who would that somebody be who would bring this national coverage decision proposal to whatever committee it is that would say yes or no?

MR. KAY: Again, because you're the Secretary's committee, I would have to defer to whatever your process is. In general, our process is that anyone can ask for a national coverage decision. At the agency, we have our Office of Clinical Standards and Quality. We have a Coverage Analysis Group. Dr. Steve Phurrough, who has attended a number of the meetings in the past, heads up that group. We'd be happy to provide further details on exactly how one would do this, but basically if you look at some of the examples, and they're wide-ranging, basically folks come and provide the specific request and why they think we should make the request and what data/studies are available to support that request.

DR. KHOURY: Just some quick comments about family history. Two years ago we had an expert panel at CDC to talk about family history as a genetic test, and at that time the thinking was, and it sort of propelled us in the next phase, that if you approach family history like a genetic test that deserves its own evaluation for its utility and whether or not there is a genetic test that goes along with it -- in other words, how good is it analytically, sort of the ACCE paradigm -- it's very obvious that family history per se is sort of a mixed bag. When you take family history, even if it's reliable, sometimes it leads you to genetic testing, sometimes it doesn't. Just to elaborate on the evidence-based process, I think family history deserves its own evidence-based process.

Now, we all think that family history is good, that we need to collect it, but in terms of whether or not it will be associated with genetic testing, it's not always obvious. I mean, a simple case in point would be BRCA1 testing. There are a lot more women that have a family history of breast cancer, maybe 10 to 1, as to the small fraction of women that are in the range of being even considered for BRCA1 testing.

So I think we can use family history as a launching pad for coverage decisions, but I guess following the same rules of engagement for genetic tests. I'm not suggesting any change to the wording here, but at least the good thing is that you put family history as somewhat up there for the decisionmaking process.

MS. BERRY: I should point out as well that the first recommendation talking about legislative change, we had discussed a bill, the Medicare Preventive Services Coverage Act, S. 2535, H.R. 4898. That

legislation would add preventive services as a Medicare benefit category and enable CMS to determine through its national coverage decisionmaking process and after assessing the evidence so that they could determine whether an item or service is reasonable or necessary. So even with the congressional authorization option, there still would have to be an assessment of the evidence in order to lead to a national coverage decision.

So in many respects, 1 and 2 are closely related. In fact, I don't think you could -- number 1 isn't going to do you any good at all if there is no evidence to support coverage, so they're closely aligned. The question is whether we can get to number 2 on its own in the absence of any congressional activity.

MS. HARRISON: I was just going to say that number 3 makes me cringe a little bit. We haven't talked about it very much, but I just didn't know if we were definitely talking about 2, and my feelings were going toward 2 and a combination of 1, if we can do that. But just to kind of put out there that these presymptomatic and predictive tests are diagnostic for the mutation that may be found but not diagnostic of a disease per se, and the limited genetic literacy sometimes that we're dealing with out there, just to be wary of that. So as I kind of get our conversations off of 3 if we ever do go there, to just be careful about that.

DR. WINN-DEEN: So I think in response to Terrence's comment that one thing that we might think about is really what tests do we know about today that we would want to see Medicare apply this to, and I'm having a hard time really thinking about anything where you would screen someone at a genetics level where you would wait until they were 65 to do that. So I think we need to think as a committee about are there some specific examples that we might put forward instead of a more broad strategy? Is this really just a few smaller things that we should put forward specifically? It's different than the concept of cholesterol screening or mammograms, where it's something that you test for every year and you continue to be tested as you age. It's really only a question of what age do you start screening.

Genetics is a once in a lifetime test, and the question I would ask is, under Medicare, how often would we want to be doing this once in a lifetime test in that population? Having participated in this program, I feel sort of stupid bringing that up at this point in time, but it helps I think to be specific, and then you can work to a specific example of what you'd like to see and why you'd like to see it.

MS. MASNY: Just as a follow-up to your comments, Emily, I know that in what Terrence had mentioned about the case by case basis for looking at some of these screening tests, that the testing for breast and ovarian cancer is now approved by Medicare, and they approve it under a specific code that states hereditary breast/ovarian cancer. So maybe in view of looking at the use of the family history as a tool, one of the things that we do in a cancer setting is actually give a diagnosis to the family history as to whether it looks sporadic, which would be more the acquired mutations, something familial that we're not really sure of, we're seeing something going on in the family or something that truly fits a hereditary pattern where you're actually then providing a specific, sort of almost diagnostic category to that, sort of giving credence to why someone would go on then for a screening test, and that even in many of the situations when Medicare first approved the coverage for the testing for breast and ovarian cancer, the woman had to have had breast cancer. So she already had the disease, but then it was actually to find whether a gene was present, most probably to help other members of the family.

DR. WINN-DEEN: That's not screening. That's in the presence of signs and symptoms, which would already be normally covered.

MS. MASNY: They did expand it now to cover people without the disease. So that would be screening.

MS. BERRY: This would be a small percentage of the population, but Medicare is also for the disabled. If they're on Medicare because of disability, they obviously have signs and symptoms of some problem, but theoretically I suppose there could be a screening component where they have other problems going on. But then that's probably a very small percentage of the population that we're dealing with.

MS. HARRISON: I was just saying that we even here had an example of that with Mr. Hardt, who had hemophilia and then also has a risk for Huntington's disease. It can happen. You may have people who are on Medicaid who need a screening test for something that may be unrelated, but they have a positive family history.

MS. BERRY: Barbara had raised some angst about number 3. Does anyone want to comment on that? Do they share her concerns about that? Does someone feel strongly that we need to keep number 3 on the table as a possible recommendation? For those who maybe can't see it there in the back, number 3 is for CMS to redefine predisposition and predictive genetic tests as diagnostic laboratory tests through a rulemaking process or a national coverage decision. So in the presence of a strong family history of disease, these tests would be considered diagnostic and not subject to the screening exclusion.

MR. MARGUS: Can you just clarify what we're trying to do here? From a very, very, very big picture, we're trying to get coverage for tests that sometimes don't get covered, right? So we have four shots at it. You told us that number 4 is a really, really, pretty much long shot. Number 1 is certainly not trivial. That leaves us with 2 and 3, and 2 says that you make tests that are, as long as there's family history involved, reasonable and necessary, and I take it that makes it qualified to be reimbursed. And 3, instead of being reasonable and necessary, that makes it the same as a diagnostic test, in which case it gets covered.

But do you have to pick one of these, or can you give them both 2 and 3, ask them to pick one, and it gives us multiple shots on goal?

MS. BERRY: Correct me if I'm wrong, but my impression is that these are not mutually exclusive. There may be some on here that people just don't want to pursue and don't feel as a committee that we should put that forward as a recommendation to the Secretary. On the other hand, if we think, hey, why not, all of these seem viable and it's, of course, possible, perhaps even likely that the Secretary would reject, for example, number 4 and just say, look, that's just not feasible, that's his decision to make and we are at least offering that up as something to consider.

MR. MARGUS: Did the task force or the staff see any negatives to any of these four? I mean, we're just hoping one of those could come to pass, right? Is there any negative?

MS. GOODWIN: If you turn to page 53, we tried to list some of the limitations of each one. Certainly, there's a point to that for the first one in terms of the legislative process, which isn't always the simplest.

In terms of doing a national coverage decision, my understanding of that process is it's generally done on a technology by technology basis. So doing an across the board change or making a change to the family history definition isn't something that's generally done. Certainly correct me if I'm wrong, Terrence, on that, but it's probably something that the Medicare Coverage Advisory Committee doesn't typically proceed in that manner. It's usually done on a case by case basis with different technologies.

But maybe you can clarify whether 2 and 3 are approaches that could be taken even though they are not your typical process.

MR. KAY: I guess my comment would be that I could see it would be within the realm of possibilities. It's clearly not typical. Believe me, the agency is very interested in making sure that our beneficiaries have access to proper medical care and new technology and all that kind of thing, so clearly I would want to be in a position to rule in or out anything that you would recommend. But again, just sort of reemphasizing what I said earlier, I've been at the agency since the mid-1980s, so I've had the advantage of kind of seeing -- I mean, Medicare is usually described as not having a lot of preventive services, but it turns out that if you take a look at all the preventive services that the U.S. Preventive Services Task Force has recommended, I think Medicare has just about gotten to the point where we cover just about every one of them.

So we're very interested in making sure beneficiaries have proper care. Congress has clearly made changes, and it just seems strategy-wise, whether you're going to make recommendations for congressional change or for the agency to make a change, still folks need a comfort level of what really would it mean to make a reg change like that, because at the agency we get the Department's clearance, of course, we have the Office of Management and Budget to deal with, and in all of these clearances we would need specifics about what would likely happen if we would do this change.

That's why I'm not saying that you should only focus on individual services, but if you want to go broader, I think it still is helpful to identify some list of specific examples to show there is a big problem here, Medicare doesn't cover these services as an example, and why care would be better if Medicare covered them. Then I think it's easier to have the more theoretical conversation about some of these things. But I think regardless of whether you go general or specific, it's helpful to have a few crisp examples of why there's a problem in Medicare and what changes should be made to improve the program.

MS. BERRY: And that gets to the point that Emily had raised earlier, which is is the committee aware of specific tests that are needed by the Medicare population that are not currently covered because of the screening exclusion? And if there are, should we perhaps identify them in this report and list them as partial justification for this change in policy? Because you're right, Terrence, I can't imagine a federal agency would just up and change their policy without any real demonstration of need based on concrete practice.

MS. GOODWIN: Also, another important consideration, actually for all of them. Here you're actually talking about changing how CMS deals with preventive services in general, and certainly that's one aspect of a preventive service. But from CMS' perspective, you also have to take into consideration that in making this change for genetic tests, you're essentially opening a can of worms for other tests that might

not necessarily be genetic tests but that would fall under the rubric of a preventive service.

MR. MARGUS: Would pharmacogenomic tests be considered preventive? Because that would be one you might foresee an elderly population participating in a lot more than disease screening. So if you had tests that predicted whether someone was going to have an adverse reaction to a drug or have efficacy, would that be a test that would also fall in this category of maybe not being covered?

MR. KAY: That type of service, as you describe it to me, sounds like the kind of service that Medicare could potentially cover, and we probably already do. I think we'll probably get increasingly more attention on that kind of an issue, because in 2006 Medicare will have the prescription drug benefit. So to date, we've had a limited list of drugs we cover, but as you describe it, I don't see why those kind of services would not already be potentially coverable.
MS. BERRY: There was someone in the audience. Yes, sir. Over there; does that microphone work?

DR. ROLLINS: My name is Jim Rollins, and I'm a medical officer for CMS. Some of the questions that you've been asking, Terrence has given you a lot of information that I basically want to reiterate.

One of the questions that was just asked a minute ago about pharmacogenetics, that is probably something that would be covered because it's considered diagnostic as opposed to a screening test. If a person already has the illness and tests are being done to determine whether or not an appropriate medication level or medication is being applied, yes, that would fall under what we would consider as a diagnostic test. So that's something that would be covered.

In terms of the four recommendations, 1 and 4 would be very difficult for the agency to pursue simply because of the current mandates which are in place. As far as number 2 goes, a national coverage decision, that is something that can be initiated by an individual, an organization, a group, and we would take into consideration the evidence that's in the literature to determine whether or not there is sufficient evidence to support its use. So that would be an option to try to address this particular issue.

MR. MARGUS: Am I right in detecting that your message to us is that you'd prefer to have it case by case or test by test rather than this is being so broad stroke that it's likely to be too scary to them?

DR. ROLLINS: I think that it would have to be on an individual case per case basis, just like we look at all technologies, the same way.

DR. McCABE: I'd just comment, though, that if there are a thousand tests and we had to do this test by test, I think part of the concern of the committee is that it could take a decade or two to work through. So I think part of the goal was to try and get, while more difficult, a broader discussion of these issues and see if more than one test at a time could be rolled into this. Is that not possible?

DR. ROLLINS: It's possible. I would also hope that in submitting those tests, perhaps the frequency or the prevalence of the particular disease within a population might be something that might be used as a means of determining which ones you may want to pursue first.

MS. GOODWIN: I had a follow-up question regarding the pharmacogenetic tests. I understand that

CMS is currently considering developing some sort of guidance document specific to pharmacogenetic testing, and I wonder if you could elaborate on that.

MR. KAY: We had an open-door meeting recently on a requirement in the Medicare Modernization Act to provide guidance documents, and we asked for public comment and suggestions. That was maybe two or three weeks ago. So we're still sort of in the process of evaluating our comments and we have not decided on specific documents. Clearly this is an example of the kind of guidance we might provide in the future depending on the review of our public comments.

MS. GOODWIN: But could you articulate what would be the need for that document? Would it simply clarify what pharmacogenetic tests would and would not be covered under current CMS statutes and policies?

MR. KAY: Right. Our guidance documents basically would be to provide the information to the public about our current coverage and coverage processes. It would not be the mechanism to make refinements.

DR. LEONARD: I'd like to go back to the point that Barbara raised. Having had Barbara make that statement about being concerned about redefining predisposition of predictive genetic tests as diagnostic laboratory tests, I'm worried about number 3 because when you're doing predisposition predictive, it is not diagnostic because diagnostic by definition means there are symptoms present, and I don't want to create worries for the patient, and confusion. So I don't know if there's a different way to word that other than redefining them as diagnostic laboratory tests. Maybe you could say redefining them as I don't know what, but that worries me. And if 2 is doing the same thing, maybe we can just get rid of 3, because they're both basically asking for --

MS. BERRY: I think 3 is just a creative way to get around statutory language. I mean, it's wordsmithing and creative argument. I think it's nothing more than that, and I agree because it certainly could open up doors that we don't want opened. If we can accomplish the same goal with recommendations, we might want to go that route. Unless anyone feels very wedded to number 3, it sounds like there's some concern about that approach, and perhaps we should focus our attention on one of the other recommendations, or a few of the other recommendations.

Does anyone have any strong support and want to argue in favor of 3?

(No response.)

MS. BERRY: Let's get rid of it. Nuke it. Any other suggestions or comments?

DR. WINN-DEEN: Can I just ask on the asymptomatic testing for BRCA1 and 2, was that national coverage decision that was basically brought up and put through on a national coverage decision basis, is that the way that got to be reimbursable?

MR. KAY: Unless Dr. Rollins happens to know, I would have to look it up. Historically, what's happened is that our current coverage process has evolved, frankly, over the last five years, and without knowing the exact timing of each coverage decision, I don't know what process was followed.

DR. ROLLINS: I also can't give you specifics on that, but what I can say is I think that that was one of the preventive tests which was mandated, and for that reason it was covered. In terms of having a national coverage decision for that, I don't think we have one for that. I'm not sure.

DR. WINN-DEEN: So you think it may have gotten through on the breast cancer congressional mandate where mammograms got through as reimbursable?

MS. GOODWIN: I've actually looked at this recently. There is no national coverage decision for BRCA1 and 2 screening. There are, I believe, a couple of local coverage decisions regarding that, but there may be just one or two, and those are limited to the specific geographic areas that those local carriers cover.

MR. KAY: Unfortunately, again, without looking it up in our coverage book, which we can do and get you that for tomorrow morning if you'd like. No problem. But I just don't offhand recall the history or exactly what the policy was.

DR. WINN-DEEN: I'm just trying to think about, as we go for recommendations here, if there's any test that we would put in this category that has successfully become reimbursable and covered under Medicare, then it would be helpful to know how that process happened and if it's something we should recommend as a replication process, or if there's nothing that's ever been approved, then we have to consider everything from a de novo point of view.

MR. KAY: I was going to say I'd be happy to take a minute now to try to make some calls to the agency, if that would help the committee, just so you have those facts. I'll be happy to do that now.

MS. BERRY: Before you go, can I ask you a question? Number 4, just in the process of elimination here, would this cause a great angst at CMS if we included a recommendation such as this in this report knowing that it's a very unlikely option to occur? Is there a real downside to including it in there, or is CMS recommending that it's so unlikely and worse than unlikely that it just shouldn't be included in the report? Is there a downside to that?

MR. KAY: I guess, again, as sort of CMS staff, we're not really wanting to rule anything in or out. If the committee wants to go in a certain direction, we'll give it a look. I'd say that the advice you've received in the past, it would be a tough hurdle to get the agency to agree to number 4, I would agree. That would be tough to do. 1862, the medical necessity portion of the law, is just a major issue for Medicare in determining what it does or doesn't cover. I think it could be very difficult to craft a policy change in some way that just didn't have wide-ranging implications for just about anything else. As you can imagine, at the agency we get a lot of requests for a lot of services, and that portion of the law is one of the major defining elements of what Medicare does or doesn't cover.

DR. TUCKSON: Just remember also, Terrence, that anything that we send you will come through the Secretary, not straight from us. So you've got a little immunization, if that's what you're worried about. You don't have to craft a deal for your agency right here on the spot.

DR. McCABE: I just wanted to comment that if there has not been a national coverage decision on BRCA1 and BRCA2, I think that illustrates some of the problems that we face. The Preventive Services Task Force is a very good mechanism, but it's also quite conservative in what it considers evidence. So I think that's sort of an example and point of what the problems are. We heard this morning from individuals who have utilized this test or are afraid to utilize this test in their family. It's available, it's recognized to be beneficial to the health of individuals. So I think that would be one that I'd be very concerned if it hasn't had a national coverage decision.

MS. BERRY: Does anyone have a proposal in terms of what we would like to put forward in the report? I'm sorry, Ellen. Did you have a comment?

DR. FOX: Just on page 34 of the report, it says, "Of the approximately 274 national coverage decisions issued by CMS, only one relates to genetic tests and services, cytogenetic analyses for monitoring acute leukemia, myelodysplasia, and congenital abnormalities." That seems to answer the question about what national coverage decisions have been.

MS. GOODWIN: And just as an addition to that, that's not a screening test that's done in symptomatic individuals.

DR. FOX: Right, so it should be genomic, I guess, instead of genetic.

DR. CHESLEY: An observation?

MS. BERRY: Yes.

DR. CHESLEY: Just in follow-up to the comment about the U.S. Preventive Services Task Force, an observation. That group also considers the strength of the evidence, as well as the strength of recommendations that it makes. I would encourage, as the group moves forward with considering these four recommendations, to underpin them as they move forward with the strength of evidence that would support them. It might allow the group to think about levels of recommendation. If you feel strongly, for example, amongst the four in terms of how you would position them as you move them forward towards the Secretary, you might use the evidence that would underpin, for example, recommendation 2 or 3 in order to more solidify the strength of that recommendation that you move forward to the Secretary. I think, from the perspective of a federal agency, that's going to be a critical component of making the case.

DR. TUCKSON: In that regard, Muin, if you could update us just real quickly on the family history deal. Apparently, I think I'm somehow aware that CDC is doing a review of the validity of family history as a clinical tool. Given that we've already a couple of times touched on that, can we find out where it is now and whether we can be updated on that progress as well?

DR. KHOURY: Sure. Actually, two years ago -- I tried to update the group earlier, but I guess I failed to do that properly. Two years ago, we convened an expert panel to take a look at family history as a tool for disease prevention. It became very quickly obvious to the group that family history, while it's good and great and it should be in everybody's medical record, and it's the ultimate genomic test, that its validity and utility for most prevention efforts have not really been evaluated.

So we embarked on a process to evaluate family history utility and validity for six common chronic diseases, three cancers, breast, ovarian and colorectal cancer, diabetes, heart disease, and stroke. We are actually in the midst of funding a controlled clinical trial as we speak that hopefully will have some results two years from now.

Now, the discussions around family history here, especially vis-a-vis genetic testing, should underscore that these two things, while they seem to be independent from each other, they both should follow evidence-based processes. I think family history may be the easiest of the two because we all have it, we just have to remember it somehow and move forward with it. But in terms of making recommendations for reimbursement, I think the underscoring principle here is evidence-based guidelines.

To echo what Francis just said earlier, the U.S. Preventive Services Task Force, as a matter of fact, is taking on BRCA1, which they never took over something like that before. They're struggling with it because the evidence base is not in the traditional strength of something that can be brought to the U.S. Preventive Services Task Force. In a way, that's one of the impetus for the EGAPP project, because the EGAPP initiative takes into account all available evidence as a first look, as Linda Bradley suggested to us earlier, and then having sort of a pre-U.S. Preventive Services Task Force recommendations, if you will, that would put these tests and practices in play while further data are being collected and further gaps are being plugged.

Inevitably, we have great knowledge gaps in both family history and genetic tests. So, in other words, if you ask today the question how many genetic tests fulfill the rigorous clinical utility look from a U.S. Preventive Services Task Force, I would say that number is between zero and minus 1, or maybe plus 1 or plus 2. There is really nothing that comes to mind that meets those rigorous criteria of the U.S. Preventive Services Task Force.

That's obviously not good enough, not good enough for the consumers, not good enough for the researchers, not good enough for access, and what we need is a process that can do this ongoing evaluation while data are continuously collected so that gaps are plugged, and we're doing that collaboratively with all the agencies both for family history and genetic tests, and occasionally for both. For example, with BRCA1, you can't do it independently of family history, and for most of the single-gene disorders, family history goes hand in hand with genetic tests.

MS. BERRY: Should we maybe consider amending 1 and 2 to bring home the point about evidence base? Because I don't think anyone is suggesting that we'll just add a benefit category for preventive services, boom, things are automatically covered, and the same thing with number 2, have an NCD family history, boom, it's justified, it's covered. I think what's implied there, what we probably need to just affirmatively state, is when the evidence supports coverage, CMS would have the flexibility, which they may not feel they currently have because of the screening exclusion, to cover these tests and services. I don't know if that does the trick or not.

DR. TUCKSON: By the way, as you all consider that, what we want to do just from a process check is let's resolve this issue, bring this little piece to closure, and then we haven't forgotten about the break. Then we'll take a break and do the rest. But I just wanted to give you that as an incentive to drive this

point to closure, nail it right now.

DR. LEONARD: I would think we would be sufficient in recommending a combination of 1 and 2 based on evidence. I'm uncomfortable recommending 4. It would be kind of like somebody recommending to this committee that we go talk to Congress. You know, it's just not something that's in our purview to do. So I feel with 4, we're making a recommendation for CMS to do something that really isn't part of their ability to do. So why do it? It makes our recommendations weaker. Whereas 1 and 2, a combination of those based on evidence that's out there, and that comes back to Emily's point, I'm not quite sure what's out there, and I don't know that we want to go making a blanket recommendation that all 30,000 genes worth of genetic tests be presymptomatically covered.

So I would say combining 1 and 2 with the evidence base into maybe even a single recommendation, because I don't think there's a lot of urgency with this. I think if maybe there are genetic tests in the future that predict late-onset Alzheimer's disease and late-onset osteoporosis and things that are degenerative in older people so that you could then prevent those, it might be useful, but right now I'm struggling to come up with specific tests that I would urgently want covered right now in the Medicare population.

DR. CHESLEY: I would second that comment, and given the work that's gone into this document to date, I think it would be worth the advisory group's effort to investigate an example or a case in the Medicare population in which this would be relevant. It would certainly make the case for CMS to then do the work that you're asking CMS to do.

DR. ROLLINS: I'm not against recommendation number 2, but I think earlier someone said that the U.S. Preventive Task Force would be the underpinning of the recommendation. I have some concerns about that only because a number of their recommendations are consensus based as opposed to evidence based.

DR. CHESLEY: Just a point of fact. They're evidence-based recommendations, but what I was trying to say is rather than use the U.S. Preventive Services Task Force, simply that this group consider the strength of the evidence that supports any recommendation that they would make, not using the U.S. Preventive Services Task Force rubric, although you could use it. The beauty of that rubric, though, is that it allows you to make a recommendation in the absence of evidence, which is different than making a recommendation when there's evidence to the contrary. So it allows you to walk through a grid in terms of the amount of evidence that exists as you try to get to a recommendation such as this.

DR. ROLLINS: I'm in agreement. I think currently the methodology which was explained in the previous lecture is something that we could use in terms of some evidence-based model for making these recommendations.

MS. BERRY: I might suggest, to go along with what Debra has offered up, actually I don't think we necessarily have to merge 1 and 2. We could leave them separate and discrete but amend number 1 to track what the legislation that has been introduced last session of Congress would do, because it does essentially merge those two things. It sets up a preventive services category but then also enables CMS to move forward with an NCD and assess the evidence and make a coverage determination.

So number 1 is a legislative fix, but it also would incorporate in that authority for CMS and weighing the

evidence and whatnot. So I think number 1 amended could be that merger that you talked about, and number 2 could just be a freestanding thing that, if they choose to do so, and should we upon further reflection and after getting testimony and other public input find that there are specific examples that urgently require CMS' action, number 2 would be a separate and discrete option that could be implemented in the absence of congressional authorization.

So I'll put forward, just for the sake of nailing this down and so Reed won't get mad at me for prolonging this, amending 1 to flesh out a little bit more what the legislation would do, and making sure that evidence-based decisionmaking is part of that, that we're not just automatically proposing automatic coverage. Then number 2, the same thing, making sure that any coverage decision is based on an analysis or an assessment of the evidence clearly supporting coverage in that circumstance, and getting rid of number 3 and number 4 and just having amended versions of 1 and 2 be part of our recommendation.

Any opposed? Suggestion? In favor? All those in favor, say aye.

(Chorus of ayes.)

MS. BERRY: No?

(No response.)

MS. BERRY: Okay, 1 and 2.

DR. TUCKSON: Great. We're going to take a 10-minute break. You all have worked hard today and you deserve the whole, full 10 minutes, so you're going to get a whole 10 doggone minutes. We stop this session at 4:30, and then we have public testimony, so that 10-minute break will be important because it only gives us a little bit of time to come back.

One thing I don't want to lose from Debra's point, and I alluded to it in my earlier comments --

PARTICIPANT: I thought we were going on break.

DR. TUCKSON: You are, you are, you are. But the point that Debra made, which is key, is I think we're going to start evaluating our success as a committee, and the way in which we're going to look at how we evaluate is how many of the things we recommend got done. So if we have a whole range of 99 things, some of which nobody can do anything about, our scorecard is going to look terrible. So Debra was right on on that. Thank you.

(Recess.)

DR. TUCKSON: By the way, I will let you know that we are quite happily aware that there are people out there on the webcast who are listening and sending emails in. They like Ed's tie, apparently. But there are people out there, so be aware that you are being watched. Thousands and trillions of people whom you cannot see are hanging on to your every word. So, Brad, behave. With that, we're going to go ahead and march through. Apparently, we've actually only gotten through two pages.

MS. BERRY: We'll motor through this.

The next section of the report deals with the national and local coverage decisionmaking processes that CMS undertakes. The local coverage decision process obviously allows a certain amount of flexibility, taking into account local practices, it's more rapid. The national coverage decisionmaking policy does preempt local policies and has broad applicability across the country.

This one, I don't want to jinx it or anything, but I really do think we might be able to get through, Reed, pretty quickly. We don't really have too much in the way of a formal recommendation. The task force and in our previous report, we recognized that really there probably always will be and probably always should be a healthy mix between local and national coverage decisionmaking processes. No one is advocating eliminating one or the other.

But in the new Medicare law that passed, there is a section, Section 731, which requires the development of a plan to evaluate new local coverage decisions to determine which of those should be adopted nationally, the idea being to provide greater consistency in Medicare coverage policy where appropriate and where possible.

So this pseudo-recommendation, I suppose, would be simply to encourage CMS to move forward with that plan as outlined in the Medicare law, to have the ultimate goal of trying where appropriate and where possible to have more consistency in Medicare coverage policy but retaining the local/national mix.

DR. McCABE: It's not stated in here. I think it's implicit. But given that somebody might look at the recommendation in isolation, do we want to say as it applies to genetic and genomic testing or something to that effect? Because as it reads, it really is all LCDs and NCDs, and I just think we ought to -- given the context of the report, it would obviously relate to genetics and genomics, but I think it would be good to state that in the recommendation.

MS. ZELLMER: I have a question, and Terry, maybe you can answer this. Do the Medicare policies as far as decisionmaking on these testing decisions have any influence at all over Medicaid? I realize Medicaid is largely state by state, but do they have any influence over Medicaid testing policies? Because obviously that would have a greater impact certainly on rare genetic testing, but that would involve children.

MR. KAY: At Central Office at CMS, we actually don't have a lot of detailed information on each state Medicaid program. They're basically state run. I've certainly heard that raised, that someone said that some Medicaid programs adopt Medicare policies, but I don't really have any information on that. I also just note as an example that maybe two or three years ago there were issues related to flu shots, completely different from today's issues, and there were concerns that Medicaid would be adopting some policies that Medicare had, and it turned out that was not the case at all, that the Medicaid programs maintained their own policies. So I don't think there's a direct correlation, but that's not to say that there couldn't be some states that adopt Medicare policies.

MS. BERRY: Any other comments or questions, suggestions?

(No response.)

MS. BERRY: How about if we include a revised version of this recommendation along the lines of what Ed had suggested where we do reference the genetic component. Any objection to that?

(No response.)

MS. BERRY: We're done.

The next one's going to be harder, genetic counseling. As we all know, the Medicare law, the Medicare statute, does not permit genetic counselors to directly bill Medicare, and the thinking is that that is, or certainly potentially is, a barrier to access. Reimbursement obviously, even if they could directly bill, would be limited by the other restrictions that we talked about earlier on screening tests.

We did, however, come up with a list of five potential recommendations to consider addressing that barrier with regard to genetic counseling. I'll just go through them very quickly.

Increase state licensure of certified genetic counselors. Adding genetic counselors to the list of non-physician providers eligible to bill Medicare directly. A demonstration project conducted by CMS that would examine genetic counseling, its value, and effectiveness. An Institute of Medicine study to assess the effectiveness of genetic counselors.

Then the fifth recommendation is not so much a recommendation, but more a statement of the need for consensus on which health disciplines should be providing these services, what would be the appropriate level of supervision for each, and under what conditions should they be reimbursed and should they be allowed to bill Medicare.

I'll just start off, and then jump in. The first recommendation dealing with professional licensure, I think that's a piece of a solution. I don't know if they are excluded from billing directly under Medicare. You can be licensed in the states all you want and it's not going to change that. So it's not a complete solution to that problem, but the thinking would be that it may reduce or lower the barrier to direct reimbursement, but it certainly will not guarantee that.

The question I would pose is what would be the nature of our recommendation? How do you go about increasing state licensure? There may be recommendations that would be needed, subrecommendations, under that recommendation.

The second one obviously requires a legislative fix. It would require Congress to pass a law to specifically add genetic counselors to the list of non-physician providers, and we've discussed previously that that isn't easy to do. It's not that we can't recommend it. It's not that we shouldn't. But it's not something that we should expect would happen in short order.

My only other comment would be that perhaps we might consider merging number 4 and number 5, and it doesn't have to be the Institute of Medicine, but that's something that we came up with. It could be

somebody else, but the idea would be when you study and examine genetic counselors and the services they provide and their effectiveness and their value, you might as well go ahead and do an assessment of all the health professions that provide these types of services if that's the route that we want to go.

The question is do we feel that that is necessary or is that make-work? I mean, is there any question that genetic counselors provide valuable services? I think we've spoken quite a bit in past meetings about access to genetic tests and technologies as long as people have access to the appropriate counseling, that a genetic test by itself, a consumer having direct access without the requisite counseling and medical guidance, isn't the best scenario and could do more harm than good.

So I throw that out. Those are the comments that I had on those recommendations and throw that out for discussion.

DR. WINN-DEEN: So again, I question why this is limited to Medicare. Is there something unique about Medicare that we only want to talk about genetic counseling reimbursement under Medicare or should we put this under sort of the all insurers umbrella and talk about it as a generic problem of getting reimbursement for genetic counseling? Because I think the only thing that's under the public and private thing is UPINs, which I don't think is necessarily the only issue that's coming up with genetic counseling in the private insurance sector.

So I almost think that the whole genetic counseling section should be under the umbrella of public and private, and then we can talk about if there's anything specifically unique to Medicare that we would need to make a recommendation specific for Medicare separate from a generic recommendation that counseling should be recognized and reimbursed for the value that it brings to the care.

MS. BERRY: I had the same reaction, but I thought maybe this bounced back and forth. I could be wrong, but I thought it was under the other section and there was some consideration.

I agree with you. It makes sense except to the extent that we're looking at number 2. Medicare not allowing them to bill directly is a Medicare-specific barrier, but there's nothing that precludes us, if we decided to include the genetic counseling section in the part of the report that covers both public and private, identifying what are the Medicare-specific barriers and then a potential recommendation that applies only to Medicare. I don't think we're precluded from that at all.

DR. LEONARD: My question is do we have the ability to influence private insurance companies and what they pay for and what they cover? I thought that we were directing it at Medicare because if we can change what Medicare does, then other insurers are likely to follow, but we can't mandate what they -- I mean, I don't know that we have any influence in that arena. We can make a statement that genetic counseling services are useful and will be increasingly useful and need to be covered as medical services, but I don't think we can influence that other than through Medicare.

DR. WINN-DEEN: I agree with that, but what we have under public and private insurers is just this UPIN issue, and I don't think that that's really the only issue under public and private, that the whole issue of genetic counselors' current status seems to vary by state, some have licensure, some don't -- you know, we need to get a more unified, maybe national, level of approach to it.

DR. TUCKSON: That's a great question, Debra. I think that we've got testimony and knowledge that says that one of the barriers for all payers in this area, whether it's public or private, is the concern around who is actually qualified to do genetic counseling and what is the scope of practice for them and what is the licensure.

So to the extent that we can say that we recognize this is a generic issue for all payers, and then be able to say that while there will be some specific Medicare recommendations, but that at some point this licensure issue and definition of scope of practice issue are germane to the whole field, we could solve it that way, I think.

Which sort of leads me to my comments on this. I think that what the section doesn't get at adequately enough is what are in fact the qualifications for these genetic counselors to be able to say that this is a real genetic counselor and this is what their scope of practice is.

I think there is a lot of language around distinctiveness or effectiveness of genetic counselors, and I think that we need to be a little more precise. I think that we all understand that there is, just on the face of it, a valuable role. So that's mother, God, and country in a way, but the question is who is a legitimate counselor and what should we expect? I would urge that we spend a little more attention to that.

The fifth recommendation is also I think good in that what I think it tries to get to is also this sense of what happens if you have the genetic counselor bills for the genetic counseling and then the doc decides to bill for genetic counseling and then the advanced practice nurse bills for the genetic counseling all on the same patient? Who is the accountable entity? I mean, how do you work that out? So you've got all three of them arguing with each other around who did what for the patient. So just some thought there.

But I think the real core here is if we could maybe at least be able to describe where we are with the national standards for what is a genetic counselor, and then hope that that gets translated down to the state level, maybe that's one of them.

DR. McCABE: These are both more really more technical editorial comments, but first of all, I like your bullets as opposed to these long -- I think it would be good, perhaps, to use those as headings, the kind of things you have on the board here, so that they're almost like the executive summary of the recommendation. They are the action item, as opposed to the whole paragraph for some of these.

Secondly, I would suggest that these be collapsed into one recommendation with parts A, B, C, D, E, perhaps, because by my count, so far we have two recommendations, or if we included these we'd have seven, five of which would be related to genetic counseling, and I think they're intertwined. So you could say something like "Because SACGHS recognizes that genetic counselors are critically important to the effective delivery of genetic services, therefore we make the following recommendation," A, B, et cetera, to tie them together but have them really be one recommendation with subparts.

MS. HARRISON: Just to respond to Reed's comment, I think a jumping off point that we can use as to who is qualified to do genetic counseling, I think a logical answer to that would be certified genetic counselors certified by the American Board of Genetic Counseling to do such work. That could be a

jumping off point.

Now, whether we identify some auxiliary people who can do that as well if they have particular qualifications, that's a discussion point, but I think at its very core we can identify the qualified people as being certified genetic counselors certified by the American Board.

DR. LEONARD: A question about that and then I have a different question. ISONG has genetic counselors that do cancer-related genetic counseling, and I can imagine that if there's a nurse that works in a particular type of genetic clinic, she or he may be qualified to do genetic counseling for a specific disease. How do you link those in without being a broadly trained genetic counselor? Because they actually may know more about that specific disease and the genetic counseling than a generally trained genetic counselor, and so their medical services are equally valid, and they wouldn't, I don't think, be certified by the American Board of Genetic Counseling.

MS. HARRISON: I certainly agree with that. I think we do have some kind of sister allied health people like, especially, nurses in oncology I think really provide quite a bit of information that we're not even always trained to give.

So in that, you know, I don't think particularly I have an answer for you, except that I can definitely see how we would need to make allowances for that, and I don't know if maybe with conversations between the genetics nurses and the American Board, maybe we can come to some agreement about that. I'm not sure.

DR. McCABE: I saw that as being covered under 3 and 4. I think that's where that issue would be considered and evaluated.

DR. LEONARD: Right. I was just concerned about the definition of -- I mean, my sense of genetic counselor is you think of a genetic counselor that does all kinds of genetic counseling, as opposed to these other allied health professionals that can do very focused types of genetic counseling, and I don't know how you bring them in, but I don't think they should be left out.

MS. MASNY: Well, just sort of as a clarification, even to Reed's point and your questions regarding nurses and the qualifications issue, is that the International Society of Nurses in Genetics does have a certification for nurses working in this whole area, both at an advanced practice level and at a basic level, trying to incorporate some of the recommendations of NCHPEG that all health professionals should be at least trained how to take a good family history. So there's a whole scope and standards of practice that are put out by the International Society sort of giving recommendations for what is the scope of practice for a nurse at the basic level and at the advanced level, and that a nurse who's actually practicing in this field could go on for certification. So then they would meet specific qualifications for certification, and the International Society of Nurses in Genetics has their scope and standards of practice approved through the American Nurses Association.

So I think that the other aspect or the first comment here about increasing the licensure is that nurses already are licensed in their own state, so that it gives them specific qualifications. They already have scope and standards of practice in which to work, and counseling and health promotion fall under the

purview of the nurses' field. So in many instances, nurses are being trained to provide some genetic counseling services.

So I think we would have to look at all of these aspects before we start to kind of specify just one particular discipline, and I think as Barbara pointed out, there is lots of work already underfoot looking at the collaborations between specifically nurses and genetic counselors.

PARTICIPANT: Are nurses reimbursed?

MS. MASNY: Nurse practitioners and advanced practice nurses in some states can be reimbursed for their services, but unfortunately, in the field of genetics, the question just comes that there's not reimbursement for the genetic counseling, is what the problem is.

DR. TUCKSON: But then that's the key thing here, I think, is that we're just basically saying, then, that how to get -- and I think this is a narrower part of the debate, and that is simply we're not so much interested in who can do the counseling as who can bill independently for their services, and I just don't see, quite frankly, any way that anybody is going to allow somebody to pay a bill for a professional service if there is not sufficient evidence that that person is qualified to act independently of anyone else and provide that service. I think that it's going to be a tough row to hoe.

So I think our decision that we have to make here is whether we call for this certification, as it were, to become clarified and acknowledge that that's the rate-limiting step and then sort of call for that to get fixed, or whether we can domino over that, jump over that, and say, okay, in the absence of that certification, we recommend that you just pay people. That's what I see the ultimate argument boiling down to here.

DR. FEETHAM: My comments build off of everything that you said, and that's what struck me, Reed, building off of what you just said, as I read this, is the beginning of the document focuses on access and we've talked about primary care as a point of entry and that genetic counseling and genetic services are fundamental to what is needed and the reimbursement is a major piece of it. So I would encourage, and the theme I'm now hearing, is that there's the genetic issue of access to genetic services, of which genetic counseling and bottom-line reimbursement is the basic line that, rather than focusing on just a discipline, it seems like we've jumped a whole lot and lost some of the basic essence we've been talking about in our other meetings.

DR. WINN-DEEN: So I guess the question is do we want to encourage that there is some mechanism put in place that recognizes that certain licensed health professionals -- let's just leave it open for right now -- do provide genetic counseling, that this is valued, and should be reimbursed, because certainly when you go for a genetic counseling session, this is not an office visit brief. This is generally an extended interaction and maybe even multiple interactions by the time you get through a couple of counseling sessions pre- and post-testing.

So we want to make sure that that's valued and that those people are being properly compensated. So the question I guess is do we need them to be separately billable or is the umbrella, you know, a physician supervising them the same way that the nurses in the physician offices are typically part of the "overhead"

rate that physician charges? The nurses aren't billing you for the time that you spent with them getting your height and weight taken and your blood pressure and all that.

I think we need to be very concrete about what it is that we think as a committee we want to recommend, keeping in mind that we all understand and I think there's consensus that genetic counseling is a valuable service and that it needs to be part of the continuum as appropriate for the severity of a specific condition.

MR. MARGUS: So these all seem like no-brainers to me. I think we all agree. I don't think we disagree. It's absurd that after all these sessions we've sat through that we would think that genetic counselors can't even find a box on that thing representing them.

That's a given, and then there's this more complicated thing. We've heard testimony from people who have come and talked about physicians or nurses that couldn't bill for their time. So they were running people out of their offices instead of providing counseling.

But all that being said, I appreciate whoever's idea it was to stick the couple of last bullets on where you're going to build a case for it because, to play the greedy businessman for a second, I still think you're going to get pushback where someone's going to say tomorrow we're reimbursing this whole new class of people called genetic counselors, a different species, and then we've also got a whole new category by which nurses and physicians can bill, and even if they did only the same amount of counseling that they're doing today -- a paltry amount that isn't enough, but even if they are, how much more does it cost, and then now, if the floodgates are open, how much could it really amount to that this could end up costing the system?

It's pretty daunting. I can see people justifying needing, yes, days with patients to walk them through genetics, and there's a real concern about that kind of cost and people are going to just push back completely.

So I'm 100 percent in favor of the first three, but the last two, and I don't know if the IOM -- and the big question there is, and those of you with more history on these committees can maybe say it, but maybe this has already been done numerous times, where economic modeling has been done and people have figured out what the impact would be, but if hasn't been or hasn't been done recently, it seems if you don't build a better case for it from a very cold economic point of view and quantity of care point of view, you're not going to ever sell it.

That's the scary part. I always try to think about what could the other side be thinking, as you did earlier with Congress, and why would anyone be opposed to genetic counseling if someone's got a devastating possible diagnosis or risk suddenly and why can't that be part of the treatment? The scary thing must be that there's this whole new area that people are afraid might end up costing an awful lot and it might not be properly regulated or there aren't enough standards. You know, have standards been set up that if you have this and this possible genetic risk now, you're entitled to 1.7 hours of counseling? I don't know how it works, but have those things been put in place?

People are going to be concerned that you're putting the cart in front of the horse if you haven't figured all that out, and so I'm putting a plug in for the last two, which seem to be premised on building a case for it.

MS. BERRY: Related to that, I wanted to ask and get some input on number 3, which calls upon CMS to conduct a demonstration project, and theoretically that could be part of, Brad, the idea of building a case. I question whether that's the most effective way to build the case or whether an assessment, a comprehensive study, is the most effective way to build the case.

MR. MARGUS: That one just felt really slow to me.

MS. BERRY: Which one?

MR. MARGUS: Number 3, compared to number 4.

MS. BERRY: So that's why I asked. Is that something that we feel is effective? They're not necessarily mutually exclusive, but if one is more effective than the other and if the goal in the end is the same, which is to build a case and demonstrate that there is this service that is being provided or should be provided, that it's a value to people, it helps improve access to care, and improves the quality of care, what is the best way to achieve that result? Is it through a limited demonstration project in a few cities or is it through a more comprehensive look, a la an IOM assessment?

DR. FEETHAM: Just as a reminder, I forgot to mention, remember Dr. Judith Cooksey presented last year on the study being funded through HRSA on genetic services and with counselors and other genetic specialists, and part of that study is winding down or is about at its end, but just a reminder that here we're moving forward suggesting additional activities when you may be having data from that that would help address some of the things that you're talking about.

MS. BERRY: Francis, did you have a comment?

DR. CHESLEY: Yes, and reflecting on your question, it occurs to me that what the IOM would do is look at extant evidence. So if there is evidence to demonstrate the effectiveness of such services, they will be able to put that in a neat package with a bow and present it to you. If the question is demonstrating the effectiveness through a demonstration study, that comes at it from a different perspective. Those data don't exist. We need to do a study to generate those data, which is slower.

So I think the fundamental question for the group is whether or not those data exist already -- a quick query on that and that's easy to do -- and if those data exist, that points a direction in terms of getting the underpinning for such recommendations.

DR. WINN-DEEN: So I see sort of two things. One is about Recommendation 1, which sort of deals with the whole who's qualified issue, and I think we could make a very specific recommendation that's sort of no-brainer. Anyone who is certified with some bullet point list of organizations should be qualified to deliver genetic counseling. This doesn't say get paid for it, but at least that is our endorsement, if you will, of those programs as providing some specific level of training that gives people the right qualifications.

The next question is this whole direct billing, and I think it's almost going to be impossible to justify

direct billing without having a specific set of data to support that.

So I'm less inclined to try and go for number 2 or put that as sort of the third level. So the first is identify what we believe are legitimate qualification programs, if you will. The second is where's the data that shows that this service has value, and the third then is what's the best mechanism for reimbursement? Is it still under the umbrella of the physician or should they be allowed to separately bill for some specific services?

I don't think anyone who sits down and chats with you and gives you informed consent forms to sign should necessarily be billing, but I do think that people who spend their lives working on counseling people should be able to get recognition and compensation for that.

DR. LEONARD: That was one of my concerns, is where does this go when we move toward genomic medicine? Because we think of genetic counselors now more with single-gene disease counseling and as we move more toward genomic medicine, where you can do genetic counseling for asthma and hypertension and congestive heart failure and who knows what, will that require the same level of genetic counseling and we're imagining that that will be done by primary care physicians potentially? And then we get back into the education issue, and if people aren't educated, then can they -- but in the current health care economic situation, anything else you can bill for would be highly valuable because you could get more money for the undercompensated services that we currently provide.

DR. WINN-DEEN: Right.

DR. LEONARD: So would this end up being a mechanism that would be used?

DR. WINN-DEEN: Correct me if I'm wrong. I thought PCPs really have a mechanism, although it may still be inadequate, because they can bill for office visits of varying duration. So if they are providing that discussion with the patient -- about anything, about cancer, about heart disease, about a highly-penetrant genetic disorder -- that they have a mechanism to be recognized as qualified and to get paid for the time they spend with someone. That would be really sort of focusing on the counseling part, where the counselors right now are not either officially recognized or officially able to bill and directly get compensated.

So I don't want to just miss the fact that PCPs are hopefully going to provide a lot of this counseling and certainly, in some situations, and I'd say probably in the cancer scenarios, the referring oncologist is probably providing a lot of the counseling.

DR. TUCKSON: Well, there's a question that we're really entering a whole new area, and I'm sure primary care physicians are frustrated with how much they are able to get reimbursed for cognitive, as opposed to procedural, oriented services. That's one of the tough challenges about being a primary care doc. You don't get to do stuff and the more stuff you do, the more you make, and the more you talk, the less you make.

So there's a certain imbalance in there, but I think that where I sort of hear Debra and Emily going here, and also actually Brad's point, was if you sort of fast forward this thing, when genetics is the essence of

medicine, basically then it means almost every encounter ultimately is going to have a genetic component. If this family history deal is a terrific success, then by the day after Thanksgiving, everybody is into their doc's office talking about Cousin Sue and then what do I do? And then, well, does the doc sit there and chat with you or do you refer that out like you would do a nutritional consult and expect that somebody would need to be compensated for those services?

You've just opened up a heck of box, which I don't think we're going to solve, and so I think what this really boils back down to again is, and I'm just putting something out there as a way of giving you something to shoot at for the report, do we acknowledge that this a big issue, this independent reimbursement?

Well, first of all, as you went through, Emily, that first we do support and recognize that it is important to have genetic counseling being done? Not only for all the good reasons that are mother, God, and country, but Brad, for the business reason, is in the absence of it you're going to get a lot of inappropriate testing, which costs a whole lot of money to people.

So genetic counseling, done properly, ought to decrease health care costs, not actually increase it in the aggregate. That would be the hypothesis. So we ought to encourage that.

Number 2, though, is the independent billing, we might have to acknowledge we cannot solve that problem in the absence of certain things that must be in place, and therefore, given that this is important, we would urge the logical steps to get these things in place.

I think finally, to be more complete than we are here, would be to acknowledge those efforts that are ongoing now to resolve this matter. I still have to go back and reread, I guess, the testimony from the counseling community. I just don't remember what else is going on to get national standards and get them implemented. I can't imagine that it's just laying dormant as an issue. Somebody's got to be working on this.

We might need to reference the work that's going on and then finally add to that the call for some of these demonstrative studies that we think need to be done to finally get that done. Then when that body of work is complete, then the issue of independent billing may be able to be addressed and we will take that up as events progress. That's one way to get at it.

MS. HARRISON: I just want to at least acknowledge that two states, Utah and California, have put through their state legislature processes for licensure for genetic counselors, and also there is an effort -- you're right, it's not laying dormant -- in order for genetic counselors to be able to be assigned UPIN numbers in I think 2007 when that process goes through.

But importantly, I guess I'd just be very curious to see what this final wording is because I think licensure without a strong argument for billing behind it doesn't mean very much. You know, if we're licensed, then fine, but I think it's been made very clear, as in previous testimony, that counselors are doing this job now and they're not getting reimbursed, and that's a threat to the genetic counseling community, to the patients, et cetera. So as self-serving as it may be, I think it's very important that we don't just say, well, we can't do anything about the billing, so we'll just make sure licensure is in place.

DR. TUCKSON: Well, I think that's really important. I think we need to acknowledge that explicitly as part of our narrative, that without reimbursement -- it's almost like which comes first, the chicken or the egg? One thing, what I've been saying is I guess without the licensure, it's going to be hard to get reimbursed. I think we ought to give equal credence to the fact that without reimbursement, who cares about licensure, is basically I guess what it comes down to, and we don't want to kill off or stifle the growth in the number of counselors, given that we have too few now.

I think also your point about some of the examples that are going on, hopefully we could appendix this in the report because I think probably very few people are aware of these groundbreaking initiatives in a couple of states and we ought to appendicize those to try to move this forward.

So I guess, Barbara, where I would agree with you, and at least as one person's opinion about this, is we do want to move this faster and I don't want it look like we're putting a lot of steps in place that look like we're not seeing the urgency of it. So I think we ought to be urgent and give examples of concrete things that we ought to build upon.

MR. MARGUS: Just as the complaints right now about the discrimination bill are that not enough people are being harmed, I think it would be great if we could also point to things that aren't right because genetic counselors aren't reimbursed.

I mean, you keep having Congress up there. Congress needs to respond to a problem that won't go away, and as long as it's not a very noisy problem right now, I don't think they're going to react. If we could point to serious problems and damage that's being done because people aren't getting counseling, and I'm sure there are as many good anecdotes for that as there are for discrimination and maybe more, it would be probably very helpful in stating our case.

DR. TUCKSON: Cindi, could you try to just give us, then, given that we've got to stop here and -- not got to. We're excited to stop and turn to public testimony in the last half-hour. We've given them each of them their amount of time and we're supposed to stop at 5:00. Can we challenge you right now to sort of frame where you think we might be and maybe just start right at this point tomorrow morning bright and early? But give us something to sort of help to see whether we need to noodle over this a little bit more tonight and then come right in and attack this or do you want to put this part of the report to closure right now?

MS. BERRY: I think maybe we should rework based on this discussion. I think we can merge some recommendations. I also think that what I heard, and if I did hear it correctly I agree with it, that number 2, the congressional component, is probably premature, given that we're calling for these other things as sort of a prerequisite. So we could maybe take that one out. Maybe we could noodle around and come up with some revised recommendations and put them up on the screen first thing in the morning. I'll just get with Suzanne and others and we'll try to come up with something, and then hopefully, with the limited amount of discussion tomorrow morning, we can close that out.

DR. LEONARD: But while you're doing that, I think the recommendation on page 61 about the UPIN, I mean, if we're going to talk about reimbursement, that's kind of stuck off there by itself. Can that be

incorporated into these genetic counseling recommendations as a whole?

And I really don't agree with dropping number 2 because I really think that if there is licensure, there should be payment and they have to be recognized and they have to get UPIN numbers in order to get paid. So I don't think we can take 2 off, but maybe make that that basically once there's licensure, that's the state saying these people are qualified to do this medical service that they're doing. They should get paid for it.

DR. TUCKSON: All right. Well, Cindi has got some stuff to look at tonight, and we'll watch and see how many glasses of wine she has at dinner and see what she's able to do. So thank you for that, and Cindi, thank you for volunteering to take even more responsibility.

Let me thank the committee for your hard work so far today, and now let's turn with great attentiveness to the public comment. One of our critical functions is to be able to receive input from the public, and we appreciate the views that they're sharing with us. We also have received written comments that can be found in your table folders. In the interest of time, commentators are to keep their remarks to five minutes, and as I said, we do have your written testimony which we will be looking at very carefully.

Let me say that I've got right now on my list first Kelly Ormond from the National Society of Genetic Counselors -- well timed -- Sharon Terry from the Genetic Alliance, Miriam O'Day from the Alpha-1 Association, Gary Martucci from Myriad Genetics, Christine Broderick from the National Partnership for Women and Families, and Donald Horton, director of public policy and advocacy for Laboratory Corporation of America. I'm sure that Sarah will get me if I've missed anybody else.

Let's start with Kelly Ormond from the National Society of Genetic Counselors.

MS. ORMOND: Thank you. It's a pleasure to be speaking here today. I'm Kelly Ormond, president of the National Society of Genetic Counselors.

As you are aware, the NSGC is the leading voice, authority, and advocate for the genetic counseling profession and represents over 2,000 members. Together, our members provide genetic counseling for prenatal, pediatric, and adult genetic indications, as well as work in academia, research, and biotechnology companies. A high percentage of our clinically practicing members offer some form of predispositional genetic testing on a regular basis, whether carrier testing or presymptomatic testing, for adult-onset disorders.

Today, we would like to primarily address two issues related to the provision of genetic services, genetic discrimination, and coverage and reimbursement of genetic counseling services.

Issues of billing and reimbursement are among the most pressing that face members of the NSGC and it is one of the three areas prioritized in our recent strategic plan. Through our past testimonies, this committee is already aware that coverage and reimbursement for genetic counseling services are limited by the lack of CPT codes and ineligibility for non-physician provider identification. While some payers contract directly with the health plans to include genetic counseling as a covered service and some services are covered by Medicaid and Medicare when provided to individuals with disabilities, the bulk of

genetic counseling services are not currently reimbursed.

While we have only preliminarily reviewed these newest draft recommendations, NSGC is pleased to see that SACGHS and the Secretary's Office consider ways to address these two points. In particular, we are heartened to see that SACGHS is promoting the development and funding of evidence-based studies around clinical genetic services through any agencies. NSGC offers its strong support in developing and conducting such studies and have repeatedly been told that studies documenting such value will be critical. We are also pleased that SACGHS is continuing to advocate for the inclusion of masters-trained genetic counselors as recognized providers in both private health plans and national provider identification systems. If NSGC can be of additional help as SACGHS works on these issues, including offering formal testimony on our efforts towards licensure or documentation of the value of genetic counseling, please do not hesitate to contact us.